

Predicting the effects of coding non-synonymous variants SIFT algorithm

Nature Protocols

4, 1073-1081

DOI: [10.1038/nprot.2009.86](https://doi.org/10.1038/nprot.2009.86)

Citation Report

#	ARTICLE	IF	CITATIONS
1	Visual repair of congenital aortic stenosis during hypothermia. <i>The Journal of Thoracic Surgery</i> , 1958, 35, 139-153.	0.7	33
2	Kenya: Language Situation. , 2006, , 180-182.		0
3	Nephrin mutations cause childhood- and adult-onset focal segmental glomerulosclerosis. <i>Kidney International</i> , 2009, 76, 1268-1276.	2.6	111
4	A New Era in Clinical Genetic Testing for Hypertrophic Cardiomyopathy. <i>Journal of Cardiovascular Translational Research</i> , 2009, 2, 381-391.	1.1	25
5	Recessive Mutations of the Gene TRPM1 Abrogate ON Bipolar Cell Function and Cause Complete Congenital Stationary Night Blindness in Humans. <i>American Journal of Human Genetics</i> , 2009, 85, 711-719.	2.6	172
6	Prevalence of Connexin 26 (GJB2) and Pendred (SLC26A4) Mutations in a Population of Adult Cochlear Implant Candidates. <i>Otology and Neurotology</i> , 2010, 31, 919-922.	0.7	16
7	Somatic FAS mutations are common in patients with genetically undefined autoimmune lymphoproliferative syndrome. <i>Blood</i> , 2010, 115, 5164-5169.	0.6	126
8	Brown-Vialetto-Van Laere Syndrome, a Ponto-Bulbar Palsy with Deafness, Is Caused by Mutations in C20orf54. <i>American Journal of Human Genetics</i> , 2010, 86, 485-489.	2.6	161
9	Common Variants of Large Effect in F12, KNG1, and HRC Are Associated with Activated Partial Thromboplastin Time. <i>American Journal of Human Genetics</i> , 2010, 86, 626-631.	2.6	81
10	Mutations in SCARF2 Are Responsible for Van Den Ende-Gupta Syndrome. <i>American Journal of Human Genetics</i> , 2010, 87, 553-559.	2.6	52
11	Familial tumoral calcinosis and hyperostosis hyperphosphataemia syndrome are different manifestations of the same disease: novel missense mutations in GALNT3. <i>Skeletal Radiology</i> , 2010, 39, 63-68.	1.2	32
12	WRN mutations in Werner syndrome patients: genomic rearrangements, unusual intronic mutations and ethnic-specific alterations. <i>Human Genetics</i> , 2010, 128, 103-111.	1.8	87
13	Prioritization of candidate SNPs in colon cancer using bioinformatics tools: An alternative approach for a cancer biologist. <i>Interdisciplinary Sciences, Computational Life Sciences</i> , 2010, 2, 320-346.	2.2	11
14	Variations in the Coding Region of the Agouti Signaling Protein Gene Do Not Explain Agouti/Non-agouti Phenotypes in Macaques. <i>Journal of Mammalian Evolution</i> , 2010, 17, 211-214.	1.0	4
15	Identification of a novel BRCA1 nucleotide 4803delCC/c.4684delCC mutation and a nucleotide 249T>A/c.130T>A (p.Cys44Ser) mutation in two Greenlandic Inuit families: implications for genetic screening of Greenlandic Inuit families with high risk for breast and/or ovarian cancer. <i>Breast Cancer Research and Treatment</i> , 2010, 124, 259-264.	1.1	10
16	Human allelic variation: perspective from protein function, structure, and evolution. <i>Current Opinion in Structural Biology</i> , 2010, 20, 342-350.	2.6	63
17	Protein annotation in the era of personal genomics. <i>Current Opinion in Structural Biology</i> , 2010, 20, 335-341.	2.6	1
18	Development and characterization of an oat TILLING-population and identification of mutations in lignin and Î ² -glucan biosynthesis genes. <i>BMC Plant Biology</i> , 2010, 10, 86.	1.6	90

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19	Unexpected allelic heterogeneity and spectrum of mutations in Fowler syndrome revealed by next-generation exome sequencing. <i>Human Mutation</i> , 2010, 31, 918-923.	1.1	116
20	Polymorphisms in inflammatory pathway genes and their association with colorectal cancer risk. <i>International Journal of Cancer</i> , 2010, 127, 2822-2830.	2.3	16
21	Mutations in the <i>G6PC3</i> gene cause Dursun syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2609-2611.	0.7	37
22	Mutations in <i>GRIN2A</i> and <i>GRIN2B</i> encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. <i>Nature Genetics</i> , 2010, 42, 1021-1026.	9.4	431
23	Identification of a novel mutation in the human <i>PDE6A</i> gene in autosomal recessive retinitis pigmentosa: homology with the <i>nmf28/nmf28</i> mice model. <i>Clinical Genetics</i> , 2010, 78, 495-498.	1.0	21
24	SPOT: a web-based tool for using biological databases to prioritize SNPs after a genome-wide association study. <i>Nucleic Acids Research</i> , 2010, 38, W201-W209.	6.5	57
25	PENALIZED REGRESSION FOR GENOME-WIDE ASSOCIATION SCREENING OF SEQUENCE DATA. , 2010, , 106-117.		20
26	Single nucleotide polymorphisms in <i>Wnt</i> signaling and cell death pathway genes and susceptibility to colorectal cancer. <i>Carcinogenesis</i> , 2010, 31, 1381-1386.	1.3	37
27	Feature Fusion and Selection for Recognizing Cancer-Related Mutations from Common Polymorphisms. , 2010, , .		4
28	Overcoming the Curse of Dimensionality in a Statistical Geometry Based Computational Protein Mutagenesis. , 2010, , .		0
29	Oligogenic basis of isolated gonadotropin-releasing hormone deficiency. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 15140-15144.	3.3	313
30	Novel mutations in the long isoform of the <i>USH2A</i> gene in patients with Usher syndrome type II or non-syndromic retinitis pigmentosa. <i>Journal of Medical Genetics</i> , 2010, 47, 499-506.	1.5	133
31	Diversity and evolution of 11 innate immune genes in <i>Bos taurus taurus</i> and <i>Bos taurus indicus</i> cattle. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 151-156.	3.3	81
32	Axenfeld-Rieger Syndrome Associated with Congenital Glaucoma and Cytochrome P4501B1 Gene Mutations. <i>Case Reports in Medicine</i> , 2010, 2010, 1-6.	0.3	41
33	PICMI: mapping point mutations on genomes. <i>Bioinformatics</i> , 2010, 26, 2904-2905.	1.8	4
34	Single nucleotide polymorphisms in the human $\text{Na}^+\text{-dicarboxylate}$ cotransporter affect transport activity and protein expression. <i>American Journal of Physiology - Renal Physiology</i> , 2010, 299, F704-F711.	1.3	24
35	Functional evidence implicating a novel <i>TOR1A</i> mutation in idiopathic, late-onset focal dystonia. <i>Journal of Medical Genetics</i> , 2010, 47, 646-650.	1.5	68
36	Application of second-generation sequencing to cancer genomics. <i>Briefings in Bioinformatics</i> , 2010, 11, 524-534.	3.2	42

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37	Hereditary spastic paraplegia due to SPAST mutations in 151 Dutch patients: new clinical aspects and 27 novel mutations. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, 1073-1078.	0.9	45
38	Surfactant Protein C Mutations Are the Basis of a Significant Portion of Adult Familial Pulmonary Fibrosis in a Dutch Cohort. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2010, 182, 1419-1425.	2.5	252
39	Molecular analysis of the AGL gene: Identification of 25 novel mutations and evidence of genetic heterogeneity in patients with Glycogen Storage Disease Type III. <i>Genetics in Medicine</i> , 2010, 12, 424-430.	1.1	41
40	Association of NPAS3 exonic variation with schizophrenia. <i>Schizophrenia Research</i> , 2010, 120, 143-149.	1.1	41
41	Bovipain-2, the falcipain-2 ortholog, is expressed in intraerythrocytic stages of the tick-transmitted hemoparasite <i>Babesia bovis</i> . <i>Parasites and Vectors</i> , 2010, 3, 113.	1.0	18
42	Profound biotinidase deficiency: a rare disease among native Swedes. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 175-180.	1.7	21
43	Malignant and benign mutations in familial cardiomyopathies: Insights into mutations linked to complex cardiovascular phenotypes. <i>Journal of Molecular and Cellular Cardiology</i> , 2010, 48, 899-909.	0.9	69
44	Sequencing and analysis of an Irish human genome. <i>Genome Biology</i> , 2010, 11, R91.	13.9	36
45	Great expectations: using massively parallel sequencing to solve inherited disorders. <i>Expert Review of Molecular Diagnostics</i> , 2010, 10, 833-836.	1.5	1
46	Genetics of osteoporosis: perspectives for personalized medicine. <i>Personalized Medicine</i> , 2010, 7, 655-668.	0.8	13
47	Identification of cis-regulatory sequence variations in individual genome sequences. <i>Genome Medicine</i> , 2011, 3, 65.	3.6	17
48	Copy number and targeted mutational analysis reveals novel somatic events in metastatic prostate tumors. <i>Genome Research</i> , 2011, 21, 47-55.	2.4	148
49	De Novo SYNGAP1 Mutations in Nonsyndromic Intellectual Disability and Autism. <i>Biological Psychiatry</i> , 2011, 69, 898-901.	0.7	164
50	Organic Cation Transporters (OCTs, MATEs), In Vitro and In Vivo Evidence for the Importance in Drug Therapy. <i>Handbook of Experimental Pharmacology</i> , 2011, , 105-167.	0.9	312
51	Detection of novel mutations and review of published data suggests that hereditary spastic paraplegia caused by spastin (SPAST) mutations is found more often in males. <i>Journal of the Neurological Sciences</i> , 2011, 306, 62-65.	0.3	31
52	EX-HOM (EXome HOMozygosity): A Proof of Principle. <i>Human Heredity</i> , 2011, 72, 45-53.	0.4	27
53	Mutation Analysis of SLC26A4 for Pendred Syndrome and Nonsyndromic Hearing Loss by High-Resolution Melting. <i>Journal of Molecular Diagnostics</i> , 2011, 13, 416-426.	1.2	20
54	Drug Transporters. <i>Handbook of Experimental Pharmacology</i> , 2011, , .	0.9	17

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55	NBEAL2 is mutated in gray platelet syndrome and is required for biogenesis of platelet α -granules. <i>Nature Genetics</i> , 2011, 43, 732-734.	9.4	223
56	Detection and characterization of interleukin-6 gene variants in <i>Canis familiaris</i> : Association studies with periodontal disease. <i>Gene</i> , 2011, 485, 139-145.	1.0	10
57	Nasal embryonic LHRH factor (NELF) mutations in patients with normosmic hypogonadotropic hypogonadism and Kallmann syndrome. <i>Fertility and Sterility</i> , 2011, 95, 1613-1620.e7.	0.5	57
58	Inherited mutation of the luteinizing hormone/choriogonadotropin receptor (LHCGR) in empty follicle syndrome. <i>Fertility and Sterility</i> , 2011, 96, e125-e130.	0.5	70
59	The prevalence of digenic mutations in patients with normosmic hypogonadotropic hypogonadism and Kallmann syndrome. <i>Fertility and Sterility</i> , 2011, 96, 1424-1430.e6.	0.5	89
60	Next-generation sequencing entering the clinical arena. <i>Molecular and Cellular Probes</i> , 2011, 25, 206-211.	0.9	30
61	Analysis of Disease-Linked Rhodopsin Mutations Based on Structure, Function, and Protein Stability Calculations. <i>Journal of Molecular Biology</i> , 2011, 405, 584-606.	2.0	86
62	Annotating individual human genomes. <i>Genomics</i> , 2011, 98, 233-241.	1.3	21
63	Clinical consequences of compound heterozygosity for protein S mutation Heerlen and p.Cys252Gly protein S mutation. <i>Thrombosis Research</i> , 2011, 128, 498-500.	0.8	0
64	Glucocerebrosidase mutations in diffuse Lewy body disease. <i>Parkinsonism and Related Disorders</i> , 2011, 17, 55-57.	1.1	43
65	A Novel Mutation of LAMB2 in a Multigenerational Mennonite Family Reveals a New Phenotypic Variant of Pierson Syndrome. <i>Ophthalmology</i> , 2011, 118, 1137-1144.	2.5	35
66	Targeted Next-Generation Sequencing for the Molecular Genetic Diagnostics of Cardiomyopathies. <i>Circulation: Cardiovascular Genetics</i> , 2011, 4, 110-122.	5.1	155
67	Targeted genomic capture and massively parallel sequencing to identify genes for hereditary hearing loss in middle eastern families. <i>Genome Biology</i> , 2011, 12, R89.	13.9	183
68	A probabilistic disease-gene finder for personal genomes. <i>Genome Research</i> , 2011, 21, 1529-1542.	2.4	182
69	Predicting Phenotypic Severity of Uncertain Gene Variants in the RET Proto-Oncogene. <i>PLoS ONE</i> , 2011, 6, e18380.	1.1	30
70	Molecular Evolutionary Analysis of ABCB5: The Ancestral Gene Is a Full Transporter with Potentially Deleterious Single Nucleotide Polymorphisms. <i>PLoS ONE</i> , 2011, 6, e16318.	1.1	24
71	Variation in LPA Is Associated with Lp(a) Levels in Three Populations from the Third National Health and Nutrition Examination Survey. <i>PLoS ONE</i> , 2011, 6, e16604.	1.1	34
72	Path to Facilitate the Prediction of Functional Amino Acid Substitutions in Red Blood Cell Disorders – A Computational Approach. <i>PLoS ONE</i> , 2011, 6, e24607.	1.1	36

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73	Identification and Characterization of Genes Required for Compensatory Growth in <i>Drosophila</i> . <i>Genetics</i> , 2011, 189, 1309-1326.	1.2	21
74	Differential Allelic Distribution of V-ets Erythroblastosis Virus E26 Oncogene Homolog2 (ETS2) Functional Polymorphisms in Different Group of Patients. <i>Gene Expression</i> , 2011, 15, 61-73.	0.5	3
75	The contribution of common CYP2A6 alleles to variation in nicotine metabolism among European-Americans. <i>Pharmacogenetics and Genomics</i> , 2011, 21, 403-416.	0.7	97
77	Identification of three novel plasminogen (PLG) gene mutations in a series of 23 patients with low PLG activity. <i>Thrombosis and Haemostasis</i> , 2011, 105, 454-460.	1.8	42
78	Germline mutation in BRAF codon 600 is compatible with human development: de novo p.V600G mutation identified in a patient with CFC syndrome. <i>Clinical Genetics</i> , 2011, 79, 468-474.	1.0	21
79	Exome sequencing in a family segregating for celiac disease. <i>Clinical Genetics</i> , 2011, 80, 138-147.	1.0	16
80	Mutations in the <i>PSTPIP1</i> gene and aberrant splicing variants in patients with pyoderma gangrenosum. <i>Clinical and Experimental Dermatology</i> , 2011, 36, 889-895.	0.6	46
81	HSPB1 and HSPB8 in inherited neuropathies: study of an Italian cohort of dHMN and CMT2 patients. <i>Journal of the Peripheral Nervous System</i> , 2011, 16, 287-294.	1.4	57
82	Resequencing of positional candidates identifies low frequency IL23R coding variants protecting against inflammatory bowel disease. <i>Nature Genetics</i> , 2011, 43, 43-47.	9.4	175
83	Mutations in origin recognition complex gene ORC4 cause Meier-Gorlin syndrome. <i>Nature Genetics</i> , 2011, 43, 360-364.	9.4	156
84	Exome sequencing as a tool for Mendelian disease gene discovery. <i>Nature Reviews Genetics</i> , 2011, 12, 745-755.	7.7	1,484
85	Human balanced translocation and mouse gene inactivation implicate Basonuclin 2 in distal urethral development. <i>European Journal of Human Genetics</i> , 2011, 19, 540-546.	1.4	28
86	Somatic coding mutations in human induced pluripotent stem cells. <i>Nature</i> , 2011, 471, 63-67.	18.7	1,147
87	Association of LRRK2 exonic variants with susceptibility to Parkinson's disease: a case-control study. <i>Lancet Neurology</i> , The, 2011, 10, 898-908.	4.9	294
88	Mutations in IL36RN/IL1F5 Are Associated with the Severe Episodic Inflammatory Skin Disease Known as Generalized Pustular Psoriasis. <i>American Journal of Human Genetics</i> , 2011, 89, 432-437.	2.6	468
89	Improving the Assessment of the Outcome of Nonsynonymous SNVs with a Consensus Deleteriousness Score, Condel. <i>American Journal of Human Genetics</i> , 2011, 88, 440-449.	2.6	736
90	Exome Sequencing Identifies Mitochondrial Alanine-tRNA Synthetase Mutations in Infantile Mitochondrial Cardiomyopathy. <i>American Journal of Human Genetics</i> , 2011, 88, 635-642.	2.6	229
91	DASH: A Method for Identical-by-Descent Haplotype Mapping Uncovers Association with Recent Variation. <i>American Journal of Human Genetics</i> , 2011, 88, 706-717.	2.6	77

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92	Mutations in PRDM5 in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. <i>American Journal of Human Genetics</i> , 2011, 88, 767-777.	2.6	106
93	Rare-Variant Association Testing for Sequencing Data with the Sequence Kernel Association Test. <i>American Journal of Human Genetics</i> , 2011, 89, 82-93.	2.6	2,060
94	Mutations Causing Familial Biparental Hydatidiform Mole Implicate C6orf221 as a Possible Regulator of Genomic Imprinting in the Human Oocyte. <i>American Journal of Human Genetics</i> , 2011, 89, 451-458.	2.6	207
95	ST3GAL3 Mutations Impair the Development of Higher Cognitive Functions. <i>American Journal of Human Genetics</i> , 2011, 89, 407-414.	2.6	89
96	XX Ovarian Dysgenesis Is Caused by a PSMC3IP/HOP2 Mutation that Abolishes Coactivation of Estrogen-Driven Transcription. <i>American Journal of Human Genetics</i> , 2011, 89, 572-579.	2.6	99
97	Abundant Pleiotropy in Human Complex Diseases and Traits. <i>American Journal of Human Genetics</i> , 2011, 89, 607-618.	2.6	478
98	Finding Disease Variants in Mendelian Disorders By Using Sequence Data: Methods and Applications. <i>American Journal of Human Genetics</i> , 2011, 89, 701-712.	2.6	50
99	Non-truncating hMLH1 variants identified in Slovenian gastric cancer patients are not associated with Lynch Syndrome: a functional analysis report. <i>Familial Cancer</i> , 2011, 10, 255-263.	0.9	6
100	BRIP1, PALB2, and RAD51C mutation analysis reveals their relative importance as genetic susceptibility factors for breast cancer. <i>Breast Cancer Research and Treatment</i> , 2011, 127, 853-859.	1.1	95
101	Hyperuricemia cosegregating with osteogenesis imperfecta is associated with a mutation in GPATCH8. <i>Human Genetics</i> , 2011, 130, 671-683.	1.8	8
102	Homozygosity for the MTX1 c.184T>A (p.S63T) alteration modifies the age of onset in GBA-associated Parkinson's disease. <i>Neurogenetics</i> , 2011, 12, 325-332.	0.7	15
103	Personalized Medicine and Cardiovascular Disease: From Genome to Bedside. <i>Current Cardiovascular Risk Reports</i> , 2011, 5, 542-551.	0.8	1
104	Ryanodine receptor type 1 gene mutations found in the Canadian malignant hyperthermia population. <i>Canadian Journal of Anaesthesia</i> , 2011, 58, 504-513.	0.7	44
105	Cataloguing functionally relevant polymorphisms in gene DNA ligase I: a computational approach. <i>3 Biotech</i> , 2011, 1, 47-56.	1.1	5
106	Computational analysis of a novel mutation in ETFDH gene highlights its long-range effects on the FAD-binding motif. <i>BMC Structural Biology</i> , 2011, 11, 43.	2.3	24
107	Incorporating predicted functions of nonsynonymous variants into gene-based analysis of exome sequencing data: a comparative study. <i>BMC Proceedings</i> , 2011, 5, S20.	1.8	18
108	PileLine: a toolbox to handle genome position information in next-generation sequencing studies. <i>BMC Bioinformatics</i> , 2011, 12, 31.	1.2	10
109	Changes in predicted protein disorder tendency may contribute to disease risk. <i>BMC Genomics</i> , 2011, 12, S2.	1.2	15

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110	Whole-exome sequencing identifies compound heterozygous mutations in <i>WDR62</i> in siblings with recurrent polymicrogyria. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2071-2077.	0.7	38
111	Incorporating biological information into association studies of sequencing data. <i>Genetic Epidemiology</i> , 2011, 35, S29-34.	0.6	4
112	Novel genomic techniques open new avenues in the analysis of monogenic disorders. <i>Human Mutation</i> , 2011, 32, 144-151.	1.1	102
113	dbNSFP: A lightweight database of human nonsynonymous SNPs and their functional predictions. <i>Human Mutation</i> , 2011, 32, 894-899.	1.1	706
114	CUBN Is a Gene Locus for Albuminuria. <i>Journal of the American Society of Nephrology: JASN</i> , 2011, 22, 555-570.	3.0	208
115	GnRH-Deficient Phenotypes in Humans and Mice with Heterozygous Variants in <i>KISS1</i> . <i>Journal of Clinical Endocrinology and Metabolism</i> , 2011, 96, E1771-E1781.	1.8	59
116	Clinical Value of NPHS2 Analysis in Early- and Adult-Onset Steroid-Resistant Nephrotic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2011, 6, 344-354.	2.2	65
117	GAMES identifies and annotates mutations in next-generation sequencing projects. <i>Bioinformatics</i> , 2011, 27, 9-13.	1.8	28
118	Identification of a novel candidate gene for non-syndromic autosomal recessive intellectual disability: the WASH complex member SWIP. <i>Human Molecular Genetics</i> , 2011, 20, 2585-2590.	1.4	78
119	ICSNPathway: identify candidate causal SNPs and pathways from genome-wide association study by one analytical framework. <i>Nucleic Acids Research</i> , 2011, 39, W437-W443.	6.5	69
120	<i>DICER1</i> ; Mutations in Familial Multinodular Goiter With and Without Ovarian Sertoli-Leydig Cell Tumors. <i>JAMA - Journal of the American Medical Association</i> , 2011, 305, 68.	3.8	284
121	Three novel mutations in <i>ASIP</i> associated with black fibre in alpacas (<i>Vicugna pacos</i>). <i>Journal of Agricultural Science</i> , 2011, 149, 529-538.	0.6	47
122	Diazoxide-Unresponsive Congenital Hyperinsulinism in Children With Dominant Mutations of the β -Cell Sulfonylurea Receptor SUR1. <i>Diabetes</i> , 2011, 60, 1797-1804.	0.3	66
123	A Survey of DNA Variation of <i>C2ORF71</i> in Proband with Progressive Autosomal Recessive Retinal Degeneration and Controls. , 2011, 52, 1880.		12
124	The zebrafish <i>dag1</i> mutant: a novel genetic model for dystroglycanopathies. <i>Human Molecular Genetics</i> , 2011, 20, 1712-1725.	1.4	101
125	Molecular Analysis of Mutant Alleles for Elevated Palmitate Concentration in Soybean. <i>Crop Science</i> , 2011, 51, 2554-2560.	0.8	6
126	Positive Signature-Tagged Mutagenesis in <i>Pseudomonas aeruginosa</i> : Tracking Patho-Adaptive Mutations Promoting Airways Chronic Infection. <i>PLoS Pathogens</i> , 2011, 7, e1001270.	2.1	55
127	Interplay between BRCA1 and RHAMM Regulates Epithelial Apicobasal Polarization and May Influence Risk of Breast Cancer. <i>PLoS Biology</i> , 2011, 9, e1001199.	2.6	91

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128	A Genome-Wide Metabolic QTL Analysis in Europeans Implicates Two Loci Shaped by Recent Positive Selection. <i>PLoS Genetics</i> , 2011, 7, e1002270.	1.5	132
129	Matrix metalloproteinase genes on chromosome 11q22 and the risk of anterior cruciate ligament (ACL) rupture. <i>British Journal of Sports Medicine</i> , 2011, 45, 321-321.	3.1	0
130	PileLineGUI: a desktop environment for handling genome position files in next-generation sequencing studies. <i>Nucleic Acids Research</i> , 2011, 39, W562-W566.	6.5	6
131	Integrating Rare-Variant Testing, Function Prediction, and Gene Network in Composite Resequencing-Based Genome-Wide Association Studies (CR-GWAS). <i>G3: Genes, Genomes, Genetics</i> , 2011, 1, 233-243.	0.8	16
132	Sun-Induced Nonsynonymous p53 Mutations Are Extensively Accumulated and Tolerated in Normal Appearing Human Skin. <i>Journal of Investigative Dermatology</i> , 2011, 131, 504-508.	0.3	49
133	Mapping of the Disease Locus and Identification of ADAMTS10 As a Candidate Gene in a Canine Model of Primary Open Angle Glaucoma. <i>PLoS Genetics</i> , 2011, 7, e1001306.	1.5	96
134	Identity-by-descent filtering of exome sequence data for disease-associated gene identification in autosomal recessive disorders. <i>Bioinformatics</i> , 2011, 27, 829-836.	1.8	30
135	Distinct genetic association at the PLCE1 locus with oesophageal squamous cell carcinoma in the South African population. <i>Carcinogenesis</i> , 2012, 33, 2155-2161.	1.3	44
136	Novel Mutations in the Glucocerebrosidase Gene of Brazilian Patients with Gaucher Disease. <i>JIMD Reports</i> , 2012, 9, 7-16.	0.7	19
137	A SEL1L Mutation Links a Canine Progressive Early-Onset Cerebellar Ataxia to the Endoplasmic Reticulum-associated Protein Degradation (ERAD) Machinery. <i>PLoS Genetics</i> , 2012, 8, e1002759.	1.5	52
138	HmtDB, a genomic resource for mitochondrion-based human variability studies. <i>Nucleic Acids Research</i> , 2012, 40, D1150-D1159.	6.5	82
139	Disease-Associated Mutations Disrupt Functionally Important Regions of Intrinsic Protein Disorder. <i>PLoS Computational Biology</i> , 2012, 8, e1002709.	1.5	123
140	Rare Copy Number Variations in Adults with Tetralogy of Fallot Implicate Novel Risk Gene Pathways. <i>PLoS Genetics</i> , 2012, 8, e1002843.	1.5	149
141	Recessive Mutations in SPTBN2 Implicate Î²-III Spectrin in Both Cognitive and Motor Development. <i>PLoS Genetics</i> , 2012, 8, e1003074.	1.5	94
142	Genomics of Acute Myeloid Leukemia: The Next Generation. <i>Frontiers in Oncology</i> , 2012, 2, 40.	1.3	32
143	Phylogenetic and Physicochemical Analyses Enhance the Classification of Rare Nonsynonymous Single Nucleotide Variants in Type 1 and 2 Long-QT Syndrome. <i>Circulation: Cardiovascular Genetics</i> , 2012, 5, 519-528.	5.1	61
144	Heuristic Methods for Finding Pathogenic Variants in Gene Coding Sequences. <i>Journal of the American Heart Association</i> , 2012, 1, e002642.	1.6	12
145	Taxonomizing, sizing, and overcoming the incidentalome. <i>Genetics in Medicine</i> , 2012, 14, 399-404.	1.1	102

#	ARTICLE	IF	CITATIONS
146	Mutations in <i>TMEM231</i> cause Joubert syndrome in French Canadians. <i>Journal of Medical Genetics</i> , 2012, 49, 636-641.	1.5	72
147	Use of whole exome and genome sequencing in the identification of genetic causes of primary immunodeficiencies. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2012, 12, 623-628.	1.1	65
148	Heterozygous <i>TBK1</i> mutations impair TLR3 immunity and underlie herpes simplex encephalitis of childhood. <i>Journal of Experimental Medicine</i> , 2012, 209, 1567-1582.	4.2	231
149	A conserved eEF2 coding variant in SCA26 leads to loss of translational fidelity and increased susceptibility to proteostatic insult. <i>Human Molecular Genetics</i> , 2012, 21, 5472-5483.	1.4	58
150	Exploiting the Mutanome for Tumor Vaccination. <i>Cancer Research</i> , 2012, 72, 1081-1091.	0.4	706
151	Recessive Mutations in <i>TSPAN12</i> Cause Retinal Dysplasia and Severe Familial Exudative Vitreoretinopathy (FEVR). , 2012, 53, 2873.		64
152	â€˜Cone dystrophy with supranormal rod responseâ€™™ in children. <i>British Journal of Ophthalmology</i> , 2012, 96, 422-426.	2.1	34
153	TREAT: a bioinformatics tool for variant annotations and visualizations in targeted and exome sequencing data. <i>Bioinformatics</i> , 2012, 28, 277-278.	1.8	59
154	A comprehensive framework for prioritizing variants in exome sequencing studies of Mendelian diseases. <i>Nucleic Acids Research</i> , 2012, 40, e53-e53.	6.5	229
155	The Next Generation of Complex Lung Genetic Studies. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2012, 186, 1087-1094.	2.5	18
156	Disruption of <i>RAB40AL</i> function leads to Martinâ€™s Probst syndrome, a rare X-linked multisystem neurodevelopmental human disorder. <i>Journal of Medical Genetics</i> , 2012, 49, 332-340.	1.5	17
157	Fine mapping and conditional analysis identify a new mutation in the autoimmunity susceptibility gene <i>BLK</i> that leads to reduced half-life of the <i>BLK</i> protein. <i>Annals of the Rheumatic Diseases</i> , 2012, 71, 1219-1226.	0.5	33
158	Whole genome sequencing of matched primary and metastatic acral melanomas. <i>Genome Research</i> , 2012, 22, 196-207.	2.4	155
159	Application of Genomics in the Prevention, Treatment and Management of Achilles Tendinopathy and Anterior Cruciate Ligament Ruptures. <i>Recent Patents on DNA & Gene Sequences</i> , 2012, 6, 216-223.	0.7	41
160	Mutations in <i>DYNC1H1</i> cause severe intellectual disability with neuronal migration defects. <i>Journal of Medical Genetics</i> , 2012, 49, 179-183.	1.5	151
161	Hirschsprungâ€™s disease and variants in genes that regulate enteric neural crest cell proliferation, migration and differentiation. <i>Journal of Human Genetics</i> , 2012, 57, 485-493.	1.1	30
162	Explore coronary artery disease related microRNA clusters by combing single nucleotide polymorphisms with microRNA microarray. , 2012, , .		0
163	Mutations in the <i>CYP1B1</i> gene may contribute to juvenile-onset open-angle glaucoma. <i>Eye</i> , 2012, 26, 1369-1377.	1.1	26

#	ARTICLE	IF	CITATIONS
164	Germline BRCA1 mutations increase prostate cancer risk. British Journal of Cancer, 2012, 106, 1697-1701.	2.9	251
165	PARADIGM-SHIFT predicts the function of mutations in multiple cancers using pathway impact analysis. Bioinformatics, 2012, 28, i640-i646.	1.8	94
166	Paternal uniparental isodisomy of chromosome 22 in a patient with metachromatic leukodystrophy. Journal of Human Genetics, 2012, 57, 687-690.	1.1	10
167	The NPM1 wild-type OCI-AML2 and the NPM1-mutated OCI-AML3 cell lines carry DNMT3A mutations. Leukemia, 2012, 26, 554-557.	3.3	44
168	A Genotype-Phenotype Comparison of <i>ADAMTSL4</i> and <i>FBN1</i> in Isolated Ectopia Lentis. , 2012, 53, 4889.		58
169	SNPnexus: a web server for functional annotation of novel and publicly known genetic variants (2012) Tj ETQq1 1 0.784314 rgBT /Overl 6.5 181		
170	Whole-exome sequencing of human pancreatic cancers and characterization of genomic instability caused by <i>MLH1</i> haploinsufficiency and complete deficiency. Genome Research, 2012, 22, 208-219.	2.4	107
171	Cardiac Structural and Sarcomere Genes Associated With Cardiomyopathy Exhibit Marked Intolerance of Genetic Variation. Circulation: Cardiovascular Genetics, 2012, 5, 602-610.	5.1	59
172	Genome and transcriptome sequencing of lung cancers reveal diverse mutational and splicing events. Genome Research, 2012, 22, 2315-2327.	2.4	177
173	Genetic Overlap in Kallmann Syndrome, Combined Pituitary Hormone Deficiency, and Septo-Optic Dysplasia. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E694-E699.	1.8	136
174	Exome Analysis of a Family With Pleiotropic Congenital Heart Disease. Circulation: Cardiovascular Genetics, 2012, 5, 175-182.	5.1	65
175	Study of FTMT and ABCA4 genes in a patient affected by age-related macular degeneration: identification and analysis of new mutations. Clinical Chemistry and Laboratory Medicine, 2012, 50, 1021-9.	1.4	24
176	A Bioinformatics Procedure to Identify and Annotate Somatic Mutations in Whole-Exome Sequencing Data. Lecture Notes in Computer Science, 2012, , 73-82.	1.0	0
177	Functional impact bias reveals cancer drivers. Nucleic Acids Research, 2012, 40, e169-e169.	6.5	304
178	Diagnosis of Fanconi Anemia: Mutation Analysis by Next-Generation Sequencing. Anemia, 2012, 2012, 1-7.	0.5	44
179	Analyzing Effects of Naturally Occurring Missense Mutations. Computational and Mathematical Methods in Medicine, 2012, 2012, 1-15.	0.7	111
180	Computational Refinement of Functional Single Nucleotide Polymorphisms Associated with ATM Gene. PLoS ONE, 2012, 7, e34573.	1.1	38
181	Human POLB Gene Is Mutated in High Percentage of Colorectal Tumors. Journal of Biological Chemistry, 2012, 287, 23830-23839.	1.6	83

#	ARTICLE	IF	CITATIONS
182	Performance of computational tools in evaluating the functional impact of laboratory-induced amino acid mutations. <i>Bioinformatics</i> , 2012, 28, 2093-2096.	1.8	51
183	Predominance of pathogenic missense variants in the RAD51C gene occurring in breast and ovarian cancer families. <i>Human Molecular Genetics</i> , 2012, 21, 2889-2898.	1.4	84
184	Incorporating Prior Biologic Information for High-Dimensional Rare Variant Association Studies. <i>Human Heredity</i> , 2012, 74, 184-195.	0.4	19
185	Advances in genetics show the need for extending screening strategies for autosomal dominant hypercholesterolaemia. <i>European Heart Journal</i> , 2012, 33, 1360-1366.	1.0	76
186	Recessive germline <i>SDHA</i> and <i>SDHB</i> mutations causing leukodystrophy and isolated mitochondrial complex II deficiency. <i>Journal of Medical Genetics</i> , 2012, 49, 569-577.	1.5	100
187	Mutations in DNMT3A and loss of RKIP are independent events in acute monocytic leukemia. <i>Haematologica</i> , 2012, 97, 1936-1937.	1.7	10
188	α 1-Antitrypsin Deficiency in Fraternal Twins Born With Familial Spontaneous Pneumothorax. <i>Chest</i> , 2012, 141, 239-241.	0.4	5
189	MSV3d: database of human MisSense variants mapped to 3D protein structure. <i>Database: the Journal of Biological Databases and Curation</i> , 2012, 2012, bas018-bas018.	1.4	24
190	PharmGKB summary. <i>Pharmacogenetics and Genomics</i> , 2012, 22, 159-165.	0.7	141
191	An Elderly Japanese Patient with Adult-onset Type II Citrullinemia with a Novel D493G Mutation in the <i>SLC25A13</i> Gene. <i>Internal Medicine</i> , 2012, 51, 2131-2134.	0.3	10
192	Mitochondrial DNA Sequence Variation Associated with Dementia and Cognitive Function in the Elderly. <i>Journal of Alzheimer's Disease</i> , 2012, 32, 357-372.	1.2	37
193	Identification of the transcript isoforms and expression characteristics for chicken Lpin1. <i>Animal</i> , 2012, 6, 1897-1903.	1.3	9
194	Novel Compound Heterozygous Mutations in the Cathepsin K Gene in Japanese Female Siblings with Pyknodysostosis. <i>Molecular Syndromology</i> , 2011, 2, 254-258.	0.3	5
195	Whole-genome sequencing of multiple myeloma from diagnosis to plasma cell leukemia reveals genomic initiating events, evolution, and clonal tides. <i>Blood</i> , 2012, 120, 1060-1066.	0.6	357
196	A bioinformatics strategy for detecting the complexity of Chronic Obstructive Pulmonary Disease in Northern Chinese Han Population. <i>Genes and Genetic Systems</i> , 2012, 87, 197-209.	0.2	3
197	Genome-Wide Patterns of Intrahuman Dengue Virus Diversity Reveal Associations with Viral Phylogenetic Clade and Interhost Diversity. <i>Journal of Virology</i> , 2012, 86, 8546-8558.	1.5	78
198	A novel autosomal dominant <i>GDAP1</i> mutation in an Italian CMT2 family. <i>Journal of the Peripheral Nervous System</i> , 2012, 17, 351-355.	1.4	13
199	A genome-wide association study of venous thromboembolism identifies risk variants in chromosomes 1q24.2 and 9q. <i>Journal of Thrombosis and Haemostasis</i> , 2012, 10, 1521-1531.	1.9	138

#	ARTICLE	IF	CITATIONS
200	Short communication: A new bovine milk-protein variant: Î± ₁ -Lactalbumin variant D. <i>Journal of Dairy Science</i> , 2012, 95, 2165-2169.	1.4	17
201	So many doggone traits: mapping genetics of multiple phenotypes in the domestic dog. <i>Human Molecular Genetics</i> , 2012, 21, R52-R57.	1.4	32
202	A Two-Stage Evaluation of Genetic Variation in Immune and Inflammation Genes with Risk of Non-Hodgkin Lymphoma Identifies New Susceptibility Locus in 6p21.3 Region. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012, 21, 1799-1806.	1.1	22
203	Mutations in the profilin 1 gene cause familial amyotrophic lateral sclerosis. <i>Nature</i> , 2012, 488, 499-503.	13.7	522
204	Association of melanocortin 1 receptor gene (MC1R) polymorphisms with skin reflectance and freckles in Japanese. <i>Journal of Human Genetics</i> , 2012, 57, 700-708.	1.1	31
205	Predicting the effects of frameshifting indels. <i>Genome Biology</i> , 2012, 13, R9.	13.9	99
206	The genetic landscape of mutations in Burkitt lymphoma. <i>Nature Genetics</i> , 2012, 44, 1321-1325.	9.4	517
207	Mutations in potassium channel <i>kcnd3</i> cause spinocerebellar ataxia type 19. <i>Annals of Neurology</i> , 2012, 72, 870-880.	2.8	121
208	The Expanding Role of MBD Genes in Autism: Identification of a MECP2 Duplication and Novel Alterations in MBD5, MBD6, and SETDB1. <i>Autism Research</i> , 2012, 5, 385-397.	2.1	81
209	The insulin-like growth factor 1 receptor (IGF1R) contributes to reduced size in dogs. <i>Mammalian Genome</i> , 2012, 23, 780-790.	1.0	98
210	Novel C3 mutation p.Lys65Gln in aHUS affects complement factor H binding. <i>Pediatric Nephrology</i> , 2012, 27, 1519-1524.	0.9	38
211	Genetic analysis of familial hypercholesterolaemia in Western Australia. <i>Atherosclerosis</i> , 2012, 224, 430-434.	0.4	61
212	Biochemical and molecular characterization of GALT gene from Indian galactosemia patients: Identification of 10 novel mutations and their structural and functional implications. <i>Clinica Chimica Acta</i> , 2012, 414, 191-196.	0.5	16
213	In silico prediction of a disease-associated STIL mutant and its affect on the recruitment of centromere protein J (CENPJ). <i>FEBS Open Bio</i> , 2012, 2, 285-293.	1.0	53
214	Molecular genetic studies of complex phenotypes. <i>Translational Research</i> , 2012, 159, 64-79.	2.2	121
215	Novel pathogenic mutations in the glucocerebrosidase locus. <i>Molecular Genetics and Metabolism</i> , 2012, 106, 495-497.	0.5	5
216	MuSiC: Identifying mutational significance in cancer genomes. <i>Genome Research</i> , 2012, 22, 1589-1598.	2.4	586
217	ColoSeq Provides Comprehensive Lynch and Polyposis Syndrome Mutational Analysis Using Massively Parallel Sequencing. <i>Journal of Molecular Diagnostics</i> , 2012, 14, 357-366.	1.2	179

#	ARTICLE	IF	CITATIONS
218	Identifying disease mutations in genomic medicine settings: current challenges and how to accelerate progress. <i>Genome Medicine</i> , 2012, 4, 58.	3.6	68
219	SNPEffect 4.0: on-line prediction of molecular and structural effects of protein-coding variants. <i>Nucleic Acids Research</i> , 2012, 40, D935-D939.	6.5	235
220	SIFT web server: predicting effects of amino acid substitutions on proteins. <i>Nucleic Acids Research</i> , 2012, 40, W452-W457.	6.5	1,838
221	Ensembl 2012. <i>Nucleic Acids Research</i> , 2012, 40, D84-D90.	6.5	840
222	The 1000 Genomes Project: data management and community access. <i>Nature Methods</i> , 2012, 9, 459-462.	9.0	308
223	Ensembl 2013. <i>Nucleic Acids Research</i> , 2012, 41, D48-D55.	6.5	856
224	COMPUTER-AIDED PROTEIN DIRECTED EVOLUTION: A REVIEW OF WEB SERVERS, DATABASES AND OTHER COMPUTATIONAL TOOLS FOR PROTEIN ENGINEERING. <i>Computational and Structural Biotechnology Journal</i> , 2012, 2, e201209008.	1.9	52
225	Screening of the SOD1, FUS, TARDBP, ANG, and OPTN mutations in Korean patients with familial and sporadic ALS. <i>Neurobiology of Aging</i> , 2012, 33, 1017.e17-1017.e23.	1.5	74
226	Retinal Structure, Function, and Molecular Pathologic Features in Gyrate Atrophy. <i>Ophthalmology</i> , 2012, 119, 596-605.	2.5	89
227	Genotype-phenotype correlations in THAP1 dystonia: Molecular foundations and description of new cases. <i>Parkinsonism and Related Disorders</i> , 2012, 18, 414-425.	1.1	74
228	A single mutation in MCCC1 or MCCC2 as a potential cause of positive screening for 3-methylcrotonyl-CoA carboxylase deficiency. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 602-606.	0.5	24
229	Biochemical, molecular, and clinical characteristics of children with short chain acyl-CoA dehydrogenase deficiency detected by newborn screening in California. <i>Molecular Genetics and Metabolism</i> , 2012, 106, 55-61.	0.5	65
230	The Results of CHD7 Analysis in Clinically Well-Characterized Patients with Kallmann Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012, 97, E858-E862.	1.8	69
231	Exome sequencing identifies a novel multiple sclerosis susceptibility variant in the TYK2 gene. <i>Neurology</i> , 2012, 79, 406-411.	1.5	56
232	Relevance of SOX17 Variants for Hypomyelinating Leukodystrophies and Congenital Anomalies of the Kidney and Urinary Tract (CAKUT). <i>Annals of Human Genetics</i> , 2012, 76, 261-267.	0.3	5
233	Prevalence of Sequence Variants in the RAS-Mitogen Activated Protein Kinase Signaling Pathway in Pre-Adolescent Children With Hypertrophic Cardiomyopathy. <i>Circulation: Cardiovascular Genetics</i> , 2012, 5, 317-326.	5.1	23
234	Applying next-generation sequencing to pancreatic cancer treatment. <i>Nature Reviews Gastroenterology and Hepatology</i> , 2012, 9, 477-486.	8.2	41
235	Globozoospermia is mainly due to DPY19L2 deletion via non-allelic homologous recombination involving two recombination hotspots. <i>Human Molecular Genetics</i> , 2012, 21, 3695-3702.	1.4	100

#	ARTICLE	IF	CITATIONS
236	HOXB1 Founder Mutation in Humans Recapitulates the Phenotype of Hoxb1 Mice. American Journal of Human Genetics, 2012, 91, 171-179.	2.6	72
237	Epistasis between the HSD17B4 and TG polymorphisms is associated with premature ovarian failure. Fertility and Sterility, 2012, 97, 968-973.	0.5	13
238	Missense substitutions associated with behavioural disturbances in Alzheimer's disease (AD). Brain Research Bulletin, 2012, 88, 394-405.	1.4	6
239	Identification of rare and frequent variants of the CASR gene by high-resolution melting. Clinica Chimica Acta, 2012, 413, 605-611.	0.5	16
240	Single-Cell Exome Sequencing Reveals Single-Nucleotide Mutation Characteristics of a Kidney Tumor. Cell, 2012, 148, 886-895.	13.5	622
241	Single-Cell Exome Sequencing and Monoclonal Evolution of a JAK2-Negative Myeloproliferative Neoplasm. Cell, 2012, 148, 873-885.	13.5	503
242	Polymorphisms in MUC1, MUC2, MUC5B and MUC6 genes are not associated with the risk of chronic atrophic gastritis. European Journal of Cancer, 2012, 48, 114-120.	1.3	10
243	Three novel BRCA1/BRCA2 mutations in breast/ovarian cancer families in Croatia. Gene, 2012, 498, 169-176.	1.0	10
244	Computational investigation of pathogenic nsSNPs in CEP63 protein. Gene, 2012, 503, 75-82.	1.0	47
245	Excess of Rare Variants in Non-Genome-Wide Association Study Candidate Genes in Patients With Hypertriglyceridemia. Circulation: Cardiovascular Genetics, 2012, 5, 66-72.	5.1	79
246	Mutations in axonemal dynein assembly factor DNAAF3 cause primary ciliary dyskinesia. Nature Genetics, 2012, 44, 381-389.	9.4	231
247	Analysis of the <i>WISP3</i> gene in Indian families with progressive pseudorheumatoid dysplasia. American Journal of Medical Genetics, Part A, 2012, 158A, 2820-2828.	0.7	63
248	Rare variants in <i>TMEM132D</i> in a case-control sample for panic disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 896-907.	1.1	25
249	SNP Set Association Analysis for Familial Data. Genetic Epidemiology, 2012, 36, 797-810.	0.6	100
250	Chromosome dynamic changes in two cultured chinese human embryonic stem cell lines: Single nucleotide polymorphism, copy number variation and loss of heterozygosity. Journal of Cellular Biochemistry, 2012, 113, 3520-3527.	1.2	7
251	In Silico profiling of deleterious amino acid substitutions of potential pathological importance in haemophilia A and haemophilia B. Journal of Biomedical Science, 2012, 19, 30.	2.6	18
252	Heterozygosity for E292V in ABCA3, lung function and COPD in 64,000 individuals. Respiratory Research, 2012, 13, 67.	1.4	18
253	Personal receptor repertoires: olfaction as a model. BMC Genomics, 2012, 13, 414.	1.2	92

#	ARTICLE	IF	CITATIONS
254	Comparative genomics and transcriptomics of trait-gene association. <i>BMC Genomics</i> , 2012, 13, 669.	1.2	16
255	A diploid wheat TILLING resource for wheat functional genomics. <i>BMC Plant Biology</i> , 2012, 12, 205.	1.6	79
256	Founder mutations characterise the mutation panorama in 200 Swedish index cases referred for Long QT syndrome genetic testing. <i>BMC Cardiovascular Disorders</i> , 2012, 12, 95.	0.7	25
257	Linkage disequilibrium analysis reveals an albuminuria risk haplotype containing three missense mutations in the cubilin gene with striking differences among European and African ancestry populations. <i>BMC Nephrology</i> , 2012, 13, 142.	0.8	7
258	SNPs in the coding region of the metastasis-inducing gene <i>MACC1</i> and clinical outcome in colorectal cancer. <i>Molecular Cancer</i> , 2012, 11, 49.	7.9	31
259	A High-Coverage Genome Sequence from an Archaic Denisovan Individual. <i>Science</i> , 2012, 338, 222-226.	6.0	1,695
260	Common variants within 6p21.31 locus are associated with chronic lymphocytic leukaemia and, potentially, other non-Hodgkin lymphoma subtypes. <i>British Journal of Haematology</i> , 2012, 159, n/a-n/a.	1.2	13
261	Spectra of <i>BRCA1</i> and <i>BRCA2</i> mutations in Korean patients with breast cancer: the importance of whole-gene sequencing. <i>Journal of Human Genetics</i> , 2012, 57, 212-215.	1.1	12
262	An integrated approach for classifying mitochondrial DNA variants: one clinical diagnostic laboratory's experience. <i>Genetics in Medicine</i> , 2012, 14, 620-626.	1.1	39
263	Doubling the referral rate of monogenic diabetes through a nationwide information campaign - update on glucokinase gene mutations in a Polish cohort. <i>Clinical Genetics</i> , 2012, 82, 587-590.	1.0	9
264	Congenital Hypothyroidism with Goiter in Tenterfield Terriers. <i>Journal of Veterinary Internal Medicine</i> , 2012, 26, 1350-1357.	0.6	14
265	Frequent somatic mutations in <i>MAP3K5</i> and <i>MAP3K9</i> in metastatic melanoma identified by exome sequencing. <i>Nature Genetics</i> , 2012, 44, 165-169.	9.4	170
266	Applying In Silico Integrative Genomics to Genetic Studies of Human Disease. <i>International Review of Neurobiology</i> , 2012, 103, 133-156.	0.9	1
267	Personalizing rare disease research: how genomics is revolutionizing the diagnosis and treatment of rare disease. <i>Personalized Medicine</i> , 2012, 9, 805-819.	0.8	8
268	Next-generation sequencing: impact of exome sequencing in characterizing Mendelian disorders. <i>Journal of Human Genetics</i> , 2012, 57, 621-632.	1.1	177
269	Sub-cellular localization analysis of <i>MSH6</i> missense mutations does not reveal an overt <i>MSH6</i> nuclear transport impairment. <i>Familial Cancer</i> , 2012, 11, 675-680.	0.9	2
270	Dysfunction of lipid sensor <i>GPR120</i> leads to obesity in both mouse and human. <i>Nature</i> , 2012, 483, 350-354.	13.7	572
271	Meta-analyses identify 13 loci associated with age at menopause and highlight DNA repair and immune pathways. <i>Nature Genetics</i> , 2012, 44, 260-268.	9.4	303

#	ARTICLE	IF	CITATIONS
272	Heterozygous de-novo mutations in ATP1A3 in patients with alternating hemiplegia of childhood: a whole-exome sequencing gene-identification study. <i>Lancet Neurology</i> , The, 2012, 11, 764-773.	4.9	223
273	RBPJ Mutations Identified in Two Families Affected by Adams-Oliver Syndrome. <i>American Journal of Human Genetics</i> , 2012, 91, 391-395.	2.6	106
274	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. <i>American Journal of Human Genetics</i> , 2012, 91, 541-547.	2.6	167
275	Loss of SUFU Function in Familial Multiple Meningioma. <i>American Journal of Human Genetics</i> , 2012, 91, 520-526.	2.6	137
276	Autosomal-Recessive Congenital Cerebellar Ataxia Is Caused by Mutations in Metabotropic Glutamate Receptor 1. <i>American Journal of Human Genetics</i> , 2012, 91, 553-564.	2.6	81
277	Loss-of-Function Mutations in HOXC13 Cause Pure Hair and Nail Ectodermal Dysplasia. <i>American Journal of Human Genetics</i> , 2012, 91, 906-911.	2.6	64
278	Autosomal mutations and human spermatogenic failure. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2012, 1822, 1873-1879.	1.8	11
279	A new mutational mechanism for hypertrophic cardiomyopathy. <i>Gene</i> , 2012, 507, 165-169.	1.0	10
280	Single nucleotide polymorphisms in genes that are common targets of luteotropin and luteolysin in primate corpus luteum: Computational exploration. <i>Gene</i> , 2012, 511, 353-357.	1.0	5
281	Brownâ€“Vialetoâ€“van Laere and Fazioâ€“Londe overlap syndromes: A clinical, biochemical and genetic study. <i>Neuromuscular Disorders</i> , 2012, 22, 1075-1082.	0.3	36
282	Current challenges in genome annotation through structural biology and bioinformatics. <i>Current Opinion in Structural Biology</i> , 2012, 22, 594-601.	2.6	14
283	Absence of de novo point mutations in exons of GRIN2B in a large schizophrenia trio sample. <i>Schizophrenia Research</i> , 2012, 141, 274-276.	1.1	5
284	Severe disseminated mycobacterial infection in a boy with a novel mutation leading to IFN-Î³R2 deficiency. <i>Journal of Infection</i> , 2012, 65, 568-572.	1.7	17
285	Computational screening and molecular dynamics simulation of disease associated nsSNPs in CENP-E. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2012, 738-739, 28-37.	0.4	62
286	Some Phenotype Association Tools in Galaxy: Looking for Disease SNPs in a Full Genome. <i>Current Protocols in Bioinformatics</i> , 2012, 39, Unit15.2.	25.8	2
287	Predicting the impact of deleterious single point mutations in SMAD gene family using structural bioinformatics approach. <i>Interdisciplinary Sciences, Computational Life Sciences</i> , 2012, 4, 103-115.	2.2	6
288	Mitochondrial DNA sequence variation is associated with free-living activity energy expenditure in the elderly. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2012, 1817, 1691-1700.	0.5	13
289	Distribution, Frequency and Variation of Stripe Rust Resistance Loci Yr10, Lr34/Yr18 and Yr36 in Chinese Wheat Cultivars. <i>Journal of Genetics and Genomics</i> , 2012, 39, 587-592.	1.7	24

#	ARTICLE	IF	CITATIONS
290	A soybean cyst nematode resistance gene points to a new mechanism of plant resistance to pathogens. <i>Nature</i> , 2012, 492, 256-260.	13.7	332
291	Structural Modelling Pipelines in Next Generation Sequencing Projects. <i>Advances in Protein Chemistry and Structural Biology</i> , 2012, 89, 117-167.	1.0	19
292	Validation of the use of multiple internal control genes, and the application of real-time quantitative PCR, to study esterase gene expression in <i>Oenococcus oeni</i> . <i>Applied Microbiology and Biotechnology</i> , 2012, 96, 1039-1047.	1.7	34
293	Novel variations in the adiponectin gene (ADIPOQ) may affect distribution of oligomeric complexes. <i>SpringerPlus</i> , 2012, 1, 66.	1.2	3
294	Computational analysis of deleterious missense mutations in aspartoacylase that cause Canavanâ€™s disease. <i>Science China Life Sciences</i> , 2012, 55, 1109-1119.	2.3	6
295	A transforming <i>KIF5B</i> and <i>RET</i> gene fusion in lung adenocarcinoma revealed from whole-genome and transcriptome sequencing. <i>Genome Research</i> , 2012, 22, 436-445.	2.4	433
296	Variants of the Lamin A/C (LMNA) Gene in Non-Valvular Atrial Fibrillation Patients. <i>Molecular Diagnosis and Therapy</i> , 2012, 16, 99-107.	1.6	24
297	Transcriptome profiling of UPF3B/NMD-deficient lymphoblastoid cells from patients with various forms of intellectual disability. <i>Molecular Psychiatry</i> , 2012, 17, 1103-1115.	4.1	97
299	Structural effect of P278A mutation conferring breast cancer susceptibility in the p53 DNA-binding core domain. <i>BMC Proceedings</i> , 2012, 6, .	1.8	0
300	Next-generation sequencing in health-care delivery: lessons from the functional analysis of rhodopsin. <i>Genetics in Medicine</i> , 2012, 14, 891-899.	1.1	28
301	A Missense Mutation in the Extracellular Domain of Fas: The Most Common Change in Argentinean Patients with Autoimmune Lymphoproliferative Syndrome Represents a Founder Effect. <i>Journal of Clinical Immunology</i> , 2012, 32, 1197-1203.	2.0	11
302	Improving the prediction of the functional impact of cancer mutations by baseline tolerance transformation. <i>Genome Medicine</i> , 2012, 4, 89.	3.6	91
303	Sequencing Analysis of SLX4/FANCP Gene in Italian Familial Breast Cancer Cases. <i>PLoS ONE</i> , 2012, 7, e31038.	1.1	10
304	Identification of Novel Mutations in HEXA Gene in Children Affected with Tay Sachs Disease from India. <i>PLoS ONE</i> , 2012, 7, e39122.	1.1	37
305	Prevalence and Clinical Features of Hearing Loss Patients with CDH23 Mutations: A Large Cohort Study. <i>PLoS ONE</i> , 2012, 7, e40366.	1.1	61
306	Rare Variants in Ischemic Stroke: An Exome Pilot Study. <i>PLoS ONE</i> , 2012, 7, e35591.	1.1	34
307	Genetic Background Analysis of Protein C Deficiency Demonstrates a Recurrent Mutation Associated with Venous Thrombosis in Chinese Population. <i>PLoS ONE</i> , 2012, 7, e35773.	1.1	42
308	Rare Primary Mitochondrial DNA Mutations and Probable Synergistic Variants in Leberâ€™s Hereditary Optic Neuropathy. <i>PLoS ONE</i> , 2012, 7, e42242.	1.1	73

#	ARTICLE	IF	CITATIONS
309	Investigating the Structural Impacts of I64T and P311S Mutations in APE1-DNA Complex: A Molecular Dynamics Approach. PLoS ONE, 2012, 7, e31677.	1.1	41
310	E-Cadherin Destabilization Accounts for the Pathogenicity of Missense Mutations in Hereditary Diffuse Gastric Cancer. PLoS ONE, 2012, 7, e33783.	1.1	53
311	Whole-Exome Sequencing Efficiently Detects Rare Mutations in Autosomal Recessive Nonsyndromic Hearing Loss. PLoS ONE, 2012, 7, e50628.	1.1	143
312	Exome Sequencing of Only Seven Qataris Identifies Potentially Deleterious Variants in the Qatari Population. PLoS ONE, 2012, 7, e47614.	1.1	16
313	Duplication of C7orf58, WNT16 and FAM3C in an Obese Female with a t(7;22)(q32.1;q11.2) Chromosomal Translocation and Clinical Features Resembling Coffin-Siris Syndrome. PLoS ONE, 2012, 7, e52353.	1.1	5
314	Mutations and Binding Sites of Human Transcription Factors. Frontiers in Genetics, 2012, 3, 100.	1.1	11
315	Clinical Implications of Human Population Differences in Genome-Wide Rates of Functional Genotypes. Frontiers in Genetics, 2012, 3, 211.	1.1	29
316	The Novel Human p.I587V Variant in the <i>ZNF644</i> Gene Is Unlikely to Be the Pathogenic Cause of Dominantly Inherited High Myopia in a Chinese Patient. , 2012, 53, 6728.		5
317	Anderson's Disease/Chylomicron Retention Disease and Mutations in the SAR1B Gene. , 2012, , .		1
318	Novel mutation of SRD5A2 gene in a patient with 5 α -reductase 2 deficiency from India. BMJ Case Reports, 2012, 2012, bcr2012007060-bcr2012007060.	0.2	6
320	Targeted capture and massively parallel sequencing in pediatric cardiomyopathy: development of novel diagnostics. Neurology International, 2012, 2, 7.	0.2	0
321	Computational Methods to Work as First-Pass Filter in Deleterious SNP Analysis of Alkaptonuria. Scientific World Journal, The, 2012, 2012, 1-9.	0.8	10
322	Early Onset Retinal Dystrophy Due to Mutations in <i>LRAT</i> : Molecular Analysis and Detailed Phenotypic Study. , 2012, 53, 3927.		38
323	A common variant of the MACC1 gene is significantly associated with overall survival in colorectal cancer patients. BMC Cancer, 2012, 12, 20.	1.1	27
324	Concise Review: Genomic Stability of Human Induced Pluripotent Stem Cells. Stem Cells, 2012, 30, 22-27.	1.4	113
325	Effectiveness of sequencing selected exons of <i>DNAH5</i> and <i>DNAI1</i> in diagnosis of primary ciliary dyskinesia. Pediatric Pulmonology, 2012, 47, 864-875.	1.0	32
326	Variations in the exome of the LNCaP prostate cancer cell line. Prostate, 2012, 72, 1317-1327.	1.2	19
327	Evolution and Functional Impact of Rare Coding Variation from Deep Sequencing of Human Exomes. Science, 2012, 337, 64-69.	6.0	1,535

#	ARTICLE	IF	CITATIONS
328	De novo mutations revealed by whole-exome sequencing are strongly associated with autism. <i>Nature</i> , 2012, 485, 237-241.	13.7	1,863
329	PON-P: Integrated predictor for pathogenicity of missense variants. <i>Human Mutation</i> , 2012, 33, 1166-1174.	1.1	88
330	A novel classification system to predict the pathogenic effects of CHD7 missense variants in CHARGE syndrome. <i>Human Mutation</i> , 2012, 33, 1251-1260.	1.1	65
331	Paralogous annotation of disease-causing variants in long QT syndrome genes. <i>Human Mutation</i> , 2012, 33, 1188-1191.	1.1	44
332	Exome sequencing reveals <i>DNAJB6</i> mutations in dominantly inherited myopathy. <i>Annals of Neurology</i> , 2012, 71, 407-416.	2.8	148
333	Investigation of SUMO pathway genes in the etiology of nonsyndromic cleft lip with or without cleft palate. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2012, 94, 459-463.	1.6	5
334	DNA Sequencing. <i>Circulation</i> , 2012, 125, 931-944.	1.6	72
335	wANNOVAR: annotating genetic variants for personal genomes via the web. <i>Journal of Medical Genetics</i> , 2012, 49, 433-436.	1.5	366
336	The juxtapanodal proteins CNTNAP2 and TAG1 regulate diet-induced obesity. <i>Mammalian Genome</i> , 2012, 23, 431-442.	1.0	33
337	Mutations in <i>EZH2</i> Cause Weaver Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 110-118.	2.6	253
338	A Restricted Spectrum of Mutations in the <i>SMAD4</i> Tumor-Suppressor Gene Underlies Myhre Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 161-169.	2.6	77
339	Exome Sequencing Reveals Mutations in <i>TRPV3</i> as a Cause of Olmsted Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 558-564.	2.6	300
340	Mutations in <i>C5ORF42</i> Cause Joubert Syndrome in the French Canadian Population. <i>American Journal of Human Genetics</i> , 2012, 90, 693-700.	2.6	118
341	<i>PSORS2</i> Is Due to Mutations in <i>CARD14</i> . <i>American Journal of Human Genetics</i> , 2012, 90, 784-795.	2.6	365
342	Computational and molecular approaches for predicting unreported causal missense mutations in Belgian patients with haemophilia A. <i>Haemophilia</i> , 2012, 18, e331-9.	1.0	11
343	Matrix metalloproteinase genes on chromosome 11q22 and the risk of anterior cruciate ligament (ACL) rupture. <i>Scandinavian Journal of Medicine and Science in Sports</i> , 2012, 22, 523-533.	1.3	71
344	Comprehensive analysis of <i>LAMC1</i> genetic variants in advanced pelvic organ prolapse. <i>American Journal of Obstetrics and Gynecology</i> , 2012, 206, 447.e1-447.e6.	0.7	16
345	Systematic Immunohistochemistry Screening for Lynch Syndrome in Early Age-of-Onset Colorectal Cancer Patients Undergoing Surgical Resection. <i>Journal of the American College of Surgeons</i> , 2012, 214, 61-67.	0.2	32

#	ARTICLE	IF	CITATIONS
346	Two novel missense mutations in <i>FGD4/FRABIN</i> cause Charcot-Marie-Tooth type 4H (CMT4H). <i>Journal of the Peripheral Nervous System</i> , 2012, 17, 141-146.	1.4	18
347	Confirmation of an epilepsy modifier locus on mouse chromosome 11 and candidate gene analysis by RNA-seq. <i>Genes, Brain and Behavior</i> , 2012, 11, 452-460.	1.1	25
348	A phenotypic study of congenital stationary night blindness (CSNB) associated with mutations in the <i>GRM6</i> gene. <i>Acta Ophthalmologica</i> , 2012, 90, e192-7.	0.6	32
349	Identification of novel genetic variants in phosphodiesterase 8B (<i>PDE8B</i>), a cAMP-specific phosphodiesterase highly expressed in the adrenal cortex, in a cohort of patients with adrenal tumours. <i>Clinical Endocrinology</i> , 2012, 77, 195-199.	1.2	72
350	Evidence for selection at cytokine loci in a natural population of field voles (<i>Microtus</i>). <i>Trends in Ecology and Evolution</i> , 2012, 27, 582-589.	2.0	44
351	BAG3-related myofibrillar myopathy in a Chinese family. <i>Clinical Genetics</i> , 2012, 81, 394-398.	1.0	61
352	Second-generation sequencing for gene discovery in the Brassicaceae. <i>Plant Biotechnology Journal</i> , 2012, 10, 750-759.	4.1	18
353	Analysis of SLX4/FANCP in non-BRCA1/2-mutated breast cancer families. <i>BMC Cancer</i> , 2012, 12, 84.	1.1	14
354	Genetic variation in <i>APOB</i> , <i>PCSK9</i> , and <i>ANGPTL3</i> in carriers of pathogenic autosomal dominant hypercholesterolemic mutations with unexpected low LDL-C levels. <i>Human Mutation</i> , 2012, 33, 448-455.	1.1	36
355	Rare germline mutations in <i>PALB2</i> and breast cancer risk: A population-based study. <i>Human Mutation</i> , 2012, 33, 674-680.	1.1	74
356	Polymorphism in the protease-activated receptor-4 gene region associates with platelet activation and perioperative myocardial injury. <i>American Journal of Hematology</i> , 2012, 87, 161-166.	2.0	14
357	Functional analysis of variant lysosomal acid glycosidases of Anderson-Fabry and Pompe disease in a human embryonic kidney epithelial cell line (HEK 293 T). <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 325-334.	1.7	6
358	Analysis of new lactotransferrin gene variants in a case-control study related to periodontal disease in dog. <i>Molecular Biology Reports</i> , 2012, 39, 4673-4681.	1.0	6
359	Situs Inversus Totalis and a Novel <i>ZIC3</i> Mutation in a Family with X-linked Heterotaxy. <i>Congenital Heart Disease</i> , 2013, 8, E36-E40.	0.0	26
360	Intellectual disability associated with a homozygous missense mutation in <i>THOC6</i> . <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 62.	1.2	48
361	Autosomal recessive spastic ataxia of Charlevoix Saguenay (ARSACS): expanding the genetic, clinical and imaging spectrum. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 41.	1.2	147
362	Prevalence of <i>PALB2</i> mutations in Australasian multiple-case breast cancer families. <i>Breast Cancer Research</i> , 2013, 15, R17.	2.2	42
363	Integrated analysis of recurrent properties of cancer genes to identify novel drivers. <i>Genome Biology</i> , 2013, 14, R52.	13.9	33

#	ARTICLE	IF	CITATIONS
364	Genetics is a major determinant of expression of the human hepatic uptake transporter OATP1B1, but not of OATP1B3 and OATP2B1. <i>Genome Medicine</i> , 2013, 5, 1.	3.6	198
365	Germ-line DICER1 mutations do not make a major contribution to the etiology of familial testicular germ cell tumours. <i>BMC Research Notes</i> , 2013, 6, 127.	0.6	13
366	Impaired information-processing speed and working memory in leukoencephalopathy with brainstem and spinal cord involvement and elevated lactate (LBSL) and DARS2 mutations: a report of three adult patients. <i>Journal of Neurology</i> , 2013, 260, 2078-2083.	1.8	16
367	Whole-genome sequencing in an autism multiplex family. <i>Molecular Autism</i> , 2013, 4, 8.	2.6	76
368	Characterization of functional variants in 33 blood pressure loci using 1000 genomes project data. <i>Genes and Genomics</i> , 2013, 35, 387-393.	0.5	3
369	Genetic Association Study of Dickkopf-1 and Sclerostin Genes with Paget Disease of Bone. <i>Calcified Tissue International</i> , 2013, 93, 405-412.	1.5	10
370	Complete genome sequencing and variant analysis of a Pakistani individual. <i>Journal of Human Genetics</i> , 2013, 58, 622-626.	1.1	13
371	Structural and functional analysis of perforin mutations in association with clinical data of familial hemophagocytic lymphohistiocytosis type 2 (FHL2) patients. <i>Protein Science</i> , 2013, 22, 823-839.	3.1	28
372	IL-12R β 1 Deficiency: Mutation Update and Description of the <i>IL12RB1</i> Variation Database. <i>Human Mutation</i> , 2013, 34, 1329-1339.	1.1	81
373	Genetic programs in human and mouse early embryos revealed by single-cell RNA-sequencing. <i>Nature</i> , 2013, 500, 593-597.	13.7	859
374	Validating therapeutic targets through human genetics. <i>Nature Reviews Drug Discovery</i> , 2013, 12, 581-594.	21.5	548
375	Statistical Challenges in Sequence-Based Association Studies with Population- and Family-Based Designs. <i>Statistics in Biosciences</i> , 2013, 5, 54-70.	0.6	11
376	Next generation sequencing in cancer research and clinical application. <i>Biological Procedures Online</i> , 2013, 15, 4.	1.4	102
377	WEP: a high-performance analysis pipeline for whole-exome data. <i>BMC Bioinformatics</i> , 2013, 14, S11.	1.2	43
378	Candidate gene association studies: a comprehensive guide to useful in silico tools. <i>BMC Genetics</i> , 2013, 14, 39.	2.7	115
379	Identifying Mendelian disease genes with the Variant Effect Scoring Tool. <i>BMC Genomics</i> , 2013, 14, S3.	1.2	360
380	Assessment of computational methods for predicting the effects of missense mutations in human cancers. <i>BMC Genomics</i> , 2013, 14, S7.	1.2	153
381	ATR-FTIR spectroscopy reveals genomic loci regulating the tissue response in high fat diet fed BXD recombinant inbred mouse strains. <i>BMC Genomics</i> , 2013, 14, 386.	1.2	47

#	ARTICLE	IF	CITATIONS
382	Prognostic impact and landscape of NOTCH1 mutations in chronic lymphocytic leukemia (CLL): a study on 852 patients. <i>Leukemia</i> , 2013, 27, 2393-2396.	3.3	65
383	Investigation of variants within the <i>COL27A1</i> and <i>TNC</i> genes and Achilles tendinopathy in two populations. <i>Journal of Orthopaedic Research</i> , 2013, 31, 632-637.	1.2	44
384	Computational approaches to identify functional genetic variants in cancer genomes. <i>Nature Methods</i> , 2013, 10, 723-729.	9.0	161
385	Exploration of deleterious single nucleotide polymorphisms in the components of human P bodies: An in silico approach. <i>Gene</i> , 2013, 528, 360-363.	1.0	4
386	dbNSFP v2.0: A Database of Human Non-synonymous SNVs and Their Functional Predictions and Annotations. <i>Human Mutation</i> , 2013, 34, E2393-E2402.	1.1	546
387	Detecting Rare Variants for Psychiatric Disorders Using Next Generation Sequencing: A Methods Primer. <i>Current Psychiatry Reports</i> , 2013, 15, 333.	2.1	1
388	Evidence of Colorectal Cancer-Associated Mutation in MCAK: A Computational Report. <i>Cell Biochemistry and Biophysics</i> , 2013, 67, 837-851.	0.9	37
389	Pure adult-onset Spastic Paraplegia caused by a novel mutation in the KIAA0196 (SPG8) gene. <i>Journal of Neurology</i> , 2013, 260, 1765-1769.	1.8	37
390	ATL1 and REEP1 mutations in hereditary and sporadic upper motor neuron syndromes. <i>Journal of Neurology</i> , 2013, 260, 869-875.	1.8	17
391	An Italian Cohort Study Identifies Four New Pathologic Mutations in the ARSA Gene. <i>Journal of Molecular Neuroscience</i> , 2013, 50, 284-290.	1.1	8
392	Spatial and Temporal Mapping of De Novo Mutations in Schizophrenia to a Fetal Prefrontal Cortical Network. <i>Cell</i> , 2013, 154, 518-529.	13.5	507
393	Hunting human disease genes: lessons from the past, challenges for the future. <i>Human Genetics</i> , 2013, 132, 603-617.	1.8	31
394	Variants in GATA4 are a rare cause of familial and sporadic congenital diaphragmatic hernia. <i>Human Genetics</i> , 2013, 132, 285-292.	1.8	81
395	Targeted Next-Generation Sequencing Panel (ThyroSeq) for Detection of Mutations in Thyroid Cancer. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013, 98, E1852-E1860.	1.8	412
396	A Gene-Specific Method for Predicting Hemophilia-Causing Point Mutations. <i>Journal of Molecular Biology</i> , 2013, 425, 4023-4033.	2.0	30
397	Recurrent mutations in multiple components of the cohesin complex in myeloid neoplasms. <i>Nature Genetics</i> , 2013, 45, 1232-1237.	9.4	334
398	Novel CYP2B6 Enzyme Variants in a Rwandese Population: Functional Characterization and Assessment of In Silico Prediction Tools. <i>Human Mutation</i> , 2013, 34, 725-734.	1.1	28
399	Variations of the angiotensin II type 1 receptor gene are associated with extreme human longevity. <i>Age</i> , 2013, 35, 993-1005.	3.0	40

#	ARTICLE	IF	CITATIONS
400	Extrapolating the effect of deleterious nsSNPs in the binding adaptability of flavopiridol with CDK7 protein: a molecular dynamics approach. <i>Human Genomics</i> , 2013, 7, 10.	1.4	47
401	DNA Variations in Oculocutaneous Albinism: An Updated Mutation List and Current Outstanding Issues in Molecular Diagnostics. <i>Human Mutation</i> , 2013, 34, 827-835.	1.1	114
402	In silico discrimination of nsSNPs in hTERT gene by means of local DNA sequence context and regularity. <i>Journal of Molecular Modeling</i> , 2013, 19, 3517-3527.	0.8	7
403	Clonal evolution of high-grade serous ovarian carcinoma from primary to recurrent disease. <i>Journal of Pathology</i> , 2013, 229, 515-524.	2.1	88
404	Short-Rib Polydactyly and Jeune Syndromes Are Caused by Mutations in WDR60. <i>American Journal of Human Genetics</i> , 2013, 93, 515-523.	2.6	116
405	Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. <i>Nature Genetics</i> , 2013, 45, 639-647.	9.4	399
406	Cole Disease Results from Mutations in ENPP1. <i>American Journal of Human Genetics</i> , 2013, 93, 752-757.	2.6	41
407	Trimethylaminuria (fish odor syndrome): Genotype characterization among Portuguese patients. <i>Gene</i> , 2013, 527, 366-370.	1.0	16
408	Whole-exome sequencing identifies tetratricopeptide repeat domain 7A (TTC7A) mutations for combined immunodeficiency with intestinal atresias. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 132, 656-664.e17.	1.5	140
409	Long-term strain improvements accumulate mutations in regulatory elements responsible for hyper-production of cellulolytic enzymes. <i>Scientific Reports</i> , 2013, 3, 1569.	1.6	104
410	Mutations in LYRM4, encoding iron-sulfur cluster biogenesis factor ISD11, cause deficiency of multiple respiratory chain complexes. <i>Human Molecular Genetics</i> , 2013, 22, 4460-4473.	1.4	97
411	The <i>PALB2</i> Gene Is a Strong Candidate for Clinical Testing in <i>BRCA1</i> - and <i>BRCA2</i> -Negative Hereditary Breast Cancer. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2013, 22, 2323-2332.	1.1	42
412	Exome sequencing identifies secondary mutations of SETBP1 and JAK3 in juvenile myelomonocytic leukemia. <i>Nature Genetics</i> , 2013, 45, 937-941.	9.4	203
413	Neuronal ceroid lipofuscinosis type CLN2: A new rationale for the construction of phenotypic subgroups based on a survey of 25 cases in South America. <i>Gene</i> , 2013, 516, 114-121.	1.0	40
414	Database tools in genetic diseases research. <i>Genomics</i> , 2013, 101, 75-85.	1.3	18
415	Screening of MYH7, MYBPC3, and TNNT2 genes in Brazilian patients with hypertrophic cardiomyopathy. <i>American Heart Journal</i> , 2013, 166, 775-782.	1.2	39
416	A De Novo Mutation in the β -Tubulin Gene TUBB4A Results in the Leukoencephalopathy Hypomyelination with Atrophy of the Basal Ganglia and Cerebellum. <i>American Journal of Human Genetics</i> , 2013, 92, 767-773.	2.6	174
417	Assessing the phenotypic effects in the general population of rare variants in genes for a dominant Mendelian form of diabetes. <i>Nature Genetics</i> , 2013, 45, 1380-1385.	9.4	129

#	ARTICLE	IF	CITATIONS
418	Learning Natural Selection from the Site Frequency Spectrum. <i>Genetics</i> , 2013, 195, 181-193.	1.2	105
419	High-resolution network biology: connecting sequence with function. <i>Nature Reviews Genetics</i> , 2013, 14, 865-879.	7.7	92
420	Therapeutic Potential of the Poly(ADP-ribose) Polymerase Inhibitor Rucaparib for the Treatment of Sporadic Human Ovarian Cancer. <i>Molecular Cancer Therapeutics</i> , 2013, 12, 1002-1015.	1.9	93
421	Clinical Analysis and Interpretation of Cancer Genome Data. <i>Journal of Clinical Oncology</i> , 2013, 31, 1825-1833.	0.8	123
422	Clinical application of amplicon-based next-generation sequencing in cancer. <i>Cancer Genetics</i> , 2013, 206, 413-419.	0.2	98
423	Recurrent inactivation of STAG2 in bladder cancer is not associated with aneuploidy. <i>Nature Genetics</i> , 2013, 45, 1464-1469.	9.4	224
424	Novel and recurrent mutations in the TAT gene in Tunisian families affected with Richner-Hanhart Syndrome. <i>Gene</i> , 2013, 529, 45-49.	1.0	10
425	Whole-exome sequencing links caspase recruitment domain 11 (CARD11) inactivation to severe combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1376-1383.e3.	1.5	127
426	Exonic Transcription Factor Binding Directs Codon Choice and Affects Protein Evolution. <i>Science</i> , 2013, 342, 1367-1372.	6.0	267
427	Next Generation Sequencing. , 2013, , .		10
428	Novel and known nephrin gene (NPHS1) mutations in two Greek cases with congenital nephrotic syndrome including a complex genotype. <i>Journal of Genetics</i> , 2013, 92, 577-581.	0.4	1
430	Can Unknown Predisposition in Familial Breast Cancer be Family-Specific?. <i>Breast Journal</i> , 2013, 19, n/a-n/a.	0.4	26
431	Genetic mapping and exome sequencing identify 2 mutations associated with stroke protection in pediatric patients with sickle cell anemia. <i>Blood</i> , 2013, 121, 3237-3245.	0.6	59
432	Western Database of Lipid Variants (WDLV): A Catalogue of Genetic Variants in Monogenic Dyslipidemias. <i>Canadian Journal of Cardiology</i> , 2013, 29, 934-939.	0.8	22
433	A practical guide for the functional annotation of genetic variations using SNPnexus. <i>Briefings in Bioinformatics</i> , 2013, 14, 437-447.	3.2	90
434	Germline mutation in the RAD51B gene confers predisposition to breast cancer. <i>BMC Cancer</i> , 2013, 13, 484.	1.1	64
435	Whole transcriptome sequencing identifies tumor-specific mutations in human oral squamous cell carcinoma. <i>BMC Medical Genomics</i> , 2013, 6, 28.	0.7	26
436	A new mutation in the gene encoding mitochondrial seryl-tRNA synthetase as a cause of HUPRA syndrome. <i>BMC Nephrology</i> , 2013, 14, 195.	0.8	31

#	ARTICLE	IF	CITATIONS
437	Whole-genome resequencing of Hanwoo (Korean cattle) and insight into regions of homozygosity. BMC Genomics, 2013, 14, 519.	1.2	55
438	An association-adjusted consensus deleterious scheme to classify homozygous Mis-sense mutations for personal genome interpretation. BioData Mining, 2013, 6, 24.	2.2	1
439	Identifying novel oncogenes: A machine learning approach. Interdisciplinary Sciences, Computational Life Sciences, 2013, 5, 241-246.	2.2	15
440	Galaxy tools to study genome diversity. GigaScience, 2013, 2, 17.	3.3	19
441	Structure-Function Studies on Non-synonymous SNPs of Chemokine Receptor Gene Implicated in Cardiovascular Disease: A Computational Approach. Protein Journal, 2013, 32, 657-665.	0.7	6
442	Abnormal centrosome and spindle morphology in a patient with autosomal recessive primary microcephaly type 2 due to compound heterozygous WDR62 gene mutation. Orphanet Journal of Rare Diseases, 2013, 8, 178.	1.2	38
443	Profiling Deleterious Non-synonymous SNPs of Smoker's Gene CYP1A1. Cell Biochemistry and Biophysics, 2013, 67, 1391-1396.	0.9	5
444	Genetics at the verge of extinction: insights from the Iberian lynx. Molecular Ecology, 2013, 22, 5503-5515.	2.0	48
445	MLL2 mutation detection in 86 patients with Kabuki syndrome: a genotype-phenotype study. Clinical Genetics, 2013, 84, 539-545.	1.0	85
446	ABCC11/MRP8 polymorphisms affect 5-fluorouracil-induced severe toxicity and hepatic expression. Pharmacogenomics, 2013, 14, 1433-1448.	0.6	21
447	Role of the sodium channel SCN9A in genetic epilepsy with febrile seizures plus and Dravet syndrome. Epilepsia, 2013, 54, e122-6.	2.6	62
448	Economic and environmental impacts of microbial biodiesel. Nature Biotechnology, 2013, 31, 789-793.	9.4	77
449	Systematic investigation of cancer-associated somatic point mutations in SNP databases. Nature Biotechnology, 2013, 31, 787-789.	9.4	21
450	Homozygosity mapping identifies a novel GIPC3 mutation causing congenital nonsyndromic hearing loss in a Saudi family. Gene, 2013, 521, 195-199.	1.0	18
451	Genomics in Cardiovascular Disease. Journal of the American College of Cardiology, 2013, 61, 2029-2037.	1.2	37
452	Population Genomics of Human Adaptation. Annual Review of Ecology, Evolution, and Systematics, 2013, 44, 123-143.	3.8	81
453	Genetic testing in benign familial epilepsies of the first year of life: Clinical and diagnostic significance. Epilepsia, 2013, 54, 425-436.	2.6	110
454	Mutation mapping of apolipoprotein A-I structure assisted with the putative cholesterol recognition regions. Biochimica Et Biophysica Acta - Proteins and Proteomics, 2013, 1834, 2030-2035.	1.1	6

#	ARTICLE	IF	CITATIONS
455	Detection of Clinically Relevant Genetic Variants in Autism Spectrum Disorder by Whole-Genome Sequencing. <i>American Journal of Human Genetics</i> , 2013, 93, 249-263.	2.6	429
456	Small intragenic deletion in <i>FOXP2</i> associated with childhood apraxia of speech and dysarthria. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2321-2326.	0.7	75
457	Epigenetic and genetic features of 24 colon cancer cell lines. <i>Oncogenesis</i> , 2013, 2, e71-e71.	2.1	658
458	The MyD88 rs6853 and TIRAP rs8177374 polymorphic sites are associated with resistance to human pulmonary tuberculosis. <i>Genes and Immunity</i> , 2013, 14, 504-511.	2.2	24
459	In-silico screening of cancer associated mutation on PLK1 protein and its structural consequences. <i>Journal of Molecular Modeling</i> , 2013, 19, 5587-5599.	0.8	31
460	Molecular basis of protein S deficiency in China. <i>American Journal of Hematology</i> , 2013, 88, 899-905.	2.0	32
461	Mutated Ephrin Receptor Genes in Non-Small Cell Lung Carcinoma and Their Occurrence with Driver Mutations—Targeted Resequencing Study on Formalin-Fixed, Paraffin-Embedded Tumor Material of 81 Patients. <i>Genes Chromosomes and Cancer</i> , 2013, 52, 1141-1149.	1.5	14
462	Progranulin Peripheral Levels as a Screening Tool for the Identification of Subjects with Progranulin Mutations in a Portuguese Cohort. <i>Neurodegenerative Diseases</i> , 2014, 13, 214-223.	0.8	28
463	Characterization of genetic lesions in rhabdomyosarcoma using a high-density single nucleotide polymorphism array. <i>Cancer Science</i> , 2013, 104, 856-864.	1.7	33
464	Analysis and Annotation of Whole-Genome or Whole-Exome Sequencing-Derived Variants for Clinical Diagnosis. <i>Current Protocols in Human Genetics</i> , 2013, 79, Unit 9.24..	3.5	17
465	No association between COL3A1, COL6A1 or COL12A1 gene variants and range of motion. <i>Journal of Sports Sciences</i> , 2013, 31, 181-187.	1.0	10
466	<i>FGFR1</i> mutations cause Hartsfield syndrome, the unique association of holoprosencephaly and ectrodactyly. <i>Journal of Medical Genetics</i> , 2013, 50, 585-592.	1.5	75
467	Clinical Genome Sequencing. , 2013, , 102-122.		29
468	Recurrent SETBP1 mutations in atypical chronic myeloid leukemia. <i>Nature Genetics</i> , 2013, 45, 18-24.	9.4	359
469	A sensitive and specific diagnostic test for hearing loss using a microdroplet PCR-based approach and next generation sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 145-152.	0.7	61
470	Genotype and Phenotype Correlations in 417 Children With Congenital Hyperinsulinism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013, 98, E355-E363.	1.8	252
471	A clinical evaluation tool for SNP arrays, especially for autosomal recessive conditions in offspring of consanguineous parents. <i>Genetics in Medicine</i> , 2013, 15, 354-360.	1.1	52
472	The effect of granulocyte colony stimulating factor receptor gene missense single nucleotide polymorphisms on peripheral blood stem cell enrichment. <i>Cytokine</i> , 2013, 61, 572-577.	1.4	4

#	ARTICLE	IF	CITATIONS
473	Single nucleotide polymorphisms in Mycobacterium tuberculosis and the need for a curated database. Tuberculosis, 2013, 93, 30-39.	0.8	43
474	Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. Nature, 2013, 493, 216-220.	13.7	898
475	Identification of germline mutations in the cancer predisposing gene CDH1 in patients with orofacial clefts. Human Molecular Genetics, 2013, 22, 919-926.	1.4	55
476	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. Human Mutation, 2013, 34, 255-265.	1.1	80
477	Mutations in <i>SYNGAP1</i> Cause Intellectual Disability, Autism, and a Specific Form of Epilepsy by Inducing Haploinsufficiency. Human Mutation, 2013, 34, 385-394.	1.1	196
478	Novel mutation predicted to disrupt SGOL1 protein function. Egyptian Journal of Medical Human Genetics, 2013, 14, 149-155.	0.5	0
479	Mutations in PDGFRB Cause Autosomal-Dominant Infantile Myofibromatosis. American Journal of Human Genetics, 2013, 92, 1001-1007.	2.6	174
480	Severe, fatal multisystem manifestations in a patient with dolichol kinase-congenital disorder of glycosylation. Molecular Genetics and Metabolism, 2013, 110, 484-489.	0.5	18
481	The genetic spectrum of familial hypercholesterolemia in Pakistan. Clinica Chimica Acta, 2013, 421, 219-225.	0.5	12
482	Current relaxation of selection on the human genome: Tolerance of deleterious mutations on olfactory receptors. Molecular Phylogenetics and Evolution, 2013, 66, 558-564.	1.2	27
483	Genome-wide sequencing to identify the cause of hereditary cancer syndromes: With examples from familial pancreatic cancer. Cancer Letters, 2013, 340, 227-233.	3.2	19
484	News from the Protein Mutability Landscape. Journal of Molecular Biology, 2013, 425, 3937-3948.	2.0	72
485	Distinct phenotypes in zebrafish models of human startle disease. Neurobiology of Disease, 2013, 60, 139-151.	2.1	32
486	Pontocerebellar hypoplasia type 2 and TSEN2: Review of the literature and two novel mutations. European Journal of Medical Genetics, 2013, 56, 325-330.	0.7	32
487	In silico prediction of the effects of mutations in the human UDP-galactose 4-epimerase gene: Towards a predictive framework for type III galactosemia. Gene, 2013, 524, 95-104.	1.0	23
488	Determination of allelic expression of SNP rs1880676 in choline acetyltransferase gene in HeLa cells. Neuroscience Letters, 2013, 555, 215-219.	1.0	1
489	A novel mutation in the SLCO2A1 gene in a Chinese family with primary hypertrophic osteoarthropathy. Gene, 2013, 521, 191-194.	1.0	23
490	Molecular diagnosis of congenital muscular dystrophies with defective glycosylation of alpha-dystroglycan using next-generation sequencing technology. Neuromuscular Disorders, 2013, 23, 337-344.	0.3	13

#	ARTICLE	IF	CITATIONS
491	Novel missense mutations in the glycine receptor $\hat{1}^2$ subunit gene (GLRB) in startle disease. <i>Neurobiology of Disease</i> , 2013, 52, 137-149.	2.1	54
492	Congruency in the prediction of pathogenic missense mutations: state-of-the-art web-based tools. <i>Briefings in Bioinformatics</i> , 2013, 14, 448-459.	3.2	79
493	SIRP $\hat{1}\pm$ polymorphisms, but not the prion protein, control phagocytosis of apoptotic cells. <i>Journal of Experimental Medicine</i> , 2013, 210, 2539-2552.	4.2	67
494	Roadmap to determine the point mutations involved in cardiomyopathy disorder: A Bayesian approach. <i>Gene</i> , 2013, 519, 34-40.	1.0	16
495	Use of comparative genomics approaches to characterize interspecies differences in response to environmental chemicals: Challenges, opportunities, and research needs. <i>Toxicology and Applied Pharmacology</i> , 2013, 271, 372-385.	1.3	29
496	The novel heterozygous Thr377Arg MYOC mutation causes severe Juvenile Open Angle Glaucoma in a large Pakistani family. <i>Gene</i> , 2013, 528, 356-359.	1.0	19
497	G6PC3 mutations cause non-syndromic severe congenital neutropenia. <i>Molecular Genetics and Metabolism</i> , 2013, 108, 138-141.	0.5	16
498	Novel excitation-contraction uncoupled RYR1 mutations in patients with central core disease. <i>Neuromuscular Disorders</i> , 2013, 23, 120-132.	0.3	24
499	An exon 53 frameshift mutation in CUBN abrogates cubam function and causes Imlerslund-GrÅsbeck syndrome in dogs. <i>Molecular Genetics and Metabolism</i> , 2013, 109, 390-396.	0.5	22
500	High resolution melting analysis of the MMAB gene in cblB patients and in those with undiagnosed methylmalonic aciduria. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 86-89.	0.5	7
501	MECP2 Gene Study in a Large Cohort. <i>Journal of Molecular Diagnostics</i> , 2013, 15, 723-729.	1.2	4
502	KIT, NRAS, BRAF and PTEN mutations in a sample of Swedish patients with acral lentiginous melanoma. <i>Journal of Dermatological Science</i> , 2013, 72, 284-289.	1.0	86
503	Hereditary cataract of the Nakano mouse: Involvement of a hypomorphic mutation in the coproporphyrinogen oxidase gene. <i>Experimental Eye Research</i> , 2013, 112, 45-50.	1.2	13
504	PTCH1 gene polymorphisms in ovarian tumors: Potential protective role of c.3944T allele. <i>Gene</i> , 2013, 517, 55-59.	1.0	8
505	Mitochondrial NADH:ubiquinone oxidoreductase alterations are associated with endometriosis. <i>Mitochondrion</i> , 2013, 13, 782-790.	1.6	14
506	Three novel germ-line VHL mutations in Hungarian von Hippel-Lindau patients, including a nonsense mutation in a fifteen-year-old boy with renal cell carcinoma. <i>BMC Medical Genetics</i> , 2013, 14, 3.	2.1	5
507	Mutation detection in Croatian patients with Familial Hypercholesterolemia. <i>Annals of Human Genetics</i> , 2013, 77, 22-30.	0.3	12
508	Point mutations as a source of de novo genetic disease. <i>Current Opinion in Genetics and Development</i> , 2013, 23, 257-263.	1.5	44

#	ARTICLE	IF	CITATIONS
509	Null CYP1B1 Genotypes in Primary Congenital and Nondominant Juvenile Glaucoma. <i>Ophthalmology</i> , 2013, 120, 716-723.	2.5	41
510	Mutations in <i>HNF1A</i> Result in Marked Alterations of Plasma Glycan Profile. <i>Diabetes</i> , 2013, 62, 1329-1337.	0.3	97
511	Targeted massively parallel sequencing provides comprehensive genetic diagnosis for patients with disorders of sex development. <i>Clinical Genetics</i> , 2013, 83, 35-43.	1.0	72
512	Whole exome sequencing identifies a novel mutation in the transglutaminase 6 gene for spinocerebellar ataxia in a Chinese family. <i>Clinical Genetics</i> , 2013, 83, 269-273.	1.0	43
513	Identification of seven loci affecting mean telomere length and their association with disease. <i>Nature Genetics</i> , 2013, 45, 422-427.	9.4	808
514	Germline mutations of regulator of telomere elongation helicase 1, <i>RTEL1</i> , in Dyskeratosis congenita. <i>Human Genetics</i> , 2013, 132, 473-480.	1.8	198
515	Relapse-specific mutations in <i>NT5C2</i> in childhood acute lymphoblastic leukemia. <i>Nature Genetics</i> , 2013, 45, 290-294.	9.4	264
516	Cystic fibrosis testing in a referral laboratory: results and lessons from a six-year period. <i>Journal of Clinical Bioinformatics</i> , 2013, 3, 3.	1.2	2
517	Rare Pathogenic Variants in <i>IL36RN</i> Underlie a Spectrum of Psoriasis-Associated Pustular Phenotypes. <i>Journal of Investigative Dermatology</i> , 2013, 133, 1366-1369.	0.3	140
518	Next-generation sequencing-based risk stratification and identification of new genes involved in structural and sequence variations in near haploid lymphoblastic leukemia. <i>Genes Chromosomes and Cancer</i> , 2013, 52, 564-579.	1.5	20
519	Computational solutions for omics data. <i>Nature Reviews Genetics</i> , 2013, 14, 333-346.	7.7	288
520	Detection of novel genetic variation in autosomal dominant retinitis pigmentosa. <i>Clinical Genetics</i> , 2013, 84, 441-452.	1.0	9
521	Combining Highly Multiplexed PCR with Semiconductor-Based Sequencing for Rapid Cancer Genotyping. <i>Journal of Molecular Diagnostics</i> , 2013, 15, 171-176.	1.2	140
522	Rare, Low-Frequency, and Common Variants in the Protein-Coding Sequence of Biological Candidate Genes from GWASs Contribute to Risk of Rheumatoid Arthritis. <i>American Journal of Human Genetics</i> , 2013, 92, 15-27.	2.6	83
523	Exome sequencing reveals <i>CCDC111</i> mutation associated with high myopia. <i>Human Genetics</i> , 2013, 132, 913-921.	1.8	74
524	Misfolding of galactose 1-phosphate uridylyltransferase can result in type I galactosemia. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2013, 1832, 1279-1293.	1.8	44
525	Molecular characterization of 355 mucopolysaccharidosis patients reveals 104 novel mutations. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 179-187.	1.7	48
526	Management of Incidental Findings in Clinical Genomic Sequencing. <i>Current Protocols in Human Genetics</i> , 2013, 77, Unit9.23.	3.5	19

#	ARTICLE	IF	CITATIONS
527	Mutations in FGF17, IL17RD, DUSP6, SPRY4, and FLRT3 Are Identified in Individuals with Congenital Hypogonadotropic Hypogonadism. <i>American Journal of Human Genetics</i> , 2013, 92, 725-743.	2.6	227
528	General Framework for Meta-analysis of Rare Variants in Sequencing Association Studies. <i>American Journal of Human Genetics</i> , 2013, 93, 42-53.	2.6	211
530	Identification of multiple risk variants for ankylosing spondylitis through high-density genotyping of immune-related loci. <i>Nature Genetics</i> , 2013, 45, 730-738.	9.4	699
531	Autosomal Dominant Spastic Paraplegias. <i>JAMA Neurology</i> , 2013, 70, 481.	4.5	48
532	Brief Report: High-Throughput Sequencing of <i>IL23R</i> Reveals a Low-Frequency, Nonsynonymous Single-Nucleotide Polymorphism That Is Associated With Ankylosing Spondylitis in a Han Chinese Population. <i>Arthritis and Rheumatism</i> , 2013, 65, 1747-1752.	6.7	28
533	Whole-exome sequencing and imaging genetics identify functional variants for rate of change in hippocampal volume in mild cognitive impairment. <i>Molecular Psychiatry</i> , 2013, 18, 781-787.	4.1	81
534	Molecular genetic testing and the future of clinical genomics. <i>Nature Reviews Genetics</i> , 2013, 14, 415-426.	7.7	334
535	The spectrum of HNF1A gene mutations in Greek patients with MODY3: relative frequency and identification of seven novel germline mutations. <i>Pediatric Diabetes</i> , 2013, 14, 526-534.	1.2	12
536	High-Throughput RNA Sequencing in B-Cell Lymphomas. <i>Methods in Molecular Biology</i> , 2013, 971, 295-312.	0.4	5
537	Predicting the Impact of Single-Nucleotide Polymorphisms in CDK2-Flavopiridol Complex by Molecular Dynamics Analysis. <i>Cell Biochemistry and Biophysics</i> , 2013, 66, 681-695.	0.9	14
538	Combined linkage analysis and exome sequencing identifies novel genes for familial goiter. <i>Journal of Human Genetics</i> , 2013, 58, 366-377.	1.1	19
539	<i>SRGAP1</i> Is a Candidate Gene for Papillary Thyroid Carcinoma Susceptibility. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013, 98, E973-E980.	1.8	74
540	Discovery of novel non-synonymous SNP variants in 988 candidate genes from 6 centenarians by target capture and next-generation sequencing. <i>Mechanisms of Ageing and Development</i> , 2013, 134, 478-485.	2.2	21
541	Identification of candidate genes for phenolics accumulation in tomato fruit. <i>Plant Science</i> , 2013, 205-206, 87-96.	1.7	51
542	The Power of Meta-Analysis in Genome-Wide Association Studies. <i>Annual Review of Genomics and Human Genetics</i> , 2013, 14, 441-465.	2.5	107
543	A Comprehensive Assay for CFTR Mutational Analysis Using Next-Generation Sequencing. <i>Clinical Chemistry</i> , 2013, 59, 1481-1488.	1.5	44
544	Leigh syndrome associated with mitochondrial complex I deficiency due to novel mutations in NDUFV1 and NDUF2. <i>Gene</i> , 2013, 516, 162-167.	1.0	41
545	Dissecting Disease Inheritance Modes in a Three-Dimensional Protein Network Challenges the "Guilt-by-Association" Principle. <i>American Journal of Human Genetics</i> , 2013, 93, 78-89.	2.6	44

#	ARTICLE	IF	CITATIONS
546	Algorithms and Guidelines for Interpretation of DNA Variants. , 2013, , 97-112.		2
547	Large Numbers of Genetic Variants Considered to be Pathogenic are Common in Asymptomatic Individuals. Human Mutation, 2013, 34, 1216-1220.	1.1	78
548	Analysis of nucleotide diversity among alleles of the major bacterial blight resistance gene Xa27 in cultivars of rice (<i>Oryza sativa</i>) and its wild relatives. Planta, 2013, 238, 293-305.	1.6	18
549	Somatic Alterations Contributing to Metastasis of a Castration-Resistant Prostate Cancer. Human Mutation, 2013, 34, 1231-1241.	1.1	52
550	SETBP1 mutations occur in 9% of MDS/MPN and in 4% of MPN cases and are strongly associated with atypical CML, monosomy 7, isochromosome i(17)(q10), ASXL1 and CBL mutations. Leukemia, 2013, 27, 1852-1860.	3.3	164
551	Pleiotropy in complex traits: challenges and strategies. Nature Reviews Genetics, 2013, 14, 483-495.	7.7	958
552	Genotypic and phenotypic characterization of Brazilian patients with GM1 gangliosidosis. Gene, 2013, 512, 113-116.	1.0	28
553	Mutational analysis of TYR gene and its structural consequences in OCA1A. Gene, 2013, 513, 184-195.	1.0	47
554	DNA repair genes are selectively mutated in diffuse large B cell lymphomas. Journal of Experimental Medicine, 2013, 210, 1729-1742.	4.2	87
555	Two Polymorphic Variants of ABCC1 Selectively Alter Drug Resistance and Inhibitor Sensitivity of the Multidrug and Organic Anion Transporter Multidrug Resistance Protein 1. Drug Metabolism and Disposition, 2013, 41, 2187-2196.	1.7	22
556	Characterization of 2 Genetic Variants of Na ^v 1.5 Arginine 689 Found in Patients with Cardiac Arrhythmias. Journal of Cardiovascular Electrophysiology, 2013, 24, 1037-1046.	0.8	11
557	High-Resolution Mapping of Complex Traits with a Four-Parent Advanced Intercross Yeast Population. Genetics, 2013, 195, 1141-1155.	1.2	164
558	Capturing the mutational landscape of the beta-lactamase TEM-1. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 13067-13072.	3.3	228
559	Mutational landscape of gingivo-buccal oral squamous cell carcinoma reveals new recurrently-mutated genes and molecular subgroups. Nature Communications, 2013, 4, 2873.	5.8	318
560	Implications of Population History of European Romani on Genetic Susceptibility to Disease. Human Heredity, 2013, 76, 194-200.	0.4	12
561	Differential expression and function of human IL-12R β 2 polymorphic variants. Molecular Immunology, 2013, 56, 380-389.	1.0	16
562	Proteome-wide Analysis of Amino Acid Variations That Influence Protein Lysine Acetylation. Journal of Proteome Research, 2013, 12, 949-958.	1.8	17
563	Developmental Dysplasia of the Hip: Linkage Mapping and Whole Exome Sequencing Identify a Shared Variant in <i>CXCR1</i> in All Affected Members of a Large Multigeneration Family. Journal of Bone and Mineral Research, 2013, 28, 2540-2549.	3.1	47

#	ARTICLE	IF	CITATIONS
564	Integrative pathway analysis of a genome-wide association study of $\dot{V}O_2\text{max}$ response to exercise training. <i>Journal of Applied Physiology</i> , 2013, 115, 1343-1359.	1.2	45
565	INTEGRATIVE ANALYSIS OF TWO CELL LINES DERIVED FROM A NON-SMALL-LUNG CANCER PATIENT – A PANOMICS APPROACH. , 2013, , .		5
566	Combined immunodeficiency with life-threatening EBV-associated lymphoproliferative disorder in patients lacking functional CD27. <i>Haematologica</i> , 2013, 98, 473-478.	1.7	153
567	Next-generation sequencing in the clinical genetic screening of patients with pheochromocytoma and paraganglioma. <i>Endocrine Connections</i> , 2013, 2, 104-111.	0.8	37
568	Replication of genetic loci for ages at menarche and menopause in the multi-ethnic Population Architecture using Genomics and Epidemiology (PAGE) study. <i>Human Reproduction</i> , 2013, 28, 1695-1706.	0.4	64
569	<i>ARHGDI3</i> : a novel gene implicated in nephrotic syndrome. <i>Journal of Medical Genetics</i> , 2013, 50, 330-338.	1.5	92
570	Exome sequencing identifies mutations in the gene <i>TTC7A</i> in French-Canadian cases with hereditary multiple intestinal atresia. <i>Journal of Medical Genetics</i> , 2013, 50, 324-329.	1.5	119
571	De novo mutations in hereditary diffuse leukoencephalopathy with axonal spheroids (HDLS). <i>Neurology</i> , 2013, 81, 2039-2044.	1.5	62
572	Novel findings in patients with primary hyperoxaluria type III and implications for advanced molecular testing strategies. <i>European Journal of Human Genetics</i> , 2013, 21, 162-172.	1.4	71
573	Chapter 15: Disease Gene Prioritization. <i>PLoS Computational Biology</i> , 2013, 9, e1002902.	1.5	65
574	In Silico Screening and Molecular Dynamics Simulation of Disease-Associated nsSNP in TYRP1 Gene and Its Structural Consequences in OCA3. <i>BioMed Research International</i> , 2013, 2013, 1-13.	0.9	69
575	Molecular Insight into the Association Between Cartilage Regeneration and Ear Wound Healing in Genetic Mouse Models: Targeting New Genes in Regeneration. <i>G3: Genes, Genomes, Genetics</i> , 2013, 3, 1881-1891.	0.8	30
576	Mutations in GNAL cause primary torsion dystonia. <i>Nature Genetics</i> , 2013, 45, 88-92.	9.4	281
577	Sex-stratified Genome-wide Association Studies Including 270,000 Individuals Show Sexual Dimorphism in Genetic Loci for Anthropometric Traits. <i>PLoS Genetics</i> , 2013, 9, e1003500.	1.5	371
578	A Recessive Founder Mutation in Regulator of Telomere Elongation Helicase 1, RTEL1, Underlies Severe Immunodeficiency and Features of Hoyeraal Hreidarsson Syndrome. <i>PLoS Genetics</i> , 2013, 9, e1003695.	1.5	106
579	Genomic Analysis of Natural Selection and Phenotypic Variation in High-Altitude Mongolians. <i>PLoS Genetics</i> , 2013, 9, e1003634.	1.5	48
580	An Alteration in ELMOD3, an Arl2 GTPase-Activating Protein, Is Associated with Hearing Impairment in Humans. <i>PLoS Genetics</i> , 2013, 9, e1003774.	1.5	48
581	In Vivo Modeling of the Morbid Human Genome using Danio rerio. <i>Journal of Visualized Experiments</i> , 2013, , e50338.	0.2	49

#	ARTICLE	IF	CITATIONS
582	No association between VAPB mutations and familial or sporadic ALS in Sweden, Portugal and Iceland. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 620-627.	1.1	11
583	Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. Human Molecular Genetics, 2013, 22, 1417-1423.	1.4	105
584	Applications and data analysis of next-generation sequencing. Laboratoriums Medizin, 2013, 37, .	0.1	3
585	Whole Genome, Whole Population Sequencing Reveals That Loss of Signaling Networks Is the Major Adaptive Strategy in a Constant Environment. PLoS Genetics, 2013, 9, e1003972.	1.5	195
586	Positional Cloning Reveals Strain-Dependent Expression of Trim16 to Alter Susceptibility to Bleomycin-Induced Pulmonary Fibrosis in Mice. PLoS Genetics, 2013, 9, e1003203.	1.5	14
587	Causal and Synthetic Associations of Variants in the SERPINA Gene Cluster with Alpha1-antitrypsin Serum Levels. PLoS Genetics, 2013, 9, e1003585.	1.5	43
588	A Mutation in the SUV39H2 Gene in Labrador Retrievers with Hereditary Nasal Parakeratosis (HNPK) Provides Insights into the Epigenetics of Keratinocyte Differentiation. PLoS Genetics, 2013, 9, e1003848.	1.5	35
589	Low Frequency Variants, Collapsed Based on Biological Knowledge, Uncover Complexity of Population Stratification in 1000 Genomes Project Data. PLoS Genetics, 2013, 9, e1003959.	1.5	35
590	First description of phosphofructokinase deficiency in Spain: identification of a novel homozygous missense mutation in the PFKM gene. Frontiers in Physiology, 2013, 4, 393.	1.3	8
591	Mutation of the Diamond-Blackfan Anemia Gene Rps7 in Mouse Results in Morphological and Neuroanatomical Phenotypes. PLoS Genetics, 2013, 9, e1003094.	1.5	47
592	Simultaneous Identification of Multiple Driver Pathways in Cancer. PLoS Computational Biology, 2013, 9, e1003054.	1.5	231
593	Assessing association between protein truncating variants and quantitative traits. Bioinformatics, 2013, 29, 2419-2426.	1.8	12
594	From Mouse to Human: Evolutionary Genomics Analysis of Human Orthologs of Essential Genes. PLoS Genetics, 2013, 9, e1003484.	1.5	168
595	The Role of ABCA1 Gene Sequence Variants on Risk of Alzheimer's Disease. Journal of Alzheimer's Disease, 2013, 38, 897-906.	1.2	45
596	A Resequencing Analysis of Genomic Loci on Chromosomes 1q32.1, 5p15.33, and 13q22.1 Associated With Pancreatic Cancer Risk. Pancreas, 2013, 42, 209-215.	0.5	5
597	Lymphatic abnormalities are associated with <i>RASA1</i> gene mutations in mouse and man. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 8621-8626.	3.3	116
598	The contribution of common UGT2B10 and CYP2A6 alleles to variation in nicotine glucuronidation among European Americans. Pharmacogenetics and Genomics, 2013, 23, 706-716.	0.7	13
599	Exome Sequencing Finds a Novel <i>PCSK1</i> Mutation in a Child With Generalized Malabsorptive Diarrhea and Diabetes Insipidus. Journal of Pediatric Gastroenterology and Nutrition, 2013, 57, 759-767.	0.9	29

#	ARTICLE	IF	CITATIONS
600	Collagen Genes and Exercise-Associated Muscle Cramping. <i>Clinical Journal of Sport Medicine</i> , 2013, 23, 64-69.	0.9	20
601	Functional anatomy of distant-acting mammalian enhancers. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , 2013, 368, 20120359.	1.8	40
602	Substitution at <i>IL1RN</i> and Deletion at <i>SLC4A11</i> Segregating with Phenotype in Familial Keratoconus. , 2013, 54, 2207.		37
603	A mutation in the <i>FAM36A</i> gene, the human ortholog of <i>COX20</i> , impairs cytochrome c oxidase assembly and is associated with ataxia and muscle hypotonia. <i>Human Molecular Genetics</i> , 2013, 22, 656-667.	1.4	75
604	Genome-Wide Association Study Identifies 3 Genomic Loci Significantly Associated With Serum Levels of Homocysteine. <i>Circulation: Cardiovascular Genetics</i> , 2013, 6, 505-513.	5.1	54
605	Reproduction and Immunity-Driven Natural Selection in the Human <i>WFDC</i> Locus. <i>Molecular Biology and Evolution</i> , 2013, 30, 938-950.	3.5	17
606	PATH-SCAN: A REPORTING TOOL FOR IDENTIFYING CLINICALLY ACTIONABLE VARIANTS. , 2013, , .		9
607	<i>IL1B</i> and <i>NFkB</i> , NSAID use and risk of colorectal cancer in the Colon Cancer Family Registry. <i>Carcinogenesis</i> , 2013, 34, 79-85.	1.3	43
608	Inherited human <i>OX40</i> deficiency underlying classic Kaposi sarcoma of childhood. <i>Journal of Experimental Medicine</i> , 2013, 210, 1743-1759.	4.2	119
609	Loss of metabotropic glutamate receptor 2 escalates alcohol consumption. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 16963-16968.	3.3	105
610	A Scan for Human-Specific Relaxation of Negative Selection Reveals Unexpected Polymorphism in Proteasome Genes. <i>Molecular Biology and Evolution</i> , 2013, 30, 1808-1815.	3.5	23
611	<i>RYR1</i> Mutations as a Cause of Ophthalmoplegia, Facial Weakness, and Malignant Hyperthermia. <i>JAMA Ophthalmology</i> , 2013, 131, 1532.	1.4	26
612	Matrix Gla Protein Gene Polymorphism Is Associated With Increased Coronary Artery Calcification Progression. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2013, 33, 645-651.	1.1	22
613	Short Read (Next-Generation) Sequencing. <i>Circulation: Cardiovascular Genetics</i> , 2013, 6, 427-434.	5.1	23
614	Correlation of Ventricular Arrhythmias With Genotype in Arrhythmogenic Right Ventricular Cardiomyopathy. <i>Circulation: Cardiovascular Genetics</i> , 2013, 6, 552-556.	5.1	51
615	Genome-wide association study meta-analysis of chronic widespread pain: evidence for involvement of the 5p15.2 region. <i>Annals of the Rheumatic Diseases</i> , 2013, 72, 427-436.	0.5	112
616	Functional Recurrent Mutations in the Human Mitochondrial Phylogeny: Dual Roles in Evolution and Disease. <i>Genome Biology and Evolution</i> , 2013, 5, 876-890.	1.1	60
617	Mapping the Signal Peptide Binding and Oligomer Contact Sites of the Core Subunit of the Pea Twin Arginine Protein Translocase. <i>Plant Cell</i> , 2013, 25, 999-1015.	3.1	37

#	ARTICLE	IF	CITATIONS
618	Evolution in Fast Forward: a Potential Role for Mutators in Accelerating <i>Staphylococcus aureus</i> Pathoadaptation. <i>Journal of Bacteriology</i> , 2013, 195, 615-628.	1.0	33
619	Next-generation sequencing of paired tyrosine kinase inhibitor-sensitive and -resistant EGFR mutant lung cancer cell lines identifies spectrum of DNA changes associated with drug resistance. <i>Genome Research</i> , 2013, 23, 1434-1445.	2.4	48
620	Mutation spectrum in South American Lynch syndrome families. <i>Hereditary Cancer in Clinical Practice</i> , 2013, 11, 18.	0.6	26
621	Examination of Rare Missense Variants in the <i>CHRNA5</i> Gene Cluster to Level of Response to Alcohol in the San Diego Sibling Pair Study. <i>Alcoholism: Clinical and Experimental Research</i> , 2013, 37, 1311-1316.	1.4	14
622	Succinate dehydrogenase deficiency in pediatric and adult gastrointestinal stromal tumors. <i>Frontiers in Oncology</i> , 2013, 3, 117.	1.3	44
623	Lessons from postgenome-wide association studies: functional analysis of cancer predisposition loci. <i>Journal of Internal Medicine</i> , 2013, 274, 414-424.	2.7	24
624	Genetic screening in adolescents with steroid-resistant nephrotic syndrome. <i>Kidney International</i> , 2013, 84, 206-213.	2.6	77
625	Pelger-Huet anomaly and a mild skeletal phenotype secondary to mutations in <i>LBR</i> . <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2066-2073.	0.7	27
626	Identification and biochemical analysis of a novel <i>APOB</i> mutation that causes autosomal dominant hypercholesterolemia. <i>Molecular Genetics & Genomic Medicine</i> , 2013, 1, 155-161.	0.6	40
627	Cytoplasmic Mislocalization of <i>POU3F4</i> Due to Novel Mutations Leads to Deafness in Humans and Mice. <i>Human Mutation</i> , 2013, 34, 1102-1110.	1.1	20
628	Clinical and mutation analysis of 51 probands with anophthalmia and/or severe microphthalmia from a single center. <i>Molecular Genetics & Genomic Medicine</i> , 2013, 1, 15-31.	0.6	79
629	Identification of novel point mutations in splicing sites integrating whole-exome and <i>RNA-seq</i> data in myeloproliferative diseases. <i>Molecular Genetics & Genomic Medicine</i> , 2013, 1, 246-259.	0.6	17
630	The Genetics of Dilated Cardiomyopathy: A Prioritized Candidate Gene Study of <i>LMNA</i> , <i>TNNT2</i> , <i>TCAP</i> , and <i>PLN</i> . <i>Clinical Cardiology</i> , 2013, 36, 628-633.	0.7	21
631	Novel mutations in <i>ADAMTSL2</i> gene underlying geleophysic dysplasia in families from United Arab Emirates. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2013, 97, 764-769.	1.6	8
632	Association of type XI collagen genes with chronic Achilles tendinopathy in independent populations from South Africa and Australia. <i>British Journal of Sports Medicine</i> , 2013, 47, 569-574.	3.1	38
633	<i>ASXL1</i> exon 12 mutations are frequent in AML with intermediate risk karyotype and are independently associated with an adverse outcome. <i>Leukemia</i> , 2013, 27, 82-91.	3.3	179
634	A phylomedicine approach to understanding the evolution of auditory sensory perception and disease in mammals. <i>Evolutionary Applications</i> , 2013, 6, 412-422.	1.5	14
635	Novel <i>GNE</i> Mutations in Autosomal Recessive Hereditary Inclusion Body Myopathy Patients. <i>Genetic Testing and Molecular Biomarkers</i> , 2013, 17, 376-382.	0.3	11

#	ARTICLE	IF	CITATIONS
636	Mutations in the autoregulatory domain of β -tubulin 4a cause hereditary dystonia. <i>Annals of Neurology</i> , 2013, 73, 546-553.	2.8	148
637	Comprehensive molecular diagnosis of 179 Leber congenital amaurosis and juvenile retinitis pigmentosa patients by targeted next generation sequencing. <i>Journal of Medical Genetics</i> , 2013, 50, 674-688.	1.5	139
638	Genetic analysis of the <i>FUS</i> / <i>TLS</i> gene in essential tremor. <i>European Journal of Neurology</i> , 2013, 20, 534-539.	1.7	32
639	A novel missense mutation in the <i>NYX</i> gene associated with high myopia. <i>Ophthalmic and Physiological Optics</i> , 2013, 33, 346-353.	1.0	25
640	The phenotypic spectrum of <i>ZIC3</i> mutations includes isolated de novo transposition of the great arteries and double outlet right ventricle. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 792-802.	0.7	32
641	Novel <i>ATP2A2</i> mutations in a large sample of individuals with <i>D</i> arier disease. <i>Journal of Dermatology</i> , 2013, 40, 259-266.	0.6	26
642	Gene \times smoking interactions on human brain gene expression: finding common mechanisms in adolescents and adults. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , 2013, 54, 1109-1119.	3.1	15
643	Novel <i>FOXF1</i> Mutations in Sporadic and Familial Cases of Alveolar Capillary Dysplasia with Misaligned Pulmonary Veins Imply a Role for its DNA Binding Domain. <i>Human Mutation</i> , 2013, 34, 801-811.	1.1	97
644	Targeted resequencing of candidate genes reveals novel variants associated with severe Behçet's uveitis. <i>Experimental and Molecular Medicine</i> , 2013, 45, e49-e49.	3.2	18
645	High-resolution loss of heterozygosity screening implicates <i>PTPRJ</i> as a potential tumor suppressor gene that affects susceptibility to non-Hodgkin's lymphoma. <i>Genes Chromosomes and Cancer</i> , 2013, 52, 467-479.	1.5	19
646	Next-generation <i>DNA</i> sequencing of <i>HEXA</i> : a step in the right direction for carrier screening. <i>Molecular Genetics & Genomic Medicine</i> , 2013, 1, 260-268.	0.6	26
647	Insight into TPMT ²³ mutation mis-folding using molecular dynamics simulation and protein structure analysis. <i>Journal of Biomolecular Structure and Dynamics</i> , 2013, 31, 1066-1076.	2.0	2
648	Phenotype versus Genotype Methods for Copy Number Variant Analysis of Glutathione S-Transferases M1. <i>Annals of Human Genetics</i> , 2013, 77, 409-415.	0.3	6
649	Sequence Diversity of Pan troglodytes Subspecies and the Impact of WFDC6 Selective Constraints in Reproductive Immunity. <i>Genome Biology and Evolution</i> , 2013, 5, 2512-2523.	1.1	1
650	Personalized genomic disease risk of volunteers. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 16957-16962.	3.3	44
651	DGKE Variants Cause a Glomerular Microangiopathy That Mimics Membranoproliferative GN. <i>Journal of the American Society of Nephrology: JASN</i> , 2013, 24, 377-384.	3.0	130
652	Mutation in <i>SNAP25</i> as a novel genetic cause of epilepsy and intellectual disability. <i>Rare Diseases (Austin, Tex)</i> , 2013, 1, e26314.	1.8	55
653	Understanding Human Glycosylation Disorders: Biochemistry Leads the Charge. <i>Journal of Biological Chemistry</i> , 2013, 288, 6936-6945.	1.6	184

#	ARTICLE	IF	CITATIONS
654	Next generation diagnostics of cystic fibrosis and CFTR-related disorders by targeted multiplex high-coverage resequencing of CFTR. <i>Journal of Medical Genetics</i> , 2013, 50, 455-462.	1.5	39
655	The TBC1D1 Gene. <i>Vitamins and Hormones</i> , 2013, 91, 77-95.	0.7	15
656	Functional diversity of the glutathione peroxidase gene family among human populations: implications for genetic predisposition to disease and drug response. <i>Pharmacogenomics</i> , 2013, 14, 1037-1045.	0.6	7
657	Clinical and genetic features in Italian Bietti crystalline dystrophy patients. <i>British Journal of Ophthalmology</i> , 2013, 97, 174-179.	2.1	48
658	Genetic Analysis through OtoSeq of Pakistani Families Segregating Prelingual Hearing Loss. <i>Otolaryngology - Head and Neck Surgery</i> , 2013, 149, 478-487.	1.1	22
659	The mutational landscape of phosphorylation signaling in cancer. <i>Scientific Reports</i> , 2013, 3, 2651.	1.6	149
660	IntOGen-mutations identifies cancer drivers across tumor types. <i>Nature Methods</i> , 2013, 10, 1081-1082.	9.0	517
661	Heterozygous non-synonymous ROBO2 variants are unlikely to be sufficient to cause familial vesicoureteric reflux. <i>Kidney International</i> , 2013, 84, 327-337.	2.6	12
662	Mutational Spectrum of the ZEB1 Gene in Corneal Dystrophies Supports a Genotype-Phenotype Correlation. , 2013, 54, 3215.		65
663	A novel mutation, outside of the candidate region for diagnosis, in the inverted formin 2 gene can cause focal segmental glomerulosclerosis. <i>Kidney International</i> , 2013, 83, 153-159.	2.6	12
664	Mutational analysis of TSC1 and TSC2 in Japanese patients with tuberous sclerosis complex revealed higher incidence of TSC1 patients than previously reported. <i>Journal of Human Genetics</i> , 2013, 58, 216-225.	1.1	19
665	Phenotypic variability of CLDN14 mutations causing DFNB29 hearing loss in the Pakistani population. <i>Journal of Human Genetics</i> , 2013, 58, 102-108.	1.1	22
666	Novel rare variants in congenital cardiac arrhythmia genes are frequent in drug-induced torsades de pointes. <i>Pharmacogenomics Journal</i> , 2013, 13, 325-329.	0.9	61
667	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. <i>Nature Genetics</i> , 2013, 45, 1244-1248.	9.4	289
668	Production of a High-Efficiency TILLING Population through Polyploidization. <i>Plant Physiology</i> , 2013, 161, 1604-1614.	2.3	48
669	A novel intellectual disability syndrome caused by GPI anchor deficiency due to homozygous mutations in PIGT. <i>Journal of Medical Genetics</i> , 2013, 50, 521-528.	1.5	108
670	Evidence for Diversifying Selection in a Set of Mycobacterium tuberculosis Genes in Response to Antibiotic- and Nonantibiotic-Related Pressure. <i>Molecular Biology and Evolution</i> , 2013, 30, 1326-1336.	3.5	43
671	Genome-Wide Patterns of Genetic Variation in Two Domestic Chickens. <i>Genome Biology and Evolution</i> , 2013, 5, 1376-1392.	1.1	65

#	ARTICLE	IF	CITATIONS
672	Identification of a Loss-of-Function Mutation in <i>Ube2l6</i> Associated With Obesity Resistance. <i>Diabetes</i> , 2013, 62, 2784-2795.	0.3	14
674	Heterozygous <i>IGFALS</i> Gene Variants in Idiopathic Short Stature and Normal Children: Impact on Height and the IGF System. <i>Hormone Research in Paediatrics</i> , 2013, 80, 413-423.	0.8	35
675	Somatic <i>MYH7</i> , <i>MYBPC3</i> , <i>TPM1</i> , <i>TNNT2</i> and <i>TNNI3</i> Mutations in Sporadic Hypertrophic Cardiomyopathy. <i>Circulation Journal</i> , 2013, 77, 2358-2365.	0.7	15
676	A novel homozygous mutation of the nicotinamide nucleotide transhydrogenase gene in a Japanese patient with familial glucocorticoid deficiency. <i>Endocrine Journal</i> , 2013, 60, 855-859.	0.7	20
677	Missense mutations of <i>MLH1</i> and <i>MSH2</i> genes detected in patients with gastrointestinal cancer are associated with exonic splicing enhancers and silencers. <i>Oncology Letters</i> , 2013, 5, 1710-1718.	0.8	9
678	<i>CASQ1</i> Gene Is an Unlikely Candidate for Malignant Hyperthermia Susceptibility in the North American Population. <i>Anesthesiology</i> , 2013, 118, 344-349.	1.3	29
679	Identification of a Novel Mutation p.L240T in the <i>FRMD7</i> gene in a Family with Congenital Nystagmus. <i>Scientific Reports</i> , 2013, 3, 3084.	1.6	11
680	Investigating the function of single nucleotide polymorphisms in the <i>CTSB</i> gene: a computational approach. <i>Future Neurology</i> , 2013, 8, 469-483.	0.9	5
681	Mutation in porcine Zip4-like zinc transporter is associated with pancreatic zinc concentration and apparent zinc absorption. <i>British Journal of Nutrition</i> , 2013, 109, 969-976.	1.2	5
682	Genome-wide Association Study Signal at the 12q12 Locus for Crohn's Disease May Represent Associations with the <i>MUC19</i> Gene. <i>Inflammatory Bowel Diseases</i> , 2013, 19, 1254-1259.	0.9	21
683	Contemporary Approaches for Identifying Rare Bone Disease Causing Genes. <i>Bone Research</i> , 2013, 1, 301-310.	5.4	1
684	Intronic rs2147363 Variant in <i>ATP7B</i> Transcription Factor-Binding Site Associated with Alzheimer's Disease. <i>Journal of Alzheimer's Disease</i> , 2013, 37, 453-459.	1.2	24
685	Genetic Variations of <i>NR2A1</i> in Asian Populations: Implications in Pharmacogenetics Studies. <i>Drug Metabolism and Pharmacokinetics</i> , 2013, 28, 278-288.	1.1	4
686	Delineating the genetic heterogeneity of ALS using targeted high-throughput sequencing. <i>Journal of Medical Genetics</i> , 2013, 50, 776-783.	1.5	151
687	<i>In silico</i> prediction of functional loss of <i>cst3</i> gene in hereditary cerebral amyloid angiopathy. <i>Bangladesh Journal of Pharmacology</i> , 2013, 8, .	0.1	0
688	<i>MLH1</i> gene: An <i>in silico</i> analysis. <i>Journal of Computational Biology and Bioinformatics Research</i> , 2013, 5, 1-5.	0.0	2
689	New-generation sequencing (NGS) in hematologic oncology laboratories. <i>Hematologie</i> , 2013, 19, 112-122.	0.0	2
690	Complementation Test of <i>Rpe65</i> Knockout and <i>Tvrn148</i> . , 2013, 54, 5111.		9

#	ARTICLE	IF	CITATIONS
691	Sequencing ASMT Identifies Rare Mutations in Chinese Han Patients with Autism. PLoS ONE, 2013, 8, e53727.	1.1	26
692	Whole Exome Sequencing Suggests Much of Non-BRCA1/BRCA2 Familial Breast Cancer Is Due to Moderate and Low Penetrance Susceptibility Alleles. PLoS ONE, 2013, 8, e55681.	1.1	95
693	Interactions between Genetic Variants in AMH and AMHR2 May Modify Age at Natural Menopause. PLoS ONE, 2013, 8, e59819.	1.1	21
694	A COL11A2 Mutation in Labrador Retrievers with Mild Disproportionate Dwarfism. PLoS ONE, 2013, 8, e60149.	1.1	37
695	Whole Genome and Transcriptome Sequencing of a B3 Thymoma. PLoS ONE, 2013, 8, e60572.	1.1	28
696	Functional and Structural Analysis of C-Terminal BRCA1 Missense Variants. PLoS ONE, 2013, 8, e61302.	1.1	16
697	Minor Hypospadias: The "Tip of the Iceberg" of the Partial Androgen Insensitivity Syndrome. PLoS ONE, 2013, 8, e61824.	1.1	36
698	ARID1A Alterations Are Associated with FGFR3-Wild Type, Poor-Prognosis, Urothelial Bladder Tumors. PLoS ONE, 2013, 8, e62483.	1.1	52
699	Next-Generation Sequencing Identifies Transportin 3 as the Causative Gene for LGMD1F. PLoS ONE, 2013, 8, e63536.	1.1	69
700	Targeted Sequencing of Cancer-Related Genes in Colorectal Cancer Using Next-Generation Sequencing. PLoS ONE, 2013, 8, e64271.	1.1	71
701	Exome Sequencing of 47 Chinese Families with Cone-Rod Dystrophy: Mutations in 25 Known Causative Genes. PLoS ONE, 2013, 8, e65546.	1.1	52
702	Prediction of Disease Causing Non-Synonymous SNPs by the Artificial Neural Network Predictor NetDiseaseSNP. PLoS ONE, 2013, 8, e68370.	1.1	20
703	Functional Coding Variants in SLC6A15, a Possible Risk Gene for Major Depression. PLoS ONE, 2013, 8, e68645.	1.1	11
704	Whole Genome SNP Genotyping and Exome Sequencing Reveal Novel Genetic Variants and Putative Causative Genes in Congenital Hyperinsulinism. PLoS ONE, 2013, 8, e68740.	1.1	25
705	Targeted Re-Sequencing Identified rs3106189 at the 5' UTR of TAPBP and rs1052918 at the 3' UTR of TCF3 to Be Associated with the Overall Survival of Colorectal Cancer Patients. PLoS ONE, 2013, 8, e70307.	1.1	8
706	Functional Properties of Rare Missense Variants of Human CDH13 Found in Adult Attention Deficit/Hyperactivity Disorder (ADHD) Patients. PLoS ONE, 2013, 8, e71445.	1.1	29
707	Prevalence of PALB2 Mutations in Breast Cancer Patients in Multi-Ethnic Asian Population in Malaysia and Singapore. PLoS ONE, 2013, 8, e73638.	1.1	27
708	Evolutionary Reconstruction and Population Genetics Analysis of Aurora Kinases. PLoS ONE, 2013, 8, e75763.	1.1	3

#	ARTICLE	IF	CITATIONS
709	Identification and Characterization of CDH1 Germline Variants in Sporadic Gastric Cancer Patients and in Individuals at Risk of Gastric Cancer. PLoS ONE, 2013, 8, e77035.	1.1	32
710	Assessment of the Geographic Origins of Pinewood Nematode Isolates via Single Nucleotide Polymorphism in Effector Genes. PLoS ONE, 2013, 8, e83542.	1.1	27
711	Detection of Haplotypes Associated with Prenatal Death in Dairy Cattle and Identification of Deleterious Mutations in GART, SHBG and SLC37A2. PLoS ONE, 2013, 8, e65550.	1.1	137
712	Etiology and Audiological Outcomes at 3 Years for 364 Children in Australia. PLoS ONE, 2013, 8, e59624.	1.1	42
713	Comparison of responses of human melanoma cell lines to MEK and BRAF inhibitors. Frontiers in Genetics, 2013, 4, 66.	1.1	40
714	The Growing Importance of CNVs: New Insights for Detection and Clinical Interpretation. Frontiers in Genetics, 2013, 4, 92.	1.1	49
715	Prediction of Deleterious Nonsynonymous Single-Nucleotide Polymorphism for Human Diseases. Scientific World Journal, The, 2013, 2013, 1-10.	0.8	53
716	Computational and Bioinformatics Frameworks for Next-Generation Whole Exome and Genome Sequencing. Scientific World Journal, The, 2013, 2013, 1-10.	0.8	39
717	BRAF Mutant Gastrointestinal Stromal Tumor: First report of regression with BRAF inhibitor dabrafenib (GSK2118436) and whole exomic sequencing for analysis of acquired resistance. Oncotarget, 2013, 4, 310-315.	0.8	140
718	A Novel Angiotensin I-Converting Enzyme Mutation (S333W) Impairs N-Domain Enzymatic Cleavage of the Anti-Fibrotic Peptide, AcSDKP. PLoS ONE, 2014, 9, e88001.	1.1	19
719	Uncovering the Rare Variants of DLC1 Isoform 1 and Their Functional Effects in a Chinese Sporadic Congenital Heart Disease Cohort. PLoS ONE, 2014, 9, e90215.	1.1	14
720	Exome Sequencing of 18 Chinese Families with Congenital Cataracts: A New Sight of the NHS Gene. PLoS ONE, 2014, 9, e100455.	1.1	47
721	Integration of Sequence Data from a Consanguineous Family with Genetic Data from an Outbred Population Identifies PLB1 as a Candidate Rheumatoid Arthritis Risk Gene. PLoS ONE, 2014, 9, e87645.	1.1	34
722	Integrative Genetic Characterization and Phenotype Correlations in Pheochromocytoma and Paraganglioma Tumours. PLoS ONE, 2014, 9, e86756.	1.1	32
723	Whole Genome Analyses of a Well-Differentiated Liposarcoma Reveals Novel SYT1 and DDR2 Rearrangements. PLoS ONE, 2014, 9, e87113.	1.1	14
724	PRODH Polymorphisms, Cortical Volumes and Thickness in Schizophrenia. PLoS ONE, 2014, 9, e87686.	1.1	14
725	Quantitative Trait Loci Affecting Atherosclerosis at the Aortic Root Identified in an Intercross between DBA2J and 129S6 Apolipoprotein E-Null Mice. PLoS ONE, 2014, 9, e88274.	1.1	15
726	Phenotypic Diversity of Breast Cancer-Related Mutations in Metalloproteinase-Disintegrin ADAM12. PLoS ONE, 2014, 9, e92536.	1.1	11

#	ARTICLE	IF	CITATIONS
727	Bovine Exome Sequence Analysis and Targeted SNP Genotyping of Recessive Fertility Defects BH1, HH2, and HH3 Reveal a Putative Causative Mutation in SMC2 for HH3. PLoS ONE, 2014, 9, e92769.	1.1	69
728	Bioinformatics Pipelines for Targeted Resequencing and Whole-Exome Sequencing of Human and Mouse Genomes: A Virtual Appliance Approach for Instant Deployment. PLoS ONE, 2014, 9, e95217.	1.1	17
729	Novel Mutations in BMPR2, ACVRL1 and KCNA5 Genes and Hemodynamic Parameters in Patients with Pulmonary Arterial Hypertension. PLoS ONE, 2014, 9, e100261.	1.1	43
730	RYR2 Sequencing Reveals Novel Missense Mutations in a Kazakh Idiopathic Ventricular Tachycardia Study Cohort. PLoS ONE, 2014, 9, e101059.	1.1	7
731	In Silico Analysis of Functional Single Nucleotide Polymorphisms in the Human TRIM22 Gene. PLoS ONE, 2014, 9, e101436.	1.1	15
732	Non-Coding Polymorphisms in Nucleotide Binding Domain 1 in ABCC1 Gene Associate with Transcript Level and Survival of Patients with Breast Cancer. PLoS ONE, 2014, 9, e101740.	1.1	14
733	Identification of Novel GRM1 Mutations and Single Nucleotide Polymorphisms in Prostate Cancer Cell Lines and Tissues. PLoS ONE, 2014, 9, e103204.	1.1	14
734	Computational Screening and Molecular Dynamic Simulation of Breast Cancer Associated Deleterious Non-Synonymous Single Nucleotide Polymorphisms in TP53 Gene. PLoS ONE, 2014, 9, e104242.	1.1	40
735	Targeted Next-Generation Sequencing Reveals Novel USH2A Mutations Associated with Diverse Disease Phenotypes: Implications for Clinical and Molecular Diagnosis. PLoS ONE, 2014, 9, e105439.	1.1	25
736	Two Novel Tyrosinase (TYR) Gene Mutations with Pathogenic Impact on Oculocutaneous Albinism Type 1 (OCA1). PLoS ONE, 2014, 9, e106656.	1.1	34
737	ErbB2 Is Required for Cardiac Atrial Electrical Activity during Development. PLoS ONE, 2014, 9, e107041.	1.1	7
738	Combining Structural Modeling with Ensemble Machine Learning to Accurately Predict Protein Fold Stability and Binding Affinity Effects upon Mutation. PLoS ONE, 2014, 9, e107353.	1.1	71
739	Exome Sequencing Reveals Novel and Recurrent Mutations with Clinical Significance in Inherited Retinal Dystrophies. PLoS ONE, 2014, 9, e116176.	1.1	16
740	Computational Analysis Reveals the Association of Threonine 118 Methionine Mutation in PMP22 Resulting in CMT-1A. Advances in Bioinformatics, 2014, 2014, 1-10.	5.7	82
741	Impaired 8-Hydroxyguanine Repair Activity of MUTYH Variant p.Arg109Trp Found in a Japanese Patient with Early-Onset Colorectal Cancer. Oxidative Medicine and Cellular Longevity, 2014, 2014, 1-12.	1.9	10
742	AUTO-MUTE 2.0: A Portable Framework with Enhanced Capabilities for Predicting Protein Functional Consequences upon Mutation. Advances in Bioinformatics, 2014, 2014, 1-7.	5.7	53
743	Genetic polymorphisms and haplotypes of the organic cation transporter 1 gene (SLC22A1) in the Xhosa population of South Africa. Genetics and Molecular Biology, 2014, 37, 350-359.	0.6	15
744	Lessons and Implications from Genome-Wide Association Studies (GWAS) Findings of Blood Cell Phenotypes. Genes, 2014, 5, 51-64.	1.0	12

#	ARTICLE	IF	CITATIONS
745	Architecture of Inherited Susceptibility to Colorectal Cancer: A Voyage of Discovery. <i>Genes</i> , 2014, 5, 270-284.	1.0	13
746	SDS, a structural disruption score for assessment of missense variant deleteriousness. <i>Frontiers in Genetics</i> , 2014, 5, 82.	1.1	13
748	c.G2114A MYH9 mutation (DFNA17) causes non-syndromic autosomal dominant hearing loss in a Brazilian family. <i>Genetics and Molecular Biology</i> , 2014, 37, 616-621.	0.6	7
749	Discovery of somatic mutations in the progression of chronic myeloid leukemia by whole-exome sequencing. <i>Genetics and Molecular Research</i> , 2014, 13, 945-953.	0.3	11
752	Computational screening of disease associated mutations on NPC1 gene and its structural consequence in Niemann-Pick type-C1. <i>Frontiers in Biology</i> , 2014, 9, 410-421.	0.7	2
754	Germline Alterations in <i>RASAL1</i> in Cowden Syndrome Patients Presenting with Follicular Thyroid Cancer and in Individuals with Apparently Sporadic Epithelial Thyroid Cancer. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014, 99, E1316-E1321.	1.8	20
755	Association mapping for wood quality and growth traits in <i>Eucalyptus globulus</i> ssp. <i>globulus</i> Labill identifies nine stable marker-trait associations for seven traits. <i>Tree Genetics and Genomes</i> , 2014, 10, 1661-1678.	0.6	34
756	Multiple Nonglycemic Genomic Loci Are Newly Associated With Blood Level of Glycated Hemoglobin in East Asians. <i>Diabetes</i> , 2014, 63, 2551-2562.	0.3	61
757	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. <i>Journal of Neuromuscular Diseases</i> , 2014, 1, 135-149.	1.1	25
758	Modification of PCR Conditions and Design of Exon-Specific Primers for the Efficient Molecular Diagnosis of PKD1 Mutations. <i>Kidney and Blood Pressure Research</i> , 2014, 39, 536-545.	0.9	5
759	The Role of Molecular Genetic Analysis in the Diagnosis of Primary Ciliary Dyskinesia. <i>Annals of the American Thoracic Society</i> , 2014, 11, 351-359.	1.5	47
760	A novel 2-bp indel within Krüppel-like factor 15 gene (<i>KLF15</i>) and its associations with chicken growth and carcass traits. <i>British Poultry Science</i> , 2014, 55, 427-434.	0.8	15
761	Whole exome sequencing of a single osteosarcoma case—integrative analysis with whole transcriptome RNA-seq data. <i>Human Genomics</i> , 2014, 8, 20.	1.4	27
762	VAS: a convenient web portal for efficient integration of genomic features with millions of genetic variants. <i>BMC Genomics</i> , 2014, 15, 886.	1.2	1
763	Identification of <i>CNGA3</i> Mutations in 46 Families. <i>JAMA Ophthalmology</i> , 2014, 132, 1076.	1.4	29
764	Application of high-throughput sequencing for studying genomic variations in congenital heart disease. <i>Briefings in Functional Genomics</i> , 2014, 13, 51-65.	1.3	16
765	FLAGS, frequently mutated genes in public exomes. <i>BMC Medical Genomics</i> , 2014, 7, 64.	0.7	108
766	MotorPlex provides accurate variant detection across large muscle genes both in single myopathic patients and in pools of DNA samples. <i>Acta Neuropathologica Communications</i> , 2014, 2, 100.	2.4	76

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767	Bioinformatics in otolaryngology research. Part one: concepts in DNA sequencing and gene expression analysis. <i>Journal of Laryngology and Otology</i> , 2014, 128, 848-858.	0.4	1
768	Postzygotic single-nucleotide mosaicisms in whole-genome sequences of clinically unremarkable individuals. <i>Cell Research</i> , 2014, 24, 1311-1327.	5.7	54
769	Identifying rare variants for genetic risk through a combined pedigree and phenotype approach: application to suicide and asthma. <i>Translational Psychiatry</i> , 2014, 4, e471-e471.	2.4	8
770	A novel locus for episodic ataxia:UBR4 the likely candidate. <i>European Journal of Human Genetics</i> , 2014, 22, 505-510.	1.4	70
771	Respiratory failure in a term newborn due to compound heterozygous ABCA3 mutation: the case report of another lethal variant. <i>Journal of Perinatology</i> , 2014, 34, 951-953.	0.9	7
772	Meta-analysis of genome-wide association studies in East Asian-ancestry populations identifies four new loci for body mass index. <i>Human Molecular Genetics</i> , 2014, 23, 5492-5504.	1.4	192
773	Mutation Analysis of Seven Known Glaucoma-Associated Genes in Chinese Patients With Glaucoma. , 2014, 55, 3594.		50
774	Malignant effects of multiple rare variants in sarcomere genes on the prognosis of patients with hypertrophic cardiomyopathy. <i>European Journal of Heart Failure</i> , 2014, 16, 950-957.	2.9	53
775	Mutations in <i>NTRK3</i> Suggest a Novel Signaling Pathway in Human Congenital Heart Disease. <i>Human Mutation</i> , 2014, 35, 1459-1468.	1.1	17
776	Comprehensive Functional Annotation of 77 Prostate Cancer Risk Loci. <i>PLoS Genetics</i> , 2014, 10, e1004102.	1.5	167
777	VarWalker: Personalized Mutation Network Analysis of Putative Cancer Genes from Next-Generation Sequencing Data. <i>PLoS Computational Biology</i> , 2014, 10, e1003460.	1.5	96
778	Hypoxia Adaptations in the Grey Wolf (<i>Canis lupus chanco</i>) from Qinghai-Tibet Plateau. <i>PLoS Genetics</i> , 2014, 10, e1004466.	1.5	169
779	A framework for organizing cancer-related variations from existing databases, publications and NGS data using a High-performance Integrated Virtual Environment (HIVE). <i>Database: the Journal of Biological Databases and Curation</i> , 2014, 2014, bau022.	1.4	62
780	The relative mRNA expression of p53 isoforms in breast cancer is associated with clinical features and outcome. <i>Carcinogenesis</i> , 2014, 35, 586-596.	1.3	67
781	Rare Variants in Genes Encoding MuRF1 and MuRF2 Are Modifiers of Hypertrophic Cardiomyopathy. <i>International Journal of Molecular Sciences</i> , 2014, 15, 9302-9313.	1.8	39
782	Use of Long Term Molecular Dynamics Simulation in Predicting Cancer Associated SNPs. <i>PLoS Computational Biology</i> , 2014, 10, e1003318.	1.5	103
783	Identification of Rare Causal Variants in Sequence-Based Studies: Methods and Applications to VPS13B, a Gene Involved in Cohen Syndrome and Autism. <i>PLoS Genetics</i> , 2014, 10, e1004729.	1.5	45
784	Integrating <i>In Silico</i> Prediction Methods, Molecular Docking, and Molecular Dynamics Simulation to Predict the Impact of ALK Missense Mutations in Structural Perspective. <i>BioMed Research International</i> , 2014, 2014, 1-14.	0.9	40

#	ARTICLE	IF	CITATIONS
785	Genomic View of Bipolar Disorder Revealed by Whole Genome Sequencing in a Genetic Isolate. <i>PLoS Genetics</i> , 2014, 10, e1004229.	1.5	69
786	Mapping Small Effect Mutations in <i>Saccharomyces cerevisiae</i> : Impacts of Experimental Design and Mutational Properties. <i>G3: Genes, Genomes, Genetics</i> , 2014, 4, 1205-1216.	0.8	22
787	GeneSV – an Approach to Help Characterize Possible Variations in Genomic and Protein Sequences. <i>Bioinformatics and Biology Insights</i> , 2014, 8, BBI.S13076.	1.0	5
788	The Impairment of MAGMAS Function in Human Is Responsible for a Severe Skeletal Dysplasia. <i>PLoS Genetics</i> , 2014, 10, e1004311.	1.5	34
789	A Mutation in the FAM83G Gene in Dogs with Hereditary Footpad Hyperkeratosis (HFH). <i>PLoS Genetics</i> , 2014, 10, e1004370.	1.5	43
790	Analysis of Stop-Gain and Frameshift Variants in Human Innate Immunity Genes. <i>PLoS Computational Biology</i> , 2014, 10, e1003757.	1.5	32
791	Integrated Pathway-Based Approach Identifies Association between Genomic Regions at CTCF and CACNB2 and Schizophrenia. <i>PLoS Genetics</i> , 2014, 10, e1004345.	1.5	44
792	DEFLATE Compression Algorithm Corrects for Overestimation of Phylogenetic Diversity by Grantham Approach to Single-Nucleotide Polymorphism Classification. <i>International Journal of Molecular Sciences</i> , 2014, 15, 8491-8508.	1.8	1
793	CanvasDB: a local database infrastructure for analysis of targeted- and whole genome re-sequencing projects. <i>Database: the Journal of Biological Databases and Curation</i> , 2014, 2014, bau098-bau098.	1.4	21
794	Biosynthetic and functional defects in newly identified SLC4A11 mutants and absence of COL8A2 mutations in Fuchs endothelial corneal dystrophy. <i>Journal of Human Genetics</i> , 2014, 59, 444-453.	1.1	32
795	Complement Mutations in Diacylglycerol Kinase – Associated Atypical Hemolytic Uremic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2014, 9, 1611-1619.	2.2	61
796	In silico identification of genetic variants in glucocerebrosidase (GBA) gene involved in Gaucher's disease using multiple software tools. <i>Frontiers in Genetics</i> , 2014, 5, 148.	1.1	18
797	Evolution- and Structure-Based Computational Strategy Reveals the Impact of Deleterious Missense Mutations on MODY 2 (Maturity-Onset Diabetes of the Young, Type 2). <i>Theranostics</i> , 2014, 4, 366-385.	4.6	48
798	HCV&TB: a comprehensive online resource on human genes and genetic variants associated with tuberculosis. <i>Database: the Journal of Biological Databases and Curation</i> , 2014, 2014, bau112-bau112.	1.4	3
799	Canine Hereditary Ataxia in Old English Sheepdogs and Gordon Setters Is Associated with a Defect in the Autophagy Gene Encoding RAB24. <i>PLoS Genetics</i> , 2014, 10, e1003991.	1.5	33
800	Integrating Multiple Genomic Data to Predict Disease-Causing Nonsynonymous Single Nucleotide Variants in Exome Sequencing Studies. <i>PLoS Genetics</i> , 2014, 10, e1004237.	1.5	50
801	Expanding the spectrum of HEXA mutations in Indian patients with Tay – Sachs disease. <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 425-430.	0.4	10
802	Next-Generation Sequencing (NGS) in Anatomic Pathology Discovery and Practice. <i>Methods in Pharmacology and Toxicology</i> , 2014, , 219-257.	0.1	1

#	ARTICLE	IF	CITATIONS
803	Functional Characterization and Categorization of Missense Mutations that Cause Methylmalonyl-CoA Mutase (MUT) Deficiency. Human Mutation, 2014, 35, 1449-1458.	1.1	40
804	Genetic characterization of Greek population isolates reveals strong genetic drift at missense and trait-associated variants. Nature Communications, 2014, 5, 5345.	5.8	60
805	Constitutional Mismatch Repair-deficiency and Whole-exome Sequencing as the Means of the Rapid Detection of the Causative MSH6 Defect. Klinische Padiatrie, 2014, 226, 357-361.	0.2	13
806	The road from next-generation sequencing to personalized medicine. Personalized Medicine, 2014, 11, 523-544.	0.8	40
807	Paralogue annotation identifies novel pathogenic variants in patients with Brugada syndrome and catecholaminergic polymorphic ventricular tachycardia. Journal of Medical Genetics, 2014, 51, 35-44.	1.5	44
808	Insights into the Maize Pan-Genome and Pan-Transcriptome. Plant Cell, 2014, 26, 121-135.	3.1	498
809	Pitfalls in Genetic Analysis of Pheochromocytomas/Paragangliomas—Case Report. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 2321-2326.	1.8	8
810	Challenges using diagnostic next-generation sequencing in the clinical environment for inherited retinal disorders. Personalized Medicine, 2014, 11, 99-111.	0.8	7
811	Dyskeratosis congenita caused by a novel TERT point mutation in siblings with pancytopenia and exudative retinopathy. Pediatric Blood and Cancer, 2014, 61, 2302-2304.	0.8	17
812	The Lotus japonicus Genome. Compendium of Plant Genomes, 2014, , .	0.3	7
813	Contribution of high-throughput DNA sequencing to the study of primary immunodeficiencies. European Journal of Immunology, 2014, 44, 2854-2861.	1.6	56
814	Copy number alterations and neoplasia-specific mutations in MELK, PDCD1LG2, TLN1, and PAX5 at 9p in different neoplasias. Genes Chromosomes and Cancer, 2014, 53, 579-588.	1.5	14
815	Novel IRF6 mutations in families with Van Der Woude syndrome and popliteal pterygium syndrome from sub-Saharan Africa. Molecular Genetics & Genomic Medicine, 2014, 2, 254-260.	0.6	24
816	Mutations in STAP1 Are Associated With Autosomal Dominant Hypercholesterolemia. Circulation Research, 2014, 115, 552-555.	2.0	146
817	Mutations of NOTCH3 in childhood pulmonary arterial hypertension. Molecular Genetics & Genomic Medicine, 2014, 2, 229-239.	0.6	47
818	Linked Charcot-Marie-Tooth disease predominates in a cohort of multiethnic Malaysian patients. Muscle and Nerve, 2014, 49, 198-201.	1.0	9
819	Mitochondrial serine protease HTRA2 p.G399S in a kindred with essential tremor and Parkinson disease. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 18285-18290.	3.3	147
820	Neonatal diabetes, gallbladder agenesis, duodenal atresia, and intestinal malrotation caused by a novel homozygous mutation in RFX6. Pediatric Diabetes, 2014, 15, 67-72.	1.2	57

#	ARTICLE	IF	CITATIONS
821	Global and disease-associated genetic variation in the human Fanconi anemia gene family. <i>Human Molecular Genetics</i> , 2014, 23, 6815-6825.	1.4	12
822	Osteoporosis Caused by Mutations in <i>PLS3</i> : Clinical and Bone Tissue Characteristics. <i>Journal of Bone and Mineral Research</i> , 2014, 29, 1805-1814.	3.1	78
823	Diamond's Blackfan anemia with mandibulofacial dystostosis is heterogeneous, including the novel DBA genes <i>TSR2</i> and <i>RPS28</i> . <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2240-2249.	0.7	121
824	ALK fusion and its association with other driver gene mutations in Finnish non-small cell lung cancer patients. <i>Genes Chromosomes and Cancer</i> , 2014, 53, 895-901.	1.5	14
825	17 α -Hydroxylase Deficiency Diagnosed in Early Infancy Caused by a Novel Mutation of the <i>CYP17A1</i> Gene. <i>Hormone Research in Paediatrics</i> , 2014, 81, 350-355.	0.8	5
826	Functional implications of the p.Cys680Arg mutation in the MLH1 mismatch repair protein. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 352-355.	0.6	1
827	<i>BRDT</i> gene sequence in human testicular pathologies and the implication of its single nucleotide polymorphism (rs3088232) on fertility. <i>Andrology</i> , 2014, 2, 641-647.	1.9	9
828	Disease-related mutations among Caribbean Hispanics with familial dementia. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 430-437.	0.6	36
829	The role of the interactome in the maintenance of deleterious variability in human populations. <i>Molecular Systems Biology</i> , 2014, 10, 752.	3.2	28
830	Haploinsufficiency of insulin gene enhancer protein 1 (<i>ISL1</i>) is associated with transposition of the great arteries. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 341-351.	0.6	14
831	Regularized Rare Variant Enrichment Analysis for Case-Control Exome Sequencing Data. <i>Genetic Epidemiology</i> , 2014, 38, 104-113.	0.6	7
832	Characterization of <i>F8</i> defects in haemophilia A in Pakistan: investigation of correlation between mutation type and the <i>in vitro</i> thrombin generation assay. <i>Haemophilia</i> , 2014, 20, 287-293.	1.0	5
833	Deciphering intrafamilial phenotypic variability by exome sequencing in a Bardet-Biedl family. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 124-133.	0.6	13
834	A Novel Ribosomopathy Caused by Dysfunction of RPL10 Disrupts Neurodevelopment and Causes X-Linked Microcephaly in Humans. <i>Genetics</i> , 2014, 198, 723-733.	1.2	92
835	Two Novel Mutations in the <i>BCKDK</i> (Branched-Chain Keto-Acid Dehydrogenase Kinase) Gene Are Responsible for a Neurobehavioral Deficit in Two Pediatric Unrelated Patients. <i>Human Mutation</i> , 2014, 35, 470-477.	1.1	70
836	Personalized pharmacogenomics profiling using whole-genome sequencing. <i>Pharmacogenomics</i> , 2014, 15, 1223-1234.	0.6	90
837	Exome Sequencing as a Diagnostic Tool for Pediatric Onset Ataxia. <i>Human Mutation</i> , 2014, 35, 45-49.	1.1	91
838	Genetic associations of nonsynonymous exonic variants with psychophysiological endophenotypes. <i>Psychophysiology</i> , 2014, 51, 1300-1308.	1.2	21

#	ARTICLE	IF	CITATIONS
839	Gain-of-function mutation in <i>TASK4</i> channels and severe cardiac conduction disorder. <i>EMBO Molecular Medicine</i> , 2014, 6, 937-951.	3.3	60
840	Mapping genetic modifiers of survival in a mouse model of Dravet syndrome. <i>Genes, Brain and Behavior</i> , 2014, 13, 163-172.	1.1	145
841	Homozygous and Compound-Heterozygous Mutations in <i>TGDS</i> Cause Catel-Manzke Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 763-770.	2.6	37
842	From the era of genome analysis to the era of genomic drug discovery: a pioneering example of rheumatoid arthritis. <i>Clinical Genetics</i> , 2014, 86, 432-440.	1.0	16
843	Whole exome sequencing identifies variation in <i>CYB5A</i> and <i>RNF10</i> associated with adiposity and type 2 diabetes. <i>Obesity</i> , 2014, 22, 984-988.	1.5	37
844	Mild nasal clefting may be predictive for <i>ALX4</i> heterozygotes. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2054-2058.	0.7	9
845	Evolutionary genetics and implications of small size and twinning in callitrichine primates. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 1467-1472.	3.3	66
846	Neural tube defects and atypical deletion on 22q11.2. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2701-2706.	0.7	6
847	Novel mutations in the glucocerebrosidase gene of Indian patients with Gaucher disease. <i>Journal of Human Genetics</i> , 2014, 59, 223-228.	1.1	21
848	Genome-Wide Association Study of Primary Dentition Pit-and-Fissure and Smooth Surface Caries. <i>Caries Research</i> , 2014, 48, 330-338.	0.9	38
849	Molecular Screening of Keratoconus Susceptibility Sequence Variants in <i>VSX1</i> , <i>TGFBI</i> , <i>DOCK9</i> , <i>STK24</i> , and <i>IPO5</i> Genes in Polish Patients and Novel <i>TGFBI</i> Variant Identification. <i>Ophthalmic Genetics</i> , 2016, 37, 1-7.	0.5	38
850	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. <i>Genome Biology</i> , 2014, 15, R53.	13.9	101
851	Choice of transcripts and software has a large effect on variant annotation. <i>Genome Medicine</i> , 2014, 6, 26.	3.6	158
852	A web-based interactive framework to assist in the prioritization of disease candidate genes in whole-exome sequencing studies. <i>Nucleic Acids Research</i> , 2014, 42, W88-W93.	6.5	39
853	<i>OTX2</i> mutations cause autosomal dominant pattern dystrophy of the retinal pigment epithelium. <i>Journal of Medical Genetics</i> , 2014, 51, 797-805.	1.5	40
854	Systems genomics evaluation of the SH-SY5Y neuroblastoma cell line as a model for Parkinson's disease. <i>BMC Genomics</i> , 2014, 15, 1154.	1.2	126
855	Unraveling <i>Mycobacterium tuberculosis</i> genomic diversity and evolution in Lisbon, Portugal, a highly drug resistant setting. <i>BMC Genomics</i> , 2014, 15, 991.	1.2	52
856	Exome sequencing helped the fine diagnosis of two siblings afflicted with atypical Timothy syndrome (TS2). <i>BMC Medical Genetics</i> , 2014, 15, 48.	2.1	43

#	ARTICLE	IF	CITATIONS
857	A novel insertion mutation identified in exon 10 of the MEFV gene associated with Familial Mediterranean Fever. <i>BMC Medical Genetics</i> , 2014, 15, 74.	2.1	2
858	Combined examination of sequence and copy number variations in human deafness genes improves diagnosis for cases of genetic deafness. <i>BMC Ear, Nose and Throat Disorders</i> , 2014, 14, 9.	2.6	24
859	Mutation analysis of PALB2 in BRCA1 and BRCA2-negative breast and/or ovarian cancer families from Eastern Ontario, Canada. <i>Hereditary Cancer in Clinical Practice</i> , 2014, 12, 19.	0.6	19
860	Non-synonymous genetic variation in exonic regions of canine Toll-like receptors. <i>Canine Genetics and Epidemiology</i> , 2014, 1, 11.	2.9	9
861	Expanding the phenotype of PRPS1 syndromes in females: neuropathy, hearing loss and retinopathy. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 190.	1.2	31
862	MSEA: detection and quantification of mutation hotspots through mutation set enrichment analysis. <i>Genome Biology</i> , 2014, 15, 489.	3.8	54
863	Computational approaches to interpreting genomic sequence variation. <i>Genome Medicine</i> , 2014, 6, 87.	3.6	33
864	The Functional Significance of Common Polymorphisms in Zinc Finger Transcription Factors. <i>G3: Genes, Genomes, Genetics</i> , 2014, 4, 1647-1655.	0.8	9
865	In Silico Analysis Identification of a Novel Germ-Line VHL Mutation in a Patient of Malignant Pheochromocytoma. <i>Endocrine Practice</i> , 2014, 20, e96-e101.	1.1	1
866	Late-Onset Lattice Corneal Dystrophy Without Typical Lattice Lines Caused by a Novel Mutation in the TGFBI Gene. <i>Cornea</i> , 2014, 33, 294-299.	0.9	14
867	Inherited biallelic CSF3R mutations in severe congenital neutropenia. <i>Blood</i> , 2014, 123, 3811-3817.	0.6	79
868	Mutation Screen of LOXL1 in Patients With Female Pelvic Organ Prolapse. <i>Female Pelvic Medicine and Reconstructive Surgery</i> , 2014, 20, 316-321.	0.6	10
869	Interleukin 23 in Crohn's Disease. <i>Inflammatory Bowel Diseases</i> , 2014, 20, 587-595.	0.9	35
870	The cavefish genome reveals candidate genes for eye loss. <i>Nature Communications</i> , 2014, 5, 5307.	5.8	256
871	Functional and Clinical Impact of Novel <i>Tmprss6</i> Variants in Iron-Refractory Iron-Deficiency Anemia Patients and Genotype-Phenotype Studies. <i>Human Mutation</i> , 2014, 35, n/a-n/a.	1.1	53
872	NECTAR: a database of codon-centric missense variant annotations. <i>Nucleic Acids Research</i> , 2014, 42, D1013-D1019.	6.5	3
873	Ataxia and hypogonadism caused by the loss of ubiquitin ligase activity of the U box protein CHIP. <i>Human Molecular Genetics</i> , 2014, 23, 1013-1024.	1.4	136
874	Haploinsufficiency of <i>CSF1R</i> and clinicopathologic characterization in patients with HDLS. <i>Neurology</i> , 2014, 82, 139-148.	1.5	103

#	ARTICLE	IF	CITATIONS
875	Hoyeraal-Hreidarsson syndrome caused by a germline mutation in the TEL patch of the telomere protein TPP1. <i>Genes and Development</i> , 2014, 28, 2090-2102.	2.7	106
876	Complement Factor B Mutations in Atypical Hemolytic Uremic Syndrome—Disease-Relevant or Benign?. <i>Journal of the American Society of Nephrology: JASN</i> , 2014, 25, 2053-2065.	3.0	107
877	An integrated computational approach can classify VHL missense mutations according to risk of clear cell renal carcinoma. <i>Human Molecular Genetics</i> , 2014, 23, 5976-5988.	1.4	21
878	AVIA: an interactive web-server for annotation, visualization and impact analysis of genomic variations. <i>Bioinformatics</i> , 2014, 30, 1013-1014.	1.8	5
879	Post-GWAS Analyses. <i>Statistics in the Health Sciences</i> , 2014, , 285-327.	0.2	0
880	Clinical Bioinformatics. <i>Methods in Molecular Biology</i> , 2014, , .	0.4	6
881	Variation in Genes that Regulate Blood Pressure Are Associated with Glomerular Filtration Rate in Chinese. <i>PLoS ONE</i> , 2014, 9, e92468.	1.1	9
882	Mutation in <i>TMEM98</i> in a Large White Kindred With Autosomal Dominant Nanophthalmos Linked to 17p12-q12. <i>JAMA Ophthalmology</i> , 2014, 132, 970.	1.4	54
883	Phenotypic Overlap Between Familial Exudative Vitreoretinopathy and Microcephaly, Lymphedema, and Chorioretinal Dysplasia Caused by <i>KIF11</i> Mutations. <i>JAMA Ophthalmology</i> , 2014, 132, 1393.	1.4	95
884	A High-Definition View of Functional Genetic Variation from Natural Yeast Genomes. <i>Molecular Biology and Evolution</i> , 2014, 31, 872-888.	3.5	328
885	Expansion of Ocular Phenotypic Features Associated With Mutations in <i>ADAMTS18</i> . <i>JAMA Ophthalmology</i> , 2014, 132, 996.	1.4	15
886	Identification of Novel ROR2 Gene Mutations in Indian Children with Robinow Syndrome. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , 2014, 6, 79-83.	0.4	7
887	Considerations for rare variants in drug metabolism genes and the clinical implications. <i>Expert Opinion on Drug Metabolism and Toxicology</i> , 2014, 10, 873-884.	1.5	22
888	Re-Sequencing Data for Refining Candidate Genes and Polymorphisms in QTL Regions Affecting Adiposity in Chicken. <i>PLoS ONE</i> , 2014, 9, e111299.	1.1	11
889	Cancer Evolution Is Associated with Pervasive Positive Selection on Globally Expressed Genes. <i>PLoS Genetics</i> , 2014, 10, e1004239.	1.5	93
890	RING Finger Protein RNF207, a Novel Regulator of Cardiac Excitation. <i>Journal of Biological Chemistry</i> , 2014, 289, 33730-33740.	1.6	38
891	Whole-Exome Sequencing Data – Identifying Somatic Mutations. , 2014, , 419-427.		0
892	A programmable method for massively parallel targeted sequencing. <i>Nucleic Acids Research</i> , 2014, 42, e88-e88.	6.5	13

#	ARTICLE	IF	CITATIONS
893	Novel Mutation in <i>ABCA3</i> Resulting in Fatal Congenital Surfactant Deficiency in Two Siblings. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2014, 189, 750-752.	2.5	2
894	Clinical and Molecular Characterization of Enhanced S-Cone Syndrome in Children. <i>JAMA Ophthalmology</i> , 2014, 132, 1341.	1.4	39
895	A Comprehensive In Silico Analysis of the Functional and Structural Impact of Nonsynonymous SNPs in the <i>ABCA1</i> Transporter Gene. <i>Cholesterol</i> , 2014, 2014, 1-19.	1.6	12
896	Mutations in the Gene That Encodes the F-Actin Binding Protein Anillin Cause FSGS. <i>Journal of the American Society of Nephrology: JASN</i> , 2014, 25, 1991-2002.	3.0	124
897	Clinical predictors of a positive genetic test in hypertrophic cardiomyopathy in the Brazilian population. <i>BMC Cardiovascular Disorders</i> , 2014, 14, 36.	0.7	6
898	Genome-wide linkage and exome analyses identify variants of <i>HMCN1</i> for splenic epidermoid cyst. <i>BMC Medical Genetics</i> , 2014, 15, 115.	2.1	3
899	Functional Impact of <i>ZEB1</i> Mutations Associated With Posterior Polymorphous and Fuchs' Endothelial Corneal Dystrophies. , 2014, 55, 6159.		34
900	A web tool for the design and management of panels of genes for targeted enrichment and massive sequencing for clinical applications. <i>Nucleic Acids Research</i> , 2014, 42, W83-W87.	6.5	6
901	BCR-ABL residues interacting with ponatinib are critical to preserve the tumorigenic potential of the oncoprotein. <i>FASEB Journal</i> , 2014, 28, 1221-1236.	0.2	29
902	A novel frameshift mutation and infrequent clinical findings in two cases with Dyggve-Melchior-Clausen syndrome. <i>Clinical Dysmorphology</i> , 2014, 23, 1-7.	0.1	5
903	Neuropathologic Features of Pontocerebellar Hypoplasia Type 6. <i>Journal of Neuropathology and Experimental Neurology</i> , 2014, 73, 1009-1025.	0.9	28
904	Mutations in the collagen XII gene define a new form of extracellular matrix-related myopathy. <i>Human Molecular Genetics</i> , 2014, 23, 2353-2363.	1.4	79
905	A mutation in the human <i>CBP4</i> ortholog <i>UQCC3</i> impairs complex III assembly, activity and cytochrome b stability. <i>Human Molecular Genetics</i> , 2014, 23, 6356-6365.	1.4	69
906	Storage and secretion of naturally occurring von Willebrand factor A domain variants. <i>British Journal of Haematology</i> , 2014, 167, 529-540.	1.2	9
907	National mutation study among Danish patients with hereditary haemorrhagic telangiectasia. <i>Clinical Genetics</i> , 2014, 86, 123-133.	1.0	42
908	Exome sequencing in multiplex autism families suggests a major role for heterozygous truncating mutations. <i>Molecular Psychiatry</i> , 2014, 19, 784-790.	4.1	110
909	Loss of <i>IL36RN</i> Function Does Not Confer Susceptibility to Psoriasis Vulgaris. <i>Journal of Investigative Dermatology</i> , 2014, 134, 271-273.	0.3	25
910	Monitoring of residual disease by next-generation deep-sequencing of <i>RUNX1</i> mutations can identify acute myeloid leukemia patients with resistant disease. <i>Leukemia</i> , 2014, 28, 129-137.	3.3	93

#	ARTICLE	IF	CITATIONS
911	Pharmacogenomics of 17-alpha hydroxyprogesterone caproate for recurrent preterm birth prevention. American Journal of Obstetrics and Gynecology, 2014, 210, 321.e1-321.e21.	0.7	23
912	Novel SCN3A variants associated with focal epilepsy in children. Neurobiology of Disease, 2014, 62, 313-322.	2.1	74
913	WFS1 and non-syndromic low-frequency sensorineural hearing loss: A novel mutation in a Portuguese case. Gene, 2014, 538, 288-291.	1.0	12
914	Forward genetics defines Xylt1 as a key, conserved regulator of early chondrocyte maturation and skeletal length. Developmental Biology, 2014, 385, 67-82.	0.9	44
915	Analysis of CYP27B1 in multiple sclerosis. Journal of Neuroimmunology, 2014, 266, 64-66.	1.1	18
916	Mutations in PCYT1A, Encoding a Key Regulator of Phosphatidylcholine Metabolism, Cause Spondylometaphyseal Dysplasia with Cone-Rod Dystrophy. American Journal of Human Genetics, 2014, 94, 105-112.	2.6	53
917	An Expressed Retrogene of the Master Embryonic Stem Cell Gene POU5F1 Is Associated with Prostate Cancer Susceptibility. American Journal of Human Genetics, 2014, 94, 395-404.	2.6	29
918	ILDR1: Novel mutation and a rare cause of congenital deafness in the Saudi Arabian population. European Journal of Medical Genetics, 2014, 57, 253-258.	0.7	20
919	Clinical and genetic analysis of MAPT, GRN, and C9orf72 genes in Korean patients with frontotemporal dementia. Neurobiology of Aging, 2014, 35, 1213.e13-1213.e17.	1.5	35
920	Computational Screening of Disease-Associated Mutations in OCA2 Gene. Cell Biochemistry and Biophysics, 2014, 68, 97-109.	0.9	53
921	Exome Sequencing Revealed Novel Germline Mutations in Chinese Peutzâ€“Jeghers Syndrome Patients. Digestive Diseases and Sciences, 2014, 59, 64-71.	1.1	15
922	A Novel Missense Mutation in AFG3L2 Associated with Late Onset and Slow Progression of Spinocerebellar Ataxia Type 28. Journal of Molecular Neuroscience, 2014, 52, 493-496.	1.1	22
923	Computational identification and analysis of functional polymorphisms involved in the activation and detoxification genes implicated in endometriosis. Gene, 2014, 542, 89-97.	1.0	13
924	exomeSuite: Whole exome sequence variant filtering tool for rapid identification of putative disease causing SNVs/indels. Genomics, 2014, 103, 169-176.	1.3	22
925	The landscape of somatic mutations in epigenetic regulators across 1,000 paediatric cancer genomes. Nature Communications, 2014, 5, 3630.	5.8	342
926	Low prevalence of germline <i>PALB2</i> mutations in Australian tripleâ€“negative breast cancer. International Journal of Cancer, 2014, 134, 301-305.	2.3	13
927	Computational Approaches and Resources in Single Amino Acid Substitutions Analysis Toward Clinical Research. Advances in Protein Chemistry and Structural Biology, 2014, 94, 365-423.	1.0	22
928	Design, Analysis, and Interpretation of Genome-Wide Association Scans. Statistics in the Health Sciences, 2014, , .	0.2	16

#	ARTICLE	IF	CITATIONS
929	Molecular Docking and Molecular Dynamics Study on the Effect of ERCC1 Deleterious Polymorphisms in ERCC1-XPF Heterodimer. <i>Applied Biochemistry and Biotechnology</i> , 2014, 172, 1265-1281.	1.4	12
930	X-linked Charcot-Marie-Tooth disease, Arts syndrome, and prelingual non-syndromic deafness form a disease continuum: evidence from a family with a novel PRPS1 mutation. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 24.	1.2	42
931	Gene structure and spatio-temporal expression of chicken LPIN2. <i>Molecular Biology Reports</i> , 2014, 41, 4081-4091.	1.0	2
932	Insights into the influence of 5-HT2c aminoacidic variants with the inhibitory action of serotonin inverse agonists and antagonists. <i>Journal of Molecular Modeling</i> , 2014, 20, 2120.	0.8	15
933	Identification of Late Blight Resistance-Related Metabolites and Genes in Potato through Nontargeted Metabolomics. <i>Plant Molecular Biology Reporter</i> , 2014, 32, 584-595.	1.0	65
934	Male-specific genetic effect on hypertension and metabolic disorders. <i>Human Genetics</i> , 2014, 133, 311-319.	1.8	29
935	Human testis-specific genes are under relaxed negative selection. <i>Molecular Genetics and Genomics</i> , 2014, 289, 37-45.	1.0	5
936	Structural and functional in silico analysis of LRRK2 missense substitutions. <i>Molecular Biology Reports</i> , 2014, 41, 2529-2542.	1.0	19
937	Using single cell sequencing data to model the evolutionary history of a tumor. <i>BMC Bioinformatics</i> , 2014, 15, 27.	1.2	58
938	Exome sequencing of extended families with autism reveals genes shared across neurodevelopmental and neuropsychiatric disorders. <i>Molecular Autism</i> , 2014, 5, 1.	2.6	246
939	Application of Evolutionary Based in Silico Methods to Predict the Impact of Single Amino Acid Substitutions in Vitelliform Macular Dystrophy. <i>Advances in Protein Chemistry and Structural Biology</i> , 2014, 94, 177-267.	1.0	10
940	Whole-genome haplotyping using long reads and statistical methods. <i>Nature Biotechnology</i> , 2014, 32, 261-266.	9.4	170
941	Update of the Spectrum of <i>GJB2</i> Mutations in 107 Patients with Nonsyndromic Hearing Loss in the Fujian Population of China. <i>Annals of Human Genetics</i> , 2014, 78, 235-242.	0.3	11
942	Genomic tools in acute myeloid leukemia: From the bench to the bedside. <i>Cancer</i> , 2014, 120, 1134-1144.	2.0	21
943	DR-GAS: A database of functional genetic variants and their phosphorylation states in human DNA repair systems. <i>DNA Repair</i> , 2014, 16, 97-103.	1.3	4
944	Cardiac channelopathy testing in 274 ethnically diverse sudden unexplained deaths. <i>Forensic Science International</i> , 2014, 237, 90-99.	1.3	70
945	FGB mutations leading to congenital quantitative fibrinogen deficiencies: An update and report of four novel mutations. <i>Thrombosis Research</i> , 2014, 133, 868-874.	0.8	23
946	Fitness costs of rifampicin resistance in <i>Mycobacterium tuberculosis</i> are amplified under conditions of nutrient starvation and compensated by mutation in the β subunit of rRNA polymerase. <i>Molecular Microbiology</i> , 2014, 91, 1106-1119.	1.2	85

#	ARTICLE	IF	CITATIONS
947	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. <i>Cell</i> , 2014, 157, 636-650.	13.5	189
948	Somatic mutations found in the healthy blood compartment of a 115-yr-old woman demonstrate oligoclonal hematopoiesis. <i>Genome Research</i> , 2014, 24, 733-742.	2.4	136
949	Novel point mutations in survival motor neuron 1 gene expand the spectrum of phenotypes observed in spinal muscular atrophy patients. <i>Neuromuscular Disorders</i> , 2014, 24, 617-623.	0.3	27
950	Whole-genome sequencing of six dog breeds from continuous altitudes reveals adaptation to high-altitude hypoxia. <i>Genome Research</i> , 2014, 24, 1308-1315.	2.4	235
951	SeqReporter. <i>Journal of Molecular Diagnostics</i> , 2014, 16, 11-22.	1.2	26
952	Mitochondrial DNA sequence associations with dementia and amyloid- β^2 in elderly African Americans. <i>Neurobiology of Aging</i> , 2014, 35, 442.e1-442.e8.	1.5	27
953	Protein aggregation due to nsSNP resulting in P56S VABP protein is associated with amyotrophic lateral sclerosis. <i>Journal of Theoretical Biology</i> , 2014, 354, 72-80.	0.8	19
954	Systems biology approach for mutational and site-specific structural investigation of DNA repair genes for xeroderma pigmentosum. <i>Gene</i> , 2014, 543, 108-117.	1.0	8
955	Combined immunodeficiency associated with homozygous MALT1 mutations. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 1458-1462.e7.	1.5	103
956	LRP4 third β^2 -propeller domain mutations cause novel congenital myasthenia by compromising agrin-mediated MuSK signaling in a position-specific manner. <i>Human Molecular Genetics</i> , 2014, 23, 1856-1868.	1.4	96
957	Next-generation sequencing in childhood disorders. <i>Archives of Disease in Childhood</i> , 2014, 99, 284-290.	1.0	20
958	Non-destructive micro-analytical differentiation of copper pigments in paint layers of works of art using laboratory-based techniques. <i>Spectrochimica Acta - Part A: Molecular and Biomolecular Spectroscopy</i> , 2014, 132, 514-525.	2.0	28
959	Large-scale genetic study in East Asians identifies six new loci associated with colorectal cancer risk. <i>Nature Genetics</i> , 2014, 46, 533-542.	9.4	212
960	Computational Identification of Pathogenic Associated nsSNPs and its Structural Impact in UROD Gene: A Molecular Dynamics Approach. <i>Cell Biochemistry and Biophysics</i> , 2014, 70, 735-746.	0.9	3
961	De novo mutation in the <i>GNAL</i> gene causing seemingly sporadic dystonia in a Serbian patient. <i>Movement Disorders</i> , 2014, 29, 1190-1193.	2.2	13
962	Prioritizing Disease-Linked Variants, Genes, and Pathways with an Interactive Whole-Genome Analysis Pipeline. <i>Human Mutation</i> , 2014, 35, 537-547.	1.1	23
963	An Integrated in Silico Approach to Analyze the Involvement of Single Amino Acid Polymorphisms in FANCD1/BRCA2-PALB2 and FANCD1/BRCA2-RAD51 Complex. <i>Cell Biochemistry and Biophysics</i> , 2014, 70, 939-956.	0.9	8
964	Performance of Protein Disorder Prediction Programs on Amino Acid Substitutions. <i>Human Mutation</i> , 2014, 35, 794-804.	1.1	20

#	ARTICLE	IF	CITATIONS
965	Expanding the phenotypic spectrum of <i>ECEL1</i> -related congenital contracture syndromes. <i>Clinical Genetics</i> , 2014, 85, 562-567.	1.0	27
966	Colorectal cancer risk and patients' survival: influence of polymorphisms in genes somatically mutated in colorectal tumors. <i>Cancer Causes and Control</i> , 2014, 25, 759-769.	0.8	15
967	The deleterious mutation load is insensitive to recent population history. <i>Nature Genetics</i> , 2014, 46, 220-224.	9.4	279
968	DECIPHER: database for the interpretation of phenotype-linked plausibly pathogenic sequence and copy-number variation. <i>Nucleic Acids Research</i> , 2014, 42, D993-D1000.	6.5	195
969	Pathological Unfoldomics of Uncontrolled Chaos: Intrinsically Disordered Proteins and Human Diseases. <i>Chemical Reviews</i> , 2014, 114, 6844-6879.	23.0	231
970	Ancient and Recent Adaptive Evolution in the Antiviral TRIM22 Gene: Identification of a Single-Nucleotide Polymorphism That Impacts TRIM22 Function. <i>Human Mutation</i> , 2014, 35, 1072-1081.	1.1	8
971	DNAJC13 mutations in Parkinson disease. <i>Human Molecular Genetics</i> , 2014, 23, 1794-1801.	1.4	258
972	Intragenic mutations in SMN1 may contribute more significantly to clinical severity than SMN2 copy numbers in some spinal muscular atrophy (SMA) patients. <i>Brain and Development</i> , 2014, 36, 914-920.	0.6	39
973	Clinical and pathological impact of <i>VHL</i> , <i>PBRM1</i> , <i>BAP1</i> , <i>SETD2</i> , <i>KDM6A</i> , and <i>JARID1c</i> in clear cell renal cell carcinoma. <i>Genes Chromosomes and Cancer</i> , 2014, 53, 38-51.	1.5	107
974	A large-scale screen for coding variants predisposing to psoriasis. <i>Nature Genetics</i> , 2014, 46, 45-50.	9.4	183
975	Accurate molecular diagnosis of phenylketonuria and tetrahydrobiopterin-deficient hyperphenylalaninemia using high-throughput targeted sequencing. <i>European Journal of Human Genetics</i> , 2014, 22, 528-534.	1.4	36
976	Whole exome sequencing unravels disease-causing genes in consanguineous families in Qatar. <i>Clinical Genetics</i> , 2014, 86, 134-141.	1.0	77
977	The mutational spectrum of the NF1 gene in neurofibromatosis type I patients from UAE. <i>Child's Nervous System</i> , 2014, 30, 1183-1189.	0.6	8
978	Genetics of sudden cardiac death caused by ventricular arrhythmias. <i>Nature Reviews Cardiology</i> , 2014, 11, 96-111.	6.1	59
979	Mutation of the human mitochondrial phenylalanine-tRNA synthetase causes infantile-onset epilepsy and cytochrome c oxidase deficiency. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2014, 1842, 56-64.	1.8	61
980	Functional annotation of noncoding sequence variants. <i>Nature Methods</i> , 2014, 11, 294-296.	9.0	493
981	Identifying driver mutations from sequencing data of heterogeneous tumors in the era of personalized genome sequencing. <i>Briefings in Bioinformatics</i> , 2014, 15, 244-255.	3.2	38
982	Somatic Mutations in MLH1 and MSH2 Are a Frequent Cause of Mismatch-Repair Deficiency in Lynch Syndrome-Like Tumors. <i>Gastroenterology</i> , 2014, 146, 643-646.e8.	0.6	294

#	ARTICLE	IF	CITATIONS
983	Simultaneous identification and prioritization of variants in familial, de novo, and somatic genetic disorders with VariantMaster. <i>Genome Research</i> , 2014, 24, 349-355.	2.4	36
984	Use of in silico tools for classification of novel missense mutations identified in dystrophin gene in developing countries. <i>Gene</i> , 2014, 535, 250-254.	1.0	6
985	Inactivating CUX1 mutations promote tumorigenesis. <i>Nature Genetics</i> , 2014, 46, 33-38.	9.4	111
986	Exome sequencing of Bardet-Biedl syndrome patient identifies a null mutation in the BBSome subunit <i>BBIP1</i> (<i>BBS18</i>). <i>Journal of Medical Genetics</i> , 2014, 51, 132-136.	1.5	124
987	Comprehensive Genomic Analysis of Rhabdomyosarcoma Reveals a Landscape of Alterations Affecting a Common Genetic Axis in Fusion-Positive and Fusion-Negative Tumors. <i>Cancer Discovery</i> , 2014, 4, 216-231.	7.7	596
988	ALK Mutations Confer Differential Oncogenic Activation and Sensitivity to ALK Inhibition Therapy in Neuroblastoma. <i>Cancer Cell</i> , 2014, 26, 682-694.	7.7	302
989	Next-generation sequencing of salivary high-grade neuroendocrine carcinomas identifies alterations in <i>RB1</i> and the mTOR pathway. <i>Experimental and Molecular Pathology</i> , 2014, 97, 572-578.	0.9	10
990	The Use of Next-Generation Sequencing in Molecular Diagnosis of Neurofibromatosis Type 1: A Validation Study. <i>Genetic Testing and Molecular Biomarkers</i> , 2014, 18, 722-735.	0.3	33
991	Inferring non-synonymous single nucleotide polymorphisms-disease associations via integration of multiple similarity networks. <i>IET Systems Biology</i> , 2014, 8, 33-40.	0.8	1
992	Whole genome sequencing identifies a novel occludin mutation in microcephaly with band-like calcification and polymicrogyria that extends the phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1614-1617.	0.7	14
993	Rare variant of unknown significance in <i>POLG1</i> and diagnostic dilemma. <i>Journal of Neurology</i> , 2014, 261, 2218-2220.	1.8	1
994	Recurrent central nervous system white matter changes in charcot-Marie-Tooth type X disease. <i>Muscle and Nerve</i> , 2014, 49, 451-454.	1.0	19
995	$\text{I}\kappa\text{B}$ Kinase $\text{I}\kappa\text{B}$ Mutations in Lymphomas That Constitutively Activate Canonical Nuclear Factor $\text{I}\kappa\text{B}$ ($\text{NF}\kappa\text{B}$) Signaling. <i>Journal of Biological Chemistry</i> , 2014, 289, 26960-26972.	1.6	20
996	Guidelines for genetic studies in single patients: lessons from primary immunodeficiencies. <i>Journal of Experimental Medicine</i> , 2014, 211, 2137-2149.	4.2	218
997	Whole-Genome Sequencing of Tibetan Macaque (<i>Macaca thibetana</i>) Provides New Insight into the Macaque Evolutionary History. <i>Molecular Biology and Evolution</i> , 2014, 31, 1475-1489.	3.5	49
998	Genetic profiling of thymic carcinoma using targeted next-generation sequencing. <i>Lung Cancer</i> , 2014, 86, 174-179.	0.9	28
999	Personalized Diagnosis and Management of Congenital Cataract by Next-Generation Sequencing. <i>Ophthalmology</i> , 2014, 121, 2124-2137.e2.	2.5	153
1000	Characteristics of Neutral and Deleterious Protein-Coding Variation among Individuals and Populations. <i>American Journal of Human Genetics</i> , 2014, 95, 421-436.	2.6	89

#	ARTICLE	IF	CITATIONS
1001	Nueva mutación en SPG11 en una paciente con paraplejía espástica hereditaria complicada: hallazgos clínicos-electrofisiológicos y moleculares. <i>Neurología Argentina</i> , 2014, 6, 155-159.	0.1	0
1002	PHENOTYPIC AND GENOTYPIC CONVERGENCES ARE INFLUENCED BY HISTORICAL CONTINGENCY AND ENVIRONMENT IN YEAST. <i>Evolution; International Journal of Organic Evolution</i> , 2014, 68, 772-790.	1.1	46
1003	Intrafamilial variability of the primary dystonia DYT6 phenotype caused by p.Cys5Trp mutation in THAP1 gene. <i>Neurologia I Neurocirurgia Polska</i> , 2014, 48, 254-257.	0.6	0
1004	Prediction of the influences of missense mutations on cholesteryl ester transfer protein structure. <i>Archives of Biochemistry and Biophysics</i> , 2014, 564, 67-73.	1.4	4
1005	Identifying driver mutations in sequenced cancer genomes: computational approaches to enable precision medicine. <i>Genome Medicine</i> , 2014, 6, 5.	3.6	186
1006	FamAnn: an automated variant annotation pipeline to facilitate target discovery for family-based sequencing studies. <i>Bioinformatics</i> , 2014, 30, 1175-1176.	1.8	7
1007	TLR3 deficiency in herpes simplex encephalitis. <i>Neurology</i> , 2014, 83, 1888-1897.	1.5	128
1008	BAP1 germline mutation in two first grade family members with uveal melanoma. <i>British Journal of Ophthalmology</i> , 2014, 98, 224-227.	2.1	25
1009	Crossover Heterogeneity in the Absence of Hotspots in <i>Caenorhabditis elegans</i> . <i>Genetics</i> , 2014, 196, 137-148.	1.2	62
1010	Genomic Resource Projects. , 2014, , 153-171.		0
1011	Macronodular Adrenal Hyperplasia due to Mutations in an Armadillo Repeat Containing 5 (<i>ARMC5</i>) Gene: A Clinical and Genetic Investigation. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014, 99, E1113-E1119.	1.8	127
1012	<i>MEFV</i> mutations affecting pyrin amino acid 577 cause autosomal dominant autoinflammatory disease. <i>Annals of the Rheumatic Diseases</i> , 2014, 73, 455-461.	0.5	101
1013	Cordova: Web-based management of genetic variation data. <i>Bioinformatics</i> , 2014, 30, 3438-3439.	1.8	3
1014	Variants in Nicotinamide Adenine Dinucleotide Phosphate Oxidase Complex Components Determine Susceptibility to Very Early Onset Inflammatory Bowel Disease. <i>Gastroenterology</i> , 2014, 147, 680-689.e2.	0.6	106
1015	Comparative Functional Analysis of <i>DPYD</i> Variants of Potential Clinical Relevance to Dihydropyrimidine Dehydrogenase Activity. <i>Cancer Research</i> , 2014, 74, 2545-2554.	0.4	136
1016	Genetic Variants in ICAM1, PPARGC1A and MTHFR Are Potentially Associated with Different Phenotypes of Diabetic Retinopathy. <i>Ophthalmologica</i> , 2014, 232, 156-162.	1.0	20
1017	An activating NLR4 inflammasome mutation causes autoinflammation with recurrent macrophage activation syndrome. <i>Nature Genetics</i> , 2014, 46, 1140-1146.	9.4	585
1018	A survey of tools for variant analysis of next-generation genome sequencing data. <i>Briefings in Bioinformatics</i> , 2014, 15, 256-278.	3.2	480

#	ARTICLE	IF	CITATIONS
1019	Pathway analysis of genome-wide association study on serum prostate-specific antigen levels. <i>Gene</i> , 2014, 551, 86-91.	1.0	16
1020	Mutations in POLE and survival of colorectal cancer patients – link to disease stage and treatment. <i>Cancer Medicine</i> , 2014, 3, 1527-1538.	1.3	56
1022	Rare-Variant Association Analysis: Study Designs and Statistical Tests. <i>American Journal of Human Genetics</i> , 2014, 95, 5-23.	2.6	837
1023	The common marmoset genome provides insight into primate biology and evolution. <i>Nature Genetics</i> , 2014, 46, 850-857.	9.4	225
1024	Next-generation sequencing for mitochondrial disorders. <i>British Journal of Pharmacology</i> , 2014, 171, 1837-1853.	2.7	51
1025	Annotation of functional variation within non-MHC MS susceptibility loci through bioinformatics analysis. <i>Genes and Immunity</i> , 2014, 15, 466-476.	2.2	8
1026	Molecular Analysis, Pathogenic Mechanisms, and Readthrough Therapy on a Large Cohort of <i>abuki</i> Syndrome Patients. <i>Human Mutation</i> , 2014, 35, 841-850.	1.1	87
1027	Novel β -Actinin 2 Variant Associated With Familial Hypertrophic Cardiomyopathy and Juvenile Atrial Arrhythmias. <i>Circulation: Cardiovascular Genetics</i> , 2014, 7, 741-750.	5.1	74
1028	Somatic aberrations of mismatch repair genes as a cause of microsatellite-unstable cancers. <i>Journal of Pathology</i> , 2014, 234, 548-559.	2.1	134
1029	Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families. <i>Human Mutation</i> , 2014, 35, 1203-1210.	1.1	75
1030	Rare functional variants in genome-wide association identified candidate genes for nonsyndromic clefts in the African population. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2567-2571.	0.7	35
1031	The genome of the stress-tolerant wild tomato species <i>Solanum pennellii</i> . <i>Nature Genetics</i> , 2014, 46, 1034-1038.	9.4	391
1032	Mutation profile of BBS genes in Iranian patients with Bardet-Biedl syndrome: genetic characterization and report of nine novel mutations in five BBS genes. <i>Journal of Human Genetics</i> , 2014, 59, 368-375.	1.1	33
1033	Genome-wide association analyses identify variants in developmental genes associated with hypospadias. <i>Nature Genetics</i> , 2014, 46, 957-963.	9.4	97
1034	Targeted next-generation sequencing of deafness genes in hearing-impaired individuals uncovers informative mutations. <i>Genetics in Medicine</i> , 2014, 16, 945-953.	1.1	85
1035	Molecular pathogenesis of congenital diaphragmatic hernia revealed by exome sequencing, developmental data, and bioinformatics. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 12450-12455.	3.3	49
1036	Patterns of somatic alterations between matched primary and metastatic colorectal tumors characterized by whole-genome sequencing. <i>Genomics</i> , 2014, 104, 234-241.	1.3	58
1037	Genome Editing: A Tool For Research and Therapy: Towards a functional understanding of variants for molecular diagnostics using genome editing. <i>Nature Medicine</i> , 2014, 20, 1103-1104.	15.2	14

#	ARTICLE	IF	CITATIONS
1038	Bioinformatics Analysis of Pancreas Cancer Genome in High-Throughput Genomic Technologies. , 2014, , 93-131.		1
1039	CUX1, a haploinsufficient tumour suppressor gene overexpressed in advanced cancers. Nature Reviews Cancer, 2014, 14, 673-682.	12.8	100
1040	Fine-mapping quantitative trait loci affecting murine external ear tissue regeneration in the LG/J by SM/J advanced intercross line. Heredity, 2014, 112, 508-518.	1.2	27
1041	The Phenotypic Variability of Retinal Dystrophies Associated With Mutations in CRX, With Report of a Novel Macular Dystrophy Phenotype. Investigative Ophthalmology and Visual Science, 2014, 55, 6934-6944.	3.3	59
1042	Whole genome and exome sequencing of monozygotic twins discordant for Crohn's disease. BMC Genomics, 2014, 15, 564.	1.2	39
1043	Evaluation of gene-based association tests for analyzing rare variants using Genetic Analysis Workshop 18 data. BMC Proceedings, 2014, 8, S9.	1.8	7
1044	Genetic Analysis Workshop 18 single-nucleotide variant prioritization based on protein impact, sequence conservation, and gene annotation. BMC Proceedings, 2014, 8, S11.	1.8	10
1045	De novo mutations discovered in 8 Mexican American families through whole genome sequencing. BMC Proceedings, 2014, 8, S24.	1.8	11
1046	Understanding the sequence requirements of protein families: insights from the BioVis 2013 contests. BMC Proceedings, 2014, 8, S1.	1.8	4
1047	Conserved recurrent gene mutations correlate with pathway deregulation and clinical outcomes of lung adenocarcinoma in never-smokers. BMC Medical Genomics, 2014, 7, 32.	0.7	49
1048	Whole genome sequencing reveals potential targets for therapy in patients with refractory KRASmutated metastatic colorectal cancer. BMC Medical Genomics, 2014, 7, 36.	0.7	18
1049	Whole-exome sequencing identifies a de novo TUBA1A mutation in a patient with sporadic malformations of cortical development: a case report. BMC Research Notes, 2014, 7, 465.	0.6	19
1050	Biotinidase deficiency: clinical and genetic studies of 38 Brazilian patients. BMC Medical Genetics, 2014, 15, 96.	2.1	16
1051	MBASED: allele-specific expression detection in cancer tissues and cell lines. Genome Biology, 2014, 15, 405.	3.8	112
1052	Metastatic tumor evolution and organoid modeling implicate TGFBR2as a cancer driver in diffuse gastric cancer. Genome Biology, 2014, 15, 428.	3.8	110
1053	De novo mutations in schizophrenia implicate chromatin remodeling and support a genetic overlap with autism and intellectual disability. Molecular Psychiatry, 2014, 19, 652-658.	4.1	332
1054	Full-gene sequencing analysis of NAT2 and its relationship with isoniazid pharmacokinetics in Venezuelan children with tuberculosis. Pharmacogenomics, 2014, 15, 285-296.	0.6	14
1055	HLA SNPs and amino acid variants are associated with nasopharyngeal carcinoma in Malaysian Chinese. International Journal of Cancer, 2015, 136, 678-687.	2.3	48

#	ARTICLE	IF	CITATIONS
1056	Functional analysis of novel alpha-1 antitrypsin variants G320R and V321F. <i>Molecular Biology Reports</i> , 2014, 41, 6133-6141.	1.0	3
1057	A novel TSC2 mutation in a Korean patient with tuberous sclerosis complex. <i>Neurological Sciences</i> , 2014, 35, 1487-1489.	0.9	0
1059	Effects of hematopoietic stem cell transplantation on acyl-CoA oxidase deficiency: a sibling comparison study. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 791-799.	1.7	17
1060	Characterization of the fan1 locus in soybean line A5 and development of molecular assays for high-throughput genotyping of FAD3 genes. <i>Molecular Breeding</i> , 2014, 33, 895-907.	1.0	18
1061	Whole-genome sequencing of 234 bulls facilitates mapping of monogenic and complex traits in cattle. <i>Nature Genetics</i> , 2014, 46, 858-865.	9.4	697
1062	A novel mutation in TTC8 is associated with progressive retinal atrophy in the golden retriever. <i>Canine Genetics and Epidemiology</i> , 2014, 1, 4.	2.9	30
1063	Resequencing the susceptibility gene, ITGAM, identifies two functionally deleterious rare variants in systemic lupus erythematosus cases. <i>Arthritis Research and Therapy</i> , 2014, 16, R114.	1.6	22
1064	Excess of rare novel loss-of-function variants in synaptic genes in schizophrenia and autism spectrum disorders. <i>Molecular Psychiatry</i> , 2014, 19, 872-879.	4.1	160
1065	Hypomyelination with atrophy of the basal ganglia and cerebellum: further delineation of the phenotype and genotype-phenotype correlation. <i>Brain</i> , 2014, 137, 1921-1930.	3.7	161
1066	Contribution of <i>RIT1</i> mutations to the pathogenesis of Noonan syndrome: Four new cases and further evidence of heterogeneity. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2310-2316.	0.7	42
1067	Enrichment of pathogenic alleles in the brittle cornea gene, ZNF469, in keratoconus. <i>Human Molecular Genetics</i> , 2014, 23, 5527-5535.	1.4	56
1068	Strategies to Design and Analyze Targeted Sequencing Data. <i>Circulation: Cardiovascular Genetics</i> , 2014, 7, 335-343.	5.1	18
1069	The MOGE(S) Classification of Cardiomyopathy for Clinicians. <i>Journal of the American College of Cardiology</i> , 2014, 64, 304-318.	1.2	158
1070	Genetic signature of differential sensitivity to stevioside in the Italian population. <i>Genes and Nutrition</i> , 2014, 9, 401.	1.2	33
1071	Computational pipeline to identify and characterize functional mutations in ornithine transcarbamylase deficiency. <i>3 Biotech</i> , 2014, 4, 621-634.	1.1	8
1072	Mitochondrial genomic variation associated with higher mitochondrial copy number: the Cache County Study on Memory Health and Aging. <i>BMC Bioinformatics</i> , 2014, 15, S6.	1.2	15
1073	EFIN: predicting the functional impact of nonsynonymous single nucleotide polymorphisms in human genome. <i>BMC Genomics</i> , 2014, 15, 455.	1.2	21
1074	A community-based resource for automatic exome variant-calling and annotation in Mendelian disorders. <i>BMC Genomics</i> , 2014, 15, S5.	1.2	17

#	ARTICLE	IF	CITATIONS
1075	CONSTANS is a photoperiod regulated activator of flowering in sorghum. <i>BMC Plant Biology</i> , 2014, 14, 148.	1.6	83
1076	Compound heterozygous mutations in glycyI-tRNA synthetase are a proposed cause of systemic mitochondrial disease. <i>BMC Medical Genetics</i> , 2014, 15, 36.	2.1	41
1077	Genome instability in blood cells of a BRCA1 + breast cancer family. <i>BMC Cancer</i> , 2014, 14, 342.	1.1	8
1078	Seeking genetic signature of radiosensitivity - a novel method for data analysis in case of small sample sizes. <i>Theoretical Biology and Medical Modelling</i> , 2014, 11, S2.	2.1	4
1079	Screening of mutations in <i>GNAL</i> in sporadic dystonia patients. <i>Movement Disorders</i> , 2014, 29, 1193-1196.	2.2	19
1080	A specific missense mutation in GTF2I occurs at high frequency in thymic epithelial tumors. <i>Nature Genetics</i> , 2014, 46, 844-849.	9.4	208
1081	A Mutation of COX6A1 Causes a Recessive Axonal or Mixed Form of Charcot-Marie-Tooth Disease. <i>American Journal of Human Genetics</i> , 2014, 95, 294-300.	2.6	65
1082	Targeted Sequencing of a Pediatric Metabolic Bone Gene Panel Using a Desktop Semiconductor Next-Generation Sequencer. <i>Calcified Tissue International</i> , 2014, 95, 323-331.	1.5	22
1083	FGF5 is a crucial regulator of hair length in humans. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 10648-10653.	3.3	132
1084	Novel heterozygous ABCB4 gene mutation causing recurrent first-trimester intrahepatic cholestasis of pregnancy. <i>Journal of Perinatology</i> , 2014, 34, 711-712.	0.9	17
1085	Phevor Combines Multiple Biomedical Ontologies for Accurate Identification of Disease-Causing Alleles in Single Individuals and Small Nuclear Families. <i>American Journal of Human Genetics</i> , 2014, 94, 599-610.	2.6	175
1086	Accumulation of Somatic Mutations in TP53 in Gastric Epithelium With <i>Helicobacter pylori</i> Infection. <i>Gastroenterology</i> , 2014, 147, 407-417.e3.	0.6	121
1087	Compound heterozygous mutations in the C6 gene of a child with recurrent infections. <i>Molecular Immunology</i> , 2014, 58, 201-205.	1.0	14
1088	Novel compound heterozygous mutations of POLR3A revealed by whole-exome sequencing in a patient with hypomyelination. <i>Brain and Development</i> , 2014, 36, 315-321.	0.6	20
1089	Two novel mutations in the SLCO2A1 gene in a Chinese patient with primary hypertrophic osteoarthropathy. <i>Gene</i> , 2014, 534, 421-423.	1.0	20
1090	Interplay between base excision repair activity and toxicity of 3-methyladenine DNA glycosylases in an <i>E. coli</i> complementation system. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2014, 763-764, 64-73.	0.4	3
1091	A novel missense mutation in POMT1 modulates the severe congenital muscular dystrophy phenotype associated with POMT1 nonsense mutations. <i>Neuromuscular Disorders</i> , 2014, 24, 312-320.	0.3	14
1092	Standardized decision support in next generation sequencing reports of somatic cancer variants. <i>Molecular Oncology</i> , 2014, 8, 859-873.	2.1	80

#	ARTICLE	IF	CITATIONS
1093	Variant Association Tools for Quality Control and Analysis of Large-Scale Sequence and Genotyping Array Data. <i>American Journal of Human Genetics</i> , 2014, 94, 770-783.	2.6	71
1094	Category fluency, latent semantic analysis and schizophrenia: a candidate gene approach. <i>Cortex</i> , 2014, 55, 182-191.	1.1	67
1095	A newly discovered LGI1 mutation in Korean family with autosomal dominant lateral temporal lobe epilepsy. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2014, 23, 69-73.	0.9	13
1096	AP1S3 Mutations Are Associated with Pustular Psoriasis and Impaired Toll-like Receptor 3 Trafficking. <i>American Journal of Human Genetics</i> , 2014, 94, 790-797.	2.6	153
1097	A novel trafficking-defective HCN4 mutation is associated with early-onset atrial fibrillation. <i>Heart Rhythm</i> , 2014, 11, 1055-1062.	0.3	64
1098	Results of fibrillin-1 gene analysis in children from inbred families with lens subluxation. <i>Journal of AAPOS</i> , 2014, 18, 134-139.	0.2	9
1099	Extensive molecular genetic survey of Taiwanese patients with amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2014, 35, 2423.e1-2423.e6.	1.5	46
1100	Collection, integration and analysis of cancer genomic profiles: from data to insight. <i>Current Opinion in Genetics and Development</i> , 2014, 24, 92-98.	1.5	22
1101	A case-control study between interleukin-10 gene variants and periodontal disease in dogs. <i>Gene</i> , 2014, 539, 75-81.	1.0	6
1102	In silico investigation of the ATP7B gene: insights from functional prediction of non-synonymous substitution to protein structure. <i>BioMetals</i> , 2014, 27, 53-64.	1.8	21
1103	In silico analysis of TTR gene (coding and non-coding regions, and interactive network) and its implications in transthyretin-related amyloidosis. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2014, 21, 154-162.	1.4	16
1104	Unraveling Cellular Phenotypes of Novel TorsinA/TOR1A Mutations. <i>Human Mutation</i> , 2014, 35, 1114-1122.	1.1	34
1105	Novel somatic and germline mutations in intracranial germ cell tumours. <i>Nature</i> , 2014, 511, 241-245.	13.7	181
1106	Whole-Exome Sequencing Identifies KIZ as a Ciliary Gene Associated with Autosomal-Recessive Rod-Cone Dystrophy. <i>American Journal of Human Genetics</i> , 2014, 94, 625-633.	2.6	52
1107	Novel Calmodulin Mutations Associated With Congenital Arrhythmia Susceptibility. <i>Circulation: Cardiovascular Genetics</i> , 2014, 7, 466-474.	5.1	165
1108	GWAS identifies novel SLE susceptibility genes and explains the association of the HLA region. <i>Genes and Immunity</i> , 2014, 15, 347-354.	2.2	109
1109	GABAA receptor biogenesis is impaired by the Î²2 subunit febrile seizure-associated mutation, GABRG2(R177G). <i>Neurobiology of Disease</i> , 2014, 69, 215-224.	2.1	29
1110	Genetic variation in the CYP1A1 gene is related to circulating PCB118 levels in a population-based sample. <i>Environmental Research</i> , 2014, 133, 135-140.	3.7	11

#	ARTICLE	IF	CITATIONS
1111	Significant survival impact of MACC1 polymorphisms in HER2 positive breast cancer patients. <i>European Journal of Cancer</i> , 2014, 50, 2134-2141.	1.3	29
1112	A novel LITAF/SIMPLE mutation within a family with a demyelinating form of Charcot-Marie-Tooth disease. <i>Journal of the Neurological Sciences</i> , 2014, 343, 183-186.	0.3	15
1113	Genetic association of KCNJ10 rs1130183 with seizure susceptibility and computational analysis of deleterious non-synonymous SNPs of KCNJ10 gene. <i>Gene</i> , 2014, 536, 247-253.	1.0	22
1114	Analysis of all subunits, SDHA, SDHB, SDHC, SDHD, of the succinate dehydrogenase complex in KIT/PDGFR α wild-type GIST. <i>European Journal of Human Genetics</i> , 2014, 22, 32-39.	1.4	90
1115	Dipeptide analysis of p53 mutations and evolution of p53 family proteins. <i>Biochimica Et Biophysica Acta - Proteins and Proteomics</i> , 2014, 1844, 198-206.	1.1	10
1116	Exome sequencing reveals a novel mutation, p.L325H, in the KRT5 gene associated with autosomal dominant Epidermolysis Bullosa Simplex Koebner type in a large family from western India. <i>Human Genome Variation</i> , 2014, 1, 14007.	0.4	8
1117	The Molecular Basis of Retinal Dystrophies in Pakistan. <i>Genes</i> , 2014, 5, 176-195.	1.0	20
1118	Application of Massively Parallel Sequencing in the Clinical Diagnostic Testing of Inherited Cardiac Conditions. <i>Medical Sciences (Basel, Switzerland)</i> , 2014, 2, 98-126.	1.3	2
1119	Novel KRAS Gene Mutations in Sporadic Colorectal Cancer. <i>PLoS ONE</i> , 2014, 9, e113350.	1.1	11
1122	The F-BAR protein PSTPIP1 controls extracellular matrix degradation and filopodia formation in macrophages. <i>Blood</i> , 2014, 123, 2703-2714.	0.6	83
1123	Mutations in GATA2 are rare in juvenile myelomonocytic leukemia. <i>Blood</i> , 2014, 123, 1426-1427.	0.6	12
1124	Whole-exome sequencing and functional studies identify RPS29 as a novel gene mutated in multicase Diamond-Blackfan anemia families. <i>Blood</i> , 2014, 124, 24-32.	0.6	79
1125	Sequencing of Charcot-Marie-Tooth disease genes in a toxic polyneuropathy. <i>Annals of Neurology</i> , 2014, 76, 727-737.	2.8	63
1126	Inflammation-Related Genetic Variations and Survival in Patients With Advanced Non-Small Cell Lung Cancer Receiving First-Line Chemotherapy. <i>Clinical Pharmacology and Therapeutics</i> , 2014, 96, 360-369.	2.3	16
1127	Rare key functional domain missense substitutions in MRE11A, RAD50, and NBN contribute to breast cancer susceptibility: results from a Breast Cancer Family Registry case-control mutation-screening study. <i>Breast Cancer Research</i> , 2014, 16, R58.	2.2	99
1128	Mutation Update for UBE3A Variants in Angelman Syndrome. <i>Human Mutation</i> , 2014, 35, 1407-1417.	1.1	56
1129	Expanding genotype/phenotype of neuromuscular diseases by comprehensive target capture/NGS. <i>Neurology: Genetics</i> , 2015, 1, e14.	0.9	48
1130	Genome-wide variation in the pinewood nematode <i>Bursaphelenchus xylophilus</i> and its relationship with pathogenic traits. <i>BMC Genomics</i> , 2015, 16, 845.	1.2	27

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1149	Novel common variants and susceptible haplotype for exfoliation glaucoma specific to Asian population. <i>Scientific Reports</i> , 2015, 4, 5340.	1.6	23
1150	Identification of two novel mutations in CDHR1 in consanguineous Spanish families with autosomal recessive retinal dystrophy. <i>Scientific Reports</i> , 2015, 5, 13902.	1.6	30
1151	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. <i>Scientific Reports</i> , 2015, 4, 7132.	1.6	29
1152	Development of a novel pink-eyed dilution mouse model showing progressive darkening of the eyes and coat hair with aging. <i>Experimental Animals</i> , 2015, 64, 207-220.	0.7	6
1153	Oculodentodigital Dysplasia with Massive Brain Calcification and a New Mutation of GJA1 Gene. <i>Journal of Alzheimer's Disease</i> , 2015, 49, 27-30.	1.2	11
1154	DYNC2L1 mutations broaden the clinical spectrum of dynein-2 defects. <i>Scientific Reports</i> , 2015, 5, 11649.	1.6	28
1155	Î2-cateninC429S mice exhibit sterility consequent to spatiotemporally sustained Wnt signalling in the internal genitalia. <i>Scientific Reports</i> , 2014, 4, 6959.	1.6	7
1156	Prioritization Of Nonsynonymous Single Nucleotide Variants For Exome Sequencing Studies Via Integrative Learning On Multiple Genomic Data. <i>Scientific Reports</i> , 2015, 5, 14955.	1.6	10
1157	Investigation of Pathogenic Genes in Chinese sporadic Hypertrophic Cardiomyopathy Patients by Whole Exome Sequencing. <i>Scientific Reports</i> , 2015, 5, 16609.	1.6	39
1158	Identification of two novel Darier disease-associated mutations in the ATP2A2 gene. <i>Molecular Medicine Reports</i> , 2015, 12, 1845-1849.	1.1	4
1159	A patient with PMP22-related hereditary neuropathy and DBH-gene-related dysautonomia. <i>Journal of Neurology</i> , 2015, 262, 2373-2381.	1.8	8
1160	CATCHing putative causative variants in consanguineous families. <i>BMC Bioinformatics</i> , 2015, 16, 310.	1.2	12
1161	Variants in the interleukin-1 alpha and beta genes, and the risk for periodontal disease in dogs. <i>Journal of Genetics</i> , 2015, 94, 651-659.	0.4	7
1162	Interpretation of personal genome sequencing data in terms of disease ranks based on mutual information. <i>BMC Medical Genomics</i> , 2015, 8, S4.	0.7	6
1163	BigQ: a NoSQL based framework to handle genomic variants in i2b2. <i>BMC Bioinformatics</i> , 2015, 16, 415.	1.2	20
1164	Runs of homozygosity reveal signatures of positive selection for reproduction traits in breed and non-breed horses. <i>BMC Genomics</i> , 2015, 16, 764.	1.2	125
1165	Germline activating MTOR mutation arising through gonadal mosaicism in two brothers with megalencephaly and neurodevelopmental abnormalities. <i>BMC Medical Genetics</i> , 2015, 16, 102.	2.1	23
1166	CoagVDb: a comprehensive database for coagulation factors and their associated SAPs. <i>Biological Research</i> , 2015, 48, 35.	1.5	4

#	ARTICLE	IF	CITATIONS
1167	Minimally symptomatic mcardle disease, expanding the genotypeâ€“phenotype spectrum. <i>Muscle and Nerve</i> , 2015, 52, 891-895.	1.0	6
1168	Infantile onset Vanishing White Matter disease associated with a novel <i>EIF2B5</i> variant, remarkably long life span, severe epilepsy, and hypopituitarism. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 826-830.	0.7	5
1169	A de novo Mutation in <i>KMT2A</i> (<i>MLL</i>) in monozygotic twins with Wiedemannâ€“Steiner Syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2182-2187.	0.7	23
1170	Novel <i>CDKN2A</i> mutations in Austrian melanoma patients. <i>Melanoma Research</i> , 2015, 25, 412-420.	0.6	10
1171	Relapsing encephalopathy with cerebellar ataxia related to an <i>ATP1A3</i> mutation. <i>Developmental Medicine and Child Neurology</i> , 2015, 57, 1183-1186.	1.1	78
1172	Inverted formin 2â€“related Charcotâ€“Marieâ€“Tooth disease: extension of the mutational spectrum and pathological findings in Schwann cells and axons. <i>Journal of the Peripheral Nervous System</i> , 2015, 20, 52-59.	1.4	21
1173	<i>FOXG1</i> Mutation is a Low-Incidence Genetic Cause in Atypical Rett Syndrome. <i>Child Neurology Open</i> , 2015, 2, 2329048X1456815.	0.5	5
1174	Integrative network-based approach identifies key genetic elements in breast invasive carcinoma. <i>BMC Genomics</i> , 2015, 16, S2.	1.2	30
1175	Comparison of GENCODE and RefSeq gene annotation and the impact of reference geneset on variant effect prediction. <i>BMC Genomics</i> , 2015, 16, S2.	1.2	80
1176	Insights from GWAS: emerging landscape of mechanisms underlying complex trait disease. <i>BMC Genomics</i> , 2015, 16, S4.	1.2	16
1178	Detection and analysis of disease-associated single nucleotide polymorphism influencing post-translational modification. <i>BMC Medical Genomics</i> , 2015, 8, S7.	0.7	36
1179	Crowdsourced direct-to-consumer genomic analysis of a family quartet. <i>BMC Genomics</i> , 2015, 16, 910.	1.2	20
1180	Next-generation-sequencing-based identification of familial hypercholesterolemia-related mutations in subjects with increased LDLâ€“C levels in a latvian population. <i>BMC Medical Genetics</i> , 2015, 16, 86.	2.1	21
1181	Establishing disease causality for a novel gene variant in familial dilated cardiomyopathy using a functional in-vitro assay of regulated thin filaments and human cardiac myosin. <i>BMC Medical Genetics</i> , 2015, 16, 97.	2.1	4
1182	Adult-onset autosomal dominant spastic paraplegia linked to a GTPase-effector domain mutation of dynamin 2. <i>BMC Neurology</i> , 2015, 15, 223.	0.8	39
1183	Whole exome sequencing of microdissected splenic marginal zone lymphoma: a study to discover novel tumor-specific mutations. <i>BMC Cancer</i> , 2015, 15, 773.	1.1	33
1184	Somatic loss of function mutations in neurofibromin 1 and MYC associated factor X genes identified by exome-wide sequencing in a wild-type GIST case. <i>BMC Cancer</i> , 2015, 15, 887.	1.1	30
1185	Defects in fatty acid amide hydrolase 2 in a male with neurologic and psychiatric symptoms. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 38.	1.2	19

#	ARTICLE	IF	CITATIONS
1186	A hypomorphic BMPR1B mutation causes du Pan acromesomelic dysplasia. Orphanet Journal of Rare Diseases, 2015, 10, 84.	1.2	18
1187	Next-generation sequencing applied to a large French cone and cone-rod dystrophy cohort: mutation spectrum and new genotype-phenotype correlation. Orphanet Journal of Rare Diseases, 2015, 10, 85.	1.2	79
1188	Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 128.	1.2	46
1189	Phenotype-driven strategies for exome prioritization of human Mendelian disease genes. Genome Medicine, 2015, 7, 81.	3.6	97
1190	Identifying Children With Poor Cochlear Implantation Outcomes Using Massively Parallel Sequencing. Medicine (United States), 2015, 94, e1073.	0.4	50
1191	An Interdomain <i>KCNH2</i> Mutation Produces an Intermediate Long QT Syndrome. Human Mutation, 2015, 36, 764-773.	1.1	4
1192	Performance of In Silico Tools for the Evaluation of <i>UGT1A1</i> Missense Variants. Human Mutation, 2015, 36, 1215-1225.	1.1	21
1193	Trans-ethnic Meta-Analysis Identifies Common and Rare Variants Associated with Hepatocyte Growth Factor Levels in the Multi-ethnic Study of Atherosclerosis (MESA). Annals of Human Genetics, 2015, 79, 264-274.	0.3	13
1194	<i>SLC1A4</i> mutations cause a novel disorder of intellectual disability, progressive microcephaly, spasticity and thin corpus callosum. Clinical Genetics, 2015, 88, 327-335.	1.0	49
1195	Genetic analysis should be included in clinical practice when screening for antithrombin deficiency. Thrombosis and Haemostasis, 2015, 113, 262-271.	1.8	29
1196	Atypical haemolytic uraemic syndrome in a Japanese patient with DGKE genetic mutations. Thrombosis and Haemostasis, 2015, 114, 862-863.	1.8	24
1197	Spondyloepiphyseal dysplasia congenita caused by double heterozygous mutations in <i>COL2A1</i> . American Journal of Medical Genetics, Part A, 2015, 167, 1578-1581.	0.7	5
1198	Exome analysis of a family with Wolff-Parkinson-White syndrome identifies a novel disease locus. American Journal of Medical Genetics, Part A, 2015, 167, 2975-2984.	0.7	17
1199	Whole-Genome Sequencing and Integrative Genomic Analysis Approach on Two 22q11.2 Deletion Syndrome Family Trios for Genotype to Phenotype Correlations. Human Mutation, 2015, 36, 797-807.	1.1	16
1200	Functional Analysis of BARD1 Missense Variants in Homology-Directed Repair of DNA Double Strand Breaks. Human Mutation, 2015, 36, 1205-1214.	1.1	27
1201	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. Human Mutation, 2015, 36, 1113-1127.	1.1	185
1202	Improved inherited peripheral neuropathy genetic diagnosis by whole-exome sequencing. Molecular Genetics & Genomic Medicine, 2015, 3, 143-154.	0.6	59
1203	Cylindrical spirals associated with severe congenital muscle weakness and epileptic encephalopathy. Muscle and Nerve, 2015, 52, 895-899.	1.0	5

#	ARTICLE	IF	CITATIONS
1204	<i>In silico</i> identification and three-dimensional modelling of the missense mutation in <i>ADAMTS2</i> in a sheep flock with dermatosparaxis. <i>Veterinary Dermatology</i> , 2015, 26, 49.	0.4	9
1205	<i>In Silico</i> Prediction of the Effects of Mutations in the Human Mevalonate Kinase Gene: Towards a Predictive Framework for Mevalonate Kinase Deficiency. <i>Annals of Human Genetics</i> , 2015, 79, 451-459.	0.3	21
1206	Twenty-One Novel EGFR Kinase Domain variants in Patients with Nonsmall Cell Lung Cancer. <i>Annals of Human Genetics</i> , 2015, 79, 385-393.	0.3	2
1207	<i>CAV3</i> gene sequence variations: National Genome Database and clinics. <i>Acta Neurologica Scandinavica</i> , 2015, 132, 185-190.	1.0	4
1208	Genetic variation in the human cytochrome P450 supergene family. <i>Pharmacogenetics and Genomics</i> , 2015, 25, 584-594.	0.7	127
1209	Genetic loci that regulate ectopic calcification in response to knee trauma in LG/J by SM/J advanced intercross mice. <i>Journal of Orthopaedic Research</i> , 2015, 33, 1412-1423.	1.2	30
1210	Genetic Heterogeneity and Clinical Variability in Musculocontractural Ehlers-Danlos Syndrome Caused by Impaired Dermatan Sulfate Biosynthesis. <i>Human Mutation</i> , 2015, 36, 535-547.	1.1	65
1211	Genomic diagnosis by whole genome sequencing in a Korean family with atypical progeroid syndrome. <i>Journal of Dermatology</i> , 2015, 42, 1149-1152.	0.6	4
1212	The <i>b^c</i> allele of <i>TYRP1</i> is causative for the recessive brown (liver) colour in German Shepherd dogs. <i>Animal Genetics</i> , 2015, 46, 588-589.	0.6	9
1213	Metastatic pheochromocytoma in a 23-year-old woman with an unclassified variant in the von Hippel Lindau disease gene: how can the pathogenicity of this variant be determined?. <i>Clinical Endocrinology</i> , 2015, 83, 15-19.	1.2	3
1214	Detection of novel germline mutations for breast cancer in non- <i>BRCA1/2</i> families. <i>FEBS Journal</i> , 2015, 282, 3424-3437.	2.2	50
1215	Whole exome sequencing identifies rare protein-coding variants in Behçet's disease. <i>Arthritis and Rheumatology</i> , 2015, 68, n/a-n/a.	2.9	9
1216	A frequent hypofunctional IRAK2 variant is associated with reduced spontaneous hepatitis C virus clearance. <i>Hepatology</i> , 2015, 62, 1375-1387.	3.6	25
1217	Alström Syndrome: Mutation Spectrum of <i>ALMS1</i> . <i>Human Mutation</i> , 2015, 36, 660-668.	1.1	117
1218	<i>CHCHD10</i> variant p.(Gly66Val) causes axonal Charcot-Marie-Tooth disease. <i>Neurology: Genetics</i> , 2015, 1, e1.	0.9	62
1219	Maxillary carcinosarcoma: Identification of a novel <i>MET</i> mutation in both carcinomatous and sarcomatous components through next generation sequencing. <i>Head and Neck</i> , 2015, 37, E179-85.	0.9	10
1220	Novel Homozygous Mutation of the Internal Translation Initiation Start Site of <i>VHL</i> is Exclusively Associated with Erythrocytosis: Indications for Distinct Functional Roles of von Hippel-Lindau Tumor Suppressor Isoforms. <i>Human Mutation</i> , 2015, 36, 1039-1042.	1.1	8
1221	Management of Incidental Findings in Clinical Genomic Sequencing. <i>Current Protocols in Human Genetics</i> , 2015, 87, 9.23.1-9.23.16.	3.5	12

#	ARTICLE	IF	CITATIONS
1222	Mutations in <i>FARS2</i> and non-fatal mitochondrial dysfunction in two siblings. American Journal of Medical Genetics, Part A, 2015, 167, 1147-1151.	0.7	33
1223	<i>SFTPA2</i> Mutations in Familial and Sporadic Idiopathic Interstitial Pneumonia. American Journal of Respiratory and Critical Care Medicine, 2015, 192, 1249-1252.	2.5	72
1224	Two candidate genes for two quantitative trait loci epistatically attenuate hypertension in a novel pathway. Journal of Hypertension, 2015, 33, 1791-1801.	0.3	12
1225	RASA3 is a critical inhibitor of RAP1-dependent platelet activation. Journal of Clinical Investigation, 2015, 125, 1419-1432.	3.9	113
1226	DNaseq Workflow in a Diagnostic Context and an Example of a User Friendly Implementation. BioMed Research International, 2015, 2015, 1-11.	0.9	10
1227	Associations of single nucleotide polymorphisms in the Pygo2 coding sequence with idiopathic oligospermia and azoospermia. Genetics and Molecular Research, 2015, 14, 9053-9061.	0.3	4
1228	Review Computational characterisation of cancer molecular profiles derived using next generation sequencing. Wspolczesna Onkologia, 2015, 1A, 78-91.	0.7	4
1229	Systematic 3D Screening of Amino Acid Mutations in Pharmacogenes. Current Pharmacogenomics and Personalized Medicine, 2015, 12, 209-226.	0.2	0
1230	Genetic Diagnosis via Whole Exome Sequencing in Taiwanese Patients with Hypertriglyceridemia. Journal of Atherosclerosis and Thrombosis, 2015, 22, 887-900.	0.9	6
1231	Targeted Next Generation Sequencing Identifies Novel Mutations in <i>RP1</i> as a Relatively Common Cause of Autosomal Recessive Rod-Cone Dystrophy. BioMed Research International, 2015, 2015, 1-11.	0.9	25
1233	Analysis of genetic variation and potential applications in genome-scale metabolic modeling. Frontiers in Bioengineering and Biotechnology, 2015, 3, 13.	2.0	30
1234	Efflux-mediated fluoroquinolone resistance in the multidrug-resistant <i>Pseudomonas aeruginosa</i> clinical isolate PA7: identification of a novel MexS variant involved in upregulation of the mexEF-oprN multidrug efflux operon. Frontiers in Microbiology, 2015, 6, 8.	1.5	49
1235	Whole Exome Sequencing Reveals DYSF, FKTN, and ISPD Mutations in Congenital Muscular Dystrophy Without Brain or Eye Involvement. Journal of Neuromuscular Diseases, 2015, 2, 87-92.	1.1	13
1236	Exome Sequencing on 298 Proband With Early-Onset High Myopia: Approximately One-Fourth Show Potential Pathogenic Mutations in RetNet Genes. , 2015, 56, 8365.		77
1237	Molecular Characterization of <i>FZD4</i> , <i>LRP5</i> , and <i>TSPAN12</i> in Familial Exudative Vitreoretinopathy. , 2015, 56, 5143.		46
1238	Affected Kindred Analysis of Human X Chromosome Exomes to Identify Novel X-Linked Intellectual Disability Genes. PLoS ONE, 2015, 10, e0116454.	1.1	49
1239	Exome Sequencing of 75 Individuals from Multiply Affected Coeliac Families and Large Scale Resequencing Follow Up. PLoS ONE, 2015, 10, e0116845.	1.1	8
1240	Single Nucleotide Variations in CLCN6 Identified in Patients with Benign Partial Epilepsies in Infancy and/or Febrile Seizures. PLoS ONE, 2015, 10, e0118946.	1.1	13

#	ARTICLE	IF	CITATIONS
1241	Rare Variants in MYD88, IRAK4 and IKBKG and Susceptibility to Invasive Pneumococcal Disease: A Population-Based Case-Control Study. PLoS ONE, 2015, 10, e0123532.	1.1	8
1242	Variants in Nebulin (NEB) Are Linked to the Development of Familial Primary Angle Closure Glaucoma in Basset Hounds. PLoS ONE, 2015, 10, e0126660.	1.1	21
1243	Genetic Testing of Korean Familial Hypercholesterolemia Using Whole-Exome Sequencing. PLoS ONE, 2015, 10, e0126706.	1.1	24
1244	Fine Mapping and Characterization of Candidate Genes that Control Resistance to <i>Cercospora sojina</i> K. Hara in Two Soybean Germplasm Accessions. PLoS ONE, 2015, 10, e0126753.	1.1	27
1245	The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. PLoS Genetics, 2015, 11, e1005378.	1.5	331
1246	DMD Mutations in 576 Dystrophinopathy Families: A Step Forward in Genotype-Phenotype Correlations. PLoS ONE, 2015, 10, e0135189.	1.1	109
1247	Discovery and Fine-Mapping of Glycaemic and Obesity-Related Trait Loci Using High-Density Imputation. PLoS Genetics, 2015, 11, e1005230.	1.5	77
1248	Multiple Changes of Gene Expression and Function Reveal Genomic and Phenotypic Complexity in SLE-like Disease. PLoS Genetics, 2015, 11, e1005248.	1.5	21
1249	Point Mutations in Centromeric Histone Induce Post-zygotic Incompatibility and Uniparental Inheritance. PLoS Genetics, 2015, 11, e1005494.	1.5	91
1250	Mosaic and Intronic Mutations in TSC1/TSC2 Explain the Majority of TSC Patients with No Mutation Identified by Conventional Testing. PLoS Genetics, 2015, 11, e1005637.	1.5	209
1251	Tyro3 Modulates Mertk-Associated Retinal Degeneration. PLoS Genetics, 2015, 11, e1005723.	1.5	55
1252	Atomic Insight into the Altered O6-Methylguanine-DNA Methyltransferase Protein Architecture in Gastric Cancer. PLoS ONE, 2015, 10, e0127741.	1.1	9
1253	Shared Segment Analysis and Next-Generation Sequencing Implicates the Retinoic Acid Signaling Pathway in Total Anomalous Pulmonary Venous Return (TAPVR). PLoS ONE, 2015, 10, e0131514.	1.1	16
1254	Bioinformatic Challenges in Clinical Diagnostic Application of Targeted Next Generation Sequencing: Experience from Pheochromocytoma. PLoS ONE, 2015, 10, e0133210.	1.1	11
1255	FROG - Fingerprinting Genomic Variation Ontology. PLoS ONE, 2015, 10, e0134693.	1.1	3
1256	Identification of Medically Actionable Secondary Findings in the 1000 Genomes. PLoS ONE, 2015, 10, e0135193.	1.1	74
1257	Association of the I264T Variant in the Sulfide Quinone Reductase-Like (SQRD) Gene with Osteoporosis in Korean Postmenopausal Women. PLoS ONE, 2015, 10, e0135285.	1.1	12
1258	Genetic and Informatic Analyses Implicate Kif12 as a Candidate Gene within the Mpkd2 Locus That Modulates Renal Cystic Disease Severity in the Cys1cpk Mouse. PLoS ONE, 2015, 10, e0135678.	1.1	13

#	ARTICLE	IF	CITATIONS
1259	Whole-Exome Sequencing in a South American Cohort Links ALDH1A3, FOXN1 and Retinoic Acid Regulation Pathways to Autism Spectrum Disorders. PLoS ONE, 2015, 10, e0135927.	1.1	27
1260	Targeted Sequencing of the Mitochondrial Genome of Women at High Risk of Breast Cancer without Detectable Mutations in BRCA1/2. PLoS ONE, 2015, 10, e0136192.	1.1	11
1261	Investigating the Molecular Basis of Retinal Degeneration in a Familial Cohort of Pakistani Decent by Exome Sequencing. PLoS ONE, 2015, 10, e0136561.	1.1	33
1262	Heterogeneous Mechanisms of Secondary Resistance and Clonal Selection in Sarcoma during Treatment with Nutlin. PLoS ONE, 2015, 10, e0137794.	1.1	12
1263	Prioritizing Clinically Relevant Copy Number Variation from Genetic Interactions and Gene Function Data. PLoS ONE, 2015, 10, e0139656.	1.1	9
1264	Fine Dissection of Human Mitochondrial DNA Haplogroup HV Lineages Reveals Paleolithic Signatures from European Glacial Refugia. PLoS ONE, 2015, 10, e0144391.	1.1	23
1265	Analysis of BRCA gene missense mutations. Journal of Biomedical Engineering and Informatics, 2015, 2, 91.	0.2	0
1266	Intermediate MCAD Deficiency Associated with a Novel Mutation of the ACADM Gene: c.1052C>T. Case Reports in Genetics, 2015, 2015, 1-4.	0.1	3
1267	A Novel PHEX Mutation in Japanese Patients with X-Linked Hypophosphatemic Rickets. Case Reports in Genetics, 2015, 2015, 1-5.	0.1	2
1268	Identifying Highly Penetrant Disease Causal Mutations Using Next Generation Sequencing: Guide to Whole Process. BioMed Research International, 2015, 2015, 1-16.	0.9	7
1269	Targeted Next-Generation Sequencing Reveals Hot Spots and Doubly Heterozygous Mutations in Chinese Patients with Familial Cardiomyopathy. BioMed Research International, 2015, 2015, 1-11.	0.9	11
1271	Dominant Red Coat Color in Holstein Cattle Is Associated with a Missense Mutation in the Coatmer Protein Complex, Subunit Alpha (COPA) Gene. PLoS ONE, 2015, 10, e0128969.	1.1	30
1272	Potential protein activity modifications of amino acid variants in the human transcriptome. Acta Biochimica Polonica, 2015, 62, 57-61.	0.3	0
1273	A Coding Variant of ANO10, Affecting Volume Regulation of Macrophages, Is Associated with Borrelia Seropositivity. Molecular Medicine, 2015, 21, 26-37.	1.9	49
1274	EpilepsyGene: a genetic resource for genes and mutations related to epilepsy. Nucleic Acids Research, 2015, 43, D893-D899.	6.5	71
1275	A novel truncation mutation in GJA1 associated with open angle glaucoma and microcornea in a large Chinese family. Eye, 2015, 29, 972-977.	1.1	11
1276	Association of exome sequences with plasma C-reactive protein levels in >9000 participants. Human Molecular Genetics, 2015, 24, 559-571.	1.4	36
1277	Early Onset Heart Failure, Alopecia, and Cutaneous Abnormalities Associated with a Novel Compound Heterozygous Mutation in Desmoplakin. Pediatric Dermatology, 2015, 32, 102-108.	0.5	14

#	ARTICLE	IF	CITATIONS
1278	Correlates between Models of Virulence for Mycobacterium tuberculosis among Isolates of the Central Asian Lineage: a Case for Lysozyme Resistance Testing?. Infection and Immunity, 2015, 83, 2213-2223.	1.0	2
1279	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	2.6	109
1280	AMH mutations with reduced in vitro bioactivity are related to premature ovarian insufficiency. Human Reproduction, 2015, 30, 1196-1202.	0.4	50
1281	Whole-exome sequencing identifies de novo mutation in the COL1A1 gene to underlie the severe osteogenesis imperfecta. Human Genomics, 2015, 9, 6.	1.4	10
1282	Impaired osteoblast and osteoclast function characterize the osteoporosis of Snyder - Robinson syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 27.	1.2	27
1283	Whole-exome SNP array identifies 15 new susceptibility loci for psoriasis. Nature Communications, 2015, 6, 6793.	5.8	118
1284	<i>CYP2R1</i> Mutations Impair Generation of 25-hydroxyvitamin D and Cause an Atypical Form of Vitamin D Deficiency. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1005-E1013.	1.8	94
1285	Mutated genes and driver pathways involved in myelodysplastic syndromesâ€”a transcriptome sequencing based approach. Molecular BioSystems, 2015, 11, 2158-2166.	2.9	8
1286	Germline Mutations in the <i>CDKN2B</i> Tumor Suppressor Gene Predispose to Renal Cell Carcinoma. Cancer Discovery, 2015, 5, 723-729.	7.7	88
1287	It's more than stamp collecting: how genome sequencing can unify biological research. Trends in Genetics, 2015, 31, 411-421.	2.9	37
1288	Mutation of the nuclear lamin gene <i>LMNB2</i> in progressive myoclonus epilepsy with early ataxia. Human Molecular Genetics, 2015, 24, 4483-4490.	1.4	41
1289	Recessive Osteogenesis Imperfecta Caused by Missense Mutations in SPARC. American Journal of Human Genetics, 2015, 96, 979-985.	2.6	107
1290	In Silico Post Genome-Wide Association Studies Analysis of C-Reactive Protein Loci Suggests an Important Role for Interferons. Circulation: Cardiovascular Genetics, 2015, 8, 487-497.	5.1	24
1291	An ancient protein-DNA interaction underlying metazoan sex determination. Nature Structural and Molecular Biology, 2015, 22, 442-451.	3.6	93
1292	Oncotator: Cancer Variant Annotation Tool. Human Mutation, 2015, 36, E2423-E2429.	1.1	448
1293	GRASP v2.0: an update on the Genome-Wide Repository of Associations between SNPs and phenotypes. Nucleic Acids Research, 2015, 43, D799-D804.	6.5	143
1294	Molecular Genetic Testing in Clinical Diagnostic Assessments That Demonstrate Correlations in Patients With Autosomal Recessive Inherited Retinal Dystrophy. JAMA Ophthalmology, 2015, 133, 427.	1.4	19
1295	Worm in the Eye. JAMA Ophthalmology, 2015, 133, 139.	1.4	0

#	ARTICLE	IF	CITATIONS
1296	Juvenile myelomonocytic leukemia due to a germline CBL Y371C mutation: 35-year follow-up of a large family. <i>Human Genetics</i> , 2015, 134, 775-787.	1.8	21
1297	Homozygosity for Frameshift Mutations in XYLT2 Result in a Spondylo-Ocular Syndrome with Bone Fragility, Cataracts, and Hearing Defects. <i>American Journal of Human Genetics</i> , 2015, 96, 971-978.	2.6	65
1298	Familial Mediterranean fever gene mutations in north-eastern part of Anatolia with special respect to rare mutations. <i>Gene</i> , 2015, 568, 170-175.	1.0	7
1299	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. <i>Lancet, The</i> , 2015, 385, 1305-1314.	6.3	651
1300	Deconstructing the genetic basis of spent sulphite liquor tolerance using deep sequencing of genome-shuffled yeast. <i>Biotechnology for Biofuels</i> , 2015, 8, 53.	6.2	25
1301	Exome sequencing reveals mutation in GJA1 as a cause of keratoderma-hypotrichosis-leukonychia totalis syndrome. <i>Human Molecular Genetics</i> , 2015, 24, 243-250.	1.4	44
1302	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adamsâ€“Oliver Syndrome With Variable Cardiac Anomalies. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 572-581.	5.1	84
1303	Insights into Disease-Associated Mutations in the Human Proteome through Protein Structural Analysis. <i>Structure</i> , 2015, 23, 1362-1369.	1.6	103
1304	Rare genetic variants in Tunisian Jewish patients suffering from age-related macular degeneration. <i>Journal of Medical Genetics</i> , 2015, 52, 484-492.	1.5	19
1305	Using genome-wide measures of coancestry to maintain diversity and fitness in endangered and domestic pig populations. <i>Genome Research</i> , 2015, 25, 970-981.	2.4	77
1306	Oncogenic mutation profiling in new lung cancer and mesothelioma cell lines. <i>OncoTargets and Therapy</i> , 2015, 8, 195.	1.0	4
1307	Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. <i>Human Mutation</i> , 2015, 36, 1197-1204.	1.1	161
1308	Functional consequences of transferrin receptorâ€“2 mutations causing hereditary hemochromatosis type 3. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 221-232.	0.6	19
1309	Familial cortical dysplasia type <sc>IIA</sc> caused by a germline mutation in <i><sc>DEPDC</sc>5</i>. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 575-580.	1.7	95
1310	Facilitating Collaboration in Rare Genetic Disorders Through Effective Matchmaking in DECIPHER. <i>Human Mutation</i> , 2015, 36, 941-949.	1.1	38
1311	Network-Informed Gene Ranking Tackles Genetic Heterogeneity in Exome-Sequencing Studies of Monogenic Disease. <i>Human Mutation</i> , 2015, 36, 1135-1144.	1.1	7
1312	The CXCR3-CXCL11 signaling axis mediates macrophage recruitment and dissemination of mycobacterial infection. <i>DMM Disease Models and Mechanisms</i> , 2015, 8, 253-69.	1.2	129
1313	Mutational analysis of pulmonary tumours with neuroendocrine features using targeted massive parallel sequencing: a comparison of a neglected tumour group. <i>British Journal of Cancer</i> , 2015, 113, 1704-1711.	2.9	61

#	ARTICLE	IF	CITATIONS
1314	Comparison of predicted and actual consequences of missense mutations. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E5189-98.	3.3	200
1315	OVA: integrating molecular and physical phenotype data from multiple biomedical domain ontologies with variant filtering for enhanced variant prioritization. Bioinformatics, 2015, 31, 3822-3829.	1.8	24
1316	Targeted sequencing of the Paget's disease associated 14q32 locus identifies several missense coding variants in RIN3 that predispose to Paget's disease of bone. Human Molecular Genetics, 2015, 24, 3286-3295.	1.4	29
1317	An N-terminal formyl methionine on COX 1 is required for the assembly of cytochrome c oxidase. Human Molecular Genetics, 2015, 24, 4103-4113.	1.4	22
1318	Increased burden of <i>de novo</i> predicted deleterious variants in complex congenital diaphragmatic hernia. Human Molecular Genetics, 2015, 24, 4764-4773.	1.4	65
1319	Assessing the impact of mutations found in next generation sequencing data over human signaling pathways. Nucleic Acids Research, 2015, 43, W270-W275.	6.5	16
1320	Babelomics 5.0: functional interpretation for new generations of genomic data. Nucleic Acids Research, 2015, 43, W117-W121.	6.5	114
1321	BayesPI-BAR: a new biophysical model for characterization of regulatory sequence variations. Nucleic Acids Research, 2015, 43, gkv733.	6.5	17
1322	Next generation sequencing of triple negative breast cancer to find predictors for chemotherapy response. Breast Cancer Research, 2015, 17, 134.	2.2	58
1323	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. American Journal of Human Genetics, 2015, 97, 886-893.	2.6	171
1324	Identification of Variant-Specific Functions of <i>PIK3CA</i> by Rapid Phenotyping of Rare Mutations. Cancer Research, 2015, 75, 5341-5354.	0.4	130
1325	The African Turquoise Killifish Genome Provides Insights into Evolution and Genetic Architecture of Lifespan. Cell, 2015, 163, 1539-1554.	13.5	200
1326	Sixteen new lung function signals identified through 1000 Genomes Project reference panel imputation. Nature Communications, 2015, 6, 8658.	5.8	108
1327	Copy number variation in CEP57L1 predisposes to congenital absence of bilateral ACL and PCL ligaments. Human Genomics, 2015, 9, 31.	1.4	9
1328	Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. American Journal of Human Genetics, 2015, 97, 790-800.	2.6	63
1329	Reads meet rotamers: structural biology in the age of deep sequencing. Current Opinion in Structural Biology, 2015, 35, 125-134.	2.6	6
1330	Recurrent ACADVL molecular findings in individuals with a positive newborn screen for very long chain acyl-coA dehydrogenase (VLCAD) deficiency in the United States. Molecular Genetics and Metabolism, 2015, 116, 139-145.	0.5	65
1331	Next-generation diagnostics and disease-gene discovery with the Exomiser. Nature Protocols, 2015, 10, 2004-2015.	5.5	296

#	ARTICLE	IF	CITATIONS
1332	Late onset variants in Fabry disease: Results in high risk population screenings in Argentina. <i>Molecular Genetics and Metabolism Reports</i> , 2015, 4, 19-24.	0.4	12
1333	A novel Fanconi anaemia subtype associated with a dominant-negative mutation in RAD51. <i>Nature Communications</i> , 2015, 6, 8829.	5.8	130
1334	Characterization of clinically identified mutations in NDUFV1, the flavin-binding subunit of respiratory complex I, using a yeast model system. <i>Human Molecular Genetics</i> , 2015, 24, 6350-6360.	1.4	48
1335	Exome Sequencing Identifies a c.148-1G>C Mutation of TBX5 in a Holt-Oram Family with Unusual Genotype-Phenotype Correlations. <i>Cellular Physiology and Biochemistry</i> , 2015, 37, 1066-1074.	1.1	11
1336	Mutation analysis of the genes associated with anterior segment dysgenesis, microcornea and microphthalmia in 257 patients with glaucoma. <i>International Journal of Molecular Medicine</i> , 2015, 36, 1111-1117.	1.8	20
1337	Reckoning the SIX1 mutation's effects in branchio-oto-renal syndrome – A bioinformatics approach. <i>Frontiers in Biology</i> , 2015, 10, 448-457.	0.7	0
1338	Whole genome sequencing as a means to assess pathogenic mutations in medical genetics and cancer. <i>Cellular and Molecular Life Sciences</i> , 2015, 72, 1463-1471.	2.4	20
1339	Comparison and integration of deleteriousness prediction methods for nonsynonymous SNVs in whole exome sequencing studies. <i>Human Molecular Genetics</i> , 2015, 24, 2125-2137.	1.4	892
1340	Conformational dynamics of nonsynonymous variants at protein interfaces reveals disease association. <i>Proteins: Structure, Function and Bioinformatics</i> , 2015, 83, 428-435.	1.5	30
1341	A novel mutation in TFL1 homolog affecting determinacy in cowpea (<i>Vigna unguiculata</i>). <i>Molecular Genetics and Genomics</i> , 2015, 290, 55-65.	1.0	33
1342	Gene mutations in primary tumors and corresponding patient-derived xenografts derived from non-small cell lung cancer. <i>Cancer Letters</i> , 2015, 357, 179-185.	3.2	81
1343	Lessons and implications from association studies and post-GWAS analyses of cervical cancer. <i>Trends in Genetics</i> , 2015, 31, 41-54.	2.9	45
1344	Predicting survival in head and neck squamous cell carcinoma from TP53 mutation. <i>Human Genetics</i> , 2015, 134, 497-507.	1.8	31
1345	Refinement of schizophrenia GWAS loci using methylome-wide association data. <i>Human Genetics</i> , 2015, 134, 77-87.	1.8	25
1346	De Novo Mutations in the Motor Domain of KIF1A Cause Cognitive Impairment, Spastic Paraparesis, Axonal Neuropathy, and Cerebellar Atrophy. <i>Human Mutation</i> , 2015, 36, 69-78.	1.1	114
1347	Mutations of epigenetic regulatory genes are common in thymic carcinomas. <i>Scientific Reports</i> , 2014, 4, 7336.	1.6	109
1348	Cole-Carpenter Syndrome Is Caused by a Heterozygous Missense Mutation in P4HB. <i>American Journal of Human Genetics</i> , 2015, 96, 425-431.	2.6	92
1349	CHCHD2 mutations in autosomal dominant late-onset Parkinson's disease: a genome-wide linkage and sequencing study. <i>Lancet Neurology</i> , The, 2015, 14, 274-282.	4.9	285

#	ARTICLE	IF	CITATIONS
1350	A nonsynonymous SNP in BANK1 is associated with serum LDL cholesterol levels in three Korean populations. <i>Journal of Human Genetics</i> , 2015, 60, 113-118.	1.1	6
1351	Somatic Diseases (Cancer). , 2015, , 343-360.		0
1352	Evolution of Darwin's finches and their beaks revealed by genome sequencing. <i>Nature</i> , 2015, 518, 371-375.	13.7	766
1353	Evaluation of somatic mutations in tibial pseudarthrosis samples in neurofibromatosis type 1. <i>Journal of Medical Genetics</i> , 2015, 52, 256-261.	1.5	27
1354	Exome Sequencing for the Diagnosis of 46,XY Disorders of Sex Development. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E333-E344.	1.8	172
1355	MADGiC: a model-based approach for identifying driver genes in cancer. <i>Bioinformatics</i> , 2015, 31, 1526-1535.	1.8	27
1356	Somatic Mutations and Genetic Heterogeneity at the CDKN1B Locus in Small Intestinal Neuroendocrine Tumors. <i>Annals of Surgical Oncology</i> , 2015, 22, 1428-1435.	0.7	62
1357	Mechismo: predicting the mechanistic impact of mutations and modifications on molecular interactions. <i>Nucleic Acids Research</i> , 2015, 43, e10-e10.	6.5	95
1358	Testing the burden of rare variation in arrhythmia-susceptibility genes provides new insights into molecular diagnosis for Brugada syndrome. <i>Human Molecular Genetics</i> , 2015, 24, 2757-2763.	1.4	130
1359	Kuwaiti population subgroup of nomadic Bedouin ancestry's Whole genome sequence and analysis. <i>Genomics Data</i> , 2015, 3, 116-127.	1.3	26
1360	From ventriculomegaly to severe muscular atrophy: Expansion of the clinical spectrum related to mutations in AIFM1. <i>Mitochondrion</i> , 2015, 21, 12-18.	1.6	51
1361	Loss-of-Function Mutations in CAST Cause Peeling Skin, Leukonychia, Acral Punctate Keratoses, Cheilitis, and Knuckle Pads. <i>American Journal of Human Genetics</i> , 2015, 96, 440-447.	2.6	36
1362	Recombination affects accumulation of damaging and disease-associated mutations in human populations. <i>Nature Genetics</i> , 2015, 47, 400-404.	9.4	84
1363	mit-o-matic: A Comprehensive Computational Pipeline for Clinical Evaluation of Mitochondrial Variations from Next-Generation Sequencing Datasets. <i>Human Mutation</i> , 2015, 36, 419-424.	1.1	26
1364	The aldo-keto reductases (AKRs): Overview. <i>Chemico-Biological Interactions</i> , 2015, 234, 236-246.	1.7	348
1365	SeqHBase: a big data toolset for family based sequencing data analysis. <i>Journal of Medical Genetics</i> , 2015, 52, 282-288.	1.5	17
1366	Whole-exome characterization of pancreatic neuroendocrine tumor cell lines BON-1 and QGP-1. <i>Journal of Molecular Endocrinology</i> , 2015, 54, 137-147.	1.1	83
1367	Clinical Validation of Targeted Next-Generation Sequencing for Inherited Disorders. <i>Archives of Pathology and Laboratory Medicine</i> , 2015, 139, 204-210.	1.2	41

#	ARTICLE	IF	CITATIONS
1368	Mutations in Bruton's tyrosine kinase impair IgA responses. <i>International Journal of Hematology</i> , 2015, 101, 305-313.	0.7	19
1369	PXK locus in systemic lupus erythematosus: fine mapping and functional analysis reveals novel susceptibility gene ABHD6. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, e14-e14.	0.5	24
1370	Exome sequencing reveals three novel candidate predisposition genes for diffuse gastric cancer. <i>Familial Cancer</i> , 2015, 14, 241-246.	0.9	50
1371	<i>Somatic Diseases (Cancer)</i> , 2015, , 297-319.		2
1372	Identification of six novel F9 mutations among haemophilia B patients from Macedonia and Bulgaria. <i>Haemophilia</i> , 2015, 21, e144-6.	1.0	0
1373	Gastric cancer in three relatives of a patient with a biallelic IL12RB1 mutation. <i>Familial Cancer</i> , 2015, 14, 89-94.	0.9	14
1374	Recoded organisms engineered to depend on synthetic amino acids. <i>Nature</i> , 2015, 518, 89-93.	13.7	288
1375	Comprehensive analysis of patients with Stargardt macular dystrophy reveals new genotype-phenotype correlations and unexpected diagnostic revisions. <i>Genetics in Medicine</i> , 2015, 17, 262-270.	1.1	41
1376	High variability of clinical symptoms in a Polish family with a novel THAP1 mutation. <i>International Journal of Neuroscience</i> , 2015, 125, 755-759.	0.8	8
1377	Large effect QTL explain natural phenotypic variation for the developmental timing of vegetative phase change in maize (<i>Zea mays</i> L.). <i>Theoretical and Applied Genetics</i> , 2015, 128, 529-538.	1.8	28
1378	mirTrios: an integrated pipeline for detection of de novo and rare inherited mutations from trios-based next-generation sequencing. <i>Journal of Medical Genetics</i> , 2015, 52, 275-281.	1.5	35
1379	BRF1 mutations alter RNA polymerase III-dependent transcription and cause neurodevelopmental anomalies. <i>Genome Research</i> , 2015, 25, 155-166.	2.4	85
1380	Comprehensive screening for mutations associated with colorectal cancer in unselected cases reveals penetrant and nonpenetrant mutations. <i>International Journal of Cancer</i> , 2015, 136, E559-68.	2.3	21
1381	Mutations in the intellectual disability gene KDM5C reduce protein stability and demethylase activity. <i>Human Molecular Genetics</i> , 2015, 24, 2861-2872.	1.4	69
1382	Next-Generation Sequencing and Novel Variant Determination in a Cohort of 92 Familial Exudative Vitreoretinopathy Patients. , 2015, 56, 1937.		84
1383	Whole exome sequencing reveals recurrent mutations in BRCA2 and FAT genes in acinar cell carcinomas of the pancreas. <i>Scientific Reports</i> , 2015, 5, 8829.	1.6	73
1384	Variability in pathogenicity prediction programs: impact on clinical diagnostics. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 99-110.	0.6	44
1385	Animal and in silico models for the study of sarcomeric cardiomyopathies. <i>Cardiovascular Research</i> , 2015, 105, 439-448.	1.8	45

#	ARTICLE	IF	CITATIONS
1386	Targeted Next-Generation Sequencing Improves the Diagnosis of Autosomal Dominant Retinitis Pigmentosa in Spanish Patients. , 2015, 56, 2173.		44
1387	X-linked myotubular myopathy in Rottweiler dogs is caused by a missense mutation in Exon 11 of the MTM1 gene. Skeletal Muscle, 2015, 5, 1.	1.9	46
1388	IGSF1 variants in boys with familial delayed puberty. European Journal of Pediatrics, 2015, 174, 687-692.	1.3	19
1389	MAP4-Dependent Regulation of Microtubule Formation Affects Centrosome, Cilia, and Golgi Architecture as a Central Mechanism in Growth Regulation. Human Mutation, 2015, 36, 87-97.	1.1	21
1390	A new compound heterozygous mutation in GJB2 causes nonsyndromic hearing loss in a consanguineous Iranian family. International Journal of Pediatric Otorhinolaryngology, 2015, 79, 553-556.	0.4	5
1391	Driver Gene Mutations of Non-Small-Cell Lung Cancer are Rare in Primary Carcinoids of the Lung: NGS Study by Ion Torrent. Lung, 2015, 193, 303-308.	1.4	25
1392	Alterations of DNA repair genes in the NCI-60 cell lines and their predictive value for anticancer drug activity. DNA Repair, 2015, 28, 107-115.	1.3	55
1393	Bayesian models for syndrome- and gene-specific probabilities of novel variant pathogenicity. Genome Medicine, 2015, 7, 5.	3.6	22
1394	Sequence and analysis of a whole genome from Kuwaiti population subgroup of Persian ancestry. BMC Genomics, 2015, 16, 92.	1.2	34
1395	Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in Medicine, 2015, 17, 405-424.	1.1	20,455
1396	Targeted Next-generation Sequencing Reveals Novel EYS Mutations in Chinese Families with Autosomal Recessive Retinitis Pigmentosa. Scientific Reports, 2015, 5, 8927.	1.6	28
1397	Mutations in DYNC2L1 disrupt cilia function and cause short rib polydactyly syndrome. Nature Communications, 2015, 6, 7092.	5.8	79
1398	Mutational Spectrum and Clinical Features of Patients With <i>ACTG1</i> Mutations Identified by Massively Parallel DNA Sequencing. Annals of Otolaryngology, Rhinology and Laryngology, 2015, 124, 84S-93S.	0.6	23
1399	Novel scripts for improved annotation and selection of variants from whole exome sequencing in cancer research. MethodsX, 2015, 2, 145-153.	0.7	5
1400	A novel OPA1 mutation causing variable age of onset autosomal dominant optic atrophy plus in an Australian family. Journal of Neurology, 2015, 262, 2323-2328.	1.8	10
1401	<i>SNiPA</i> : an interactive, genetic variant-centered annotation browser. Bioinformatics, 2015, 31, 1334-1336.	1.8	273
1402	Genetic epidemiology, prevalence, and genotype-phenotype correlations in the Swedish population with osteogenesis imperfecta. European Journal of Human Genetics, 2015, 23, 1042-1050.	1.4	126
1403	Genotype-Phenotype Correlations in Ornithine Transcarbamylase Deficiency: A Mutation Update. Journal of Genetics and Genomics, 2015, 42, 181-194.	1.7	111

#	ARTICLE	IF	CITATIONS
1404	Using large-scale genomics data to identify driver mutations in lung cancer: methods and challenges. <i>Pharmacogenomics</i> , 2015, 16, 1149-1160.	0.6	15
1405	Loss of function of PGAP1 as a cause of severe encephalopathy identified by Whole Exome Sequencing: Lessons of the bioinformatics pipeline. <i>Molecular and Cellular Probes</i> , 2015, 29, 323-329.	0.9	24
1406	A novel type of rhizomelic chondrodysplasia punctata, RCDP5, is caused by loss of the PEX5 long isoform. <i>Human Molecular Genetics</i> , 2015, 24, 5845-5854.	1.4	73
1407	First Reported Case of Unexpected Response to an Epidermal Growth Factor Receptor Tyrosine Kinase Inhibitor in the I744M Uncommon EGFR Mutation. <i>Clinical Lung Cancer</i> , 2015, 16, e259-e261.	1.1	5
1408	A rule-based expert system for inferring functional annotation. <i>Applied Soft Computing Journal</i> , 2015, 35, 373-385.	4.1	5
1409	Targeted Sequencing in Chromosome 17q Linkage Region Identifies Familial Glioma Candidates in the Gliogene Consortium. <i>Scientific Reports</i> , 2015, 5, 8278.	1.6	22
1410	Whole exome sequencing in extended families with autism spectrum disorder implicates four candidate genes. <i>Human Genetics</i> , 2015, 134, 1055-1068.	1.8	49
1411	Massive parallel sequencing applied to the molecular autopsy in sudden cardiac death in the young. <i>Forensic Science International: Genetics</i> , 2015, 18, 160-170.	1.6	37
1412	Utilizing Multiple in Silico Analyses to Identify Putative Causal SCN5A Variants in Brugada Syndrome. <i>Scientific Reports</i> , 2014, 4, 3850.	1.6	21
1413	The genetics of human infertility by functional interrogation of SNPs in mice. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 10431-10436.	3.3	62
1414	De novo missense mutations in the NAA10 gene cause severe non-syndromic developmental delay in males and females. <i>European Journal of Human Genetics</i> , 2015, 23, 602-609.	1.4	72
1415	Analyses of the mitochondrial mutations in the Chinese patients with sporadic Creutzfeldt-Jakob disease. <i>European Journal of Human Genetics</i> , 2015, 23, 86-91.	1.4	12
1416	Efficiency of Exome Sequencing for the Molecular Diagnosis of Pseudoxanthoma Elasticum. <i>Journal of Investigative Dermatology</i> , 2015, 135, 992-998.	0.3	25
1417	Analysis of exome sequence in 604 trios for recessive genotypes in schizophrenia. <i>Translational Psychiatry</i> , 2015, 5, e607-e607.	2.4	35
1418	Association of PALB2 sequence variants with the risk of familial and early-onset breast cancer in a South-American population. <i>BMC Cancer</i> , 2015, 15, 30.	1.1	18
1419	Functional Characterization of Rare Variants Implicated in Susceptibility to Lone Atrial Fibrillation. <i>Circulation: Arrhythmia and Electrophysiology</i> , 2015, 8, 1095-1104.	2.1	44
1420	Non-ocular Stickler Syndrome With a Novel Mutation in <i>COL11A2</i> Diagnosed by Massively Parallel Sequencing in Japanese Hearing Loss Patients. <i>Annals of Otolaryngology and Laryngology</i> , 2015, 124, 111S-117S.	0.6	8
1421	Systematic Cell-Based Phenotyping of Missense Alleles Empowers Rare Variant Association Studies: A Case for LDLR and Myocardial Infarction. <i>PLoS Genetics</i> , 2015, 11, e1004855.	1.5	50

#	ARTICLE	IF	CITATIONS
1422	Concurrent exome-targeted next-generation sequencing and single nucleotide polymorphism array to identify the causative genetic aberrations of isolated Mayer-Rokitansky-Kuster-Hauser syndrome. <i>Human Reproduction</i> , 2015, 30, 1732-1742.	0.4	19
1423	MiR-204 is responsible for inherited retinal dystrophy associated with ocular coloboma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, E3236-45.	3.3	90
1424	Pooled Sequencing of 531 Genes in Inflammatory Bowel Disease Identifies an Associated Rare Variant in <i>BTNL2</i> and Implicates Other Immune Related Genes. <i>PLoS Genetics</i> , 2015, 11, e1004955.	1.5	59
1425	Evolutionary Constraint and Disease Associations of Post-Translational Modification Sites in Human Genomes. <i>PLoS Genetics</i> , 2015, 11, e1004919.	1.5	69
1426	College of American Pathologists' Laboratory Standards for Next-Generation Sequencing Clinical Tests. <i>Archives of Pathology and Laboratory Medicine</i> , 2015, 139, 481-493.	1.2	315
1427	R331W Missense Mutation of Oncogene <i>YAP1</i> Is a Germline Risk Allele for Lung Adenocarcinoma With Medical Actionability. <i>Journal of Clinical Oncology</i> , 2015, 33, 2303-2310.	0.8	77
1429	Lysoplex: An efficient toolkit to detect DNA sequence variations in the autophagy-lysosomal pathway. <i>Autophagy</i> , 2015, 11, 928-938.	4.3	47
1430	<i>MYOC</i> Mutations in Black South African Patients with Primary Open-angle Glaucoma: Genetic Testing and Cascade Screening. <i>Ophthalmic Genetics</i> , 2015, 36, 31-38.	0.5	8
1431	Genome-wide Association Study Identifies Shared Risk Loci Common to Two Malignancies in Golden Retrievers. <i>PLoS Genetics</i> , 2015, 11, e1004922.	1.5	66
1432	Targeted next generation sequencing application in cardiac channelopathies: Analysis of a cohort of autopsy-negative sudden unexplained deaths. <i>Forensic Science International</i> , 2015, 254, 5-11.	1.3	35
1433	An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. <i>Nature Cell Biology</i> , 2015, 17, 1074-1087.	4.6	215
1434	The Contribution of Missense Mutations in Core and Rim Residues of Protein-Protein Interfaces to Human Disease. <i>Journal of Molecular Biology</i> , 2015, 427, 2886-2898.	2.0	105
1435	Exome sequencing identifies somatic mutations of <i>DDX3X</i> in natural killer/T-cell lymphoma. <i>Nature Genetics</i> , 2015, 47, 1061-1066.	9.4	315
1436	Emergence of a New Highly Successful Acapsular Group A <i>Streptococcus</i> Clade of Genotype <i>emm</i> 89 in the United Kingdom. <i>MBio</i> , 2015, 6, e00622.	1.8	126
1437	Exome Sequencing Reveals <i>AMER1</i> as a Frequently Mutated Gene in Colorectal Cancer. <i>Clinical Cancer Research</i> , 2015, 21, 4709-4718.	3.2	52
1438	Systematic Analysis of the Genetic Variability That Impacts SUMO Conjugation and Their Involvement in Human Diseases. <i>Scientific Reports</i> , 2015, 5, 10900.	1.6	11
1439	Novel germline <i>ERCC5</i> mutations identified in a xeroderma pigmentosum complementation group G pedigree. <i>JAAD Case Reports</i> , 2015, 1, 66-70.	0.4	3
1440	Activating <i>CARD14</i> Mutations Are Associated with Generalized Pustular Psoriasis but Rarely Account for Familial Recurrence in Psoriasis Vulgaris. <i>Journal of Investigative Dermatology</i> , 2015, 135, 2964-2970.	0.3	89

#	ARTICLE	IF	CITATIONS
1441	Impact of I30T and I30M substitution in MPZ gene associated with Dejerineâ€“Sottas syndrome type B (DSSB): A molecular modeling and dynamics. <i>Journal of Theoretical Biology</i> , 2015, 382, 23-33.	0.8	12
1442	A Novel Single-Nucleotide Deletion (c.1020delA) in NSUN2 Causes Intellectual Disability in an Emirati Child. <i>Journal of Molecular Neuroscience</i> , 2015, 57, 393-399.	1.1	39
1443	A Shared Genetic Basis for Self-Limited Delayed Puberty and Idiopathic Hypogonadotropic Hypogonadism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E646-E654.	1.8	91
1444	Novel <i>POC1A</i> mutation in primordial dwarfism reveals new insights for centriole biogenesis. <i>Human Molecular Genetics</i> , 2015, 24, 5378-5387.	1.4	26
1445	Functional and in silico assessment of MAX variants of unknown significance. <i>Journal of Molecular Medicine</i> , 2015, 93, 1247-1255.	1.7	25
1446	Next generation sequencing search for uromodulin gene variants related with impaired renal function. <i>Molecular Biology Reports</i> , 2015, 42, 1353-1358.	1.0	1
1447	Beta-catenin in schizophrenia: Possibly deleterious novel mutation. <i>Psychiatry Research</i> , 2015, 228, 843-848.	1.7	30
1448	Whole genome sequencing and analysis of Swarna, a widely cultivated indica rice variety with low glycemic index. <i>Scientific Reports</i> , 2015, 5, 11303.	1.6	33
1449	Accuracy of Hereditary Diffuse Gastric Cancer Testing Criteria and Outcomes in Patients With a Germline Mutation in CDH1. <i>Gastroenterology</i> , 2015, 149, 897-906.e19.	0.6	70
1450	Detailed Hearing and Vestibular Profiles in the Patients with COCH Mutations. <i>Annals of Otology, Rhinology and Laryngology</i> , 2015, 124, 100S-110S.	0.6	25
1451	Massively Parallel DNA Sequencing Successfully Identified Seven Families With Deafness-Associated <i>MYO6</i> Mutations. <i>Annals of Otology, Rhinology and Laryngology</i> , 2015, 124, 148S-157S.	0.6	27
1452	Germinal Mosaicism in a Family With BO Syndrome. <i>Annals of Otology, Rhinology and Laryngology</i> , 2015, 124, 118S-122S.	0.6	7
1453	<i>De novo</i> point mutations in patients diagnosed with ataxic cerebral palsy. <i>Brain</i> , 2015, 138, 1817-1832.	3.7	129
1455	Mutational dichotomy in desmoplastic malignant melanoma corroborated by multigene panel analysis. <i>Modern Pathology</i> , 2015, 28, 895-903.	2.9	15
1456	Identification and Functional Characterization of G6PC2 Coding Variants Influencing Glycemic Traits Define an Effector Transcript at the G6PC2-ABCB11 Locus. <i>PLoS Genetics</i> , 2015, 11, e1004876.	1.5	95
1457	The Patients Associated With <i>TMPRSS3</i> Mutations Are Good Candidates for Electric Acoustic Stimulation. <i>Annals of Otology, Rhinology and Laryngology</i> , 2015, 124, 193S-204S.	0.6	32
1458	Novel mutations expand the clinical spectrum of <i>DYNC1H1</i> -associated spinal muscular atrophy. <i>Neurology</i> , 2015, 84, 668-679.	1.5	106
1459	Compound RYR1 heterozygosity resulting in a complex phenotype of malignant hyperthermia susceptibility and a core myopathy. <i>Neuromuscular Disorders</i> , 2015, 25, 567-576.	0.3	28

#	ARTICLE	IF	CITATIONS
1460	Mutations in the genes encoding eukaryotic translation initiation factor 2B in Japanese patients with vanishing white matter disease. <i>Brain and Development</i> , 2015, 37, 960-966.	0.6	10
1461	Cysteine-Sparing CADASIL Mutations in <i>NOTCH3</i> Show Proaggregatory Properties In Vitro. <i>Stroke</i> , 2015, 46, 786-792.	1.0	46
1462	Targeted next-generation sequencing (NGS) of nine candidate genes with custom AmpliSeq in patients and a cardiomyopathy risk group. <i>Clinica Chimica Acta</i> , 2015, 446, 132-140.	0.5	37
1463	Defining neurodegeneration on <i>G</i> by targeted genomic sequencing. <i>Annals of Neurology</i> , 2015, 77, 458-468.	2.8	63
1464	Molecular basis of primary hyperoxaluria and strategies for diagnosis. <i>Expert Opinion on Orphan Drugs</i> , 2015, 3, 663-673.	0.5	3
1465	Computational analyses and prediction of guanylin deleterious SNPs. <i>Peptides</i> , 2015, 69, 92-102.	1.2	31
1466	Germline TP53 Variants and Susceptibility to Osteosarcoma. <i>Journal of the National Cancer Institute</i> , 2015, 107, .	3.0	109
1467	Candidate colorectal cancer predisposing gene variants in Chinese early-onset and familial cases. <i>World Journal of Gastroenterology</i> , 2015, 21, 4136.	1.4	10
1468	IL36RN mutations define a severe autoinflammatory phenotype of generalized pustular psoriasis. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 135, 1067-1070.e9.	1.5	115
1469	Mutational analysis of MATR3 in Taiwanese patients with amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2015, 36, 2005.e1-2005.e4.	1.5	64
1470	Personal exposure to PM2.5, genetic variants and DNA damage: A multi-center population-based study in Chinese. <i>Toxicology Letters</i> , 2015, 235, 172-178.	0.4	34
1471	Whole-exome sequencing and its impact in hereditary hearing loss. <i>Genetical Research</i> , 2015, 97, e4.	0.3	43
1472	Novel <i>PTPRQ</i> Mutations Identified in Three Congenital Hearing Loss Patients With Various Types of Hearing Loss. <i>Annals of Otology, Rhinology and Laryngology</i> , 2015, 124, 184S-192S.	0.6	19
1473	Mutations in <i>LOXHD1</i> Gene Cause Various Types and Severities of Hearing Loss. <i>Annals of Otology, Rhinology and Laryngology</i> , 2015, 124, 135S-141S.	0.6	24
1474	Whole-exome sequencing revealed two novel mutations in Usher syndrome. <i>Gene</i> , 2015, 563, 215-218.	1.0	6
1475	Novel Mutations in <i>GRXCR1</i> at DFNB25 Lead to Progressive Hearing Loss and Dizziness. <i>Annals of Otology, Rhinology and Laryngology</i> , 2015, 124, 129S-134S.	0.6	8
1476	Genetic Variation of <i>MT-ND</i> Genes in Frontotemporal Lobar Degeneration: Biochemical Phenotype-Genotype Correlation. <i>Neurodegenerative Diseases</i> , 2015, 15, 70-80.	0.8	1
1477	Dental Abnormalities Caused by Novel Compound Heterozygous <i>CTSK</i> Mutations. <i>Journal of Dental Research</i> , 2015, 94, 674-681.	2.5	26

#	ARTICLE	IF	CITATIONS
1478	Next generation sequencing on patients with LGMD and nonspecific myopathies: Findings associated with ANO5 mutations. <i>Neuromuscular Disorders</i> , 2015, 25, 533-541.	0.3	65
1479	Characterization and review of MTHFD1 deficiency: four new patients, cellular delineation and response to folic and folinic acid treatment. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 863-872.	1.7	39
1480	A novel POLE mutation associated with cancers of colon, pancreas, ovaries and small intestine. <i>Familial Cancer</i> , 2015, 14, 437-448.	0.9	67
1481	Whole Genome Sequencing Reveals a Chromosome 9p Deletion Causing DOCK8 Deficiency in an Adult Diagnosed with Hyper IgE Syndrome Who Developed Progressive Multifocal Leukoencephalopathy. <i>Journal of Clinical Immunology</i> , 2015, 35, 92-96.	2.0	16
1482	Whole genome analysis of a Vietnamese trio. <i>Journal of Biosciences</i> , 2015, 40, 113-124.	0.5	4
1483	Association of CG Genotype at rs4950928 Promoter in CHI3L1 Gene with YKL-40 Levels and Asthma Susceptibility in North Indian Asthma Patients. <i>Indian Journal of Clinical Biochemistry</i> , 2015, 30, 403-411.	0.9	3
1484	Fundus albipunctatus: review of the literature and report of a novel RDH5 gene mutation affecting the invariant tyrosine (p.Tyr175Phe). <i>Journal of Applied Genetics</i> , 2015, 56, 317-327.	1.0	27
1485	Exome sequencing reveals novel BCS1L mutations in siblings with hearing loss and hypotrichosis. <i>Gene</i> , 2015, 566, 84-88.	1.0	7
1486	Mutations in PYCR2, Encoding Pyrroline-5-Carboxylate Reductase 2, Cause Microcephaly and Hypomyelination. <i>American Journal of Human Genetics</i> , 2015, 96, 709-719.	2.6	60
1487	Enhanced Classification of Brugada Syndrome—Associated and Long-QT Syndrome—Associated Genetic Variants in the <i>SCN5A</i> -Encoded Na ^v 1.5 Cardiac Sodium Channel. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 582-595.	5.1	87
1488	The clinical and genetic features in a cohort of mainland Chinese patients with thyrotoxic periodic paralysis. <i>BMC Neurology</i> , 2015, 15, 38.	0.8	14
1489	The genotypic and phenotypic spectrum of PIGA deficiency. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 23.	1.2	70
1490	Rare variant association studies: considerations, challenges and opportunities. <i>Genome Medicine</i> , 2015, 7, 16.	3.6	176
1491	Multiethnic Genome-Wide Association Study of Cerebral White Matter Hyperintensities on MRI. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 398-409.	5.1	162
1492	Evaluation of BRCA1/2 mutational status among German and Austrian women with triple-negative breast cancer. <i>Journal of Cancer Research and Clinical Oncology</i> , 2015, 141, 2005-2012.	1.2	23
1493	Identification of the Inflammasome <i>Nlrp1b</i> as the Candidate Gene Conferring Diabetes Risk at the <i>Idd4.1</i> Locus in the Nonobese Diabetic Mouse. <i>Journal of Immunology</i> , 2015, 194, 5663-5673.	0.4	6
1494	New Mutations in the <i>RAB28</i> Gene in 2 Spanish Families With Cone-Rod Dystrophy. <i>JAMA Ophthalmology</i> , 2015, 133, 133.	1.4	28
1495	Homozygosity mapping and sequencing identify two genes that might contribute to pointing behavior in hunting dogs. <i>Canine Genetics and Epidemiology</i> , 2015, 2, 5.	2.9	11

#	ARTICLE	IF	CITATIONS
1496	Skeletal dysplasia in a consanguineous clan from the island of Nias/Indonesia is caused by a novel mutation in B3GAT3. <i>Human Genetics</i> , 2015, 134, 691-704.	1.8	27
1497	Integrated analysis of whole-exome sequencing and transcriptome profiling in males with autism spectrum disorders. <i>Molecular Autism</i> , 2015, 6, 21.	2.6	106
1498	Mutational landscape determines sensitivity to PD-1 blockade in non-“small cell lung cancer. <i>Science</i> , 2015, 348, 124-128.	6.0	6,756
1499	Evaluation of von Willebrand factor phenotypes and genotypes in Hemophilia A patients with and without identified F8 mutations. <i>Journal of Thrombosis and Haemostasis</i> , 2015, 13, 1036-1042.	1.9	14
1500	Improved feature-based prediction of SNPs in human cytochrome P450 enzymes. <i>Interdisciplinary Sciences, Computational Life Sciences</i> , 2015, 7, 65-77.	2.2	9
1501	Mutations in the <i>MYO15A</i> Gene Are a Significant Cause of Nonsyndromic Hearing Loss. <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2015, 124, 158S-168S.	0.6	42
1502	Evaluation of 12 Myopia-Associated Genes in Chinese Patients With High Myopia. <i>Investigative Ophthalmology and Visual Science</i> , 2015, 56, 722-729.	3.3	60
1503	In Silico Analysis of <i>RET</i> Variants in Medullary Thyroid Cancer. <i>Otolaryngology - Head and Neck Surgery</i> , 2015, 152, 650-654.	1.1	2
1504	Homozygous MED25 mutation implicated in eye-“intellectual disability syndrome. <i>Human Genetics</i> , 2015, 134, 577-587.	1.8	18
1505	Longitudinal analysis of 25 sequential sample-pairs using a custom multiple myeloma mutation sequencing panel (M3P). <i>Annals of Hematology</i> , 2015, 94, 1205-1211.	0.8	47
1506	Absence of germline mutations in BAP1 in sporadic cases of malignant mesothelioma. <i>Gene</i> , 2015, 563, 103-105.	1.0	27
1507	Human ACAP2 is a homolog of <i>C. elegans</i> CNT-1 that promotes apoptosis in cancer cells. <i>Cell Cycle</i> , 2015, 14, 1771-1778.	1.3	8
1508	Systems Biology With High-Throughput Sequencing Reveals Genetic Mechanisms Underlying the Metabolic Syndrome in the Lyon Hypertensive Rat. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 316-326.	5.1	24
1509	Next Generation Sequencing in Cancer Research, Volume 2. , 2015, , .		4
1510	A Decision Support Framework for Genomically Informed Investigational Cancer Therapy. <i>Journal of the National Cancer Institute</i> , 2015, 107, .	3.0	168
1511	Analysis of consequences of non-synonymous SNP in feed conversion ratio associated TGF- β 2 receptor type 3 gene in chicken. <i>Meta Gene</i> , 2015, 4, 107-117.	0.3	18
1512	Estimating the mutation load in human genomes. <i>Nature Reviews Genetics</i> , 2015, 16, 333-343.	7.7	233
1513	PaPI: pseudo amino acid composition to score human protein-coding variants. <i>BMC Bioinformatics</i> , 2015, 16, 123.	1.2	44

#	ARTICLE	IF	CITATIONS
1514	Functional analysis of naturally occurring DCLRE1C mutations and correlation with the clinical phenotype of ARTEMIS deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 140-150.e7.	1.5	63
1515	Clinical utility of a next generation sequencing panel assay for Marfan and Marfan-like syndromes featuring aortopathy. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1747-1757.	0.7	52
1516	Recurrent and novel GLB1 mutations in India. <i>Gene</i> , 2015, 567, 173-181.	1.0	22
1517	Whole exome sequencing combined with linkage analysis identifies a novel 3â€‰%bp deletion in NR5A1. <i>European Journal of Human Genetics</i> , 2015, 23, 486-493.	1.4	27
1518	The variation game: Cracking complex genetic disorders with NGS and omics data. <i>Methods</i> , 2015, 79-80, 18-31.	1.9	22
1519	Ten new cases further delineate the syndromic intellectual disability phenotype caused by mutations in DYRK1A. <i>European Journal of Human Genetics</i> , 2015, 23, 1482-1487.	1.4	62
1520	Genetic Basis of Common Human Disease: Insight into the Role of Missense SNPs from Genome-Wide Association Studies. <i>Journal of Molecular Biology</i> , 2015, 427, 2271-2289.	2.0	44
1521	DEOD: uncovering dominant effects of cancer-driver genes based on a partial covariance selection method. <i>Bioinformatics</i> , 2015, 31, 2452-2460.	1.8	22
1522	Identification and Mode of Inheritance of Quantitative Trait Loci for Secondary Metabolite Abundance in Tomato. <i>Plant Cell</i> , 2015, 27, 485-512.	3.1	188
1523	Nonsyndromic Hearing Loss Caused by USH1G Mutations. <i>Ear and Hearing</i> , 2015, 36, 205-211.	1.0	20
1524	Somatic <i>POLE</i> mutations cause an ultramutated giant cell high-grade glioma subtype with better prognosis. <i>Neuro-Oncology</i> , 2015, 17, 1356-1364.	0.6	94
1525	Next generation sequencing to identify novel genetic variants causative of autosomal dominant familial hypercholesterolemia associated with increased risk of coronary heart disease. <i>Gene</i> , 2015, 565, 76-84.	1.0	31
1526	ImmunoChip Analysis Identification of 6 Additional Susceptibility Loci for Crohn's Disease in Koreans. <i>Inflammatory Bowel Diseases</i> , 2015, 21, 1-7.	0.9	60
1527	Translational Research Methods: Basics of Renal Molecular Biology. , 2015, , 1-22.		0
1528	Identification of Candidate Gene Variants in Korean MODY Families by Whole-Exome Sequencing. <i>Hormone Research in Paediatrics</i> , 2015, 83, 242-251.	0.8	206
1529	A Novel Missense Mutation of Wilms' Tumor 1 Causes Autosomal Dominant FSGS. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 831-843.	3.0	45
1530	Reducing the search space for causal genetic variants with VASP. <i>Bioinformatics</i> , 2015, 31, 2377-2379.	1.8	17
1531	Variant discovery in a QTL region on chromosome 3 associated with fatness in chickens. <i>Animal Genetics</i> , 2015, 46, 141-147.	0.6	28

#	ARTICLE	IF	CITATIONS
1532	Heterozygous germline mutations in NBS1 among Korean patients with high-risk breast cancer negative for BRCA1/2 mutation. <i>Familial Cancer</i> , 2015, 14, 365-371.	0.9	3
1533	Common variants spanning <i>PLK4</i> are associated with mitotic-origin aneuploidy in human embryos. <i>Science</i> , 2015, 348, 235-238.	6.0	106
1534	PROVEAN web server: a tool to predict the functional effect of amino acid substitutions and indels. <i>Bioinformatics</i> , 2015, 31, 2745-2747.	1.8	2,033
1535	Polymorphisms of nucleotide-binding oligomerization domain 2 (NOD2) gene in miniature dachshunds with inflammatory colorectal polyps. <i>Veterinary Immunology and Immunopathology</i> , 2015, 164, 160-169.	0.5	10
1536	Mutations in TUBGCP4 Alter Microtubule Organization via the β -Tubulin Ring Complex in Autosomal-Recessive Microcephaly with Chorioretinopathy. <i>American Journal of Human Genetics</i> , 2015, 96, 666-674.	2.6	60
1537	SIPA1L3 identified by linkage analysis and whole-exome sequencing as a novel gene for autosomal recessive congenital cataract. <i>European Journal of Human Genetics</i> , 2015, 23, 1627-1633.	1.4	15
1538	Common and rare variants associated with kidney stones and biochemical traits. <i>Nature Communications</i> , 2015, 6, 7975.	5.8	117
1539	Newly identified natural high-oleate mutant from <i>Arachis hypogaea</i> L. subsp. <i>hypogaea</i> . <i>Molecular Breeding</i> , 2015, 35, 1.	1.0	16
1540	International genome-wide meta-analysis identifies new primary biliary cirrhosis risk loci and targetable pathogenic pathways. <i>Nature Communications</i> , 2015, 6, 8019.	5.8	245
1541	ClinLabGeneticist: a tool for clinical management of genetic variants from whole exome sequencing in clinical genetic laboratories. <i>Genome Medicine</i> , 2015, 7, 77.	3.6	5
1542	Incidence of Dravet Syndrome in a US Population. <i>Pediatrics</i> , 2015, 136, e1310-e1315.	1.0	178
1543	Targeted multi-gene panel testing for the diagnosis of Bardet Biedl syndrome: Identification of nine novel mutations across BBS1, BBS2, BBS4, BBS7, BBS9, BBS10 genes. <i>European Journal of Medical Genetics</i> , 2015, 58, 689-694.	0.7	37
1544	Newborn screening and the era of medical genomics. <i>Seminars in Perinatology</i> , 2015, 39, 617-622.	1.1	17
1545	A Heritable Missense Polymorphism in <i>CDKN2A</i> Confers Strong Risk of Childhood Acute Lymphoblastic Leukemia and Is Preferentially Selected during Clonal Evolution. <i>Cancer Research</i> , 2015, 75, 4884-4894.	0.4	38
1546	High-throughput screening identified inherited genetic variations in the EGFR pathway contributing to skin toxicity of EGFR inhibitors. <i>Pharmacogenomics</i> , 2015, 16, 1605-1619.	0.6	7
1547	Complete loss of the DNAJB6 G/F domain and novel missense mutations cause distal-onset DNAJB6 myopathy. <i>Acta Neuropathologica Communications</i> , 2015, 3, 44.	2.4	45
1548	A novel epileptic encephalopathy mutation in <i>KCNB1</i> disrupts Kv2.1 ion selectivity, expression, and localization. <i>Journal of General Physiology</i> , 2015, 146, 399-410.	0.9	79
1549	Mutational analysis by next generation sequencing of gastric type dysplasia occurring in hyperplastic polyps of the stomach. <i>Experimental and Molecular Pathology</i> , 2015, 99, 468-473.	0.9	14

#	ARTICLE	IF	CITATIONS
1550	Mutation in CEP63 co-segregating with developmental dyslexia in a Swedish family. <i>Human Genetics</i> , 2015, 134, 1239-1248.	1.8	23
1551	Genome-wide association study of toxic metals and trace elements reveals novel associations. <i>Human Molecular Genetics</i> , 2015, 24, 4739-4745.	1.4	104
1552	GATA2 deficiency in children and adults with severe pulmonary alveolar proteinosis and hematologic disorders. <i>BMC Pulmonary Medicine</i> , 2015, 15, 87.	0.8	63
1553	Bent spine syndrome as an initial manifestation of late-onset multiple acyl-CoA dehydrogenase deficiency: a case report and literature review. <i>BMC Neurology</i> , 2015, 15, 114.	0.8	17
1554	The human gene damage index as a gene-level approach to prioritizing exome variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 13615-13620.	3.3	213
1555	X-linked Recessive Distal Myopathy With Hypertrophic Cardiomyopathy Caused by a Novel Mutation in the FHL1 Gene. <i>Journal of Child Neurology</i> , 2015, 30, 1211-1217.	0.7	12
1556	Congenital Visual Impairment and Progressive Microcephaly Due to Lysyl-tRNA Synthetase (<i>KARS</i>) Mutations. <i>Journal of Child Neurology</i> , 2015, 30, 1037-1043.	0.7	47
1557	Heterozygous <i>RTEL1</i> mutations are associated with familial pulmonary fibrosis. <i>European Respiratory Journal</i> , 2015, 46, 474-485.	3.1	135
1558	Investigation of androgen receptor gene mutations in a series of 21 patients with 46,XY disorders of sex development. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2015, 28, 1257-63.	0.4	11
1559	Identification of mutations, genotype-phenotype correlation and prenatal diagnosis of maple syrup urine disease in Indian patients. <i>European Journal of Medical Genetics</i> , 2015, 58, 471-478.	0.7	25
1560	Unusual clinical expression and long survival of a pseudouridylate synthase (<i>PUS1</i>) mutation into adulthood. <i>European Journal of Human Genetics</i> , 2015, 23, 880-882.	1.4	20
1561	Clinicopathological significance of somatic <i>RNF43</i> mutation and aberrant expression of ring finger protein 43 in intraductal papillary mucinous neoplasms of the pancreas. <i>Modern Pathology</i> , 2015, 28, 261-267.	2.9	64
1562	Nature and nurture: a case of transcending haematological pre-malignancies in a pair of monozygotic twins adding possible clues on the pathogenesis of cell proliferations. <i>British Journal of Haematology</i> , 2015, 169, 391-400.	1.2	4
1563	Calcification of joints and arteries: second report with novel <i>NT5E</i> mutations and expansion of the phenotype. <i>Journal of Human Genetics</i> , 2015, 60, 561-564.	1.1	25
1564	<i>ABCC8</i> R1420H Loss-of-Function Variant in a Southwest American Indian Community: Association With Increased Birth Weight and Doubled Risk of Type 2 Diabetes. <i>Diabetes</i> , 2015, 64, 4322-4332.	0.3	50
1565	Application of whole genome and RNA sequencing to investigate the genomic landscape of common variable immunodeficiency disorders. <i>Clinical Immunology</i> , 2015, 160, 301-314.	1.4	100
1566	Enhanced utility of family-centered diagnostic exome sequencing with inheritance model-based analysis: results from 500 unselected families with undiagnosed genetic conditions. <i>Genetics in Medicine</i> , 2015, 17, 578-586.	1.1	401
1567	Genotype alone does not predict the clinical course of <i>SFTPC</i> deficiency in paediatric patients. <i>European Respiratory Journal</i> , 2015, 46, 197-206.	3.1	72

#	ARTICLE	IF	CITATIONS
1568	<i>DCLRE1C</i> (ARTEMIS) mutations causing phenotypes ranging from atypical severe combined immunodeficiency to mere antibody deficiency. <i>Human Molecular Genetics</i> , 2015, 24, 7361-7372.	1.4	72
1569	Mutation screening of <i>DUOX2</i> in Chinese patients with congenital hypothyroidism. <i>Journal of Endocrinological Investigation</i> , 2015, 38, 1219-1224.	1.8	29
1570	Whole-exome sequencing of a rare case of familial childhood acute lymphoblastic leukemia reveals putative predisposing mutations in Fanconi anemia genes. <i>BMC Cancer</i> , 2015, 15, 539.	1.1	30
1571	Molecular Characterization of <i>QDPR</i> Gene in Iranian Families with BH4 Deficiency: Reporting Novel and Recurrent Mutations. <i>JIMD Reports</i> , 2015, 21, 123-128.	0.7	8
1572	Genetic Variants That Predispose to DNA Double-Strand Breaks in Lymphocytes From a Subset of Patients With Familial Colorectal Carcinomas. <i>Gastroenterology</i> , 2015, 149, 1872-1883.e9.	0.6	31
1573	Identification of hub genes and their SNP analysis in West Nile virus infection for designing therapeutic methodologies using RNA-Seq data. <i>Genes and Genomics</i> , 2015, 37, 679-691.	0.5	1
1574	An Updated Collection of Sequence Barcoded Temperature-Sensitive Alleles of Yeast Essential Genes. <i>G3: Genes, Genomes, Genetics</i> , 2015, 5, 1879-1887.	0.8	38
1575	Brachydactyly Type C patient with compound heterozygosity for p.Gly319Val and p.Ile358Thr variants in the <i>GDF5</i> proregion: benign variants or mutations?. <i>Journal of Human Genetics</i> , 2015, 60, 419-425.	1.1	4
1576	Using whole-genome sequences of the <i>LG/J</i> and <i>SM/J</i> inbred mouse strains to prioritize quantitative trait genes and nucleotides. <i>BMC Genomics</i> , 2015, 16, 415.	1.2	31
1577	A systematic screening to identify <i>de novo</i> mutations causing sporadic early-onset Parkinson's disease. <i>Human Molecular Genetics</i> , 2015, 24, 6711-6720.	1.4	59
1578	Molecular diagnosis of hypophosphatasia and differential diagnosis by targeted Next Generation Sequencing. <i>Molecular Genetics and Metabolism</i> , 2015, 116, 215-220.	0.5	54
1579	Impaired <i>PIEZO1</i> function in patients with a novel autosomal recessive congenital lymphatic dysplasia. <i>Nature Communications</i> , 2015, 6, 8329.	5.8	239
1580	Evidence for Adaptation to the Tibetan Plateau Inferred from Tibetan Loach Transcriptomes. <i>Genome Biology and Evolution</i> , 2015, 7, 2970-2982.	1.1	70
1581	The diagnostic application of targeted re-sequencing in Korean patients with retinitis pigmentosa. <i>BMC Genomics</i> , 2015, 16, 515.	1.2	23
1582	Meta-analysis of shared genetic architecture across ten pediatric autoimmune diseases. <i>Nature Medicine</i> , 2015, 21, 1018-1027.	15.2	212
1583	Mutation of <i>Rv2887</i> , a <i>marR</i> -Like Gene, Confers <i>Mycobacterium tuberculosis</i> Resistance to an Imidazopyridine-Based Agent. <i>Antimicrobial Agents and Chemotherapy</i> , 2015, 59, 6873-6881.	1.4	25
1584	Structural modeling and in silico analysis of non-synonymous single nucleotide polymorphisms of human 3 β -hydroxysteroid dehydrogenase type 2. <i>Meta Gene</i> , 2015, 5, 162-172.	0.3	37
1585	Novel <i>TBK1</i> truncating mutation in a familial amyotrophic lateral sclerosis patient of Chinese origin. <i>Neurobiology of Aging</i> , 2015, 36, 3334.e1-3334.e5.	1.5	35

#	ARTICLE	IF	CITATIONS
1586	Mutations in apoptosis-inducing factor cause X-linked recessive auditory neuropathy spectrum disorder. <i>Journal of Medical Genetics</i> , 2015, 52, 523-531.	1.5	92
1587	Identification of hub glyco genes and their nsSNP analysis from mouse RNA-Seq data. <i>Gene</i> , 2015, 574, 235-246.	1.0	2
1588	Proteins linked to autosomal dominant and autosomal recessive disorders harbor characteristic rare missense mutation distribution patterns. <i>Human Molecular Genetics</i> , 2015, 24, 5995-6002.	1.4	40
1589	MTHFR-Ala222Val and male infertility: a study in Iranian men, an updated meta-analysis and an in silico-analysis. <i>Reproductive BioMedicine Online</i> , 2015, 31, 668-680.	1.1	39
1590	Prevalence of PALB2 mutations in Australian familial breast cancer cases and controls. <i>Breast Cancer Research</i> , 2015, 17, 111.	2.2	36
1591	Defects in Nicotinamide-adenine Dinucleotide Phosphate Oxidase Genes NOX1 and DUOX2 in Very Early Onset Inflammatory Bowel Disease. <i>Cellular and Molecular Gastroenterology and Hepatology</i> , 2015, 1, 489-502.	2.3	127
1592	Next-generation sequencing: hype and hope for development of personalized radiation therapy?. <i>Radiation Oncology</i> , 2015, 10, 183.	1.2	9
1593	Assessing Rare Variation in Complex Traits. , 2015, , .		6
1594	The development of rapid and accurate screening test for RET hotspot somatic and germline mutations in MEN2 syndromes. <i>Experimental and Molecular Pathology</i> , 2015, 99, 416-425.	0.9	7
1595	Can the impact of human genetic variations be predicted?. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 11426-11427.	3.3	26
1596	Functional classification of 15 million SNPs detected from diverse chicken populations. <i>DNA Research</i> , 2015, 22, 205-217.	1.5	40
1597	Homozygous mutation in the eukaryotic translation initiation factor 2alpha phosphatase gene, <i>PPP1R15B</i> , is associated with severe microcephaly, short stature and intellectual disability. <i>Human Molecular Genetics</i> , 2015, 24, 6293-6300.	1.4	36
1598	Association between polymorphisms in Interleukin-16 gene and risk of late-onset Alzheimer's disease. <i>Journal of the Neurological Sciences</i> , 2015, 358, 324-327.	0.3	13
1599	Biotinidase deficiency: Spectrum of molecular, enzymatic and clinical information from newborn screening Ontario, Canada (2007-2014). <i>Molecular Genetics and Metabolism</i> , 2015, 116, 146-151.	0.5	22
1600	Clinical Characteristics of Uveal Melanoma in Patients With Germline <i>BAP1</i> Mutations. <i>JAMA Ophthalmology</i> , 2015, 133, 881.	1.4	99
1601	Does familial breast cancer and thymoma suggest a cancer syndrome? A family perspective. <i>Gene</i> , 2015, 573, 333-337.	1.0	3
1602	Insights into the proline hydroxylase (PHD) family, molecular evolution and its impact on human health. <i>Biochimie</i> , 2015, 116, 114-124.	1.3	17
1603	The genetics of human autoimmune disease: A perspective on progress in the field and future directions. <i>Journal of Autoimmunity</i> , 2015, 64, 1-12.	3.0	80

#	ARTICLE	IF	CITATIONS
1604	Mutations on the α -2-Globin Gene That May Trigger α -Thalassemia. <i>Hemoglobin</i> , 2015, 39, 398-402.	0.4	3
1605	<i>FGFR2</i> mutation in 46,XY sex reversal with craniosynostosis. <i>Human Molecular Genetics</i> , 2015, 24, 6699-6710.	1.4	44
1607	The transcription factors Ets1 and Sox10 interact during murine melanocyte development. <i>Developmental Biology</i> , 2015, 407, 300-312.	0.9	14
1608	Current strategies for mutation detection in phenotype-driven screens utilising next generation sequencing. <i>Mammalian Genome</i> , 2015, 26, 486-500.	1.0	28
1609	Mitochondrial Involvement in Vertebrate Speciation? The Case of Mito-nuclear Genetic Divergence in Chameleons. <i>Genome Biology and Evolution</i> , 2015, 7, 3322-3336.	1.1	49
1610	Effect of Readthrough Treatment in Fibroblasts of Patients Affected by Lysosomal Diseases Caused by Premature Termination Codons. <i>Neurotherapeutics</i> , 2015, 12, 874-886.	2.1	17
1611	The contribution of rare and common variants in 30 genes to risk nicotine dependence. <i>Molecular Psychiatry</i> , 2015, 20, 1467-1478.	4.1	64
1612	Genome-wide identification of RNA editing in hepatocellular carcinoma. <i>Genomics</i> , 2015, 105, 76-82.	1.3	40
1613	Identification and molecular characterisation of a homozygous missense mutation in the ADAMTS10 gene in a patient with Weill-Marchesani syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1186-1191.	1.4	13
1614	The human splicing code reveals new insights into the genetic determinants of disease. <i>Science</i> , 2015, 347, 1254806.	6.0	1,053
1615	Adaptive evolution at immune system genes and deep pregnancy implantation in primates. <i>Genomics</i> , 2015, 105, 17-22.	1.3	6
1616	A glycome mutation map for discovery of diseases of glycosylation. <i>Glycobiology</i> , 2015, 25, 211-224.	1.3	52
1617	Recent Progress on Structural Bioinformatics Research of Cytochrome P450 and Its Impact on Drug Discovery. <i>Advances in Experimental Medicine and Biology</i> , 2015, 827, 327-339.	0.8	1
1618	Identification of Rare, Single-Nucleotide Mutations in NDE1 and Their Contributions to Schizophrenia Susceptibility. <i>Schizophrenia Bulletin</i> , 2015, 41, 744-753.	2.3	26
1619	Tryptophan to Glycine mutation in the position 116 leads to protein aggregation and decreases the stability of the LITAF protein. <i>Journal of Biomolecular Structure and Dynamics</i> , 2015, 33, 1695-1709.	2.0	6
1620	Bioinformatics for Clinical Next Generation Sequencing. <i>Clinical Chemistry</i> , 2015, 61, 124-135.	1.5	114
1621	Rectal cancer profiling identifies distinct subtypes in India based on age at onset, genetic, epigenetic and clinicopathological characteristics. <i>Molecular Carcinogenesis</i> , 2015, 54, 1786-1795.	1.3	12
1622	Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy. <i>Nature Genetics</i> , 2015, 47, 73-77.	9.4	130

#	ARTICLE	IF	CITATIONS
1623	Concerted Evolution of Life Stage Performances Signals Recent Selection on Yeast Nitrogen Use. <i>Molecular Biology and Evolution</i> , 2015, 32, 153-161.	3.5	86
1624	Advance in Structural Bioinformatics. <i>Advances in Experimental Medicine and Biology</i> , 2015, , .	0.8	6
1625	Mutational analysis of FUS gene and its structural and functional role in amyotrophic lateral sclerosis 6. <i>Journal of Biomolecular Structure and Dynamics</i> , 2015, 33, 834-844.	2.0	46
1626	Disease-Targeted Sequencing of Ion Channel Genes identifies de novo mutations in Patients with Non-Familial Brugada Syndrome. <i>Scientific Reports</i> , 2014, 4, 6733.	1.6	54
1627	Noninvasive Prenatal Testing for Wilson Disease by Use of Circulating Single-Molecule Amplification and Resequencing Technology (cSMART). <i>Clinical Chemistry</i> , 2015, 61, 172-181.	1.5	85
1628	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. <i>Nature Genetics</i> , 2015, 47, 39-46.	9.4	245
1629	Congenital hypogonadotropic hypogonadism with split hand/foot malformation: a clinical entity with a high frequency of FGFR1 mutations. <i>Genetics in Medicine</i> , 2015, 17, 651-659.	1.1	55
1630	Constitutional Disorders. , 2015, , 271-296.		0
1631	Evidence for association of the rs605059 polymorphism of <i>HSD17B1</i> gene with recurrent spontaneous abortions. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2015, 28, 2250-2253.	0.7	6
1632	Compound heterozygous mutations in RIPPLY2 associated with vertebral segmentation defects. <i>Human Molecular Genetics</i> , 2015, 24, 1234-1242.	1.4	39
1633	Genetic and bioinformatics analysis of four novel <i>GCK</i> missense variants detected in Caucasian families with <i>GCK</i> MODY phenotype. <i>Clinical Genetics</i> , 2015, 87, 440-447.	1.0	6
1634	Polymorphisms in human heat shock factor-1 and analysis of potential biological consequences. <i>Cell Stress and Chaperones</i> , 2015, 20, 47-59.	1.2	4
1635	Cancer Evolution: Mathematical Models and Computational Inference. <i>Systematic Biology</i> , 2015, 64, e1-e25.	2.7	292
1636	Targeted next-generation sequencing in the diagnosis of neurodevelopmental disorders. <i>Clinical Genetics</i> , 2015, 88, 288-292.	1.0	35
1637	Aromatase deficiency in a Chinese adult man caused by novel compound heterozygous CYP19A1 mutations: Effects of estrogen replacement therapy on the bone, lipid, liver and glucose metabolism. <i>Molecular and Cellular Endocrinology</i> , 2015, 399, 32-42.	1.6	46
1638	Linking genotypes database with locus-specific database and genotype-phenotype correlation in phenylketonuria. <i>European Journal of Human Genetics</i> , 2015, 23, 302-309.	1.4	64
1639	Genomic Applications in Pathology. , 2015, , .		1
1640	A novel mutation of the axonemal dynein heavy chain gene 5 (DNAH5) in a Japanese neonate with asplenia syndrome. <i>Medical Molecular Morphology</i> , 2015, 48, 116-122.	0.4	4

#	ARTICLE	IF	CITATIONS
1641	DNA-based Diagnosis of Uncharacterized Inherited Macrothrombocytopenias Using Next-generation Sequencing Technology with a Candidate Gene Array. , 0, , .		0
1642	Caratterizzazione della mutazione SLC26A4 c.918+2T>C e report di una nuova variante potenzialmente a rischio. Acta Otorhinolaryngologica Italica, 2016, 36, 233-238.	0.7	1
1643	The next generation of metastatic melanoma: uncovering the genetic variants for anti-BRAF therapy response. Oncotarget, 2016, 7, 25135-25149.	0.8	6
1644	APE1 polymorphic variants cause persistent genomic stress and affect cancer cell proliferation. Oncotarget, 2016, 7, 26293-26306.	0.8	27
1645	Visual Function and Central Retinal Structure in Choroideremia. , 2016, 57, OCT377.		65
1646	Next-generation sequencing reveals somatic mutations that confer exceptional response to everolimus. Oncotarget, 2016, 7, 10547-10556.	0.8	52
1647	Design and application of a target capture sequencing of exons and conserved non-coding sequences for the rat. BMC Genomics, 2016, 17, 593.	1.2	12
1648	Reevaluation of the Retinal Dystrophy Due to Recessive Alleles of <i>RGR</i> With the Discovery of a Cis-Acting Mutation in <i>CDHR1</i> . , 2016, 57, 4806.		25
1649	Homozygosity Mapping and Whole-Genome Sequencing Links a Missense Mutation in <i>POMGNT1</i> to Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 3601.		18
1650	Exploration of association between <i>EPHX1</i> and chronic obstructive pulmonary disease on the basis of combined data mining. Genetics and Molecular Research, 2016, 15, .	0.3	4
1651	Massive parallel sequencing and digital gene expression analysis reveals potential mechanisms to overcome therapy resistance in pulmonary neuroendocrine tumors. Journal of Cancer, 2016, 7, 2165-2172.	1.2	10
1652	Identification of protein-damaging mutations in 10 swine taste receptors and 191 appetite-reward genes. BMC Genomics, 2016, 17, 685.	1.2	5
1653	A blood pressure-associated variant of the <i>SLC39A8</i> gene influences cellular cadmium accumulation and toxicity. Human Molecular Genetics, 2016, 25, 4117-4126.	1.4	53
1654	The New Immortalized Uroepithelial Cell Line HBLAK Contains Defined Genetic Aberrations Typical of Early Stage Urothelial Tumors. Bladder Cancer, 2016, 2, 449-463.	0.2	34
1655	Bioinformatics Approach for Prediction of Functional Coding/Noncoding Simple Polymorphisms (SNPs/Indels) in Human <i>BRAF</i> Gene. Advances in Bioinformatics, 2016, 2016, 1-15.	5.7	10
1656	Early Infantile Epileptic Encephalopathy in an <i>STXBP1</i> Patient with Lactic Acidemia and Normal Mitochondrial Respiratory Chain Function. Case Reports in Genetics, 2016, 2016, 1-5.	0.1	6
1657	Identification of Deleterious Mutations in <i>Myostatin</i> Gene of Rohu Carp (<i>Labeo rohita</i>) Using Modeling and Molecular Dynamic Simulation Approaches. BioMed Research International, 2016, 2016, 1-10.	0.9	8
1658	A Survey of Computational Tools to Analyze and Interpret Whole Exome Sequencing Data. International Journal of Genomics, 2016, 2016, 1-16.	0.8	37

#	ARTICLE	IF	CITATIONS
1659	Molecular Characterization of a Novel Germline VHL Mutation by Extensive In Silico Analysis in an Indian Family with Von Hippel-Lindau Disease. <i>Genetics Research International</i> , 2016, 2016, 1-9.	2.0	2
1660	<i>TINF2</i> Gene Mutation in a Patient with Pulmonary Fibrosis. <i>Case Reports in Pulmonology</i> , 2016, 2016, 1-6.	0.2	22
1661	Impacts of Nonsynonymous Single Nucleotide Polymorphisms of Adiponectin Receptor 1 Gene on Corresponding Protein Stability: A Computational Approach. <i>BioMed Research International</i> , 2016, 2016, 1-12.	0.9	15
1662	Identification and Evolutionary Analysis of Potential Candidate Genes in a Human Eating Disorder. <i>BioMed Research International</i> , 2016, 2016, 1-11.	0.9	7
1663	Multiple gene sequencing for risk assessment in patients with early-onset or familial breast cancer. <i>Oncotarget</i> , 2016, 7, 8310-8320.	0.8	83
1664	Biallelic Mutations in <i>CRB1</i> Underlie Autosomal Recessive Familial Foveal Retinoschisis. , 2016, 57, 2637.		34
1665	Genome-wide analysis reveals signatures of selection for important traits in domestic sheep from different ecoregions. <i>BMC Genomics</i> , 2016, 17, 863.	1.2	67
1666	Steric Clash in the SET Domain of Histone Methyltransferase NSD1 as a Cause of Sotos Syndrome and Its Genetic Heterogeneity in a Brazilian Cohort. <i>Genes</i> , 2016, 7, 96.	1.0	9
1667	A Novel KIT INDEL Mutation in Acute Myeloid Leukemia With t(8;21)(q22;q22); RUNX1-RUNX1T1. <i>Annals of Laboratory Medicine</i> , 2016, 36, 371-374.	1.2	3
1668	The Brugada Syndrome: A Rare Arrhythmia Disorder with Complex Inheritance. <i>Frontiers in Cardiovascular Medicine</i> , 2016, 3, 9.	1.1	48
1669	Multivariate Methods for Genetic Variants Selection and Risk Prediction in Cardiovascular Diseases. <i>Frontiers in Cardiovascular Medicine</i> , 2016, 3, 17.	1.1	11
1670	Strategies for Integrated Analysis of Genetic, Epigenetic, and Gene Expression Variation in Cancer: Addressing the Challenges. <i>Frontiers in Genetics</i> , 2016, 7, 2.	1.1	23
1671	In silico Analysis Revealed High-risk Single Nucleotide Polymorphisms in Human Pentraxin-3 Gene and their Impact on Innate Immune Response against Microbial Pathogens. <i>Frontiers in Microbiology</i> , 2016, 7, 192.	1.5	18
1672	Specific MRI Abnormalities Reveal Severe Perrault Syndrome due to CLPP Defects. <i>Frontiers in Neurology</i> , 2016, 7, 203.	1.1	25
1673	Slitrk Missense Mutations Associated with Neuropsychiatric Disorders Distinctively Impair Slitrk Trafficking and Synapse Formation. <i>Frontiers in Molecular Neuroscience</i> , 2016, 9, 104.	1.4	31
1674	Association between Genetic Variants in DNA Double-Strand Break Repair Pathways and Risk of Radiation Therapy-Induced Pneumonitis and Esophagitis in Non-Small Cell Lung Cancer. <i>Cancers</i> , 2016, 8, 23.	1.7	13
1675	Novel PIGT Variant in Two Brothers: Expansion of the Multiple Congenital Anomalies-Hypotonia Seizures Syndrome 3 Phenotype. <i>Genes</i> , 2016, 7, 108.	1.0	25
1676	tarSVM: Improving the accuracy of variant calls derived from microfluidic PCR-based targeted next generation sequencing using a support vector machine. <i>BMC Bioinformatics</i> , 2016, 17, 233.	1.2	2

#	ARTICLE	IF	CITATIONS
1677	Convert your favorite protein modeling program into a mutation predictor: <i>MODICT</i> . <i>BMC Bioinformatics</i> , 2016, 17, 425.	1.2	2
1678	Whole genome sequence analysis of the TALLYHO/Jng mouse. <i>BMC Genomics</i> , 2016, 17, 907.	1.2	13
1679	A rapid NGS strategy for comprehensive molecular diagnosis of Birt-Hogg-Dubé syndrome in patients with primary spontaneous pneumothorax. <i>Respiratory Research</i> , 2016, 17, 64.	1.4	20
1680	A Multi-scale Computational Platform to Mechanistically Assess the Effect of Genetic Variation on Drug Responses in Human Erythrocyte Metabolism. <i>PLoS Computational Biology</i> , 2016, 12, e1005039.	1.5	12
1681	A Comprehensive In Silico Analysis on the Structural and Functional Impact of SNPs in the Congenital Heart Defects Associated with NKX2-5 Gene—A Molecular Dynamic Simulation Approach. <i>PLoS ONE</i> , 2016, 11, e0153999.	1.1	49
1682	Selection Transforms the Landscape of Genetic Variation Interacting with Hsp90. <i>PLoS Biology</i> , 2016, 14, e2000465.	2.6	94
1683	DNAH6 and Its Interactions with PCD Genes in Heterotaxy and Primary Ciliary Dyskinesia. <i>PLoS Genetics</i> , 2016, 12, e1005821.	1.5	92
1684	Genetic Dissection of Cardiac Remodeling in an Isoproterenol-Induced Heart Failure Mouse Model. <i>PLoS Genetics</i> , 2016, 12, e1006038.	1.5	70
1685	High Resolution Genomic Scans Reveal Genetic Architecture Controlling Alcohol Preference in Bidirectionally Selected Rat Model. <i>PLoS Genetics</i> , 2016, 12, e1006178.	1.5	22
1686	Accelerating Gene Discovery by Phenotyping Whole-Genome Sequenced Multi-mutation Strains and Using the Sequence Kernel Association Test (SKAT). <i>PLoS Genetics</i> , 2016, 12, e1006235.	1.5	22
1687	Fine Mapping of a Dravet Syndrome Modifier Locus on Mouse Chromosome 5 and Candidate Gene Analysis by RNA-Seq. <i>PLoS Genetics</i> , 2016, 12, e1006398.	1.5	76
1688	In-Silico Computing of the Most Deleterious nsSNPs in HBA1 Gene. <i>PLoS ONE</i> , 2016, 11, e0147702.	1.1	38
1689	Whole Exome Sequencing Reveals Homozygous Mutations in RAI1, OTOF, and SLC26A4 Genes Associated with Nonsyndromic Hearing Loss in Altaian Families (South Siberia). <i>PLoS ONE</i> , 2016, 11, e0153841.	1.1	13
1690	Genome Sequence Variability Predicts Drug Precautions and Withdrawals from the Market. <i>PLoS ONE</i> , 2016, 11, e0162135.	1.1	17
1691	Genomic Profile of Chronic Lymphocytic Leukemia in Korea Identified by Targeted Sequencing. <i>PLoS ONE</i> , 2016, 11, e0167641.	1.1	27
1692	A Novel HRAS Mutation Independently Contributes to Left Ventricular Hypertrophy in a Family with a Known MYH7 Mutation. <i>PLoS ONE</i> , 2016, 11, e0168501.	1.1	13
1693	Prevalence of mutations in LEP, LEPR, and MC4R genes in individuals with severe obesity. <i>Genetics and Molecular Research</i> , 2016, 15, .	0.3	24
1694	Carotenoid Derivates in Achiote (<i>Bixa orellana</i>) Seeds: Synthesis and Health Promoting Properties. <i>Frontiers in Plant Science</i> , 2016, 7, 1406.	1.7	62

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1695	Identification of a <i>Solanum pennellii</i> Chromosome 4 Fruit Flavor and Nutritional Quality-Associated Metabolite QTL. <i>Frontiers in Plant Science</i> , 2016, 7, 1671.	1.7	35
1696	A profile-based method for identifying functional divergence of orthologous genes in bacterial genomes. <i>Bioinformatics</i> , 2016, 32, 3566-3574.	1.8	25
1697	Nonsyndromic Retinal Dystrophy due to Bi-Allelic Mutations in the Ciliary Transport Gene <i>IFT140</i> . , 2016, 57, 1053.		33
1698	Interleukin 23 in IBD Pathogenesis. , 0, , .		3
1699	Identification and prediction of the consequences of nonsynonymous SNPs in glyceraldehyde 3-phosphate dehydrogenase (GAPDH) gene of zebrafish <i>Danio rerio</i> . <i>Turkish Journal of Biology</i> , 2016, 40, 43-54.	2.1	8
1700	Evidence against <i>ZNF469</i> being causative for keratoconus in Polish patients. <i>Acta Ophthalmologica</i> , 2016, 94, 289-294.	0.6	20
1701	Characterization of <i>SPATA5</i> -related encephalopathy in early childhood. <i>Clinical Genetics</i> , 2016, 90, 437-444.	1.0	20
1702	Congenital disorders of glycosylation presenting as epileptic encephalopathy with migrating partial seizures in infancy. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 1085-1091.	1.1	33
1703	Molecular diagnosis of thrombocytopenia-absent radius syndrome using next-generation sequencing. <i>International Journal of Laboratory Hematology</i> , 2016, 38, 412-418.	0.7	10
1704	Multiple post-domestication origins of <i>kabuli</i> chickpea through allelic variation in a diversification-associated transcription factor. <i>New Phytologist</i> , 2016, 211, 1440-1451.	3.5	51
1705	Cobalamin C defect-associated hemolytic uremic syndrome caused by new mutation in <i>MMACHC</i> . <i>Pediatrics International</i> , 2016, 58, 763-765.	0.2	10
1706	Germline mutations in DNA repair genes may predict neoadjuvant therapy response in triple negative breast patients. <i>Genes Chromosomes and Cancer</i> , 2016, 55, 915-924.	1.5	16
1707	Using Whole Exome Sequencing to Identify Candidate Genes With Rare Variants In Nonsyndromic Cleft Lip and Palate. <i>Genetic Epidemiology</i> , 2016, 40, 432-441.	0.6	34
1708	Super-Transactivation TP53 Variant in the Germline of a Family with Li-Fraumeni Syndrome. <i>Human Mutation</i> , 2016, 37, 889-892.	1.1	7
1709	Mutation Update for Kabuki Syndrome Genes <i>KMT2D</i> and <i>KDM6A</i> and Further Delineation of X-Linked Kabuki Syndrome Subtype 2. <i>Human Mutation</i> , 2016, 37, 847-864.	1.1	134
1710	<i>TP53</i> Variations in Human Cancers: New Lessons from the IARC TP53 Database and Genomics Data. <i>Human Mutation</i> , 2016, 37, 865-876.	1.1	589
1711	Clinical utility of next-generation sequencing in the diagnosis of hereditary haemolytic anaemias. <i>British Journal of Haematology</i> , 2016, 174, 806-814.	1.2	71
1712	Genetic association of MYH genes with hereditary hearing loss in Korea. <i>Gene</i> , 2016, 591, 177-182.	1.0	19

#	ARTICLE	IF	CITATIONS
1713	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases. <i>Journal of Human Genetics</i> , 2016, 61, 951-958.	1.1	7
1714	Biallelic <i>CACNA1A</i> mutations cause early onset epileptic encephalopathy with progressive cerebral, cerebellar, and optic nerve atrophy. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2173-2176.	0.7	65
1715	<i>CRIP1</i> exonic deletion and a novel missense mutation in a female with short stature, dysmorphic features, microcephaly, and pigmentary abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2206-2211.	0.7	16
1716	A novel <i>MED12</i> mutation: Evidence for a fourth phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2377-2382.	0.7	31
1717	Exome-based analysis of cardiac arrhythmia, respiratory control, and epilepsy genes in sudden unexpected death in epilepsy. <i>Annals of Neurology</i> , 2016, 79, 522-534.	2.8	216
1718	A G-protein Subunit-11 Loss-of-Function Mutation, Thr54Met, Causes Familial Hypocalciuric Hypercalcemia Type 2 (FHH2). <i>Journal of Bone and Mineral Research</i> , 2016, 31, 1200-1206.	3.1	40
1719	A Comprehensive Study on the Etiology of Patients Receiving Cochlear Implantation With Special Emphasis on Genetic Epidemiology. <i>Otology and Neurotology</i> , 2016, 37, e126-e134.	0.7	61
1720	Homozygous nonsense mutation in <i>SGCA</i> is a common cause of limb-girdle muscular dystrophy in Assiut, Egypt. <i>Muscle and Nerve</i> , 2016, 54, 690-695.	1.0	12
1721	Identification of germline alterations in breast cancer predisposition genes among Malaysian breast cancer patients using panel testing. <i>Clinical Genetics</i> , 2016, 90, 315-323.	1.0	12
1722	Design and application of a 23-gene panel by next-generation sequencing for inherited coagulation bleeding disorders. <i>Haemophilia</i> , 2016, 22, 590-597.	1.0	43
1723	Prediction of the impact of coding missense and nonsense single nucleotide polymorphisms on HD5 and HBD1 antibacterial activity against <i>Escherichia coli</i> . <i>Biopolymers</i> , 2016, 106, 633-644.	1.2	28
1724	Mutational Analysis on Membrane Associated Transporter Protein (MATP) and Their Structural Consequences in Oculocutaneous Albinism Type 4 (OCA4) – A Molecular Dynamics Approach. <i>Journal of Cellular Biochemistry</i> , 2016, 117, 2608-2619.	1.2	46
1725	Mutational and phenotypical spectrum of phenylalanine hydroxylase deficiency in Denmark. <i>Clinical Genetics</i> , 2016, 90, 247-251.	1.0	11
1726	<i>CLTC</i> as a clinically novel gene associated with multiple malformations and developmental delay. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 958-966.	0.7	33
1727	Whole exome sequencing reveals a C-terminal germline variant in CEBPA-associated acute myeloid leukemia: 45-year follow up of a large family. <i>Haematologica</i> , 2016, 101, 846-852.	1.7	42
1728	An in silico approach to elucidate structure based functional evolution of oxacillinase. <i>Computational Biology and Chemistry</i> , 2016, 64, 145-153.	1.1	3
1729	DNA Diagnostics of Hereditary Hearing Loss: A Targeted Resequencing Approach Combined with a Mutation Classification System. <i>Human Mutation</i> , 2016, 37, 812-819.	1.1	76
1730	Clinicopathologic Features and Germline Sequence Variants in Young Patients (≤ 40 Years Old) With Pancreatic Ductal Adenocarcinoma. <i>Pancreas</i> , 2016, 45, 1056-1061.	0.5	20

#	ARTICLE	IF	CITATIONS
1731	Analysis of <i>FOXL2</i> detects three novel mutations and an atypical phenotype of blepharophimosis-epicanthus inversus syndrome. <i>Clinical and Experimental Ophthalmology</i> , 2016, 44, 757-762.	1.3	10
1732	Can the phenotype of inherited fibrinogen disorders be predicted?. <i>Haemophilia</i> , 2016, 22, 667-675.	1.0	25
1733	Molecular Genetic Characterization of 151 Mut-Type Methylmalonic Aciduria Patients and Identification of 41 Novel Mutations in MUT. <i>Human Mutation</i> , 2016, 37, 745-754.	1.1	37
1734	The contribution of protein intrinsic disorder to understand the role of genetic variants uncovered by autism spectrum disorders exome studies. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2016, 171, 479-491.	1.1	7
1735	Genes encoding members of the JAK-STAT pathway or epigenetic regulators are recurrently mutated in T-cell prolymphocytic leukaemia. <i>British Journal of Haematology</i> , 2016, 173, 265-273.	1.2	64
1736	Integration of genome-wide association and extant brain expression QTL identifies candidate genes influencing prepulse inhibition in inbred F1 mice. <i>Genes, Brain and Behavior</i> , 2016, 15, 260-270.	1.1	6
1737	Targeted and exome sequencing identified somatic mutations in hepatocellular carcinoma. <i>Hepatology Research</i> , 2016, 46, 1145-1151.	1.8	27
1738	dbWGFP: a database and web server of human whole-genome single nucleotide variants and their functional predictions. <i>Database: the Journal of Biological Databases and Curation</i> , 2016, 2016, baw024.	1.4	27
1739	A Carbohydrate Sulfotransferase 6 (<i>CHST6</i>) gene mutation is associated with Macular Corneal Dystrophy in Labrador Retrievers. <i>Veterinary Ophthalmology</i> , 2016, 19, 488-492.	0.6	6
1740	Construction of a large collection of small genome variations in French dairy and beef breeds using whole-genome sequences. <i>Genetics Selection Evolution</i> , 2016, 48, 87.	1.2	15
1741	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. <i>Genome Biology</i> , 2016, 17, 243.	3.8	241
1742	Role of Niemann-Pick Type C Disease Mutations in Dementia. <i>Journal of Alzheimer's Disease</i> , 2016, 55, 1249-1259.	1.2	24
1743	Mutations in human C2CD3 cause skeletal dysplasia and provide new insights into phenotypic and cellular consequences of altered C2CD3 function. <i>Scientific Reports</i> , 2016, 6, 24083.	1.6	30
1744	Natural resistance to Meningococcal Disease related to CFH loci: Meta-analysis of genome-wide association studies. <i>Scientific Reports</i> , 2016, 6, 35842.	1.6	33
1745	Var2GO: a web-based tool for gene variants selection. <i>BMC Bioinformatics</i> , 2016, 17, 376.	1.2	7
1746	Cancer genes discovery based on integrating transcriptomic data and the impact of gene length. , ,		0
1747	Novel gene function revealed by mouse mutagenesis screens for models of age-related disease. <i>Nature Communications</i> , 2016, 7, 12444.	5.8	79
1748	10 IGSF mutations dysregulate gonadotropin-releasing hormone neuronal migration resulting in delayed puberty. <i>EMBO Molecular Medicine</i> , 2016, 8, 626-642.	3.3	109

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1749	Molecular findings from 537 individuals with inherited retinal disease. <i>Journal of Medical Genetics</i> , 2016, 53, 761-767.	1.5	135
1750	A comprehensive in silico analysis of non-synonymous and regulatory SNPs of human MBL2 gene. <i>SpringerPlus</i> , 2016, 5, 811.	1.2	47
1751	A Nonsynonymous FCER1B SNP is Associated with Risk of Developing Allergic Rhinitis and with IgE Levels. <i>Scientific Reports</i> , 2016, 6, 19724.	1.6	14
1752	Association of HLA class I and II genes with cutaneous leishmaniasis: a case control study from Sri Lanka and a systematic review. <i>BMC Infectious Diseases</i> , 2016, 16, 292.	1.3	17
1753	Understanding rare and common diseases in the context of human evolution. <i>Genome Biology</i> , 2016, 17, 225.	3.8	76
1754	Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. <i>Science</i> , 2016, 354, .	6.0	464
1755	Transcriptome analysis revealed chimeric RNAs, single nucleotide polymorphisms and allele-specific expression in porcine prenatal skeletal muscle. <i>Scientific Reports</i> , 2016, 6, 29039.	1.6	8
1756	High-throughput interpretation of gene structure changes in human and nonhuman resequencing data, using ACE. <i>Bioinformatics</i> , 2017, 33, 1437-1446.	1.8	2
1757	iCAGES: integrated CAnceR GEnome Score for comprehensively prioritizing driver genes in personal cancer genomes. <i>Genome Medicine</i> , 2016, 8, 135.	3.6	45
1758	MIB2variants altering NOTCH signalling result in left ventricle hypertrabeculation/non-compaction and are associated with MÃ©nÃ©trier-like gastropathy. <i>Human Molecular Genetics</i> , 2016, 26, ddw365.	1.4	7
1759	P27/CDKN1B Translational Regulators in Pituitary Tumorigenesis. <i>Hormone and Metabolic Research</i> , 2016, 48, 840-846.	0.7	12
1760	Homozygous mutation of VPS16 gene is responsible for an autosomal recessive adolescent-onset primary dystonia. <i>Scientific Reports</i> , 2016, 6, 25834.	1.6	36
1761	A proteogenomic approach for protein-level evidence of genomic variants in cancer cells. <i>Scientific Reports</i> , 2016, 6, 35305.	1.6	14
1762	Whole Genome Sequencing Identifies a Missense Mutation in HES7 Associated with Short Tails in Asian Domestic Cats. <i>Scientific Reports</i> , 2016, 6, 31583.	1.6	25
1763	Novel genetic risk variants for pediatric celiac disease. <i>Human Genomics</i> , 2016, 10, 34.	1.4	9
1764	Coexistence of gain-of-function JAK2 germ line mutations with JAK2V617F in polycythemia vera. <i>Blood</i> , 2016, 128, 2266-2270.	0.6	21
1765	Systems Pharmacology: An Overview. <i>AAPS Advances in the Pharmaceutical Sciences Series</i> , 2016, , 53-80.	0.2	7
1766	Purifying selection shapes the coincident SNP distribution of primate coding sequences. <i>Scientific Reports</i> , 2016, 6, 27272.	1.6	5

#	ARTICLE	IF	CITATIONS
1767	Complex inheritance in Pulmonary Arterial Hypertension patients with several mutations. Scientific Reports, 2016, 6, 33570.	1.6	15
1768	Premature termination codons in modern human genomes. Scientific Reports, 2016, 6, 22468.	1.6	8
1769	BRONCO: Biomedical entity Relation ONcology COrpus for extracting gene-variant-disease-drug relations. Database: the Journal of Biological Databases and Curation, 2016, 2016, .	1.4	29
1770	<i>SLC25A46</i> is required for mitochondrial lipid homeostasis and cristae maintenance and is responsible for Leigh syndrome. EMBO Molecular Medicine, 2016, 8, 1019-1038.	3.3	141
1771	A novel ANO3 variant identified in a 53-year-old woman presenting with hyperkinetic dysarthria, blepharospasm, hyperkinesias, and complex motorÀtics. BMC Medical Genetics, 2016, 17, 93.	2.1	14
1772	Mutational and clinical analysis of the ENG gene in patients with pulmonary arterial hypertension. BMC Genetics, 2016, 17, 72.	2.7	21
1773	Analysing the diversity of the caprine melanocortin 1 receptor (MC1R) in goats with distinct geographic origins. Small Ruminant Research, 2016, 145, 7-11.	0.6	4
1774	A genetic analysis of 23 Chinese patients with hemophilia B. Scientific Reports, 2016, 6, 25024.	1.6	3
1775	Whole exome sequencing links dental tumor to an autosomal-dominant mutation in ANO5 gene associated with gnathodiaphyseal dysplasia and muscle dystrophies. Scientific Reports, 2016, 6, 26440.	1.6	25
1776	De novo EDA mutations: Variable expression in two Egyptian families. Archives of Oral Biology, 2016, 68, 21-28.	0.8	10
1777	Assessment of pathogenicity of natural IGFALS gene variants by in silico bioinformatics tools and inÀvitro functional studies. Molecular and Cellular Endocrinology, 2016, 429, 19-28.	1.6	11
1778	GTF2E2 Mutations Destabilize the General Transcription Factor Complex TFIIIE in Individuals with DNA Repair-Proficient Trichothiodystrophy. American Journal of Human Genetics, 2016, 98, 627-642.	2.6	49
1779	Animal inference on human mitochondrial diseases. Computational Biology and Chemistry, 2016, 62, 17-28.	1.1	0
1780	IMPACT: a whole-exome sequencing analysis pipeline for integrating molecular profiles with actionable therapeutics in clinical samples. Journal of the American Medical Informatics Association: JAMIA, 2016, 23, 721-730.	2.2	38
1781	In vitro selection of miltefosine resistance in promastigotes of <i>Leishmania donovani</i> from Nepal: genomic and metabolomic characterization. Molecular Microbiology, 2016, 99, 1134-1148.	1.2	67
1782	Novel <i>TGFBI</i> mutation p.(Leu558Arg) in a lattice corneal dystrophy patient. Ophthalmic Genetics, 2016, 37, 473-474.	0.5	10
1783	Mutations in HIVEP2 are associated with developmental delay, intellectual disability, and dysmorphic features. Neurogenetics, 2016, 17, 159-164.	0.7	31
1784	A novel kinase mutation in VEGFR-1 predisposes its Î±C-helix/activation loop towards allosteric activation: Atomic insights from protein simulation. European Journal of Human Genetics, 2016, 24, 1287-1293.	1.4	4

#	ARTICLE	IF	CITATIONS
1785	Common and Rare Genetic Variation in <i>CCR2</i> , <i>CCR5</i> , or <i>CX3CR1</i> and Risk of Atherosclerotic Coronary Heart Disease and Glucometabolic Traits. <i>Circulation: Cardiovascular Genetics</i> , 2016, 9, 250-258.	5.1	20
1786	Mutations in <i>C8ORF37</i> cause Bardet Biedl syndrome (BBS21). <i>Human Molecular Genetics</i> , 2016, 25, 2283-2294.	1.4	91
1787	Nonoverlapping Clinical and Mutational Patterns in Melanomas from the Female Genital Tract and Atypical Genital Nevi. <i>Journal of Investigative Dermatology</i> , 2016, 136, 1858-1865.	0.3	27
1788	Genomic screening of <i>ABCA4</i> and array CGH analysis underline the genetic variability of Greek patients with inherited retinal diseases. <i>Meta Gene</i> , 2016, 8, 37-43.	0.3	4
1789	Lack of genomic diversity in the <i>SLC47A1</i> gene within the indigenous Xhosa population. <i>Drug Metabolism and Personalized Therapy</i> , 2016, 31, 107-114.	0.3	1
1790	Early Onset Parkinson's disease due to DJ1 mutations: An Indian study. <i>Parkinsonism and Related Disorders</i> , 2016, 32, 20-24.	1.1	18
1791	Congenital afibrinogenemia: Identification and characterization of two novel homozygous fibrinogen A α and B β chain mutations in two Tunisian families. <i>Thrombosis Research</i> , 2016, 143, 11-16.	0.8	8
1792	De novo RRAGC mutation activates mTORC1 signaling in syndromic fetal dilated cardiomyopathy. <i>Human Genetics</i> , 2016, 135, 909-917.	1.8	28
1793	Carpal tunnel syndrome: The role of collagen gene variants. <i>Gene</i> , 2016, 587, 53-58.	1.0	14
1794	The role of the melanoma gene <i>MC1R</i> in Parkinson disease and REM sleep behavior disorder. <i>Neurobiology of Aging</i> , 2016, 43, 180.e7-180.e13.	1.5	12
1795	Somatic overgrowth associated with homozygous mutations in both <i>MAN1B1</i> and <i>SEC23A</i> . <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a000737.	0.5	18
1796	Molecular Drivers of the Non-T-cell-Inflamed Tumor Microenvironment in Urothelial Bladder Cancer. <i>Cancer Immunology Research</i> , 2016, 4, 563-568.	1.6	293
1797	A novel mutation in <i>FBXL4</i> in a Norwegian child with encephalomyopathic mitochondrial DNA depletion syndrome 13. <i>European Journal of Medical Genetics</i> , 2016, 59, 342-346.	0.7	16
1798	A novel <i>GBE1</i> gene variant in a child with glycogen storage disease type IV. <i>Human Pathology</i> , 2016, 54, 152-156.	1.1	7
1799	Deep sequencing of the MHC region in the Chinese population contributes to studies of complex disease. <i>Nature Genetics</i> , 2016, 48, 740-746.	9.4	188
1800	Molecular genetics of cone-rod dystrophy in Chinese patients: New data from 61 probands and mutation overview of 163 probands. <i>Experimental Eye Research</i> , 2016, 146, 252-258.	1.2	60
1801	In Silico Screening and Molecular Dynamic Study of Nonsynonymous Single Nucleotide Polymorphisms Associated with Kidney Stones in the <i>SLC26A6</i> Gene. <i>Journal of Urology</i> , 2016, 196, 118-123.	0.2	19
1802	Identification of mutations in Korean patients with amyotrophic lateral sclerosis using multigene panel testing. <i>Neurobiology of Aging</i> , 2016, 37, 209.e9-209.e16.	1.5	25

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1803	Nucleotide polymorphisms in the bovine lymphotoxin A gene and their distribution among Bos indicus zebu cattle breeds. <i>Gene</i> , 2016, 579, 82-94.	1.0	2
1804	A Standardized DNA Variant Scoring System for Pathogenicity Assessments in Mendelian Disorders. <i>Human Mutation</i> , 2016, 37, 127-134.	1.1	52
1805	Hoyeraal-Hreidarsson Syndrome due to PARN Mutations: Fourteen Years of Follow-Up. <i>Pediatric Neurology</i> , 2016, 56, 62-68.e1.	1.0	29
1806	Novel mutations in SKIV2L and TTC37 genes in Malaysian children with trichohepatoenteric syndrome. <i>Gene</i> , 2016, 586, 1-6.	1.0	24
1807	Heterozygous colon cancer-associated mutations of <i>SAMHD1</i> have functional significance. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 4723-4728.	3.3	100
1808	Advances in Genetic Testing for Hereditary Cancer Syndromes. <i>Recent Results in Cancer Research</i> , 2016, 205, 1-15.	1.8	4
1809	Novel bioinformatic developments for exome sequencing. <i>Human Genetics</i> , 2016, 135, 603-614.	1.8	37
1810	Ligase-4 Deficiency Causes Distinctive Immune Abnormalities in Asymptomatic Individuals. <i>Journal of Clinical Immunology</i> , 2016, 36, 341-353.	2.0	30
1811	Polymorphisms within base and nucleotide excision repair pathways and risk of differentiated thyroid carcinoma. <i>DNA Repair</i> , 2016, 41, 27-31.	1.3	5
1812	Pathogenic classification of LPL gene variants reported to be associated with LPL deficiency. <i>Journal of Clinical Lipidology</i> , 2016, 10, 394-409.	0.6	35
1813	A flexible method for estimating the fraction of fitness influencing mutations from large sequencing data sets. <i>Genome Research</i> , 2016, 26, 834-843.	2.4	10
1814	BAP1 Germline Mutations in Finnish Patients with Uveal Melanoma. <i>Ophthalmology</i> , 2016, 123, 1112-1117.	2.5	39
1815	Targeted next-generation sequencing identification of mutations in patients with disorders of sex development. <i>BMC Medical Genetics</i> , 2016, 17, 23.	2.1	51
1816	Mutations in CAPN1 Cause Autosomal-Recessive Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2016, 98, 1038-1046.	2.6	96
1817	Comparison of pathogenicity prediction tools on missense variants in RYR1 and CACNA1S associated with malignant hyperthermia. <i>British Journal of Anaesthesia</i> , 2016, 117, 124-128.	1.5	34
1818	Computational approaches for predicting mutant protein stability. <i>Journal of Computer-Aided Molecular Design</i> , 2016, 30, 401-412.	1.3	60
1819	Iron-refractory iron deficiency anemia (IRIDA) cases with 2 novel <i>TMPRSS6</i> mutations. <i>Pediatric Hematology and Oncology</i> , 2016, 33, 226-232.	0.3	14
1820	Phenotypic features of CRB1-associated early-onset severe retinal dystrophy and the different molecular approaches to identifying the disease-causing variants. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2016, 254, 1833-1839.	1.0	19

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1821	Germline RECQL mutations in high risk Chinese breast cancer patients. <i>Breast Cancer Research and Treatment</i> , 2016, 157, 211-215.	1.1	23
1822	A Syndromic Intellectual Disability Disorder Caused by Variants in <i>TELO2</i> , a Gene Encoding a Component of the TTT Complex. <i>American Journal of Human Genetics</i> , 2016, 98, 909-918.	2.6	35
1823	Driver Gene Mutations in Stools of Colorectal Carcinoma Patients Detected by Targeted Next-Generation Sequencing. <i>Journal of Molecular Diagnostics</i> , 2016, 18, 471-479.	1.2	10
1824	Genomic Flatlining in the Endangered Island Fox. <i>Current Biology</i> , 2016, 26, 1183-1189.	1.8	201
1825	<i>CSF1R</i> mosaicism in a family with hereditary diffuse leukoencephalopathy with spheroids. <i>Brain</i> , 2016, 139, 1666-1672.	3.7	53
1826	Identification of novel mutations including a double mutation in patients with inherited cardiomyopathy by a targeted sequencing approach using the Ion Torrent PGM system. <i>International Journal of Molecular Medicine</i> , 2016, 37, 1511-1520.	1.8	9
1827	Somatic ERCC2 mutations are associated with a distinct genomic signature in urothelial tumors. <i>Nature Genetics</i> , 2016, 48, 600-606.	9.4	352
1828	Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. <i>Nature Genetics</i> , 2016, 48, 593-599.	9.4	273
1829	How to Assess the Clinical Relevance of Novel RET Missense Variants in the Absence of Functional Studies?. <i>European Thyroid Journal</i> , 2016, 5, 73-77.	1.2	3
1830	Hematopoietic stem cell transplantation for CTLA4 deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 615-619.e1.	1.5	88
1831	Detection of Germline Mutation in Hereditary Breast and/or Ovarian Cancers by Next-Generation Sequencing on a Four-Gene Panel. <i>Journal of Molecular Diagnostics</i> , 2016, 18, 580-594.	1.2	38
1832	Targeted sequencing of refractory myeloma reveals a high incidence of mutations in <i>CRBN</i> and Ras pathway genes. <i>Blood</i> , 2016, 128, 1226-1233.	0.6	185
1833	Post Mortem Diagnostic Exome Sequencing Identifies a de novo <i>TUBB3</i> Alteration in a Newborn with Prenatally Diagnosed Hydrocephalus and Suspected Walker-Warburg Syndrome. <i>Pediatric and Developmental Pathology</i> , 0, , .	0.5	0
1834	A novel missense mutation of <i>CMT2P</i> alters transcription machinery. <i>Annals of Neurology</i> , 2016, 80, 834-845.	2.8	18
1835	Mutational signatures in esophageal adenocarcinoma define etiologically distinct subgroups with therapeutic relevance. <i>Nature Genetics</i> , 2016, 48, 1131-1141.	9.4	332
1836	Mutations in mitochondrial enzyme <i>GPT2</i> cause metabolic dysfunction and neurological disease with developmental and progressive features. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E5598-607.	3.3	51
1837	De novo <i>PHIP</i> -predicted deleterious variants are associated with developmental delay, intellectual disability, obesity, and dysmorphic features. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a001172.	0.5	42
1838	Targeted Next-Generation Sequencing Successfully Detects Causative Genes in Chinese Patients with Hereditary Hearing Loss. <i>Genetic Testing and Molecular Biomarkers</i> , 2016, 20, 660-665.	0.3	32

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1839	Structure-function analyses of microsomal triglyceride transfer protein missense mutations in abetalipoproteinemia and hypobetalipoproteinemia subjects. <i>Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids</i> , 2016, 1861, 1623-1633.	1.2	21
1840	Non lethal Raine syndrome and differential diagnosis. <i>European Journal of Medical Genetics</i> , 2016, 59, 577-583.	0.7	33
1841	Investigating regulatory signatures of human autophagy related gene 5 (ATG5) through functional in silico analysis. <i>Meta Gene</i> , 2016, 9, 237-248.	0.3	16
1842	Bioinformatics and Orphan Diseases. <i>Translational Bioinformatics</i> , 2016, , 313-338.	0.0	0
1843	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. <i>American Journal of Human Genetics</i> , 2016, 99, 1086-1105.	2.6	45
1844	Identification of SLC26A4 mutations p.L582LfsX4, p.I188T and p.E704K in a Chinese family with large vestibular aqueduct syndrome (LVAS). <i>International Journal of Pediatric Otorhinolaryngology</i> , 2016, 91, 1-5.	0.4	5
1845	Genome-wide associations for birth weight and correlations with adult disease. <i>Nature</i> , 2016, 538, 248-252.	13.7	406
1846	Comparative genomic, transcriptomic and secretomic profiling of <i>Penicillium oxalicum</i> HP7-1 and its cellulase and xylanase hyper-producing mutant EU2106, and identification of two novel regulatory genes of cellulase and xylanase gene expression. <i>Biotechnology for Biofuels</i> , 2016, 9, 203.	6.2	73
1848	Proteogenomic Analysis of Single Amino Acid Polymorphisms in Cancer Research. <i>Advances in Experimental Medicine and Biology</i> , 2016, 926, 93-113.	0.8	7
1849	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. <i>American Journal of Human Genetics</i> , 2016, 99, 860-876.	2.6	93
1850	Increased burden of ultra-rare protein-altering variants among 4,877 individuals with schizophrenia. <i>Nature Neuroscience</i> , 2016, 19, 1433-1441.	7.1	427
1851	Co-existence of phenylketonuria either with maple syrup urine disease or Sandhoff disease in two patients from Iran: emphasizing the role of consanguinity. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2016, 29, 1215-1219.	0.4	7
1852	Arts syndrome with a novel missense mutation in the PRPS1 gene: A case report. <i>Brain and Development</i> , 2016, 38, 954-958.	0.6	15
1853	Exome and genome sequencing for inborn errors of immunity. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 957-969.	1.5	187
1854	Rare variants in SQSTM1 and VCP genes and risk of sporadic inclusion body myositis. <i>Neurobiology of Aging</i> , 2016, 47, 218.e1-218.e9.	1.5	40
1855	Temple-Baraitser Syndrome and Zimmermann-Laband Syndrome: one clinical entity?. <i>BMC Medical Genetics</i> , 2016, 17, 42.	2.1	27
1856	A reverse genetic approach identifies an ancestral frameshift mutation in RP1 causing recessive progressive retinal degeneration in European cattle breeds. <i>Genetics Selection Evolution</i> , 2016, 48, 56.	1.2	25
1857	Activation induced cytidine deaminase mutant (AID-His130Pro) from Hyper IgM 2 patient retained mutagenic activity on SHM artificial substrate. <i>Molecular Immunology</i> , 2016, 79, 77-82.	1.0	4

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1858	Novel motor phenotypes in patients with <i>VRK1</i> mutations without pontocerebellar hypoplasia. <i>Neurology</i> , 2016, 87, 65-70.	1.5	38
1859	Computational identification of non-synonymous polymorphisms within regions corresponding to protein interaction sites. <i>Computers in Biology and Medicine</i> , 2016, 79, 30-35.	3.9	6
1860	Fine mapping of the GWAS loci identifies <i>SLC35D1</i> and <i>IL23R</i> as potential risk genes for leprosy. <i>Journal of Dermatological Science</i> , 2016, 84, 322-329.	1.0	4
1861	Unravelling 5-oxoprolinuria (pyroglutamic aciduria) due to bi-allelic <i>OPLAH</i> mutations: 20 new mutations in 14 families. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 44-49.	0.5	9
1862	Simul-seq: combined DNA and RNA sequencing for whole-genome and transcriptome profiling. <i>Nature Methods</i> , 2016, 13, 953-958.	9.0	34
1863	The role of next generation sequencing in understanding male and female sexual development: clinical implications. <i>Expert Review of Endocrinology and Metabolism</i> , 2016, 11, 433-443.	1.2	3
1865	The Ensembl Variant Effect Predictor. <i>Genome Biology</i> , 2016, 17, 122.	3.8	5,181
1866	REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. <i>American Journal of Human Genetics</i> , 2016, 99, 877-885.	2.6	1,555
1867	An Application of NGS for Molecular Investigations in Perrault Syndrome: Study of 14 Families and Review of the Literature. <i>Human Mutation</i> , 2016, 37, 1354-1362.	1.1	46
1868	Polymorphic variants of <i>MRP4/ABCC4</i> differentially modulate the transport of methylated arsenic metabolites and physiological organic anions. <i>Biochemical Pharmacology</i> , 2016, 120, 72-82.	2.0	32
1869	Osteomalacia induced by long-term low-dose adefovir dipivoxil: Clinical characteristics and genetic predictors. <i>Bone</i> , 2016, 93, 97-103.	1.4	11
1870	Obesity and developmental delay in a patient with uniparental disomy of chromosome 2. <i>International Journal of Obesity</i> , 2016, 40, 1935-1941.	1.6	11
1871	Loss-of-Function Mutations in <i>SERPIN8</i> Linked to Exfoliative Ichthyosis with Impaired Mechanical Stability of Intercellular Adhesions. <i>American Journal of Human Genetics</i> , 2016, 99, 430-436.	2.6	27
1872	Whole Exome Sequencing Reveals a Mutation in <i>CRYBB2</i> in a Large Mexican Family with Autosomal Dominant Pulverulent Cataract. <i>Molecular Syndromology</i> , 2016, 7, 87-92.	0.3	5
1873	Relaxed selective constraints drove functional modifications in peripheral photoreception of the cavefish <i>P. andruzzii</i> and provide insight into the time of cave colonization. <i>Heredity</i> , 2016, 117, 383-392.	1.2	35
1874	Fatal Lymphoproliferative Disease in Two Siblings Lacking Functional <i>FAAP24</i> . <i>Journal of Clinical Immunology</i> , 2016, 36, 684-692.	2.0	20
1875	An inactivating mutation in intestinal cell kinase, <i>ICK</i> , impairs hedgehog signalling and causes short rib-polydactyly syndrome. <i>Human Molecular Genetics</i> , 2016, 25, 3998-4011.	1.4	44
1876	<i>IFT52</i> mutations destabilize anterograde complex assembly, disrupt ciliogenesis and result in short rib polydactyly syndrome. <i>Human Molecular Genetics</i> , 2016, 25, 4012-4020.	1.4	44

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1877	Evaluation of artificial selection in Standard Poodles using whole-genome sequencing. <i>Mammalian Genome</i> , 2016, 27, 599-609.	1.0	13
1878	Expanding the spectrum of PEX10-related peroxisomal biogenesis disorders: slowly progressive recessive ataxia. <i>Journal of Neurology</i> , 2016, 263, 1552-1558.	1.8	26
1879	Non-human primates avoid the detrimental effects of prenatal androgen exposure in mixed-sex litters: combined demographic, behavioral, and genetic analyses. <i>American Journal of Primatology</i> , 2016, 78, 1304-1315.	0.8	7
1880	Mutations in <i>MME</i> cause an autosomal recessive Charcot-Marie-Tooth disease type 2. <i>Annals of Neurology</i> , 2016, 79, 659-672.	2.8	82
1881	Phenotypic insights into <i>ADCY5</i> associated disease. <i>Movement Disorders</i> , 2016, 31, 1033-1040.	2.2	106
1882	A multifaceted computational report on the variants effect on KIR2DL3 and IFNL3 candidate gene in HCV clearance. <i>Molecular Biology Reports</i> , 2016, 43, 1101-1117.	1.0	5
1883	Beta thalassemia in 31,734 cases with HBB gene mutations: Pathogenic and structural analysis of the common mutations; Iran as the crossroads of the Middle East. <i>Blood Reviews</i> , 2016, 30, 493-508.	2.8	31
1884	Screening of CD96 and ASXL1 in 11 patients with Opitz C or Bohring-Opitz syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 24-31.	0.7	13
1885	<i>GREM1</i> and <i>POLE</i> variants in hereditary colorectal cancer syndromes. <i>Genes Chromosomes and Cancer</i> , 2016, 55, 95-106.	1.5	40
1886	Mutations in Known Monogenic High Bone Mass Loci Only Explain a Small Proportion of High Bone Mass Cases. <i>Journal of Bone and Mineral Research</i> , 2016, 31, 640-649.	3.1	38
1887	Hutterite cataract maps to chromosome 6p21.32-p21.31, cosegregates with a homozygous mutation in <i>LEMD2</i> , and is associated with sudden cardiac death. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 77-94.	0.6	28
1888	Lynch syndrome mutation spectrum in New South Wales, Australia, including 55 novel mutations. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 223-231.	0.6	16
1889	Exome sequencing identifies variants in two genes encoding the LIM-proteins NRAP and FHL1 in an Italian patient with BAG3 myofibrillar myopathy. <i>Journal of Muscle Research and Cell Motility</i> , 2016, 37, 101-115.	0.9	23
1890	Genetic aetiology of ophthalmological manifestations in children – a focus on mitochondrial disease-related symptoms. <i>Acta Ophthalmologica</i> , 2016, 94, 83-91.	0.6	10
1891	Disrupted lymphocyte homeostasis in hepatitis-associated acquired aplastic anemia is associated with short telomeres. <i>American Journal of Hematology</i> , 2016, 91, 243-247.	2.0	11
1892	Should any genetic defect affecting α -granules in platelets be classified as gray platelet syndrome?. <i>American Journal of Hematology</i> , 2016, 91, 714-718.	2.0	25
1893	Cellular defects caused by hypomorphic variants of the Bloom syndrome helicase gene BLM. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 106-119.	0.6	12
1894	Lactoferrin gene polymorphisms in Italian patients with recurrent tonsillitis. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2016, 88, 153-156.	0.4	1

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1895	A Sorghum Mutant Resource as an Efficient Platform for Gene Discovery in Grasses. <i>Plant Cell</i> , 2016, 28, tpc.00373.2016.	3.1	113
1896	Genetic variants of the MAVS, MITA and MFN2 genes are not associated with leprosy in Han Chinese from Southwest China. <i>Infection, Genetics and Evolution</i> , 2016, 45, 105-110.	1.0	6
1897	Polymorphisms in DNA repair genes in gastrointestinal stromal tumours: susceptibility and correlation with tumour characteristics and clinical outcome. <i>Tumor Biology</i> , 2016, 37, 13413-13423.	0.8	19
1898	Homozygous inactivation of <i>CHEK2</i> is linked to a familial case of multiple primary lung cancer with accompanying cancers in other organs. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a001032.	0.5	16
1899	Acute Intermittent Porphyria: Predicted Pathogenicity of <i>HMBS</i> Variants Indicates Extremely Low Penetrance of the Autosomal Dominant Disease. <i>Human Mutation</i> , 2016, 37, 1215-1222.	1.1	129
1900	Spectrum of Mutations in Hypertrophic Cardiomyopathy Genes Among Tunisian Patients. <i>Genetic Testing and Molecular Biomarkers</i> , 2016, 20, 674-679.	0.3	11
1901	Determinants of Power in Gene-Based Burden Testing for Monogenic Disorders. <i>American Journal of Human Genetics</i> , 2016, 99, 527-539.	2.6	39
1902	Germline mutations and genotype-phenotype correlation in Asian Indian patients with pheochromocytoma and paraganglioma. <i>European Journal of Endocrinology</i> , 2016, 175, 311-323.	1.9	27
1903	Novel mutations in <i>Vicugna pacos</i> (alpaca) <i>Tyrp1</i> are not correlated with brown fibre colour phenotypes. <i>Small Ruminant Research</i> , 2016, 143, 29-34.	0.6	4
1904	Mutational Spectrum in Holoprosencephaly Shows That FGF is a New Major Signaling Pathway. <i>Human Mutation</i> , 2016, 37, 1329-1339.	1.1	56
1905	Hot spot mutations in Finnish non-small cell lung cancers. <i>Lung Cancer</i> , 2016, 99, 102-110.	0.9	21
1906	Identification of a novel <i>AGXT</i> gene mutation in primary hyperoxaluria after kidney transplantation failure. <i>Transplant Immunology</i> , 2016, 39, 60-65.	0.6	5
1907	A vitamin D pathway gene-gene interaction affects low-density lipoprotein cholesterol levels. <i>Journal of Nutritional Biochemistry</i> , 2016, 38, 12-17.	1.9	13
1908	A genetic network that suppresses genome rearrangements in <i>Saccharomyces cerevisiae</i> and contains defects in cancers. <i>Nature Communications</i> , 2016, 7, 11256.	5.8	36
1909	Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 568-580.	0.6	83
1910	Pathogenic germline <i>MCM9</i> variants are rare in Australian Lynch-like syndrome patients. <i>Cancer Genetics</i> , 2016, 209, 497-500.	0.2	8
1911	Identification of two novel mutations in the <i>GALNT3</i> gene in a Chinese family with hyperphosphatemic familial tumoral calcinosis. <i>Bone Research</i> , 2016, 4, 16038.	5.4	11
1912	Whole-genome sequencing of the endangered bovine species Gayal (<i>Bos frontalis</i>) provides new insights into its genetic features. <i>Scientific Reports</i> , 2016, 6, 19787.	1.6	32

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1913	Genetic analysis of rare coding mutations of <i>CELSR1</i> in congenital heart and neural tube defects in Chinese people. <i>Clinical Science</i> , 2016, 130, 2329-2340.	1.8	37
1914	Genomic architecture of inflammatory bowel disease in five families with multiple affected individuals. <i>Human Genome Variation</i> , 2016, 3, 15060.	0.4	14
1915	Hyperlipidemia-associated gene variations and expression patterns revealed by whole-genome and transcriptome sequencing of rabbit models. <i>Scientific Reports</i> , 2016, 6, 26942.	1.6	24
1916	Polygenic Versus Monogenic Causes of Hypercholesterolemia Ascertained Clinically. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2016, 36, 2439-2445.	1.1	174
1917	Next-generation sequencing and a novel <i>COL3A1</i> mutation associated with vascular Ehlers-Danlos syndrome with severe intestinal involvement: a case report. <i>Journal of Medical Case Reports</i> , 2016, 10, 303.	0.4	9
1918	BLM germline and somatic <i>PKMYT1</i> and <i>AHCY</i> mutations: Genetic variations beyond <i>MYCN</i> and prognosis in neuroblastoma. <i>Medical Hypotheses</i> , 2016, 97, 22-25.	0.8	10
1919	Human genetic variation database, a reference database of genetic variations in the Japanese population. <i>Journal of Human Genetics</i> , 2016, 61, 547-553.	1.1	270
1920	Mutations in <i>REEP6</i> Cause Autosomal-Recessive Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2016, 99, 1305-1315.	2.6	121
1921	Tet oncogene family member 2 gene alterations in childhood acute myeloid leukemia. <i>Journal of the Formosan Medical Association</i> , 2016, 115, 801-806.	0.8	8
1922	The Qatar genome: a population-specific tool for precision medicine in the Middle East. <i>Human Genome Variation</i> , 2016, 3, 16016.	0.4	103
1923	3-Methylcrotonyl-CoA carboxylase deficiency: Mutational spectrum derived from comprehensive newborn screening. <i>Gene</i> , 2016, 594, 203-210.	1.0	20
1925	A novel de novo mutation in <i>ATP1A3</i> and childhood-onset schizophrenia. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a001008.	0.5	46
1926	Functional polymorphism in lycopene beta-cyclase gene as a molecular marker to predict bixin production in <i>Bixa orellana</i> L. (achiote). <i>Molecular Breeding</i> , 2016, 36, 1.	1.0	12
1927	A Biobank of Breast Cancer Explants with Preserved Intra-tumor Heterogeneity to Screen Anticancer Compounds. <i>Cell</i> , 2016, 167, 260-274.e22.	13.5	376
1928	Combined variants in factor VIII and prostaglandin synthase-1 amplify hemorrhage severity across three generations of descendants. <i>Journal of Thrombosis and Haemostasis</i> , 2016, 14, 2230-2240.	1.9	6
1929	Estrogen Drives Cellular Transformation and Mutagenesis in Cells Expressing the Breast Cancer-Associated R438W DNA Polymerase Lambda Protein. <i>Molecular Cancer Research</i> , 2016, 14, 1068-1077.	1.5	12
1930	A de novo missense mutation in <i>ZMYND11</i> is associated with global developmental delay, seizures, and hypotonia. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a000851.	0.5	16
1931	Mutation spectrum of the <i>FZD-4</i> , <i>TSPAN12</i> AND <i>ZNF408</i> genes in Indian FEVR patients. <i>BMC Ophthalmology</i> , 2016, 16, 90.	0.6	28

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1932	Population-specific single nucleotide polymorphism confers increased risk of venous thromboembolism in African Americans. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 513-520.	0.6	7
1933	Assessing the general population frequency of rare coding variants in the EXT1 and EXT2 genes previously implicated in hereditary multiple exostoses. <i>Bone</i> , 2016, 92, 196-200.	1.4	12
1934	Genetic Factors of the Disease Course After Sepsis: Rare Deleterious Variants Are Predictive. <i>EBioMedicine</i> , 2016, 12, 227-238.	2.7	34
1935	NGS-based reverse genetic screen for common embryonic lethal mutations compromising fertility in livestock. <i>Genome Research</i> , 2016, 26, 1333-1341.	2.4	71
1936	A mutation in the glutamate-rich region of RNA-binding motif protein 20 causes dilated cardiomyopathy through missplicing of titin and impaired Frank-Starling mechanism. <i>Cardiovascular Research</i> , 2016, 112, 452-463.	1.8	97
1937	Structural impact analysis of missense SNPs present in the uroguanylin gene by long-term molecular dynamics simulations. <i>Journal of Theoretical Biology</i> , 2016, 410, 9-17.	0.8	11
1938	Clonal hematopoiesis in patients with dyskeratosis congenita. <i>American Journal of Hematology</i> , 2016, 91, 1227-1233.	2.0	44
1939	De Novo Mutations in CHD4, an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dismorphisms. <i>American Journal of Human Genetics</i> , 2016, 99, 934-941.	2.6	111
1940	<i>PRKG1</i> and genetic diagnosis of early-onset thoracic aortic disease. <i>European Journal of Clinical Investigation</i> , 2016, 46, 787-794.	1.7	21
1941	SZDB: A Database for Schizophrenia Genetic Research. <i>Schizophrenia Bulletin</i> , 2017, 43, sbw102.	2.3	91
1942	Spectrum of <i>SMPD1</i> mutations in Asian-Indian patients with acid sphingomyelinase (ASM)-deficient Niemann-Pick disease. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2719-2730.	0.7	15
1943	Association of Exome Sequences With Cardiovascular Traits Among Blacks in the Jackson Heart Study. <i>Circulation: Cardiovascular Genetics</i> , 2016, 9, 368-374.	5.1	8
1944	Genotype and Phenotype Characteristics in 22 Patients with Vitamin D-Dependent Rickets Type I. <i>Hormone Research in Paediatrics</i> , 2016, 85, 309-317.	0.8	24
1945	Exome sequencing of geographically diverse barley landraces and wild relatives gives insights into environmental adaptation. <i>Nature Genetics</i> , 2016, 48, 1024-1030.	9.4	259
1946	Computational analysis and enzyme assay of inhibitor response to disease single nucleotide polymorphisms (SNPs) in lipoprotein lipase. <i>Journal of Bioinformatics and Computational Biology</i> , 2016, 14, 1650028.	0.3	6
1947	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042.	9.4	218
1948	Genetic Landscape of Sporadic Unilateral Adrenocortical Adenomas Without PRKACA p.Leu206Arg Mutation. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 3526-3538.	1.8	65
1949	A preliminary study to evaluate the strategy of combining clinical criteria and next generation sequencing (NGS) for the identification of monogenic diabetes among multi-ethnic Asians. <i>Diabetes Research and Clinical Practice</i> , 2016, 119, 13-22.	1.1	37

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1950	Next-generation sequencing for molecular diagnosis of autosomal recessive polycystic kidney disease. <i>Gene</i> , 2016, 591, 214-226.	1.0	15
1951	Truncating and missense <i>PPM1D</i> mutations in early-onset and/or familial/hereditary prostate cancer patients. <i>Genes Chromosomes and Cancer</i> , 2016, 55, 954-961.	1.5	15
1952	Challenging a dogma: co-mutations exist in MAPK pathway genes in colorectal cancer. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2016, 469, 459-464.	1.4	5
1953	The Complementarity Between Protein-Specific and General Pathogenicity Predictors for Amino Acid Substitutions. <i>Human Mutation</i> , 2016, 37, 1013-1024.	1.1	42
1954	The Anatomy to Genomics (ATG) Start Genetics medical school initiative: incorporating exome sequencing data from cadavers used for Anatomy instruction into the first year curriculum. <i>BMC Medical Genomics</i> , 2016, 9, 62.	0.7	10
1955	Association study between near-MC4R variants and obesity-related variables in Portuguese young adults. <i>Gene Reports</i> , 2016, 5, 98-101.	0.4	0
1956	Spatial intratumoral heterogeneity and temporal clonal evolution in esophageal squamous cell carcinoma. <i>Nature Genetics</i> , 2016, 48, 1500-1507.	9.4	217
1957	Targeted exome sequencing resolves allelic and the genetic heterogeneity in the genetic diagnosis of nephronophthisis-related ciliopathy. <i>Experimental and Molecular Medicine</i> , 2016, 48, e251-e251.	3.2	26
1958	Dwarfism with joint laxity in Friesian horses is associated with a splice site mutation in <i>B4GALT7</i> . <i>BMC Genomics</i> , 2016, 17, 839.	1.2	31
1959	Clinical and molecular study of a pediatric patient with sodium taurocholate cotransporting polypeptide deficiency. <i>Experimental and Therapeutic Medicine</i> , 2016, 12, 3294-3300.	0.8	39
1960	Targeted Resequencing of Deafness Genes Reveals a Founder <i>MYO15A</i> Variant in Northeastern Brazil. <i>Annals of Human Genetics</i> , 2016, 80, 327-331.	0.3	17
1961	In silico SNP analysis of the breast cancer antigen NY-BR-1. <i>BMC Cancer</i> , 2016, 16, 901.	1.1	10
1963	Current Perspectives in Autism Spectrum Disorder: From Genes to Therapy. <i>Journal of Neuroscience</i> , 2016, 36, 11402-11410.	1.7	44
1964	Capturing the biology of disease severity in a PSC-based model of familial dysautonomia. <i>Nature Medicine</i> , 2016, 22, 1421-1427.	15.2	58
1965	Inactivation of TGF β ² receptors in stem cells drives cutaneous squamous cell carcinoma. <i>Nature Communications</i> , 2016, 7, 12493.	5.8	81
1966	A continuum of admixture in the Western Hemisphere revealed by the African Diaspora genome. <i>Nature Communications</i> , 2016, 7, 12522.	5.8	136
1967	Challenges and disparities in the application of personalized genomic medicine to populations with African ancestry. <i>Nature Communications</i> , 2016, 7, 12521.	5.8	68
1968	Inherited breast cancer predisposition in Asians: multigene panel testing outcomes from Singapore. <i>Npj Genomic Medicine</i> , 2016, 1, 15003.	1.7	44

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1969	Variant detection and runs of homozygosity in next generation sequencing data elucidate the genetic background of Lundehund syndrome. <i>BMC Genomics</i> , 2016, 17, 535.	1.2	12
1970	Novel mutation in the CHST6 gene causes macular corneal dystrophy in a black South African family. <i>BMC Medical Genetics</i> , 2016, 17, 47.	2.1	11
1971	Scarce evidence of the causal role of germline mutations in UNC5C in hereditary colorectal cancer and polyposis. <i>Scientific Reports</i> , 2016, 6, 20697.	1.6	9
1972	Computational Approaches to Accelerating Novel Medicine and Better Patient Care from Bedside to Benchtop. <i>Advances in Protein Chemistry and Structural Biology</i> , 2016, 102, 147-179.	1.0	2
1973	Genomic Variants Associated with Resistance to High Fat Diet Induced Obesity in a Primate Model. <i>Scientific Reports</i> , 2016, 6, 36123.	1.6	23
1975	Translational Biomedical Informatics. <i>Advances in Experimental Medicine and Biology</i> , 2016, , .	0.8	1
1976	The stepwise evolution of the exome during acquisition of docetaxel resistance in breast cancer cells. <i>BMC Genomics</i> , 2016, 17, 442.	1.2	25
1977	Frequent mutations in acetylation and ubiquitination sites suggest novel driver mechanisms of cancer. <i>Genome Medicine</i> , 2016, 8, 55.	3.6	51
1978	Spectrum of mutations in homozygous familial hypercholesterolemia in India, with four novel mutations. <i>Atherosclerosis</i> , 2016, 255, 31-36.	0.4	15
1979	PSIP1/LEDGF: a new gene likely involved in sensorineural progressive hearing loss. <i>Scientific Reports</i> , 2016, 5, 18568.	1.6	7
1980	Identification of the Photoreceptor Transcriptional Co-Repressor SAMD11 as Novel Cause of Autosomal Recessive Retinitis Pigmentosa. <i>Scientific Reports</i> , 2016, 6, 35370.	1.6	13
1981	A genome-wide association meta-analysis on apolipoprotein A-IV concentrations. <i>Human Molecular Genetics</i> , 2016, 25, 3635-3646.	1.4	46
1982	Clinical profile and molecular characterization of Galactosemia in Brazil: identification of seven novel mutations. <i>BMC Medical Genetics</i> , 2016, 17, 39.	2.1	14
1983	Next generation sequencing based identification of disease-associated mutations in Swiss patients with retinal dystrophies. <i>Scientific Reports</i> , 2016, 6, 28755.	1.6	62
1984	Next generation sequencing identifies abnormal Y chromosome and candidate causal variants in premature ovarian failure patients. <i>Genomics</i> , 2016, 108, 209-215.	1.3	12
1985	Patients with genetically heterogeneous synchronous colorectal cancer carry rare damaging germline mutations in immune-related genes. <i>Nature Communications</i> , 2016, 7, 12072.	5.8	49
1986	Stabilizing mutations of KLHL24 ubiquitin ligase cause loss of keratin 14 and human skin fragility. <i>Nature Genetics</i> , 2016, 48, 1508-1516.	9.4	101
1987	Mutation screening of GRIN2B in schizophrenia and autism spectrum disorder in a Japanese population. <i>Scientific Reports</i> , 2016, 6, 33311.	1.6	23

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1988	Natural variation in non-coding regions underlying phenotypic diversity in budding yeast. <i>Scientific Reports</i> , 2016, 6, 21849.	1.6	100
1989	Biochemical and functional characterization of glycosylation-associated mutational landscapes in colon cancer. <i>Scientific Reports</i> , 2016, 6, 23642.	1.6	39
1990	A case study of an integrative genomic and experimental therapeutic approach for rare tumors: identification of vulnerabilities in a pediatric poorly differentiated carcinoma. <i>Genome Medicine</i> , 2016, 8, 116.	3.6	15
1991	<i>MCM8</i> and <i>MCM9</i> Nucleotide Variants in Women with Primary Ovarian Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, jc.2016-2565.	1.8	68
1992	Exploring Human Diseases and Biological Mechanisms by Protein Structure Prediction and Modeling. <i>Advances in Experimental Medicine and Biology</i> , 2016, 939, 39-61.	0.8	4
1993	A <i>CD2AP</i> Mutation Associated with Focal Segmental Glomerulosclerosis in Young Adulthood. <i>Clinical Medicine Insights: Case Reports</i> , 2016, 9, CCRRep.S30867.	0.3	13
1994	Identification of Genetic Defects Underlying FXII Deficiency in Four Unrelated Chinese Patients. <i>Acta Haematologica</i> , 2016, 135, 238-240.	0.7	10
1995	RGR variants in different forms of retinal diseases: The undetermined role of truncation mutations. <i>Molecular Medicine Reports</i> , 2016, 14, 4811-4815.	1.1	5
1996	<i>SLC22A2</i> â€œ mapping genomic variations within South African indigenous and admixed populations. <i>Drug Metabolism and Personalized Therapy</i> , 2016, 31, 213-220.	0.3	2
1997	The analysis of heterotaxy patients reveals new loss-of-function variants of GRK5. <i>Scientific Reports</i> , 2016, 6, 33231.	1.6	4
1998	Role of von Willebrand Factorâ€™A1 Domain Variants P1266L, H1268D, C1272R, and C1272F in VWD. <i>Advances in Protein Chemistry and Structural Biology</i> , 2016, 102, 299-330.	1.0	1
1999	Integrative analyses reveal signaling pathways underlying familial breast cancer susceptibility. <i>Molecular Systems Biology</i> , 2016, 12, 860.	3.2	14
2000	Threeâ€œDimensional Model of Human Nicotinamide Nucleotide Transhydrogenase (NNT) and Sequenceâ€œStructure Analysis of its Diseaseâ€œCausing Variations. <i>Human Mutation</i> , 2016, 37, 1074-1084.	1.1	16
2001	Global inference of disease-causing single nucleotide variants from exome sequencing data. <i>BMC Bioinformatics</i> , 2016, 17, 468.	1.2	4
2002	Independent test assessment using the extreme value distribution theory. <i>BMC Proceedings</i> , 2016, 10, 245-249.	1.8	1
2003	Deep genome sequencing and variation analysis of 13 inbred mouse strains defines candidate phenotypic alleles, private variation and homozygous truncating mutations. <i>Genome Biology</i> , 2016, 17, 167.	3.8	70
2004	Establishing and validating regulatory regions for variant annotation and expression analysis. <i>BMC Genomics</i> , 2016, 17, 393.	1.2	14
2005	Mutation analysis of the COL1A1 and COL1A2 genes in Vietnamese patients with osteogenesis imperfecta. <i>Human Genomics</i> , 2016, 10, 27.	1.4	32

#	ARTICLE	IF	CITATIONS
2006	VaProS: a database-integration approach for protein/genome information retrieval. <i>Journal of Structural and Functional Genomics</i> , 2016, 17, 69-81.	1.2	9
2007	How Do We Make Clinical Molecular Testing for Cancer Standard of Care for Pathology Departments?. <i>Journal of the National Comprehensive Cancer Network: JNCCN</i> , 2016, 14, 787-792.	2.3	4
2008	Whole-Genome Sequencing and iPLEX MassARRAY Genotyping Map an EMS-Induced Mutation Affecting Cell Competition in <i>Drosophila melanogaster</i> . <i>G3: Genes, Genomes, Genetics</i> , 2016, 6, 3207-3217.	0.8	26
2009	<i>DNAJC6</i> Mutations Associated with Early-Onset Parkinson's Disease. <i>Annals of Neurology</i> , 2016, 79, 244-256.	2.8	148
2010	<i>MCM3AP</i> and <i>POMP</i> Mutations Cause a DNA-Repair and DNA-Damage-Signaling Defect in an Immunodeficient Child. <i>Human Mutation</i> , 2016, 37, 257-268.	1.1	18
2011	Expanding the Phenotype Associated with NAA10-Related N-Terminal Acetylation Deficiency. <i>Human Mutation</i> , 2016, 37, 755-764.	1.1	70
2012	Identification and characterization of 20 novel pathogenic variants in 60 unrelated Indian patients with mucopolysaccharidoses type I and type II. <i>Clinical Genetics</i> , 2016, 90, 496-508.	1.0	23
2013	Somatic Mosaicism for a Lethal <i>GJB2</i> Mutation Results in a Patterned Form of Spiny Hyperkeratosis without Eccrine Involvement. <i>Pediatric Dermatology</i> , 2016, 33, 322-326.	0.5	5
2014	New <i>KEL*01M</i> and <i>KEL*02M</i> alleles: structural modeling to assess the impact of amino acid changes. <i>Transfusion</i> , 2016, 56, 1223-1229.	0.8	1
2015	An exome-wide analysis of low frequency and rare variants in relation to risk of breast cancer in African American Women: the AMBER Consortium. <i>Carcinogenesis</i> , 2016, 37, 870-877.	1.3	22
2016	Evidence for adaptation of porcine Toll-like receptors. <i>Immunogenetics</i> , 2016, 68, 179-189.	1.2	7
2018	Analysis of DNAJC13 mutations in French-Canadian/French cohort of Parkinson's disease. <i>Neurobiology of Aging</i> , 2016, 45, 212.e13-212.e17.	1.5	38
2019	Very low-depth sequencing in a founder population identifies a cardioprotective <i>APOC3</i> signal missed by genome-wide imputation. <i>Human Molecular Genetics</i> , 2016, 25, 2360-2365.	1.4	21
2020	Identification of Genetic Polymorphisms of CYP2W1 in the Three Main Chinese Ethnicities: Han, Tibetan, and Uighur. <i>Drug Metabolism and Disposition</i> , 2016, 44, 1510-1515.	1.7	3
2021	STRUM: structure-based prediction of protein stability changes upon single-point mutation. <i>Bioinformatics</i> , 2016, 32, 2936-2946.	1.8	275
2022	Severe Neonatal Presentation of Mitochondrial Citrate Carrier (SLC25A1) Deficiency. <i>JIMD Reports</i> , 2016, 30, 73-79.	0.7	21
2023	Hereditary nonspherocytic hemolytic anemia caused by red cell glucose-6-phosphate isomerase (GPI) deficiency in two Portuguese patients: Clinical features and molecular study. <i>Blood Cells, Molecules, and Diseases</i> , 2016, 60, 18-23.	0.6	12
2024	Hypodysfibrinogenemia: A novel abnormal fibrinogen associated with bleeding and thrombotic complications. <i>Clinica Chimica Acta</i> , 2016, 460, 55-62.	0.5	8

#	ARTICLE	IF	CITATIONS
2025	Aberrant splicing induced by the most common <i>EPG5</i> mutation in an individual with Vici syndrome. <i>Brain</i> , 2016, 139, e52-e52.	3.7	14
2026	Mitochondrial Complex III Deficiency with Ketoacidosis and Hyperglycemia Mimicking Neonatal Diabetes. <i>JIMD Reports</i> , 2016, 31, 57-62.	0.7	5
2027	Natural History of Congenital Generalized Lipodystrophy: A Nationwide Study From Turkey. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 2759-2767.	1.8	67
2028	Transcriptional profiling of the ovine abomasal lymph node reveals a role for timing of the immune response in gastrointestinal nematode resistance. <i>Veterinary Parasitology</i> , 2016, 224, 96-108.	0.7	19
2029	A study of the mutational landscape of pediatric-type follicular lymphoma and pediatric nodal marginal zone lymphoma. <i>Modern Pathology</i> , 2016, 29, 1212-1220.	2.9	46
2030	Whole-genome re-sequencing for the identification of high contribution susceptibility gene variants in patients with type 2 diabetes. <i>Molecular Medicine Reports</i> , 2016, 13, 3735-3746.	1.1	5
2031	In silico analysis of deleterious single nucleotide polymorphisms in human BUB1 mitotic checkpoint serine/threonine kinase B gene. <i>Meta Gene</i> , 2016, 9, 142-150.	0.3	9
2032	Variants within the SP110 nuclear body protein modify risk of canine degenerative myelopathy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E3091-100.	3.3	32
2033	Carbamoylphosphate synthetase 1 (CPS1) deficiency: clinical, biochemical, and molecular characterization in Malaysian patients. <i>European Journal of Pediatrics</i> , 2016, 175, 339-346.	1.3	13
2034	Whole genome sequencing and its applications in medical genetics. <i>Quantitative Biology</i> , 2016, 4, 115-128.	0.3	7
2035	Seventeen Novel Mutations in <i>PCCA</i> and <i>PCCB</i> Genes in Indian Propionic Acidemia Patients, and Their Outcomes. <i>Genetic Testing and Molecular Biomarkers</i> , 2016, 20, 373-382.	0.3	12
2036	The Spectrum and Clinical Impact of Epigenetic Modifier Mutations in Myeloma. <i>Clinical Cancer Research</i> , 2016, 22, 5783-5794.	3.2	81
2037	Semiconductor Whole Exome Sequencing for the Identification of Genetic Variants in Colombian Patients Clinically Diagnosed with Long QT Syndrome. <i>Molecular Diagnosis and Therapy</i> , 2016, 20, 353-362.	1.6	2
2038	Identification of a novel HLA-A allele, HLA-A*01:195, in a UAE national. <i>Human Immunology</i> , 2016, 77, 605-608.	1.2	3
2039	A rare genetic variant of the ryanodine receptor in a suspected malignant hyperthermia susceptible patient. <i>Journal of Clinical Anesthesia</i> , 2016, 33, 144-146.	0.7	1
2040	Alpha-1-antitrypsin (SERPINA1) mutation spectrum: Three novel variants and haplotype characterization of rare deficiency alleles identified in Portugal. <i>Respiratory Medicine</i> , 2016, 116, 8-18.	1.3	53
2041	Autosomal-Dominant Corneal Endothelial Dystrophies CHED1 and PPCD1 Are Allelic Disorders Caused by Non-coding Mutations in the Promoter of OVOL2. <i>American Journal of Human Genetics</i> , 2016, 98, 75-89.	2.6	70
2042	Genetic loci for Epstein-Barr virus nuclear antigen-1 are associated with risk of multiple sclerosis. <i>Multiple Sclerosis Journal</i> , 2016, 22, 1655-1664.	1.4	44

#	ARTICLE	IF	CITATIONS
2043	Filtering genetic variants and placing informative priors based on putative biological function. BMC Genetics, 2016, 17, 8.	2.7	6
2044	Exome sequencing in one family with gastric- and rectal cancer. BMC Genetics, 2016, 17, 41.	2.7	31
2045	The genomic architecture of resistance to Campylobacter jejuni intestinal colonisation in chickens. BMC Genomics, 2016, 17, 293.	1.2	28
2046	Variants of PLCXD3 are not associated with variant or sporadic Creutzfeldt-Jakob disease in a large international study. BMC Medical Genetics, 2016, 17, 28.	2.1	3
2047	Pathogenic copy number variants and SCN1A mutations in patients with intellectual disability and childhood-onset epilepsy. BMC Medical Genetics, 2016, 17, 34.	2.1	23
2048	Evaluation of pleiotropic effects among common genetic loci identified for cardio-metabolic traits in a Korean population. Cardiovascular Diabetology, 2016, 15, 20.	2.7	20
2049	Investigation of SLA4A3 as a candidate gene for human retinal disease. Journal of Negative Results in BioMedicine, 2016, 15, 11.	1.4	1
2050	High-performance web services for querying gene and variant annotation. Genome Biology, 2016, 17, 91.	3.8	166
2051	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. Cilia, 2016, 5, 8.	1.8	37
2052	Genetic studies of Polish migraine patients: screening for causative mutations in four migraine-associated genes. Human Genomics, 2016, 10, 3.	1.4	10
2053	Novel function discovery through sequence and structural data mining. Current Opinion in Structural Biology, 2016, 38, 53-61.	2.6	32
2054	Biallelic Mutations of VAC14 in Pediatric-Onset Neurological Disease. American Journal of Human Genetics, 2016, 99, 188-194.	2.6	45
2055	Identifying the genome-wide genetic variation between precocious trifoliolate orange and its wild type and developing new markers for genetics research. DNA Research, 2016, 23, 403-414.	1.5	36
2056	The Extent of mRNA Editing Is Limited in Chicken Liver and Adipose, but Impacted by Tissue Context, Genotype, Age, and Feeding as Exemplified with a Conserved Edited Site in COG3. G3: Genes, Genomes, Genetics, 2016, 6, 321-335.	0.8	13
2057	SMPD1 Mutation Update: Database and Comprehensive Analysis of Published and Novel Variants. Human Mutation, 2016, 37, 139-147.	1.1	66
2058	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811.	1.1	92
2059	Role of <i>ATG10</i> expression quantitative trait loci in non-small cell lung cancer survival. International Journal of Cancer, 2016, 139, 1564-1573.	2.3	55
2060	In Silico Functional Annotation of Genomic Variation. Current Protocols in Human Genetics, 2016, 88, 6.15.1-6.15.17.	3.5	20

#	ARTICLE	IF	CITATIONS
2061	Target resequencing of neuromuscular disease-related genes using next-generation sequencing for patients with undiagnosed early-onset neuromuscular disorders. <i>Journal of Human Genetics</i> , 2016, 61, 931-942.	1.1	22
2062	<i>BRAT1</i> -related disease identification of a patient without early lethality. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 699-702.	0.7	24
2063	<i>EZH2</i> mutation in an adolescent with Weaver syndrome developing acute myeloid leukemia and secondary hemophagocytic lymphohistiocytosis. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1274-1277.	0.7	23
2064	Mutation screening of <i>SCN2A</i> in schizophrenia and identification of a novel loss-of-function mutation. <i>Psychiatric Genetics</i> , 2016, 26, 60-65.	0.6	45
2065	<i>CARD14</i> alterations in Tunisian patients with psoriasis and further characterization in European cohorts. <i>British Journal of Dermatology</i> , 2016, 174, 330-337.	1.4	33
2066	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. <i>Molecular Psychiatry</i> , 2016, 21, 133-148.	4.1	243
2067	Comprehensive genome-wide evaluation of lapatinib-induced liver injury yields a single genetic signal centered on known risk allele HLA-DRB1*07:01. <i>Pharmacogenomics Journal</i> , 2016, 16, 180-185.	0.9	51
2068	High incidence of unrecognized visceral/neurological late-onset Niemann-Pick disease, type C1, predicted by analysis of massively parallel sequencing data sets. <i>Genetics in Medicine</i> , 2016, 18, 41-48.	1.1	171
2069	Global molecular analysis and APOE mutations in a cohort of autosomal dominant hypercholesterolemia patients in France. <i>Journal of Lipid Research</i> , 2016, 57, 482-491.	2.0	29
2070	<i>ATP7B</i> Gene Mutations in Croatian Patients with Wilson Disease. <i>Genetic Testing and Molecular Biomarkers</i> , 2016, 20, 112-117.	0.3	11
2071	Recurrent candidiasis and early-onset gastric cancer in a patient with a genetically defined partial MYD88 defect. <i>Familial Cancer</i> , 2016, 15, 289-296.	0.9	13
2072	The mutation significance cutoff: gene-level thresholds for variant predictions. <i>Nature Methods</i> , 2016, 13, 109-110.	9.0	249
2073	Prevalence of Monogenic Causes in Pediatric Patients with Nephrolithiasis or Nephrocalcinosis. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2016, 11, 664-672.	2.2	105
2074	Mutational spectrum of Barrett's stem cells suggests paths to initiation of a precancerous lesion. <i>Nature Communications</i> , 2016, 7, 10380.	5.8	57
2075	Association of the MTHFR rs1801131 and rs1801133 variants in sporadic Parkinson's disease patients. <i>Neuroscience Letters</i> , 2016, 616, 26-31.	1.0	20
2076	A panoply of errors: polymerase proofreading domain mutations in cancer. <i>Nature Reviews Cancer</i> , 2016, 16, 71-81.	12.8	292
2077	Multicenter Feasibility Study of Tumor Molecular Profiling to Inform Therapeutic Decisions in Advanced Pediatric Solid Tumors. <i>JAMA Oncology</i> , 2016, 2, 608.	3.4	172
2078	A Multiplex Kindred with Hennekam Syndrome due to Homozygosity for a CCBE1 Mutation that does not Prevent Protein Expression. <i>Journal of Clinical Immunology</i> , 2016, 36, 19-27.	2.0	12

#	ARTICLE	IF	CITATIONS
2079	Identification of critical variants within SLC44A4, an ulcerative colitis susceptibility gene identified in a GWAS in north Indians. <i>Genes and Immunity</i> , 2016, 17, 105-109.	2.2	12
2080	Mutation Profiling of Usual Ductal Hyperplasia of the Breast Reveals Activating Mutations Predominantly at Different Levels of the PI3K/AKT/mTOR Pathway. <i>American Journal of Pathology</i> , 2016, 186, 15-23.	1.9	20
2081	267 Spanish Exomes Reveal Population-Specific Differences in Disease-Related Genetic Variation. <i>Molecular Biology and Evolution</i> , 2016, 33, 1205-1218.	3.5	78
2082	Autosomal dominant epilepsy with auditory features: a new LGI1 family including a phenocopy with cortical dysplasia. <i>Journal of Neurology</i> , 2016, 263, 11-16.	1.8	10
2083	DNM1L-related mitochondrial fission defect presenting as refractory epilepsy. <i>European Journal of Human Genetics</i> , 2016, 24, 1084-1088.	1.4	113
2084	Analysis of the genetic variability in Parkinson's disease from Southern Spain. <i>Neurobiology of Aging</i> , 2016, 37, 210.e1-210.e5.	1.5	23
2085	Using Population Genetics to Interrogate the Monogenic Nephrotic Syndrome Diagnosis in a Case Cohort. <i>Journal of the American Society of Nephrology: JASN</i> , 2016, 27, 1970-1983.	3.0	41
2086	Search for the potential "second-hit" mechanism underlying the onset of familial hemophagocytic lymphohistiocytosis type 2 by whole-exome sequencing analysis. <i>Translational Research</i> , 2016, 170, 26-39.	2.2	10
2087	Multilocus Inherited Neoplasia Alleles Syndrome. <i>JAMA Oncology</i> , 2016, 2, 373.	3.4	43
2088	Association study of MMP8 gene in osteoarthritis. <i>Connective Tissue Research</i> , 2016, 57, 44-52.	1.1	16
2089	Clinical, biochemical and mutation profile in Indian patients with Sandhoff disease. <i>Journal of Human Genetics</i> , 2016, 61, 163-166.	1.1	7
2090	SIFT missense predictions for genomes. <i>Nature Protocols</i> , 2016, 11, 1-9.	5.5	1,053
2091	Mutation spectra of the ITGB2 gene in Iranian families with leukocyte adhesion deficiency type 1. <i>Human Immunology</i> , 2016, 77, 191-195.	1.2	23
2092	DIDA: A curated and annotated digenic diseases database. <i>Nucleic Acids Research</i> , 2016, 44, D900-D907.	6.5	84
2093	Exploring the complete mutational space of the LDL receptor LA5 domain using molecular dynamics: linking SNPs with disease phenotypes in familial hypercholesterolemia. <i>Human Molecular Genetics</i> , 2016, 25, 1233-1246.	1.4	9
2094	Functional cellular analyses reveal energy metabolism defect and mitochondrial DNA depletion in a case of mitochondrial aconitase deficiency. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 28-34.	0.5	32
2095	Characterization of two de novo KCNT1 mutations in children with malignant migrating partial seizures in infancy. <i>Molecular and Cellular Neurosciences</i> , 2016, 72, 54-63.	1.0	77
2096	Developing maps of fitness consequences for plant genomes. <i>Current Opinion in Plant Biology</i> , 2016, 30, 101-107.	3.5	13

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2097	Expansion of phenotype and genotypic data in CRB2-related syndrome. <i>European Journal of Human Genetics</i> , 2016, 24, 1436-1444.	1.4	36
2098	Perspectives in Polycystic Ovary Syndrome: From Hair to Eternity. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 759-768.	1.8	71
2099	Mutations in the mammalian target of rapamycin pathway regulators <i>NPRL2</i> and <i>NPRL3</i> cause focal epilepsy. <i>Annals of Neurology</i> , 2016, 79, 120-131.	2.8	190
2100	Unmasking targets of antitumor immunity via high-throughput antigen profiling. <i>Current Opinion in Biotechnology</i> , 2016, 42, 92-97.	3.3	9
2101	Identification and Functional Characterization of <i>CLCN1</i> Mutations Found in Nondystrophic Myotonia Patients. <i>Human Mutation</i> , 2016, 37, 74-83.	1.1	23
2102	The albinism of the feral Asinara white donkeys (<i>Equus asinus</i>) is determined by a missense mutation in a highly conserved position of the tyrosinase (<i>TYR</i>) gene deduced protein. <i>Animal Genetics</i> , 2016, 47, 120-124.	0.6	34
2103	Exome and genome sequencing: a revolution for the discovery and diagnosis of monogenic disorders. <i>Journal of Internal Medicine</i> , 2016, 279, 3-15.	2.7	71
2104	Whole-genome sequencing in multiplex families with psychoses reveals mutations in the <i>SHANK2</i> and <i>SMARCA1</i> genes segregating with illness. <i>Molecular Psychiatry</i> , 2016, 21, 1690-1695.	4.1	88
2105	Integrating population variation and protein structural analysis to improve clinical interpretation of missense variation: application to the WD40 domain. <i>Human Molecular Genetics</i> , 2016, 25, 927-935.	1.4	26
2106	Targeted resequencing identifies <i>TRPM4</i> as a major gene predisposing to progressive familial heart block type I. <i>International Journal of Cardiology</i> , 2016, 207, 349-358.	0.8	62
2107	Forty-eight novel mutations causing biotinidase deficiency. <i>Molecular Genetics and Metabolism</i> , 2016, 117, 369-372.	0.5	16
2108	De novo <i>PIK3R2</i> variant causes polymicrogyria, corpus callosum hyperplasia and focal cortical dysplasia. <i>European Journal of Human Genetics</i> , 2016, 24, 1359-1362.	1.4	26
2109	Whole-genome sequence analyses of Western Central African Pygmy hunter-gatherers reveal a complex demographic history and identify candidate genes under positive natural selection. <i>Genome Research</i> , 2016, 26, 279-290.	2.4	54
2110	Targeted resequencing identifies <i>PTCH1</i> as a major contributor to ocular developmental anomalies and extends the <i>SOX2</i> regulatory network. <i>Genome Research</i> , 2016, 26, 474-485.	2.4	37
2111	Analysis of <i>CH25H</i> in multiple sclerosis and neuromyelitis optica. <i>Journal of Neuroimmunology</i> , 2016, 291, 70-72.	1.1	21
2112	Genome-wide association study identifies multiple susceptibility loci for craniofacial microsomia. <i>Nature Communications</i> , 2016, 7, 10605.	5.8	40
2113	g2pDB: A Database Mapping Protein Post-Translational Modifications to Genomic Coordinates. <i>Journal of Proteome Research</i> , 2016, 15, 983-990.	1.8	5
2114	Influence of <i>TYK2</i> in systemic sclerosis susceptibility: a new locus in the IL-12 pathway. <i>Annals of the Rheumatic Diseases</i> , 2016, 75, 1521-1526.	0.5	41

#	ARTICLE	IF	CITATIONS
2115	Germline mutations in <i>PMS2</i> and <i>MLH1</i> in individuals with solitary loss of PMS2 expression in colorectal carcinomas from the Colon Cancer Family Registry Cohort. <i>BMJ Open</i> , 2016, 6, e010293.	0.8	33
2116	Next-Generation Sequencing for the Analysis of Cancer Specimens. , 2016, , 911-931.		0
2117	Polymorphism of 3' UTR of MAMLD1 gene is also associated with increased risk of isolated hypospadias in Indian children: a preliminary report. <i>Pediatric Surgery International</i> , 2016, 32, 515-524.	0.6	11
2118	Hotspot mutations in polyomavirus positive and negative Merkel cell carcinomas. <i>Cancer Genetics</i> , 2016, 209, 30-35.	0.2	26
2119	Effectiveness of whole-exome sequencing and costs of the traditional diagnostic trajectory in children with intellectual disability. <i>Genetics in Medicine</i> , 2016, 18, 949-956.	1.1	148
2120	Disturbed cofactor binding by a novel mutation in UDP-galactose 4-epimerase results in a type III galactosemia phenotype at birth. <i>RSC Advances</i> , 2016, 6, 17297-17301.	1.7	2
2121	Investigating the structural impact of S311C mutation in DRD2 receptor by molecular dynamics & docking studies. <i>Biochimie</i> , 2016, 123, 52-64.	1.3	10
2122	One SNP at a Time: Moving beyond GWAS in Psoriasis. <i>Journal of Investigative Dermatology</i> , 2016, 136, 567-573.	0.3	48
2123	Prominent scapulae mimicking an inherited myopathy expands the phenotype of CHD7-related disease. <i>European Journal of Human Genetics</i> , 2016, 24, 1216-1219.	1.4	2
2124	Discovery of a frameshift mutation in podocalyxin-like (PODXL) gene, coding for a neural adhesion molecule, as causal for autosomal-recessive juvenile Parkinsonism. <i>Journal of Medical Genetics</i> , 2016, 53, 450-456.	1.5	37
2125	Unbiased targeted next-generation sequencing molecular approach for primary immunodeficiency diseases. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 137, 1780-1787.	1.5	115
2126	Identification of six novel mutations in Iranian patients with maple syrup urine disease and their in silico analysis. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2016, 786, 34-40.	0.4	12
2127	DNAH11 Localization in the Proximal Region of Respiratory Cilia Defines Distinct Outer Dynein Arm Complexes. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2016, 55, 213-224.	1.4	107
2128	ECHS1 Deficiency as a Cause of Severe Neonatal Lactic Acidosis. <i>JIMD Reports</i> , 2016, 30, 33-37.	0.7	26
2129	Deficiency of Adenosine Deaminase 2 Causes Antibody Deficiency. <i>Journal of Clinical Immunology</i> , 2016, 36, 179-186.	2.0	78
2130	Leukocyte Adhesion Deficiency-I: Clinical and Molecular Characterization in an Indian Population. <i>Indian Journal of Pediatrics</i> , 2016, 83, 799-804.	0.3	14
2131	Common variation in BRCA1 may have a role in progression to lethal prostate cancer after radiation treatment. <i>Prostate Cancer and Prostatic Diseases</i> , 2016, 19, 197-201.	2.0	6
2132	From Genetics to Genomics: A Short Introduction for Pediatric Neurologists. <i>Neuropediatrics</i> , 2016, 47, 005-011.	0.3	1

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2133	In Silico Analysis of FMR1 Gene Missense SNPs. <i>Cell Biochemistry and Biophysics</i> , 2016, 74, 109-127.	0.9	10
2134	Identification of Susceptibility Loci and Genes for Colorectal Cancer Risk. <i>Gastroenterology</i> , 2016, 150, 1633-1645.	0.6	97
2135	Oligonucleotide-directed mutagenesis screen to identify pathogenic Lynch syndrome-associated MSH2 DNA mismatch repair gene variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 4128-4133.	3.3	28
2136	Congenital hypothyroidism and thyroid dysmorphogenesis: a case report of siblings with a newly identified mutation in thyroperoxidase. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2016, 29, 627-31.	0.4	6
2137	Germline BAP1 mutations misreported as somatic based on tumor-only testing. <i>Familial Cancer</i> , 2016, 15, 327-330.	0.9	13
2138	A Statistical Approach for Testing Cross-Phenotype Effects of Rare Variants. <i>American Journal of Human Genetics</i> , 2016, 98, 525-540.	2.6	75
2139	Genomic Analysis of <i>Salmonella enterica</i> Serovar Typhimurium Characterizes Strain Diversity for Recent U.S. Salmonellosis Cases and Identifies Mutations Linked to Loss of Fitness under Nitrosative and Oxidative Stress. <i>MBio</i> , 2016, 7, e00154.	1.8	26
2140	Role of Cytochrome P450 2C8 in Drug Metabolism and Interactions. <i>Pharmacological Reviews</i> , 2016, 68, 168-241.	7.1	175
2141	Novel Genetic Loci Associated With Retinal Microvascular Diameter. <i>Circulation: Cardiovascular Genetics</i> , 2016, 9, 45-54.	5.1	28
2142	Pathogenic and likely pathogenic variant prevalence among the first 10,000 patients referred for next-generation cancer panel testing. <i>Genetics in Medicine</i> , 2016, 18, 823-832.	1.1	227
2143	Next-generation sequencing of ABCA4: High frequency of complex alleles and novel mutations in patients with retinal dystrophies from Central Europe. <i>Experimental Eye Research</i> , 2016, 145, 93-99.	1.2	31
2144	Systems genetics identifies a convergent gene network for cognition and neurodevelopmental disease. <i>Nature Neuroscience</i> , 2016, 19, 223-232.	7.1	131
2145	Integrating 400 million variants from 80,000 human samples with extensive annotations: towards a knowledge base to analyze disease cohorts. <i>BMC Bioinformatics</i> , 2016, 17, 24.	1.2	13
2146	Landscape of somatic mutations in sporadic GH-secreting pituitary adenomas. <i>European Journal of Endocrinology</i> , 2016, 174, 363-372.	1.9	100
2147	The role of next-generation sequencing in understanding the genomic basis of diffuse large B cell lymphoma and advancing targeted therapies. <i>Expert Review of Hematology</i> , 2016, 9, 255-269.	1.0	12
2148	Machine Learning in Genomic Medicine: A Review of Computational Problems and Data Sets. <i>Proceedings of the IEEE</i> , 2016, 104, 176-197.	16.4	186
2149	Basic Bioinformatic Analyses of NGS Data. , 2016, , 19-37.		0
2150	Progression of neuropsychiatric and cognitive features due to exons 2 to 5 deletion in the epsilon-sarcoglycan gene: a case report. <i>Neurocase</i> , 2016, 22, 215-219.	0.2	4

#	ARTICLE	IF	CITATIONS
2151	<i>CYP2B6*6</i> and <i>CYP2B6*18</i> Predict Long-Term Efavirenz Exposure Measured in Hair Samples in HIV-Positive South African Women. <i>AIDS Research and Human Retroviruses</i> , 2016, 32, 529-538.	0.5	16
2152	Novel and recurrent AID mutations underlie prevalent autosomal recessive form of HIGM in consanguineous patients. <i>Immunogenetics</i> , 2016, 68, 19-28.	1.2	14
2153	Genetic screening of the makorin ring finger 3 gene in girls with idiopathic central precocious puberty. <i>Clinical Chemistry and Laboratory Medicine</i> , 2016, 54, e93-6.	1.4	14
2154	Spectrum of mutations in Glutaryl-CoA dehydrogenase gene in glutaric aciduria type I “ Study from South India. <i>Brain and Development</i> , 2016, 38, 54-60.	0.6	16
2155	Pilot whole-exome sequencing of a German early-onset Alzheimer's disease cohort reveals a substantial frequency of PSEN2 variants. <i>Neurobiology of Aging</i> , 2016, 37, 208.e11-208.e17.	1.5	38
2156	Detection of low-prevalence somatic TSC2 mutations in sporadic pulmonary lymphangioleiomyomatosis tissues by deep sequencing. <i>Human Genetics</i> , 2016, 135, 61-68.	1.8	16
2157	A novel common large genomic deletion and two new missense mutations identified in the Romanian phenylketonuria population. <i>Gene</i> , 2016, 576, 182-188.	1.0	12
2158	Concomitant partial exon skipping by a unique missense mutation of RPS6KA3 causes Coffin“Lowry syndrome. <i>Gene</i> , 2016, 575, 42-47.	1.0	8
2159	Candidate DNA repair susceptibility genes identified by exome sequencing in high-risk pancreatic cancer. <i>Cancer Letters</i> , 2016, 370, 302-312.	3.2	47
2160	AGXT and ERCC2 polymorphisms are associated with clinical outcome in metastatic colorectal cancer patients treated with 5-FU/oxaliplatin. <i>Pharmacogenomics Journal</i> , 2016, 16, 272-279.	0.9	16
2161	Exome sequencing identifies rare variants in multiple genes in atrioventricular septal defect. <i>Genetics in Medicine</i> , 2016, 18, 189-198.	1.1	39
2162	Adaptive Evolution Favoring KLK4 Downregulation in East Asians. <i>Molecular Biology and Evolution</i> , 2016, 33, 93-108.	3.5	5
2163	Gnaq: An ENU-Induced Mutant Allele Affecting Pigmentation in the Mouse. <i>Journal of Investigative Dermatology</i> , 2016, 136, 334-336.	0.3	1
2164	Identification of candidate genes for familial early-onset essential tremor. <i>European Journal of Human Genetics</i> , 2016, 24, 1009-1015.	1.4	36
2165	Unmasking a novel disease gene NEO1 associated with autism spectrum disorders by a hemizygous deletion on chromosome 15 and a functional polymorphism. <i>Behavioural Brain Research</i> , 2016, 300, 135-142.	1.2	11
2166	Dense genotyping of immune-related loci in idiopathic inflammatory myopathies confirms HLA alleles as the strongest genetic risk factor and suggests different genetic background for major clinical subgroups. <i>Annals of the Rheumatic Diseases</i> , 2016, 75, 1558-1566.	0.5	127
2168	Detection of Unknown Amino Acid Substitutions Using Error-Tolerant Database Search. <i>Methods in Molecular Biology</i> , 2016, 1362, 247-264.	0.4	3
2169	Matrix metalloproteinase genes on chromosome 11q22 and risk of carpal tunnel syndrome. <i>Rheumatology International</i> , 2016, 36, 413-419.	1.5	9

#	ARTICLE	IF	CITATIONS
2170	Bovine Genome Database: new tools for gleaning function from the <i>Bos taurus</i> genome. <i>Nucleic Acids Research</i> , 2016, 44, D834-D839.	6.5	87
2171	A targeted next-generation sequencing assay for the molecular diagnosis of genetic disorders with orodental involvement. <i>Journal of Medical Genetics</i> , 2016, 53, 98-110.	1.5	100
2172	Targeted massively parallel sequencing of a panel of putative breast cancer susceptibility genes in a large cohort of multiple-case breast and ovarian cancer families. <i>Journal of Medical Genetics</i> , 2016, 53, 34-42.	1.5	63
2173	Prioritization and burden analysis of rare variants in 208 candidate genes suggest they do not play a major role in CAKUT. <i>Kidney International</i> , 2016, 89, 476-486.	2.6	78
2174	Interpreting functional effects of coding variants: challenges in proteome-scale prediction, annotation and assessment. <i>Briefings in Bioinformatics</i> , 2016, 17, 841-862.	3.2	23
2175	Replication study of MATR3 in familial and sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2016, 37, 209.e17-209.e21.	1.5	53
2176	Analysis of <i>SLC16A11</i> Variants in 12,811 American Indians: Genotype-Obesity Interaction for Type 2 Diabetes and an Association With <i>RNASEK</i> Expression. <i>Diabetes</i> , 2016, 65, 510-519.	0.3	23
2177	Targeted next-generation sequencing of the ATP7B gene for molecular diagnosis of Wilson disease. <i>Clinical Biochemistry</i> , 2016, 49, 166-171.	0.8	16
2178	A novel mutation of GATA4 (K300T) associated with familial atrial septal defect. <i>Gene</i> , 2016, 575, 473-477.	1.0	34
2179	Deep Sequencing of Three Loci Implicated in Large-Scale Genome-Wide Association Study Smoking Meta-Analyses. <i>Nicotine and Tobacco Research</i> , 2016, 18, 626-631.	1.4	10
2180	Exome Sequence Data From Multigenerational Families Implicate AMPA Receptor Trafficking in Neurocognitive Impairment and Schizophrenia Risk. <i>Schizophrenia Bulletin</i> , 2016, 42, 288-300.	2.3	22
2181	Rare variants in tenascin genes in a cohort of children with primary vesicoureteric reflux. <i>Pediatric Nephrology</i> , 2016, 31, 247-253.	0.9	10
2182	TULP1 Missense Mutations Induces the Endoplasmic Reticulum Unfolded Protein Response Stress Complex (ER-UPR). <i>Advances in Experimental Medicine and Biology</i> , 2016, 854, 223-230.	0.8	7
2183	Parallelism and Epistasis in Skeletal Evolution Identified through Use of Phylogenomic Mapping Strategies. <i>Molecular Biology and Evolution</i> , 2016, 33, 162-173.	3.5	32
2184	Driver Gene and Novel Mutations in Asbestos-Exposed Lung Adenocarcinoma and Malignant Mesothelioma Detected by Exome Sequencing. <i>Lung</i> , 2016, 194, 125-135.	1.4	34
2185	Identification and Characterization of Mutations in the CLCN7 Gene in a Taiwanese Patient with Infantile Malignant Osteopetrosis. <i>Pediatrics and Neonatology</i> , 2016, 57, 155-157.	0.3	3
2186	Advances in computational approaches for prioritizing driver mutations and significantly mutated genes in cancer genomes. <i>Briefings in Bioinformatics</i> , 2016, 17, 642-656.	3.2	120
2187	High-Throughput Genetic Testing for Thrombotic Microangiopathies and C3 Glomerulopathies. <i>Journal of the American Society of Nephrology: JASN</i> , 2016, 27, 1245-1253.	3.0	89

#	ARTICLE	IF	CITATIONS
2188	Burden of Common Complex Disease Variants in the Exomes of Two Healthy Centenarian Brothers. <i>Gerontology</i> , 2016, 62, 58-62.	1.4	7
2189	POLE and POLD1 mutations in 529 kindred with familial colorectal cancer and/or polyposis: review of reported cases and recommendations for genetic testing and surveillance. <i>Genetics in Medicine</i> , 2016, 18, 325-332.	1.1	209
2190	Whole-genome sequencing identifies a novel ABCB7 gene mutation for X-linked congenital cerebellar ataxia in a large family of Mongolian ancestry. <i>European Journal of Human Genetics</i> , 2016, 24, 550-555.	1.4	28
2191	Comprehensive analysis via exome sequencing uncovers genetic etiology in autosomal recessive nonsyndromic deafness in a large multiethnic cohort. <i>Genetics in Medicine</i> , 2016, 18, 364-371.	1.1	124
2192	Genetic screening in sudden cardiac death in the young can save future lives. <i>International Journal of Legal Medicine</i> , 2016, 130, 59-66.	1.2	54
2193	Identification of rare variants in TNNI3 with atrial fibrillation in a Chinese GeneID population. <i>Molecular Genetics and Genomics</i> , 2016, 291, 79-92.	1.0	13
2194	Genes with de novo mutations are shared by four neuropsychiatric disorders discovered from NPdenovo database. <i>Molecular Psychiatry</i> , 2016, 21, 290-297.	4.1	167
2195	WHATIF: An open-source desktop application for extraction and management of the incidental findings from next-generation sequencing variant data. <i>Computers in Biology and Medicine</i> , 2016, 68, 165-169.	3.9	3
2196	A rare variant in MCF2L identified using exclusion linkage in a pedigree with premature atherosclerosis. <i>European Journal of Human Genetics</i> , 2016, 24, 86-91.	1.4	12
2197	Are all the previously reported genetic variants in limb girdle muscular dystrophy genes pathogenic?. <i>European Journal of Human Genetics</i> , 2016, 24, 73-77.	1.4	21
2198	A gain-of-function mutation in the cardiac pacemaker HCN4 channel increasing cAMP sensitivity is associated with familial Inappropriate Sinus Tachycardia. <i>European Heart Journal</i> , 2017, 38, 280-288.	1.0	73
2199	The OPA1 Gene Mutations Are Frequent in Han Chinese Patients with Suspected Optic Neuropathy. <i>Molecular Neurobiology</i> , 2017, 54, 1622-1630.	1.9	12
2200	Functional genomics of candidate genes derived from genome-wide association studies for five common neurological diseases. <i>International Journal of Neuroscience</i> , 2017, 127, 118-123.	0.8	14
2201	Diagnóstico molecular de cavernomatosis cerebral. <i>Neurología</i> , 2017, 32, 540-545.	0.3	7
2202	Mutation Analysis of Cell-Free DNA and Single Circulating Tumor Cells in Metastatic Breast Cancer Patients with High Circulating Tumor Cell Counts. <i>Clinical Cancer Research</i> , 2017, 23, 88-96.	3.2	186
2203	Expanding the genotypic spectrum of Perrault syndrome. <i>Clinical Genetics</i> , 2017, 91, 302-312.	1.0	68
2204	The novel <i>EDAR</i> p.L397H missense mutation causes autosomal dominant hypohidrotic ectodermal dysplasia. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2017, 31, e17-e20.	1.3	8
2205	Pure Cerebellar Ataxia with Homozygous Mutations in the PNPLA6 Gene. <i>Cerebellum</i> , 2017, 16, 262-267.	1.4	26

#	ARTICLE	IF	CITATIONS
2206	Mutation Screening of the CHCHD10 Gene in Chinese Patients with Amyotrophic Lateral Sclerosis. <i>Molecular Neurobiology</i> , 2017, 54, 3189-3194.	1.9	19
2207	Molecular Diagnostics of Gliomas Using Next Generation Sequencing of a Glioma-tailored Gene Panel. <i>Brain Pathology</i> , 2017, 27, 146-159.	2.1	130
2208	Prediction of functionally significant single nucleotide polymorphisms in <i>PTEN</i> tumor suppressor gene: An <i>in silico</i> approach. <i>Biotechnology and Applied Biochemistry</i> , 2017, 64, 657-666.	1.4	15
2209	Mutational analysis of <i>HOXA10</i> gene in Chinese patients with cryptorchidism. <i>Andrologia</i> , 2017, 49, e12592.	1.0	6
2210	Analysis of the FGF gene family provides insights into aquatic adaptation in cetaceans. <i>Scientific Reports</i> , 2017, 7, 40233.	1.6	21
2211	<i>PROM1</i> gene variations in Brazilian patients with macular dystrophy. <i>Ophthalmic Genetics</i> , 2017, 38, 39-42.	0.5	8
2212	RNF43 germline and somatic mutation in serrated neoplasia pathway and its association with BRAF mutation. <i>Gut</i> , 2017, 66, 1645-1656.	6.1	157
2213	Novel founder mutation in French-Canadian families with Naxos disease. <i>Clinical Genetics</i> , 2017, 92, 451-453.	1.0	14
2214	Update of the spectrum of mucopolysaccharidoses type III in Tunisia: identification of three novel mutations and <i>in silico</i> structural analysis of the missense mutations. <i>World Journal of Pediatrics</i> , 2017, 13, 374-380.	0.8	9
2215	Inflammatory pathway genes associated with inter-individual variability in the trajectories of morning and evening fatigue in patients receiving chemotherapy. <i>Cytokine</i> , 2017, 91, 187-210.	1.4	31
2216	Epilepsy in patients with <i>GRIN2A</i> alterations: Genetics, neurodevelopment, epileptic phenotype and response to anticonvulsive drugs. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 530-541.	0.7	37
2217	A human immunodeficiency syndrome caused by mutations in <i>CARMIL2</i> . <i>Nature Communications</i> , 2017, 8, 14209.	5.8	103
2218	Functional and clinical relevance of novel and known <i>PCSK1</i> variants for childhood obesity and glucose metabolism. <i>Molecular Metabolism</i> , 2017, 6, 295-305.	3.0	26
2219	Combined alpha-delta platelet storage pool deficiency is associated with mutations in <i>GFI1B</i> . <i>Molecular Genetics and Metabolism</i> , 2017, 120, 288-294.	0.5	22
2220	Mutations in <i>ATP6V1E1</i> or <i>ATP6V1A</i> Cause Autosomal-Recessive Cutis Laxa. <i>American Journal of Human Genetics</i> , 2017, 100, 216-227.	2.6	82
2221	Bundle Branch Re-Entrant Ventricular Tachycardia. <i>JACC: Clinical Electrophysiology</i> , 2017, 3, 276-288.	1.3	27
2222	Common Variable Immunodeficiency patients with a phenotypic profile of immunosenescence present with thrombocytopenia. <i>Scientific Reports</i> , 2017, 7, 39710.	1.6	31
2223	Identifying Novel Inborn Errors of the Immune System. , 2017, , .		0

#	ARTICLE	IF	CITATIONS
2224	Identification and functional characterization of a novel MTFMT mutation associated with selective vulnerability of the visual pathway and a mild neurological phenotype. <i>Neurogenetics</i> , 2017, 18, 97-103.	0.7	11
2225	Mutation screening of ACKR3 and COPS8 in kidney cancer cases from the CONFIRM study. <i>Familial Cancer</i> , 2017, 16, 411-416.	0.9	6
2226	Molecular genetics of familial hypercholesterolemia in Israel—revisited. <i>Atherosclerosis</i> , 2017, 257, 55-63.	0.4	19
2227	ARLTS1, potential candidate gene in familial aggregation of hematological malignancies. <i>Bulletin Du Cancer</i> , 2017, 104, 123-127.	0.6	10
2228	Structural analysis of pathogenic mutations in the <i>DYRK1A</i> gene in patients with developmental disorders. <i>Human Molecular Genetics</i> , 2017, 26, ddw409.	1.4	33
2229	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. <i>Nature Genetics</i> , 2017, 49, 249-255.	9.4	88
2230	JIMD Reports, Volume 31. <i>JIMD Reports</i> , 2017, . .	0.7	0
2231	A targeted next-generation sequencing in the molecular risk stratification of adult acute myeloid leukemia: implications for clinical practice. <i>Cancer Medicine</i> , 2017, 6, 349-360.	1.3	48
2232	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. <i>American Journal of Human Genetics</i> , 2017, 100, 334-342.	2.6	26
2233	Osteoporosis-pseudoglioma syndrome: Report of two cases and a manifesting carrier. <i>Ophthalmic Genetics</i> , 2017, 38, 473-479.	0.5	7
2235	Severe neurodegenerative disease in brothers with homozygous mutation in POLR1A. <i>European Journal of Human Genetics</i> , 2017, 25, 315-323.	1.4	23
2236	Absence of Hisheshi, a nuclear transporter for heat-shock protein HSP70, causes infantile hypomyelinating leukoencephalopathy. <i>European Journal of Human Genetics</i> , 2017, 25, 366-370.	1.4	11
2237	The ExAC browser: displaying reference data information from over 60 000 exomes. <i>Nucleic Acids Research</i> , 2017, 45, D840-D845.	6.5	587
2238	Biallelic <i>SCN10A</i> mutations in neuromuscular disease and epileptic encephalopathy. <i>Annals of Clinical and Translational Neurology</i> , 2017, 4, 26-35.	1.7	20
2239	Deleterious variants in Asian rice and the potential cost of domestication. <i>Molecular Biology and Evolution</i> , 2017, 34, msw296.	3.5	68
2240	In silico analysis of the deleterious nsSNPs (missense) in the homeobox domain of human <i>HOXB13</i> gene responsible for hereditary prostate cancer. <i>Chemical Biology and Drug Design</i> , 2017, 90, 188-199.	1.5	9
2241	InterVar: Clinical Interpretation of Genetic Variants by the 2015 ACMG-AMP Guidelines. <i>American Journal of Human Genetics</i> , 2017, 100, 267-280.	2.6	717
2242	NLRC5 polymorphism is associated with susceptibility to chronic periodontitis. <i>Immunobiology</i> , 2017, 222, 704-708.	0.8	17

#	ARTICLE	IF	CITATIONS
2243	Axial spondylometaphyseal dysplasia is also caused by NEK1 mutations. <i>Journal of Human Genetics</i> , 2017, 62, 503-506.	1.1	25
2244	Next Generation Sequencing of Circulating Cell-Free DNA for Evaluating Mutations and Gene Amplification in Metastatic Breast Cancer. <i>Clinical Chemistry</i> , 2017, 63, 532-541.	1.5	81
2245	A de novo mutation in the X-linked PAK3 gene is the underlying cause of intellectual disability and macrocephaly in monozygotic twins. <i>European Journal of Medical Genetics</i> , 2017, 60, 212-216.	0.7	17
2246	JAK1 gain-of-function causes an autosomal dominant immune dysregulatory and hypereosinophilic syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 2016-2020.e5.	1.5	101
2247	A low-frequency variant in SMAD7 modulates TGF β ² signaling and confers risk for colorectal cancer in Chinese population. <i>Molecular Carcinogenesis</i> , 2017, 56, 1798-1807.	1.3	48
2248	Systematic screening for <i>CYP3A4</i> genetic polymorphisms in a Han Chinese population. <i>Pharmacogenomics</i> , 2017, 18, 369-379.	0.6	51
2249	Characterization of BRCA1 and BRCA2 variants in multi-ethnic Asian cohort from a Malaysian case-control study. <i>BMC Cancer</i> , 2017, 17, 149.	1.1	5
2250	Exome sequencing identifies SLC26A4, GJB2, SCARB2 and DUOX2 mutations in 2 siblings with Pendred syndrome in a Malaysian family. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 40.	1.2	7
2251	Autosomal dominant cutis laxa with progeroid features due to a novel, de novo mutation in ALDH18A1. <i>Journal of Human Genetics</i> , 2017, 62, 661-663.	1.1	12
2252	A novel <i>DNAJB6</i> mutation causes dominantly inherited distal-onset myopathy and compromises <i>DNAJB6</i> function. <i>Clinical Genetics</i> , 2017, 92, 150-157.	1.0	16
2253	Assessment of plasma chitotriosidase activity, CCL18/PARC concentration and NP-C suspicion index in the diagnosis of Niemann-Pick disease type C: a prospective observational study. <i>Journal of Translational Medicine</i> , 2017, 15, 43.	1.8	19
2254	Lung disease caused by <i>ABCA3</i> mutations. <i>Thorax</i> , 2017, 72, 213-220.	2.7	110
2255	Variant discovery in the sheep milk transcriptome using RNA sequencing. <i>BMC Genomics</i> , 2017, 18, 170.	1.2	44
2256	Genetic analysis of impaired trimethylamine metabolism using whole exome sequencing. <i>BMC Medical Genetics</i> , 2017, 18, 11.	2.1	9
2257	GAVIN: Gene-Aware Variant INTERpretation for medical sequencing. <i>Genome Biology</i> , 2017, 18, 6.	3.8	55
2258	A girl with developmental delay, ataxia, cranial nerve palsies, severe respiratory problems in infancy—Expanding NDST1 syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 712-715.	0.7	14
2259	Transmembrane Domain Single-Nucleotide Polymorphisms Impair Expression and Transport Activity of ABC Transporter ABCG2. <i>Pharmaceutical Research</i> , 2017, 34, 1626-1636.	1.7	24
2260	Functional annotation of sixty-five type-2 diabetes risk SNPs and its application in risk prediction. <i>Scientific Reports</i> , 2017, 7, 43709.	1.6	4

#	ARTICLE	IF	CITATIONS
2261	Molecular Analysis of Twelve Pakistani Families with Nonsyndromic or Syndromic Hearing Loss. Genetic Testing and Molecular Biomarkers, 2017, 21, 316-321.	0.3	13
2262	A New <i>COL3A1</i> Mutation in Ehlers-Danlos Syndrome Vascular Type With Different Phenotypes in the Same Family. Vascular and Endovascular Surgery, 2017, 51, 141-145.	0.3	11
2263	Impacts of Neanderthal-Introgressed Sequences on the Landscape of Human Gene Expression. Cell, 2017, 168, 916-927.e12.	13.5	136
2264	Frequency, spectrum, and functional significance of TP53 mutations in patients with diffuse large B-cell lymphoma. Molecular Biology, 2017, 51, 53-60.	0.4	9
2265	Strain-specific single-nucleotide polymorphisms in hypertensive ISIAH rats. Biochemistry (Moscow), 2017, 82, 224-235.	0.7	4
2266	Mutation in mitochondrial complex IV subunit COX5A causes pulmonary arterial hypertension, lactic acidemia, and failure to thrive. Human Mutation, 2017, 38, 692-703.	1.1	32
2267	Identification of a genetically defined ultra-high-risk group in relapsed pediatric T-lymphoblastic leukemia. Blood Cancer Journal, 2017, 7, e523-e523.	2.8	69
2268	De novo and rare mutations in the HSPA1L heat shock gene associated with inflammatory bowel disease. Genome Medicine, 2017, 9, 8.	3.6	27
2269	Compound heterozygous <i>POMT1</i> mutations in a Chinese family with autosomal recessive muscular dystrophy/dystroglycanopathy C1. Journal of Cellular and Molecular Medicine, 2017, 21, 1388-1393.	1.6	20
2270	<i>RIC3</i> variants are not associated with Parkinson's disease in French-Canadians and French. Neurobiology of Aging, 2017, 53, 194.e9-194.e11.	1.5	5
2271	Structural and functional impacts of amino acid substitutions that create blood group antigens: implications for immunogenicity. Transfusion, 2017, 57, 541-553.	0.8	6
2272	<i>FOXP2</i> variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum. Journal of Medical Genetics, 2017, 54, 64-72.	1.5	67
2273	Rapid and cost-effective high-throughput sequencing for identification of germline mutations of BRCA1 and BRCA2. Journal of Human Genetics, 2017, 62, 561-567.	1.1	17
2274	De novo missense variants in <i>HECW2</i> are associated with neurodevelopmental delay and hypotonia. Journal of Medical Genetics, 2017, 54, 84-86.	1.5	46
2275	<i>SLC13A5</i> is the second gene associated with Kohlschütter-Janz syndrome. Journal of Medical Genetics, 2017, 54, 54-62.	1.5	45
2276	The landscape of sex-differential transcriptome and its consequent selection in human adults. BMC Biology, 2017, 15, 7.	1.7	212
2277	Somatic MAP2K1 Mutations Are Associated with Extracranial Arteriovenous Malformation. American Journal of Human Genetics, 2017, 100, 546-554.	2.6	215
2278	Two cases of Legg-Perthes and intellectual disability in Tricho-Rhino-Phalangeal syndrome type 1 associated with novel <i>TRPS1</i> mutations. American Journal of Medical Genetics, Part A, 2017, 173, 1663-1667.	0.7	4

#	ARTICLE	IF	CITATIONS
2279	Genome-wide analysis of differential RNA editing in epilepsy. <i>Genome Research</i> , 2017, 27, 440-450.	2.4	73
2280	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. <i>Nature Neuroscience</i> , 2017, 20, 602-611.	7.1	691
2281	Common coding variant in <i>SERPINA1</i> increases the risk for large artery stroke. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, 3613-3618.	3.3	46
2282	Aminoglycoside resistance of <i>Pseudomonas aeruginosa</i> in cystic fibrosis results from convergent evolution in the <i>mexZ</i> gene. <i>Thorax</i> , 2017, 72, 40-47.	2.7	49
2283	X-linked primary ciliary dyskinesia due to mutations in the cytoplasmic axonemal dynein assembly factor PIH1D3. <i>Nature Communications</i> , 2017, 8, 14279.	5.8	133
2284	Impact of ERG3 mutations and expression of ergosterol genes controlled by UPC2 and NDT80 in <i>Candida parapsilosis</i> azole resistance. <i>Clinical Microbiology and Infection</i> , 2017, 23, 575.e1-575.e8.	2.8	42
2285	Homozygous and compound heterozygous mutations in the <i>FBN1</i> gene: unexpected findings in molecular diagnosis of Marfan syndrome. <i>Journal of Medical Genetics</i> , 2017, 54, 100-103.	1.5	30
2286	Opportunities and challenges of whole-genome and -exome sequencing. <i>BMC Genetics</i> , 2017, 18, 14.	2.7	160
2287	A comprehensive computational study on pathogenic mis-sense mutations spanning the RING2 and REP domains of Parkin protein. <i>Gene</i> , 2017, 610, 49-58.	1.0	3
2288	Postmortem genetic testing should be recommended in sudden cardiac death cases due to thoracic aortic dissection. <i>International Journal of Legal Medicine</i> , 2017, 131, 1211-1219.	1.2	13
2289	Mutations in proteasome-related genes are associated with thyroid hemiagenesis. <i>Endocrine</i> , 2017, 56, 279-285.	1.1	18
2290	Melanoma during pregnancy: a report of 60 pregnancies complicated by melanoma. <i>Melanoma Research</i> , 2017, 27, 218-223.	0.6	16
2291	Does the distribution pattern of brain metastases during BRAF inhibitor therapy reflect phenotype switching?. <i>Melanoma Research</i> , 2017, 27, 231-237.	0.6	15
2292	Molecular outcomes, clinical consequences, and genetic diagnosis of Oculocutaneous Albinism in Pakistani population. <i>Scientific Reports</i> , 2017, 7, 44185.	1.6	25
2293	Identification of a novel missense mutation in <i>FGFR3</i> gene in an Iranian family with LADD syndrome by Next-Generation Sequencing. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2017, 97, 192-196.	0.4	12
2294	Association between the use of surveillance PET/CT and the detection of potentially salvageable occult recurrences among patients with resected high-risk melanoma. <i>Melanoma Research</i> , 2017, 27, 335-341.	0.6	27
2295	Psychological characteristics of early-stage melanoma patients: a cross-sectional study on 204 patients. <i>Melanoma Research</i> , 2017, 27, 277-280.	0.6	17
2296	Biotin-thiamine responsive basal ganglia disease: Identification of a pyruvate peak on brain spectroscopy, novel mutation in <i>SLC19A3</i> , and calculation of prevalence based on allele frequencies from aggregated next-generation sequencing data. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1502-1513.	0.7	15

#	ARTICLE	IF	CITATIONS
2297	Genetic and phenotypic characteristics of four Chinese families with fundus albipunctatus. <i>Scientific Reports</i> , 2017, 7, 46285.	1.6	18
2298	Sudden death due to catecholaminergic polymorphic ventricular tachycardia following negative stress-test outcome: genetics and clinical implications. <i>Forensic Science, Medicine, and Pathology</i> , 2017, 13, 217-225.	0.6	5
2299	Prediction of a highly deleterious mutation E17K in AKT-1 gene: An in silico approach. <i>Biochemistry and Biophysics Reports</i> , 2017, 10, 260-266.	0.7	7
2300	Mutations in <i>TGDS</i> associated with additional malformations of the middle fingers and halluces: Atypical Catelâ€“Manzke syndrome in a fetus. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1694-1697.	0.7	7
2301	Critical points of T1 stage in primary melanoma. <i>Melanoma Research</i> , 2017, 27, 399-399.	0.6	2
2302	Whole-Exome Sequencing of Congenital Glaucoma Patients Reveals Hypermorphic Variants in GPATCH3, a New Gene Involved in Ocular and Craniofacial Development. <i>Scientific Reports</i> , 2017, 7, 46175.	1.6	22
2303	Delivering Clinical Grade Sequencing and Genetic Test Interpretation for Cardiovascular Medicine. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	11
2304	Cassava haplotype map highlights fixation of deleterious mutations during clonal propagation. <i>Nature Genetics</i> , 2017, 49, 959-963.	9.4	208
2305	A case of splenomegaly in CBL syndrome. <i>European Journal of Medical Genetics</i> , 2017, 60, 374-379.	0.7	10
2306	PET/CT surveillance detects asymptomatic recurrences in stage IIIB and IIIC melanoma patients: a prospective cohort study. <i>Melanoma Research</i> , 2017, 27, 251-257.	0.6	16
2307	BRAF inhibitor discontinuation and rechallenge in advanced melanoma patients with a complete initial treatment response. <i>Melanoma Research</i> , 2017, 27, 281-287.	0.6	25
2308	<i>PIK3CA</i> Mutations Contribute to Acquired Cetuximab Resistance in Patients with Metastatic Colorectal Cancer. <i>Clinical Cancer Research</i> , 2017, 23, 4602-4616.	3.2	72
2309	Combating mutations in genetic disease and drug resistance: understanding molecular mechanisms to guide drug design. <i>Expert Opinion on Drug Discovery</i> , 2017, 12, 553-563.	2.5	26
2310	Nextâ€“generation sequencing targeted disease panel in rodâ€“cone retinal dystrophies in M“ori and Polynesian reveals novel changes and a common founder mutation. <i>Clinical and Experimental Ophthalmology</i> , 2017, 45, 901-910.	1.3	15
2311	Functional implications of Neandertal introgression in modern humans. <i>Genome Biology</i> , 2017, 18, 61.	3.8	81
2312	A common NHE3 single-nucleotide polymorphism has normal function and sensitivity to regulatory ligands. <i>American Journal of Physiology - Renal Physiology</i> , 2017, 313, G129-G137.	1.6	4
2313	Determination of disease phenotypes and pathogenic variants from exome sequence data in the CAGI 4 gene panel challenge. <i>Human Mutation</i> , 2017, 38, 1201-1216.	1.1	5
2314	Next Generation Sequencing Based Clinical Molecular Diagnosis of Human Genetic Disorders. , 2017, , .		2

#	ARTICLE	IF	CITATIONS
2315	VarScan2 analysis of de novo variants in monozygotic twins discordant for schizophrenia. <i>Psychiatric Genetics</i> , 2017, 27, 62-70.	0.6	29
2316	Targeted sequencing identifies novel variants involved in autosomal recessive hereditary hearing loss in Qatari families. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2017, 800-802, 29-36.	0.4	23
2317	A Novel Kleefstra Syndrome-associated Variant That Affects the Conserved TPLX Motif within the Ankyrin Repeat of EHMT1 Leads to Abnormal Protein Folding. <i>Journal of Biological Chemistry</i> , 2017, 292, 3866-3876.	1.6	18
2318	<i>ADAMTSL4</i> assessment in ectopia lentis reveals a recurrent founder mutation in Polynesians. <i>Ophthalmic Genetics</i> , 2017, 38, 537-543.	0.5	10
2319	Next-Generation Sequencing Reveals Novel Mutations in X-linked Intellectual Disability. <i>OMICS A Journal of Integrative Biology</i> , 2017, 21, 295-303.	1.0	34
2320	Mutation of the Planar Cell Polarity Gene <i>VANGL1</i> in Adolescent Idiopathic Scoliosis. <i>Spine</i> , 2017, 42, E702-E707.	1.0	16
2321	Epidermolytic Ichthyosis Sine Epidermolysis. <i>American Journal of Dermatopathology</i> , 2017, 39, 440-444.	0.3	11
2322	Correlation of <i>FCGRT</i> genomic structure with serum immunoglobulin, albumin and farletuzumab pharmacokinetics in patients with first relapsed ovarian cancer. <i>Genomics</i> , 2017, 109, 251-257.	1.3	10
2323	Identification of Somatic Mutations in Primary Cutaneous Diffuse Large B-Cell Lymphoma, Leg Type by Massive Parallel Sequencing. <i>Journal of Investigative Dermatology</i> , 2017, 137, 1984-1994.	0.3	93
2324	Sirtuin 2 mutations in human cancers impair its function in genome maintenance. <i>Journal of Biological Chemistry</i> , 2017, 292, 9919-9931.	1.6	29
2325	Mutational screening of <i>SLC39A5</i> , <i>LEPREL1</i> and <i>LRPAP1</i> in a cohort of 187 high myopia patients. <i>Scientific Reports</i> , 2017, 7, 1120.	1.6	21
2326	Expanding the phenotypic spectrum of <i>GABRG2</i> variants: a recurrent <i>GABRG2</i> missense variant associated with a severe phenotype. <i>Journal of Neurogenetics</i> , 2017, 31, 30-36.	0.6	11
2327	Recessive <i>TAF1A</i> mutations reveal ribosomopathy in siblings with end-stage pediatric dilated cardiomyopathy. <i>Human Molecular Genetics</i> , 2017, 26, 2874-2881.	1.4	23
2328	Genomic Characterization of Low- and High-Grade Pancreatic Mucinous Cystic Neoplasms Reveals Recurrent <i>KRAS</i> Alterations in High-Risk Lesions. <i>Pancreas</i> , 2017, 46, 665-671.	0.5	29
2329	Whole-genome sequencing identifies homozygous <i>BRCA2</i> deletion guiding treatment in dedifferentiated prostate cancer. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a001362.	0.5	9
2330	A novel molecular diagnostics platform for somatic and germline precision oncology. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 336-359.	0.6	12
2331	Molecular Genetic Testing and the Future of Clinical Genomics. , 2017, , 263-282.		1
2332	Variable Susceptibility to Cigarette Smoke-Induced Emphysema in 34 Inbred Strains of Mice Implicates <i>Abi3bp</i> in Emphysema Susceptibility. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2017, 57, 367-375.	1.4	22

#	ARTICLE	IF	CITATIONS
2333	Identification of Sequence Variants within Experimentally Validated Protein Interaction Sites Provides New Insights into Molecular Mechanisms of Disease Development. <i>Molecular Informatics</i> , 2017, 36, 1700017.	1.4	4
2334	Novel neuro-audiological findings and further evidence for TWNK involvement in Perrault syndrome. <i>Journal of Translational Medicine</i> , 2017, 15, 25.	1.8	36
2335	Functional Investigations of <i>HNF1A</i> Identify Rare Variants as Risk Factors for Type 2 Diabetes in the General Population. <i>Diabetes</i> , 2017, 66, 335-346.	0.3	54
2336	Ensembl 2017. <i>Nucleic Acids Research</i> , 2017, 45, D635-D642.	6.5	535
2337	Lacrimal Gland Involvement in Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome. <i>Ophthalmology</i> , 2017, 124, 399-406.	2.5	12
2338	Whole exome sequencing of thymic neuroendocrine tumor with ectopic ACTH syndrome. <i>European Journal of Endocrinology</i> , 2017, 176, 187-194.	1.9	10
2339	Role of a heterotrimeric G α protein, Gi2, in the corticogenesis: possible involvement in periventricular nodular heterotopia and intellectual disability. <i>Journal of Neurochemistry</i> , 2017, 140, 82-95.	2.1	13
2340	Novel Zinc Finger Protein Gene 469 (ZNF469) Variants in Advanced Keratoconus. <i>Current Eye Research</i> , 2017, 42, 1396-1400.	0.7	16
2341	Identification of rare nonsynonymous variants in SYNE1/CPG2 in bipolar affective disorder. <i>Psychiatric Genetics</i> , 2017, 27, 81-88.	0.6	6
2342	Colorectal Cancer Cell Line Proteomes Are Representative of Primary Tumors and Predict Drug Sensitivity. <i>Gastroenterology</i> , 2017, 153, 1082-1095.	0.6	55
2343	Exploring the global landscape of genetic variation in coagulation factor XI deficiency. <i>Blood</i> , 2017, 130, e1-e6.	0.6	41
2344	The phenotypic spectrum of ARHGEF9 includes intellectual disability, focal epilepsy and febrile seizures. <i>Journal of Neurology</i> , 2017, 264, 1421-1425.	1.8	20
2345	Structural insights into the function of ZRANB3 in replication stress response. <i>Nature Communications</i> , 2017, 8, 15847.	5.8	41
2346	Exome sequencing identifies a novel UNC5D mutation in a severe myopic anisometropia family. <i>Medicine (United States)</i> , 2017, 96, e7138.	0.4	8
2347	<i>WT1</i> Haploinsufficiency Supports Milder Renal Manifestation in Two Patients with Denys-Drash Syndrome. <i>Sexual Development</i> , 2017, 11, 34-39.	1.1	5
2348	Common sequence variants affect molecular function more than rare variants?. <i>Scientific Reports</i> , 2017, 7, 1608.	1.6	20
2349	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. <i>Neurobiology of Aging</i> , 2017, 57, 247.e9-247.e13.	1.5	108
2350	A genome-wide association meta-analysis on lipoprotein (a) concentrations adjusted for apolipoprotein (a) isoforms. <i>Journal of Lipid Research</i> , 2017, 58, 1834-1844.	2.0	114

#	ARTICLE	IF	CITATIONS
2351	CAGI4 SickKids clinical genomes challenge: A pipeline for identifying pathogenic variants. <i>Human Mutation</i> , 2017, 38, 1169-1181.	1.1	11
2352	Induced pluripotent stem cell modelling of HLHS underlines the contribution of dysfunctional NOTCH signalling to impaired cardiogenesis. <i>Human Molecular Genetics</i> , 2017, 26, 3031-3045.	1.4	56
2353	GeMSTONE: orchestrated prioritization of human germline mutations in the cloud. <i>Nucleic Acids Research</i> , 2017, 45, W207-W214.	6.5	2
2354	Computational Investigation of Growth Hormone Receptor Trp169Arg Heterozygous Mutation in a Child With Short Stature. <i>Journal of Cellular Biochemistry</i> , 2017, 118, 4762-4771.	1.2	8
2355	Comparison and optimization of in silico algorithms for predicting the pathogenicity of sodium channel variants in epilepsy. <i>Epilepsia</i> , 2017, 58, 1190-1198.	2.6	14
2356	Molecular and functional characterization of the BMPR2 gene in Pulmonary Arterial Hypertension. <i>Scientific Reports</i> , 2017, 7, 1923.	1.6	16
2357	<i>CNTNAP1</i> mutations cause CNS hypomyelination and neuropathy with or without arthrogyriposis. <i>Neurology: Genetics</i> , 2017, 3, e144.	0.9	24
2358	The Phenotypic Spectrum of Nephropathies Associated with Mutations in Diacylglycerol Kinase β . <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 3066-3075.	3.0	50
2359	CAGI4 Crohn's exome challenge: Marker SNP versus exome variant models for assigning risk of Crohn disease. <i>Human Mutation</i> , 2017, 38, 1225-1234.	1.1	15
2360	Exome Sequencing in the Clinical Setting. , 2017, , 305-320.		0
2361	Germline Genetic Variants and Lung Cancer Survival in African Americans. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2017, 26, 1288-1295.	1.1	7
2362	al mena: a comprehensive resource of human genetic variants integrating genomes and exomes from Arab, Middle Eastern and North African populations. <i>Journal of Human Genetics</i> , 2017, 62, 889-894.	1.1	26
2363	Frequent <i>COL4A3BP</i> mutations in familial microhematuria accompanied by later-onset Alport nephropathy due to focal segmental glomerulosclerosis. <i>Clinical Genetics</i> , 2017, 92, 517-527.	1.0	35
2364	Infection Exposure Promotes <i>ETV6-RUNX1</i> Precursor B-cell Leukemia via Impaired H3K4 Demethylases. <i>Cancer Research</i> , 2017, 77, 4365-4377.	0.4	76
2365	Somatic mutations in clonally expanded cytotoxic T lymphocytes in patients with newly diagnosed rheumatoid arthritis. <i>Nature Communications</i> , 2017, 8, 15869.	5.8	83
2366	Mutation Spectrum in the CACNA1A Gene in 49 Patients with Episodic Ataxia. <i>Scientific Reports</i> , 2017, 7, 2514.	1.6	36
2367	A <i>MUTYH</i> germline mutation is associated with small intestinal neuroendocrine tumors. <i>Endocrine-Related Cancer</i> , 2017, 24, 427-443.	1.6	49
2368	The detection of a novel insertion mutation in exon 2 of the MEFV gene associated with familial mediterranean fever in a moroccan family. <i>Human Genome Variation</i> , 2017, 4, 17023.	0.4	1

#	ARTICLE	IF	CITATIONS
2369	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a <i>TUBB3</i> mutation. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a000984.	0.5	18
2370	Modeling gene regulation from paired expression and chromatin accessibility data. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, E4914-E4923.	3.3	167
2371	ProtVista: visualization of protein sequence annotations. <i>Bioinformatics</i> , 2017, 33, 2040-2041.	1.8	58
2372	Loss-of-function mutations in the <i>ATP13A2/PARK9</i> gene cause complicated hereditary spastic paraplegia (SPG78). <i>Brain</i> , 2017, 140, 287-305.	3.7	135
2373	Genetics of Genome-Wide Recombination Rate Evolution in Mice from an Isolated Island. <i>Genetics</i> , 2017, 206, 1841-1852.	1.2	13
2374	Genome-wide association study identifies multiple risk loci for renal cell carcinoma. <i>Nature Communications</i> , 2017, 8, 15724.	5.8	106
2375	Semi-automated cancer genome analysis using high-performance computing. <i>Human Mutation</i> , 2017, 38, 1325-1335.	1.1	9
2376	<i>RXR</i> B Is an MHC-Encoded Susceptibility Gene Associated with Anti-Topoisomerase γ Antibody-Positive Systemic Sclerosis. <i>Journal of Investigative Dermatology</i> , 2017, 137, 1878-1886.	0.3	3
2377	Functional analysis by minigene assay of putative splicing variants found in Bardet-Biedl syndrome patients. <i>Journal of Cellular and Molecular Medicine</i> , 2017, 21, 2268-2275.	1.6	5
2378	Biallelic Mutations in <i>CFAP43</i> and <i>CFAP44</i> Cause Male Infertility with Multiple Morphological Abnormalities of the Sperm Flagella. <i>American Journal of Human Genetics</i> , 2017, 100, 854-864.	2.6	220
2379	3-Hydroxy-3-methylglutaryl-coenzyme A lyase deficiency: Clinical presentation and outcome in a series of 37 patients. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 206-215.	0.5	32
2380	Protein-altering and regulatory genetic variants near <i>GATA4</i> implicated in bicuspid aortic valve. <i>Nature Communications</i> , 2017, 8, 15481.	5.8	90
2381	Express with caution: Epitope tags and cDNA variants effects on hERG channel trafficking, half-life and function. <i>Journal of Cardiovascular Electrophysiology</i> , 2017, 28, 1070-1082.	0.8	7
2382	<i>ACSS2</i> gene variant associated with cleft lip and palate in two independent Hispanic populations. <i>Laryngoscope</i> , 2017, 127, E336-E339.	1.1	2
2383	Whole-exome sequencing identifies recurrent <i>SF3B1</i> R625 mutation and comutation of <i>NF1</i> and <i>KIT</i> in mucosal melanoma. <i>Melanoma Research</i> , 2017, 27, 189-199.	0.6	121
2384	iSeq: A New Double-Barcode Method for Detecting Dynamic Genetic Interactions in Yeast. <i>G3: Genes, Genomes, Genetics</i> , 2017, 7, 143-153.	0.8	32
2385	Clinical and Genetic Diagnosis of Nonischemic Sudden Cardiac Death. <i>Revista Espanola De Cardiologia (English Ed)</i> , 2017, 70, 808-816.	0.4	8
2386	Compound heterozygous <i>KCNQ1</i> mutations (A300T/P535T) in a child with sudden unexplained death: Insights into possible molecular mechanisms based on protein modeling. <i>Gene</i> , 2017, 627, 40-48.	1.0	7

#	ARTICLE	IF	CITATIONS
2387	Whole-genome sequencing of monozygotic twins discordant for schizophrenia indicates multiple genetic risk factors for schizophrenia. <i>Journal of Genetics and Genomics</i> , 2017, 44, 295-306.	1.7	36
2388	Identification of candidate genes involved in the etiology of sporadic Tourette syndrome by exome sequencing. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2017, 174, 712-723.	1.1	17
2389	Unravelling the genetics of inherited retinal dystrophies: Past, present and future. <i>Progress in Retinal and Eye Research</i> , 2017, 59, 53-96.	7.3	85
2390	A Computational Approach to Identify a Potential Alternative Drug With Its Positive Impact Toward PMP22. <i>Journal of Cellular Biochemistry</i> , 2017, 118, 3730-3743.	1.2	9
2391	PNPLA1 defects in patients with autosomal recessive congenital ichthyosis and KO mice sustain PNPLA1 irreplaceable function in epidermal omega-O-acylceramide synthesis and skin permeability barrier. <i>Human Molecular Genetics</i> , 2017, 26, 1787-1800.	1.4	47
2392	Replicated evidence for aminoacylase 3 and nephrin gene variations to predict antihypertensive drug responses. <i>Pharmacogenomics</i> , 2017, 18, 445-458.	0.6	18
2393	Molecular diagnosis in children with fractures but no extraskeletal signs of osteogenesis imperfecta. <i>Osteoporosis International</i> , 2017, 28, 2095-2101.	1.3	29
2394	Diagn�stico cl�nico y gen�tico de la muerte s�bita cardiaca de origen no isqu�mico. <i>Revista Espanola De Cardiologia</i> , 2017, 70, 808-816.	0.6	9
2395	Malignant hyperthermia susceptibility in patients with exertional rhabdomyolysis: a retrospective cohort study and updated systematic review. <i>Canadian Journal of Anaesthesia</i> , 2017, 64, 736-743.	0.7	31
2396	Whole exome sequencing with genomic triangulation implicates <i>CDH2</i> -encoded N-cadherin as a novel pathogenic substrate for arrhythmogenic cardiomyopathy. <i>Congenital Heart Disease</i> , 2017, 12, 226-235.	0.0	46
2397	Identification of new BMP6 pro-peptide mutations in patients with iron overload. <i>American Journal of Hematology</i> , 2017, 92, 562-568.	2.0	35
2398	In silico prediction of the effects of mutations in the human triose phosphate isomerase gene: Towards a predictive framework for TPI deficiency. <i>European Journal of Medical Genetics</i> , 2017, 60, 289-298.	0.7	7
2399	Breast ductal carcinoma in situ carry mutational driver events representative of invasive breast cancer. <i>Modern Pathology</i> , 2017, 30, 952-963.	2.9	50
2400	Successful Application of Whole Genome Sequencing in a Medical Genetics Clinic. <i>Journal of Pediatric Genetics</i> , 2017, 06, 061-076.	0.3	54
2401	In silico analysis of non-synonymous single nucleotide polymorphisms in human <i>DAZL</i> gene associated with male infertility. <i>Systems Biology in Reproductive Medicine</i> , 2017, 63, 248-258.	1.0	23
2402	Novel de novo variant in <i>EBF3</i> is likely to impact DNA binding in a patient with a neurodevelopmental disorder and expanded phenotypes: patient report, in silico functional assessment, and review of published cases. <i>Journal of Physical Education and Sports Management</i> , 2017. 3. a001743.	0.5	22
2403	Screening of the <i>Filamin C</i> Gene in a Large Cohort of Hypertrophic Cardiomyopathy Patients. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	68
2404	Short stature and hypoparathyroidism in a child with Kenny-Caffey syndrome type 2 due to a novel mutation in <i>FAM111A</i> gene. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2017, 2017, 1.	1.6	27

#	ARTICLE	IF	CITATIONS
2405	Microtubule-associated defects caused by <i>EFHC1</i> mutations in juvenile myoclonic epilepsy. <i>Human Mutation</i> , 2017, 38, 816-826.	1.1	7
2406	Germline mutations in <i>ABL1</i> cause an autosomal dominant syndrome characterized by congenital heart defects and skeletal malformations. <i>Nature Genetics</i> , 2017, 49, 613-617.	9.4	40
2407	Progress towards precision functional genomics in cancer. <i>Current Opinion in Systems Biology</i> , 2017, 2, 74-83.	1.3	7
2408	In silico analysis of nonsynonymous single nucleotide polymorphisms of the human adiponectin receptor 2 (<i>ADIPOR2</i>) gene. <i>Computational Biology and Chemistry</i> , 2017, 68, 175-185.	1.1	20
2409	Clinical and Molecular Variability in Patients with <i>PHKA2</i> Variants and Liver Phosphorylase b Kinase Deficiency. <i>JIMD Reports</i> , 2017, 37, 63-72.	0.7	9
2410	Analysis of <i>SOD1</i> mutations in a Chinese population with amyotrophic lateral sclerosis: a case-control study and literature review. <i>Scientific Reports</i> , 2017, 7, 44606.	1.6	23
2411	The enigmatic oncogene and tumor suppressor-like properties of <i>RAD54B</i> : Insights into genome instability and cancer. <i>Genes Chromosomes and Cancer</i> , 2017, 56, 513-523.	1.5	16
2412	Nationwide genetic analysis for molecularly unresolved cystic fibrosis patients in a multiethnic society: implications for preconception carrier screening. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 223-236.	0.6	7
2413	Nonketotic hyperglycinemia: Functional assessment of missense variants in <i>GLDC</i> to understand phenotypes of the disease. <i>Human Mutation</i> , 2017, 38, 678-691.	1.1	13
2414	In vivo characterization of <i>CYP2D6</i> *12, *29 and *84 using dextromethorphan as a probe drug: a case report. <i>Pharmacogenomics</i> , 2017, 18, 427-431.	0.6	5
2415	<i>OPA1</i> analysis in an international series of probands with bilateral optic atrophy. <i>Acta Ophthalmologica</i> , 2017, 95, 363-369.	0.6	7
2416	Alternating Hemiplegia of Childhood as a New Presentation of Adenylate Cyclase 5-Mutation-Associated Disease: A Report of Two Cases. <i>Journal of Pediatrics</i> , 2017, 181, 306-308.e1.	0.9	24
2417	The Functional Effect of Rare Variants in Complement Genes on C3b Degradation in Patients With Age-Related Macular Degeneration. <i>JAMA Ophthalmology</i> , 2017, 135, 39.	1.4	48
2418	Sequential analysis of 18 genes in polycythemia vera and essential thrombocythemia reveals an association between mutational status and clinical outcome. <i>Genes Chromosomes and Cancer</i> , 2017, 56, 354-362.	1.5	12
2419	PERCH: A Unified Framework for Disease Gene Prioritization. <i>Human Mutation</i> , 2017, 38, 243-251.	1.1	119
2420	Loss of Nardilysin, a Mitochondrial Co-chaperone for α -Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. <i>Neuron</i> , 2017, 93, 115-131.	3.8	95
2421	Hemorheological alterations in sickle cell anemia and their clinical consequences – The role of genetic modulators. <i>Clinical Hemorheology and Microcirculation</i> , 2017, 64, 859-866.	0.9	5
2422	Comprehensive population-based genome sequencing provides insight into hematopoietic regulatory mechanisms. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, E327-E336.	3.3	39

#	ARTICLE	IF	CITATIONS
2423	Exome sequencing provides additional evidence for the involvement of <i>ARHGAP29</i> in Mendelian orofacial clefting and extends the phenotypic spectrum to isolated cleft palate. <i>Birth Defects Research</i> , 2017, 109, 27-37.	0.8	49
2424	In silico analysis for predicting pathogenicity of five unclassified mitochondrial DNA mutations associated with mitochondrial cytopathies' phenotypes. <i>European Journal of Medical Genetics</i> , 2017, 60, 172-177.	0.7	10
2425	Computational prediction of the effects of non-synonymous single nucleotide polymorphisms in the human Quinone Oxidoreductase 1 (NQO1). <i>Meta Gene</i> , 2017, 11, 127-135.	0.3	6
2426	Missense mutations of CACNA1A are a frequent cause of autosomal dominant nonprogressive congenital ataxia. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 450-456.	0.7	37
2427	Mutations in <i>DYNC2H1</i> , the cytoplasmic dynein 2, heavy chain 1 motor protein gene, cause short-rib polydactyly type I, Saldino-Noonan type. <i>Clinical Genetics</i> , 2017, 92, 158-165.	1.0	21
2428	Stargardt disease-associated mutation spectrum of a Russian Federation cohort. <i>European Journal of Medical Genetics</i> , 2017, 60, 140-147.	0.7	16
2429	Intensification: A Resource for Amplifying Population-Genetic Signals with Protein Repeats. <i>Journal of Molecular Biology</i> , 2017, 429, 435-445.	2.0	2
2430	TBCD may be a causal gene in progressive neurodegenerative encephalopathy with atypical infantile spinal muscular atrophy. <i>Journal of Human Genetics</i> , 2017, 62, 473-480.	1.1	15
2431	Flexible and Scalable Full-Length CYP2D6 Long Amplicon PacBio Sequencing. <i>Human Mutation</i> , 2017, 38, 310-316.	1.1	69
2432	Personalized Medicine Through Advanced Genomics. , 2017, , 31-48.		1
2433	Compound Heterozygosity for Null Mutations and a Common Hypomorphic Risk Haplotype in <i>TBX6</i> Causes Congenital Scoliosis. <i>Human Mutation</i> , 2017, 38, 317-323.	1.1	41
2434	The Rare-Variant Generalized Disequilibrium Test for Association Analysis of Nuclear and Extended Pedigrees with Application to Alzheimer Disease WGS Data. <i>American Journal of Human Genetics</i> , 2017, 100, 193-204.	2.6	26
2435	Thymidine kinase 2 and alanyl-tRNA synthetase 2 deficiencies cause lethal mitochondrial cardiomyopathy: case reports and review of the literature. <i>Cardiology in the Young</i> , 2017, 27, 936-944.	0.4	28
2436	Genome-wide association study of primary sclerosing cholangitis identifies new risk loci and quantifies the genetic relationship with inflammatory bowel disease. <i>Nature Genetics</i> , 2017, 49, 269-273.	9.4	230
2437	Whole-genome sequencing reveals a potential causal mutation for dwarfism in the Miniature Shetland pony. <i>Mammalian Genome</i> , 2017, 28, 143-151.	1.0	17
2438	Status and future perspectives of single nucleotide polymorphisms (SNPs) markers in farmed fishes: Way ahead using next generation sequencing. <i>Gene Reports</i> , 2017, 6, 81-86.	0.4	16
2439	Mutation screening of PLA2G6 in Japanese patients with early onset dystonia-parkinsonism. <i>Journal of Neural Transmission</i> , 2017, 124, 431-435.	1.4	12
2440	Late-onset hereditary hypophosphatemic rickets with hypercalciuria (HHRH) due to mutation of SLC34A3/NPT2c. <i>Bone</i> , 2017, 97, 15-19.	1.4	30

#	ARTICLE	IF	CITATIONS
2441	Novel Hypoglycemia Phenotype in Congenital Hyperinsulinism Due to Dominant Mutations of Uncoupling Protein 2. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 942-949.	1.8	36
2442	Signatures of Selection and Interspecies Introgression in the Genome of Chinese Domestic Pigs. <i>Genome Biology and Evolution</i> , 2017, 9, 2592-2603.	1.1	43
2443	Next-generation sequencing identifies three novel missense variants in ILDR1 and MYO6 genes in an Iranian family with hearing loss with review of the literature. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2017, 103, 103-108.	0.4	13
2444	Novel <i>NR2F1</i> variants likely disrupt DNA binding: molecular modeling in two cases, review of published cases, genotype-phenotype correlation, and phenotypic expansion of the Bosch-Boonstra-Schaaf optic atrophy syndrome. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a002162.	0.5	33
2445	MAPPIN: a method for annotating, predicting pathogenicity and mode of inheritance for nonsynonymous variants. <i>Nucleic Acids Research</i> , 2017, 45, 10393-10402.	6.5	15
2446	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. <i>American Journal of Human Genetics</i> , 2017, 101, 833-843.	2.6	56
2447	Genomic Triangulation and Coverage Analysis in Whole-Exome Sequencing-Based Molecular Autopsies. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	14
2448	Mutator genomes decay, despite sustained fitness gains, in a long-term experiment with bacteria. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, E9026-E9035.	3.3	87
2449	Whole exome sequencing in inborn errors of immunity: use the power but mind the limits. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2017, 17, 421-430.	1.1	12
2450	Novel pathogenic mutations in C1QTNF5 support a dominant negative disease mechanism in late-onset retinal degeneration. <i>Scientific Reports</i> , 2017, 7, 12147.	1.6	30
2451	Novel PANK2 mutation in the first Greek compound heterozygote patient with pantothenate-kinase-associated neurodegeneration. <i>SAGE Open Medical Case Reports</i> , 2017, 5, 2050313X1772010.	0.2	0
2452	Functional analysis of novel <i>DEAF1</i> variants identified through clinical exome sequencing expands <i>DEAF1</i> -associated neurodevelopmental disorder (DAND) phenotype. <i>Human Mutation</i> , 2017, 38, 1774-1785.	1.1	20
2453	A novel PAX9 mutation causing oligodontia. <i>Archives of Oral Biology</i> , 2017, 84, 100-105.	0.8	8
2454	Evidence of Early-Stage Selection on EPAS1 and GPR126 Genes in Andean High Altitude Populations. <i>Scientific Reports</i> , 2017, 7, 13042.	1.6	29
2455	Machine Learning and Rare Variant Adjudication in Type 1 Long QT Syndrome. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	3
2456	Computational study of putative functional variants in human kisspeptin. <i>Journal of Genetic Engineering and Biotechnology</i> , 2017, 15, 419-422.	1.5	3
2457	A Missense Variant in PLEC Increases Risk of Atrial Fibrillation. <i>Journal of the American College of Cardiology</i> , 2017, 70, 2157-2168.	1.2	73
2458	Evolutionary genomics of grape (<i>Vitis vinifera</i> ssp. <i>vinifera</i>) domestication. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, 11715-11720.	3.3	236

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2459	Exploiting whole genome sequence data to fine map and characterize candidate genes within a quantitative trait loci region affecting androstenone on porcine chromosome 5. <i>Animal Genetics</i> , 2017, 48, 653-659.	0.6	5
2460	Analysis and Annotation of Whole-Genome or Whole-Exome Sequencing Derived Variants for Clinical Diagnosis. <i>Current Protocols in Human Genetics</i> , 2017, 95, 9.24.1-9.24.28.	3.5	20
2461	A Novel Mutation in the TBG Gene Producing Partial Thyroxine-Binding Globulin Deficiency (Glencoe) Identified in 2 Families. <i>European Thyroid Journal</i> , 2017, 6, 138-142.	1.2	8
2462	Novel mutations in the <i>LRP5</i> gene in patients with Osteoporosis-pseudoglioma syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3132-3135.	0.7	13
2463	The Genetics and Genomics of Plant Domestication. <i>BioScience</i> , 2017, 67, 971-982.	2.2	83
2464	Exome sequencing and network analysis identifies shared mechanisms underlying spinocerebellar ataxia. <i>Brain</i> , 2017, 140, 2860-2878.	3.7	98
2465	Evidence for the Selective Basis of Transition-to-Transversion Substitution Bias in Two RNA Viruses. <i>Molecular Biology and Evolution</i> , 2017, 34, 3205-3215.	3.5	128
2466	Association analysis identifies 65 new breast cancer risk loci. <i>Nature</i> , 2017, 551, 92-94.	13.7	1,099
2467	A novel p.T139M mutation in HSPB1 highlighting the phenotypic spectrum in a family. <i>Brain and Behavior</i> , 2017, 7, e00774.	1.0	12
2468	A novel Pyrin-Associated Autoinflammation with Neutrophilic Dermatitis mutation further defines 14-3-3 binding of pyrin and distinction to Familial Mediterranean Fever. <i>Annals of the Rheumatic Diseases</i> , 2017, 76, 2085-2094.	0.5	118
2469	Characterisation of the novel deleterious RAD51C p.Arg312Trp variant and prioritisation criteria for functional analysis of RAD51C missense changes. <i>British Journal of Cancer</i> , 2017, 117, 1048-1062.	2.9	12
2470	A Scalable Bayesian Method for Integrating Functional Information in Genome-wide Association Studies. <i>American Journal of Human Genetics</i> , 2017, 101, 404-416.	2.6	63
2471	Panel sequencing of 264 candidate susceptibility genes and segregation analysis in a cohort of non-BRCA1, non-BRCA2 breast cancer families. <i>Breast Cancer Research and Treatment</i> , 2017, 166, 937-949.	1.1	16
2472	FIRE: functional inference of genetic variants that regulate gene expression. <i>Bioinformatics</i> , 2017, 33, 3895-3901.	1.8	30
2473	Assessing predictions of fitness effects of missense mutations in SUMO-conjugating enzyme UBE2L. <i>Human Mutation</i> , 2017, 38, 1051-1063.	1.1	12
2474	A missense mutation in PFAS (phosphoribosylformylglycinamide synthase) is likely causal for embryonic lethality associated with the MH1 haplotype in MontbÃ©liarde dairy cattle. <i>Journal of Dairy Science</i> , 2017, 100, 8176-8187.	1.4	24
2475	High-Throughput Genetic Screening of 51 Pediatric Cataract Genes Identifies Causative Mutations in Inherited Pediatric Cataract in South Eastern Australia. <i>G3: Genes, Genomes, Genetics</i> , 2017, 7, 3257-3268.	0.8	20
2476	SETD6 dominant negative mutation in familial colorectal cancer type X. <i>Human Molecular Genetics</i> , 2017, 26, 4481-4493.	1.4	23

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2477	1-CMDb: A Curated Database of Genomic Variations of the One-Carbon Metabolism Pathway. <i>Public Health Genomics</i> , 2017, 20, 136-141.	0.6	2
2478	NDUFAF4 variants are associated with Leigh syndrome and cause a specific mitochondrial complex I assembly defect. <i>European Journal of Human Genetics</i> , 2017, 25, 1273-1277.	1.4	25
2479	Whole Exome Sequencing Identified a Novel <i>IGFBP6</i> Variant in a Disc Degeneration Pedigree. <i>Genetic Testing and Molecular Biomarkers</i> , 2017, 21, 580-585.	0.3	6
2480	Ultra-rare mutations in <i>SRCAP</i> segregate in Caribbean Hispanic families with Alzheimer disease. <i>Neurology: Genetics</i> , 2017, 3, e178.	0.9	8
2481	The mtDNA replication-related genes TFAM and POLG are associated with leprosy in Han Chinese from Southwest China. <i>Journal of Dermatological Science</i> , 2017, 88, 349-356.	1.0	8
2482	Clinical Yield of Familial Screening After Sudden Death in Young Subjects. <i>Circulation: Arrhythmia and Electrophysiology</i> , 2017, 10, .	2.1	29
2483	Discovery of Variants Underlying Host Susceptibility to Virus Infection Using Whole-Exome Sequencing. <i>Methods in Molecular Biology</i> , 2017, 1656, 209-227.	0.4	0
2484	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. <i>European Journal of Human Genetics</i> , 2017, 25, 1246-1252.	1.4	34
2485	Replication and validation of genetic polymorphisms associated with survival after allogeneic blood or marrow transplant. <i>Blood</i> , 2017, 130, 1585-1596.	0.6	45
2486	Mutations in <i>KARS</i> cause early-onset hearing loss and leukoencephalopathy: Potential pathogenic mechanism. <i>Human Mutation</i> , 2017, 38, 1740-1750.	1.1	25
2487	Prevalence of common hereditary risk factors for thrombophilia in Somalia and identification of a novel Gln544Arg mutation in coagulation factor V. <i>Journal of Thrombosis and Thrombolysis</i> , 2017, 44, 536-543.	1.0	8
2488	A Phase II Study of Arginine Deiminase (ADI-PEG20) in Relapsed/Refractory or Poor-Risk Acute Myeloid Leukemia Patients. <i>Scientific Reports</i> , 2017, 7, 11253.	1.6	52
2489	<i>HLX</i> is a candidate gene for a pattern of anomalies associated with congenital diaphragmatic hernia, short bowel, and asplenia. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3070-3074.	0.7	10
2490	Combining Traditional Mutagenesis with New High-Throughput Sequencing and Genome Editing to Reveal Hidden Variation in Polyploid Wheat. <i>Annual Review of Genetics</i> , 2017, 51, 435-454.	3.2	100
2491	Annotating pathogenic non-coding variants in genic regions. <i>Nature Communications</i> , 2017, 8, 236.	5.8	122
2492	Interleukin-6, tumor necrosis factor-alpha and receptor activator of nuclear factor kappa ligand are elevated in hypertrophic gastric mucosa of pachydermoperiostosis. <i>Scientific Reports</i> , 2017, 7, 9686.	1.6	4
2493	Cscape: a tool for predicting oncogenic single-point mutations in the cancer genome. <i>Scientific Reports</i> , 2017, 7, 11597.	1.6	52
2494	Genomic landscape of human erythroleukemia K562 cell line, as determined by next-generation sequencing and cytogenetics. <i>Acta Haematologica Polonica</i> , 2017, 48, 343-349.	0.1	3

#	ARTICLE	IF	CITATIONS
2495	Genetic association study of common variants in TGFB1 and IL-6 with developmental dysplasia of the hip in Han Chinese population. <i>Scientific Reports</i> , 2017, 7, 10287.	1.6	15
2496	Polygenic determinants in extremes of high-density lipoprotein cholesterol. <i>Journal of Lipid Research</i> , 2017, 58, 2162-2170.	2.0	49
2497	An overview of posttraumatic stress disorder genetic studies by analyzing and integrating genetic data into genetic database PTSDgene. <i>Neuroscience and Biobehavioral Reviews</i> , 2017, 83, 647-656.	2.9	17
2498	The correlation between CRB1 variants and the clinical severity of Brazilian patients with different inherited retinal dystrophy phenotypes. <i>Scientific Reports</i> , 2017, 7, 8654.	1.6	17
2499	Identification of Novel Clinically Relevant Variants in 70 Southern Chinese patients with Thoracic Aortic Aneurysm and Dissection by Next-generation Sequencing. <i>Scientific Reports</i> , 2017, 7, 10035.	1.6	29
2500	A novel rare variant R292H in RTN4R affects growth cone formation and possibly contributes to schizophrenia susceptibility. <i>Translational Psychiatry</i> , 2017, 7, e1214-e1214.	2.4	25
2501	A strategy for molecular diagnostics of Fanconi anemia in Brazilian patients. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 360-372.	0.6	8
2502	A deep intronic CLRN1 (USH3A) founder mutation generates an aberrant exon and underlies severe Usher syndrome on the Arabian Peninsula. <i>Scientific Reports</i> , 2017, 7, 1411.	1.6	33
2503	Molecular characterization of haemophilia B patients in southern Brazil. <i>Haemophilia</i> , 2017, 23, e457-e461.	1.0	4
2504	Neural tube defects in Waardenburg syndrome: A case report and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2472-2477.	0.7	25
2505	<i>TEX15</i> : A DNA repair gene associated with prostate cancer risk in Han Chinese. <i>Prostate</i> , 2017, 77, 1271-1278.	1.2	9
2506	Clinical features and mutation spectrum in Chinese patients with <i>CADASIL</i> : A multicenter retrospective study. <i>CNS Neuroscience and Therapeutics</i> , 2017, 23, 707-716.	1.9	41
2507	Combined tumor genomic profiling and exome sequencing in a breast cancer family implicates <i>ATM</i> in tumorigenesis: A proof of principle study. <i>Genes Chromosomes and Cancer</i> , 2017, 56, 788-799.	1.5	5
2508	Homozygous mutations in <i>PJVK</i> and <i>MYO15A</i> genes associated with non-syndromic hearing loss in Moroccan families. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2017, 101, 25-29.	0.4	18
2509	Clinical and molecular characterization of cystinuria in a French cohort: relevance of assessing large-scale rearrangements and splicing variants. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 373-389.	0.6	22
2510	A recurrent, non-penetrant sequence variant, p.Arg266Cys in Growth/Differentiation Factor 3 (<i>GDF3</i>) in a female with unilateral anophthalmia and skeletal anomalies. <i>American Journal of Ophthalmology Case Reports</i> , 2017, 7, 102-106.	0.4	4
2511	Identification and characterization of a novel recessive <i>KCNQ1</i> mutation associated with Romano-Ward Long-QT syndrome in two Iranian families. <i>Journal of Electrocardiology</i> , 2017, 50, 912-918.	0.4	6
2512	Comparative genomic and phylogenetic analysis of a toxigenic clinical isolate of <i>Corynebacterium diphtheriae</i> strain B-D-16-78 from Malaysia. <i>Infection, Genetics and Evolution</i> , 2017, 54, 263-270.	1.0	19

#	ARTICLE	IF	CITATIONS
2513	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. <i>Nature Neuroscience</i> , 2017, 20, 1217-1224.	7.1	212
2514	Mutation screening in genes known to be responsible for Retinitis Pigmentosa in 98 Small Han Chinese Families. <i>Scientific Reports</i> , 2017, 7, 1948.	1.6	34
2515	SMARCB1/INI1 Involvement in Pediatric Chordoma. <i>American Journal of Surgical Pathology</i> , 2017, 41, 56-61.	2.1	64
2516	Squamous proliferations on the legs of women: Qualitative examination of histopathology, TP53 sequencing, and implications for diagnosis in a series of 30 cases. <i>Journal of the American Academy of Dermatology</i> , 2017, 77, 1126-1132.e1.	0.6	10
2517	Characterization of SNPs in the dopamine- β -hydroxylase gene providing new insights into its structure-function relationship. <i>Neurogenetics</i> , 2017, 18, 155-168.	0.7	9
2518	Biochemical characteristics of newborns with carnitine transporter defect identified by newborn screening in California. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 76-84.	0.5	23
2519	Genetic variation in the gene <i>LRP2</i> increases relapse risk in multiple sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 864-868.	0.9	21
2520	Functionally pathogenic <i>EARS2</i> variants in vitro may not manifest a phenotype in vivo. <i>Neurology: Genetics</i> , 2017, 3, e162.	0.9	11
2521	Use of exome sequencing to determine the full profile of genetic variants in the fluoropyrimidine pathway in colorectal cancer patients affected by severe toxicity. <i>Pharmacogenomics</i> , 2017, 18, 1215-1223.	0.6	12
2522	<i>IDUA</i> mutational profile and genotype-phenotype relationships in UK patients with Mucopolysaccharidosis Type I. <i>Human Mutation</i> , 2017, 38, 1555-1568.	1.1	16
2523	One amino acid makes a difference—Characterization of a new TPMT allele and the influence of SAM on TPMT stability. <i>Scientific Reports</i> , 2017, 7, 46428.	1.6	16
2524	Application of whole-exome sequencing to direct the specific functional testing and diagnosis of rare inherited bleeding disorders in patients from the Å–resund Region, Scandinavia. <i>British Journal of Haematology</i> , 2017, 179, 308-322.	1.2	49
2525	Deep sequencing reveals variations in somatic cell mosaic mutations between monozygotic twins with discordant psychiatric disease. <i>Human Genome Variation</i> , 2017, 4, 17032.	0.4	22
2526	Pitfalls of exome sequencing: a case study of the attribution of HBP2 rs7080536 in familial non-medullary thyroid cancer. <i>Npj Genomic Medicine</i> , 2017, 2, .	1.7	7
2527	Leveraging multiple genomic data to prioritize disease-causing indels from exome sequencing data. <i>Scientific Reports</i> , 2017, 7, 1804.	1.6	3
2528	Predicting the functional consequences of non-synonymous single nucleotide polymorphisms in IL8 gene. <i>Scientific Reports</i> , 2017, 7, 6525.	1.6	75
2529	Novel copy number variation of POMGNT1 associated with muscle-eye-brain disease detected by next-generation sequencing. <i>Scientific Reports</i> , 2017, 7, 7056.	1.6	13
2530	Characterization of a novel <i>HESX1</i> mutation in a pediatric case of septo-optic dysplasia. <i>Clinical Case Reports (discontinued)</i> , 2017, 5, 463-470.	0.2	4

#	ARTICLE	IF	CITATIONS
2531	TP53 alterations in Wilms tumour represent progression events with strong intratumour heterogeneity that are closely linked but not limited to anaplasia. <i>Journal of Pathology: Clinical Research</i> , 2017, 3, 234-248.	1.3	53
2532	Lessons from the CAGIâ€™ Hopkins clinical panel challenge. <i>Human Mutation</i> , 2017, 38, 1155-1168.	1.1	6
2533	Evolution and diversity studies of innate immune genes in Indian buffalo (<i>Bubalus bubalis</i>) breeds using next generation sequencing. <i>Genes and Genomics</i> , 2017, 39, 1237-1247.	0.5	1
2534	Reappraisal of the genetic diversity and pharmacogenetic assessment of <i>CES1</i> . <i>Pharmacogenomics</i> , 2017, 18, 1241-1257.	0.6	4
2535	Blocking protein quality control to counter hereditary cancers. <i>Genes Chromosomes and Cancer</i> , 2017, 56, 823-831.	1.5	23
2536	Clinical, Endocrine, and Molecular Genetic Analysis of a Large Cohort of Saudi Arabian Patients with Laron Syndrome. <i>Hormone Research in Paediatrics</i> , 2017, 88, 119-126.	0.8	4
2537	Loss-of-Function <i>KCNE2</i> Variants. <i>Circulation: Arrhythmia and Electrophysiology</i> , 2017, 10, .	2.1	31
2538	<i>CNGB3</i> mutation spectrum including copy number variations in 552 achromatopsia patients. <i>Human Mutation</i> , 2017, 38, 1579-1591.	1.1	52
2539	Identification of a novel variant of the RET proto-oncogene in a novel family with Hirschsprungâ€™s disease. <i>Pediatric Surgery International</i> , 2017, 33, 1041-1046.	0.6	3
2540	BRCA1/2 missense mutations and the value of in-silico analyses. <i>European Journal of Medical Genetics</i> , 2017, 60, 572-577.	0.7	7
2541	Settling the score: variant prioritization and Mendelian disease. <i>Nature Reviews Genetics</i> , 2017, 18, 599-612.	7.7	213
2542	Speech and Language: Translating the Genome. <i>Trends in Genetics</i> , 2017, 33, 642-656.	2.9	57
2543	Germline MSH6 Mutation in a Patient With Two Independent Primary Glioblastomas. <i>Journal of Neuropathology and Experimental Neurology</i> , 2017, 76, 848-853.	0.9	4
2544	Yeast Reciprocal Hemizygoty to Confirm the Causality of a Quantitative Trait Loci-Associated Gene. <i>Cold Spring Harbor Protocols</i> , 2017, 2017, pdb.prot089078.	0.2	7
2545	Human CRMP4 mutation and disrupted Crmp4 expression in mice are associated with ASD characteristics and sexual dimorphism. <i>Scientific Reports</i> , 2017, 7, 16812.	1.6	18
2546	Soluble epoxide hydrolase (sEH) as a potential target for arterial hypertension therapy. <i>Russian Journal of Genetics</i> , 2017, 53, 972-981.	0.2	3
2547	Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 965-976.	2.6	41
2548	Identification of potentially oncogenic alterations from tumor-only samples reveals Fanconi anemia pathway mutations in bladder carcinomas. <i>Npj Genomic Medicine</i> , 2017, 2, 29.	1.7	14

#	ARTICLE	IF	CITATIONS
2549	The Genetic Spectrum of Familial Hypercholesterolemia (FH) in the Iranian Population. <i>Scientific Reports</i> , 2017, 7, 17087.	1.6	15
2550	Novel Mutation in <i>FLNC</i> (Filamin C) Causes Familial Restrictive Cardiomyopathy. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	62
2551	Computational prediction of the phenotypic effects of genetic variants: basic concepts and some application examples in <i>Drosophila</i> nervous system genes. <i>Journal of Neurogenetics</i> , 2017, 31, 307-319.	0.6	2
2552	Exome array analysis identifies GPR35 as a novel susceptibility gene for anthracycline-induced cardiotoxicity in childhood cancer. <i>Pharmacogenetics and Genomics</i> , 2017, 27, 445-453.	0.7	22
2553	Pointed rhythmic theta waves: a unique EEG pattern in KCNQ2-related neonatal epileptic encephalopathy. <i>Epileptic Disorders</i> , 2017, 19, 351-356.	0.7	5
2554	Factor XI gene variants in factor XI-deficient patients of Southern Italy: identification of a novel mutation and genotype-phenotype relationship. <i>Human Genome Variation</i> , 2017, 4, 17043.	0.4	6
2555	Refactoring the Genetic Code for Increased Evolvability. <i>MBio</i> , 2017, 8, .	1.8	17
2557	Prevalence and detection of low-allele-fraction variants in clinical cancer samples. <i>Nature Communications</i> , 2017, 8, 1377.	5.8	137
2558	Genomic Subtypes of Non-invasive Bladder Cancer with Distinct Metabolic Profile and Female Gender Bias in KDM6A Mutation Frequency. <i>Cancer Cell</i> , 2017, 32, 701-715.e7.	7.7	224
2559	Fatal <i>CTLA4</i> heterozygosity with autoimmunity and recurrent infections: a de novo mutation. <i>Clinical Case Reports (discontinued)</i> , 2017, 5, 2066-2070.	0.2	5
2560	Factor VII deficiency: a novel missense variant and genotype-phenotype correlation in patients from Southern Italy. <i>Human Genome Variation</i> , 2017, 4, 17048.	0.4	6
2561	Novel Homozygous <i>LRP5</i> Mutations in Mexican Patients with Osteoporosis-Pseudoglioma Syndrome. <i>Genetic Testing and Molecular Biomarkers</i> , 2017, 21, 742-746.	0.3	6
2562	Dysfunction of Myosin Light Chain 4 (MYL4) Leads to Heritable Atrial Cardiomyopathy With Electrical, Contractile, and Structural Components: Evidence From Genetically Engineered Rats. <i>Journal of the American Heart Association</i> , 2017, 6, .	1.6	52
2563	Genome-wide linkage and association study implicates the 10q26 region as a major genetic contributor to primary nonsyndromic vesicoureteric reflux. <i>Scientific Reports</i> , 2017, 7, 14595.	1.6	17
2564	Molecular diagnosis in cerebral cavernous malformations. <i>Neurologia (English Edition)</i> , 2017, 32, 540-545.	0.2	2
2565	Autoimmunity/inflammation in a monogenic primary immunodeficiency cohort. <i>Clinical and Translational Immunology</i> , 2017, 6, e155.	1.7	27
2566	Percolation transition of cooperative mutational effects in colorectal tumorigenesis. <i>Nature Communications</i> , 2017, 8, 1270.	5.8	28
2567	<i>Caenorhabditis elegans</i> DAF-2 as a Model for Human Insulin Receptoropathies. <i>G3: Genes, Genomes, Genetics</i> , 2017, 7, 257-268.	0.8	10

#	ARTICLE	IF	CITATIONS
2568	Clinical presentations, metabolic abnormalities and end-organ complications in patients with familial partial lipodystrophy. <i>Metabolism: Clinical and Experimental</i> , 2017, 72, 109-119.	1.5	54
2569	<i>ATM</i> , radiation, and the risk of second primary breast cancer. <i>International Journal of Radiation Biology</i> , 2017, 93, 1121-1127.	1.0	34
2570	Protein structure and phenotypic analysis of pathogenic and population missense variants in <i>STXBP1</i> . <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 495-507.	0.6	29
2571	In silico analyses of deleterious missense SNPs of human apolipoprotein E3. <i>Scientific Reports</i> , 2017, 7, 2509.	1.6	24
2572	Clinical presentation and outcome in a series of 32 patients with 2-methylacetoacetyl-coenzyme A thiolase (MAT) deficiency. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 67-75.	0.5	24
2573	A variation in <i>PANK2</i> gene is causing Pantothenate kinase-associated Neurodegeneration in a family from Jammu and Kashmir “India. <i>Scientific Reports</i> , 2017, 7, 4834.	1.6	6
2574	Analysis of <i>GJB6</i> (Δ ₃₀) and <i>GJB3</i> (Δ ₃₁) genes in deaf patients with monoallelic mutations in <i>GJB2</i> (Δ ₂₆) gene in the Sakha Republic (Yakutia). <i>Russian Journal of Genetics</i> , 2017, 53, 688-697.	0.2	6
2575	Novel mutations in patients with hereditary red blood cell membrane disorders using next-generation sequencing. <i>Gene</i> , 2017, 627, 556-562.	1.0	25
2576	13 novel putative mutations in <i>ATP7A</i> found in a cohort of 25 Italian families. <i>Metabolic Brain Disease</i> , 2017, 32, 1173-1183.	1.4	7
2577	Functional analyses of a novel missense and other mutations of the vitamin D receptor in association with alopecia. <i>Scientific Reports</i> , 2017, 7, 5102.	1.6	14
2578	DoriTool: A Bioinformatics Integrative Tool for Post-Association Functional Annotation. <i>Public Health Genomics</i> , 2017, 20, 126-135.	0.6	4
2579	Soft Sweeps Are the Dominant Mode of Adaptation in the Human Genome. <i>Molecular Biology and Evolution</i> , 2017, 34, 1863-1877.	3.5	164
2580	The UCL low-density lipoprotein receptor gene variant database: pathogenicity update. <i>Journal of Medical Genetics</i> , 2017, 54, 217-223.	1.5	75
2581	Mutations of conserved non-coding elements of <i>PITX2</i> in patients with ocular dysgenesis and developmental glaucoma. <i>Human Molecular Genetics</i> , 2017, 26, 3630-3638.	1.4	28
2582	Heterologous expression and characterization of functional mushroom tyrosinase (AbPPO4). <i>Scientific Reports</i> , 2017, 7, 1810.	1.6	85
2583	Natural variation in <i>Arabidopsis thaliana</i> Cd responses and the detection of quantitative trait loci affecting Cd tolerance. <i>Scientific Reports</i> , 2017, 7, 3693.	1.6	19
2584	Ancient selection for derived alleles at a <i>GDF5</i> enhancer influencing human growth and osteoarthritis risk. <i>Nature Genetics</i> , 2017, 49, 1202-1210.	9.4	77
2585	Human Genome Sequencing at the Population Scale: A Primer on High-Throughput DNA Sequencing and Analysis. <i>American Journal of Epidemiology</i> , 2017, 186, 1000-1009.	1.6	63

#	ARTICLE	IF	CITATIONS
2586	Looking into a whale's heart: investigating a genetic basis for cardiomyopathy in a non-model species. <i>Genome</i> , 2017, 60, 695-705.	0.9	0
2587	Diagnosis implications of the whole genome sequencing in a large Lebanese family with hyaline fibromatosis syndrome. <i>BMC Genetics</i> , 2017, 18, 3.	2.7	8
2588	Whole genome sequencing identifies missense mutation in MTBP in Shar-Pei affected with Autoinflammatory Disease (SPAID). <i>BMC Genomics</i> , 2017, 18, 348.	1.2	9
2589	A combination of genetic and biochemical analyses for the diagnosis of PI3K-AKT-mTOR pathway-associated megalencephaly. <i>BMC Medical Genetics</i> , 2017, 18, 4.	2.1	21
2590	Sequence variations of the EGR4 gene in Korean men with spermatogenesis impairment. <i>BMC Medical Genetics</i> , 2017, 18, 47.	2.1	4
2591	Novel missense mutation in the bZIP transcription factor, MAF, associated with congenital cataract, developmental delay, seizures and hearing loss (Aymaç-Gripp syndrome). <i>BMC Medical Genetics</i> , 2017, 18, 52.	2.1	21
2592	KCNJ11, ABCC8 and TCF7L2 polymorphisms and the response to sulfonylurea treatment in patients with type 2 diabetes: a bioinformatics assessment. <i>BMC Medical Genetics</i> , 2017, 18, 64.	2.1	21
2593	Lost in translation: returning germline genetic results in genome-scale cancer research. <i>Genome Medicine</i> , 2017, 9, 41.	3.6	27
2594	Delineating the genetic heterogeneity of OCA in Hungarian patients. <i>European Journal of Medical Research</i> , 2017, 22, 20.	0.9	4
2595	Whole-exome sequencing identifies novel candidate predisposition genes for familial polycythemia vera. <i>Human Genomics</i> , 2017, 11, 6.	1.4	11
2596	Variant effect prediction tools assessed using independent, functional assay-based datasets: implications for discovery and diagnostics. <i>Human Genomics</i> , 2017, 11, 10.	1.4	68
2597	DUOX2 Mutations Are Associated With Congenital Hypothyroidism With Ectopic Thyroid Gland. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 4060-4071.	1.8	48
2598	Korean Variant Archive (KOVA): a reference database of genetic variations in the Korean population. <i>Scientific Reports</i> , 2017, 7, 4287.	1.6	60
2599	Determinants of postprandial plasma bile acid kinetics in human volunteers. <i>American Journal of Physiology - Renal Physiology</i> , 2017, 313, G300-G312.	1.6	38
2600	Evaluating Mendelian nephrotic syndrome genes for evidence for risk alleles or oligogenicity that explain heritability. <i>Pediatric Nephrology</i> , 2017, 32, 467-476.	0.9	9
2601	Genome-wide genetic screening with chemically mutagenized haploid embryonic stem cells. <i>Nature Chemical Biology</i> , 2017, 13, 12-14.	3.9	36
2602	Exome-chip association analysis reveals an Asian-specific missense variant in PAX4 associated with type 2 diabetes in Chinese individuals. <i>Diabetologia</i> , 2017, 60, 107-115.	2.9	19
2603	Human RECQ Helicase Pathogenic Variants, Population Variation and Missing Diseases. <i>Human Mutation</i> , 2017, 38, 193-203.	1.1	24

#	ARTICLE	IF	CITATIONS
2604	<i>De novo GABRG2</i> mutations associated with epileptic encephalopathies. <i>Brain</i> , 2017, 140, 49-67.	3.7	80
2605	Novel Deleterious Dihydropyrimidine Dehydrogenase Variants May Contribute to 5-Fluorouracil Sensitivity in an East African Population. <i>Clinical Pharmacology and Therapeutics</i> , 2017, 101, 382-390.	2.3	31
2606	A novel TPM2 gene splice-site mutation causes severe congenital myopathy with arthrogryposis and dysmorphic features. <i>Journal of Applied Genetics</i> , 2017, 58, 199-203.	1.0	13
2607	A cross-ethnic survey of CFB and SLC44A4, Indian ulcerative colitis GWAS hits, underscores their potential role in disease susceptibility. <i>European Journal of Human Genetics</i> , 2017, 25, 111-122.	1.4	13
2608	Novel BRCA1 and BRCA2 Tumor Test as Basis for Treatment Decisions and Referral for Genetic Counselling of Patients with Ovarian Carcinomas. <i>Human Mutation</i> , 2017, 38, 226-235.	1.1	55
2609	Calpain 12 Function Revealed through the Study of an Atypical Case of Autosomal Recessive Congenital Ichthyosis. <i>Journal of Investigative Dermatology</i> , 2017, 137, 385-393.	0.3	19
2610	Frequency of pathogenic germline mutation in CHEK2, PALB2, MRE11, and RAD50 in patients at high risk for hereditary breast cancer. <i>Breast Cancer Research and Treatment</i> , 2017, 161, 95-102.	1.1	28
2611	Using GWAS to identify novel therapeutic targets for osteoporosis. <i>Translational Research</i> , 2017, 181, 15-26.	2.2	45
2612	Acute Lung Injury and Repair. <i>Respiratory Medicine</i> , 2017, , .	0.1	1
2613	In silico analysis of novel mutations in maple syrup urine disease patients from Iran. <i>Metabolic Brain Disease</i> , 2017, 32, 105-113.	1.4	16
2614	Genetic diagnosis in hemophilia and von Willebrand disease. <i>Blood Reviews</i> , 2017, 31, 47-56.	2.8	47
2615	EFHC1 variants in juvenile myoclonic epilepsy: reanalysis according to NHGRI and ACMG guidelines for assigning disease causality. <i>Genetics in Medicine</i> , 2017, 19, 144-156.	1.1	34
2616	Accumulation of Deleterious Mutations on the Neo-Y Chromosome of Japan Sea Stickleback (<i>Gasterosteus nipponicus</i>). <i>Journal of Heredity</i> , 2017, 108, 63-68.	1.0	12
2617	Rare Genetic Variants of the Transthyretin Gene Are Associated with Alzheimer's Disease in Han Chinese. <i>Molecular Neurobiology</i> , 2017, 54, 5192-5200.	1.9	24
2618	<i>mlc</i>-based powdery mildew resistance in hexaploid bread wheat generated by a non-transgenic <sc>TILLING</sc> approach. <i>Plant Biotechnology Journal</i> , 2017, 15, 367-378.	4.1	163
2619	In silico screening, molecular docking, and molecular dynamics studies of SNP-derived human P5CR mutants. <i>Journal of Biomolecular Structure and Dynamics</i> , 2017, 35, 2441-2453.	2.0	12
2620	Using large sequencing data sets to refine intragenic disease regions and prioritize clinical variant interpretation. <i>Genetics in Medicine</i> , 2017, 19, 496-504.	1.1	15
2621	Molecular dynamics-based analyses of the structural instability and secondary structure of the fibrinogen gamma chain protein with the D356V mutation. <i>Journal of Biomolecular Structure and Dynamics</i> , 2017, 35, 2714-2724.	2.0	39

#	ARTICLE	IF	CITATIONS
2622	Digenic inheritance in epidermolysis bullosa simplex involving two novel mutations in <i>KRT5</i> and <i>KRT14</i> . <i>British Journal of Dermatology</i> , 2017, 177, 262-264.	1.4	9
2623	Prediction of deleterious single nucleotide polymorphisms and their effect on the sequence and structure of the human <i>CCND1</i> gene. <i>Journal of Taibah University Medical Sciences</i> , 2017, 12, 221-228.	0.5	1
2624	Targeted next generation sequencing: the diagnostic value in early-onset epileptic encephalopathy. <i>Acta Neurologica Belgica</i> , 2017, 117, 131-138.	0.5	33
2625	Variants in <i>SKP1</i> , <i>PROB1</i> , and <i>IL17B</i> genes at keratoconus 5q31.1q35.3 susceptibility locus identified by whole-exome sequencing. <i>European Journal of Human Genetics</i> , 2017, 25, 73-78.	1.4	19
2626	<i>INPPL1</i> gene mutations in opsismodysplasia. <i>Journal of Human Genetics</i> , 2017, 62, 135-140.	1.1	13
2627	A Comprehensive Functional Analysis of <i>NTRK1</i> Missense Mutations Causing Hereditary Sensory and Autonomic Neuropathy Type IV (HSAN IV). <i>Human Mutation</i> , 2017, 38, 55-63.	1.1	29
2628	First evidence of Smith-Magenis syndrome in mother and daughter due to a novel <i>RAI</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 231-238.	0.7	17
2629	Expanding the genotype-phenotype spectrum in hereditary colorectal cancer by gene panel testing. <i>Familial Cancer</i> , 2017, 16, 195-203.	0.9	55
2630	Targeted Next-Generation Sequencing for Comprehensive Genetic Profiling of Pharmacogenes. <i>Clinical Pharmacology and Therapeutics</i> , 2017, 101, 396-405.	2.3	54
2631	Novel <i>Col12A1</i> variant expands the clinical picture of congenital myopathies with extracellular matrix defects. <i>Muscle and Nerve</i> , 2017, 55, 277-281.	1.0	31
2632	Next generation semiconductor based sequencing of bitter taste receptor genes in different pig populations and association analysis using a selective DNA pool-seq approach. <i>Animal Genetics</i> , 2017, 48, 97-102.	0.6	14
2633	Screening of Living Kidney Donors for Genetic Diseases Using a Comprehensive Genetic Testing Strategy. <i>American Journal of Transplantation</i> , 2017, 17, 401-410.	2.6	27
2634	Pulmonary Vasculopathy Associated with <i>FIGF</i> Gene Mutation. <i>American Journal of Pathology</i> , 2017, 187, 25-32.	1.9	8
2635	<i>EGR2</i> mutations define a new clinically aggressive subgroup of chronic lymphocytic leukemia. <i>Leukemia</i> , 2017, 31, 1547-1554.	3.3	46
2636	Genomic approaches to the assessment of human spina bifida risk. <i>Birth Defects Research</i> , 2017, 109, 120-128.	0.8	23
2637	Computational Screening and Exploration of Disease-Associated Genes in Alzheimer's Disease. <i>Journal of Cellular Biochemistry</i> , 2017, 118, 1471-1479.	1.2	8
2638	Prognostic predictive value of gene mutations in Japanese patients with hypertrophic cardiomyopathy. <i>Heart and Vessels</i> , 2017, 32, 700-707.	0.5	13
2639	Cerebrospinal Fluid Progranulin, but Not Serum Progranulin, Is Reduced in <i>GRN</i>-Negative Frontotemporal Dementia. <i>Neurodegenerative Diseases</i> , 2017, 17, 83-88.	0.8	23

#	ARTICLE	IF	CITATIONS
2640	Clinical genomic profiling identifies <i>TYK2</i> mutation and overexpression in patients with neurofibromatosis type 1-associated malignant peripheral nerve sheath tumors. <i>Cancer</i> , 2017, 123, 1194-1201.	2.0	25
2641	Genome-Wide Transcriptome Profiling of the Neoplastic Giant Cell Tumor of Bone Stromal Cells by RNA Sequencing. <i>Journal of Cellular Biochemistry</i> , 2017, 118, 1349-1360.	1.2	7
2642	Gene panel sequencing in familial breast/ovarian cancer patients identifies multiple novel mutations also in genes others than BRCA1/2. <i>International Journal of Cancer</i> , 2017, 140, 95-102.	2.3	99
2643	Beta-Binomial Model for the Detection of Rare Mutations in Pooled Next-Generation Sequencing Experiments. <i>Journal of Computational Biology</i> , 2017, 24, 357-367.	0.8	7
2644	Consecutive analysis of mutation spectrum in the dystrophin gene of 507 Korean boys with Duchenne/Becker muscular dystrophy in a single center. <i>Muscle and Nerve</i> , 2017, 55, 727-734.	1.0	35
2645	A novel multisystem disease associated with recessive mutations in the tyrosyl-tRNA synthetase (<i>YARS</i>) gene. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 126-134.	0.7	36
2646	Confirmation of the GNB4 gene as causal for Charcot-Marie-Tooth disease by a novel de novo mutation in a Czech patient. <i>Neuromuscular Disorders</i> , 2017, 27, 57-60.	0.3	9
2647	Network-Based Identification of Adaptive Pathways in Evolved Ethanol-Tolerant Bacterial Populations. <i>Molecular Biology and Evolution</i> , 2017, 34, 2927-2943.	3.5	16
2648	Genetic polymorphism analysis of cytochrome P4502E1 (CYP2E1) in a Chinese Tibetan population. <i>Medicine (United States)</i> , 2017, 96, e8855.	0.4	7
2649	Two novel mutations identified in ADCC families impair crystallin protein distribution and induce apoptosis in human lens epithelial cells. <i>Scientific Reports</i> , 2017, 7, 17848.	1.6	28
2650	Enrichment of putatively damaging rare variants in the DYX2 locus and the reading-related genes CCDC136 and FLNC. <i>Human Genetics</i> , 2017, 136, 1395-1405.	1.8	9
2651	Genetic variations in Bestrophin-1 and associated clinical findings in two Chinese patients with juvenile-onset and adult-onset best vitelliform macular dystrophy. <i>Molecular Medicine Reports</i> , 2017, 17, 225-233.	1.1	11
2652	High Prevalence of Diabetes-Predisposing Variants in MODY Genes Among Danish Women With Gestational Diabetes Mellitus. <i>Journal of the Endocrine Society</i> , 2017, 1, 681-690.	0.1	32
2653	Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency: Unique Presenting Laboratory Values and a Review of Biochemical and Clinical Features. <i>JIMD Reports</i> , 2017, 40, 63-69.	0.7	27
2654	Identification of a novel synaptic protein, TMTC3, involved in periventricular nodular heterotopia with intellectual disability and epilepsy. <i>Human Molecular Genetics</i> , 2017, 26, 4278-4289.	1.4	36
2655	Analysis of decision tree performance in predicting the relationship between a scored outcome and multiple single nucleotide polymorphisms. , 2017, , .		0
2656	High-resolution analysis of selection sweeps identified between fine-wool Merino and coarse-wool Churra sheep breeds. <i>Genetics Selection Evolution</i> , 2017, 49, 81.	1.2	35
2657	A missense variant in the coil1A domain of the keratin 25 gene is associated with the dominant curly hair coat trait (Crd) in horse. <i>Genetics Selection Evolution</i> , 2017, 49, 85.	1.2	47

#	ARTICLE	IF	CITATIONS
2658	Abnormal creatine transport of mutations in monocarboxylate transporter 12 (MCT12) found in patients with age-related cataract can be partially rescued by exogenous chaperone CD147. <i>Human Molecular Genetics</i> , 2017, 26, 4203-4214.	1.4	17
2659	Exome-wide association study reveals novel psoriasis susceptibility locus at TNFSF15 and rare protective alleles in genes contributing to type I IFN signalling. <i>Human Molecular Genetics</i> , 2017, 26, 4301-4313.	1.4	41
2660	Heterogeneous Image Stylization Using Neural Networks. , 2017, , .		0
2661	DriverFinder: A Gene Length-Based Network Method to Identify Cancer Driver Genes. <i>Complexity</i> , 2017, 2017, 1-10.	0.9	12
2662	A molecular inversion probe-based next-generation sequencing panel to detect germline mutations in Chinese early-onset colorectal cancer patients. <i>Oncotarget</i> , 2017, 8, 24533-24547.	0.8	12
2663	Molecular Analysis of Factor VIII and Factor IX Genes in Hemophilia Patients: Identification of Novel Mutations and Molecular Dynamics Studies. <i>Journal of Clinical Medicine Research</i> , 2017, 9, 317-331.	0.6	6
2664	IMPACT OF DELETERIOUS NON-SYNONYMOUS SINGLE NUCLEOTIDE POLYMORPHISMS OF CYTOKINE GENES ON NON-CLASSICAL HYDROGEN BONDS PREDISPOSING TO CARDIOVASCULAR DISEASE: AN IN SILICO APPROACH. <i>Asian Journal of Pharmaceutical and Clinical Research</i> , 2017, 10, 214.	0.3	1
2665	Erythrocyte \hat{I}^2 spectrin can be genetically targeted to protect mice from malaria. <i>Blood Advances</i> , 2017, 1, 2624-2636.	2.5	16
2666	The Cys83Gly amino acid substitution in feather keratin is associated with pigeon performance in long-distance races. <i>Veterinari Medicina</i> , 2017, 62, 221-225.	0.2	7
2667	Contribution of Mutations in Known Mendelian Glaucoma Genes to Advanced Early-Onset Primary Open-Angle Glaucoma. , 2017, 58, 1537.		13
2668	Mutation in HvCBP20 (Cap Binding Protein 20) Adapts Barley to Drought Stress at Phenotypic and Transcriptomic Levels. <i>Frontiers in Plant Science</i> , 2017, 8, 942.	1.7	48
2669	Four novel &em>ARSA&em> gene mutations with pathogenic impacts on metachromatic leukodystrophy: a bioinformatics approach to predict pathogenic mutations. <i>Therapeutics and Clinical Risk Management</i> , 2017, Volume 13, 725-731.	0.9	9
2670	Novel EDA or EDAR Mutations Identified in Patients with X-Linked Hypohidrotic Ectodermal Dysplasia or Non-Syndromic Tooth Agenesis. <i>Genes</i> , 2017, 8, 259.	1.0	27
2671	Predicting the Functional Impact of CDH1 Missense Mutations in Hereditary Diffuse Gastric Cancer. <i>International Journal of Molecular Sciences</i> , 2017, 18, 2687.	1.8	47
2672	Caucasian Families Exhibit Significant Linkage of Myopia to Chromosome 11p. , 2017, 58, 3547.		11
2673	Fibrinogen as a Pleiotropic Protein Causing Human Diseases: The Mutational Burden of $A^{\hat{1}\pm}$, $B^{\hat{1}2}$, and \hat{I}^3 Chains. <i>International Journal of Molecular Sciences</i> , 2017, 18, 2711.	1.8	36
2674	Novel Genetic Variants Associated with Child Refractory Esophageal Stricture with Food Allergy by Exome Sequencing. <i>Nutrients</i> , 2017, 9, 390.	1.7	1
2675	Ubiquitin Specific Peptidase 22 Regulates Histone H2B Mono-Ubiquitination and Exhibits Both Oncogenic and Tumor Suppressor Roles in Cancer. <i>Cancers</i> , 2017, 9, 167.	1.7	43

#	ARTICLE	IF	CITATIONS
2676	Clinical and Genetic Evaluation of a Cohort of Pediatric Patients with Severe Inherited Retinal Dystrophies. <i>Genes</i> , 2017, 8, 280.	1.0	23
2677	A Strategy for Identifying Quantitative Trait Genes Using Gene Expression Analysis and Causal Analysis. <i>Genes</i> , 2017, 8, 347.	1.0	16
2678	Big Data Analytics for Genomic Medicine. <i>International Journal of Molecular Sciences</i> , 2017, 18, 412.	1.8	121
2679	Neuropeptide VGF Promotes Maturation of Hippocampal Dendrites That Is Reduced by Single Nucleotide Polymorphisms. <i>International Journal of Molecular Sciences</i> , 2017, 18, 612.	1.8	21
2680	Exome Sequencing Identified a Novel FBN2 Mutation in a Chinese Family with Congenital Contractural Arachnodactyly. <i>International Journal of Molecular Sciences</i> , 2017, 18, 626.	1.8	8
2681	Incomplete Segregation of MSH6 Frameshift Variants with Phenotype of Lynch Syndrome. <i>International Journal of Molecular Sciences</i> , 2017, 18, 999.	1.8	13
2682	Validation and Utilization of a Clinical Next-Generation Sequencing Panel for Selected Cardiovascular Disorders. <i>Frontiers in Cardiovascular Medicine</i> , 2017, 4, 11.	1.1	8
2683	A Review of Pathway-Based Analysis Tools That Visualize Genetic Variants. <i>Frontiers in Genetics</i> , 2017, 8, 174.	1.1	67
2684	Clinical and Molecular Heterogeneity of RTEL1 Deficiency. <i>Frontiers in Immunology</i> , 2017, 8, 449.	2.2	35
2685	Family History of Early Infant Death Correlates with Earlier Age at Diagnosis But Not Shorter Time to Diagnosis for Severe Combined Immunodeficiency. <i>Frontiers in Immunology</i> , 2017, 8, 808.	2.2	34
2686	Resources for Interpreting Variants in Precision Genomic Oncology Applications. <i>Frontiers in Oncology</i> , 2017, 7, 214.	1.3	18
2687	Exome sequencing reveals novel IRXI mutation in congenital heart disease. <i>Molecular Medicine Reports</i> , 2017, 15, 3193-3197.	1.1	11
2688	Idiopathic hypereosinophilia is clonal disorder? Clonality identified by targeted sequencing. <i>PLoS ONE</i> , 2017, 12, e0185602.	1.1	27
2689	Three Novel Mutations in the <i>NPHS1</i> Gene in Vietnamese Patients with Congenital Nephrotic Syndrome. <i>Case Reports in Genetics</i> , 2017, 2017, 1-7.	0.1	3
2690	Genotype-Phenotype Characterization of Novel Variants in Six Italian Patients with Familial Exudative Vitreoretinopathy. <i>Journal of Ophthalmology</i> , 2017, 2017, 1-10.	0.6	5
2691	De Novo <i>PTEN</i> Mutation in a Young Boy with Cutaneous Vasculitis. <i>Case Reports in Pediatrics</i> , 2017, 2017, 1-4.	0.2	12
2692	Bioinformatically predicted deleterious mutations reveal complementation in the interior spruce hybrid complex. <i>BMC Genomics</i> , 2017, 18, 970.	1.2	16
2693	Predicting treatable traits for long-acting bronchodilators in patients with stable COPD. <i>International Journal of COPD</i> , 2017, Volume 12, 3557-3565.	0.9	8

#	ARTICLE	IF	CITATIONS
2694	Disease-associated mitochondrial mutations and the evolution of primate mitogenomes. PLoS ONE, 2017, 12, e0177403.	1.1	7
2695	Candidate genes on murine chromosome 8 are associated with susceptibility to Staphylococcus aureus infection in mice and are involved with Staphylococcus aureus septicemia in humans. PLoS ONE, 2017, 12, e0179033.	1.1	5
2696	Genetic architecture of atherosclerosis dissected by QTL analyses in three F2 intercrosses of apolipoprotein E-null mice on C57BL6/J, DBA/2J and 129S6/SvEvTac backgrounds. PLoS ONE, 2017, 12, e0182882.	1.1	4
2697	Molecular diagnosis of patients with epilepsy and developmental delay using a customized panel of epilepsy genes. PLoS ONE, 2017, 12, e0188978.	1.1	56
2698	Role of GALNT12 in the genetic predisposition to attenuated adenomatous polyposis syndrome. PLoS ONE, 2017, 12, e0187312.	1.1	10
2699	Unique Features of Germline Variation in Five Egyptian Familial Breast Cancer Families Revealed by Exome Sequencing. PLoS ONE, 2017, 12, e0167581.	1.1	17
2700	Investigation of mutations in the HBB gene using the 1,000 genomes database. PLoS ONE, 2017, 12, e0174637.	1.1	29
2701	Beyond the MHC: A canine model of dermatomyositis shows a complex pattern of genetic risk involving novel loci. PLoS Genetics, 2017, 13, e1006604.	1.5	15
2702	Rare coding variants pinpoint genes that control human hematological traits. PLoS Genetics, 2017, 13, e1006925.	1.5	39
2703	Candidate lethal haplotypes and causal mutations in Angus cattle. BMC Genomics, 2017, 18, 799.	1.2	42
2704	Within-breed and multi-breed GWAS on imputed whole-genome sequence variants reveal candidate mutations affecting milk protein composition in dairy cattle. Genetics Selection Evolution, 2017, 49, 68.	1.2	111
2705	Fine mapping of a QTL affecting levels of skatole on pig chromosome 7. BMC Genetics, 2017, 18, 85.	2.7	6
2706	Genotype-phenotype correlations and expansion of the molecular spectrum of AP4M1-related hereditary spastic paraplegia. Orphanet Journal of Rare Diseases, 2017, 12, 172.	1.2	17
2707	Identification of GAA variants through whole exome sequencing targeted to a cohort of 606 patients with unexplained limb-girdle muscle weakness. Orphanet Journal of Rare Diseases, 2017, 12, 173.	1.2	21
2708	Strong epistatic and additive effects of linked candidate SNPs for Drosophila pigmentation have implications for analysis of genome-wide association studies results. Genome Biology, 2017, 18, 126.	3.8	11
2709	Linkage, whole genome sequence, and biological data implicate variants in RAB10 in Alzheimer's disease resilience. Genome Medicine, 2017, 9, 100.	3.6	67
2710	Mutational analysis of COL1A1 and COL1A2 genes among Estonian osteogenesis imperfecta patients. Human Genomics, 2017, 11, 19.	1.4	32
2711	Variant Ranker: a web-tool to rank genomic data according to functional significance. BMC Bioinformatics, 2017, 18, 341.	1.2	21

#	ARTICLE	IF	CITATIONS
2712	Mutation status coupled with RNA-sequencing data can efficiently identify important non-significantly mutated genes serving as diagnostic biomarkers of endometrial cancer. BMC Bioinformatics, 2017, 18, 472.	1.2	5
2713	Classifying cancer genome aberrations by their mutually exclusive effects on transcription. BMC Medical Genomics, 2017, 10, 66.	0.7	7
2714	Gene-Targeted Analysis of Clinically Diagnosed Long QT Russian Families. International Heart Journal, 2017, 58, 81-87.	0.5	20
2715	Genome-wide meta-analysis in Japanese populations identifies novel variants at the TMC6 and SIX3-SIX2 loci associated with HbA1c. Scientific Reports, 2017, 7, 16147.	1.6	28
2717	Targeted next-generation sequencing reveals two novel mutations of NBAS in a patient with infantile liver failure syndrome. Molecular Medicine Reports, 2017, 17, 2245-2250.	1.1	18
2718	Whole Genome Sequencing and Analysis of Godawee, a Salt Tolerant Indica Rice Variety. Rice Research Open Access, 2017, 05, .	0.4	10
2719	BRCA2, EGFR, and NTRK mutations in mismatch repair-deficient colorectal cancers with MSH2 or MLH1 mutations. Oncotarget, 2017, 8, 39945-39962.	0.8	29
2720	Polymorphism of stearoyl-CoA desaturase (SCD1) gene in Indonesian local cattle. Journal of the Indonesian Tropical Animal Agriculture, 2017, 42, 1.	0.1	1
2721	Sodium taurocholate cotransporting polypeptide (NTCP) deficiency: Identification of a novel SLC10A1 mutation in two unrelated infants presenting with neonatal indirect hyperbilirubinemia and remarkable hypercholanemia. Oncotarget, 2017, 8, 106598-106607.	0.8	25
2722	Multiple-gene panel analysis in a case series of 255 women with hereditary breast and ovarian cancer. Oncotarget, 2017, 8, 47064-47075.	0.8	68
2723	Screening RB1 Gene in Algerian Patients and Predicting the Pathogenicity of Variations by In Silico Analysis. Journal of Clinical & Experimental Ophthalmology, 2017, 08, .	0.1	0
2724	Identification of a novel de novo ANK1 R1426* nonsense mutation in a Chinese family with hereditary spherocytosis by NGS. Oncotarget, 2017, 8, 96791-96797.	0.8	9
2725	Whole exome sequencing in 75 high-risk families with validation and replication in independent case-control studies identifies TANGO2, OR5H14, and CHAD as new prostate cancer susceptibility genes. Oncotarget, 2017, 8, 1495-1507.	0.8	11
2726	The Genetic Causes of Nonsyndromic Congenital Retinal Detachment: A Genetic and Phenotypic Study of Pakistani Families. , 2017, 58, 1028.		15
2727	Effects of Type 1 Diabetes Risk Alleles on Immune Cell Gene Expression. Genes, 2017, 8, 167.	1.0	17
2728	Identification of novel mutations in endometrial cancer patients by whole-exome sequencing. International Journal of Oncology, 2017, 50, 1778-1784.	1.4	43
2729	Novel Complex ABCA4 Alleles in Brazilian Patients With Stargardt Disease: Genotype-Phenotype Correlation. , 2017, 58, 5723.		12
2730	Elucidation of MRAS-mediated Noonan syndrome with cardiac hypertrophy. JCI Insight, 2017, 2, e91225.	2.3	66

#	ARTICLE	IF	CITATIONS
2731	MUTATIONAL PROFILES OF F8 AND F9 IN A COHORT OF HAEMOPHILIA A AND HAEMOPHILIA B PATIENTS IN THE MULTI-ETHNIC MALAYSIAN POPULATION. <i>Mediterranean Journal of Hematology and Infectious Diseases</i> , 2017, 10, e2018056.	0.5	5
2732	Germline mutation within COL2A1 associated with lethal chondrodysplasia in a polled Holstein family. <i>BMC Genomics</i> , 2017, 18, 762.	1.2	9
2734	Elucidation of the genetic and epigenetic landscape alterations in RNA binding proteins in glioblastoma. <i>Oncotarget</i> , 2017, 8, 16650-16668.	0.8	36
2735	Autosomal dominant deficiency of the interleukin-17F in recurrent aphthous stomatitis: Possible novel mutation in a new entity. <i>Gene</i> , 2018, 654, 64-68.	1.0	5
2736	Outcome of adult patients with X-linked hypophosphatemia caused by <i>PHEX</i> gene mutations. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 865-876.	1.7	103
2737	Phenotypic Spectrum of <i>HCN4</i> Mutations. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002033.	1.6	18
2738	Exome sequencing reveals blended phenotype of double heterozygous <i>FBN1</i> and <i>FBN2</i> variants in a fetus. <i>European Journal of Medical Genetics</i> , 2018, 61, 399-402.	0.7	8
2739	Genetic heterogeneity of primary lesion and metastasis in small intestine neuroendocrine tumors. <i>Scientific Reports</i> , 2018, 8, 3811.	1.6	42
2740	Clinical heterogeneity of hyperinsulinism due to <i>HNF1A</i> and <i>HNF4A</i> mutations. <i>Pediatric Diabetes</i> , 2018, 19, 910-916.	1.2	23
2741	A Mild <i>PUM1</i> Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. <i>Cell</i> , 2018, 172, 924-936.e11.	13.5	103
2742	Inborn Errors of RNA Lariat Metabolism in Humans with Brainstem Viral Infection. <i>Cell</i> , 2018, 172, 952-965.e18.	13.5	92
2743	Network analysis of coronary artery disease risk genes elucidates disease mechanisms and druggable targets. <i>Scientific Reports</i> , 2018, 8, 3434.	1.6	43
2744	Genetics of dementia in a Finnish cohort. <i>European Journal of Human Genetics</i> , 2018, 26, 827-837.	1.4	6
2745	Human <i>TGF-β1</i> deficiency causes severe inflammatory bowel disease and encephalopathy. <i>Nature Genetics</i> , 2018, 50, 344-348.	9.4	95
2746	Clinical testing of <i>BRCA1</i> and <i>BRCA2</i> : a worldwide snapshot of technological practices. <i>Npj Genomic Medicine</i> , 2018, 3, 7.	1.7	44
2747	Myelin abnormality in Charcot-Marie-Tooth type 4J recapitulates features of acquired demyelination. <i>Annals of Neurology</i> , 2018, 83, 756-770.	2.8	28
2748	Recessive mutations in <i>VPS13D</i> cause childhood onset movement disorders. <i>Annals of Neurology</i> , 2018, 83, 1089-1095.	2.8	104
2749	Partial growth hormone insensitivity and dysregulatory immune disease associated with de novo germline activating <i>STAT3</i> mutations. <i>Molecular and Cellular Endocrinology</i> , 2018, 473, 166-177.	1.6	38

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2750	Natural polyphenols as sirtuin 6 modulators. <i>Scientific Reports</i> , 2018, 8, 4163.	1.6	109
2751	Kidney enlargement and multiple liver cyst formation implicate mutations in <i>PKD1/2</i> in adult sporadic polycystic kidney disease. <i>Clinical Genetics</i> , 2018, 94, 125-131.	1.0	16
2752	Whole Exome Sequencing Reveals a Monogenic Cause of Disease in 43% of 35 Families With Midaortic Syndrome. <i>Hypertension</i> , 2018, 71, 691-699.	1.3	22
2753	<i>KRAS</i> mutation is predictive of outcome in patients with pulmonary sarcomatoid carcinoma. <i>Histopathology</i> , 2018, 73, 207-214.	1.6	32
2754	Genotypic effect of a mutation of the <i>MYBPC3</i> gene and two phenotypes with different patterns of inheritance. <i>Journal of Clinical Laboratory Analysis</i> , 2018, 32, e22419.	0.9	6
2755	Variants Associated with Infantile Cholestatic Syndromes Detected in Extrahepatic Biliary Atresia by Whole Exome Studies: A 20-Case Series from Thailand. <i>Journal of Pediatric Genetics</i> , 2018, 07, 067-073.	0.3	21
2756	Statistical Validation of Rare Complement Variants Provides Insights into the Molecular Basis of Atypical Hemolytic Uremic Syndrome and C3 Glomerulopathy. <i>Journal of Immunology</i> , 2018, 200, 2464-2478.	0.4	130
2757	Heterozygous <i>SSBP1</i> start loss mutation co-segregates with hearing loss and the m.1555A>G mtDNA variant in a large multigenerational family. <i>Brain</i> , 2018, 141, 55-62.	3.7	19
2758	The contribution of <i>CACNA1A</i> , <i>ATP1A2</i> and <i>SCN1A</i> mutations in hemiplegic migraine: A clinical and genetic study in Finnish migraine families. <i>Cephalalgia</i> , 2018, 38, 1849-1863.	1.8	38
2759	Genomic Locus Modulating IOP in the BXD RI Mouse Strains. <i>G3: Genes, Genomes, Genetics</i> , 2018, 8, 1571-1578.	0.8	14
2760	<i>URAT1</i> and <i>GLUT9</i> mutations in Spanish patients with renal hypouricemia. <i>Clinica Chimica Acta</i> , 2018, 481, 83-89.	0.5	42
2761	Panel Sequencing Shows Recurrent Genetic <i>FAS</i> Alterations in Primary Cutaneous Marginal Zone Lymphoma. <i>Journal of Investigative Dermatology</i> , 2018, 138, 1573-1581.	0.3	41
2762	Variant Linkage Analysis Using <i>de Novo</i> Transcriptome Sequencing Identifies a Conserved Phosphine Resistance Gene in Insects. <i>Genetics</i> , 2018, 209, 281-290.	1.2	34
2763	<i>GNAO1</i> Mutation Induced Pediatric Dystonic Storm Rescue With Pallidal Deep Brain Stimulation. <i>Journal of Child Neurology</i> , 2018, 33, 413-416.	0.7	31
2764	No novel, high penetrant gene might remain to be found in Japanese patients with unknown MODY. <i>Journal of Human Genetics</i> , 2018, 63, 821-829.	1.1	3
2765	<i>PLCB3</i> Loss of Function Reduces <i>Pseudomonas aeruginosa</i> -Dependent IL-8 Release in Cystic Fibrosis. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2018, 59, 428-436.	1.4	15
2766	An epistatic effect of <i>KRT25</i> on <i>SP6</i> is involved in curly coat in horses. <i>Scientific Reports</i> , 2018, 8, 6374.	1.6	18
2767	Detection of functional protein domains by unbiased genome-wide forward genetic screening. <i>Scientific Reports</i> , 2018, 8, 6161.	1.6	14

#	ARTICLE	IF	CITATIONS
2768	Dysregulation of Epstein-Barr Virus Infection in Hypomorphic ZAP70 Mutation. <i>Journal of Infectious Diseases</i> , 2018, 218, 825-834.	1.9	22
2769	Whole genome sequencing of Caribbean Hispanic families with late-onset Alzheimer's disease. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 406-417.	1.7	42
2770	Canine NAPEPLD-associated models of human myelin disorders. <i>Scientific Reports</i> , 2018, 8, 5818.	1.6	14
2771	A novel compound heterozygous variant identified in GLDC gene in a Chinese family with non-ketotic hyperglycinemia. <i>BMC Medical Genetics</i> , 2018, 19, 5.	2.1	6
2772	NIPS, a 3D network-integrated predictor of deleterious protein SAPs, and its application in cancer prognosis. <i>Scientific Reports</i> , 2018, 8, 6021.	1.6	1
2773	<i>MERTK</i> mutation update in inherited retinal diseases. <i>Human Mutation</i> , 2018, 39, 887-913.	1.1	41
2774	Novel valosin-containing protein mutations associated with multisystem proteinopathy. <i>Neuromuscular Disorders</i> , 2018, 28, 491-501.	0.3	20
2775	Truncation- and motif-based pan-cancer analysis reveals tumor-suppressing kinases. <i>Science Signaling</i> , 2018, 11, .	1.6	10
2776	Germline Genetic IKZF1 Variation and Predisposition to Childhood Acute Lymphoblastic Leukemia. <i>Cancer Cell</i> , 2018, 33, 937-948.e8.	7.7	142
2777	Exome chip analyses identify genes affecting mortality after HLA-matched unrelated-donor blood and marrow transplantation. <i>Blood</i> , 2018, 131, 2490-2499.	0.6	21
2778	Pathogenic Germline Variants in 10,389 Adult Cancers. <i>Cell</i> , 2018, 173, 355-370.e14.	13.5	620
2779	In silico characterization of functional single nucleotide polymorphisms of folate pathway genes. <i>Annals of Human Genetics</i> , 2018, 82, 186-199.	0.3	6
2780	Genomic and Molecular Landscape of DNA Damage Repair Deficiency across The Cancer Genome Atlas. <i>Cell Reports</i> , 2018, 23, 239-254.e6.	2.9	801
2781	m6ASNP: a tool for annotating genetic variants by m6A function. <i>GigaScience</i> , 2018, 7, .	3.3	36
2782	VarCards: an integrated genetic and clinical database for coding variants in the human genome. <i>Nucleic Acids Research</i> , 2018, 46, D1039-D1048.	6.5	148
2783	Targeted Next-generation Sequencing and Bioinformatics Pipeline to Evaluate Genetic Determinants of Constitutional Disease. <i>Journal of Visualized Experiments</i> , 2018, , .	0.2	17
2784	Pleiotropic mapping and annotation selection in genome-wide association studies with penalized Gaussian mixture models. <i>Bioinformatics</i> , 2018, 34, 2797-2807.	1.8	34
2785	Single, short indel, and copy number variations detection in monogenic dyslipidemia using a next-generation sequencing strategy. <i>Clinical Genetics</i> , 2018, 94, 132-140.	1.0	19

#	ARTICLE	IF	CITATIONS
2786	A clinical guidance to DFNA22 drawn from a Korean cohort study with an autosomal dominant deaf population: A retrospective cohort study. <i>Journal of Gene Medicine</i> , 2018, 20, e3019.	1.4	7
2787	Clinical and biochemical features of different molecular etiologies of familial chylomicronemia. <i>Journal of Clinical Lipidology</i> , 2018, 12, 920-927.e4.	0.6	97
2788	Germline but not somatic de novo mutations are common in human congenital diaphragmatic hernia. <i>Birth Defects Research</i> , 2018, 110, 610-617.	0.8	12
2789	Genome evolution across 1,011 <i>Saccharomyces cerevisiae</i> isolates. <i>Nature</i> , 2018, 556, 339-344.	13.7	952
2790	Mutations in CFAP43 and CFAP44 cause male infertility and flagellum defects in <i>Trypanosoma</i> and human. <i>Nature Communications</i> , 2018, 9, 686.	5.8	173
2791	Whole-exome sequencing reveals a rare interferon gamma receptor 1 mutation associated with myasthenia gravis. <i>Neurological Sciences</i> , 2018, 39, 717-724.	0.9	3
2792	Yield of the <i>RYR2</i> Genetic Test in Suspected Catecholaminergic Polymorphic Ventricular Tachycardia and Implications for Test Interpretation. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e001424.	1.6	30
2793	Frequent mutations of <i>RetNet</i> genes in eoHM: Further confirmation in 325 probands and comparison with late-onset high myopia based on exome sequencing. <i>Experimental Eye Research</i> , 2018, 171, 76-91.	1.2	36
2794	Pediatric Dilated Cardiomyopathy-Associated <i>LRRC10</i> (Leucine-Rich Repeat-Containing 10) Variant Reveals <i>LRRC10</i> as an Auxiliary Subunit of Cardiac Ca^{2+} Channels. <i>Journal of the American Heart Association</i> , 2018, 7, .	1.6	16
2795	Association of elevated homocysteine levels and Methylenetetrahydrofolate reductase (<i>MTHFR</i>) 1298 A>C polymorphism with Vitiligo susceptibility in Gujarat. <i>Journal of Dermatological Science</i> , 2018, 90, 112-122.	1.0	16
2796	Probability of phenotypically detectable protein damage by ENU-induced mutations in the Mutagenetix database. <i>Nature Communications</i> , 2018, 9, 441.	5.8	43
2797	Mapping Causal Variants with Single-Nucleotide Resolution Reveals Biochemical Drivers of Phenotypic Change. <i>Cell</i> , 2018, 172, 478-490.e15.	13.5	62
2798	A novel variant of osteogenesis imperfecta type IV and low serum phosphorus level caused by a Val94Asp mutation in <i>COL1A1</i> . <i>Molecular Medicine Reports</i> , 2018, 17, 4433-4439.	1.1	0
2799	The first two confirmed sub-Saharan African families with germline <i>TP53</i> mutations causing Li-Fraumeni syndrome. <i>Familial Cancer</i> , 2018, 17, 607-613.	0.9	6
2800	Loss of function mutations in <i>EPHB4</i> are responsible for vein of Galen aneurysmal malformation. <i>Brain</i> , 2018, 141, 979-988.	3.7	47
2801	Neonatal Onset of Epilepsy of Infancy with Migrating Focal Seizures Associated with a Novel <i>GABRB3</i> Variant in Monozygotic Twins. <i>Neuropediatrics</i> , 2018, 49, 204-208.	0.3	15
2802	Somatic <i>APC</i> mosaicism and oligogenic inheritance in genetically unsolved colorectal adenomatous polyposis patients. <i>European Journal of Human Genetics</i> , 2018, 26, 387-395.	1.4	26
2803	Cancer genetics meets biomolecular mechanism-bridging an age-old gulf. <i>FEBS Letters</i> , 2018, 592, 463-474.	1.3	9

#	ARTICLE	IF	CITATIONS
2804	Experience with genomic sequencing in pediatric patients with congenital cardiac defects in a large community hospital. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 200-212.	0.6	12
2805	Development of new preclinical models to advance adrenocortical carcinoma research. <i>Endocrine-Related Cancer</i> , 2018, 25, 437-451.	1.6	45
2806	A genome-wide association study in the Japanese population identifies the 12q24 locus for habitual coffee consumption: The J-MICC Study. <i>Scientific Reports</i> , 2018, 8, 1493.	1.6	32
2807	Novel and recurrent RNF213 variants in Japanese pediatric patients with moyamoya disease. <i>Human Genome Variation</i> , 2018, 5, 17060.	0.4	12
2808	A Case of Maturity Onset Diabetes of the Young (MODY3) in a Family with a Novel HNF1A Gene Mutation in Five Generations. <i>Diabetes Therapy</i> , 2018, 9, 413-420.	1.2	12
2809	Accumulation of Mutational Load at the Edges of a Species Range. <i>Molecular Biology and Evolution</i> , 2018, 35, 781-791.	3.5	86
2810	Rare missense mutations in <i>RECQL</i> and <i>POLG</i> associate with inherited predisposition to breast cancer. <i>International Journal of Cancer</i> , 2018, 142, 2286-2292.	2.3	15
2811	<i>MYH9</i> macrothrombocytopenia caused by a novel variant (E1421K) initially presenting as apparent neonatal alloimmune thrombocytopenia. <i>Pediatric Blood and Cancer</i> , 2018, 65, e26949.	0.8	5
2812	BRCA1 and BRCA2 germline variants in breast cancer patients from the Republic of Macedonia. <i>Breast Cancer Research and Treatment</i> , 2018, 168, 745-753.	1.1	8
2813	The protective role of DOT1L in UV-induced melanomagenesis. <i>Nature Communications</i> , 2018, 9, 259.	5.8	63
2814	Spectrum of APC and MUTYH germline mutations in Russian patients with colorectal malignancies. <i>Clinical Genetics</i> , 2018, 93, 1015-1021.	1.0	16
2815	Genetic and functional analysis of SHROOM1-4 in a Chinese neural tube defect cohort. <i>Human Genetics</i> , 2018, 137, 195-202.	1.8	19
2816	Cerebellar ataxia-dominant phenotype in patients with ERCC4 mutations. <i>Journal of Human Genetics</i> , 2018, 63, 417-423.	1.1	15
2817	Genome-Wide Association Study in African Americans with Acute Respiratory Distress Syndrome Identifies the Selectin P Ligand Gene as a Risk Factor. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2018, 197, 1421-1432.	2.5	50
2818	Two microcephaly-associated novel missense mutations in CASK specifically disrupt the CASK-neurexin interaction. <i>Human Genetics</i> , 2018, 137, 231-246.	1.8	29
2819	A Novel FOXL2 Mutation Implying Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome Type I. <i>Cellular Physiology and Biochemistry</i> , 2018, 45, 203-211.	1.1	5
2820	Identification of genomic variants causing sperm abnormalities and reduced male fertility. <i>Animal Reproduction Science</i> , 2018, 194, 57-62.	0.5	32
2821	Orchid: a novel management, annotation and machine learning framework for analyzing cancer mutations. <i>Bioinformatics</i> , 2018, 34, 936-942.	1.8	16

#	ARTICLE	IF	CITATIONS
2822	A novel <i>PKLR</i> gene mutation identified using advanced molecular techniques. <i>Pediatric Transplantation</i> , 2018, 22, e13143.	0.5	6
2823	High Yield of Pathogenic Germline Mutations Causative or Likely Causative of the Cancer Phenotype in Selected Children with Cancer. <i>Clinical Cancer Research</i> , 2018, 24, 1594-1603.	3.2	52
2824	The Plantain Proteome, a Focus on Allele Specific Proteins Obtained from Plantain Fruits. <i>Proteomics</i> , 2018, 18, 1700227.	1.3	10
2825	Inherited Cardiovascular Conditions: Phenotype-Genotype Data Mining and Sharing, and Databases. , 2018, , 869-877.		1
2826	Mitochondrial Mutations in Cholestatic Liver Disease with Biliary Atresia. <i>Scientific Reports</i> , 2018, 8, 905.	1.6	29
2827	Key apoptotic genes APAF1 and CASP9 implicated in recurrent folate-resistant neural tube defects. <i>European Journal of Human Genetics</i> , 2018, 26, 420-427.	1.4	20
2828	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. <i>Brain</i> , 2018, 141, 698-712.	3.7	72
2829	An Atypical Rett Syndrome Phenotype Due to a Novel Missense Mutation in CACNA1A. <i>Journal of Child Neurology</i> , 2018, 33, 286-289.	0.7	18
2830	Constitutional <i>SAMD9L</i> mutations cause familial myelodysplastic syndrome and transient monosomy 7. <i>Haematologica</i> , 2018, 103, 427-437.	1.7	83
2831	Low syndrome: A particularly severe phenotype without clinical kidney involvement. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 460-464.	0.7	4
2832	Spectrum of mutations in index patients with familial hypercholesterolemia in Singapore: Single center study. <i>Atherosclerosis</i> , 2018, 269, 106-116.	0.4	22
2833	Familial Esophageal Squamous Cell Carcinoma with damaging rare/germline mutations in KCNJ12/KCNJ18 and GPRIN2 genes. <i>Cancer Genetics</i> , 2018, 221, 46-52.	0.2	20
2834	Genomic Analysis Reveals Distinct Subtypes in Two Rare Cases of Primary Ovarian Lymphoma. <i>Pathology Research and Practice</i> , 2018, 214, 593-598.	1.0	5
2835	Evaluating the contribution of rare variants to type 2 diabetes and related traits using pedigrees. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 379-384.	3.3	28
2836	Identification of somatic mutations in postmortem human brains by whole genome sequencing and their implications for psychiatric disorders. <i>Psychiatry and Clinical Neurosciences</i> , 2018, 72, 280-294.	1.0	9
2837	Variants in GNPTAB, GNPTG and NAGPA genes are associated with stutterers. <i>Gene</i> , 2018, 647, 93-100.	1.0	15
2838	Interactome INSIDER: a structural interactome browser for genomic studies. <i>Nature Methods</i> , 2018, 15, 107-114.	9.0	133
2839	Protein-altering variants of <i>PTPN2</i> in childhood-onset Type 1A diabetes. <i>Diabetic Medicine</i> , 2018, 35, 376-380.	1.2	7

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2840	Missense Variants in RHOBTB2 Cause a Developmental and Epileptic Encephalopathy in Humans, and Altered Levels Cause Neurological Defects in Drosophila. <i>American Journal of Human Genetics</i> , 2018, 102, 44-57.	2.6	49
2841	MERRF Classification: Implications for Diagnosis and Clinical Trials. <i>Pediatric Neurology</i> , 2018, 80, 8-23.	1.0	58
2842	Rare loss of function mutations in N-methyl-d-aspartate glutamate receptors and their contributions to schizophrenia susceptibility. <i>Translational Psychiatry</i> , 2018, 8, 12.	2.4	41
2843	Characteristics of MUTYH variants in Japanese colorectal polyposis patients. <i>International Journal of Clinical Oncology</i> , 2018, 23, 497-503.	1.0	10
2844	Large-scale exome datasets reveal a new class of adaptor-related protein complex 2 sigma subunit (AP2lf) mutations, located at the interface with the AP2 alpha subunit, that impair calcium-sensing receptor signalling. <i>Human Molecular Genetics</i> , 2018, 27, 901-911.	1.4	15
2845	Variation in Cilia Protein Genes and Progression of Lung Disease in Cystic Fibrosis. <i>Annals of the American Thoracic Society</i> , 2018, 15, 440-448.	1.5	14
2846	Mutation in barley ERA1 (Enhanced Response to ABA1) gene confers better photosynthesis efficiency in response to drought as revealed by transcriptomic and physiological analysis. <i>Environmental and Experimental Botany</i> , 2018, 148, 12-26.	2.0	17
2847	Modeling mutant/wild-type interactions to ascertain pathogenicity of PROKR2 missense variants in patients with isolated GnRH deficiency. <i>Human Molecular Genetics</i> , 2018, 27, 338-350.	1.4	21
2848	Improvement of Self-Injury With Dopamine and Serotonin Replacement Therapy in a Patient With a Hemizygous <i>PAK3</i> Mutation: A New Therapeutic Strategy for Neuropsychiatric Features of an Intellectual Disability Syndrome. <i>Journal of Child Neurology</i> , 2018, 33, 106-113.	0.7	20
2849	A frame-shift deletion in the PURA gene associates with a new clinical finding: Hypoglycorrhachia. Is GLUT1 a new PURA target?. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 331-336.	0.5	17
2850	DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport. <i>Human Molecular Genetics</i> , 2018, 27, 529-545.	1.4	45
2851	Serum caffeine and metabolites are reliable biomarkers of early Parkinson disease. <i>Neurology</i> , 2018, 90, e404-e411.	1.5	70
2852	A survey of functional genomic variation in domesticated chickens. <i>Genetics Selection Evolution</i> , 2018, 50, 17.	1.2	27
2853	Compound heterozygous variants in the multiple PDZ domain protein (MPDZ) cause a case of mild non-progressive communicating hydrocephalus. <i>BMC Medical Genetics</i> , 2018, 19, 34.	2.1	12
2854	Fabry disease in the Spanish population: observational study with detection of 77 patients. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 52.	1.2	20
2855	The use of whole exome sequencing and murine patient derived xenografts as a method of chemosensitivity testing in sarcoma. <i>Clinical Sarcoma Research</i> , 2018, 8, 4.	2.3	4
2856	A novel, homozygous mutation in desert hedgehog (DHH) in a 46, XY patient with dysgenetic testes presenting with primary amenorrhoea: a case report. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2018, 2018, 2.	1.6	14
2857	Germline variants in pancreatic cancer patients with a personal or family history of cancer fulfilling the revised Bethesda guidelines. <i>Journal of Gastroenterology</i> , 2018, 53, 1159-1167.	2.3	7

#	ARTICLE	IF	CITATIONS
2858	A 2-Year-Old Child with Bilateral Ectopis Lentis and a Novel FBN1 Gene Variant Cys129Ser. <i>Journal of Pediatric Genetics</i> , 2018, 07, 083-085.	0.3	1
2859	Endothelial nitric oxide synthase genotype is associated with pulmonary hypertension severity in left heart failure patients. <i>Pulmonary Circulation</i> , 2018, 8, 1-8.	0.8	10
2860	Screening for germline KCNQ1 and KCNE2 mutations in a set of somatotropinoma patients. <i>Endocrine Connections</i> , 2018, 7, 645-652.	0.8	0
2861	Sequencing of intraductal biopsies is feasible and potentially impacts clinical management of patients with indeterminate biliary stricture and cholangiocarcinoma. <i>Clinical and Translational Gastroenterology</i> , 2018, 9, e151.	1.3	11
2862	Survey and evaluation of mutations in the human KLF1 transcription unit. <i>Scientific Reports</i> , 2018, 8, 6587.	1.6	5
2863	Whole-exome sequencing and microRNA profiling reveal PI3K/AKT pathway's involvement in juvenile myelomonocytic leukemia. <i>Quantitative Biology</i> , 2018, 6, 85-97.	0.3	1
2864	Profiling and Leveraging Relatedness in a Precision Medicine Cohort of 92,455 Exomes. <i>American Journal of Human Genetics</i> , 2018, 102, 874-889.	2.6	58
2865	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. <i>Brain</i> , 2018, 141, 1622-1636.	3.7	38
2866	Genetic burden and associations with adverse neurodevelopment in neonates with congenital heart disease. <i>American Heart Journal</i> , 2018, 201, 33-39.	1.2	19
2867	Prognostic value of chromosomal imbalances, gene mutations, and BAP1 expression in uveal melanoma. <i>Genes Chromosomes and Cancer</i> , 2018, 57, 387-400.	1.5	21
2868	Whole-Exome Sequencing Reveals Uncaptured Variation and Distinct Ancestry in the Southern African Population of Botswana. <i>American Journal of Human Genetics</i> , 2018, 102, 731-743.	2.6	38
2869	MoBiDiC Prioritization Algorithm, a Free, Accessible, and Efficient Pipeline for Single-Nucleotide Variant Annotation and Prioritization for Next-Generation Sequencing Routine Molecular Diagnosis. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 465-473.	1.2	13
2870	Whole-Genome Sequence Accuracy Is Improved by Replication in a Population of Mutagenized Sorghum. <i>G3: Genes, Genomes, Genetics</i> , 2018, 8, 1079-1094.	0.8	33
2871	Missense Variants in HIF1A and LACC1 Contribute to Leprosy Risk in Han Chinese. <i>American Journal of Human Genetics</i> , 2018, 102, 794-805.	2.6	42
2872	System for Informatics in the Molecular Pathology Laboratory. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 522-532.	1.2	8
2873	A missense mutation in EBF2 was segregated with imperforate anus in a family across three generations. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1632-1636.	0.7	2
2874	Persistence of the ABCC6 genes and the emergence of the bony skeleton in vertebrates. <i>Scientific Reports</i> , 2018, 8, 6027.	1.6	7
2875	A homozygous mutation p.Arg2167Trp in FREM2 causes isolated cryptophthalmos. <i>Human Molecular Genetics</i> , 2018, 27, 2357-2366.	1.4	14

#	ARTICLE	IF	CITATIONS
2876	CoERG11 A395T mutation confers azole resistance in <i>Candida orthopsilosis</i> clinical isolates. <i>Journal of Antimicrobial Chemotherapy</i> , 2018, 73, 1815-1822.	1.3	19
2877	Mutations in <i>SZT2</i> result in early-onset epileptic encephalopathy and leukoencephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1443-1448.	0.7	15
2878	Analysis of Genes Associated With Monogenic Primary Immunodeficiency Identifies Rare Variants in XIAP in Patients With Crohn's Disease. <i>Gastroenterology</i> , 2018, 154, 2165-2177.	0.6	26
2879	New workflow for classification of genetic variants' pathogenicity applied to hereditary recurrent fevers by the International Study Group for Systemic Autoinflammatory Diseases (INSAID). <i>Journal of Medical Genetics</i> , 2018, 55, 530-537.	1.5	117
2880	Rare human Caspase-6-R65W and Caspase-6-G66R variants identify a novel regulatory region of Caspase-6 activity. <i>Scientific Reports</i> , 2018, 8, 4428.	1.6	9
2881	Weighted Burden Analysis of Exome-Sequenced Case-Control Sample Implicates Synaptic Genes in Schizophrenia Aetiology. <i>Behavior Genetics</i> , 2018, 48, 198-208.	1.4	23
2882	Functional annotation of genomic variants in studies of late-onset Alzheimer's disease. <i>Bioinformatics</i> , 2018, 34, 2724-2731.	1.8	30
2883	Unexplained cardiac arrest: a tale of conflicting interpretations of <i>KCNQ1</i> genetic test results. <i>Clinical Research in Cardiology</i> , 2018, 107, 670-678.	1.5	6
2884	Exome sequencing in neonates: diagnostic rates, characteristics, and time to diagnosis. <i>Genetics in Medicine</i> , 2018, 20, 1468-1471.	1.1	31
2885	Whole-genome sequencing of Atacama skeleton shows novel mutations linked with dysplasia. <i>Genome Research</i> , 2018, 28, 423-431.	2.4	19
2886	Novel Mutation in <i>CECR1</i> Leads to Deficiency of ADA2 with Associated Neutropenia. <i>Journal of Clinical Immunology</i> , 2018, 38, 273-277.	2.0	32
2887	Clinical genetic strategies for early onset neurodegenerative diseases. <i>Molecular and Cellular Toxicology</i> , 2018, 14, 123-142.	0.8	20
2888	Late diagnosis of a truncating <i>WISP3</i> mutation entails a severe phenotype of progressive pseudorheumatoid dysplasia. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a002139.	0.5	6
2889	Advanced Whole-Genome Sequencing and Analysis of Fetal Genomes from Amniotic Fluid. <i>Clinical Chemistry</i> , 2018, 64, 715-725.	1.5	10
2890	Prediction and interpretation of deleterious coding variants in terms of protein structural stability. <i>Scientific Reports</i> , 2018, 8, 4480.	1.6	64
2891	APC:T1556fs and <i>STK11</i> mutations in duodenal adenomas and adenocarcinomas. <i>Surgery Today</i> , 2018, 48, 765-772.	0.7	9
2892	Rare variant analysis in multiply affected families, association studies and functional analysis suggest a role for the <i>ITG14</i> gene in schizophrenia and bipolar disorder. <i>Schizophrenia Research</i> , 2018, 199, 181-188.	1.1	11
2893	Adaptive evolution to a high purine and fat diet of carnivorans revealed by gut microbiomes and host genomes. <i>Environmental Microbiology</i> , 2018, 20, 1711-1722.	1.8	61

#	ARTICLE	IF	CITATIONS
2894	Association of CTH variant with sinusoidal obstruction syndrome in children receiving intravenous busulfan and cyclophosphamide before hematopoietic stem cell transplantation. <i>Pharmacogenomics Journal</i> , 2018, 18, 64-69.	0.9	13
2895	<i>CASP9</i> germline mutation in a family with multiple brain tumors. <i>Brain Pathology</i> , 2018, 28, 94-102.	2.1	11
2896	Global genetic variation of select opiate metabolism genes in self-reported healthy individuals. <i>Pharmacogenomics Journal</i> , 2018, 18, 281-294.	0.9	8
2897	TMEM230 Mutations Are Rare in Han Chinese Patients with Autosomal Dominant Parkinson's Disease. <i>Molecular Neurobiology</i> , 2018, 55, 2851-2855.	1.9	8
2898	Genomic Assessment of Blitz Nevi Suggests Classification as a Subset of Blue Nevus Rather Than Spitz Nevus: Clinical, Histopathologic, and Molecular Analysis of 18 Cases. <i>American Journal of Dermatopathology</i> , 2018, 40, 118-124.	0.3	1
2899	Human <i>MTHFR</i> -G1793A transition may be a protective mutation against male infertility: a genetic association study and <i>in silico</i> analysis. <i>Human Fertility</i> , 2018, 21, 128-136.	0.7	22
2900	Beneficial effect of ustekinumab in familial pityriasis rubra pilaris with a new missense mutation in <i>CARD14</i> . <i>British Journal of Dermatology</i> , 2018, 178, 969-972.	1.4	38
2901	Simulation Based Investigation of Deleterious nsSNPs in ATXN2 Gene and Its Structural Consequence Toward Spinocerebellar Ataxia. <i>Journal of Cellular Biochemistry</i> , 2018, 119, 499-510.	1.2	16
2903	Postmortem Diagnostic Exome Sequencing Identifies a De Novo <i>TUBB3</i> Alteration in a Newborn With Prenatally Diagnosed Hydrocephalus and Suspected Walker-Warburg Syndrome. <i>Pediatric and Developmental Pathology</i> , 2018, 21, 319-323.	0.5	7
2904	Potentially pathogenic germline CHEK2 c.319+2>A among multiple early-onset cancer families. <i>Familial Cancer</i> , 2018, 17, 141-153.	0.9	12
2905	An Integrated Computational Framework to Assess the Mutational Landscape of α -L-iduronidase <i>IDUA</i> Gene. <i>Journal of Cellular Biochemistry</i> , 2018, 119, 555-565.	1.2	10
2906	Novel biallelic <i>SZT2</i> mutations in 3 cases of early-onset epileptic encephalopathy. <i>Clinical Genetics</i> , 2018, 93, 266-274.	1.0	25
2907	Analysis of selected genes associated with cardiomyopathy by next-generation sequencing. <i>Journal of Clinical Laboratory Analysis</i> , 2018, 32, .	0.9	12
2908	Double somatic mutations in mismatch repair genes are frequent in colorectal cancer after Hodgkin's lymphoma treatment. <i>Gut</i> , 2018, 67, 447-455.	6.1	27
2909	In Silico Analysis of nsSNPs of Carp TLR22 Gene Affecting its Binding Ability with Poly I:C. <i>Interdisciplinary Sciences, Computational Life Sciences</i> , 2018, 10, 641-652.	2.2	10
2910	Mutation Spectrum and Genotype-Phenotype Analyses in a Pakistani Cohort With Hemophilia B. <i>Clinical and Applied Thrombosis/Hemostasis</i> , 2018, 24, 741-748.	0.7	6
2911	Novel clinical and molecular findings in Spanish patients with naevoid basal cell carcinoma syndrome. <i>British Journal of Dermatology</i> , 2018, 178, 198-206.	1.4	5
2912	Serine proteases of the complement lectin pathway and their genetic variations in ischaemic stroke. <i>Journal of Clinical Pathology</i> , 2018, 71, 141-147.	1.0	13

#	ARTICLE	IF	CITATIONS
2913	<i><sc>NDUFA9</sc></i> point mutations cause a variable mitochondrial complex I assembly defect. <i>Clinical Genetics</i> , 2018, 93, 111-118.	1.0	36
2914	Neurexin gene family variants as risk factors for autism spectrum disorder. <i>Autism Research</i> , 2018, 11, 37-43.	2.1	53
2915	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis and nephrocalcinosis. <i>Kidney International</i> , 2018, 93, 204-213.	2.6	133
2916	A profound computational study to prioritize the disease-causing mutations in PRPS1 gene. <i>Metabolic Brain Disease</i> , 2018, 33, 589-600.	1.4	49
2917	Utility of genetics for risk stratification in pediatric hypertrophic cardiomyopathy. <i>Clinical Genetics</i> , 2018, 93, 310-319.	1.0	56
2918	Mutation spectrum of <i>NDP</i>, <i>FZD4</i> and <i>TSPAN12</i> genes in Indian patients with retinopathy of prematurity. <i>British Journal of Ophthalmology</i> , 2018, 102, 276-281.	2.1	17
2919	m6AVar: a database of functional variants involved in m6A modification. <i>Nucleic Acids Research</i> , 2018, 46, D139-D145.	6.5	181
2920	A novel mutation of the EYA4 gene associated with post-lingual hearing loss in a proband is co-segregating with a novel PAX3 mutation in two congenitally deaf family members. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2018, 104, 88-93.	0.4	12
2921	Whole Exome Sequencing allows the identification of two novel groups of Xeroderma pigmentosum in Tunisia, XP-D and XP-E: Impact on molecular diagnosis. <i>Journal of Dermatological Science</i> , 2018, 89, 172-180.	1.0	20
2922	Whole exome sequencing identified a pathogenic mutation in RYR2 in a Chinese family with unexplained sudden death. <i>Journal of Electrocardiology</i> , 2018, 51, 309-315.	0.4	9
2923	Catching hidden variation: systematic correction of reference minor allele annotation in clinical variant calling. <i>Genetics in Medicine</i> , 2018, 20, 360-364.	1.1	26
2924	A de novo mutation in RPL10 causes a rare X-linked ribosomopathy characterized by syndromic intellectual disability and epilepsy: A new case and review of the literature. <i>European Journal of Medical Genetics</i> , 2018, 61, 89-93.	0.7	22
2925	Clinical efficacy of a next-generation sequencing gene panel for primary immunodeficiency diagnostics. <i>Clinical Genetics</i> , 2018, 93, 647-655.	1.0	63
2926	Full-gene haplotypes refine CYP2D6 metabolizer phenotype inferences. <i>International Journal of Legal Medicine</i> , 2018, 132, 1007-1024.	1.2	11
2927	Functional mutant GATA4 identification and potential application in preimplantation diagnosis of congenital heart diseases. <i>Gene</i> , 2018, 641, 349-354.	1.0	15
2928	Novel Mutation of <i>LRP6</i> Identified in Chinese Han Population Links Canonical WNT Signaling to Neural Tube Defects. <i>Birth Defects Research</i> , 2018, 110, 63-71.	0.8	22
2929	DICER1 syndrome: Approach to testing and management at a large pediatric tertiary care center. <i>Pediatric Blood and Cancer</i> , 2018, 65, e26720.	0.8	33
2930	Mutational landscape of RNA-binding proteins in human cancers. <i>RNA Biology</i> , 2018, 15, 115-129.	1.5	87

#	ARTICLE	IF	CITATIONS
2931	Diffuse mesangial sclerosis in a PDSS2 mutation-induced coenzyme Q10 deficiency. <i>Pediatric Nephrology</i> , 2018, 33, 439-446.	0.9	18
2932	A pleiotropic effect of the <i>APOE</i> gene: association of <i>APOE</i> polymorphisms with multibacillary leprosy in Han Chinese from Southwest China. <i>British Journal of Dermatology</i> , 2018, 178, 931-939.	1.4	15
2933	Analysis of clinically relevant somatic mutations in high-risk head and neck cutaneous squamous cell carcinoma. <i>Modern Pathology</i> , 2018, 31, 275-287.	2.9	37
2934	Insights From Molecular Characterization of Adult Patients of Families With Multigenerational Diabetes. <i>Diabetes</i> , 2018, 67, 137-145.	0.3	23
2935	Missense variants in the chromatin remodeler <i>CHD1</i> are associated with neurodevelopmental disability. <i>Journal of Medical Genetics</i> , 2018, 55, 561-566.	1.5	49
2936	Biallelic variants in <i>LINGO1</i> are associated with autosomal recessive intellectual disability, microcephaly, speech and motor delay. <i>Genetics in Medicine</i> , 2018, 20, 778-784.	1.1	21
2937	Prospective cohort study for identification of underlying genetic causes in neonatal encephalopathy using whole-exome sequencing. <i>Genetics in Medicine</i> , 2018, 20, 486-494.	1.1	38
2938	Missense mutations of <i>NCPAG</i> gene affect calving ease in Piedmontese cattle: preliminary evidences. <i>Italian Journal of Animal Science</i> , 2018, 17, 301-305.	0.8	0
2939	Malignant Hyperthermia in the Post-Genomics Era. <i>Anesthesiology</i> , 2018, 128, 168-180.	1.3	120
2940	Exome array analysis identifies <i>ETFB</i> as a novel susceptibility gene for anthracycline-induced cardiotoxicity in cancer patients. <i>Breast Cancer Research and Treatment</i> , 2018, 167, 249-256.	1.1	23
2941	Loss-of-Function <i>GRHL3</i> Variants Detected in African Patients with Isolated Cleft Palate. <i>Journal of Dental Research</i> , 2018, 97, 41-48.	2.5	28
2942	Construction of an Exome-Wide Risk Score for Schizophrenia Based on a Weighted Burden Test. <i>Annals of Human Genetics</i> , 2018, 82, 11-22.	0.3	8
2943	Distinct Patterns of Acral Melanoma Based on Site and Relative Sun Exposure. <i>Journal of Investigative Dermatology</i> , 2018, 138, 384-393.	0.3	44
2944	Association Between Germline Mutations in <i>BRF1</i> , a Subunit of the RNA Polymerase III Transcription Complex, and Hereditary Colorectal Cancer. <i>Gastroenterology</i> , 2018, 154, 181-194.e20.	0.6	32
2945	Investigating <i>CCNF</i> mutations in a Taiwanese cohort with amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2018, 62, 243.e1-243.e6.	1.5	13
2946	Diagnosis and treatment of MYH9-RD in an Australasian cohort with thrombocytopenia. <i>Platelets</i> , 2018, 29, 793-800.	1.1	15
2947	<i>OPTN</i> p.Met468Arg and <i>ATXN2</i> intermediate length polyQ extension in families with C9orf72 mediated amyotrophic lateral sclerosis and frontotemporal dementia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018, 177, 75-85.	1.1	12
2948	Impact of Receiving Secondary Results from Genomic Research: A 12-Month Longitudinal Study. <i>Journal of Genetic Counseling</i> , 2018, 27, 709-722.	0.9	26

#	ARTICLE	IF	CITATIONS
2949	Sequence analysis of the <i>N</i> -acetyltransferase 2 gene (NAT2) among Jordanian volunteers. <i>Libyan Journal of Medicine</i> , 2018, 13, 1408381.	0.8	19
2950	Pharmacogenomic Biomarkers for Improved Drug Therapy—Recent Progress and Future Developments. <i>AAPS Journal</i> , 2018, 20, 4.	2.2	106
2951	Evaluation of exome filtering techniques for the analysis of clinically relevant genes. <i>Human Mutation</i> , 2018, 39, 197-201.	1.1	13
2952	In Silico Approach to Investigate the Structural and Functional Attributes of Familial Hypercholesterolemia Variants Reported in the Saudi Population. <i>Journal of Computational Biology</i> , 2018, 25, 170-181.	0.8	4
2953	A novel homozygous <i>SLC25A1</i> mutation with impaired mitochondrial complex V: Possible phenotypic expansion. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 330-336.	0.7	14
2954	Mutation burden profile in familial Alzheimer's disease cases from India. <i>Neurobiology of Aging</i> , 2018, 64, 158.e7-158.e13.	1.5	11
2955	Properties of human genes guided by their enrichment in rare and common variants. <i>Human Mutation</i> , 2018, 39, 365-370.	1.1	13
2956	Analysis of TSC1 mutation spectrum in mucosal melanoma. <i>Journal of Cancer Research and Clinical Oncology</i> , 2018, 144, 257-267.	1.2	12
2957	A targeted sequencing panel identifies rare damaging variants in multiple genes in the cranial neural tube defect, anencephaly. <i>Clinical Genetics</i> , 2018, 93, 870-879.	1.0	29
2958	Predicted activity of UGT2B7, ABCB1, OPRM1, and COMT using full-gene haplotypes and their association with the CYP2D6-inferred metabolizer phenotype. <i>Forensic Science International: Genetics</i> , 2018, 33, 48-58.	1.6	4
2959	Delayed diagnosis in X-linked agammaglobulinemia and its relationship to the occurrence of mutations in BTK non-kinase domains. <i>Expert Review of Clinical Immunology</i> , 2018, 14, 83-93.	1.3	8
2960	Genetic study of non-syndromic tooth agenesis through the screening of paired box 9, msh homeobox 1, axin 2, and Wnt family member 10A genes: a case-series. <i>European Journal of Oral Sciences</i> , 2018, 126, 24-32.	0.7	8
2961	Recurrence of reported <i>CDH23</i> mutations causing DFNB12 in a special cohort of South Indian hearing impaired assortative mating families—an evaluation. <i>Annals of Human Genetics</i> , 2018, 82, 119-126.	0.3	12
2962	Therapeutic Potential of Afatinib for Cancers with <i>ERBB2</i> (<i>HER2</i>) Transmembrane Domain Mutations G660D and V659E. <i>Oncologist</i> , 2018, 23, 150-154.	1.9	25
2963	Tarsal-carpal coalition syndrome: Report of a novel missense mutation in <i>NOG</i> gene and phenotypic delineation. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 219-224.	0.7	12
2964	Exome Sequencing Identifies LOXL2 Mutation as a Cause of Familial Intracranial Aneurysm. <i>World Neurosurgery</i> , 2018, 109, e812-e818.	0.7	22
2965	Activating <i>FGFR1</i> Mutations in Sporadic Pheochromocytomas. <i>World Journal of Surgery</i> , 2018, 42, 482-489.	0.8	13
2966	Identification of <i>de novo</i> germline mutations and causal genes for sporadic diseases using trio-based whole-exome/genome sequencing. <i>Biological Reviews</i> , 2018, 93, 1014-1031.	4.7	35

#	ARTICLE	IF	CITATIONS
2967	Genotype phenotype correlation in Asian Indian von Hippel-Lindau (VHL) syndrome patients with pheochromocytoma/paraganglioma. <i>Familial Cancer</i> , 2018, 17, 441-449.	0.9	10
2968	Identification of a novel mutation in the FGFR3 gene in a Chinese family with Hypochondroplasia. <i>Gene</i> , 2018, 641, 355-360.	1.0	10
2969	Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2018, 13, 53-62.	2.2	170
2970	How chromosomal deletions can unmask recessive mutations? Deletions in 10q11.2 associated with <i>CHAT</i> or <i>SLC18A3</i> mutations lead to congenital myasthenic syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 151-155.	0.7	16
2971	SNPs in the Toll1 receptor of <i>Litopenaeus vannamei</i> are associated with immune response. <i>Fish and Shellfish Immunology</i> , 2018, 72, 410-417.	1.6	13
2972	A Missense Variant p.Ala117Ser in the Transthyretin Gene of a Han Chinese Family with Familial Amyloid Polyneuropathy. <i>Molecular Neurobiology</i> , 2018, 55, 4911-4917.	1.9	17
2973	Neurodevelopmental disorders in children with macrocephaly: A prevalence study and PTEN gene analysis. <i>Brain and Development</i> , 2018, 40, 36-41.	0.6	14
2974	Plasma cell deficiency in human subjects with heterozygous mutations in Sec61 translocon alpha 1 subunit (SEC61A1). <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 1427-1438.	1.5	63
2975	Analysis of <i>KERA</i> in four families with cornea plana identifies two novel mutations. <i>Acta Ophthalmologica</i> , 2018, 96, e87-e91.	0.6	4
2976	Hereditary angioedema with a mutation in the plasminogen gene. <i>Allergy: European Journal of Allergy and Clinical Immunology</i> , 2018, 73, 442-450.	2.7	225
2977	<i>TP53</i> Germline Variations Influence the Predisposition and Prognosis of B-Cell Acute Lymphoblastic Leukemia in Children. <i>Journal of Clinical Oncology</i> , 2018, 36, 591-599.	0.8	121
2978	Ensembl variation resources. Database: the Journal of Biological Databases and Curation, 2018, 2018, .	1.4	377
2979	Mitochondrial gene sequence variants in children with severe malaria anaemia with or without lactic acidosis: a case control study. <i>Malaria Journal</i> , 2018, 17, 467.	0.8	3
2980	Representativeness of variation benchmark datasets. <i>BMC Bioinformatics</i> , 2018, 19, 461.	1.2	18
2981	Variant information systems for precision oncology. <i>BMC Medical Informatics and Decision Making</i> , 2018, 18, 107.	1.5	10
2982	The genomics of desmoplastic small round cell tumor reveals the deregulation of genes related to DNA damage response, epithelial-mesenchymal transition, and immune response. <i>Cancer Communications</i> , 2018, 38, 1-14.	3.7	25
2983	Analysis of <i>VSX1</i> variations in Brazilian subjects with keratoconus. <i>Journal of Ophthalmic and Vision Research</i> , 2018, 13, 266.	0.7	6
2984	Spontaneous Spongiform Brainstem Degeneration in a Young Mouse Lemur (<i>Microcebus murinus</i>) with Conspicuous Behavioral, Motor, Growth, and Ocular Pathologies. <i>Comparative Medicine</i> , 2018, 68, 489-495.	0.4	2

#	ARTICLE	IF	CITATIONS
2985	Phenotype Prediction of Pathogenic Nonsynonymous Single Nucleotide Polymorphisms in Insulin with Bioinformatics Tools. , 2018, , .		0
2986	Comparative Analysis of Tools for Predicting the Functional Impact of mtDNA Variants. , 2018, , .		0
2987	dbCPM: a manually curated database for exploring the cancer passenger mutations. Briefings in Bioinformatics, 2018, , .	3.2	10
2988	Personalized Clinical Decision Making Through Implementation of a Molecular Tumor Board: A German Single-Center Experience. JCO Precision Oncology, 2018, 2, 1-16.	1.5	41
2989	A multi-omic atlas of the human frontal cortex for aging and Alzheimer's disease research. Scientific Data, 2018, 5, 180142.	2.4	357
2990	Status of BRCA1/2 Genetic Testing Practices in Korea (2014). Laboratory Medicine Online, 2018, 8, 107.	0.0	2
2991	Identification of the Genetic Variation and Gene Exchange between Citrus Trifoliata and Citrus Clementina. Biomolecules, 2018, 8, 182.	1.8	4
2992	Mutational analysis of selected high-grade malignancies in a premenopausal gynecologic cancer population: a potential for targeted therapies?. Applied Cancer Research, 2018, 38, .	1.0	1
2993	Mutation Based Structural Modelling and Dynamics Study of Alpha Fetoprotein: An Insight to Inhibitory Mechanism in Breast Cancer. Journal of Proteomics and Bioinformatics, 2018, 11, .	0.4	1
2994	Assessment of coding region variants in Kuwaiti population: implications for medical genetics and population genomics. Scientific Reports, 2018, 8, 16583.	1.6	26
2995	Recent developments in statistical methods for GWAS and high-throughput sequencing association studies of complex traits. Biostatistics and Epidemiology, 2018, 2, 132-159.	0.4	3
2996	Targeted next generation sequencing in patients with maturity-onset diabetes of the young (MODY). Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 1295-1304.	0.4	23
2997	Cell-Free DNA From Metastatic Pancreatic Neuroendocrine Tumor Patients Contains Tumor-Specific Mutations and Copy Number Variations. Frontiers in Oncology, 2018, 8, 467.	1.3	25
2998	Next generation sequencing-based molecular diagnosis in familial congenital cataract expands the mutational spectrum in known congenital cataract genes. American Journal of Medical Genetics, Part A, 2018, 176, 2637-2645.	0.7	22
2999	High-throughput sequencing for the molecular diagnosis of Usher syndrome reveals 42 novel mutations and consolidates CEP250 as Usher-like disease causative. Scientific Reports, 2018, 8, 17113.	1.6	30
3000	Helios is a key transcriptional regulator of outer hair cell maturation. Nature, 2018, 563, 696-700.	13.7	90
3001	Molecular characterization of Axenfeld-Rieger spectrum and other anterior segment dysgeneses in a sample of Mexican patients. Ophthalmic Genetics, 2018, 39, 728-734.	0.5	3
3002	Role of variant allele fraction and rare SNP filtering to improve cellular DNA repair endpoint association. PLoS ONE, 2018, 13, e0206632.	1.1	2

#	ARTICLE	IF	CITATIONS
3003	Loss of tubulin deglutamylase <sc>CCP</sc> 1 causes infantile-onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
3004	<i>PSMD12</i> haploinsufficiency in a neurodevelopmental disorder with autistic features. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 736-745.	1.1	23
3005	Exploring the Impact of Single-Nucleotide Polymorphisms on Translation. Frontiers in Genetics, 2018, 9, 507.	1.1	128
3006	Integration of genome wide association studies and whole genome sequencing provides novel insights into fat deposition in chicken. Scientific Reports, 2018, 8, 16222.	1.6	29
3007	Functional characterization of TRPM4 variants identified in sudden unexpected natural death. Forensic Science International, 2018, 293, 37-46.	1.3	11
3008	Structural Biology Helps Interpret Variants of Uncertain Significance in Genes Causing Endocrine and Metabolic Disorders. Journal of the Endocrine Society, 2018, 2, 842-854.	0.1	7
3009	Report of the Third Family with Multiple Mitochondrial Dysfunctions Syndrome 5 Caused by the Founder Variant p.(Glu87Lys) in ISCA1. Journal of Pediatric Genetics, 2018, 07, 130-133.	0.3	8
3010	Identification of Compound Heterozygous EYS Variants in a Korean Patient with Retinitis Pigmentosa. Laboratory Medicine Online, 2018, 8, 66.	0.0	1
3011	Molecular and clinical analysis of 27 German patients with Leber congenital amaurosis. PLoS ONE, 2018, 13, e0205380.	1.1	38
3012	Inherited selective cobalamin malabsorption in Komondor dogs associated with a CUBN splice site variant. BMC Veterinary Research, 2018, 14, 418.	0.7	9
3013	Calculating the statistical significance of rare variants causal for Mendelian and complex disorders. BMC Medical Genomics, 2018, 11, 53.	0.7	13
3014	Identification of deleterious and regulatory genomic variations in known asthma loci. Respiratory Research, 2018, 19, 248.	1.4	5
3015	The evolutionary pattern of mutations in glioblastoma reveals therapy-mediated selection. Oncotarget, 2018, 9, 7844-7858.	0.8	29
3016	Whole-Genome Sequencing Identifies a Novel Variation of WAS Gene Coordinating With Heterozygous Germline Mutation of APC to Enhance Hepatoblastoma Oncogenesis. Frontiers in Genetics, 2018, 9, 668.	1.1	11
3017	Mutations in the <i>MRPS28</i> gene encoding the small mitoribosomal subunit protein bS1m in a patient with intrauterine growth retardation, craniofacial dysmorphism and multisystemic involvement. Human Molecular Genetics, 2019, 28, 1445-1462.	1.4	19
3018	Analysis of human MutS homolog 2 missense mutations in patients with colorectal cancer. Oncology Letters, 2018, 15, 6275-6282.	0.8	0
3019	Novel variants in the ACTA2 and MYH11 genes in a Cypriot family with thoracic aortic aneurysms: a case report. BMC Medical Genetics, 2018, 19, 208.	2.1	5
3020	A resource of variant effect predictions of single nucleotide variants in model organisms. Molecular Systems Biology, 2018, 14, e8430.	3.2	84

#	ARTICLE	IF	CITATIONS
3021	Association between Polymorphisms in Inflammatory Response-Related Genes and the Susceptibility, Progression and Prognosis of the Diffuse Histological Subtype of Gastric Cancer. <i>Genes</i> , 2018, 9, 631.	1.0	18
3022	High-resolution clonal mapping of multi-organ metastasis in triple negative breast cancer. <i>Nature Communications</i> , 2018, 9, 5079.	5.8	91
3023	Whole Genome Sequencing of a Vietnamese Family from a Dioxin Contamination Hotspot Reveals Novel Variants in the Son with Undiagnosed Intellectual Disability. <i>International Journal of Environmental Research and Public Health</i> , 2018, 15, 2629.	1.2	4
3024	Expanding clinical phenotype in <i>CACNA1C</i> related disorders: From neonatal onset severe epileptic encephalopathy to late-onset epilepsy. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2733-2739.	0.7	30
3025	Combined CNV, haplotyping and whole exome sequencing enable identification of two distinct novel EYS mutations causing RP in a single inbred tribe. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2695-2703.	0.7	3
3026	Atrial Structural Remodeling Gene Variants in Patients with Atrial Fibrillation. <i>BioMed Research International</i> , 2018, 2018, 1-12.	0.9	11
3027	Novel combined variants of LDLR and LDLRAP1 genes causing severe familial hypercholesterolemia. <i>Atherosclerosis</i> , 2018, 277, 425-433.	0.4	15
3028	GWAS and eQTL analysis identifies a SNP associated with both residual feed intake and GFRA2 expression in beef cattle. <i>Scientific Reports</i> , 2018, 8, 14301.	1.6	48
3029	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. <i>Human Molecular Genetics</i> , 2019, 28, 258-268.	1.4	19
3030	Dusp6 is a genetic modifier of growth through enhanced ERK activity. <i>Human Molecular Genetics</i> , 2018, 28, 279-289.	1.4	6
3031	Urea cycle disorders in India: clinical course, biochemical and genetic investigations, and prenatal testing. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 174.	1.2	24
3032	Systematic analysis of genetic variants in patients with essential tremor. <i>Brain and Behavior</i> , 2018, 8, e01100.	1.0	8
3033	The clinical spectrum of CASQ1-related myopathy. <i>Neurology</i> , 2018, 91, e1629-e1641.	1.5	14
3034	Burden Testing of Rare Variants Identified through Exome Sequencing via Publicly Available Control Data. <i>American Journal of Human Genetics</i> , 2018, 103, 522-534.	2.6	132
3035	Addition of triple negativity of breast cancer as an indicator for germline mutations in predisposing genes increases sensitivity of clinical selection criteria. <i>BMC Cancer</i> , 2018, 18, 926.	1.1	16
3036	Fourteen new mutations of BCKDHA, BCKDHB and DBT genes associated with maple syrup urine disease (MSUD) in Malaysian population. <i>Molecular Genetics and Metabolism Reports</i> , 2018, 17, 22-30.	0.4	14
3037	Early-Onset Invasive Infection Due to <i>Corynespora cassicola</i> Associated with Compound Heterozygous CARD9 Mutations in a Colombian Patient. <i>Journal of Clinical Immunology</i> , 2018, 38, 794-803.	2.0	40
3038	Identification of a novel LDLR disease-causing variant using capture-based next-generation sequencing screening of familial hypercholesterolemia patients in Taiwan. <i>Atherosclerosis</i> , 2018, 277, 440-447.	0.4	11

#	ARTICLE	IF	CITATIONS
3039	Identification of a novel B allele with multiple missense mutations in a Chinese family with a Bw phenotype. <i>Transfusion</i> , 2018, 58, 2741-2742.	0.8	0
3040	HIF-2 α -pVHL complex reveals broad genotype-phenotype correlations in HIF-2 α -driven disease. <i>Nature Communications</i> , 2018, 9, 3359.	5.8	26
3041	Pathogenic variants in glutamyl-tRNA ^{Gln} amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. <i>Nature Communications</i> , 2018, 9, 4065.	5.8	44
3042	Bi-allelic Loss-of-Function Variants in DNMBP Cause Infantile Cataracts. <i>American Journal of Human Genetics</i> , 2018, 103, 568-578.	2.6	29
3043	9p24 triplication in syndromic hydrocephalus with diffuse villous hyperplasia of the choroid plexus. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003145.	0.5	8
3044	<i>GRIN2D</i> variants in three cases of developmental and epileptic encephalopathy. <i>Clinical Genetics</i> , 2018, 94, 538-547.	1.0	17
3045	BRCA1/2 mutations are not a common cause of malignant melanoma in the Polish population. <i>PLoS ONE</i> , 2018, 13, e0204768.	1.1	6
3046	ENTPRISE-X: Predicting disease-associated frameshift and nonsense mutations. <i>PLoS ONE</i> , 2018, 13, e0196849.	1.1	20
3047	Association of single-nucleotide polymorphisms of the KEAP1 gene with the risk of various human diseases and its functional impact using in silico analysis. <i>Pharmacological Research</i> , 2018, 137, 205-218.	3.1	10
3048	De novo Mutations (DNMs) in Autism Spectrum Disorder (ASD): Pathway and Network Analysis. <i>Frontiers in Genetics</i> , 2018, 9, 406.	1.1	40
3049	Phenotypic expression of a spectrum of Neurofibromatosis Type 1 (NF1) mutations identified through NGS and MLPA. <i>Journal of the Neurological Sciences</i> , 2018, 395, 95-105.	0.3	29
3050	Prioritization of Variants Detected by Next Generation Sequencing According to the Mutation Tolerance and Mutational Architecture of the Corresponding Genes. <i>International Journal of Molecular Sciences</i> , 2018, 19, 1584.	1.8	16
3052	Evidence of peripheral olfactory impairment in the domestic silkworms: insight from the comparative transcriptome and population genetics. <i>BMC Genomics</i> , 2018, 19, 788.	1.2	14
3053	Exome sequencing in large, multiplex bipolar disorder families from Cuba. <i>PLoS ONE</i> , 2018, 13, e0205895.	1.1	13
3054	Autosomal dominant tubulointerstitial kidney disease-UMOD is the most frequent non polycystic genetic kidney disease. <i>BMC Nephrology</i> , 2018, 19, 301.	0.8	39
3055	Novel sequence variants in the TLR6 gene associated with advanced breast cancer risk in the Saudi Arabian population. <i>PLoS ONE</i> , 2018, 13, e0203376.	1.1	16
3056	Identification of a Homozygous Missense Mutation in the TYR Gene in a Chinese Family with OCA1. <i>Current Medical Science</i> , 2018, 38, 932-936.	0.7	1
3057	Genomic landscapes of Chinese sporadic autism spectrum disorders revealed by whole-genome sequencing. <i>Journal of Genetics and Genomics</i> , 2018, 45, 527-538.	1.7	33

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3058	BRCA-analyzer: Automatic workflow for processing NGS reads of BRCA1 and BRCA2 genes. <i>Computational Biology and Chemistry</i> , 2018, 77, 297-306.	1.1	10
3059	Phenotypic and transcriptomic characterization of a wheat tall mutant carrying an induced mutation in the C-terminal PFYRE motif of RHT-B1b. <i>BMC Plant Biology</i> , 2018, 18, 253.	1.6	15
3060	Impact of structural prior knowledge in SNV prediction: Towards causal variant finding in rare disease. <i>PLoS ONE</i> , 2018, 13, e0204101.	1.1	3
3061	Purging of Strongly Deleterious Mutations Explains Long-Term Persistence and Absence of Inbreeding Depression in Island Foxes. <i>Current Biology</i> , 2018, 28, 3487-3494.e4.	1.8	140
3062	Variants in <i>NKX2-5</i> and <i>FLNC</i> Cause Dilated Cardiomyopathy and Sudden Cardiac Death. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002151.	1.6	27
3063	Unique aspects of sequence variant interpretation for inborn errors of metabolism (IEM): The ClinGen IEM Working Group and the Phenylalanine Hydroxylase Gene. <i>Human Mutation</i> , 2018, 39, 1569-1580.	1.1	50
3064	A recessive lethal chondrodysplasia in a miniature zebu family results from an insertion affecting the chondroitin sulfat domain of aggrecan. <i>BMC Genetics</i> , 2018, 19, 91.	2.7	5
3065	Syndromic hearing loss molecular diagnosis: Application of massive parallel sequencing. <i>Hearing Research</i> , 2018, 370, 181-188.	0.9	6
3066	Gain-of-function <i>IKBKB</i> mutation causes human combined immune deficiency. <i>Journal of Experimental Medicine</i> , 2018, 215, 2715-2724.	4.2	69
3067	Genome Sequence of Peacock Reveals the Peculiar Case of a Glittering Bird. <i>Frontiers in Genetics</i> , 2018, 9, 392.	1.1	32
3068	3T MRI study discloses high intrafamilial variability in CADASIL due to a novel NOTCH3 mutation. <i>Journal of Clinical Neuroscience</i> , 2018, 58, 25-29.	0.8	2
3069	Sudden unexpected death in the young – Value of massive parallel sequencing in postmortem genetic analyses. <i>Forensic Science International</i> , 2018, 293, 70-76.	1.3	17
3070	Molecular diagnosis of inherited peripheral neuropathies by targeted next-generation sequencing: molecular spectrum delineation. <i>BMJ Open</i> , 2018, 8, e021632.	0.8	36
3071	Genetics of Monogenic Diabetes: Present Clinical Challenges. <i>Current Diabetes Reports</i> , 2018, 18, 141.	1.7	50
3072	In silico approach to identify non-synonymous SNPs with highest predicted deleterious effect on protein function in human obesity related gene, neuronal growth regulator 1 (NEGR1). <i>3 Biotech</i> , 2018, 8, 466.	1.1	9
3073	Deciphering the Emerging Complexities of Molecular Mechanisms at GWAS Loci. <i>American Journal of Human Genetics</i> , 2018, 103, 637-653.	2.6	93
3074	Biallelic Mutations in LRRC56, Encoding a Protein Associated with Intraflagellar Transport, Cause Mucociliary Clearance and Laterality Defects. <i>American Journal of Human Genetics</i> , 2018, 103, 727-739.	2.6	49
3075	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. <i>American Journal of Human Genetics</i> , 2018, 103, 752-768.	2.6	40

#	ARTICLE	IF	CITATIONS
3076	A de novo variant in ADGRL2 suggests a novel mechanism underlying the previously undescribed association of extreme microcephaly with severely reduced sulcation and rhombencephalosynapsis. <i>Acta Neuropathologica Communications</i> , 2018, 6, 109.	2.4	20
3077	Two Novel Candidate Genes for Insulin Secretion Identified by Comparative Genomics of Multiple Backcross Mouse Populations. <i>Genetics</i> , 2018, 210, 1527-1542.	1.2	17
3078	DermaGene and VitmiRS: a comprehensive systems analysis of genetic dermatological disorders. <i>Biomedical Dermatology</i> , 2018, 2, .	7.6	1
3079	HS6ST1 Insufficiency Causes Self-Limited Delayed Puberty in Contrast With Other GnRH Deficiency Genes. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 3420-3429.	1.8	38
3080	A Bayesian framework for efficient and accurate variant prediction. <i>PLoS ONE</i> , 2018, 13, e0203553.	1.1	12
3081	Selection at a genomic region of major effect is responsible for evolution of complex life histories in anadromous steelhead. <i>BMC Evolutionary Biology</i> , 2018, 18, 140.	3.2	41
3082	A case of perinatal hypophosphatasia with a novel mutation in the <i>ALPL</i> gene: clinical course and review of the literature. <i>Clinical Pediatric Endocrinology</i> , 2018, 27, 179-186.	0.4	14
3083	Identification of a novel <i>MYO6</i> mutation associated with autosomal dominant non-syndromic hearing loss in a Chinese family by whole-exome sequencing. <i>Genes and Genetic Systems</i> , 2018, 93, 171-179.	0.2	8
3084	Differences in clinical characteristics and mutational pattern between synchronous and metachronous colorectal liver metastases. <i>Cancer Management and Research</i> , 2018, Volume 10, 2871-2881.	0.9	11
3085	Utility of trio-based exome sequencing in the elucidation of the genetic basis of isolated syndromic intellectual disability: illustrative cases. <i>The Application of Clinical Genetics</i> , 2018, Volume 11, 93-98.	1.4	34
3086	Identification of novel pathogenic variants and novel gene-phenotype correlations in Mexican subjects with microphthalmia and/or anophthalmia by next-generation sequencing. <i>Journal of Human Genetics</i> , 2018, 63, 1169-1180.	1.1	16
3087	Functional Characterization of a GGPPS Variant Identified in Atypical Femoral Fracture Patients and Delineation of the Role of GGPPS in Bone-Relevant Cell Types. <i>Journal of Bone and Mineral Research</i> , 2018, 33, 2091-2098.	3.1	21
3088	Minimal functional driver gene heterogeneity among untreated metastases. <i>Science</i> , 2018, 361, 1033-1037.	6.0	223
3089	Novel and de novo mutations in pediatric refractory epilepsy. <i>Molecular Brain</i> , 2018, 11, 48.	1.3	70
3090	Assessing Human Genetic Variations in Glucose Transporter SLC2A10 and Their Role in Altering Structural and Functional Properties. <i>Frontiers in Genetics</i> , 2018, 9, 276.	1.1	12
3091	Positive and balancing selection on <i>SLC18A1</i> gene associated with psychiatric disorders and human-unique personality traits. <i>Evolution Letters</i> , 2018, 2, 499-510.	1.6	16
3092	Genomic screening in rare disorders: New mutations and phenotypes, highlighting <i>ALG14</i> as a novel cause of severe intellectual disability. <i>Clinical Genetics</i> , 2018, 94, 528-537.	1.0	29
3093	Schnyder corneal dystrophy and associated phenotypes caused by novel and recurrent mutations in the UBIAD1 gene. <i>BMC Ophthalmology</i> , 2018, 18, 250.	0.6	9

#	ARTICLE	IF	CITATIONS
3094	Balancing selection on a recessive lethal deletion with pleiotropic effects on two neighboring genes in the porcine genome. <i>PLoS Genetics</i> , 2018, 14, e1007661.	1.5	39
3095	Genomic Landscape and Mutational Signatures of Deafness-Associated Genes. <i>American Journal of Human Genetics</i> , 2018, 103, 484-497.	2.6	214
3096	Accurate classification of BRCA1 variants with saturation genome editing. <i>Nature</i> , 2018, 562, 217-222.	13.7	570
3097	Analytical Approaches for Exome Sequence Data. <i>Translational Bioinformatics</i> , 2018, , 121-136.	0.0	0
3098	Correlation of genomic alterations between tumor tissue and circulating tumor DNA by next-generation sequencing. <i>Journal of Cancer Research and Clinical Oncology</i> , 2018, 144, 2167-2175.	1.2	9
3099	Screening of known disease genes in congenital scoliosis. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 966-974.	0.6	20
3100	Identification of a pathogenic PMP2 variant in a multi-generational family with CMT type 1: Clinical gene panels versus genome-wide approaches to molecular diagnosis. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 302-304.	0.5	13
3101	Case Report: Identification of an HNF1B p.Arg527Gln mutation in a Maltese patient with atypical early onset diabetes and diabetic nephropathy. <i>BMC Endocrine Disorders</i> , 2018, 18, 28.	0.9	11
3102	[18F]fluorodeoxyglucose-positron emission tomography study of genetically confirmed patients with Dravet syndrome. <i>Epilepsy Research</i> , 2018, 147, 9-14.	0.8	11
3103	Applied Computational Genomics. <i>Translational Bioinformatics</i> , 2018, , .	0.0	0
3104	Novel missense and 3' UTR splice site variants in LHFPL5 cause autosomal recessive nonsyndromic hearing impairment. <i>Journal of Human Genetics</i> , 2018, 63, 1099-1107.	1.1	3
3105	Heat in Wheat: Exploit Reverse Genetic Techniques to Discover New Alleles Within the Triticum durum sHsp26 Family. <i>Frontiers in Plant Science</i> , 2018, 9, 1337.	1.7	38
3106	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. <i>Human Mutation</i> , 2018, 39, 1875-1884.	1.1	23
3107	Somatic EP300-G211S mutations are associated with overall somatic mutational patterns and breast cancer specific survival in triple-negative breast cancer. <i>Breast Cancer Research and Treatment</i> , 2018, 172, 339-351.	1.1	11
3108	A portrait of germline mutation in Brazilian at-risk for hereditary breast cancer. <i>Breast Cancer Research and Treatment</i> , 2018, 172, 637-646.	1.1	19
3109	Genetic polymorphisms of GZMB and vitiligo: A genetic association study based on Chinese Han population. <i>Scientific Reports</i> , 2018, 8, 13001.	1.6	24
3110	Targeted capture sequencing identifies novel genetic variations in Chinese patients with idiopathic inflammatory myopathies. <i>International Journal of Rheumatic Diseases</i> , 2018, 21, 1619-1626.	0.9	6
3111	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. <i>PLoS Genetics</i> , 2018, 14, e1007504.	1.5	25

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3112	MECOM-associated syndrome: a heterogeneous inherited bone marrow failure syndrome with amegakaryocytic thrombocytopenia. <i>Blood Advances</i> , 2018, 2, 586-596.	2.5	75
3113	Computational insights of K1444N substitution in GAP-related domain of NF1 gene associated with neurofibromatosis type 1 disease: a molecular modeling and dynamics approach. <i>Metabolic Brain Disease</i> , 2018, 33, 1443-1457.	1.4	24
3114	Frequency of genetic variants associated with arrhythmogenic right ventricular cardiomyopathy in the genome aggregation database. <i>European Journal of Human Genetics</i> , 2018, 26, 1312-1318.	1.4	31
3115	ActiveDriverDB: human disease mutations and genome variation in post-translational modification sites of proteins. <i>Nucleic Acids Research</i> , 2018, 46, D901-D910.	6.5	82
3116	Genome-wide association meta-analysis highlights light-induced signaling as a driver for refractive error. <i>Nature Genetics</i> , 2018, 50, 834-848.	9.4	239
3117	Common and Rare Coding Genetic Variation Underlying the Electrocardiographic PR Interval. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002037.	1.6	19
3118	Diagnostic Yield of Genetic Testing in Young Athletes With T-Wave Inversion. <i>Circulation</i> , 2018, 138, 1184-1194.	1.6	43
3119	Analysis of the exome aggregation consortium (ExAC) database suggests that the <i>BAP1</i> tumor predisposition syndrome is underreported in cancer patients. <i>Genes Chromosomes and Cancer</i> , 2018, 57, 478-481.	1.5	6
3120	ABO genotyping with next-generation sequencing to resolve heterogeneity in donors with serology discrepancies. <i>Transfusion</i> , 2018, 58, 2232-2242.	0.8	37
3121	Distinctive regional-specific <i>PROS1</i> mutation spectrum in Southern China. <i>Journal of Thrombosis and Thrombolysis</i> , 2018, 46, 120-124.	1.0	3
3122	Skin cells for use in an alternate diagnostic method for Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , 2018, 28, 553-563.	0.3	5
3123	A mild form of haemophilia A is associated with two <i>factor VIII</i> missense mutations in German Fleckvieh cattle. <i>Animal Genetics</i> , 2018, 49, 350-351.	0.6	0
3124	Diagnostic Yield of Next-generation Sequencing in Very Early-onset Inflammatory Bowel Diseases: A Multicentre Study. <i>Journal of Crohn's and Colitis</i> , 2018, 12, 1104-1112.	0.6	68
3125	Functional Assays Are Essential for Interpretation of Missense Variants Associated with Variable Expressivity. <i>American Journal of Human Genetics</i> , 2018, 102, 1062-1077.	2.6	69
3126	A novel <i>ZC4H2</i> gene mutation, K209N, in Japanese siblings with arthrogryposis multiplex congenita and intellectual disability: characterization of the K209N mutation and clinical findings. <i>Brain and Development</i> , 2018, 40, 760-767.	0.6	14
3127	Screening for Wiskott-Aldrich syndrome by flow cytometry. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 333-335.e8.	1.5	20
3128	Autoinflammatory mutation in <i>NLRC4</i> reveals a leucine-rich repeat (LRR) LRR oligomerization interface. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 1956-1967.e6.	1.5	52
3129	Evaluating the causality of novel sequence variants in the prion protein gene by example. <i>Neurobiology of Aging</i> , 2018, 71, 265.e1-265.e7.	1.5	9

#	ARTICLE	IF	CITATIONS
3130	An initiator codon mutation in SDE2 causes recessive embryonic lethality in Holstein cattle. <i>Journal of Dairy Science</i> , 2018, 101, 6220-6231.	1.4	23
3131	De novo FBXO11 mutations are associated with intellectual disability and behavioural anomalies. <i>Human Genetics</i> , 2018, 137, 401-411.	1.8	29
3132	Mutational analysis in patients with Autosomal Dominant Polycystic Kidney Disease (ADPKD): Identification of five mutations in the PKD1 gene. <i>Gene</i> , 2018, 671, 28-35.	1.0	3
3133	Gain-of-function KCNJ6 Mutation in a Severe Hyperkinetic Movement Disorder Phenotype. <i>Neuroscience</i> , 2018, 384, 152-164.	1.1	18
3134	Identification of novel mutations causing pediatric cataract in Bhutan, Cambodia, and Sri Lanka. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 555-564.	0.6	15
3135	In silico analysis of the functional non-synonymous single nucleotide polymorphisms in the human CYP27B1 gene. <i>Egyptian Journal of Medical Human Genetics</i> , 2018, 19, 367-378.	0.5	8
3136	Genotype-phenotype correlation of hereditary erythrocytosis mutations, a single center experience. <i>American Journal of Hematology</i> , 2018, 93, 1029-1041.	2.0	38
3137	<i>SCN5A</i> (Na ^v 1.5) Variant Functional Perturbation and Clinical Presentation. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002095.	1.6	36
3138	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. <i>Neuron</i> , 2018, 99, 302-314.e4.	3.8	112
3139	Further evidence for the involvement of <i>EFL1</i> in a Shwachman-Diamond-like syndrome and expansion of the phenotypic features. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003046.	0.5	29
3140	PopViz: a webserver for visualizing minor allele frequencies and damage prediction scores of human genetic variations. <i>Bioinformatics</i> , 2018, 34, 4307-4309.	1.8	55
3141	A Novel Heterozygous Missense Mutation in <i>GNAT1</i> Leads to Autosomal Dominant Riggs Type of Congenital Stationary Night Blindness. <i>BioMed Research International</i> , 2018, 2018, 1-10.	0.9	8
3142	Analysis of CARD10 and CARD11 somatic mutations in patients with ovarian endometriosis. <i>Oncology Letters</i> , 2018, 16, 491-496.	0.8	7
3143	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2018, 103, 154-162.	2.6	56
3144	Dissecting KMT2D missense mutations in Kabuki syndrome patients. <i>Human Molecular Genetics</i> , 2018, 27, 3651-3668.	1.4	49
3145	The genetic component of preeclampsia: A whole-exome sequencing study. <i>PLoS ONE</i> , 2018, 13, e0197217.	1.1	21
3146	Validation of CZE CANCA (CZEch CAncer paNel for Clinical Application) for targeted NGS-based analysis of hereditary cancer syndromes. <i>PLoS ONE</i> , 2018, 13, e0195761.	1.1	31
3147	Mutation goals in the vitamin D receptor predicted by computational methods. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 2018, 183, 210-220.	1.2	0

#	ARTICLE	IF	CITATIONS
3148	Whole genome sequencing and mutation rate analysis of trios with paternal dioxin exposure. <i>Human Mutation</i> , 2018, 39, 1384-1392.	1.1	14
3149	Genetic Variation in the Syntaxin-Binding Protein STXBP5 in Type 1 von Willebrand Disease Patients. <i>Thrombosis and Haemostasis</i> , 2018, 118, 1382-1389.	1.8	7
3150	Innate Genetic Evolution of Lung Cancers and Spatial Heterogeneity: Analysis of Treatment-Naïve Lesions. <i>Journal of Thoracic Oncology</i> , 2018, 13, 1496-1507.	0.5	22
3151	De novo ITPR1 variants are a recurrent cause of early-onset ataxia, acting via loss of channel function. <i>European Journal of Human Genetics</i> , 2018, 26, 1623-1634.	1.4	32
3152	Accurate prediction of functional, structural, and stability changes in PITX2 mutations using in silico bioinformatics algorithms. <i>PLoS ONE</i> , 2018, 13, e0195971.	1.1	35
3153	The Utility of Next-Generation Sequencing for Primary Immunodeficiency Disorders: Experience from a Clinical Diagnostic Laboratory. <i>BioMed Research International</i> , 2018, 2018, 1-5.	0.9	37
3154	Comparative genomic investigation of high-elevation adaptation in ectothermic snakes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 8406-8411.	3.3	119
3155	A Novel Mutation of KIF11 in a Child with 22q11.2 Deletion Syndrome Associated with MCLMR. <i>Molecular Syndromology</i> , 2018, 9, 266-270.	0.3	3
3156	Systematic evaluation of a targeted gene capture sequencing panel for molecular diagnosis of retinitis pigmentosa. <i>PLoS ONE</i> , 2018, 13, e0185237.	1.1	19
3157	Molecular genetic study of Calpainopathy in Iran. <i>Gene</i> , 2018, 677, 259-265.	1.0	4
3158	C3 Glomerulopathy: Ten Years' Experience at Mayo Clinic. <i>Mayo Clinic Proceedings</i> , 2018, 93, 991-1008.	1.4	82
3159	Long QT molecular autopsy in sudden unexplained death in the young (1-40 years old): Lessons learnt from an eight year experience in New Zealand. <i>PLoS ONE</i> , 2018, 13, e0196078.	1.1	24
3160	Phenotype-driven gene prioritization for rare diseases using graph convolution on heterogeneous networks. <i>BMC Medical Genomics</i> , 2018, 11, 57.	0.7	30
3161	Detection of variants in dystroglycanopathy-associated genes through the application of targeted whole-exome sequencing analysis to a large cohort of patients with unexplained limb-girdle muscle weakness. <i>Skeletal Muscle</i> , 2018, 8, 23.	1.9	40
3162	Mutations in <i>WDR4</i> as a new cause of Galloway-Mowat syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2460-2465.	0.7	56
3163	Mutations in the gene <i>PDE6C</i> encoding the catalytic subunit of the cone photoreceptor phosphodiesterase in patients with achromatopsia. <i>Human Mutation</i> , 2018, 39, 1366-1371.	1.1	18
3164	In silico mapping of quantitative trait loci (QTL) regulating the milk ionome in mice identifies a milk iron locus on chromosome 1. <i>Mammalian Genome</i> , 2018, 29, 632-655.	1.0	5
3165	FANCM Limits Meiotic Crossovers in Brassica Crops. <i>Frontiers in Plant Science</i> , 2018, 9, 368.	1.7	41

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3166	Identification of a <sc>BRCA</sc>2 mutation in a Turkish family with early-onset breast cancer. <i>Clinical Case Reports (discontinued)</i> , 2018, 6, 1751-1755.	0.2	1
3167	Autozygosity mapping of methylmalonic acidemia associated genes by short tandem repeat markers facilitates the identification of five novel mutations in an Iranian patient cohort. <i>Metabolic Brain Disease</i> , 2018, 33, 1689-1697.	1.4	5
3168	Genetic polymorphisms of <i>CYP2S1</i>, <i>CYP2J2</i> and <i>CYP2R1</i> genes in three Chinese populations: Han, Tibetan and Uighur. <i>Pharmacogenomics</i> , 2018, 19, 961-977.	0.6	5
3169	Nuclear Factor κ B Activation in a Type V Pityriasis Rubra Pilaris Patient Harboring Multiple CARD14 Variants. <i>Frontiers in Immunology</i> , 2018, 9, 1564.	2.2	9
3170	PR interval genome-wide association meta-analysis identifies 50 loci associated with atrial and atrioventricular electrical activity. <i>Nature Communications</i> , 2018, 9, 2904.	5.8	71
3171	Genome data uncover four synergistic key regulators for extremely small body size in horses. <i>BMC Genomics</i> , 2018, 19, 492.	1.2	18
3172	Identification of 613 new loci associated with heel bone mineral density and a polygenic risk score for bone mineral density, osteoporosis and fracture. <i>PLoS ONE</i> , 2018, 13, e0200785.	1.1	137
3173	Performance evaluation of pathogenicity-computation methods for missense variants. <i>Nucleic Acids Research</i> , 2018, 46, 7793-7804.	6.5	168
3174	Mutational analysis of epidermal and hyperproliferative type I keratins in mild and moderate psoriasis vulgaris patients: a possible role in the pathogenesis of psoriasis along with disease severity. <i>Human Genomics</i> , 2018, 12, 27.	1.4	18
3175	Heritability and genome-wide association study of diffusing capacity of the lung. <i>European Respiratory Journal</i> , 2018, 52, 1800647.	3.1	18
3176	Functional confirmation that the R1488* variant in SCN9A results in complete loss-of-function of Nav1.7. <i>BMC Medical Genetics</i> , 2018, 19, 124.	2.1	6
3177	The genomic impact of historical hybridization with massive mitochondrial DNA introgression. <i>Genome Biology</i> , 2018, 19, 91.	3.8	71
3178	<i>LAMA2</i> gene mutation update: Toward a more comprehensive picture of the laminin- α 2 variome and its related phenotypes. <i>Human Mutation</i> , 2018, 39, 1314-1337.	1.1	71
3179	Novel mutation in the RNASEH1 gene in a chronic progressive external ophthalmoplegia patient. <i>Canadian Journal of Ophthalmology</i> , 2018, 53, e203-e205.	0.4	11
3180	Three novel mutations in the ATP7B gene of unrelated Vietnamese patients with Wilson disease. <i>BMC Medical Genetics</i> , 2018, 19, 104.	2.1	5
3181	Using Multi-Scale Genetic, Neuroimaging and Clinical Data for Predicting Alzheimer's Disease and Reconstruction of Relevant Biological Mechanisms. <i>Scientific Reports</i> , 2018, 8, 11173.	1.6	59
3182	Novel aggrecan variant, p. Gln2364Pro, causes severe familial nonsyndromic adult short stature and poor growth hormone response in Chinese children. <i>BMC Medical Genetics</i> , 2018, 19, 79.	2.1	14
3183	Association Between LTF Polymorphism and Risk of HIV-1 Transmission Among Zambian Seropositive Mothers. <i>Current HIV Research</i> , 2018, 16, 52-57.	0.2	1

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3184	Hb Oslo [Î²42(CD1)Pheâ†Ile; <i>HBB</i>: c.127T>A]: A Novel Unstable Hemoglobin Variant Found in a Norwegian Patient. <i>Hemoglobin</i> , 2018, 42, 78-83.	0.4	3
3185	Discovering novel SNPs that are correlated with patient outcome in a Singaporean cancer patient cohort treated with gemcitabine-based chemotherapy. <i>BMC Cancer</i> , 2018, 18, 555.	1.1	16
3186	Comparative Whole-Genomic Analysis of an Ancient L2 Lineage Mycobacterium tuberculosis Reveals a Novel Phylogenetic Clade and Common Genetic Determinants of Hypervirulent Strains. <i>Frontiers in Cellular and Infection Microbiology</i> , 2018, 7, 539.	1.8	9
3187	Integrative Bioinformatics Approaches for Identification of Drug Targets in Hypertension. <i>Frontiers in Cardiovascular Medicine</i> , 2018, 5, 25.	1.1	3
3188	Comprehensive Characterization of the RNA Editomes in Cancer Development and Progression. <i>Frontiers in Genetics</i> , 2017, 8, 230.	1.1	4
3189	Evaluating the Genetics of Common Variable Immunodeficiency: Monogenetic Model and Beyond. <i>Frontiers in Immunology</i> , 2018, 9, 636.	2.2	142
3190	Missense Gamma-Aminobutyric Acid Receptor Polymorphisms Are Associated with Reaction Time, Motor Time, and Ethanol Effects in Vivo. <i>Frontiers in Cellular Neuroscience</i> , 2018, 12, 10.	1.8	6
3191	Structure/Function Studies of the Î±4 Subunit Reveal Evolutionary Loss of a GlyR Subtype Involved in Startle and Escape Responses. <i>Frontiers in Molecular Neuroscience</i> , 2018, 11, 23.	1.4	16
3192	Exome Chip Analysis Identifies Low-Frequency and Rare Variants in <i>MRPL38</i> for White Matter Hyperintensities on Brain Magnetic Resonance Imaging. <i>Stroke</i> , 2018, 49, 1812-1819.	1.0	17
3193	Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. <i>American Journal of Human Genetics</i> , 2018, 103, 100-114.	2.6	34
3194	Utilizing ExAC to assess the hidden contribution of variants of unknown significance to Sanfilippo Type B incidence. <i>PLoS ONE</i> , 2018, 13, e0200008.	1.1	10
3195	Molecular Modeling of Drugâ€“Transporter Interactionsâ€”An International Transporter Consortium Perspective. <i>Clinical Pharmacology and Therapeutics</i> , 2018, 104, 818-835.	2.3	43
3196	Rothmund-Thomson Syndrome: Insights from New Patients on the Genetic Variability Underpinning Clinical Presentation and Cancer Outcome. <i>International Journal of Molecular Sciences</i> , 2018, 19, 1103.	1.8	20
3197	What Does This Mutation Mean? The Tools and Pitfalls of Variant Interpretation in Lymphoid Malignancies. <i>International Journal of Molecular Sciences</i> , 2018, 19, 1251.	1.8	11
3198	Effects of genetic variants in the TSPO gene on protein structure and stability. <i>PLoS ONE</i> , 2018, 13, e0195627.	1.1	19
3199	A New Integrated Newborn Screening Workflow Can Provide a Shortcut to Differential Diagnosis and Confirmation of Inherited Metabolic Diseases. <i>Yonsei Medical Journal</i> , 2018, 59, 652.	0.9	9
3200	Computational analysis of deleterious SNPs of SLC45A2 involved in oculocutaneous albinism type 4. <i>Gene Reports</i> , 2018, 12, 248-254.	0.4	0
3201	Basonuclin 1 deficiency is a cause of primary ovarian insufficiency. <i>Human Molecular Genetics</i> , 2018, 27, 3787-3800.	1.4	38

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3202	Comprehensive genomic diagnosis of non-syndromic and syndromic hereditary hearing loss in Spanish patients. <i>BMC Medical Genomics</i> , 2018, 11, 58.	0.7	65
3203	Prioritization and functional assessment of noncoding variants associated with complex diseases. <i>Genome Medicine</i> , 2018, 10, 53.	3.6	33
3204	Copy number variation analysis and targeted NGS in 77 families with suspected Lynch syndrome reveals novel potential causative genes. <i>International Journal of Cancer</i> , 2018, 143, 2800-2813.	2.3	11
3205	A Functional Mutation in HDAC8 Gene as Novel Diagnostic Marker for Cornelia De Lange Syndrome. <i>Cellular Physiology and Biochemistry</i> , 2018, 47, 2388-2395.	1.1	11
3206	Report of second case and clinical and molecular characterization of Eiken syndrome. <i>Clinical Genetics</i> , 2018, 94, 457-460.	1.0	13
3207	Molecular genetics of tetrahydrobiopterin deficiency in Chinese patients. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 911-916.	0.4	18
3208	Analysis of causal effect of <i>APOA5</i> variants on premature coronary artery disease. <i>Annals of Human Genetics</i> , 2018, 82, 437-447.	0.3	8
3209	Spectrum of genomic variations in Indian patients with progressive familial intrahepatic cholestasis. <i>BMC Gastroenterology</i> , 2018, 18, 107.	0.8	9
3210	Distal renal tubular acidosis caused by <i>tryptophan</i> aspartate repeat domain 72 (<i>WDR72</i>) mutations. <i>Clinical Genetics</i> , 2018, 94, 409-418.	1.0	54
3211	Cardiomyopathy and Preeclampsia. <i>Circulation</i> , 2018, 138, 2359-2366.	1.6	60
3212	Identification of genetic variants for clinical management of familial colorectal tumors. <i>BMC Medical Genetics</i> , 2018, 19, 26.	2.1	18
3213	Targeted sequencing reveals complex, phenotype-correlated genotypes in cystic fibrosis. <i>BMC Medical Genomics</i> , 2018, 11, 13.	0.7	24
3214	Performance of in silico prediction tools for the classification of rare BRCA1/2 missense variants in clinical diagnostics. <i>BMC Medical Genomics</i> , 2018, 11, 35.	0.7	78
3216	Determination of the mutational landscape in Taiwanese patients with papillary thyroid cancer by whole-exome sequencing. <i>Human Pathology</i> , 2018, 78, 151-158.	1.1	9
3217	Functional Invalidation of Putative Sudden Infant Death Syndrome-Associated Variants in the KCNH2-Encoded Kv11.1 Channel. <i>Circulation: Arrhythmia and Electrophysiology</i> , 2018, 11, e005859.	2.1	6
3218	A multiple myeloma-specific capture sequencing platform discovers novel translocations and frequent, risk-associated point mutations in IGLL5. <i>Blood Cancer Journal</i> , 2018, 8, 35.	2.8	41
3219	Identification of somatic mutations in monozygotic twins discordant for psychiatric disorders. <i>NPJ Schizophrenia</i> , 2018, 4, 7.	2.0	16
3220	Germline breast cancer susceptibility gene mutations and breast cancer outcomes. <i>BMC Cancer</i> , 2018, 18, 315.	1.1	61

#	ARTICLE	IF	CITATIONS
3221	Deleterious genetic variants in ciliopathy genes increase risk of ritodrine-induced cardiac and pulmonary side effects. <i>BMC Medical Genomics</i> , 2018, 11, 4.	0.7	16
3222	Spectrum of low-density lipoprotein receptor (LDLR) mutations in a cohort of Sri Lankan patients with familial hypercholesterolemia – a preliminary report. <i>Lipids in Health and Disease</i> , 2018, 17, 100.	1.2	9
3223	Detection rate of causal variants in severe childhood epilepsy is highest in patients with seizure onset within the first four weeks of life. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 71.	1.2	18
3224	Prevalence of <i>Plasmodium falciparum</i> delayed clearance associated polymorphisms in adaptor protein complex 2 mu subunit (pfap2mu) and ubiquitin specific protease 1 (pfubp1) genes in Ghanaian isolates. <i>Parasites and Vectors</i> , 2018, 11, 175.	1.0	27
3225	The VAAST Variant Prioritizer (VVP): ultrafast, easy to use whole genome variant prioritization tool. <i>BMC Bioinformatics</i> , 2018, 19, 57.	1.2	29
3226	Genetic analysis of Wnt/PCP genes in neural tube defects. <i>BMC Medical Genomics</i> , 2018, 11, 38.	0.7	43
3227	Identification of novel candidate genes for 46,XY disorders of sex development (DSD) using a C57BL/6J-Y POS mouse model. <i>Biology of Sex Differences</i> , 2018, 9, 8.	1.8	14
3228	Harnessing Omics Big Data in Nine Vertebrate Species by Genome-Wide Prioritization of Sequence Variants with the Highest Predicted Deleterious Effect on Protein Function. <i>OMICS A Journal of Integrative Biology</i> , 2018, 22, 410-421.	1.0	7
3229	Targeted exome sequencing reveals homozygous TREM2 R47C mutation presenting with behavioral variant frontotemporal dementia without bone involvement. <i>Neurobiology of Aging</i> , 2018, 68, 160.e15-160.e19.	1.5	14
3230	AARS2-related ovarioleukodystrophy: Clinical and neuroimaging features of three new cases. <i>Acta Neurologica Scandinavica</i> , 2018, 138, 278-283.	1.0	22
3231	Mutation analysis of SLC26A4 (Pendrin) gene in a Brazilian sample of hearing-impaired subjects. <i>BMC Medical Genetics</i> , 2018, 19, 73.	2.1	12
3232	Next generation sequencing panel based on single molecule molecular inversion probes for detecting genetic variants in children with hypopituitarism. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 514-525.	0.6	17
3233	Detection of Molecular Alterations in Taiwanese Patients with Medullary Thyroid Cancer Using Whole-Exome Sequencing. <i>Endocrine Pathology</i> , 2018, 29, 324-331.	5.2	23
3234	A novel ZIC3 gene mutation identified in patients with heterotaxy and congenital heart disease. <i>Scientific Reports</i> , 2018, 8, 12386.	1.6	9
3235	Analytical –bake-off– of whole genome sequencing quality for the Genome Russia project using a small cohort for autoimmune hepatitis. <i>PLoS ONE</i> , 2018, 13, e0200423.	1.1	7
3236	Spectrum of <i>MNX1</i> Pathogenic Variants and Associated Clinical Features in Korean Patients with Currarino Syndrome. <i>Annals of Laboratory Medicine</i> , 2018, 38, 242-248.	1.2	7
3237	A Review of Tools to Automatically Infer Chromosomal Positions From dbSNP and HGVS Genetic Variants. , 2018, , 133-156.		1
3238	The Role of Genetic Variation in CYP2R1, the Principal Vitamin D 25-Hydroxylase, in Vitamin D Homeostasis. , 2018, , 303-315.		2

#	ARTICLE	IF	CITATIONS
3239	Characterization of three ciliopathy pedigrees expands the phenotype associated with biallelic C2CD3 variants. <i>European Journal of Human Genetics</i> , 2018, 26, 1797-1809.	1.4	19
3240	Case report of familial sudden cardiac death caused by a DSG2 p.F531C mutation as genetic background when carrying with heterozygous KCNE5 p.D92E/E93X mutation. <i>BMC Medical Genetics</i> , 2018, 19, 148.	2.1	9
3241	Inherited CARD9 Deficiency: Invasive Disease Caused by Ascomycete Fungi in Previously Healthy Children and Adults. <i>Journal of Clinical Immunology</i> , 2018, 38, 656-693.	2.0	130
3242	Whole-exome sequencing identifies mutations in FSIP2 as a recurrent cause of multiple morphological abnormalities of the sperm flagella. <i>Human Reproduction</i> , 2018, 33, 1973-1984.	0.4	93
3243	Bridging Genomics to Phenomics at Atomic Resolution through Variation Spatial Profiling. <i>Cell Reports</i> , 2018, 24, 2013-2028.e6.	2.9	33
3244	IDGenetics: a comprehensive database for genes and mutations of intellectual disability related disorders. <i>Neuroscience Letters</i> , 2018, 685, 96-101.	1.0	10
3245	Cenani-Lenz syndactyly syndrome - a case report of a family with isolated syndactyly. <i>BMC Medical Genetics</i> , 2018, 19, 125.	2.1	9
3246	Detection of Familial Hypercholesterolemia Using Next Generation Sequencing in Two Population-Based Cohorts. <i>Chonnam Medical Journal</i> , 2018, 54, 31.	0.5	10
3247	The characteristics of digenic familial exudative vitreoretinopathy. <i>Graefes' Archive for Clinical and Experimental Ophthalmology</i> , 2018, 256, 2149-2156.	1.0	18
3248	Multiethnic meta-analysis identifies ancestry-specific and cross-ancestry loci for pulmonary function. <i>Nature Communications</i> , 2018, 9, 2976.	5.8	85
3249	A putative human infertility allele of the meiotic recombinase DMC1 does not affect fertility in mice. <i>Human Molecular Genetics</i> , 2018, 27, 3911-3918.	1.4	14
3250	A Novel p.G141R Mutation in <i>ILDR1</i> Leads to Recessive Nonsyndromic Deafness DFNB42 in Two Chinese Han Families. <i>Neural Plasticity</i> , 2018, 2018, 1-6.	1.0	6
3251	Targeted next-generation sequencing identifies two novel COL2A1 gene mutations in Stickler syndrome with bilateral retinal detachment. <i>International Journal of Molecular Medicine</i> , 2018, 42, 1819-1826.	1.8	3
3252	Breast cancer patients suggestive of Li-Fraumeni syndrome: mutational spectrum, candidate genes, and unexplained heredity. <i>Breast Cancer Research</i> , 2018, 20, 87.	2.2	9
3253	Multiple alleles of <i>ACAN</i> associated with chondrodysplastic dwarfism in Miniature horses. <i>Animal Genetics</i> , 2018, 49, 413-420.	0.6	10
3254	NDDVD: an integrated and manually curated Neurodegenerative Diseases Variation Database. <i>Database: the Journal of Biological Databases and Curation</i> , 2018, 2018, .	1.4	5
3255	Expanding the spectrum of PEX16 mutations and novel insights into disease mechanisms. <i>Molecular Genetics and Metabolism Reports</i> , 2018, 16, 46-51.	0.4	21
3256	Familial Cancer Variant Prioritization Pipeline version 2 (FCVPPv2) applied to a papillary thyroid cancer family. <i>Scientific Reports</i> , 2018, 8, 11635.	1.6	30

#	ARTICLE	IF	CITATIONS
3257	Application of High-Throughput Sequencing in the Diagnosis of Inherited Thrombocytopenia. <i>Clinical and Applied Thrombosis/Hemostasis</i> , 2018, 24, 94S-103S.	0.7	7
3258	Genotype-phenotype correlation among Malaysian patients with osteogenesis imperfecta. <i>Clinica Chimica Acta</i> , 2018, 484, 141-147.	0.5	10
3259	±-Adducin nsSNPs affect mRNA secondary structure, protein modification and stability. <i>Meta Gene</i> , 2018, 17, 153-162.	0.3	7
3260	Tailoring the American College of Medical Genetics and Genomics and the Association for Molecular Pathology Guidelines for the Interpretation of Sequenced Variants in the <i>FBN1</i> Gene for Marfan Syndrome. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002039.	1.6	20
3261	Demographic expansion and genetic load of the halophyte model plant <i>Eutrema salsugineum</i> . <i>Molecular Ecology</i> , 2018, 27, 2943-2955.	2.0	11
3262	Establishing the role of PLVAP in protein-losing enteropathy: a homozygous missense variant leads to an attenuated phenotype. <i>Journal of Medical Genetics</i> , 2018, 55, 779-784.	1.5	14
3263	Clinical heterogeneity and molecular profile of triple A syndrome: a study of seven cases. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 799-807.	0.4	13
3264	Germline variation in the oxidative DNA repair genes NUDT1 and OGG1 is not associated with hereditary colorectal cancer or polyposis. <i>Human Mutation</i> , 2018, 39, 1214-1225.	1.1	10
3265	Phosphorylation of the RSRSP stretch is critical for splicing regulation by RNA-Binding Motif Protein 20 (RBM20) through nuclear localization. <i>Scientific Reports</i> , 2018, 8, 8970.	1.6	58
3266	Atrial involvement in arrhythmogenic right ventricular cardiomyopathy patients referred for ventricular arrhythmias ablation. <i>Journal of Cardiovascular Electrophysiology</i> , 2018, 29, 1388-1395.	0.8	7
3267	Identification and in silico analysis of functional SNPs of human TAGAP protein: A comprehensive study. <i>PLoS ONE</i> , 2018, 13, e0188143.	1.1	71
3268	Estimating the mutational load for cardiovascular diseases in Pakistani population. <i>PLoS ONE</i> , 2018, 13, e0192446.	1.1	15
3269	Yeast-based assays for characterization of the functional effects of single nucleotide polymorphisms in human DNA repair genes. <i>PLoS ONE</i> , 2018, 13, e0193823.	1.1	6
3270	Rare Variants in PLD3 Increase Risk for Alzheimer's Disease in Han Chinese. <i>Journal of Alzheimer's Disease</i> , 2018, 64, 55-59.	1.2	17
3271	<i>MACROD2</i> Haploinsufficiency Impairs Catalytic Activity of PARP1 and Promotes Chromosome Instability and Growth of Intestinal Tumors. <i>Cancer Discovery</i> , 2018, 8, 988-1005.	7.7	55
3272	Comprehensive analysis of CTNNB1 in adrenocortical carcinomas: Identification of novel mutations and correlation to survival. <i>Scientific Reports</i> , 2018, 8, 8610.	1.6	22
3273	Novel Mutation in Anoctamin 5 Gene Causing Limb-Girdle Muscular Dystrophy 2L. <i>Journal of Clinical Neuromuscular Disease</i> , 2018, 19, 228-231.	0.3	2
3274	Multi-ethnic genome-wide association study for atrial fibrillation. <i>Nature Genetics</i> , 2018, 50, 1225-1233.	9.4	552

#	ARTICLE	IF	CITATIONS
3275	Role of Targeted Next Generation Sequencing in the Etiological Work-Up of Congenitally Deaf Children. <i>Otology and Neurotology</i> , 2018, 39, 732-738.	0.7	10
3277	Expanding the clinical and molecular findings in RASA1 capillary malformation-arteriovenous malformation. <i>European Journal of Human Genetics</i> , 2018, 26, 1521-1536.	1.4	42
3278	Characterization of a novel MYO3A missense mutation associated with a dominant form of late onset hearing loss. <i>Scientific Reports</i> , 2018, 8, 8706.	1.6	22
3279	IBD risk loci are enriched in multigenic regulatory modules encompassing putative causative genes. <i>Nature Communications</i> , 2018, 9, 2427.	5.8	159
3280	Ligand-induced perturbation of the HIF-2 α :ARNT dimer dynamics. <i>PLoS Computational Biology</i> , 2018, 14, e1006021.	1.5	22
3281	A Novel SCN9A Mutation (F826Y) in Primary Erythromelalgia Alters the Excitability of Nav1.7. <i>Current Molecular Medicine</i> , 2018, 17, 450-457.	0.6	7
3282	Metachronous Medulloblastoma in a Child With Successfully Treated Neuroblastoma: Case Report and Novel Findings of DNA Sequencing. <i>Journal of the National Comprehensive Cancer Network: JNCCN</i> , 2018, 16, 683-691.	2.3	2
3283	Genetic background of Japanese patients with pediatric hypertrophic and restrictive cardiomyopathy. <i>Journal of Human Genetics</i> , 2018, 63, 989-996.	1.1	26
3284	Primary Myocardial Fibrosis as an Alternative Phenotype Pathway of Inherited Cardiac Structural Disorders. <i>Circulation</i> , 2018, 137, 2716-2726.	1.6	41
3285	Cancer driver mutation prediction through Bayesian integration of multi-omic data. <i>PLoS ONE</i> , 2018, 13, e0196939.	1.1	23
3286	A recurrent de novo missense pathogenic variant in SMARCB1 causes severe intellectual disability and choroid plexus hyperplasia with resultant hydrocephalus. <i>Genetics in Medicine</i> , 2019, 21, 572-579.	1.1	24
3287	Mouse models as a tool for discovering new neurological diseases. <i>Neurobiology of Learning and Memory</i> , 2019, 165, 106902.	1.0	17
3288	A novel EXT2 mutation in a consanguineous family with severe developmental delay, microcephaly, seizures, feeding difficulties, and osteopenia extends the phenotypic spectrum of autosomal recessive EXT2-related syndrome (AREXT2). <i>European Journal of Medical Genetics</i> , 2019, 62, 259-264.	0.7	11
3289	Variation within voltage-gated calcium channel genes and antipsychotic treatment response in a South African first episode schizophrenia cohort. <i>Pharmacogenomics Journal</i> , 2019, 19, 109-114.	0.9	7
3290	The Role of Next-Generation Sequencing in Pharmacogenetics and Pharmacogenomics. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2019, 9, a033027.	2.9	49
3291	Implementing precision cancer medicine in the genomic era. <i>Seminars in Cancer Biology</i> , 2019, 55, 16-27.	4.3	24
3292	Circulating tumor DNA analysis enables molecular characterization of pediatric renal tumors at diagnosis. <i>International Journal of Cancer</i> , 2019, 144, 68-79.	2.3	37
3293	Clinical and genetic spectrum of a large cohort of children with epilepsy in China. <i>Genetics in Medicine</i> , 2019, 21, 564-571.	1.1	93

#	ARTICLE	IF	CITATIONS
3294	Predicting Non-Synonymous Single Nucleotide Variants Pathogenic Effects in Human Diseases. , 2019, , 400-409.		1
3295	Genome Annotation. , 2019, , 195-209.		3
3296	Deleterious alleles in the context of domestication, inbreeding, and selection. Evolutionary Applications, 2019, 12, 6-17.	1.5	94
3297	Candidate susceptibility variants in angioimmunoblastic T-cell lymphoma. Familial Cancer, 2019, 18, 113-119.	0.9	8
3298	Panel sequencing distinguishes monogenic forms of nephritis from nephrosis in children. Nephrology Dialysis Transplantation, 2019, 34, 474-485.	0.4	13
3299	Role of TGF- β 1 and C-Kit Mutations in the Development of Hepatocellular Carcinoma in Hepatitis C Virus-Infected Patients: in vitro Study. Biochemistry (Moscow), 2019, 84, 941-953.	0.7	8
3300	Identification of genetic factors underlying persistent pulmonary hypertension of newborns in a cohort of Chinese neonates. Respiratory Research, 2019, 20, 174.	1.4	21
3301	Individual factors influencing contractors' risk attitudes among Malaysian construction industries: the moderating role of government policy. International Journal of Construction Management, 2022, 22, 612-631.	2.2	21
3302	Gene Expression Profiling Analysis Reveals Putative Phytochemotherapeutic Target for Castration-Resistant Prostate Cancer. Frontiers in Oncology, 2019, 9, 714.	1.3	5
3303	Novel mutations and the ophthalmologic characters in Chinese patients with Wolfram Syndrome. Orphanet Journal of Rare Diseases, 2019, 14, 190.	1.2	9
3304	Family-Based Quantitative Trait Meta-Analysis Implicates Rare Noncoding Variants in DENND1A in Polycystic Ovary Syndrome. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3835-3850.	1.8	51
3305	Familial Intracranial Aneurysm in Newfoundland: Clinical and Genetic Analysis. Canadian Journal of Neurological Sciences, 2019, 46, 518-526.	0.3	2
3306	Targeted genomic profiling identifies frequent deleterious mutations in FAT4 and TP53 genes in HBV-associated hepatocellular carcinoma. BMC Cancer, 2019, 19, 789.	1.1	12
3307	Inherited glycoposphatidylinositol deficiency variant database and analysis of pathogenic variants. Molecular Genetics & Genomic Medicine, 2019, 7, e00743.	0.6	11
3308	Phenotypic severity in a family with MEND syndrome is directly associated with the accumulation of potentially functional variants of cholesterol homeostasis genes. Molecular Genetics & Genomic Medicine, 2019, 7, e931.	0.6	2
3309	<i>AMB1</i> mutations causing hypoplastic amelogenesis imperfecta and <i>Amb1</i> knockout knockin mice exhibiting failed amelogenesis and <i>Amb1</i> tissue-specificity. Molecular Genetics & Genomic Medicine, 2019, 7, e929.	0.6	23
3310	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	2.6	46
3311	In search of a genetic explanation for LDLc variability in an FH family: common SNPs and a rare mutation in MTP explain only part of LDL variability in an FH family. Journal of Lipid Research, 2019, 60, 1733-1740.	2.0	2

#	ARTICLE	IF	CITATIONS
3312	Whole genome sequencing and rare variant analysis in essential tremor families. PLoS ONE, 2019, 14, e0220512.	1.1	28
3313	Identification of genetic associations of ECHS1 gene with milk fatty acid traits in dairy cattle. Animal Genetics, 2019, 50, 430-438.	0.6	5
3314	Functional analysis of <i>GALT</i> variants found in classic galactosemia patients using a novel cell-free translation method. JIMD Reports, 2019, 48, 60-66.	0.7	2
3315	Challenges in funding and developing genomic software: roots and remedies. Genome Biology, 2019, 20, 147.	3.8	21
3316	Novel Androgen Receptor Gene Variant Containing a Premature Termination Codon in a Patient with Androgen Insensitivity Syndrome. Journal of Pediatric and Adolescent Gynecology, 2019, 32, 641-644.	0.3	1
3317	Genetics and Genomics of <i>Linum</i> . Plant Genetics and Genomics: Crops and Models, 2019, , .	0.3	14
3318	ATP1A1 mutations cause intermediate Charcot-Marie-Tooth disease. Human Mutation, 2019, 40, 2334-2343.	1.1	11
3319	Contributions of Rare Gene Variants to Familial and Sporadic FSGS. Journal of the American Society of Nephrology: JASN, 2019, 30, 1625-1640.	3.0	42
3320	Mutation Spectrum of Cancer-Associated Genes in Patients With Early Onset of Colorectal Cancer. Frontiers in Oncology, 2019, 9, 673.	1.3	36
3321	Clinical and Genetic Findings in Children with Neurofibromatosis Type 1, Legius Syndrome, and Other Related Neurocutaneous Disorders. Genes, 2019, 10, 580.	1.0	25
3322	Development, Implementation and Assessment of Molecular Diagnostics by Next Generation Sequencing in Personalized Treatment of Cancer: Experience of a Public Reference Healthcare Hospital. Cancers, 2019, 11, 1196.	1.7	13
3323	Effect of BTB gene variants on in vitro biotinidase activity. Molecular Genetics and Metabolism, 2019, 127, 361-367.	0.5	6
3324	Human DEF6 deficiency underlies an immunodeficiency syndrome with systemic autoimmunity and aberrant CTLA-4 homeostasis. Nature Communications, 2019, 10, 3106.	5.8	48
3325	Mutational analysis of BRCA1 and BRCA2 genes in women with familial breast cancer from different regions of Colombia. Hereditary Cancer in Clinical Practice, 2019, 17, 20.	0.6	6
3326	<p>Mechanisms of resistance to a PI3K inhibitor in gastrointestinal stromal tumors: an omic approach to identify novel druggable targets<p>. Cancer Management and Research, 2019, Volume 11, 6229-6244.	0.9	2
3327	Clinical utility of a targeted next generation sequencing panel in severe and pediatric onset Mendelian diseases. European Journal of Medical Genetics, 2019, 62, 103725.	0.7	5
3328	Mutations in <i>RHOT1</i> Disrupt Endoplasmic Reticulum-Mitochondria Contact Sites Interfering with Calcium Homeostasis and Mitochondrial Dynamics in Parkinson's Disease. Antioxidants and Redox Signaling, 2019, 31, 1213-1234.	2.5	56
3329	Contribution of New Adenomatous Polyposis Predisposition Genes in an Unexplained Attenuated Spanish Cohort by Multigene Panel Testing. Scientific Reports, 2019, 9, 9814.	1.6	9

#	ARTICLE	IF	CITATIONS
3330	Co-segregation of candidate polymorphism rs201204878 of the PKD1 gene in a large Iranian family with autosomal dominant polycystic disease. <i>Experimental and Therapeutic Medicine</i> , 2019, 18, 1345-1349.	0.8	0
3331	Dissecting in silico Mutation Prediction of Variants in African Genomes: Challenges and Perspectives. <i>Frontiers in Genetics</i> , 2019, 10, 601.	1.1	25
3332	Assessing computational predictions of the phenotypic effect of cystathionine- β -synthase variants. <i>Human Mutation</i> , 2019, 40, 1530-1545.	1.1	5
3333	Computational assessment of somatic and germline mutations of p16INK4a: Structural insights and implications in disease. <i>Informatics in Medicine Unlocked</i> , 2019, 17, 100208.	1.9	2
3334	Development of a genomics module within an epilepsy-specific electronic health record: Toward genomic medicine in epilepsy care. <i>Epilepsia</i> , 2019, 60, 1670-1677.	2.6	7
3335	Prediction of disease-associated mutations in the transmembrane regions of proteins with known 3D structure. <i>PLoS ONE</i> , 2019, 14, e0219452.	1.1	19
3336	Building a Hybrid Physical-Statistical Classifier for Predicting the Effect of Variants Related to Protein-Drug Interactions. <i>Structure</i> , 2019, 27, 1469-1481.e3.	1.6	6
3337	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. <i>European Journal of Human Genetics</i> , 2019, 27, 1611-1618.	1.4	45
3338	VIPdb, a genetic Variant Impact Predictor Database. <i>Human Mutation</i> , 2019, 40, 1202-1214.	1.1	24
3339	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. <i>Brain</i> , 2019, 142, 2617-2630.	3.7	31
3340	Novel SCA19/22-associated <i>KCND3</i> mutations disrupt human K _V 4.3 protein biosynthesis and channel gating. <i>Human Mutation</i> , 2019, 40, 2088-2107.	1.1	15
3341	Locating potentially lethal genes using the abnormal distributions of genotypes. <i>Scientific Reports</i> , 2019, 9, 10543.	1.6	0
3342	Genetic spectrum of renal disease for 1001 Chinese children based on a multicenter registration system. <i>Clinical Genetics</i> , 2019, 96, 402-410.	1.0	52
3343	A Deep Belief Networks Based Prediction Method for Identification of Disease-Associated Non-coding SNPs in Human Genome. <i>Lecture Notes in Computer Science</i> , 2019, , 12-24.	1.0	0
3344	A brain somatic RHEB doublet mutation causes focal cortical dysplasia type II. <i>Experimental and Molecular Medicine</i> , 2019, 51, 1-11.	3.2	46
3345	COL1A1/2 Pathogenic Variants and Phenotype Characteristics in Ukrainian Osteogenesis Imperfecta Patients. <i>Frontiers in Genetics</i> , 2019, 10, 722.	1.1	29
3346	Assessment of predicted enzymatic activity of <i>HN</i> -acetylglucosaminidase variants of unknown significance for CAGI 2016. <i>Human Mutation</i> , 2019, 40, 1519-1529.	1.1	10
3347	TAF1, associated with intellectual disability in humans, is essential for embryogenesis and regulates neurodevelopmental processes in zebrafish. <i>Scientific Reports</i> , 2019, 9, 10730.	1.6	28

#	ARTICLE	IF	CITATIONS
3348	Next-generation sequencing using microfluidic PCR enrichment for molecular autopsy. <i>BMC Cardiovascular Disorders</i> , 2019, 19, 174.	0.7	7
3349	Host Genetics, Innate Immune Responses, and Cellular Death Pathways in Poliomyelitis Patients. <i>Frontiers in Microbiology</i> , 2019, 10, 1495.	1.5	7
3350	Exome sequencing of Finnish isolates enhances rare-variant association power. <i>Nature</i> , 2019, 572, 323-328.	13.7	161
3351	Identification of most damaging nsSNPs in human <i>CCR6</i> gene: In silico analyses. <i>International Journal of Immunogenetics</i> , 2019, 46, 459-471.	0.8	21
3352	The <i>dmc1</i> Mutant Allows an Insight Into the DNA Double-Strand Break Repair During Meiosis in Barley (<i>Hordeum vulgare</i> L.). <i>Frontiers in Plant Science</i> , 2019, 10, 761.	1.7	17
3353	Molecular characterization of known and novel <i>ACVR1</i> variants in phenotypes of aberrant ossification. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1764-1777.	0.7	13
3354	A novel de novo mutation (p.Pro1310Glnfs*46) in <i>KMT2A</i> caused Wiedemann-Steiner Syndrome in a Chinese boy with postnatal growth retardation: a case report. <i>Molecular Biology Reports</i> , 2019, 46, 5555-5559.	1.0	8
3355	Diagnostic Yield and Treatment Impact of Targeted Exome Sequencing in Early-Onset Epilepsy. <i>Frontiers in Neurology</i> , 2019, 10, 434.	1.1	70
3356	Differences in Mutational Profile between Follicular Thyroid Carcinoma and Follicular Thyroid Adenoma Identified Using Next Generation Sequencing. <i>International Journal of Molecular Sciences</i> , 2019, 20, 3126.	1.8	25
3357	In silico analysis of missense mutations in exons 1-5 of the <i>F9</i> gene that cause hemophilia B. <i>BMC Bioinformatics</i> , 2019, 20, 363.	1.2	9
3358	In Silico Insights into HIV-1 Vpu-Tetherin Interactions and Its Mutational Counterparts. <i>Medical Sciences (Basel, Switzerland)</i> , 2019, 7, 74.	1.3	0
3359	Endangered island endemic plants have vulnerable genomes. <i>Communications Biology</i> , 2019, 2, 244.	2.0	28
3360	Sequence-based GWAS, network and pathway analyses reveal genes co-associated with milk cheese-making properties and milk composition in Montbéliarde cows. <i>Genetics Selection Evolution</i> , 2019, 51, 34.	1.2	46
3361	Exome and Genome Sequencing. , 2019, , 137-148.		0
3362	PhyreRisk: A Dynamic Web Application to Bridge Genomics, Proteomics and 3D Structural Data to Guide Interpretation of Human Genetic Variants. <i>Journal of Molecular Biology</i> , 2019, 431, 2460-2466.	2.0	21
3363	Leigh syndrome caused by mitochondrial DNA-maintenance defects revealed by whole exome sequencing. <i>Mitochondrion</i> , 2019, 49, 25-34.	1.6	3
3364	Rare Protein-Altering Telomere-related Gene Variants in Patients with Chronic Hypersensitivity Pneumonitis. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2019, 200, 1154-1163.	2.5	81
3365	Effects of common genetic variants associated with colorectal cancer risk on survival outcomes after diagnosis: A large population-based cohort study. <i>International Journal of Cancer</i> , 2019, 145, 2427-2432.	2.3	11

#	ARTICLE	IF	CITATIONS
3366	Genetic diagnosis in first or second trimester pregnancy loss using exome sequencing: a systematic review of human essential genes. <i>Journal of Assisted Reproduction and Genetics</i> , 2019, 36, 1539-1548.	1.2	25
3367	Whole-Exome Sequencing Identified a De Novo Mutation of <i>Junction Plakoglobin</i> (p.R577C) in a Chinese Patient with Arrhythmogenic Right Ventricular Cardiomyopathy. <i>BioMed Research International</i> , 2019, 2019, 1-6.	0.9	8
3368	Coexistence of Congenital Adrenal Hyperplasia and Autoimmune Addison's Disease. <i>Frontiers in Endocrinology</i> , 2019, 10, 648.	1.5	2
3369	Clinical exome sequencing identifies novel CREBBP variants in 18 Chinese Rubinsteinâ€Taybi Syndrome kids with high frequency of polydactyly. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e1009.	0.6	9
3370	Functional analysis of six uncharacterised mutations in LDLR gene. <i>Atherosclerosis</i> , 2019, 291, 44-51.	0.4	5
3371	Essentials of Bioinformatics, Volume II. , 2019, , .		1
3372	Large-Scale Whole-Genome Sequencing of Three Diverse Asian Populations in Singapore. <i>Cell</i> , 2019, 179, 736-749.e15.	13.5	126
3373	Pathogenicity discrimination and genetic test reference for CRX variants based on genotype-phenotype analysis. <i>Experimental Eye Research</i> , 2019, 189, 107846.	1.2	18
3374	IL-12R β 1 deficiency corresponding to concurrency of two diseases, mendelian susceptibility to mycobacterial disease and Crohn's disease. <i>Journal of Clinical Tuberculosis and Other Mycobacterial Diseases</i> , 2019, 17, 100123.	0.6	3
3375	Bi-allelic Variants in IQSEC1 Cause Intellectual Disability, Developmental Delay, and Short Stature. <i>American Journal of Human Genetics</i> , 2019, 105, 907-920.	2.6	22
3376	Contribution of SLC22A12 on hypouricemia and its clinical significance for screening purposes. <i>Scientific Reports</i> , 2019, 9, 14360.	1.6	13
3377	A novel <i>LAMP2</i> p.G93R mutation associated with mild Danon disease presenting with familial hypertrophic cardiomyopathy. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00941.	0.6	9
3378	De novo pathogenic <i>DNM1L</i> variant in a patient diagnosed with atypical hereditary sensory and autonomic neuropathy. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00961.	0.6	12
3379	A de novo SCN8A heterozygous mutation in a child with epileptic encephalopathy: a case report. <i>BMC Pediatrics</i> , 2019, 19, 400.	0.7	3
3380	The impact of missense mutation in PIGA associated to paroxysmal nocturnal hemoglobinuria and multiple congenital anomalies-hypotonia-seizures syndrome 2: A computational study. <i>Heliyon</i> , 2019, 5, e02709.	1.4	8
3381	The design of remote-control system for photographic equipment. <i>IOP Conference Series: Materials Science and Engineering</i> , 2019, 569, 042017.	0.3	0
3382	Mammals and <i>Dictyostelium</i> rictor mutations swapping reveals two essential Gly residues for mTORC2 activity and integrity. <i>Journal of Cell Science</i> , 2019, 132, .	1.2	3
3383	Linkage and exome analysis implicate multiple genes in non-syndromic intellectual disability in a large Swedish family. <i>BMC Medical Genomics</i> , 2019, 12, 156.	0.7	3

#	ARTICLE	IF	CITATIONS
3384	Bi-allelic Mutations in TTC29 Cause Male Subfertility with Asthenoteratospermia in Humans and Mice. <i>American Journal of Human Genetics</i> , 2019, 105, 1168-1181.	2.6	62
3385	Detection of a Frameshift Deletion in the SPTBN4 Gene Leads to Prevention of Severe Myopathy and Postnatal Mortality in Pigs. <i>Frontiers in Genetics</i> , 2019, 10, 1226.	1.1	6
3386	Genetic variability in Iranian limb-girdle muscular dystrophy type 2B patients: An evidence of a founder effect. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e1029.	0.6	2
3387	Ensembl 2020. <i>Nucleic Acids Research</i> , 2020, 48, D682-D688.	6.5	1,076
3388	Gpr63 is a modifier of microcephaly in Ttc21b mouse mutants. <i>PLoS Genetics</i> , 2019, 15, e1008467.	1.5	7
3389	Novel KCNJ10 Compound Heterozygous Mutations Causing EAST/SeSAME-Like Syndrome Compromise Potassium Channel Function. <i>Frontiers in Genetics</i> , 2019, 10, 912.	1.1	9
3390	Fluid-solid interaction simulation for particles and walls of arbitrary polygonal shapes with a coupled LBM-IMB-DEM method. <i>Powder Technology</i> , 2019, 356, 177-192.	2.1	14
3391	A novel <i>TYRP1</i> variant is associated with liver and tan coat colour in Lancashire Heelers. <i>Animal Genetics</i> , 2019, 50, 783-783.	0.6	8
3392	Rare Genetic Variants in Jewish Patients Suffering from Age-Related Macular Degeneration. <i>Genes</i> , 2019, 10, 825.	1.0	7
3393	Chromosomal microarray and whole-exome sequence analysis in Taiwanese patients with autism spectrum disorder. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e996.	0.6	11
3394	Genetic variations in familial hypercholesterolemia and cascade screening in East Asians. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00520.	0.6	12
3395	PGC.SNV: understanding the evolutionary and medical implications of human single nucleotide variations in diverse populations. <i>Genome Biology</i> , 2019, 20, 215.	3.8	30
3396	Perrault syndrome with neurological features in a compound heterozygote for two TWNK mutations: overlap of TWNK-related recessive disorders. <i>Journal of Translational Medicine</i> , 2019, 17, 290.	1.8	14
3397	Steroid metabolism gene variants and their genotype-phenotype correlations in Chinese early-onset hypertension patients. <i>Hypertension Research</i> , 2019, 42, 1536-1543.	1.5	2
3398	Genome-wide association scan for QTL and their positional candidate genes associated with internal organ traits in chickens. <i>BMC Genomics</i> , 2019, 20, 669.	1.2	17
3399	Molecular Evolution in Small Steps under Prevailing Negative Selection: A Nearly Universal Rule of Codon Substitution. <i>Genome Biology and Evolution</i> , 2019, 11, 2702-2712.	1.1	10
3400	The genome-wide dynamics of purging during selfing in maize. <i>Nature Plants</i> , 2019, 5, 980-990.	4.7	42
3401	Cognitive characterization of SCAR10 caused by a homozygous c.132dupA mutation in the ANO10 gene. <i>Neurocase</i> , 2019, 25, 195-201.	0.2	7

#	ARTICLE	IF	CITATIONS
3402	Screening of 31 genes involved in monogenic forms of obesity in 23 Pakistani probands with early-onset childhood obesity: a case report. <i>BMC Medical Genetics</i> , 2019, 20, 152.	2.1	3
3403	REVEL and BayesDel outperform other in silico meta-predictors for clinical variant classification. <i>Scientific Reports</i> , 2019, 9, 12752.	1.6	52
3404	MARK4 protein can explore the active-like conformations in its non-phosphorylated state. <i>Scientific Reports</i> , 2019, 9, 12967.	1.6	4
3405	Rare variants in FANCA induce premature ovarian insufficiency. <i>Human Genetics</i> , 2019, 138, 1227-1236.	1.8	56
3406	Recurrent inflammatory disease caused by a heterozygous mutation in CD48. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 1441-1445.e17.	1.5	9
3407	A germline alteration of ERBB2 increases the risk of breast cancer in Chinese Han women with a familial history of malignant tumors. <i>Oncology Letters</i> , 2019, 18, 2885-2890.	0.8	0
3408	A missense variant in PER2 is associated with delayed sleep-wake phase disorder in a Japanese population. <i>Journal of Human Genetics</i> , 2019, 64, 1219-1225.	1.1	19
3409	Mid-Frequency Hearing Loss Is Characteristic Clinical Feature of OTOA-Associated Hearing Loss. <i>Genes</i> , 2019, 10, 715.	1.0	15
3410	Functional analysis of new human Bardet-Biedl syndrome loci specific variants in the zebrafish model. <i>Scientific Reports</i> , 2019, 9, 12936.	1.6	6
3411	Multigene Panel Testing Increases the Number of Loci Associated with Gastric Cancer Predisposition. <i>Cancers</i> , 2019, 11, 1340.	1.7	19
3412	The Prevalence and Clinical Characteristics of TECTA-Associated Autosomal Dominant Hearing Loss. <i>Genes</i> , 2019, 10, 744.	1.0	17
3413	Detailed Clinical Features of Deafness Caused by a Claudin-14 Variant. <i>International Journal of Molecular Sciences</i> , 2019, 20, 4579.	1.8	4
3414	Ancestry-Dependent Enrichment of Deleterious Homozygotes in Runs of Homozygosity. <i>American Journal of Human Genetics</i> , 2019, 105, 747-762.	2.6	36
3415	Whole Genome Sequencing of the Mutamouse Model Reveals Strain- and Colony-Level Variation, and Genomic Features of the Transgene Integration Site. <i>Scientific Reports</i> , 2019, 9, 13775.	1.6	4
3416	Estimating the Frequency of Single Point Driver Mutations across Common Solid Tumours. <i>Scientific Reports</i> , 2019, 9, 13452.	1.6	6
3417	Functional rare variants influence the clinical response to anti-TNF therapy in Crohn's disease. <i>Therapeutic Advances in Gastroenterology</i> , 2019, 12, 175628481986784.	1.4	1
3418	De novo identification of essential protein domains from CRISPR-Cas9 tiling-sgRNA knockout screens. <i>Nature Communications</i> , 2019, 10, 4541.	5.8	44
3419	de novo 3.8-Mb inversion affecting the EDA and XIST genes in a heterozygous female calf with generalized hypohidrotic ectodermal dysplasia. <i>BMC Genomics</i> , 2019, 20, 715.	1.2	8

#	ARTICLE	IF	CITATIONS
3420	Clinical features and molecular genetic analysis of thanatophoric dysplasia type I in a neonate with a de novo c.2419G>A (p. Ter807Arg) (X807R) mutation in FGFR3. <i>Experimental and Molecular Pathology</i> , 2019, 111, 104297.	0.9	0
3421	Annotation of Variant Data from High-Throughput DNA Sequencing from Tumor Specimens: Filtering Strategies to Identify Driver Mutations. <i>Methods in Molecular Biology</i> , 2019, 1908, 49-60.	0.4	1
3422	Capturing variation impact on molecular interactions in the IMEx Consortium mutations data set. <i>Nature Communications</i> , 2019, 10, 10.	5.8	193
3423	EAP1 regulation of GnRH promoter activity is important for human pubertal timing. <i>Human Molecular Genetics</i> , 2019, 28, 1357-1368.	1.4	29
3424	Genome-wide association meta-analysis of 30,000 samples identifies seven novel loci for quantitative ECG traits. <i>European Journal of Human Genetics</i> , 2019, 27, 952-962.	1.4	29
3425	Molecular expression, characterization and mechanism of ALAS2 gain-of-function mutants. <i>Molecular Medicine</i> , 2019, 25, 4.	1.9	18
3426	Structure and function analysis of the <i>C. elegans</i> aminophospholipid translocase TAT1. <i>Journal of Cell Science</i> , 2019, 132, .	1.2	3
3427	Mutated SON putatively causes a cancer syndrome comprising high-risk medulloblastoma combined with café-au-lait spots. <i>Familial Cancer</i> , 2019, 18, 353-358.	0.9	4
3428	Functional Annotation and Analysis of Dual Oxidase 1 (DUOX1): a Potential Anti-pyocyanin Immune Component. <i>Interdisciplinary Sciences, Computational Life Sciences</i> , 2019, 11, 597-610.	2.2	2
3429	Significant abundance of configurations of coding variants in diploid human genomes. <i>Nucleic Acids Research</i> , 2019, 47, 2981-2995.	6.5	5
3430	GenESysV: a fast, intuitive and scalable genome exploration open source tool for variants generated from high-throughput sequencing projects. <i>BMC Bioinformatics</i> , 2019, 20, 61.	1.2	2
3431	Near-Comprehensive Resequencing of Cancer-Associated Genes in Surgically Resected Metastatic Liver Tumors of Gastric Cancer. <i>American Journal of Pathology</i> , 2019, 189, 784-796.	1.9	18
3432	deepDriver: Predicting Cancer Driver Genes Based on Somatic Mutations Using Deep Convolutional Neural Networks. <i>Frontiers in Genetics</i> , 2019, 10, 13.	1.1	72
3433	A Novel CNGA1 Gene Mutation (c.G622A) of Autosomal Recessive Retinitis Pigmentosa Leads to the CNGA1 Protein Reduction on Membrane. <i>Biochemical Genetics</i> , 2019, 57, 540-554.	0.8	5
3434	Varanto: variant enrichment analysis and annotation. <i>Bioinformatics</i> , 2019, 35, 3154-3156.	1.8	1
3435	Spectrum of ARSA variations in Asian Indian patients with Arylsulfatase A deficient metachromatic leukodystrophy. <i>Journal of Human Genetics</i> , 2019, 64, 323-331.	1.1	15
3436	Loss-of-function mutations in QRICH2 cause male infertility with multiple morphological abnormalities of the sperm flagella. <i>Nature Communications</i> , 2019, 10, 433.	5.8	108
3437	Whole-genome sequencing identifies complex contributions to genetic risk by variants in genes causing monogenic systemic lupus erythematosus. <i>Human Genetics</i> , 2019, 138, 141-150.	1.8	63

#	ARTICLE	IF	CITATIONS
3438	Comprehensive functional annotation of susceptibility SNPs prioritized 10 genes for schizophrenia. <i>Translational Psychiatry</i> , 2019, 9, 56.	2.4	20
3439	VARIFlâ€”Web-Based Automatic Variant Identification, Filtering and Annotation of Amplicon Sequencing Data. <i>Journal of Personalized Medicine</i> , 2019, 9, 10.	1.1	1
3440	Genotypic and phenotypic characterization of Chinese patients with osteogenesis imperfecta. <i>Human Mutation</i> , 2019, 40, 588-600.	1.1	19
3441	Untargeted metabolomics identifies unique though benign biochemical changes in patients with pathogenic variants in UROC1. <i>Molecular Genetics and Metabolism Reports</i> , 2019, 18, 14-18.	0.4	15
3442	Genetic basis for plasma amino acid concentrations based on absolute quantification: a genome-wide association study in the Japanese population. <i>European Journal of Human Genetics</i> , 2019, 27, 621-630.	1.4	16
3443	<p>Mutation spectrum of germline cancer susceptibility genes among unselected Chinese colorectal cancer patients</p>. <i>Cancer Management and Research</i> , 2019, Volume 11, 3721-3739.	0.9	15
3444	Structural Analysis and Conformational Dynamics of STN1 Gene Mutations Involved in Coat Plus Syndrome. <i>Frontiers in Molecular Biosciences</i> , 2019, 6, 41.	1.6	53
3445	A familial case of pseudohypoaldosteronism type II (PHA2) with a novel mutation (D564N) in the acidic motif in <i>WNK4</i>. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e705.	0.6	5
3446	Maple syrup urine disease mutation spectrum in a cohort of 40 consanguineous patients and insilico analysis of novel mutations. <i>Metabolic Brain Disease</i> , 2019, 34, 1145-1156.	1.4	11
3447	Predicting disease-causing variant combinations. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 11878-11887.	3.3	68
3448	Innovative strategies for annotating the â€œrelationSNPâ€•between variants and molecular phenotypes. <i>BioData Mining</i> , 2019, 12, 10.	2.2	6
3449	<i>GATA4</i> screening in Iranian patients of various ethnicities affected with congenital heart disease: Coâ€œoccurrence of a novel de novo translocation (5;7) and a likely pathogenic heterozygous <i>GATA4</i> mutation in a family with autosomal dominant congenital heart disease. <i>Journal of Clinical Laboratory Analysis</i> , 2019, 33, e22923.	0.9	7
3450	Computational Screening and Analysis of Lung Cancer Related Non-Synonymous Single Nucleotide Polymorphisms on the Human Kirsten Rat Sarcoma Gene. <i>Molecules</i> , 2019, 24, 1951.	1.7	32
3451	Evolutionary coupling analysis identifies the impact of disease-associated variants at less-conserved sites. <i>Nucleic Acids Research</i> , 2019, 47, e94-e94.	6.5	11
3452	Genomics of benign adrenocortical tumors. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 2019, 193, 105414.	1.2	15
3453	Identification of Three Novel Homozygous NAGLU Mutations in Egyptian Patients with Sanfilippo Syndrome B. <i>Meta Gene</i> , 2019, 21, 100580.	0.3	3
3454	Prioritization of SNPs in γ -LATâ€•1 culpable of Lysinuric protein intolerance and their mutational impacts using proteinâ€œprotein docking and molecular dynamics simulation studies. <i>Journal of Cellular Biochemistry</i> , 2019, 120, 18496-18508.	1.2	10
3455	Investigation of new candidate genes in retinoblastoma using the TruSight One â€œclinical exomeâ€•gene panel. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e785.	0.6	7

#	ARTICLE	IF	CITATIONS
3456	<sc></sc> -Serine dietary supplementation is associated with clinical improvement of loss-of-function <i>GRIN2B</i>-related pediatric encephalopathy. <i>Science Signaling</i> , 2019, 12, .	1.6	53
3457	Mutation spectrum of Charcotâ€Marieâ€Tooth disease among the Han Chinese in Taiwan. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1090-1101.	1.7	29
3458	Costâ€effective molecular inversion probeâ€based <i>ABCA4</i> sequencing reveals deepâ€intronic variants in Stargardt disease. <i>Human Mutation</i> , 2019, 40, 1749-1759.	1.1	39
3459	Association of genetic variants at 22q11.2 chromosomal region with cognitive performance in Japanese patients with schizophrenia. <i>Schizophrenia Research: Cognition</i> , 2019, 17, 100134.	0.7	0
3460	Exome Sequencing in Clinical Hepatology. <i>Hepatology</i> , 2019, 70, 2185-2192.	3.6	19
3461	NTHL1 biallelic mutations seldom cause colorectal cancer, serrated polyposis or a multi-tumor phenotype, in absence of colorectal adenomas. <i>Scientific Reports</i> , 2019, 9, 9020.	1.6	23
3462	A comprehensive analysis of NPHS1 gene mutations in patients with sporadic focal segmental glomerulosclerosis. <i>BMC Medical Genetics</i> , 2019, 20, 111.	2.1	11
3463	Genetic testing of Mucopolysaccharidoses disease using multiplex PCR- based panels of STR markers: in silico analysis of novel mutations. <i>Metabolic Brain Disease</i> , 2019, 34, 1447-1455.	1.4	5
3464	RNA sequence analysis reveals macroscopic somatic clonal expansion across normal tissues. <i>Science</i> , 2019, 364, .	6.0	369
3465	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unitâ€successes and challenges. <i>European Journal of Pediatrics</i> , 2019, 178, 1207-1218.	1.3	59
3466	A case study of a long-term glioblastoma survivor with unmethylated MGMT and hypermutated genotype. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003251.	0.5	2
3467	A truncating CLDN9 variant is associated with autosomal recessive nonsyndromic hearing loss. <i>Human Genetics</i> , 2019, 138, 1071-1075.	1.8	17
3468	Identification and in-silico analysis of a novel disease-causing variant in the GUSB gene for Mucopolysaccharidosis VII presenting as non-immune fetal hydrops. <i>Gene Reports</i> , 2019, 16, 100437.	0.4	1
3469	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. <i>Nature Communications</i> , 2019, 10, 2506.	5.8	46
3470	Identifying Candidate Druggable Targets in Canine Cancer Cell Lines Using Whole-Exome Sequencing. <i>Molecular Cancer Therapeutics</i> , 2019, 18, 1460-1471.	1.9	24
3471	EFL1 mutations impair eIF6 release to cause Shwachman-Diamond syndrome. <i>Blood</i> , 2019, 134, 277-290.	0.6	48
3472	Assessing concordance among human, in silico predictions and functional assays on genetic variant classification. <i>Bioinformatics</i> , 2019, 35, 5163-5170.	1.8	4
3473	Phenotypic characterization of the novel, nonâ€hotspot oncogenic KRAS mutants E31D and E63K. <i>Oncology Letters</i> , 2019, 18, 420-432.	0.8	7

#	ARTICLE	IF	CITATIONS
3474	Genomic signatures of extensive inbreeding in Isle Royale wolves, a population on the threshold of extinction. <i>Science Advances</i> , 2019, 5, eaau0757.	4.7	173
3475	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. <i>Nature Communications</i> , 2019, 10, 2373.	5.8	86
3476	Rare variant phasing using paired tumor:normal sequence data. <i>BMC Bioinformatics</i> , 2019, 20, 265.	1.2	2
3477	A metabolic profile of polyamines in parkinson disease: A promising biomarker. <i>Annals of Neurology</i> , 2019, 86, 251-263.	2.8	74
3478	Targeted next generation sequencing as a tool for precision medicine. <i>BMC Medical Genomics</i> , 2019, 12, 81.	0.7	54
3479	Multiple rare and common variants in APOB gene locus associated with oxidatively modified low-density lipoprotein levels. <i>PLoS ONE</i> , 2019, 14, e0217620.	1.1	9
3480	Independent Severe Cases of Heterozygous Familial Hypercholesterolemia Caused by the W483X and Novel W483G Mutations in the Low-Density Lipoprotein Receptor Gene That Were Clinically Diagnosed as Homozygous Cases. <i>Genetic Testing and Molecular Biomarkers</i> , 2019, 23, 401-408.	0.3	3
3481	RNA editing alterations in a multi-ethnic Alzheimer disease cohort converge on immune and endocytic molecular pathways. <i>Human Molecular Genetics</i> , 2019, 28, 3053-3061.	1.4	19
3482	Novel Germline Mutations of BRCA1 and BRCA2 in Korean Familial Breast Cancer Patients. <i>Chonnam Medical Journal</i> , 2019, 55, 99.	0.5	0
3483	<i>In vitro</i> and <i>in vivo</i> efficacy of a Rift Valley fever virus vaccine based on pseudovirus. <i>Human Vaccines and Immunotherapeutics</i> , 2019, 15, 2286-2294.	1.4	19
3484	Influence of genetic factors on long-term treatment related neurocognitive complications, and on anxiety and depression in survivors of childhood acute lymphoblastic leukemia: The Petale study. <i>PLoS ONE</i> , 2019, 14, e0217314.	1.1	14
3485	Whole Exome Sequencing Identified a Novel Biallelic SMARCA1 Mutation in the Extremely Rare Disease SIOD. <i>Frontiers in Genetics</i> , 2019, 10, 565.	1.1	4
3486	A catalog of single nucleotide changes distinguishing modern humans from archaic hominins. <i>Scientific Reports</i> , 2019, 9, 8463.	1.6	60
3487	A missense mutation (p.Tyr452Cys) in the CAD gene compromises reproductive success in French Normande cattle. <i>Journal of Dairy Science</i> , 2019, 102, 6340-6356.	1.4	21
3488	<i>CYP24A1</i> and <i>SLC34A1</i> genetic defects associated with idiopathic infantile hypercalcemia: from genotype to phenotype. <i>Clinical Chemistry and Laboratory Medicine</i> , 2019, 57, 1650-1667.	1.4	30
3489	Novel heterozygous <i>BPIFC</i> variant in a Chinese pedigree with hereditary trichilemmal cysts. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e697.	0.6	1
3490	<i>NF1</i> mutations identify molecular and clinical subtypes of lung adenocarcinomas. <i>Cancer Medicine</i> , 2019, 8, 4330-4337.	1.3	14
3491	Identification and characterization of rare toll-like receptor 3 variants in patients with autoimmune Addison's disease. <i>Journal of Translational Autoimmunity</i> , 2019, 1, 100005.	2.0	5

#	ARTICLE	IF	CITATIONS
3492	Mutations in <i>KARS</i> cause a severe neurological and neurosensory disease with optic neuropathy. <i>Human Mutation</i> , 2019, 40, 1826-1840.	1.1	15
3493	Massively parallel sequencing of tenosynovial giant cell tumors reveals novel CSF1 fusion transcripts and novel somatic CBL mutations. <i>International Journal of Cancer</i> , 2019, 145, 3276-3284.	2.3	28
3494	Assessment of Potential Clinical Role for Exome Sequencing in Schizophrenia. <i>Schizophrenia Bulletin</i> , 2020, 46, 328-335.	2.3	7
3495	Identification of genetic association between cardiorespiratory fitness and the trainability genes in childhood acute lymphoblastic leukemia survivors. <i>BMC Cancer</i> , 2019, 19, 443.	1.1	9
3496	Metabolic reprogramming toward oxidative phosphorylation identifies a therapeutic target for mantle cell lymphoma. <i>Science Translational Medicine</i> , 2019, 11, .	5.8	161
3497	A Chinese family with familial hemiplegic migraine type 2 due to a novel missense mutation in <i>ATP1A2</i> . <i>Cephalalgia</i> , 2019, 39, 1382-1395.	1.8	7
3498	A novel <i>WARS</i> mutation (p.Asp314Gly) identified in a Chinese distal hereditary motor neuropathy family. <i>Clinical Genetics</i> , 2019, 96, 176-182.	1.0	21
3499	The genetics of cholesteatoma study. Loss of function variants in an affected family. <i>Clinical Otolaryngology</i> , 2019, 44, 826-830.	0.6	7
3500	Increased Diagnostic Yield of Spastic Paraplegia with or Without Cerebellar Ataxia Through Whole-Genome Sequencing. <i>Cerebellum</i> , 2019, 18, 781-790.	1.4	28
3501	Novel EYA4 variant in Slovak family with late onset autosomal dominant hearing loss: a case report. <i>BMC Medical Genetics</i> , 2019, 20, 84.	2.1	9
3502	BRCA1 and BRCA2 specific in silico tools for variant interpretation in the CAGI 5 ENIGMA challenge. <i>Human Mutation</i> , 2019, 40, 1593-1611.	1.1	11
3503	Identification of novel compound heterozygous mutations in <i>ACO2</i> in a patient with progressive cerebral and cerebellar atrophy. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00698.	0.6	6
3504	<i>Transforming growth factor beta</i> -induced p.(L558P) variant is associated with autosomal dominant lattice corneal dystrophy type IV in a large cohort of Spanish patients. <i>Clinical and Experimental Ophthalmology</i> , 2019, 47, 871-880.	1.3	0
3505	Identification of novel sarcoma risk genes using a two-stage genome wide DNA sequencing strategy in cancer cluster families and population case and control cohorts. <i>BMC Medical Genetics</i> , 2019, 20, 69.	2.1	2
3506	Clinico-pathological associations and concomitant mutations of the RAS/RAF pathway in metastatic colorectal cancer. <i>Journal of Translational Medicine</i> , 2019, 17, 137.	1.8	13
3507	Mutation spectrum and clinical investigation of achromatopsia patients with mutations in the <i>GNAT2</i> gene. <i>Human Mutation</i> , 2019, 40, 1145-1155.	1.1	15
3508	BMP4 mutations in tooth agenesis and low bone mass. <i>Archives of Oral Biology</i> , 2019, 103, 40-46.	0.8	32
3509	CD91 on dendritic cells governs immunosurveillance of nascent, emerging tumors. <i>JCI Insight</i> , 2019, 4, .	2.3	17

#	ARTICLE	IF	CITATIONS
3510	Further support linking the 22q11.2 microduplication to an increased risk of bladder exstrophy and highlighting LZTR1 as a candidate gene. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e666.	0.6	9
3511	A Unique Presentation of Infantile-Onset Colitis and Eosinophilic Disease without Recurrent Infections Resulting from a Novel Homozygous CARMIL2 Variant. <i>Journal of Clinical Immunology</i> , 2019, 39, 430-439.	2.0	21
3512	Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. <i>American Journal of Ophthalmology</i> , 2019, 207, 87-98.	1.7	20
3513	Molecular Origins of Complex Heritability in Natural Genotype-to-Phenotype Relationships. <i>Cell Systems</i> , 2019, 8, 363-379.e3.	2.9	26
3514	A segregating human allele of <i>SPO11</i> modeled in mice disrupts timing and amounts of meiotic recombination, causing oligospermia and a decreased ovarian reserve. <i>Biology of Reproduction</i> , 2019, 101, 347-359.	1.2	10
3515	A mutation in Site 1 Protease is associated with a complex phenotype that includes episodic hyperCKemia and focal myoedema. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00733.	0.6	13
3516	Uncommon mutational profiles of metastatic colorectal cancer detected during routine genotyping using next generation sequencing. <i>Scientific Reports</i> , 2019, 9, 7083.	1.6	5
3517	Molecular Evolution in Large Steps—Codon Substitutions under Positive Selection. <i>Molecular Biology and Evolution</i> , 2019, 36, 1862-1873.	3.5	16
3518	Somatic variants in epigenetic modifiers can predict failure of response to imatinib but not to second-generation tyrosine kinase inhibitors. <i>Haematologica</i> , 2019, 104, 2400-2409.	1.7	37
3519	An ADAMTS3 missense variant is associated with Norwich Terrier upper airway syndrome. <i>PLoS Genetics</i> , 2019, 15, e1008102.	1.5	14
3520	Nonsense mutation in <i>CFAP43</i> causes normal-pressure hydrocephalus with ciliary abnormalities. <i>Neurology</i> , 2019, 92, e2364-e2374.	1.5	65
3521	Human collectin-11 (COLEC11) and its synergic genetic interaction with MASP2 are associated with the pathophysiology of Chagas Disease. <i>PLoS Neglected Tropical Diseases</i> , 2019, 13, e0007324.	1.3	7
3522	Identification of novel mutations in preaxial polydactyly patients through whole-exome sequencing. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e690.	0.6	7
3523	Functional characterization of 3D protein structures informed by human genetic diversity. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 8960-8965.	3.3	33
3524	Developmental delay, coarse facial features, and epilepsy in a patient with <i>EXT2</i> gene variants. <i>Clinical Case Reports (discontinued)</i> , 2019, 7, 632-637.	0.2	6
3525	Identification of a novel ANO5 missense mutation in a Chinese family with familial florid osseous dysplasia. <i>Journal of Human Genetics</i> , 2019, 64, 599-607.	1.1	6
3526	Design of a Targeted Sequencing Assay to Detect Rare Mutations in Circulating Tumor DNA. <i>Genetic Testing and Molecular Biomarkers</i> , 2019, 23, 264-269.	0.3	4
3527	Genetic Techniques Used in the Diagnosis of Inherited Platelet Disorders. <i>Seminars in Thrombosis and Hemostasis</i> , 2019, 45, 685-694.	1.5	4

#	ARTICLE	IF	CITATIONS
3528	Mitochondrial Disease Genetics. , 2019, , 41-62.		0
3529	Update on Molecular Testing in von Willebrand Disease. <i>Seminars in Thrombosis and Hemostasis</i> , 2019, 45, 708-719.	1.5	9
3530	<i>DUOX2</i> / <i>DUOXA2</i> Mutations Frequently Cause Congenital Hypothyroidism that Evades Detection on Newborn Screening in the United Kingdom. <i>Thyroid</i> , 2019, 29, 790-801.	2.4	26
3531	Targeted NGS Platforms for Genetic Screening and Gene Discovery in Primary Immunodeficiencies. <i>Frontiers in Immunology</i> , 2019, 10, 316.	2.2	42
3532	Circulating tumor DNA analyses predict progressive disease and indicate trastuzumab-resistant mechanism in advanced gastric cancer. <i>EBioMedicine</i> , 2019, 43, 261-269.	2.7	68
3533	Comparison of Predictive <i>In Silico</i> Tools on Missense Variants in <i>GJB2</i> , <i>GJB6</i> , and <i>GJB3</i> Genes Associated with Autosomal Recessive Deafness 1A (DFNB1A). <i>Scientific World Journal, The</i> , 2019, 2019, 1-9.	0.8	26
3534	Mutations in <i>PIK3C2A</i> cause syndromic short stature, skeletal abnormalities, and cataracts associated with ciliary dysfunction. <i>PLoS Genetics</i> , 2019, 15, e1008088.	1.5	45
3535	Translating genomics to the clinical diagnosis of disorders/differences of sex development. <i>Current Topics in Developmental Biology</i> , 2019, 134, 317-375.	1.0	25
3536	Sensitivity, advantages, limitations, and clinical utility of targeted next-generation sequencing panels for the diagnosis of selected lysosomal storage disorders. <i>Genetics and Molecular Biology</i> , 2019, 42, 197-206.	0.6	21
3537	Single-cell whole-genome sequencing reveals the functional landscape of somatic mutations in B lymphocytes across the human lifespan. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 9014-9019.	3.3	174
3538	Two novel <i>ANK1</i> loss-of-function mutations in Chinese families with hereditary spherocytosis. <i>Journal of Cellular and Molecular Medicine</i> , 2019, 23, 4454-4463.	1.6	10
3539	eDiVA "Classification and prioritization of pathogenic variants for clinical diagnostics. <i>Human Mutation</i> , 2019, 40, 865-878.	1.1	19
3540	Clinical and Genetic Investigation of Premature Ovarian Insufficiency Cases from Turkey. <i>Journal of Gynecology Obstetrics and Human Reproduction</i> , 2019, 48, 817-823.	0.6	6
3541	Novel mutations in <i>MYBPC1</i> are associated with myogenic tremor and mild myopathy. <i>Annals of Neurology</i> , 2019, 86, 129-142.	2.8	27
3542	Structural and functional impact of non-synonymous SNPs in the CST complex subunit TEN1: structural genomics approach. <i>Bioscience Reports</i> , 2019, 39, .	1.1	17
3543	De novo single-nucleotide and copy number variation in discordant monozygotic twins reveals disease-related genes. <i>European Journal of Human Genetics</i> , 2019, 27, 1121-1133.	1.4	37
3544	Cross-Cancer Pleiotropic Associations with Lung Cancer Risk in African Americans. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2019, 28, 715-723.	1.1	11
3545	A novel pathogenic variant in <i>OFD1</i> results in X-linked Joubert syndrome with orofacioidigital features and pituitary aplasia. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1010-1014.	0.7	9

#	ARTICLE	IF	CITATIONS
3546	How to discover new antibiotic resistance genes?. <i>Expert Review of Molecular Diagnostics</i> , 2019, 19, 349-362.	1.5	15
3547	SNP Variation of RELN Gene and Schizophrenia in a Chinese Population: A Hospital-Based Caseâ€“Control Study. <i>Frontiers in Genetics</i> , 2019, 10, 175.	1.1	6
3548	A synonymous germline variant in a gene encoding a cell adhesion molecule is associated with cutaneous mast cell tumour development in Labrador and Golden Retrievers. <i>PLoS Genetics</i> , 2019, 15, e1007967.	1.5	9
3549	Pervasive function and evidence for selection across standing genetic variation in <i>S. cerevisiae</i> . <i>Nature Communications</i> , 2019, 10, 1222.	5.8	10
3550	Systematic evaluation of gene variants linked to hearing loss based on allele frequency threshold and filtering allele frequency. <i>Scientific Reports</i> , 2019, 9, 4583.	1.6	13
3551	Loss of function mutations in essential genes cause embryonic lethality in pigs. <i>PLoS Genetics</i> , 2019, 15, e1008055.	1.5	46
3552	A de novo mutation in DHD domain of SKI causing spina bifida with no craniofacial malformation or intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 936-939.	0.7	3
3553	Functional characterization of the C7ORF76 genomic region, a prominent GWAS signal for osteoporosis in 7q21.3. <i>Bone</i> , 2019, 123, 39-47.	1.4	12
3554	A novel homozygous FBXO43 mutation associated with male infertility and teratozoospermia in a consanguineous Chinese family. <i>Fertility and Sterility</i> , 2019, 111, 909-917.e1.	0.5	18
3555	Characterization of growth hormone gene (GH) in three Egyptian goat breeds. <i>Meta Gene</i> , 2019, 20, 100556.	0.3	0
3556	Polygonal-feature-based shape context for flexible surface vision positioning. <i>Measurement Science and Technology</i> , 2019, 30, 055403.	1.4	4
3557	Making Sense of the Epigenome Using Data Integration Approaches. <i>Frontiers in Pharmacology</i> , 2019, 10, 126.	1.6	58
3558	Moving beyond simple answers to complex disorders in sarcomeric cardiomyopathies: the role of integrated systems. <i>Pflugers Archiv European Journal of Physiology</i> , 2019, 471, 661-671.	1.3	14
3559	Gliosarcoma Is Driven by Alterations in PI3K/Akt, RAS/MAPK Pathways and Characterized by Collagen Gene Expression Signature. <i>Cancers</i> , 2019, 11, 284.	1.7	18
3560	Novel compound heterozygous mutations in the SPTA1 gene, causing hereditary spherocytosis in a neonate with Coombsâ€“negative hemolytic jaundice. <i>Molecular Medicine Reports</i> , 2019, 19, 2801-2807.	1.1	7
3561	New variants in COL5A1 gene among Polish patients with Ehlers-Danlos syndrome â€“ analysis of nine cases. <i>Postepy Dermatologii I Alergologii</i> , 2019, 36, 29-33.	0.4	6
3562	Selecting variants of unknown significance through network-based gene-association significantly improves risk prediction for disease-control cohorts. <i>Scientific Reports</i> , 2019, 9, 3266.	1.6	18
3563	Genome-wide scans of myopia in Pennsylvania Amish families reveal significant linkage to 12q15, 8q21.3 and 5p15.33. <i>Human Genetics</i> , 2019, 138, 339-354.	1.8	8

#	ARTICLE	IF	CITATIONS
3564	Targeted next generation sequencing identifies novel pathogenic variants and provides molecular diagnoses in a cohort of pediatric and adult patients with unexplained mitochondrial dysfunction. <i>Mitochondrion</i> , 2019, 47, 309-317.	1.6	8
3565	A familial congenital heart disease with a possible multigenic origin involving a mutation in <i>BMPR1A</i> . <i>Scientific Reports</i> , 2019, 9, 2959.	1.6	14
3566	Novel mutations causing biotinidase deficiency in individuals identified by the newborn screening program in Minas Gerais, Brazil. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 978-982.	0.7	7
3568	A Novel <i>FOXN1</i> Variant Is Identified in Two Siblings with Nude Severe Combined Immunodeficiency. <i>Journal of Clinical Immunology</i> , 2019, 39, 144-147.	2.0	3
3569	Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling. <i>Neuromuscular Disorders</i> , 2019, 29, 601-613.	0.3	18
3570	An integrated whole genome analysis of <i>Mycobacterium tuberculosis</i> reveals insights into relationship between its genome, transcriptome and methylome. <i>Scientific Reports</i> , 2019, 9, 5204.	1.6	26
3571	The genomic mutation spectrums of breast fibroadenomas in Chinese population by whole exome sequencing analysis. <i>Cancer Medicine</i> , 2019, 8, 2372-2379.	1.3	12
3572	Mutation in NADPH oxidase 3 (<i>NOX3</i>) impairs SHH signaling and increases cerebellar neural stem/progenitor cell proliferation. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2019, 1865, 1502-1515.	1.8	10
3573	Protein structure aids predicting functional perturbation of missense variants in <i>SCN5A</i> and <i>KCNQ1</i> . <i>Computational and Structural Biotechnology Journal</i> , 2019, 17, 206-214.	1.9	19
3574	Role of protein structure in variant annotation: structural insight of mutations causing 6-pyruvoyl-tetrahydropterin synthase deficiency. <i>Pathology</i> , 2019, 51, 274-280.	0.3	7
3575	Carrier frequency estimation of Zellweger spectrum disorder using ExAC database and bioinformatics tools. <i>Genetics in Medicine</i> , 2019, 21, 1969-1976.	1.1	10
3576	Genetic analysis of patients with fructose-1,6-bisphosphatase deficiency. <i>Gene</i> , 2019, 699, 102-109.	1.0	9
3577	Risk gene-set and pathways in 22q11.2 deletion-related schizophrenia: a genealogical molecular approach. <i>Translational Psychiatry</i> , 2019, 9, 15.	2.4	13
3578	Genetic Variation in Pan Species Is Shaped by Demographic History and Harbors Lineage-Specific Functions. <i>Genome Biology and Evolution</i> , 2019, 11, 1178-1191.	1.1	15
3579	Sequence Variations in <i>pxr</i> (<i>nr1i2</i>) From Zebrafish (<i>Danio rerio</i>) Strains Affect Nuclear Receptor Function. <i>Toxicological Sciences</i> , 2019, 168, 28-39.	1.4	6
3580	Comparison of different functional prediction scores using a gene-based permutation model for identifying cancer driver genes. <i>BMC Medical Genomics</i> , 2019, 12, 22.	0.7	12
3581	The rare mutation in the endosome-associated recycling protein gene <i>VPS50</i> is associated with human neural tube defects. <i>Molecular Cytogenetics</i> , 2019, 12, 8.	0.4	9
3582	SeqSQC: A Bioconductor Package for Evaluating the Sample Quality of Next-generation Sequencing Data. <i>Genomics, Proteomics and Bioinformatics</i> , 2019, 17, 211-218.	3.0	6

#	ARTICLE	IF	CITATIONS
3583	Big data in der Diagnostik genetischer Schwerhörigkeit. <i>Laryngo- Rhino- Otologie</i> , 2019, 98, S32-S81.	0.2	6
3584	Loss of Cajal bodies in motor neurons from patients with novel mutations in VRK1. <i>Human Molecular Genetics</i> , 2019, 28, 2378-2394.	1.4	17
3585	Hyperammonemia after capecitabine associated with occult impairment of the urea cycle. <i>Cancer Medicine</i> , 2019, 8, 1996-2004.	1.3	8
3586	Atomic insight into prion disorder: An intricate detail gained by 0.5 μ s molecular dynamics simulation of preventive G127V and deleterious D178V mutation in prion protein. <i>Journal of Cellular Biochemistry</i> , 2019, 120, 14156-14164.	1.2	2
3587	Clinical diagnosis and genetic counseling of atypical ataxia-telangiectasia in a Chinese family. <i>Molecular Medicine Reports</i> , 2019, 19, 3441-3448.	1.1	3
3588	Association of XRCC1 and XPD functional gene variants with nicotine dependence and/or schizophrenia: a case-control study and in silico analysis. <i>Journal of Theoretical Social Psychology</i> , 2019, 29, 21-27.	1.2	0
3589	Population-based analysis of BAP1 germline variations in patients with uveal melanoma. <i>Human Molecular Genetics</i> , 2019, 28, 2415-2426.	1.4	17
3590	Genetic risk scores demonstrate the cumulative association of single nucleotide polymorphisms in gut microbiome-related genes with obesity phenotypes in preschool age children. <i>Pediatric Obesity</i> , 2019, 14, e12530.	1.4	2
3592	A splice donor variant in CCDC189 is associated with asthenospermia in Nordic Red dairy cattle. <i>BMC Genomics</i> , 2019, 20, 286.	1.2	21
3593	Evolution of a novel chimeric maltotriose transporter in <i>Saccharomyces eubayanus</i> from parent proteins unable to perform this function. <i>PLoS Genetics</i> , 2019, 15, e1007786.	1.5	35
3594	New Chondrosarcoma Cell Lines with Preserved Stem Cell Properties to Study the Genomic Drift During In Vitro/In Vivo Growth. <i>Journal of Clinical Medicine</i> , 2019, 8, 455.	1.0	18
3595	Multi-region sequencing unveils novel actionable targets and spatial heterogeneity in esophageal squamous cell carcinoma. <i>Nature Communications</i> , 2019, 10, 1670.	5.8	110
3596	Review: Precision medicine and driver mutations: Computational methods, functional assays and conformational principles for interpreting cancer drivers. <i>PLoS Computational Biology</i> , 2019, 15, e1006658.	1.5	83
3597	Mutation p.R356Q in the Collybistin Phosphoinositide Binding Site Is Associated With Mild Intellectual Disability. <i>Frontiers in Molecular Neuroscience</i> , 2019, 12, 60.	1.4	10
3598	Identification of novel pathogenic F13A1 mutation and novel NBEAL2 gene missense mutation in a pedigree with hereditary congenital factor XIII deficiency. <i>Gene</i> , 2019, 702, 143-147.	1.0	4
3599	Mutational analysis of the FAM175A gene in patients with premature ovarian insufficiency. <i>Reproductive BioMedicine Online</i> , 2019, 38, 943-950.	1.1	2
3600	Heterogeneous pathway activation and drug response modelled in colorectal-tumor-derived 3D cultures. <i>PLoS Genetics</i> , 2019, 15, e1008076.	1.5	59
3601	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2019, 104, 758-766.	2.6	34

#	ARTICLE	IF	CITATIONS
3602	Identification of human D lactate dehydrogenase deficiency. <i>Nature Communications</i> , 2019, 10, 1477.	5.8	62
3603	Pathogenic variants in the AFG3L2 proteolytic domain cause SCA28 through haploinsufficiency and proteostatic stress-driven OMA1 activation. <i>Journal of Medical Genetics</i> , 2019, 56, 499-511.	1.5	20
3604	Genome-wide selection footprints and deleterious variations in young Asian allotetraploid rapeseed. <i>Plant Biotechnology Journal</i> , 2019, 17, 1998-2010.	4.1	54
3605	Prioritizing candidate genes for fertility in dairy cows using gene-based analysis, functional annotation and differential gene expression. <i>BMC Genomics</i> , 2019, 20, 255.	1.2	30
3606	An enhanced workflow for variant interpretation in UniProtKB/Swiss-Prot improves consistency and reuse in ClinVar. <i>Database: the Journal of Biological Databases and Curation</i> , 2019, 2019, .	1.4	7
3607	Understanding the Impact of Aberrant Splicing in Coagulation Factor V Deficiency. <i>International Journal of Molecular Sciences</i> , 2019, 20, 910.	1.8	5
3608	Population-based identity-by-descent mapping combined with exome sequencing to detect rare risk variants for schizophrenia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2019, 180, 223-231.	1.1	2
3609	DAMPred: Recognizing Disease-Associated nsSNPs through Bayes-Guided Neural-Network Model Built on Low-Resolution Structure Prediction of Proteins and Protein-Protein Interactions. <i>Journal of Molecular Biology</i> , 2019, 431, 2449-2459.	2.0	19
3610	ALPL mutations in adults with rheumatologic disorders and low serum alkaline phosphatase activity. <i>Journal of Bone and Mineral Metabolism</i> , 2019, 37, 893-899.	1.3	9
3611	Genetic Variation in RIN3 in the Belgian Population Supports Its Involvement in the Pathogenesis of Paget's Disease of Bone and Modifies the Age of Onset. <i>Calcified Tissue International</i> , 2019, 104, 613-621.	1.5	5
3612	Clinical spectrum of <i>STX1B</i> -related epileptic disorders. <i>Neurology</i> , 2019, 92, e1238-e1249.	1.5	43
3613	Burkitt-like lymphoma with 11q aberration: a germinal center-derived lymphoma genetically unrelated to Burkitt lymphoma. <i>Haematologica</i> , 2019, 104, 1822-1829.	1.7	71
3614	Personalization of Logical Models With Multi-Omics Data Allows Clinical Stratification of Patients. <i>Frontiers in Physiology</i> , 2018, 9, 1965.	1.3	66
3615	Unknown mutations and genotype/phenotype correlations of autosomal recessive congenital ichthyosis in patients from Saudi Arabia and Pakistan. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e539.	0.6	9
3616	Identification of four novel mutations in MYO7A gene and their association with nonsyndromic deafness and Usher Syndrome 1B. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2019, 120, 166-172.	0.4	7
3617	In vivo, in vitro and in silico correlations of four de novo SCN1A missense mutations. <i>PLoS ONE</i> , 2019, 14, e0211901.	1.1	21
3618	LRP1B Polymorphisms Are Associated with Multiple Myeloma Risk in a Chinese Han Population. <i>Journal of Cancer</i> , 2019, 10, 577-582.	1.2	6
3619	Determining the Likelihood of Variant Pathogenicity Using Amino Acid-level Signal-to-Noise Analysis of Genetic Variation. <i>Journal of Visualized Experiments</i> , 2019, .	0.2	10

#	ARTICLE	IF	CITATIONS
3620	Incidence of Mutations in the <i>ALPL</i> , <i>GGPS1</i> , and <i>CYP1A1</i> Genes in Patients With Atypical Femoral Fractures. <i>JBMR Plus</i> , 2019, 3, 29-36.	1.3	23
3621	A diagnosis of discernment: Identifying a novel <i>ATRX</i> mutation in myelodysplastic syndrome with acquired β -thalassemia. <i>Cancer Genetics</i> , 2019, 231-232, 36-40.	0.2	3
3622	Phospholipase C-Gamma 2 Activity in Familial Steroid-Sensitive Nephrotic Syndrome. <i>Pediatric Research</i> , 2019, 85, 719-723.	1.1	7
3623	Genetic diversity and pathogenic variants as possible predictors of severity in a French sample of nonsyndromic heritable thoracic aortic aneurysms and dissections (nshTAAD). <i>Genetics in Medicine</i> , 2019, 21, 2015-2024.	1.1	39
3625	Phosphatidylinositol-3-kinase (PI3K)/Akt Signaling is Functionally Essential in Myxoid Liposarcoma. <i>Molecular Cancer Therapeutics</i> , 2019, 18, 834-844.	1.9	28
3626	Novel Variant of the Androgen Receptor Gene in a Patient With Complete Androgen Insensitivity Syndrome and Polyorchidism. <i>Frontiers in Endocrinology</i> , 2018, 9, 795.	1.5	0
3627	A High-Resolution Map of Wheat <i>QYr.ucw-1BL</i> , an Adult Plant Stripe Rust Resistance Locus in the Same Chromosomal Region as <i>Yr29</i> . <i>Plant Genome</i> , 2019, 12, 180055.	1.6	24
3628	Identification of a novel homozygous variant confirms <i>ITPA</i> as a developmental and epileptic encephalopathy gene. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 857-861.	0.7	14
3629	Exome sequencing in 51 early onset non-familial CRC cases. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e605.	0.6	17
3630	Missense Variants in the Histone Acetyltransferase Complex Component Gene <i>TRRAP</i> Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	2.6	30
3631	Genetic variation in thyroid folliculogenesis influences susceptibility to hypothyroidism-induced hearing impairment. <i>Mammalian Genome</i> , 2019, 30, 5-22.	1.0	2
3632	Novel Missense Mutations in <i>BEST1</i> Are Associated with Bestrophinopathies in Lebanese Patients. <i>Genes</i> , 2019, 10, 151.	1.0	7
3633	Characterization, evolution, and expression analysis of <i>TLR7</i> gene subfamily members in <i>Mastacembelus armatus</i> (Synbranchiformes: Mastacembelidae). <i>Developmental and Comparative Immunology</i> , 2019, 95, 77-88.	1.0	12
3634	Homozygous loss-of-function mutations in <i>FSIP2</i> cause male infertility with asthenoteratospermia. <i>Journal of Genetics and Genomics</i> , 2019, 46, 53-56.	1.7	31
3635	Novel mutations in the <i>RS1</i> gene in Japanese patients with X-linked congenital retinoschisis. <i>Human Genome Variation</i> , 2019, 6, 3.	0.4	18
3636	Molecular genetic analysis using targeted NGS analysis of 677 individuals with retinal dystrophy. <i>Scientific Reports</i> , 2019, 9, 1219.	1.6	76
3637	Variant <i>PADI3</i> in Central Centrifugal Cicatricial Alopecia. <i>New England Journal of Medicine</i> , 2019, 380, 833-841.	13.9	102
3638	Bi-allelic Variants in <i>TONSL</i> Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. <i>American Journal of Human Genetics</i> , 2019, 104, 422-438.	2.6	27

#	ARTICLE	IF	CITATIONS
3639	Investigation of the molecular signatures of selection on ATP synthase genes in the marine bivalve <i>Limecola balthica</i> . <i>Aquatic Living Resources</i> , 2019, 32, 3.	0.5	3
3640	Novel mutations of AXIN2 identified in a Chinese Congenital Heart Disease Cohort. <i>Journal of Human Genetics</i> , 2019, 64, 427-435.	1.1	14
3641	Effect of single amino acid mutations on C-terminal domain of breast cancer susceptible protein 1. <i>International Journal of Bioinformatics Research and Applications</i> , 2019, 15, 305.	0.1	0
3642	Performance Evaluation of in Silico Predictors for the Classification of ClinVar Variants. , 2019, , .		0
3643	Exome Sequencing Identifies Susceptibility Loci for Sarcoidosis Prognosis. <i>Frontiers in Immunology</i> , 2019, 10, 2964.	2.2	13
3644	Distinct clinical and biological implications of CUX1 in myeloid neoplasms. <i>Blood Advances</i> , 2019, 3, 2164-2178.	2.5	26
3645	Pediatric ALL relapses after allo-SCT show high individuality, clonal dynamics, selective pressure, and druggable targets. <i>Blood Advances</i> , 2019, 3, 3143-3156.	2.5	4
3646	Advances in identifying coding variants of common complex diseases. <i>Journal of Bio-X Research</i> , 2019, 2, 153-158.	0.3	0
3647	GTX.Digest.VCF: an online NGS data interpretation system based on intelligent gene ranking and large-scale text mining. <i>BMC Medical Genomics</i> , 2019, 12, 193.	0.7	3
3648	Chromatic Full-Field Stimulus Threshold and Pupillography as Functional Markers for Late-Stage, Early-Onset Retinitis Pigmentosa Caused by <i>CRB1</i> Mutations. <i>Translational Vision Science and Technology</i> , 2019, 8, 45.	1.1	13
3649	Whole exome sequencing reveals a de novo missense variant in <i>EEF1A2</i> in a Rett syndrome-like patient. <i>Clinical Case Reports (discontinued)</i> , 2019, 7, 2476-2482.	0.2	8
3650	Genetics of Congenital Isolated TSH Deficiency: Mutation Screening of the Known Causative Genes and a Literature Review. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 6229-6237.	1.8	15
3651	Application of Computational Biology and Artificial Intelligence Technologies in Cancer Precision Drug Discovery. <i>BioMed Research International</i> , 2019, 2019, 1-15.	0.9	42
3652	An epistasis between dopaminergic and oxytocinergic systems confers risk of post-traumatic stress disorder in a traumatized Chinese cohort. <i>Scientific Reports</i> , 2019, 9, 19252.	1.6	8
3653	Adult Diagnosis of Type 1 Fiber Predominance Myopathy Caused by Novel Mutations in the RYR1 Gene. <i>Journal of Clinical Neuromuscular Disease</i> , 2019, 20, 214-216.	0.3	4
3654	Fine mapping of genomic regions associated with female fertility in Nellore beef cattle based on sequence variants from segregating sires. <i>Journal of Animal Science and Biotechnology</i> , 2019, 10, 97.	2.1	7
3655	Whole-exome sequencing in multiplex preeclampsia families identifies novel candidate susceptibility genes. <i>Journal of Hypertension</i> , 2019, 37, 997-1011.	0.3	19
3656	Ataluren for the Treatment of Usher Syndrome 2A Caused by Nonsense Mutations. <i>International Journal of Molecular Sciences</i> , 2019, 20, 6274.	1.8	30

#	ARTICLE	IF	CITATIONS
3657	A Cohesin Subunit Variant Identified from a Peripheral Sclerocornea Pedigree. <i>Disease Markers</i> , 2019, 2019, 1-8.	0.6	6
3658	Phylogenetic Analysis of Bird-Virulent West Nile Virus Strain, Greece. <i>Emerging Infectious Diseases</i> , 2019, 25, 2323-2325.	2.0	11
3659	The intrinsically disordered C terminus of troponin T binds to troponin C to modulate myocardial force generation. <i>Journal of Biological Chemistry</i> , 2019, 294, 20054-20069.	1.6	23
3660	Novel SAMD9 Mutation in a Patient With Immunodeficiency, Neutropenia, Impaired Anti-CMV Response, and Severe Gastrointestinal Involvement. <i>Frontiers in Immunology</i> , 2019, 10, 2194.	2.2	12
3661	VarSight: prioritizing clinically reported variants with binary classification algorithms. <i>BMC Bioinformatics</i> , 2019, 20, 496.	1.2	14
3662	Genome-wide association analysis reveals QTL and candidate mutations involved in white spotting in cattle. <i>Genetics Selection Evolution</i> , 2019, 51, 62.	1.2	23
3663	Children with vesico-ureteric reflux have joint hypermobility and occasional TNXB sequence variants. <i>Canadian Urological Association Journal</i> , 2019, 14, E128-E136.	0.3	1
3664	VPOT: A Customizable Variant Prioritization Ordering Tool for Annotated Variants. <i>Genomics, Proteomics and Bioinformatics</i> , 2019, 17, 540-545.	3.0	10
3665	Somatic mutation signatures in primary liver tumors of workers exposed to ionizing radiation. <i>Scientific Reports</i> , 2019, 9, 18199.	1.6	8
3666	Novel variant in HDAC8 gene resulting in the severe Cornelia de Lange phenotype. <i>Clinical Dysmorphology</i> , 2019, 28, 124-128.	0.1	6
3667	In-silico investigation of coding variants potentially affecting the functioning of the glutamatergic N-methyl-D-aspartate receptor in schizophrenia. <i>Psychiatric Genetics</i> , 2019, 29, 44-50.	0.6	12
3668	Rare genetic variants in patients with cervical artery dissection. <i>European Stroke Journal</i> , 2019, 4, 355-362.	2.7	26
3669	Genetic variations in olfactory receptor gene OR2AG2 in a large multigenerational family with asthma. <i>Scientific Reports</i> , 2019, 9, 19029.	1.6	12
3670	Detection of Pathogenic Germline Variants Among Patients With Advanced Colorectal Cancer Undergoing Tumor Genomic Profiling for Precision Medicine. <i>Diseases of the Colon and Rectum</i> , 2019, 62, 429-437.	0.7	21
3671	Next Generation Sequencing Identifies Five Novel Mutations in Lebanese Patients with Bardet-Biedl and Usher Syndromes. <i>Genes</i> , 2019, 10, 1047.	1.0	6
3672	Recent genetic and functional insights in autism spectrum disorder. <i>Current Opinion in Neurology</i> , 2019, 32, 627-634.	1.8	7
3673	Corneal Perforation After Corneal Cross-Linking in Keratoconus Associated With Potentially Pathogenic ZNF469 Mutations. <i>Cornea</i> , 2019, 38, 1033-1039.	0.9	13
3674	Case report of two siblings with type 2A von Willebrand disease involving a novel mutation within the calcium-binding site of the A2 domain of von Willebrand factor. <i>Blood Coagulation and Fibrinolysis</i> , 2019, 30, 161-167.	0.5	0

#	ARTICLE	IF	CITATIONS
3675	Results of multigene panel testing in familial cancer cases without genetic cause demonstrated by single gene testing. <i>Scientific Reports</i> , 2019, 9, 18555.	1.6	13
3676	Monozygotic Twins Concordant for Common Variable Immunodeficiency: Strikingly Similar Clinical and Immune Profile Associated With a Polygenic Burden. <i>Frontiers in Immunology</i> , 2019, 10, 2503.	2.2	5
3677	Non-Redundant and Overlapping Oncogenic Readouts of Non-Canonical and Novel Colorectal Cancer KRAS and NRAS Mutants. <i>Cells</i> , 2019, 8, 1557.	1.8	7
3678	Mitochondrial DNA Variation of Leber's Hereditary Optic Neuropathy in Western Siberia. <i>Cells</i> , 2019, 8, 1574.	1.8	12
3679	Using mechanistic models for the clinical interpretation of complex genomic variation. <i>Scientific Reports</i> , 2019, 9, 18937.	1.6	20
3680	Genomic analysis of a spinal muscular atrophy (SMA) discordant family identifies a novel mutation in TLL2, an activator of growth differentiation factor 8 (myostatin): a case report. <i>BMC Medical Genetics</i> , 2019, 20, 204.	2.1	8
3681	Atypical <i>RAS</i> Mutations in Metastatic Colorectal Cancer. <i>JCO Precision Oncology</i> , 2019, 3, 1-11.	1.5	1
3682	Integrating hypertension phenotype and genotype with hybrid non-negative matrix factorization. <i>Bioinformatics</i> , 2019, 35, 1395-1403.	1.8	12
3683	MED12 somatic mutations encompassing exon 2 associated with benign breast fibroadenomas and not breast carcinoma in Indian women. <i>Journal of Cellular Biochemistry</i> , 2019, 120, 182-191.	1.2	9
3684	Variations in maternal adenylate cyclase genes are associated with congenital Zika syndrome in a cohort from Northeast, Brazil. <i>Journal of Internal Medicine</i> , 2019, 285, 215-222.	2.7	18
3685	Systematic re-evaluation of <i>SCN5A</i> variants associated with Brugada syndrome. <i>Journal of Cardiovascular Electrophysiology</i> , 2019, 30, 118-127.	0.8	36
3686	Novel CFAP43 and CFAP44 mutations cause male infertility with multiple morphological abnormalities of the sperm flagella (MMAF). <i>Reproductive BioMedicine Online</i> , 2019, 38, 769-778.	1.1	26
3687	Non-leukemic pediatric mixed phenotype acute leukemia/lymphoma: Genomic characterization and clinical outcome in a prospective trial for pediatric lymphoblastic lymphoma. <i>Genes Chromosomes and Cancer</i> , 2019, 58, 365-372.	1.5	6
3688	Investigation of deleterious effects of nsSNPs in the <i>POT1</i> gene: a structural genomics-based approach to understand the mechanism of cancer development. <i>Journal of Cellular Biochemistry</i> , 2019, 120, 10281-10294.	1.2	32
3689	Defective repair capacity of variant proteins of the DNA glycosylase NTHL1 for 5-hydroxyuracil, an oxidation product of cytosine. <i>Free Radical Biology and Medicine</i> , 2019, 131, 264-273.	1.3	11
3690	Combined Targeted Resequencing of Cytosine DNA Methylation and Mutations of DNA Repair Genes with Potential Use for Poly(ADP-Ribose) Polymerase 1 Inhibitor Sensitivity Testing. <i>Journal of Molecular Diagnostics</i> , 2019, 21, 198-213.	1.2	2
3691	Systematic prediction of familial hypercholesterolemia caused by low-density lipoprotein receptor missense mutations. <i>Atherosclerosis</i> , 2019, 281, 1-8.	0.4	10
3692	Common genetic variants contribute to incomplete penetrance: evidence from cancer-free BRCA1 mutation carriers. <i>European Journal of Cancer</i> , 2019, 107, 68-78.	1.3	10

#	ARTICLE	IF	CITATIONS
3693	A novel mutation of <i>PANK4</i> causes autosomal dominant congenital posterior cataract. <i>Human Mutation</i> , 2019, 40, 380-391.	1.1	12
3694	Noncoding rare variants of <i>TBX6</i> in congenital anomalies of the kidney and urinary tract. <i>Molecular Genetics and Genomics</i> , 2019, 294, 493-500.	1.0	8
3695	<i>CRB2</i> mutation causes autosomal recessive retinitis pigmentosa. <i>Experimental Eye Research</i> , 2019, 180, 164-173.	1.2	20
3696	Mutational Analysis of 472 Urothelial Carcinoma Across Grades and Anatomic Sites. <i>Clinical Cancer Research</i> , 2019, 25, 2458-2470.	3.2	102
3697	Whole-exome sequencing reveals <i>SALL4</i> variants in premature ovarian insufficiency: an update on genotype-phenotype correlations. <i>Human Genetics</i> , 2019, 138, 83-92.	1.8	27
3698	Spectrum of <i>SLC20A2</i> , <i>PDGFRB</i> , <i>PDGFB</i> , and <i>XPR1</i> mutations in a large cohort of patients with primary familial brain calcification. <i>Human Mutation</i> , 2019, 40, 392-403.	1.1	26
3699	Variants in the <i>PSCA</i> gene associated with risk of cancer and nonneoplastic diseases: systematic research synopsis, meta-analysis and epidemiological evidence. <i>Carcinogenesis</i> , 2019, 40, 70-83.	1.3	7
3700	Mutations in <i>PERP</i> Cause Dominant and Recessive Keratoderma. <i>Journal of Investigative Dermatology</i> , 2019, 139, 380-390.	0.3	17
3701	The Burden of Candidate Pathogenic Variants for Kidney and Genitourinary Disorders Emerging From Exome Sequencing. <i>Annals of Internal Medicine</i> , 2019, 170, 11.	2.0	60
3702	Novel homozygous <i>CFAP69</i> mutations in humans and mice cause severe asthenoteratospermia with multiple morphological abnormalities of the sperm flagella. <i>Journal of Medical Genetics</i> , 2019, 56, 96-103.	1.5	70
3703	Association of prolactin receptor (<i>PRLR</i>) variants with prolactinomas. <i>Human Molecular Genetics</i> , 2019, 28, 1023-1037.	1.4	24
3704	Mutation analysis and pathogenicity identification of Mucopolysaccharidosis type IVA in 8 south China families. <i>Gene</i> , 2019, 686, 261-269.	1.0	3
3705	Limb Girdle Muscular Dystrophy due to Digenic Inheritance of <i>DES</i> and <i>CAPN3</i> Mutations. <i>Case Reports in Neurology</i> , 2019, 10, 272-278.	0.3	8
3706	<i>Complement C7</i> is a novel risk gene for Alzheimer's disease in Han Chinese. <i>National Science Review</i> , 2019, 6, 257-274.	4.6	55
3707	Severe hypertriglyceridemia is primarily polygenic. <i>Journal of Clinical Lipidology</i> , 2019, 13, 80-88.	0.6	136
3708	A <i>de Novo EDA</i> -Variant in a Litter of Shorthaired Standard Dachshunds with X-Linked Hypohidrotic Ectodermal Dysplasia. <i>C3: Genes, Genomes, Genetics</i> , 2019, 9, 95-104.	0.8	7
3709	Genomic analyses in African populations identify novel risk loci for cleft palate. <i>Human Molecular Genetics</i> , 2019, 28, 1038-1051.	1.4	61
3710	A novel truncating variant of <i>GLI2</i> associated with Culler-Jones syndrome impairs Hedgehog signalling. <i>PLoS ONE</i> , 2019, 14, e0210097.	1.1	9

#	ARTICLE	IF	CITATIONS
3711	A study in scarlet: MC1R as the main predictor of red hair and exemplar of the flip-flop effect. <i>Human Molecular Genetics</i> , 2019, 28, 2093-2106.	1.4	11
3712	Recurrent somatic BRAF insertion (p.V504_R506dup): a tumor marker and a potential therapeutic target in pilocytic astrocytoma. <i>Oncogene</i> , 2019, 38, 2994-3002.	2.6	13
3713	Somatic Tumor Variant Filtration Strategies to Optimize Tumor-Only Molecular Profiling Using Targeted Next-Generation Sequencing Panels. <i>Journal of Molecular Diagnostics</i> , 2019, 21, 261-273.	1.2	36
3714	Genomic landscape of synchronous tubulovillous adenoma and multiple non-familial colon cancers from a single patient. <i>Cancer Genetics</i> , 2019, 231-232, 54-61.	0.2	1
3715	Characterization of Tumor-Suppressor Gene Inactivation Events in 33 Cancer Types. <i>Cell Reports</i> , 2019, 26, 496-506.e3.	2.9	21
3716	Biallelic sequence variants in INTS1 in patients with developmental delays, cataracts, and craniofacial anomalies. <i>European Journal of Human Genetics</i> , 2019, 27, 582-593.	1.4	23
3717	Proteome Analysis of Human Neutrophil Granulocytes From Patients With Monogenic Disease Using Data-independent Acquisition. <i>Molecular and Cellular Proteomics</i> , 2019, 18, 760-772.	2.5	52
3718	Finding the "hot spot": Are predictors of binding affinity changes upon mutations in protein-protein interactions ready for it?. <i>Wiley Interdisciplinary Reviews: Computational Molecular Science</i> , 2019, 9, e1410.	6.2	86
3720	Mutation analysis by deep sequencing of pancreatic juice from patients with pancreatic ductal adenocarcinoma. <i>BMC Cancer</i> , 2019, 19, 11.	1.1	18
3721	Loss-of-function mutations in caspase recruitment domain-containing protein 14 (CARD14) are associated with a severe variant of atopic dermatitis. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 173-181.e10.	1.5	60
3722	Structural and Functional Biology of Aldo-Keto Reductase Steroid-Transforming Enzymes. <i>Endocrine Reviews</i> , 2019, 40, 447-475.	8.9	73
3723	Exome sequencing in families with severe mental illness identifies novel and rare variants in genes implicated in Mendelian neuropsychiatric syndromes. <i>Psychiatry and Clinical Neurosciences</i> , 2019, 73, 11-19.	1.0	31
3724	Genetic associations of T cell cancer immune response with tumor aggressiveness in localized prostate cancer patients and disease reclassification in an active surveillance cohort. <i>OncImmunity</i> , 2019, 8, e1483303.	2.1	7
3725	Sarcomere variants in arrhythmogenic cardiomyopathy: Pathogenic factor or bystander?. <i>Gene</i> , 2019, 687, 82-89.	1.0	7
3726	The regulatory role of Toll-like receptors after ischemic stroke: neurosteroids as TLR modulators with the focus on TLR2/4. <i>Cellular and Molecular Life Sciences</i> , 2019, 76, 523-537.	2.4	50
3727	KDF1 is a novel candidate gene of non-syndromic tooth agenesis. <i>Archives of Oral Biology</i> , 2019, 97, 131-136.	0.8	17
3728	Gene-specific metrics to facilitate identification of disease genes for molecular diagnosis in patient genomes: a systematic review. <i>Briefings in Functional Genomics</i> , 2019, 18, 23-29.	1.3	6
3729	Ensembl 2019. <i>Nucleic Acids Research</i> , 2019, 47, D745-D751.	6.5	879

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3730	Next Generation Sequencing Analysis in Early Onset Dementia Patients. <i>Journal of Alzheimer's Disease</i> , 2019, 67, 243-256.	1.2	29
3731	Prenatal Diagnosis by Whole Exome Sequencing in Fetuses with Ultrasound Abnormalities. <i>Methods in Molecular Biology</i> , 2019, 1885, 267-285.	0.4	4
3732	Efficient region-based test strategy uncovers genetic risk factors for functional outcome in bipolar disorder. <i>European Neuropsychopharmacology</i> , 2019, 29, 156-170.	0.3	7
3733	Bioinformatics Tools in Clinical Genomics. , 2019, , 163-182.		0
3734	Acquisition of deleterious mutations during potato polyploidization. <i>Journal of Integrative Plant Biology</i> , 2019, 61, 7-11.	4.1	30
3736	Association of single nucleotide polymorphism c.673C>A/p.Gln225Lys in <i>SEPT12</i> gene with spermatogenesis failure in male idiopathic infertility in Northeast China. <i>Journal of International Medical Research</i> , 2019, 47, 992-998.	0.4	4
3737	Image feature extraction algorithm based on bi-dimensional local mean decomposition. <i>Optical Review</i> , 2019, 26, 43-64.	1.2	5
3738	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. <i>Human Mutation</i> , 2019, 40, 53-72.	1.1	48
3739	Bone matrix hypermineralization associated with low bone turnover in a case of Nasu-Hakola disease. <i>Bone</i> , 2019, 123, 48-55.	1.4	7
3740	DNA sequences and predicted protein structures of <i>prot6E</i> and <i>sefA</i> genes for <i>Salmonella</i> ser. Enteritidis detection. <i>Food Control</i> , 2019, 96, 271-280.	2.8	3
3741	A review study: Computational techniques for expecting the impact of non-synonymous single nucleotide variants in human diseases. <i>Gene</i> , 2019, 680, 20-33.	1.0	47
3742	A cancer associated somatic mutation in LC3B attenuates its binding to E1-like ATG7 protein and subsequent lipidation. <i>Autophagy</i> , 2019, 15, 438-452.	4.3	7
3743	Hypomorphic caspase activation and recruitment domain 11 (CARD11) mutations associated with diverse immunologic phenotypes with or without atopic disease. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 1482-1495.	1.5	116
3744	A weighted burden test using logistic regression for integrated analysis of sequence variants, copy number variants and polygenic risk score. <i>European Journal of Human Genetics</i> , 2019, 27, 114-124.	1.4	24
3745	KAT6A Syndrome: genotype-phenotype correlation in 76 patients with pathogenic KAT6A variants. <i>Genetics in Medicine</i> , 2019, 21, 850-860.	1.1	68
3746	Understanding the disease genome: gene essentiality and the interplay of selection, recombination and mutation. <i>Briefings in Bioinformatics</i> , 2019, 20, 267-273.	3.2	11
3747	Frequently used bioinformatics tools overestimate the damaging effect of allelic variants. <i>Genes and Immunity</i> , 2019, 20, 10-22.	2.2	12
3748	Image multi-target detection and segmentation algorithm based on regional proposed fast intelligent network. <i>Cluster Computing</i> , 2019, 22, 3385-3393.	3.5	1

#	ARTICLE	IF	CITATIONS
3749	Germline CDH1 mutations are a significant contributor to the high frequency of early-onset diffuse gastric cancer cases in New Zealand Māori. <i>Familial Cancer</i> , 2019, 18, 83-90.	0.9	33
3750	New GJA8 variants and phenotypes highlight its critical role in a broad spectrum of eye anomalies. <i>Human Genetics</i> , 2019, 138, 1027-1042.	1.8	38
3751	Computational Analysis of High-Risk SNPs in Human DBY Gene Responsible for Male Infertility: A Functional and Structural Impact. <i>Interdisciplinary Sciences, Computational Life Sciences</i> , 2019, 11, 412-427.	2.2	3
3752	Finding cancer driver mutations in the era of big data research. <i>Biophysical Reviews</i> , 2019, 11, 21-29.	1.5	20
3753	An autism-linked missense mutation in SHANK3 reveals the modularity of Shank3 function. <i>Molecular Psychiatry</i> , 2020, 25, 2534-2555.	4.1	61
3754	Genome-wide significant regions in 43 Utah high-risk families implicate multiple genes involved in risk for completed suicide. <i>Molecular Psychiatry</i> , 2020, 25, 3077-3090.	4.1	40
3755	Exome sequencing reveals a novel splice site variant in HUWE1 gene in patients with suspected Say-Meyer syndrome. <i>European Journal of Medical Genetics</i> , 2020, 63, 103635.	0.7	14
3756	A genome-wide association study of tramadol metabolism from post-mortem samples. <i>Pharmacogenomics Journal</i> , 2020, 20, 94-103.	0.9	3
3757	Impact of Gln94Glu mutation on the structure and function of protection of telomere 1, a cause of cutaneous familial melanoma. <i>Journal of Biomolecular Structure and Dynamics</i> , 2020, 38, 1514-1524.	2.0	29
3758	Identification of novel loci associated with infant cognitive ability. <i>Molecular Psychiatry</i> , 2020, 25, 3010-3019.	4.1	6
3759	Genetic aetiology of early infant deaths in a neonatal intensive care unit. <i>Journal of Medical Genetics</i> , 2020, 57, 169-177.	1.5	22
3760	Diagnostic interpretation of genetic studies in patients with primary immunodeficiency diseases: A working group report of the Primary Immunodeficiency Diseases Committee of the American Academy of Allergy, Asthma & Immunology. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 145, 46-69.	1.5	54
3761	The synergy between human factors and risk attitudes of Malaysian contractorsâ€™: Moderating effect of government policy. <i>Safety Science</i> , 2020, 121, 331-347.	2.6	22
3762	Multi-omics study in monozygotic twins confirm the contribution of de novo mutation to psoriasis. <i>Journal of Autoimmunity</i> , 2020, 106, 102349.	3.0	13
3763	Mutations in MTHFR and POLG impaired activity of the mitochondrial respiratory chain in 46-year-old twins with spastic paraparesis. <i>Journal of Human Genetics</i> , 2020, 65, 91-98.	1.1	5
3764	A Balanced Translocation in Kallmann Syndrome Implicates a Long Noncoding RNA, RMST, as a GnRH Neuronal Regulator. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, e231-e244.	1.8	28
3765	In silico analysis of nonsynonymous single nucleotide polymorphisms (nsSNPs) of the <i>SMPX</i> gene. <i>Annals of Human Genetics</i> , 2020, 84, 54-71.	0.3	20
3766	A Rare Mutation in <i>SMAD9</i> Associated With High Bone Mass Identifies the SMAD-Dependent BMP Signaling Pathway as a Potential Anabolic Target for Osteoporosis. <i>Journal of Bone and Mineral Research</i> , 2020, 35, 92-105.	3.1	34

#	ARTICLE	IF	CITATIONS
3767	A genome-first approach to aggregating rare genetic variants in LMNA for association with electronic health record phenotypes. <i>Genetics in Medicine</i> , 2020, 22, 102-111.	1.1	42
3768	A linkage and exome study implicates rare variants of KANK4 and CAP2 in bipolar disorder in a multiplex family. <i>Bipolar Disorders</i> , 2020, 22, 70-78.	1.1	6
3769	Frequent and Persistent PLCG1 Mutations in SÄ©zary Cells Directly Enhance PLCÎ³1 Activity and Stimulate NFÎ±B, AP-1, and NFAT Signaling. <i>Journal of Investigative Dermatology</i> , 2020, 140, 380-389.e4.	0.3	25
3770	Diagnostic Approach to Monogenic Inflammatory Bowel Disease in Clinical Practice: A Ten-Year Multicentric Experience. <i>Inflammatory Bowel Diseases</i> , 2020, 26, 720-727.	0.9	32
3771	Bayesian estimation of genetic regulatory effects in high-throughput reporter assays. <i>Bioinformatics</i> , 2020, 36, 331-338.	1.8	0
3772	<i>RPE</i> mutation frequency and phenotypic variation according to exome sequencing in a tertiary centre for genetic eye diseases in China. <i>Acta Ophthalmologica</i> , 2020, 98, e181-e190.	0.6	21
3773	Next generation sequencing panel in undifferentiated autoinflammatory diseases identifies patients with colchicine-responder recurrent fevers. <i>Rheumatology</i> , 2020, 59, 344-360.	0.9	36
3774	Genomic profiling of primary histiocytic sarcoma reveals two molecular subgroups. <i>Haematologica</i> , 2020, 105, 951-960.	1.7	53
3775	A Novel Homozygous Missense Variant in the NAGA Gene with Extreme Intrafamilial Phenotypic Heterogeneity. <i>Journal of Molecular Neuroscience</i> , 2020, 70, 45-55.	1.1	2
3776	Whole exome sequencing identifies a novel SCN1A mutation in genetic (idiopathic) generalized epilepsy and juvenile myoclonic epilepsy subtypes. <i>Neurological Sciences</i> , 2020, 41, 591-598.	0.9	6
3777	The association of the UHRF1BP1 gene with systemic lupus erythematosus was replicated in a Han Chinese population from mainland China. <i>Annals of Human Genetics</i> , 2020, 84, 221-228.	0.3	2
3778	<i>TJP2</i> hepatobiliary disorders: Novel variants and clinical diversity. <i>Human Mutation</i> , 2020, 41, 502-511.	1.1	25
3779	Resolving the genetic paradox of invasions: Preadapted genomes and postintroduction hybridization of bigheaded carps in the Mississippi River Basin. <i>Evolutionary Applications</i> , 2020, 13, 263-277.	1.5	20
3780	Multicenter study of the clinical features and mutation gene spectrum of Chinese children with Dent disease. <i>Clinical Genetics</i> , 2020, 97, 407-417.	1.0	19
3781	Mutations and Response to Rapalogs in Patients with Metastatic Renal Cell Carcinoma. <i>Molecular Cancer Therapeutics</i> , 2020, 19, 690-696.	1.9	11
3782	Matching whole genomes to rare genetic disorders: Identification of potential causative variants using phenotypeâ€weighted knowledge in the CAGI SickKids5 clinical genomes challenge. <i>Human Mutation</i> , 2020, 41, 347-362.	1.1	4
3783	Application of targeted exome and wholeâ€exome sequencing for Chinese families with Stargardt disease. <i>Annals of Human Genetics</i> , 2020, 84, 177-184.	0.3	4
3784	Whole exome sequencing identified ATP6V1C2 as a novel candidate gene for recessive distal renal tubular acidosis. <i>Kidney International</i> , 2020, 97, 567-579.	2.6	42

#	ARTICLE	IF	CITATIONS
3785	Most frequent mutational events of home box 13 gene in prostatic adenocarcinoma and correlation with tumor characteristics. <i>Meta Gene</i> , 2020, 23, 100637.	0.3	2
3786	Cyclin D1 immunohistochemical expression and somatic mutations in canine oral melanoma. <i>Veterinary and Comparative Oncology</i> , 2020, 18, 231-238.	0.8	6
3787	Rare variants in SLC6A4 cause susceptibility to major depressive disorder with suicidal ideation in Han Chinese adolescents and young adults. <i>Gene</i> , 2020, 726, 144147.	1.0	5
3788	Compound and heterozygous mutations of KCNQ1 in long QT syndrome with familial history of unexplained sudden death: Identified by analysis of whole exome sequencing and predisposing genes. <i>Annals of Noninvasive Electrocardiology</i> , 2020, 25, e12694.	0.5	2
3789	Multiple genetic mutations implicate spectrum of phenotypes in Bardet-Biedl syndrome. <i>Gene</i> , 2020, 725, 144164.	1.0	3
3790	Assessing genomic diversity and signatures of selection in Original Braunvieh cattle using whole-genome sequencing data. <i>BMC Genomics</i> , 2020, 21, 27.	1.2	47
3791	Clinical, cytogenetic and molecular genetic characterization of a tandem fusion translocation in a male Holstein cattle with congenital hypospadias and a ventricular septal defect. <i>PLoS ONE</i> , 2020, 15, e0227117.	1.1	11
3792	Biallelic variants in KYN1 cause a multisystemic syndrome with hand hyperphalangism. <i>Bone</i> , 2020, 133, 115219.	1.4	12
3793	Impact of proactive high-throughput functional assay data on BRCA1 variant interpretation in 3684 patients with breast or ovarian cancer. <i>Journal of Human Genetics</i> , 2020, 65, 209-220.	1.1	6
3794	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020, 52, 56-73.	9.4	120
3795	Alterations of Nedd4 ² binding capacity in PY ² motif of Na ^v 1.5 channel underlie long QT syndrome and Brugada syndrome. <i>Acta Physiologica</i> , 2020, 229, e13438.	1.8	9
3796	Targeted Next-Generation Sequencing in Patients with Suggestive X-Linked Intellectual Disability. <i>Genes</i> , 2020, 11, 51.	1.0	15
3797	Clinical and Genetic Analysis of a European Cohort with Pericentral Retinitis Pigmentosa. <i>International Journal of Molecular Sciences</i> , 2020, 21, 86.	1.8	25
3798	Pathogenicity Reclassification of RPE65 Missense Variants Related to Leber Congenital Amaurosis and Early-Onset Retinal Dystrophy. <i>Genes</i> , 2020, 11, 24.	1.0	14
3799	Identification of altered biological processes in heterogeneous RNA-sequencing data by discretization of expression profiles. <i>Nucleic Acids Research</i> , 2020, 48, 1730-1747.	6.5	8
3800	A novel de novo dominant mutation of <i>NOTCH1</i> gene in an Iranian family with non-syndromic congenital heart disease. <i>Journal of Clinical Laboratory Analysis</i> , 2020, 34, e23147.	0.9	13
3801	When moments matter: Finding answers with rapid exome sequencing. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1027.	0.6	12
3802	A novel heterozygous STAT5B variant in a patient with short stature and partial growth hormone insensitivity (GHI). <i>Growth Hormone and IGF Research</i> , 2020, 50, 61-70.	0.5	7

#	ARTICLE	IF	CITATIONS
3803	Sorting Variants of Unknown Significance Identified by Whole Exome Sequencing: Genetic and Laboratory Investigations of Two Novel <i>MCT8</i> Variants. <i>Thyroid</i> , 2020, 30, 463-465.	2.4	4
3804	Identification of Novel Loci and New Risk Variant in Known Loci for Colorectal Cancer Risk in East Asians. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2020, 29, 477-486.	1.1	25
3805	Analysis of spinal muscular atrophy-like patients by targeted resequencing. <i>Brain and Development</i> , 2020, 42, 148-156.	0.6	8
3806	Extensive genic and allelic heterogeneity underlying inherited retinal dystrophies in Mexican patients molecularly analyzed by next-generation sequencing. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, .	0.6	30
3807	Unraveling the genetic cause of hereditary ophthalmic disorders in Arab societies from Israel and the Palestinian Authority. <i>European Journal of Human Genetics</i> , 2020, 28, 742-753.	1.4	1
3808	DROIDS 3.0 – Detecting Genetic and Drug Class Variant Impact on Conserved Protein Binding Dynamics. <i>Biophysical Journal</i> , 2020, 118, 541-551.	0.2	18
3809	ClinVAP: a reporting strategy from variants to therapeutic options. <i>Bioinformatics</i> , 2020, 36, 2316-2317.	1.8	4
3810	Interpretation of somatic <i>POLE</i> mutations in endometrial carcinoma. <i>Journal of Pathology</i> , 2020, 250, 323-335.	2.1	203
3811	Clinicopathological and molecular characterisation of a multiple classifier™ endometrial carcinomas. <i>Journal of Pathology</i> , 2020, 250, 312-322.	2.1	205
3812	High prevalence of mutations in perilipin 1 in patients with precocious acute coronary syndrome. <i>Atherosclerosis</i> , 2020, 293, 86-91.	0.4	2
3813	A recurrent missense variant in <i>HARS2</i> results in variable sensorineural hearing loss in three unrelated families. <i>Journal of Human Genetics</i> , 2020, 65, 305-311.	1.1	5
3814	The Laboratory Domestication of Zebrafish: From Diverse Populations to Inbred Substrains. <i>Molecular Biology and Evolution</i> , 2020, 37, 1056-1069.	3.5	30
3815	Ribosomal protein gene <i>RPL9</i> variants can differentially impair ribosome function and cellular metabolism. <i>Nucleic Acids Research</i> , 2020, 48, 770-787.	6.5	28
3816	Novel <i>ADGRG2</i> truncating variants in patients with X-linked congenital absence of vas deferens. <i>Andrology</i> , 2020, 8, 618-624.	1.9	20
3817	PAK3 mutations responsible for severe intellectual disability and callosal agenesis inhibit cell migration. <i>Neurobiology of Disease</i> , 2020, 136, 104709.	2.1	14
3818	A model combining clinical and genomic factors to predict response to PD-1/PD-L1 blockade in advanced urothelial carcinoma. <i>British Journal of Cancer</i> , 2020, 122, 555-563.	2.9	59
3819	Deficiencies in vesicular transport mediated by <i>TRAPPC4</i> are associated with severe syndromic intellectual disability. <i>Brain</i> , 2020, 143, 112-130.	3.7	33
3820	High-throughput custom capture sequencing identifies novel mutations in coloboma-associated genes: Mutation in DNA-binding domain of retinoic acid receptor beta affects nuclear localization causing ocular coloboma. <i>Human Mutation</i> , 2020, 41, 678-695.	1.1	13

#	ARTICLE	IF	CITATIONS
3821	Low folate concentration impacts mismatch repair deficiency in neural tube defects. <i>Epigenomics</i> , 2020, 12, 5-18.	1.0	10
3822	Whole Exome Sequencing Identifies Candidate Genes Associated with Hereditary Predisposition to Uveal Melanoma. <i>Ophthalmology</i> , 2020, 127, 668-678.	2.5	27
3823	Genetic Susceptibility to Hepatic Sinusoidal Obstruction Syndrome in Pediatric Patients Undergoing Hematopoietic Stem Cell Transplantation. <i>Biology of Blood and Marrow Transplantation</i> , 2020, 26, 920-927.	2.0	11
3824	Six2 regulates Pax9 expression, palatogenesis and craniofacial bone formation. <i>Developmental Biology</i> , 2020, 458, 246-256.	0.9	13
3825	The relevance of tumor mutation profiling in interpretation of NGS data from cell-free DNA in non-small cell lung cancer patients. <i>Experimental and Molecular Pathology</i> , 2020, 112, 104347.	0.9	5
3826	Genomic divergences between the two polyphagous <i>Spodoptera</i> relatives provide cues for successful invasion of the fall armyworm. <i>Insect Science</i> , 2020, 27, 1257-1265.	1.5	8
3827	Genetic landscape of adult Langerhans cell histiocytosis with lung involvement. <i>European Respiratory Journal</i> , 2020, 55, 1901190.	3.1	38
3828	Germline MUTYH Mutation in a Pediatric Cancer Survivor Developing a Secondary Malignancy. <i>Journal of Pediatric Hematology/Oncology</i> , 2020, 42, e647-e654.	0.3	2
3829	Identification of pathogenic variant enriched regions across genes and gene families. <i>Genome Research</i> , 2020, 30, 62-71.	2.4	47
3830	TCTEX1D1 is a genetic modifier of disease progression in Duchenne muscular dystrophy. <i>European Journal of Human Genetics</i> , 2020, 28, 815-825.	1.4	36
3831	Functional phenotype variations of two novel K _V 7.1 mutations identified in patients with Long QT syndrome. <i>PACE - Pacing and Clinical Electrophysiology</i> , 2020, 43, 210-216.	0.5	4
3832	InMeRF: prediction of pathogenicity of missense variants by individual modeling for each amino acid substitution. <i>NAR Genomics and Bioinformatics</i> , 2020, 2, lqaa038.	1.5	16
3833	Nearly Half of TP53 Germline Variants Predicted To Be Pathogenic in Patients With Osteosarcoma Are De Novo: A Report From the Children's Oncology Group. <i>JCO Precision Oncology</i> , 2020, 4, 1187-1195.	1.5	10
3834	Do damaging variants of SLC6A9, the gene for the glycine transporter 1 (GlyT-1), protect against schizophrenia?. <i>Psychiatric Genetics</i> , 2020, 30, 150-152.	0.6	1
3835	Ovarian Cancer Risk Variants Are Enriched in Histotype-Specific Enhancers and Disrupt Transcription Factor Binding Sites. <i>American Journal of Human Genetics</i> , 2020, 107, 622-635.	2.6	14
3836	Titration of bacterial growth and chemical biosynthesis for efficient N-acetylglucosamine and N-acetylneuraminic acid bioproduction. <i>Nature Communications</i> , 2020, 11, 5078.	5.8	33
3837	A computational and bioinformatic analysis of ACE2: an elucidation of its dual role in COVID-19 pathology and finding its associated partners as potential therapeutic targets. <i>Journal of Biomolecular Structure and Dynamics</i> , 2022, 40, 1813-1829.	2.0	16
3838	Confirmed effects of candidate variants for milk production, udder health, and udder morphology in dairy cattle. <i>Genetics Selection Evolution</i> , 2020, 52, 55.	1.2	32

#	ARTICLE	IF	CITATIONS
3839	Functional imaging of RAS pathway targeting in malignant peripheral nerve sheath tumor cells and xenografts. <i>Pediatric Blood and Cancer</i> , 2020, 67, e28639.	0.8	2
3840	Haplotype-resolved genome analyses of a heterozygous diploid potato. <i>Nature Genetics</i> , 2020, 52, 1018-1023.	9.4	134
3841	Constitutional variants in POT1, TERF2IP, and ACD genes in patients with melanoma in the Polish population. <i>European Journal of Cancer Prevention</i> , 2020, 29, 511-519.	0.6	3
3842	Variants in <i>RABL2A</i> causing male infertility and ciliopathy. <i>Human Molecular Genetics</i> , 2020, 29, 3402-3411.	1.4	11
3843	Clinical and Molecular Characterization of Fanconi Anemia Patients in Turkey. <i>Molecular Syndromology</i> , 2020, 11, 183-196.	0.3	6
3844	Analysis of Whole Genome Resequencing Datasets from a Worldwide Sample of Sheep Breeds to Identify Potential Causal Mutations Influencing Milk Composition Traits. <i>Animals</i> , 2020, 10, 1542.	1.0	7
3845	Data Analysis in Rare Disease Diagnostics. <i>Journal of the Indian Institute of Science</i> , 2020, 100, 733-751.	0.9	0
3846	Cancer Predisposition Genes in Cancer-Free Families. <i>Cancers</i> , 2020, 12, 2770.	1.7	2
3847	H258R mutation in <i>KCNAB3</i> gene in a family with genetic epilepsy and febrile seizures plus. <i>Brain and Behavior</i> , 2020, 10, e01859.	1.0	8
3848	<i>NKX2-2</i> Mutation Causes Congenital Diabetes and Infantile Obesity With Paradoxical Glucose-Induced Ghrelin Secretion. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 3486-3495.	1.8	11
3849	Essential role of autophagy in restricting poliovirus infection revealed by identification of an ATG7 defect in a poliomyelitis patient. <i>Autophagy</i> , 2021, 17, 2449-2464.	4.3	10
3850	<i>CRB1</i> -related retinopathy overlapping the ocular phenotype of S-adenosylhomocysteine hydrolase deficiency. <i>Ophthalmic Genetics</i> , 2020, 41, 457-464.	0.5	3
3851	Genomic diversity revealed by whole-genome sequencing in three Danish commercial pig breeds. <i>Journal of Animal Science</i> , 2020, 98, .	0.2	9
3852	Estimation of varicocele associated human ARG2 and NOS1 proteins and computational analysis on the effect of its nsSNPs. <i>International Journal of Biological Macromolecules</i> , 2020, 164, 735-747.	3.6	3
3853	Genetics of Gland-in-situ or Hypoplastic Congenital Hypothyroidism in Macedonia. <i>Frontiers in Endocrinology</i> , 2020, 11, 413.	1.5	13
3854	Understanding the impact of DIS3 cancer-associated mutations by in silico structure modeling. <i>Gene Reports</i> , 2020, 20, 100779.	0.4	0
3855	In silico analysis of likely pathogenic variants in human GGCX gene. <i>Informatics in Medicine Unlocked</i> , 2020, 19, 100337.	1.9	2
3856	Structure-guided discovery of pharmacological chaperones targeting protein conformational and misfolding diseases. , 2020, , 281-308.		1

#	ARTICLE	IF	CITATIONS
3857	Whole exome sequencing identified mutations causing hearing loss in five consanguineous Pakistani families. <i>BMC Medical Genetics</i> , 2020, 21, 151.	2.1	8
3858	TraPS-VarI: Identifying genetic variants altering phosphotyrosine based signalling motifs. <i>Scientific Reports</i> , 2020, 10, 8453.	1.6	1
3859	Association between rare variants in specific functional pathways and human neural tube defects multiple subphenotypes. <i>Neural Development</i> , 2020, 15, 8.	1.1	14
3860	A Pilot Study for the Feasibility of Exome-Sequencing in Circulating Tumor Cells Versus Single Metastatic Biopsies in Breast Cancer. <i>International Journal of Molecular Sciences</i> , 2020, 21, 4826.	1.8	7
3861	Heterozygous missense variant in EIF6 gene: A novel form of Shwachmanâ€“Diamond syndrome?. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2010-2020.	0.7	11
3862	COVID-19 vaccines: no time for complacency. <i>Lancet, The</i> , 2020, 396, 1607.	6.3	36
3864	Impact of laparoscopic approach on the short-term outcomes of elderly patients with colorectal cancer: A nationwide Italian experience. <i>European Journal of Surgical Oncology</i> , 2020, 46, e8.	0.5	0
3866	P.240 Investigation of the behavioural effects of paroxetine discontinuation in mice. <i>European Neuropsychopharmacology</i> , 2020, 40, S136-S137.	0.3	1
3867	Synthesis, characterization and structural systematics in diorganotin complexes with O,N,O'-tris-chelating semirigid diaza-scaffolds: Mono- vs. di-nuclear compounds. <i>Journal of Organometallic Chemistry</i> , 2020, 927, 121522.	0.8	11
3869	Morphology and molecular taxonomy of the tongue worm, genus <i>Raillietiella</i> (Pentastomida) from the lungs of berber skinks <i>Eumeces schneideri</i> (Scincidae): First report. <i>Revista Argentina De Microbiologia</i> , 2021, 53, 110-123.	0.4	1
3871	Cross-market selling channel strategies in an international luxury brand's supply chain with gray markets. <i>Transportation Research, Part E: Logistics and Transportation Review</i> , 2020, 144, 102157.	3.7	17
3872	An activating germline IDH1 variant associated with a tumor entity characterized by unilateral and bilateral chondrosarcoma of the mastoid. <i>Human Genetics and Genomics Advances</i> , 2020, 1, 100006.	1.0	3
3873	In silico deleterious prediction of nonsynonymous single nucleotide polymorphisms in <i>Neurexin1</i> gene for mental disorders. <i>International Journal of Bioinformatics Research and Applications</i> , 2020, 16, 1.	0.1	3
3874	MFA: An unfulfilled promise. <i>Computer Fraud and Security</i> , 2020, 2020, 20-20.	1.3	1
3876	A rare case of dermoid cyst arising in the upper lip. <i>International Journal of Surgery Case Reports</i> , 2020, 74, 77-81.	0.2	0
3877	Doctors as soldiers in times of pandemic. <i>Journal of Arthroscopy and Joint Surgery</i> , 2020, 7, 165-166.	0.3	0
3878	Pulse Oximeter Plethysmograph Variation During Hemorrhage in Beta-Blockerâ€“Treated Swine. <i>Journal of Surgical Research</i> , 2020, 256, 468-475.	0.8	3
3879	Phenotypic and Genotypic Features of Thai Patients With Nonsyndromic Tooth Agenesis and <i>WNT10A</i> Variants. <i>Frontiers in Physiology</i> , 2020, 11, 573214.	1.3	9

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3881	Quantum backflow for dissipative two-identical-particle systems. <i>Results in Physics</i> , 2020, 19, 103426.	2.0	1
3882	Methodological considerations for longitudinal investigations of young drivers. <i>Transportation Research Part F: Traffic Psychology and Behaviour</i> , 2020, 75, 214-221.	1.8	6
3883	Precise uncertain significance prediction using latent space matrix factorization models: genomics variant and heterogeneous clinical data-driven approaches. <i>Briefings in Bioinformatics</i> , 2021, 22, .	3.2	4
3884	Characterization of an N-terminal Nav1.5 channel variant " a potential risk factor for arrhythmias and sudden death?. <i>BMC Medical Genetics</i> , 2020, 21, 227.	2.1	1
3885	Wide-angle microwave absorption properties of multilayer metamaterial fabricated by 3D printing. <i>Materials Letters</i> , 2020, 281, 128571.	1.3	34
3886	Can DLNO/DLCO ratio offset prejudicial effects of functional heterogeneities in acinar regions?. <i>Respiratory Physiology and Neurobiology</i> , 2020, 282, 103517.	0.7	1
3887	Estimation of binder quantity in a binary blend of asphalt binders using FTIR. <i>Transportation Research Procedia</i> , 2020, 48, 3756-3763.	0.8	6
3890	Formulation and combinatorial effect of <i>Pseudomonas fluorescens</i> and <i>Bacillus coagulans</i> as biocontrol agents. <i>Biocatalysis and Agricultural Biotechnology</i> , 2020, 30, 101868.	1.5	2
3891	Effects of a simple cardiac rehabilitation program on improvement of self-reported physical activity in atrial fibrillation " Data from the RACE 3 study. <i>IJC Heart and Vasculature</i> , 2020, 31, 100673.	0.6	4
3892	Familial central precocious puberty: two novel MKRN3 mutations. <i>Pediatric Research</i> , 2020, 90, 431-435.	1.1	8
3893	PTMsnp: A Web Server for the Identification of Driver Mutations That Affect Protein Post-translational Modification. <i>Frontiers in Cell and Developmental Biology</i> , 2020, 8, 593661.	1.8	7
3894	A Novel ALAS2 Missense Mutation in Two Brothers With Iron Overload and Associated Alterations in Serum Hepcidin/Erythroferrone Levels. <i>Frontiers in Physiology</i> , 2020, 11, 581386.	1.3	2
3895	A novel design of UWB low noise amplifier for 2"10"GHz wireless sensor applications. <i>Sensors International</i> , 2020, 1, 100041.	4.9	3
3896	Relaci3n m3dico-paciente en la enfermedad cr3nica. <i>EMC - Tratado De Medicina</i> , 2020, 24, 1-6.	0.0	0
3897	Sport participation may protect socioeconomically disadvantaged youths with refugee backgrounds from experiencing behavioral and emotional difficulties. <i>Journal of Adolescence</i> , 2020, 85, 148-152.	1.2	6
3898	oxLDL receptor in lymphocytes prevents atherosclerosis and predicts subclinical disease. <i>Atherosclerosis</i> , 2020, 315, e90-e91.	0.4	0
3899	Crosslinks in COVID19 as inflammation,thrombosis and dyslipidemia. <i>Atherosclerosis</i> , 2020, 315, e185.	0.4	0
3900	Biomarkers of hypoxia in patients with chronic cardiorenal syndrome. <i>Atherosclerosis</i> , 2020, 315, e265-e266.	0.4	0

#	ARTICLE	IF	CITATIONS
3901	Are tie-over bolster dressings necessary for healing or success of full thickness skin graft reconstruction, following facial skin cancer excision?. British Journal of Oral and Maxillofacial Surgery, 2020, 58, e170.	0.4	0
3902	Dataset of concentrations of mercury and methylmercury in fish from a tropical river impacted by gold mining in the Colombian Pacific. Data in Brief, 2020, 33, 106513.	0.5	1
3903	Protective Effects of Curcumin on TNF α -induced Caco-2 cell Monolayer Permeabilization. Free Radical Biology and Medicine, 2020, 159, S43.	1.3	0
3904	Usefulness of familiarity signals during recognition depends on test format: Neurocognitive evidence for a core assumption of the CLS framework. Neuropsychologia, 2020, 148, 107659.	0.7	5
3905	EVALUATION OF DENTAL CARIES EXPERIENCE IN CHILDREN WITH CONGENITAL ZIKA SYNDROME. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2020, 130, e276.	0.2	1
3907	Effect of ultrasonic activation of the adhesive system on dentin tubule penetration and the pushout bond strength of fiber posts. Journal of Prosthetic Dentistry, 2020, , .	1.1	1
3911	IPSC-derived endothelial cells from high-risk cad patients demonstrate increased inducible adhesion molecule upregulation by inflammatory stimulation and gap closure delay compared to healthy controls. Atherosclerosis, 2020, 315, e88-e89.	0.4	0
3912	Dynamic forced shear characteristics of Ti-6Al-4V alloy using flat hat-shaped specimen. Engineering Fracture Mechanics, 2020, 238, 107286.	2.0	14
3914	Monoallelic Mutations in <i>CC2D1A</i> Suggest a Novel Role in Human Heterotaxy and Ciliary Dysfunction. Circulation Genomic and Precision Medicine, 2020, 13, e003000.	1.6	4
3915	Does mass testing work?. New Scientist, 2020, 248, 14-16.	0.0	0
3917	Behavioral and brain signatures of substance use vulnerability in childhood. Developmental Cognitive Neuroscience, 2020, 46, 100878.	1.9	23
3918	New filaments with natural fillers for FDM 3D printing and their applications in biomedical field. Procedia Manufacturing, 2020, 51, 698-703.	1.9	30
3920	13640 A case of multiple aggressive granular cell tumors. Journal of the American Academy of Dermatology, 2020, 83, AB10.	0.6	0
3921	16177 Assessment of conjunctivitis occurring in patients with long-term use of dupilumab for the treatment of atopic dermatitis in clinical practice. Journal of the American Academy of Dermatology, 2020, 83, AB55.	0.6	0
3922	16184 Risk stratification of severely dysplastic nevi with the use of noninvasively obtained gene expression and mutation analyses. Journal of the American Academy of Dermatology, 2020, 83, AB55.	0.6	0
3923	18684 A scoring method to assess the gentleness of cleansers. Journal of the American Academy of Dermatology, 2020, 83, AB104.	0.6	1
3924	14087 Integrating genomics for the diagnosis and prognosis of melanoma into clinical practice. Journal of the American Academy of Dermatology, 2020, 83, AB130.	0.6	0
3925	16465 Utility of 1565-nm nonablative fractional erbium laser in treatment of pediatric patients with acne scars measured by reflectance confocal microscopy. Journal of the American Academy of Dermatology, 2020, 83, AB179.	0.6	0

#	ARTICLE	IF	CITATIONS
3927	Unconventional Natural Gas Development and Hospitalization for Heart Failure in Pennsylvania. <i>Journal of the American College of Cardiology</i> , 2020, 76, 2862-2874.	1.2	17
3929	Clinical Phenotype and Course of PDE6A-Associated Retinitis Pigmentosa Disease, Characterized in Preparation for a Gene Supplementation Trial. <i>JAMA Ophthalmology</i> , 2020, 138, 1241.	1.4	9
3930	Peripherally inserted central venous catheter placed and maintained by a dedicated nursing team for the administration of antimicrobial therapy vs. another type of catheter: a retrospective case-control study. <i>Enfermedades Infecciosas Y Microbiología Clínica</i> , 2020, 38, 425-430.	0.3	5
3931	Novel nonsense mutation p. Gln264Ter in the ANK1 confirms causative role for hereditary spherocytosis: a case report. <i>BMC Medical Genetics</i> , 2020, 21, 223.	2.1	5
3932	Mitchell-Riley syndrome iPSC exhibit reduced pancreatic endoderm differentiation due to an RFX6 mutation. <i>Development (Cambridge)</i> , 2020, 147, .	1.2	10
3934	Binding numbers and restricted fractional $\frac{1}{x^2} = \frac{1}{x^2} + \frac{1}{x^2} + \frac{1}{x^2} + \dots$ in graphs. <i>Discrete Applied Mathematics</i> , 2021, 305, 350-356.	0.5	23
3938	Chronic In Vivo Administration of MnTnBuOE-2-PyP5+ Does Not Decrease Hypertensive Blood Pressure. <i>Free Radical Biology and Medicine</i> , 2020, 159, S97.	1.3	0
3939	Effects of graded levels of dietary squalene supplementation on the growth performance, plasma biochemical parameters, antioxidant capacity, and meat quality in broiler chickens. <i>Poultry Science</i> , 2020, 99, 5915-5924.	1.5	15
3940	Estimated prevalence of mucopolysaccharidoses from population-based exomes and genomes. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 324.	1.2	8
3941	rs2274911 polymorphism in GPRC6A associated with serum E2 and PSA in a Southern Chinese male population. <i>Gene</i> , 2020, 763, 145067.	1.0	4
3942	Correlation of luminescence and Judd-Ofelt intensity parameters in red ZrO2: Eu3+, Al3+ phosphor: The influences of Al3+ ions. <i>Materials Science and Engineering B: Solid-State Materials for Advanced Technology</i> , 2020, 262, 114794.	1.7	6
3943	Economic regulation and E-scooter networks in the USA. <i>Research in Transportation Economics</i> , 2020, 84, 100973.	2.2	29
3945	Eco-friendly silver nanoparticles (AgNPs) fabricated by green synthesis using the crude extract of marine polychaete, <i>Marphysa moribidii</i> : biosynthesis, characterisation, and antibacterial applications. <i>Heliyon</i> , 2020, 6, e05462.	1.4	27
3946	Carbonate diagenesis in fourth-order sequences: A case study of yingshan formation (Lower Tertiary). <i>Engineering</i> , 2020, 195, 107756.	2.1	9
3947	Knowledge-based Design Method of Forging Dies based on the Stereotypes of Die Structures and the Functions of Forming Surfaces. <i>Procedia Manufacturing</i> , 2020, 50, 475-482.	1.9	3
3948	Room-temperature deposition of low H-content SiNx/SiNxOy thin films using a specially designed PECVD system. <i>Surface and Coatings Technology</i> , 2020, 402, 126506.	2.2	4
3950	Commentary: Back to the future in cervical screening: applying a contemporary lens to an old controversy. <i>Journal of Clinical Epidemiology</i> , 2020, 127, 218-219.	2.4	2
3951	Clinical and genetic characteristics and prenatal diagnosis of patients presented GDD/ID with rare monogenic causes. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 317.	1.2	5

#	ARTICLE	IF	CITATIONS
3953	Toxicological implications of amplifying the antibacterial activity of gallic acid by immobilisation on silica particles: A study on <i>C. elegans</i> . <i>Environmental Toxicology and Pharmacology</i> , 2020, 80, 103492.	2.0	13
3954	Antimicrobial assessment of polyphenolic extracts from onion (<i>Allium cepa</i> L.) skin of fifteen cultivars by sonication-assisted extraction method. <i>Heliyon</i> , 2020, 6, e05478.	1.4	24
3955	A comparison of spent resin degradation by Fenton and O3-Fenton process. <i>Progress in Nuclear Energy</i> , 2020, 130, 103566.	1.3	9
3956	Investigation of Genetic Modifiers of Copper Toxicosis in Labrador Retrievers. <i>Life</i> , 2020, 10, 266.	1.1	10
3957	Volumes in the EFC series list. , 2020, , xi-xv.		0
3959	Autophagic cell death in viral infection: Do TAM receptors play a role?. <i>International Review of Cell and Molecular Biology</i> , 2020, 357, 123-168.	1.6	3
3960	Deformation and fracture of non-metallic inclusions in steel at different temperatures. <i>Journal of Materials Research and Technology</i> , 2020, 9, 15016-15022.	2.6	21
3965	Genomic Convergence in the Adaptation to Extreme Environments. <i>Plant Communications</i> , 2020, 1, 100117.	3.6	49
3966	Impact of amino acid substitution in the kinase domain of Bruton tyrosine kinase and its association with X-linked agammaglobulinemia. <i>International Journal of Biological Macromolecules</i> , 2020, 164, 2399-2408.	3.6	18
3967	Informing patients about their mutation tests: CDKN2A c.256G>A in melanoma as an example. <i>Hereditary Cancer in Clinical Practice</i> , 2020, 18, 15.	0.6	3
3968	CAMT-MPL: congenital amegakaryocytic thrombocytopenia caused by MPL mutations - heterogeneity of a monogenic disorder - a comprehensive analysis of 56 patients. <i>Haematologica</i> , 2021, 106, 2439-2448.	1.7	20
3969	An International Multicenter Evaluation of Inheritance Patterns, Arrhythmic Risks, and Underlying Mechanisms of <i>CASQ2</i> Catecholaminergic Polymorphic Ventricular Tachycardia. <i>Circulation</i> , 2020, 142, 932-947.	1.6	44
3970	Computational approach for predicting the functional effects of missense variants on Speckle-type BTB/POZ protein and association with prostate cancer. <i>Journal of Proteins and Proteomics</i> , 2020, 11, 205-212.	1.0	0
3971	Molecular diagnosis of epileptic encephalopathy of the first year of life applying a customized gene panel in a group of Argentinean patients. <i>Epilepsy and Behavior</i> , 2020, 111, 107322.	0.9	4
3972	Definition of a New Metric With Mutual Exclusivity and Coverage for Identifying Cancer Driver Modules. <i>IEEE Access</i> , 2020, 8, 133767-133776.	2.6	2
3973	Biallelic ZNF407 mutations in a neurodevelopmental disorder with ID, short stature and variable microcephaly, hypotonia, ocular anomalies and facial dysmorphism. <i>Journal of Human Genetics</i> , 2020, 65, 1115-1123.	1.1	5
3974	Analysis of exome-sequenced UK Biobank subjects implicates genes affecting risk of hyperlipidaemia. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 277-283.	0.5	5
3975	MISTIC: A prediction tool to reveal disease-relevant deleterious missense variants. <i>PLoS ONE</i> , 2020, 15, e0236962.	1.1	26

#	ARTICLE	IF	CITATIONS
3976	parSMURF, a high-performance computing tool for the genome-wide detection of pathogenic variants. <i>GigaScience</i> , 2020, 9, .	3.3	11
3977	Insights into changes in binding affinity caused by disease mutations in protein-protein complexes. <i>Computers in Biology and Medicine</i> , 2020, 123, 103829.	3.9	21
3978	The utility of whole exome sequencing for identification of the molecular etiology in autosomal recessive developmental and epileptic encephalopathies. <i>Neurological Sciences</i> , 2020, 41, 3729-3739.	0.9	5
3979	Molecular genetic analysis of AKR1C2-4 and HSD17B6 genes in subjects 46,XY with hypospadias. <i>Journal of Pediatric Urology</i> , 2020, 16, 689.e1-689.e12.	0.6	6
3980	Clinical features and underlying genetic causes in neonatal encephalopathy: A large cohort study. <i>Clinical Genetics</i> , 2020, 98, 365-373.	1.0	11
3981	Selective sweep analysis reveals extensive parallel selection traits between large white and Duroc pigs. <i>Evolutionary Applications</i> , 2020, 13, 2807-2820.	1.5	14
3982	RNA-Seq based genetic variant discovery provides new insights into controlling fat deposition in the tail of sheep. <i>Scientific Reports</i> , 2020, 10, 13525.	1.6	31
3983	Expansion of germline <i>RPS20</i> mutation phenotype to include Diamond-Blackfan anemia. <i>Human Mutation</i> , 2020, 41, 1918-1930.	1.1	13
3985	A gall-inducing infection of <i>Lepista</i> spp. in Norfolk by <i>Mycosymbiocytes mycenophila</i> - first record for Britain. <i>Field Mycology</i> , 2020, 21, 119-123.	0.0	1
3987	Amplification of SNRPE promotes tumor proliferation and invasion in triple-negative breast cancer by activating mTOR signaling. <i>European Journal of Cancer</i> , 2020, 138, S31-S32.	1.3	0
3988	Secondary resistance to the PI3K inhibitor copanlisib in marginal zone lymphoma. <i>European Journal of Cancer</i> , 2020, 138, S40.	1.3	5
3989	Early evidence of dose-dependent pharmacodynamic activity following treatment with SY-5609, a highly selective and potent oral CDK7 inhibitor, in patients with advanced solid tumors. <i>European Journal of Cancer</i> , 2020, 138, S50.	1.3	1
3992	Understanding Everyday Needs of Older Adults with Cognitive Impairments: Insights from Subject Matter Experts. <i>Archives of Physical Medicine and Rehabilitation</i> , 2020, 101, e10.	0.5	0
3993	Creating a Spanish-language Translation of Problem-Solving Training (PST) for Hispanic/Latinx Care Partners. <i>Archives of Physical Medicine and Rehabilitation</i> , 2020, 101, e76-e77.	0.5	0
3994	Emotion regulation in social interaction: Physiological and emotional responses associated with social inhibition. <i>International Journal of Psychophysiology</i> , 2020, 158, 62-72.	0.5	11
3995	Applying Antiracist Concepts to Clinical Practice. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2020, 59, 1299-1300.	0.3	0
3996	A Standardized and Comprehensive Approach to the Management of Cardiogenic Shock. <i>JACC: Heart Failure</i> , 2020, 8, 879-891.	1.9	132
3997	Gastrointestinal potassium binding in hemodialysis. <i>Kidney International</i> , 2020, 98, 1095-1097.	2.6	5

#	ARTICLE	IF	CITATIONS
3998	Reenvisioning the Adult Nephrology Workforce: The Future of Kidney Care in the United States. <i>Advances in Chronic Kidney Disease</i> , 2020, 27, 279-280.	0.6	1
3999	The genetic architecture of appendicular lean mass characterized by association analysis in the UK Biobank study. <i>Communications Biology</i> , 2020, 3, 608.	2.0	83
4000	Computational Modeling of NLRP3 Identifies Enhanced ATP Binding and Multimerization in Cryopyrin-Associated Periodic Syndromes. <i>Frontiers in Immunology</i> , 2020, 11, 584364.	2.2	9
4001	Landscape of driver mutations and their clinical impacts in pediatric B-cell precursor acute lymphoblastic leukemia. <i>Blood Advances</i> , 2020, 4, 5165-5173.	2.5	33
4002	Multiple neoplasia in a patient with Gitelman syndrome harboring germline monoallelic MUTYH mutation. <i>Npj Genomic Medicine</i> , 2020, 5, 39.	1.7	3
4003	A loss-of-function NLI2 mutation in humans causes anencephaly due to impaired Hippo-YAP signaling. <i>Journal of Experimental Medicine</i> , 2020, 217, .	4.2	25
4004	Mutations in the exocyst component EXOC2 cause severe defects in human brain development. <i>Journal of Experimental Medicine</i> , 2020, 217, .	4.2	17
4005	Genome sequencing unveils mutational landscape of the familial Mediterranean fever: Potential implications of IL33/ST2 signalling. <i>Journal of Cellular and Molecular Medicine</i> , 2020, 24, 11294-11306.	1.6	7
4006	m ⁶ A RNA modification modulates PI3K/Akt/mTOR signal pathway in Gastrointestinal Cancer. <i>Theranostics</i> , 2020, 10, 9528-9543.	4.6	62
4007	The genomic landscape of Mongolian hepatocellular carcinoma. <i>Nature Communications</i> , 2020, 11, 4383.	5.8	55
4008	Molecular and phenotypic investigation of a New Zealand cohort of childhood-onset retinal dystrophy. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 708-717.	0.7	12
4009	Predicting the most deleterious missense nsSNPs of the protein isoforms of the human HLA-G gene and in silico evaluation of their structural and functional consequences. <i>BMC Genetics</i> , 2020, 21, 94.	2.7	22
4010	Whole genome analysis of water buffalo and global cattle breeds highlights convergent signatures of domestication. <i>Nature Communications</i> , 2020, 11, 4739.	5.8	50
4011	De novo mutations of <i>SCN1A</i> are responsible for arthrogryposis broadening the <i>SCN1A</i> -related phenotypes. <i>Journal of Medical Genetics</i> , 2021, 58, 737-742.	1.5	13
4012	Family-Based Whole Genome Sequencing Identified Novel Variants in ABCA5 Gene in a Patient with Idiopathic Ventricular Tachycardia. <i>Pediatric Cardiology</i> , 2020, 41, 1783-1794.	0.6	1
4013	Molecular basis for a new bovine model of Niemann-Pick type C disease. <i>PLoS ONE</i> , 2020, 15, e0238697.	1.1	4
4014	Two Novel Pathogenic Variants Confirm RMND1 Causative Role in Perrault Syndrome with Renal Involvement. <i>Genes</i> , 2020, 11, 1060.	1.0	16
4015	A Membrane-Tethered Ubiquitination Pathway Regulates Hedgehog Signaling and Heart Development. <i>Developmental Cell</i> , 2020, 55, 432-449.e12.	3.1	21

#	ARTICLE	IF	CITATIONS
4016	Evaluating the informativeness of deep learning annotations for human complex diseases. <i>Nature Communications</i> , 2020, 11, 4703.	5.8	21
4017	In silico analysis predicting effects of deleterious SNPs of human RASSF5 gene on its structure and functions. <i>Scientific Reports</i> , 2020, 10, 14542.	1.6	51
4018	Genotype-phenotype correlation of 33 patients with maple syrup urine disease. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2486-2500.	0.7	6
4019	Primary Driver Mutations in GTF2I Specific to the Development of Thymomas. <i>Cancers</i> , 2020, 12, 2032.	1.7	17
4020	The Application of Next-Generation Sequencing (NGS) in Neonatal-Onset Urea Cycle Disorders (UCDs): Clinical Course, Metabolomic Profiling, and Genetic Findings in Nine Chinese Hyperammonemia Patients. <i>BioMed Research International</i> , 2020, 2020, 1-11.	0.9	4
4021	Molecular analysis of 76 Chinese hemophilia B pedigrees and the identification of 10 novel mutations. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1482.	0.6	7
4022	Transcript Assembly and Quantification by RNA-Seq Reveals Significant Differences in Gene Expression and Genetic Variants in Mosquitoes of the <i>Culex pipiens</i> (Diptera: Culicidae) Complex. <i>Journal of Medical Entomology</i> , 2021, 58, 139-145.	0.9	7
4023	A Novel Duplication Mutation in the Myelin Protein Zero Gene Causing Mild, Nonprogressive Demyelinating Neuropathy. <i>Case Reports in Neurology</i> , 2020, 12, 255-259.	0.3	0
4024	Specifications of the ACMG/AMP standards and guidelines for mitochondrial DNA variant interpretation. <i>Human Mutation</i> , 2020, 41, 2028-2057.	1.1	84
4025	A Clinicopathological Study of 29 Spitzoid Melanocytic Lesions With ALK Fusions, Including Novel Fusion Variants, Accompanied by Fluorescence In Situ Hybridization Analysis for Chromosomal Copy Number Changes, and Both TERT Promoter and Next-Generation Sequencing Mutation Analysis. <i>American Journal of Dermatopathology</i> , 2020, 42, 578-592.	0.3	11
4026	A novel homozygous variant in REN in a family presenting with classic features of disorders involving the renin-angiotensin pathway, without renal tubular dysgenesis. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2284-2290.	0.7	3
4027	A method for scoring the cell type-specific impacts of noncoding variants in personal genomes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 21364-21372.	3.3	16
4028	The Needle in the Haystack—Searching for Genetic and Epigenetic Differences in Monozygotic Twins Discordant for Tetralogy of Fallot. <i>Journal of Cardiovascular Development and Disease</i> , 2020, 7, 55.	0.8	10
4029	Molecular analysis of GALT gene in Argentinian population: Correlation with enzyme activity and characterization of a novel Duarte-like allele. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100695.	0.4	5
4030	Rare single-nucleotide DAB1 variants and their contribution to Schizophrenia and autism spectrum disorder susceptibility. <i>Human Genome Variation</i> , 2020, 7, 37.	0.4	7
4031	A flexible computational pipeline for research analyses of unsolved clinical exome cases. <i>Npj Genomic Medicine</i> , 2020, 5, 54.	1.7	4
4032	Mutational analysis in familial Alzheimer's disease of Han Chinese in Taiwan with a predominant mutation PSEN1 p.Met146Ile. <i>Scientific Reports</i> , 2020, 10, 19769.	1.6	7
4033	Status Quo of Glycosylation in Cancer: What Is, What Is Not and What Is to Be. <i>Applied Sciences (Switzerland)</i> , 2020, 10, 8401.	1.3	0

#	ARTICLE	IF	CITATIONS
4034	APC Mutations Are Not Confined to Hotspot Regions in Early-Onset Colorectal Cancer. <i>Cancers</i> , 2020, 12, 3829.	1.7	14
4035	Tracing selection signatures in the pig genome gives evidence for selective pressures on a unique curly hair phenotype in Mangalitza. <i>Scientific Reports</i> , 2020, 10, 22142.	1.6	8
4036	Background levels of OCPs, PCBs, and PAHs in soils from the eastern Pamirs, China, an alpine region influenced by westerly atmospheric transport. <i>Journal of Environmental Sciences</i> , 2022, 115, 453-464.	3.2	16
4037	Tensor products of quandles and 1-handles attached to surface-links. <i>Topology and Its Applications</i> , 2020, 301, 107520.	0.2	0
4038	A computational model for classification of BRCA2 variants using mouse embryonic stem cell-based functional assays. <i>Npj Genomic Medicine</i> , 2020, 5, 52.	1.7	18
4039	Role of Gender and Race in Patient-Reported Outcomes and Satisfaction. <i>Anesthesiology Clinics</i> , 2020, 38, 417-431.	0.6	11
4040	Capacity analysis of ship-tugging operations in a large container port. <i>Asian Transport Studies</i> , 2020, 6, 100011.	0.7	5
4041	Construction of a virtual supply chain using enterprise e-catalogues. <i>Procedia CIRP</i> , 2020, 93, 688-693.	1.0	9
4042	Novel case of percutaneous access of afferent limb of Roux-en-Y hepaticojejunostomy for biliary decompression and jejunoplasty in a patient with primary sclerosing cholangitis. <i>Radiology Case Reports</i> , 2020, 15, 2681-2686.	0.2	1
4043	Meningoradiculitis due to varicella zoster virus reactivation in a patient treated with ixekizumab. <i>Annales De Dermatologie Et De Venereologie</i> , 2020, 147, 667-671.	0.5	1
4044	The relation between mitogen activated protein kinase (MAPK) pathway and different genes expression in patients with beta Thalassemia. <i>Biochemistry and Biophysics Reports</i> , 2020, 24, 100836.	0.7	1
4045	Impact of a program with intensified physical exercise and nutritional support on body composition in patients with gastroesophageal cancer during peri-operative treatment. <i>Clinical Nutrition ESPEN</i> , 2020, 40, 463.	0.5	0
4046	On the growth rate of geodesic chords. <i>Differential Geometry and Its Applications</i> , 2020, 73, 101668.	0.2	1
4047	Les fractures du col du radius par accident d'â©quitation chez l'enfant: une étude comparative. <i>Revue De Chirurgie Orthopedique Et Traumatologique</i> , 2020, 106, 804-809.	0.0	0
4048	Adult Gender Dysphoria with Coronary Disease: Case Report and Literature Review. <i>Revista Colombiana De Psiquiatría (English Ed)</i> , 2020, 49, 211-215.	0.1	1
4049	128: Outcomes Following SBRT for Head and Neck Skin Cancer: A Single Institutional Experience. <i>Radiotherapy and Oncology</i> , 2020, 150, S56.	0.3	0
4051	Cachexia and procachectic targets in oncology patients. <i>Clinical Nutrition ESPEN</i> , 2020, 40, 553-554.	0.5	0
4052	Nutritional compliance to medical nutrition within different groups of oncology patients: a real life comparison. <i>Clinical Nutrition ESPEN</i> , 2020, 40, 558.	0.5	0

#	ARTICLE	IF	CITATIONS
4053	Performance improvement of a new proposed Savonius hydrokinetic turbine: a numerical investigation. <i>Energy Reports</i> , 2020, 6, 3051-3066.	2.5	27
4054	Aging-Associated Extracellular Vesicles Contain Immune Regulatory microRNAs Alleviating Hyperinflammatory State and Immune Dysfunction in the Elderly. <i>IScience</i> , 2020, 23, 101520.	1.9	24
4055	Material point method after 25 years: Theory, implementation, and applications. <i>Advances in Applied Mechanics</i> , 2020, 53, 185-398.	1.4	74
4056	Intravesical instillation of hyaluronic acid and chondroitin sulfate for recurrent urinary tract infections. <i>Actas Urológicas Españolas (English Edition)</i> , 2020, 44, 568-569.	0.2	0
4058	Multiple independent mechanisms link gene polymorphisms in the region of ZEB2 with risk of coronary artery disease. <i>Atherosclerosis</i> , 2020, 311, 20-29.	0.4	9
4059	Combining structure and genomics to understand antimicrobial resistance. <i>Computational and Structural Biotechnology Journal</i> , 2020, 18, 3377-3394.	1.9	17
4060	Identification of cross-reactive markers to strengthen the development of immunodiagnostic methods for angiostrongyliasis and other parasitic infections. <i>Experimental Parasitology</i> , 2020, 218, 107999.	0.5	4
4061	Control Law Design for MISO System in the Presence of Actuator Position Limit Constraint. <i>IFAC-PapersOnLine</i> , 2020, 53, 164-169.	0.5	1
4062	Isolation and characterization of ergosterol from <i>Monascus anka</i> for anti-lipid peroxidation properties. <i>Journal De Mycologie Medicale</i> , 2020, 30, 101038.	0.7	10
4063	Estrategia de rehabilitación de la escoliosis idiopática de la adolescencia. <i>EMC - Kinesiterapia - Medicina Física</i> , 2020, 41, 1-15.	0.1	0
4064	Unsupervised Clustering of Missense Variants in HNF1A Using Multidimensional Functional Data Aids Clinical Interpretation. <i>American Journal of Human Genetics</i> , 2020, 107, 670-682.	2.6	25
4065	Fluid expulsion system and tectonic architecture of the incipient Cascadia convergent margin as revealed by Nd, Sr and stable isotope composition of mid-Eocene methane seep carbonates. <i>Chemical Geology</i> , 2020, 558, 119872.	1.4	12
4066	Quantum chemical calculation of intrinsic reaction coordinates from trans to cis structure of flvoxamine. <i>Computational and Theoretical Chemistry</i> , 2020, 1192, 113051.	1.1	0
4067	The influence of workload on the nurses performance at Ambon general hospital. <i>Enfermería Clínica</i> , 2020, 30, 419-422.	0.1	2
4069	An enhanced quasicrystalline Ti1.4V0.6Ni alloy electrode modified by uniformly covered RGO for nickel metal hydride battery. <i>Intermetallics</i> , 2020, 127, 106972.	1.8	7
4070	Hot deformation behaviors of WE71 alloy under plain strain compression at elevated temperature. <i>Progress in Natural Science: Materials International</i> , 2020, 30, 526-532.	1.8	3
4071	Whole-exome sequencing in patients with protein aggregate myopathies reveals causative mutations associated with novel atypical phenotypes. <i>Neurological Sciences</i> , 2021, 42, 2819-2827.	0.9	2
4072	A model that adopts human fixations explains individual differences in multiple object tracking. <i>Cognition</i> , 2020, 205, 104418.	1.1	2

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4075	HER2DX: a tool that might inform treatment choices for HER2-positive breast cancer. <i>Lancet Oncology, The</i> , 2020, 21, 1392-1393.	5.1	1
4077	122 Point of Care Ultrasound Reduces Time to Diagnosis and Treatment of Ruptured Ectopic Pregnancy. <i>Annals of Emergency Medicine</i> , 2020, 76, S48.	0.3	0
4078	145 Physician Perceptions Impacting Snake Envenomation Treatment. <i>Annals of Emergency Medicine</i> , 2020, 76, S56-S57.	0.3	1
4079	168 Insights on Ultrasound Training for Ultrasound Naive Flight Paramedics and Nurses. <i>Annals of Emergency Medicine</i> , 2020, 76, S65-S66.	0.3	0
4080	Advance Care Planning Video Intervention. <i>Caring for the Ages</i> , 2020, 21, 19.	0.0	0
4081	Managing variable renewables with biomass in the European electricity system: Emission targets and investment preferences. <i>Energy</i> , 2020, 213, 118786.	4.5	19
4083	Effects of sacubitril valsartan on clinical and echocardiographic parameters of outpatients with heart failure and reduced ejection fraction. <i>IJC Heart and Vasculature</i> , 2020, 31, 100656.	0.6	15
4084	In Reply to Vitiello etÅal.: Acute Optic Neuritis Diagnosed by Bedside Ultrasound in an Emergency Department: Follow-Up and Clarification. <i>Journal of Emergency Medicine</i> , 2020, 59, 449-451.	0.3	0
4085	Precise phase control of resonant MOEMS mirrors by comb-drive current feedback. <i>Mechatronics</i> , 2020, 71, 102420.	2.0	10
4086	Nomophobia and self-reported smartphone use while driving: An investigation into whether nomophobia can increase the likelihood of illegal smartphone use while driving. <i>Transportation Research Part F: Traffic Psychology and Behaviour</i> , 2020, 74, 212-224.	1.8	17
4088	Profiling cardio-active compound response between high-throughput 2D monolayers, anisotropic and 3-dimensional human induced pluripotent stem cell-derived cardiomyocytes. <i>Journal of Pharmacological and Toxicological Methods</i> , 2020, 105, 106860.	0.3	0
4089	NGS-based expanded carrier screening for genetic disorders in North Indian population reveals unexpected results â€“ a pilot study. <i>BMC Medical Genetics</i> , 2020, 21, 216.	2.1	9
4090	The genetic landscape of axonal neuropathies in the middle-aged and elderly. <i>Neurology</i> , 2020, 95, e3163-e3179.	1.5	19
4091	Impaired cognitive performance under psychosocial stress in cannabis-dependent men is associated with attenuated precuneus activity. <i>Journal of Psychiatry and Neuroscience</i> , 2020, 45, 88-97.	1.4	9
4093	Prediction of Protein Mutational Free Energy: Benchmark and Sampling Improvements Increase Classification Accuracy. <i>Frontiers in Bioengineering and Biotechnology</i> , 2020, 8, 558247.	2.0	55
4094	Behavioral and Social Research to Accelerate the Geroscience Translation Agenda. <i>Ageing Research Reviews</i> , 2020, 63, 101146.	5.0	23
4095	The relation between neurofunctional and neurostructural determinants of phonological processing in pre-readers. <i>Developmental Cognitive Neuroscience</i> , 2020, 46, 100874.	1.9	5
4096	Increasing gaps between materials demand and materials recycling rates: A historical perspective for evolution of consumer products and waste quantities. <i>Journal of Environmental Management</i> , 2020, 276, 111196.	3.8	15

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4097	Compact maglev stage system for nanometer-scale positioning. <i>Precision Engineering</i> , 2020, 66, 519-530.	1.8	9
4099	Papel de la imagen multimodal en la periartritis relacionada con la inmunoglobulina G4. <i>Revista Espanola De Cardiologia</i> , 2020, 73, 948.	0.6	0
4100	Variant discovery using next-generation sequencing and its future role in pharmacogenetics. <i>Pharmacogenomics</i> , 2020, 21, 471-486.	0.6	9
4101	RP1L1 and inherited photoreceptor disease: A review. <i>Survey of Ophthalmology</i> , 2020, 65, 725-739.	1.7	25
4102	Loss-of-function variants in C3ORF52 result in localized autosomal recessive hypotrichosis. <i>Genetics in Medicine</i> , 2020, 22, 1227-1234.	1.1	12
4103	The evolutionary history of extinct and living lions. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 10927-10934.	3.3	70
4104	PG-path: Modeling and personalizing pharmacogenomics-based pathways. <i>PLoS ONE</i> , 2020, 15, e0230950.	1.1	3
4105	Single-fiber studies for assigning pathogenicity of eight mitochondrial DNA variants associated with mitochondrial diseases. <i>Human Mutation</i> , 2020, 41, 1394-1406.	1.1	4
4106	Genomically Aided Diagnosis of Severe Developmental Disorders. <i>Annual Review of Genomics and Human Genetics</i> , 2020, 21, 327-349.	2.5	3
4107	Inactivation of <i>Fbxw7</i> Impairs dsRNA Sensing and Confers Resistance to PD-1 Blockade. <i>Cancer Discovery</i> , 2020, 10, 1296-1311.	7.7	49
4108	Identifying disease-causing mutations in genomes of single patients by computational approaches. <i>Human Genetics</i> , 2020, 139, 769-776.	1.8	12
4109	Clinical characterization of patients with leucine-rich repeat kinase 2 genetic variants in Japan. <i>Journal of Human Genetics</i> , 2020, 65, 771-781.	1.1	15
4110	Role of SPRED1 in keratinocyte proliferation in psoriasis. <i>Journal of Dermatology</i> , 2020, 47, 735-742.	0.6	5
4111	Identification and functional characterization of CYP4V2 genetic variants exhibiting decreased activity of lauric acid metabolism. <i>Annals of Human Genetics</i> , 2020, 84, 400-411.	0.3	4
4112	Genetic and Clinical Heterogeneity in Thirteen New Cases with Aceruloplasminemia. Atypical Anemia as a Clue for an Early Diagnosis. <i>International Journal of Molecular Sciences</i> , 2020, 21, 2374.	1.8	25
4113	Burden of Rare Variants in ALS and Axonal Hereditary Neuropathy Genes Influence Survival in ALS: Insights from a Next Generation Sequencing Study of an Italian ALS Cohort. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3346.	1.8	11
4114	Myotonia congenita and periodic hypokalemia paralysis in a consanguineous marriage pedigree: Coexistence of a novel CLCN1 mutation and an SCN4A mutation. <i>PLoS ONE</i> , 2020, 15, e0233017.	1.1	4
4115	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. <i>American Journal of Human Genetics</i> , 2020, 106, 893-904.	2.6	29

#	ARTICLE	IF	CITATIONS
4116	Research on Remote Sensing Image Target Recognition Based on Deep Convolution Neural Network. International Journal of Pattern Recognition and Artificial Intelligence, 2020, 34, 2054015.	0.7	8
4117	Genetic homozygosity in a diverse population: An experience of long QT syndrome. International Journal of Cardiology, 2020, 316, 117-124.	0.8	2
4118	Frequency of Usher gene mutations in non-syndromic hearing loss: higher variability of the Usher phenotype. Journal of Human Genetics, 2020, 65, 855-864.	1.1	6
4119	Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction. Nature Communications, 2020, 11, 2542.	5.8	59
4120	Identification of ACKR1 variants associated with altered Duffy phenotype expression in blood donors from southern Brazil. Transfusion and Apheresis Science, 2020, 59, 102768.	0.5	1
4121	Genetic testing strategies in the newborn. Journal of Perinatology, 2020, 40, 1007-1016.	0.9	9
4122	Genomic and Transcriptomic Characteristics According to Size of Papillary Thyroid Microcarcinoma. Cancers, 2020, 12, 1345.	1.7	12
4123	Dysregulated expression of androgen metabolism genes and genetic analysis in hypospadias. Molecular Genetics & Genomic Medicine, 2020, 8, e1346.	0.6	10
4124	New DPYD variants causing DPD deficiency in patients treated with fluoropyrimidine. Cancer Chemotherapy and Pharmacology, 2020, 86, 45-54.	1.1	13
4125	<i>GJB4</i> and <i>GJC3</i> variants in non-syndromic hearing impairment in Ghana. Experimental Biology and Medicine, 2020, 245, 1355-1367.	1.1	4
4126	Mitochondrial DNA intra-individual variation in a bumblebee species: A challenge for evolutionary studies and molecular identification. Mitochondrion, 2020, 53, 243-254.	1.6	9
4127	Focal epilepsy in <i>SCN1A</i> mutation carrying patients: is there a role for epilepsy surgery?. Developmental Medicine and Child Neurology, 2020, 62, 1331-1335.	1.1	20
4128	A Bayesian method to estimate variant-induced disease penetrance. PLoS Genetics, 2020, 16, e1008862.	1.5	11
4129	Pancreatic ductal adenocarcinomas from Mexican patients present a distinct genomic mutational pattern. Molecular Biology Reports, 2020, 47, 5175-5184.	1.0	3
4130	Identification of a novel titin-cap/telethonin mutation in a Portuguese family with hypertrophic cardiomyopathy. Revista Portuguesa De Cardiologia, 2020, 39, 317-327.	0.2	4
4131	Granularity of <i>SERPINA1</i> alleles by DNA sequencing in CanCOLD. European Respiratory Journal, 2020, 56, 2000958.	3.1	13
4132	Hemizygous mutations in L1CAM in two unrelated male probands with childhood onset psychosis. Psychiatric Genetics, 2020, 30, 73-82.	0.6	2
4133	Genetic locus responsible for diabetic phenotype in the insulin hyposecretion (ihs) mouse. PLoS ONE, 2020, 15, e0234132.	1.1	4

#	ARTICLE	IF	CITATIONS
4134	SEMA3A and IGSF10 Are Novel Contributors to Combined Pituitary Hormone Deficiency (CPHD). <i>Frontiers in Endocrinology</i> , 2020, 11, 368.	1.5	13
4135	Insights into the Evolution of Neoteny from the Genome of the Asian Icefish <i>Protosalanx chinensis</i> . <i>IScience</i> , 2020, 23, 101267.	1.9	7
4136	Progressive cavitating leukoencephalopathy associated with a homozygous POLG mutation of 264C>G (p.F88L). <i>Radiology Case Reports</i> , 2020, 15, 908-913.	0.2	1
4137	Whole exome sequencing identifies compound heterozygous variants of CR2 gene in monozygotic twin patients with common variable immunodeficiency. <i>SAGE Open Medicine</i> , 2020, 8, 205031212092265.	0.7	1
4138	Lamotrigine induced Brugada-pattern in a patient with genetic epilepsy associated with a novel variant in SCN9A. <i>Gene</i> , 2020, 754, 144847.	1.0	14
4139	Gene-Wise Burden of Coding Variants Correlates to Noncoding Pharmacogenetic Risk Variants. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3091.	1.8	5
4140	Whole exome sequencing reveals the different responsiveness to Enterovirus 71 vaccination in Chinese children. <i>International Journal of Infectious Diseases</i> , 2020, 97, 47-53.	1.5	1
4141	Non-random distribution of deleterious mutations in the DNA and protein-binding domains of IRF6 are associated with Van Der Woude syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1355.	0.6	13
4142	A novel COQ8A missense variant associated with a mild form of primary coenzyme Q10 deficiency type 4. <i>Clinical Biochemistry</i> , 2020, 84, 93-98.	0.8	5
4143	Benchmarking analysis of deleterious SNP prediction tools on CYP2D6 enzyme. <i>Chemical Biology and Drug Design</i> , 2020, 96, 984-994.	1.5	6
4144	Testing association of rare genetic variants with resistance to three common antiseizure medications. <i>Epilepsia</i> , 2020, 61, 657-666.	2.6	22
4145	A Missense Mutation in the UGDH Gene Is Associated With Developmental Delay and Axial Hypotonia. <i>Frontiers in Pediatrics</i> , 2020, 8, 71.	0.9	15
4146	KRGDB: the large-scale variant database of 1722 Koreans based on whole genome sequencing. <i>Database: the Journal of Biological Databases and Curation</i> , 2020, 2020, .	1.4	36
4147	Association of CHRNA5 Gene Variants with Crack Cocaine Addiction. <i>NeuroMolecular Medicine</i> , 2020, 22, 384-390.	1.8	9
4148	m7GHub: deciphering the location, regulation and pathogenesis of internal mRNA N7-methylguanosine (m7G) sites in human. <i>Bioinformatics</i> , 2020, 36, 3528-3536.	1.8	85
4149	A decision tree to improve identification of pathogenic mutations in clinical practice. <i>BMC Medical Informatics and Decision Making</i> , 2020, 20, 52.	1.5	20
4150	Compound heterozygous mutations in ABCG5 or ABCG8 causing Chinese familial Sitosterolemia. <i>Journal of Gene Medicine</i> , 2020, 22, e3185.	1.4	8
4151	Biallelic variants in PSMB1 encoding the proteasome subunit β 26 cause impairment of proteasome function, microcephaly, intellectual disability, developmental delay and short stature. <i>Human Molecular Genetics</i> , 2020, 29, 1132-1143.	1.4	30

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4152	Inherited Missense Mutation Occurring in Arginine76 of the <i>SRY</i> Gene Does Not Account for Familial 46, XY Sex Reversal. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 1355-1365.	1.8	1
4153	Identification and clinical implications of a novel pathogenic variant in the <i>GJB2</i> gene causes autosomal recessive non-syndromic hearing loss in a consanguineous Iranian family. <i>Intractable and Rare Diseases Research</i> , 2020, 9, 30-34.	0.3	2
4154	Variants in saposin D domain of prosaposin gene linked to Parkinson's disease. <i>Brain</i> , 2020, 143, 1190-1205.	3.7	72
4155	Heterogeneity of chondrosarcomas response to irradiations with X-rays and carbon ions: A comparative study on five cell lines. <i>Journal of Bone Oncology</i> , 2020, 22, 100283.	1.0	10
4156	Pathway Mutations in Breast Cancer Using Whole-Exome Sequencing. <i>Oncology Research</i> , 2020, 28, 107-116.	0.6	15
4157	Mutation screening of 17 candidate genes in a cohort of 67 probands with early-onset high myopia. <i>Ophthalmic and Physiological Optics</i> , 2020, 40, 271-280.	1.0	16
4158	Wolff-Parkinson-White syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1387-1399.	0.7	14
4159	First-line exome sequencing in Palestinian and Israeli Arabs with neurological disorders is efficient and facilitates disease gene discovery. <i>European Journal of Human Genetics</i> , 2020, 28, 1034-1043.	1.4	20
4160	Rapid Identification of Biallelic <i>SPTB</i> Mutation in a Neonate with Severe Congenital Hemolytic Anemia and Liver Failure. <i>Molecular Syndromology</i> , 2020, 11, 50-55.	0.3	9
4161	Identification of novel missense mutations associated with non-syndromic syndactyly in two vietnamese trios by whole exome sequencing. <i>Clinica Chimica Acta</i> , 2020, 506, 16-21.	0.5	4
4162	Unraveling the Influence of Common von Willebrand factor variants on von Willebrand Disease Phenotype: An Exploratory Study on the Molecular and Clinical Profile of von Willebrand Disease in Spain Cohort. <i>Thrombosis and Haemostasis</i> , 2020, 120, 437-448.	1.8	2
4163	The European Association for Haemophilia and Allied Disorders (EAHAD) Coagulation Factor Variant Databases: Important resources for haemostasis clinicians and researchers. <i>Haemophilia</i> , 2020, 26, 306-313.	1.0	55
4164	Investigation of metastasis-associated in colon cancer-1 genetic variants in the development and clinicopathological characteristics of uterine cervical cancer in Taiwanese women. <i>International Journal of Medical Sciences</i> , 2020, 17, 490-497.	1.1	5
4165	Differential Frequency of <i>CYP2R1</i> Variants Across Populations Reveals Pathway Selection for Vitamin D Homeostasis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 1302-1315.	1.8	5
4166	Direct coupling analysis of epistasis in allosteric materials. <i>PLoS Computational Biology</i> , 2020, 16, e1007630.	1.5	14
4167	Mutations in the coat complex II component SEC23B promote colorectal cancer metastasis. <i>Cell Death and Disease</i> , 2020, 11, 157.	2.7	7
4168	Low- γ GGT intrahepatic cholestasis associated with biallelic <i>USP53</i> variants: Clinical, histological and ultrastructural characterization. <i>Liver International</i> , 2020, 40, 1142-1150.	1.9	34
4169	The molecular landscape of osteogenesis imperfecta in a Brazilian tertiary service cohort. <i>Osteoporosis International</i> , 2020, 31, 1341-1352.	1.3	5

#	ARTICLE	IF	CITATIONS
4170	Whole-genome sequencing of European autochthonous and commercial pig breeds allows the detection of signatures of selection for adaptation of genetic resources to different breeding and production systems. <i>Genetics Selection Evolution</i> , 2020, 52, 33.	1.2	45
4171	New germline mutations in non-BRCA genes among breast cancer women of Mongoloid origin. <i>Molecular Biology Reports</i> , 2020, 47, 5315-5321.	1.0	4
4172	Variants in <i>CHRNA4</i> and <i>CHRNA4</i> Identified in Patients with Insular Epilepsy. <i>Canadian Journal of Neurological Sciences</i> , 2020, 47, 800-809.	0.3	7
4173	Investigation on the role of biallelic variants in <i>VEGFA</i> found in a patient affected by Milroy's lymphedema. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1389.	0.6	6
4174	Differences in the molecular profile of endometrial cancers from British White and British South Asian women. <i>PLoS ONE</i> , 2020, 15, e0233900.	1.1	6
4175	Association Between Glucocerebrosidase Mutations and Parkinson's Disease in Ireland. <i>Frontiers in Neurology</i> , 2020, 11, 527.	1.1	17
4176	HTRA1-Related Cerebral Small Vessel Disease: A Review of the Literature. <i>Frontiers in Neurology</i> , 2020, 11, 545.	1.1	52
4177	Developmental delay, intellectual disability, short stature, subglottic stenosis, hearing impairment, onychodysplasia of the index fingers, and distinctive facial features: A newly reported autosomal recessive syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1865-1872.	0.7	2
4178	PTEN Protein Loss and Loss-of-Function Mutations in Gastric Cancers: The Relationship with Microsatellite Instability, EBV, HER2, and PD-L1 Expression. <i>Cancers</i> , 2020, 12, 1724.	1.7	13
4179	Characterization of Exome Variants and Their Metabolic Impact in 6,716 American Indians from the Southwest US. <i>American Journal of Human Genetics</i> , 2020, 107, 251-264.	2.6	12
4180	Heterogeneous phenotype of Hereditary Xerocytosis in association with PIEZO1 variants. <i>Blood Cells, Molecules, and Diseases</i> , 2020, 82, 102413.	0.6	4
4181	Novel MNX1 mutations and genotype-phenotype analysis of patients with Currarino syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 155.	1.2	3
4182	WDR34 mutation from anencephaly patients impaired both SHH and PCP signaling pathways. <i>Journal of Human Genetics</i> , 2020, 65, 985-993.	1.1	5
4183	Statistical methods for SNP heritability estimation and partition: A review. <i>Computational and Structural Biotechnology Journal</i> , 2020, 18, 1557-1568.	1.9	41
4184	The association between Single Nucleotide Polymorphisms of Klotho Gene and Mortality in Elderly Men: The MrOS Sweden Study. <i>Scientific Reports</i> , 2020, 10, 10243.	1.6	3
4185	A novel loss-of-function mutation of PBK associated with human kidney stone disease. <i>Scientific Reports</i> , 2020, 10, 10282.	1.6	3
4186	Different Clinical Phenotypes Caused by Three <i>F8</i> Missense Mutations in Three Chinese Families with Moderate Hemophilia A. <i>DNA and Cell Biology</i> , 2020, 39, 1685-1690.	0.9	0
4187	Comparative Genomics Reveals Evolution of a Beak Morphology Locus in a High-Altitude Songbird. <i>Molecular Biology and Evolution</i> , 2020, 37, 2983-2988.	3.5	6

#	ARTICLE	IF	CITATIONS
4188	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A (<i>KIF1A</i>) in patients with developmental delay. <i>Journal of Medical Genetics</i> , 2020, 57, 107-111.	1.1	16
4189	Genetic factors for short life span associated with evolution of the loss of flight ability. <i>Ecology and Evolution</i> , 2020, 10, 6020-6029.	0.8	1
4190	Clinical and Molecular Investigation of Familial Multiple Lipomatosis: Variants in the <i>HMGGA2</i> Gene. <i>Clinical, Cosmetic and Investigational Dermatology</i> , 2020, Volume 13, 1-10.	0.8	6
4191	Genomic and Transcriptomic Characterization of Sporadic Medullary Thyroid Carcinoma. <i>Thyroid</i> , 2020, 30, 1025-1036.	2.4	10
4192	Rare Co-occurrence of Beta-Thalassemia and Pseudoxanthoma elasticum: Novel Biomolecular Findings. <i>Frontiers in Medicine</i> , 2020, 6, 322.	1.2	24
4193	Human FCHO1 deficiency reveals role for clathrin-mediated endocytosis in development and function of T cells. <i>Nature Communications</i> , 2020, 11, 1031.	5.8	23
4194	Novel pathogenic alterations in pediatric and adult desmoid-type fibromatosis – A systematic analysis of 204 cases. <i>Scientific Reports</i> , 2020, 10, 3368.	1.6	29
4195	NOX1 Regulates Collective and Planktonic Cell Migration: Insights From Patients With Pediatric-Onset IBD and NOX1 Deficiency. <i>Inflammatory Bowel Diseases</i> , 2020, 26, 1166-1176.	0.9	9
4196	S100A7/Ran-binding protein 9 coevolution in mammals. <i>Immunogenetics</i> , 2020, 72, 155-164.	1.2	1
4197	A polygenic predictor of treatment-resistant depression using whole exome sequencing and genome-wide genotyping. <i>Translational Psychiatry</i> , 2020, 10, 50.	2.4	33
4198	Expanding the phenotypic spectrum in RDH12-associated retinal disease. <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a004754.	0.5	16
4199	Neurological Involvement in Glycogen Storage Disease Type IXa due to PHKA2 Mutation. <i>Canadian Journal of Neurological Sciences</i> , 2020, 47, 400-403.	0.3	7
4200	Ellis-van Creveld syndrome: Report of a case and recurrent variant. <i>Journal of Gene Medicine</i> , 2020, 22, e3175.	1.4	3
4201	An autosomal dominant ERLIN2 mutation leads to a pure HSP phenotype distinct from the autosomal recessive ERLIN2 mutations (SPG18). <i>Scientific Reports</i> , 2020, 10, 3295.	1.6	11
4202	The genetics of situs inversus without primary ciliary dyskinesia. <i>Scientific Reports</i> , 2020, 10, 3677.	1.6	37
4203	A novel compound heterozygous mutation in <i>DGKE</i> in a Chinese patient causes atypical hemolytic uremic syndrome. <i>Hematology</i> , 2020, 25, 101-107.	0.7	4
4204	Leveraging Human Genetics to Identify Safety Signals Prior to Drug Marketing Approval and Clinical Use. <i>Drug Safety</i> , 2020, 43, 567-582.	1.4	9
4205	Missense PALB2 germline variant disrupts nuclear localization of PALB2 in a patient with breast cancer. <i>Familial Cancer</i> , 2020, 19, 123-131.	0.9	3

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4206	Whole genome sequencing unveils genetic heterogeneity in optic nerve hypoplasia. <i>PLoS ONE</i> , 2020, 15, e0228622.	1.1	6
4207	A mutation map for human glycoside hydrolase genes. <i>Glycobiology</i> , 2020, 30, 500-515.	1.3	6
4208	<i>LACC1</i> gene mutation in three sisters with polyarthritis without systemic features. <i>Annals of the Rheumatic Diseases</i> , 2020, 79, 425-426.	0.5	8
4209	An International Multicenter Evaluation of Type 5 Long QT Syndrome. <i>Circulation</i> , 2020, 141, 429-439.	1.6	39
4210	The Rare <i>IL22RA2</i> Signal Peptide Coding Variant rs28385692 Decreases Secretion of IL-22BP Isoform-1, -2 and -3 and Is Associated with Risk for Multiple Sclerosis. <i>Cells</i> , 2020, 9, 175.	1.8	1
4211	Familial "Diplegic" Migraine" Description of a Family With a Novel <i>CACNA1A</i> Mutation. <i>Headache</i> , 2020, 60, 600-606.	1.8	4
4212	A protein-centric approach for exome variant aggregation enables sensitive association analysis with clinical outcomes. <i>Human Mutation</i> , 2020, 41, 934-945.	1.1	3
4213	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing. <i>Eye and Vision (London, England)</i> , 2020, 7, 3.	1.4	5
4214	The polygenic nature of mild-to-moderate hypertriglyceridemia. <i>Journal of Clinical Lipidology</i> , 2020, 14, 28-34.e2.	0.6	32
4215	A new mutational hotspot in the <i>SKI</i> gene in the context of MFS/TAA molecular diagnosis. <i>Human Genetics</i> , 2020, 139, 461-472.	1.8	8
4216	Cardio-facio-cutaneous syndrome-associated pathogenic <i>MAP2K1</i> variants activate autophagy. <i>Gene</i> , 2020, 733, 144369.	1.0	14
4217	Biallelic variants/mutations of <i>IL1RAP</i> in patients with steroid-sensitive nephrotic syndrome. <i>International Immunology</i> , 2020, 32, 283-292.	1.8	3
4218	In-silico Analysis of <i>NF1</i> Missense Variants in ClinVar: Translating Variant Predictions into Variant Interpretation and Classification. <i>International Journal of Molecular Sciences</i> , 2020, 21, 721.	1.8	9
4219	Molecular basis of Leigh syndrome: a current look. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 31.	1.2	62
4220	Compound heterozygous mutations in <i>FBN1</i> in a large family with Marfan syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1116.	0.6	5
4221	Comparative genomics shows differences in the electron transport and carbon metabolic pathways of <i>Mycobacterium africanum</i> relative to <i>Mycobacterium tuberculosis</i> and suggests an adaptation to low oxygen tension. <i>Tuberculosis</i> , 2020, 120, 101899.	0.8	15
4222	Stepwise evolution and convergent recombination underlie the global dissemination of carbapenemase-producing <i>Escherichia coli</i> . <i>Genome Medicine</i> , 2020, 12, 10.	3.6	40
4223	Dating genomic variants and shared ancestry in population-scale sequencing data. <i>PLoS Biology</i> , 2020, 18, e3000586.	2.6	127

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4224	Autosomal Dominant Tubulointerstitial Kidney Disease—Uromodulin Misclassified as Focal Segmental Glomerulosclerosis or Hereditary Glomerular Disease. <i>Kidney International Reports</i> , 2020, 5, 519-529.	0.4	14
4225	RPE65 and retinal dystrophy: Report of new and recurrent mutations. <i>Journal of Gene Medicine</i> , 2020, 22, e3154.	1.4	7
4226	Application of targeted panel sequencing and whole exome sequencing for 76 Chinese families with retinitis pigmentosa. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1131.	0.6	27
4227	Brown-Vialetto-Van Laere syndrome and Fazio-Londe syndrome: A novel mutation and in silico analyses. <i>Journal of Clinical Neuroscience</i> , 2020, 72, 342-349.	0.8	5
4228	A novel TAB2 nonsense mutation (p.S149X) causing autosomal dominant congenital heart defects: a case report of a Chinese family. <i>BMC Cardiovascular Disorders</i> , 2020, 20, 27.	0.7	14
4229	Computational analyses prioritize and reveal the deleterious nsSNPs in human angiotensinogen gene. <i>Computational Biology and Chemistry</i> , 2020, 84, 107199.	1.1	8
4230	Genetic analysis of familial hypercholesterolemia in Asian Indians: A single-center study. <i>Journal of Clinical Lipidology</i> , 2020, 14, 35-45.	0.6	10
4231	Genetic Analyses in Dent Disease and Characterization of CLCN5 Mutations in Kidney Biopsies. <i>International Journal of Molecular Sciences</i> , 2020, 21, 516.	1.8	17
4232	Variants in CAPZA2, a member of an F-actin capping complex, cause intellectual disability and developmental delay. <i>Human Molecular Genetics</i> , 2020, 29, 1537-1546.	1.4	15
4233	Four novel compound heterozygous mutations in C5orf42 gene in patients with pure and mild Joubert syndrome. <i>International Journal of Developmental Neuroscience</i> , 2020, 80, 455-463.	0.7	6
4234	Rare variants lowering the levels of coagulation factor X are protective against ischemic heart disease. <i>Haematologica</i> , 2020, 105, e365-e369.	1.7	9
4235	Bardet-Biedl syndrome in two unrelated patients with identical compound heterozygous SCLT1 mutations. <i>CEN Case Reports</i> , 2020, 9, 260-265.	0.5	11
4236	Partial thyroxine-binding globulin deficiency in a family with coding region mutations in the TBG gene. <i>Journal of Endocrinological Investigation</i> , 2020, 43, 1703-1710.	1.8	2
4237	Loss-of-Function Variants in SERPINA12 Underlie Autosomal Recessive Palmoplantar Keratoderma. <i>Journal of Investigative Dermatology</i> , 2020, 140, 2178-2187.	0.3	14
4238	A comparison of genomic diagnostics in adults and children with epilepsy and comorbid intellectual disability. <i>European Journal of Human Genetics</i> , 2020, 28, 1066-1077.	1.4	30
4239	Significant loss of genetic diversity and accumulation of deleterious genetic variation in a critically endangered azalea species, <i>Rhododendron boninense</i> , growing on the Bonin Islands. <i>Plant Species Biology</i> , 2020, 35, 166-174.	0.6	4
4240	<i>PRKDC</i>: new biomarker and drug target for checkpoint blockade immunotherapy. , 2020, 8, e000485.		32
4241	Tumour characteristics provide evidence for germline mismatch repair missense variant pathogenicity. <i>Journal of Medical Genetics</i> , 2020, 57, 62-69.	1.5	11

#	ARTICLE	IF	CITATIONS
4242	A Novel Cosegregating DCTN1 Splice Site Variant in a Family with Bipolar Disorder May Hold the Key to Understanding the Etiology. <i>Genes</i> , 2020, 11, 446.	1.0	1
4243	Genetic variants of uncertain significance: How to match scientific rigour and standard of proof in sudden cardiac death?. <i>Legal Medicine</i> , 2020, 45, 101712.	0.6	22
4244	Comprehensive molecular analysis of 61 Egyptian families with hereditary nonsyndromic hearing loss. <i>Clinical Genetics</i> , 2020, 98, 32-42.	1.0	22
4245	Oncogenic and drug-sensitive RET mutations in human epithelial ovarian cancer. <i>Journal of Experimental and Clinical Cancer Research</i> , 2020, 39, 53.	3.5	3
4246	<i>C</i> Scape-somatic: distinguishing driver and passenger point mutations in the cancer genome. <i>Bioinformatics</i> , 2020, 36, 3637-3644.	1.8	19
4247	Hypermutator <i>Pseudomonas aeruginosa</i> Exploits Multiple Genetic Pathways To Develop Multidrug Resistance during Long-Term Infections in the Airways of Cystic Fibrosis Patients. <i>Antimicrobial Agents and Chemotherapy</i> , 2020, 64, .	1.4	47
4248	Collagen VI-related limb-girdle syndrome caused by frequent mutation in COL6A3 gene with conflicting reports of pathogenicity. <i>Neuromuscular Disorders</i> , 2020, 30, 483-491.	0.3	3
4249	Spectrum of Genetic Variants Associated with Anterior Segment Dysgenesis in South Florida. <i>Genes</i> , 2020, 11, 350.	1.0	14
4250	Compound heterozygous variants of the <i>FBXO7</i> gene resulting in infantile-onset Parkinsonian-pyramidal syndrome in siblings of a Chinese family. <i>Journal of Clinical Laboratory Analysis</i> , 2020, 34, e23324.	0.9	8
4251	A novel hemizygous loss-of-function mutation in ADGRG2 causes male infertility with congenital bilateral absence of the vas deferens. <i>Journal of Assisted Reproduction and Genetics</i> , 2020, 37, 1421-1429.	1.2	11
4252	Novel insights into the organic solute transporter alpha/beta, OST α/β : From the bench to the bedside. , 2020, 211, 107542.		38
4253	CDH1 gene mutation in early-onset, colorectal signet-ring cell carcinoma. <i>Pathology Research and Practice</i> , 2020, 216, 152912.	1.0	7
4254	Proteogenomic Characterization of Ovarian HGSC Implicates Mitotic Kinases, Replication Stress in Observed Chromosomal Instability. <i>Cell Reports Medicine</i> , 2020, 1, 100004.	3.3	46
4255	Neuronal Calcium Sensor GCAP1 Encoded by <i>GUCA1A</i> Exhibits Heterogeneous Functional Properties in Two Cases of Retinitis Pigmentosa. <i>ACS Chemical Neuroscience</i> , 2020, 11, 1458-1470.	1.7	8
4256	Somatic mutations in the DNA repairome in prostate cancers in African Americans and Caucasians. <i>Oncogene</i> , 2020, 39, 4299-4311.	2.6	30
4257	Curaçao diagnostic criteria for hereditary hemorrhagic telangiectasia is highly predictive of a pathogenic variant in ENG or ACVRL1 (HHT1 and HHT2). <i>Genetics in Medicine</i> , 2020, 22, 1201-1205.	1.1	37
4258	Variations in cag pathogenicity island genes of <i>Helicobacter pylori</i> from Latin American groups may influence neoplastic progression to gastric cancer. <i>Scientific Reports</i> , 2020, 10, 6570.	1.6	11
4259	Novel MYH8 mutations in 152 Han Chinese samples with ovarian endometriosis. <i>Gynecological Endocrinology</i> , 2020, 36, 632-635.	0.7	1

#	ARTICLE	IF	CITATIONS
4260	High Complexity and Degree of Genetic Variation in <i>Brettanomyces bruxellensis</i> Population. <i>Genome Biology and Evolution</i> , 2020, 12, 795-807.	1.1	18
4261	Molecular analysis and clinical diversity of distal hereditary motor neuropathy. <i>European Journal of Neurology</i> , 2020, 27, 1319-1326.	1.7	28
4262	Genetic Polymorphism of Experimentally Produced Forms of Arterial Hypertension. <i>Russian Journal of Genetics</i> , 2020, 56, 213-225.	0.2	2
4263	Characterisation of genetic regulatory effects for osteoporosis risk variants in human osteoclasts. <i>Genome Biology</i> , 2020, 21, 80.	3.8	36
4264	Early infantile epileptic-dyskinetic encephalopathy due to biallelic PIGP mutations. <i>Neurology: Genetics</i> , 2020, 6, e387.	0.9	26
4265	Exome-Wide Association Study Reveals Several Susceptibility Genes and Pathways Associated With Acute Coronary Syndromes in Han Chinese. <i>Frontiers in Genetics</i> , 2020, 11, 336.	1.1	3
4266	Genetic diagnosis of autoinflammatory disease patients using clinical exome sequencing. <i>European Journal of Medical Genetics</i> , 2020, 63, 103920.	0.7	15
4267	Cellular analysis of a novel mutation p. Ser287Tyr in TOR1A in late-onset isolated dystonia. <i>Neurobiology of Disease</i> , 2020, 140, 104851.	2.1	1
4268	Whole Exome Sequencing reveals NOTCH1 mutations in anaplastic large cell lymphoma and points to Notch both as a key pathway and a potential therapeutic target. <i>Haematologica</i> , 2021, 106, 1693-1704.	1.7	40
4269	Human X chromosome exome sequencing identifies <i>BCORL1</i> as contributor to spermatogenesis. <i>Journal of Medical Genetics</i> , 2021, 58, 56-65.	1.5	13
4270	A biallelic pathogenic variant in the <i>OGDH</i> gene results in a neurological disorder with features of a mitochondrial disease. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 388-400.	1.7	24
4271	Prevalence and associated phenotypes of DUSP6, IL17RD and SPRY4 variants in a large Chinese cohort with isolated hypogonadotropic hypogonadism. <i>Journal of Medical Genetics</i> , 2021, 58, 66-72.	1.5	5
4272	The identified clinical features of Parkinson's disease in homo-, heterozygous and digenic variants of PINK1. <i>Neurobiology of Aging</i> , 2021, 97, 146.e1-146.e13.	1.5	14
4273	Delineating the folding perturbations and molecular mechanisms of Thr-Ala 642 mutation in Rab-GTPase activating protein Akt substrate of 160kDa and its impact on the aetiology of diabetes. <i>Journal of Biomolecular Structure and Dynamics</i> , 2021, 39, 409-420.	2.0	2
4274	Carcinogen soil radon enrichment in a geothermal area: Case of G ⁴ zel [±] -Davutlar district of Aydı ⁿ city, western Turkey. <i>Ecotoxicology and Environmental Safety</i> , 2021, 208, 111466.	2.9	11
4275	Tunable agglomeration of Co ₃ O ₄ nanowires as the growing core for in-situ formation of Co ₂ NiO ₄ assembled with polyaniline-derived carbonaceous fibers as the high-performance asymmetric supercapacitors. <i>Journal of Alloys and Compounds</i> , 2021, 853, 157210.	2.8	47
4276	<i>STK11</i> ; p.F354L Germline Mutation in a Case of Multiple Gastrointestinal Tumors. <i>Case Reports in Gastroenterology</i> , 2021, 14, 547-553.	0.3	2
4278	Failure analysis of a welded stainless-steel piping system with premature pitting. <i>Engineering Failure Analysis</i> , 2021, 119, 104986.	1.8	7

#	ARTICLE	IF	CITATIONS
4279	Isolation of new secondary metabolites from the liana <i>Landolphia lucida</i> K. Schum. (Apocynaceae). <i>Phytochemistry Letters</i> , 2021, 41, 27-33.	0.6	5
4280	Space-filling curves for numerical approximation and visualization of solutions to systems of nonlinear inequalities with applications in robotics. <i>Applied Mathematics and Computation</i> , 2021, 390, 125660.	1.4	14
4281	Energy savings in greenhouses by transition from high-pressure sodium to LED lighting. <i>Applied Energy</i> , 2021, 281, 116019.	5.1	70
4282	A computational model applied to myocardial perfusion in the human heart: From large coronaries to microvasculature. <i>Journal of Computational Physics</i> , 2021, 424, 109836.	1.9	23
4283	Characterization of four major degradation products in metformin by 2D LC-QTOF/MS/MS. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2021, 192, 113662.	1.4	8
4284	A size-dependent financial evaluation of green hydrogen-oxygen co-production. <i>Renewable Energy</i> , 2021, 163, 2165-2177.	4.3	32
4285	Bifunctional NiAlFe LDH-coated membrane for oil-in-water emulsion separation and photocatalytic degradation of antibiotic. <i>Science of the Total Environment</i> , 2021, 751, 141660.	3.9	41
4286	Evolution between 2008 and 2018 of mothers'™ perception regarding vaccination and infant vaccine coverage in France. <i>Infectious Diseases Now</i> , 2021, 51, 153-158.	0.7	4
4287	Evolution of the mammalian insulin (Ins) gene; Changes in proteolytic processing. <i>Peptides</i> , 2021, 135, 170435.	1.2	6
4288	Lamb wave-based mapping of plate structures via frontier exploration. <i>Ultrasonics</i> , 2021, 110, 106282.	2.1	20
4290	Regulatory role of microRNAs on PTEN signaling. <i>Biomedicine and Pharmacotherapy</i> , 2021, 133, 110986.	2.5	52
4291	Capital flows under global uncertainties: Evidence from Turkey. <i>Borsa Istanbul Review</i> , 2021, 21, 175-185.	2.4	9
4292	Hydro-mechanical modeling of the first and second hydraulic stimulations in a fractured geothermal reservoir in Pohang, South Korea. <i>Geothermics</i> , 2021, 89, 101982.	1.5	14
4293	Multi-wall carbon nanotubes decorated MnCo ₂ O _{4.5} hexagonal nanoplates with enhanced electrochemical behavior for high-performance electrochemical capacitors. <i>Journal of Industrial and Engineering Chemistry</i> , 2021, 94, 292-301.	2.9	14
4294	Pricing strategy and coordination of automobile manufacturers based on government intervention and carbon emission reduction. <i>Energy Policy</i> , 2021, 148, 111919.	4.2	77
4295	Can microaeration boost the biotransformation of parabens in high-rate anaerobic systems?. <i>Chemical Engineering Research and Design</i> , 2021, 145, 255-261.	2.7	11
4296	Large-scale genome-wide association study, using historical data, identifies conserved genetic architecture of cyanogenic glucoside content in cassava (<i>Manihot esculenta</i> Crantz) root. <i>Plant Journal</i> , 2021, 105, 754-770.	2.8	26
4297	Different approaches to long-term treatment of aHUS due to MCP mutations: a multicenter analysis. <i>Pediatric Nephrology</i> , 2021, 36, 463-471.	0.9	6

#	ARTICLE	IF	CITATIONS
4298	Palmoplantar keratoderma caused by a missense variant in <i>CTSB</i> encoding cathepsin B. <i>Clinical and Experimental Dermatology</i> , 2021, 46, 103-108.	0.6	5
4299	Clinically and biologically relevant subgroups of Wilms tumour defined by genomic and epigenomic analyses. <i>British Journal of Cancer</i> , 2021, 124, 437-446.	2.9	16
4300	Prospective phenotyping of long-term survivors of generalized arterial calcification of infancy (GACI). <i>Genetics in Medicine</i> , 2021, 23, 396-407.	1.1	44
4301	RMDisease: a database of genetic variants that affect RNA modifications, with implications for epitranscriptome pathogenesis. <i>Nucleic Acids Research</i> , 2021, 49, D1396-D1404.	6.5	65
4302	CARD8 polymorphism rs2043211 protects against noise-induced hearing loss by causing the dysfunction of CARD8 protein. <i>Environmental Science and Pollution Research</i> , 2021, 28, 8626-8636.	2.7	7
4303	Study on sleep-wake disorders in patients with genetic and non-genetic amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 96-102.	0.9	8
4304	Mutations in membrane fusion subunit of spike glycoprotein play crucial role in the recent outbreak of COVID-19. <i>Journal of Medical Virology</i> , 2021, 93, 2790-2798.	2.5	9
4305	Novel loss-of-function variants in <i>DNAH17</i> cause multiple morphological abnormalities of the sperm flagella in humans and mice. <i>Clinical Genetics</i> , 2021, 99, 176-186.	1.0	26
4306	Frequent loss of BTG1 activity and impaired interactions with the Caf1 subunit of the Ccr4-Not deadenylase in non-Hodgkin lymphoma. <i>Leukemia and Lymphoma</i> , 2021, 62, 281-290.	0.6	9
4307	Disease-specific variant pathogenicity prediction significantly improves variant interpretation in inherited cardiac conditions. <i>Genetics in Medicine</i> , 2021, 23, 69-79.	1.1	39
4309	Influence of co-existing social isolation and homebound status on medical care utilization and expenditure among older adults in Japan. <i>Archives of Gerontology and Geriatrics</i> , 2021, 93, 104286.	1.4	10
4311	Robotic Urology: The Next Frontier. <i>Urologic Clinics of North America</i> , 2021, 48, i.	0.8	1
4313	The results of optical guidance of flowing particles to the probe volume of Dual-Beam Velocimetry. <i>Optik</i> , 2021, 225, 165771.	1.4	3
4314	Smartphone sensing of social interactions in people with and without schizophrenia. <i>Journal of Psychiatric Research</i> , 2021, 137, 613-620.	1.5	39
4315	Direct demand-pull and indirect certification effects of public procurement for innovation. <i>Technovation</i> , 2021, 101, 102198.	4.2	13
4316	Characterizing the use of anticoagulants in children using the American Thrombosis and Hemostasis Network Dataset (ATHNdataset). <i>Thrombosis Research</i> , 2021, 197, 84-87.	0.8	9
4317	Genotoxicity, oxidative stress, and biochemical biomarkers of exposure to green synthesized cadmium nanoparticles in <i>Oreochromis niloticus</i> (L.). <i>Comparative Biochemistry and Physiology Part - C: Toxicology and Pharmacology</i> , 2021, 242, 108942.	1.3	15
4318	Flexural failure of fabric reinforced cementitious mortar (FRCM) plates under punctual loads: Experimental test, analytical approach and numerical simulation. <i>Construction and Building Materials</i> , 2021, 272, 121651.	3.2	4

#	ARTICLE	IF	CITATIONS
4319	Three-dimensional modeling on lightning induced overvoltage for photovoltaic arrays installed on mountain. <i>Journal of Cleaner Production</i> , 2021, 288, 125084.	4.6	16
4320	Evaluation of tensile strength of painted steel with local corrosion at structural connections. <i>Journal of Constructional Steel Research</i> , 2021, 177, 106449.	1.7	6
4321	Re: Hoffer et al.: Update on intraocular lens power calculation study protocols: the better way to design and report clinical trials (Ophthalmology. 2020 Jul 9 [Epub ahead of print]). <i>Ophthalmology</i> , 2021, 128, e17-e18.	2.5	5
4322	A rare cause of syndromic short stature: 3M syndrome in three families. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 461-468.	0.7	7
4323	Pathogenic variants causing ABL1 malformation syndrome cluster in a myristoyl-binding pocket and increase tyrosine kinase activity. <i>European Journal of Human Genetics</i> , 2021, 29, 593-603.	1.4	7
4324	Identification and functional study of genetic polymorphisms in cyclic nucleotide phosphodiesterase 3A (PDE3A). <i>Annals of Human Genetics</i> , 2021, 85, 80-91.	0.3	3
4325	A bioinformatic approach to investigating cytokine genes and their receptor variants in relation to COVID-19 progression. <i>International Journal of Immunogenetics</i> , 2021, 48, 211-218.	0.8	17
4326	Identification of a novel missense c.386G>A variant in a boy with the POMGNT1-related muscular dystrophy-dystroglycanopathy. <i>Acta Neurologica Belgica</i> , 2021, 121, 143-151.	0.5	11
4327	A novel mutation in the aspartate beta-hydroxylase (ASPH) gene is associated with a rare form of Traboulsi syndrome. <i>Ophthalmic Genetics</i> , 2021, 42, 28-34.	0.5	5
4328	Predictive significance of selected gene mutations in relapsed and refractory chronic lymphocytic leukemia patients treated with ibrutinib. <i>European Journal of Haematology</i> , 2021, 106, 320-326.	1.1	2
4329	Rare variants discovery by extensive whole-genome sequencing of the Han Chinese population in Taiwan: Applications to cardiovascular medicine. <i>Journal of Advanced Research</i> , 2021, 30, 147-158.	4.4	13
4330	Molecular docking simulation reveals ACE2 polymorphisms that may increase the affinity of ACE2 with the SARS-CoV-2 Spike protein. <i>Biochimie</i> , 2021, 180, 143-148.	1.3	53
4331	The Frailty Syndrome. <i>Critical Care Clinics</i> , 2021, 37, 151-174.	1.0	7
4332	The fold illusion: The origins and implications of ogives on silicic lavas. <i>Earth and Planetary Science Letters</i> , 2021, 553, 116643.	1.8	9
4333	Clinical heterogeneity of the SLC26A4 gene in UAE patients with hearing loss and bioinformatics investigation of DFNB4/Pendred syndrome missense mutations. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2021, 140, 110467.	0.4	10
4334	Euler scheme for density dependent stochastic differential equations. <i>Journal of Differential Equations</i> , 2021, 274, 996-1014.	1.1	6
4335	La pandemia COVID-19 y su repercusión en la unidad de radiología intervencionista: nuestra experiencia. <i>Radiología</i> , 2021, 63, 170-179.	0.3	1
4336	Performance of DG methods based on different variables for low Mach number flows. <i>Communications in Nonlinear Science and Numerical Simulation</i> , 2021, 95, 105580.	1.7	1

#	ARTICLE	IF	CITATIONS
4337	Outcomes following potentially curative therapies for older patients with metastatic colorectal cancer. <i>European Journal of Surgical Oncology</i> , 2021, 47, 591-596.	0.5	0
4338	Rapid misalignment correction method in reflective fourier ptychographic microscopy for full field of view reconstruction. <i>Optics and Lasers in Engineering</i> , 2021, 138, 106418.	2.0	11
4339	Novel variants broaden the phenotypic spectrum of PLEKHA7-associated neuropathies. <i>European Journal of Neurology</i> , 2021, 28, 1344-1355.	1.7	4
4340	INSAID Variant Classification and Eurofever Criteria Guide Optimal Treatment Strategy in Patients with TRAPS: Data from the Eurofever Registry. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 783-791.e4.	2.0	16
4341	Chemoplasticity of the polar lipid profile of the microalgae <i>Chlorella vulgaris</i> grown under heterotrophic and autotrophic conditions. <i>Algal Research</i> , 2021, 53, 102128.	2.4	24
4342	On a reduced order modeling of the nuclear reactor dynamics. <i>Applied Mathematics and Computation</i> , 2021, 393, 125819.	1.4	3
4343	Disorder engineering in transition metal dichalcogenides toward efficient high current density reduction electrocatalysts. <i>Current Opinion in Electrochemistry</i> , 2021, 25, 100639.	2.5	4
4344	Effect of matrix on aromatics production by cracking and dehydrocyclization of n-pentane using Ga ion-exchanged ZSM-5-alumina composite catalysts. <i>Fuel Processing Technology</i> , 2021, 213, 106679.	3.7	27
4345	Mutation analysis of <i>MFSD8</i> in an amyotrophic lateral sclerosis cohort from mainland China. <i>European Journal of Neuroscience</i> , 2021, 53, 1197-1206.	1.2	2
4346	A multi-institutional study of patient-derived gender-based discrimination experienced by resident physicians. <i>American Journal of Surgery</i> , 2021, 221, 309-314.	0.9	6
4347	Onset of Nucleate Boiling Model for Rectangular Upward Narrow Channel: CFD Based Approach. <i>International Journal of Heat and Mass Transfer</i> , 2021, 165, 120715.	2.5	16
4348	Microhotplate gas sensors incorporated with Al electrodes and 3D hierarchical structured PdO/PdO ₂ -SnO ₂ :Sb materials for sensitive VOC detection. <i>Sensors and Actuators B: Chemical</i> , 2021, 329, 128984.	4.0	23
4349	Happy analysts. <i>Accounting, Organizations and Society</i> , 2021, 90, 101199.	1.4	12
4350	A parametric divergence-free vector field method for the optimization of composite structures with curvilinear fibers. <i>Computer Methods in Applied Mechanics and Engineering</i> , 2021, 373, 113574.	3.4	20
4351	“Ripe for decision” Tiering in environmental assessment. <i>Environmental Impact Assessment Review</i> , 2021, 87, 106520.	4.4	13
4352	Graphs that contain multiply transitive matchings. <i>European Journal of Combinatorics</i> , 2021, 92, 103236.	0.5	0
4353	Reducing the cadmium, inorganic arsenic and dimethylarsinic acid content of rice through food-safe chemical cooking pre-treatment. <i>Food Chemistry</i> , 2021, 338, 127842.	4.2	13
4354	The epidemiology of atrial fibrillation in Chinese postmenopausal women and its association with age of menopause. <i>Maturitas</i> , 2021, 143, 151-156.	1.0	5

#	ARTICLE	IF	CITATIONS
4355	EDITORIAL COMMENT. Urology, 2021, 147, 210-211.	0.5	0
4356	Patisiran in hereditary transthyretin-mediated amyloidosis. Lancet Neurology, The, 2021, 20, 21-23.	4.9	9
4357	Potential pathways of zinc deficiency-promoted tumorigenesis. Biomedicine and Pharmacotherapy, 2021, 133, 110983.	2.5	21
4358	Competitive oxidation of methane and C_2 hydrocarbons discerned by isotopic labeling and laser absorption spectroscopy of CO isotopologues in shock-heated mixtures. Combustion and Flame, 2021, 224, 54-65.	2.8	7
4359	Refinement of coding SNPs in the human aryl hydrocarbon receptor gene using ISNPranker: An integrative-SNP ranking web-tool. Computational Biology and Chemistry, 2021, 90, 107416.	1.1	2
4360	Offline ion source for laser spectroscopy of RI at the SLOWRI. Nuclear Instruments & Methods in Physics Research B, 2021, 486, 48-54.	0.6	5
4361	A study of the role of forceful behaviour in evacuations via microscopic modelling of evacuation drills. Safety Science, 2021, 134, 105018.	2.6	6
4362	Preservation Rhinoplasty. Facial Plastic Surgery Clinics of North America, 2021, 29, i.	0.9	0
4364	Effects of passenger load, road grade, and congestion level on real-world fuel consumption and emissions from compressed natural gas and diesel urban buses. Applied Energy, 2021, 282, 116195.	5.1	57
4365	Carbonation characteristics of cement-based materials under the uniform distribution of pore water. Construction and Building Materials, 2021, 275, 121450.	3.2	12
4366	"Now it is mostly done through stashes, to do it in person one has to trust you": Understanding the retail injection drug market in Dnipro, Ukraine. International Journal of Drug Policy, 2021, 87, 102988.	1.6	5
4367	Mapping synergies and trade-offs between energy and the sustainable development goals: A case study of off-grid solar energy in Rwanda. Energy Policy, 2021, 149, 112028.	4.2	60
4368	Area deprivation, perceived neighbourhood cohesion and mental health at older ages: A cross lagged analysis of UK longitudinal data. Health and Place, 2021, 67, 102470.	1.5	19
4369	X-linked cellular mosaicism underlies age-dependent occurrence of seizure-like events in mouse models of CDKL5 deficiency disorder. Neurobiology of Disease, 2021, 148, 105176.	2.1	21
4370	A review on applications of artificial intelligence in modeling and optimization of laser beam machining. Optics and Laser Technology, 2021, 135, 106721.	2.2	53
4371	Age-proofing a traffic saturated metropolis – Evaluating the influences on walking behaviour in older adults in Ho Chi Minh City. Travel Behaviour & Society, 2021, 23, 1-12.	2.4	8
4372	Design of tethered bilayer lipid membranes, using wet chemistry via aryldiazonium sulfonic acid spontaneous grafting on silicon and chrome. Colloids and Surfaces B: Biointerfaces, 2021, 197, 111427.	2.5	5
4373	Optimizing Safety for Surgical Patients Undergoing Interhospital Transfer. Surgical Clinics of North America, 2021, 101, 57-69.	0.5	3

#	ARTICLE	IF	CITATIONS
4374	SQSTM1 gene as a potential genetic modifier of CADASIL phenotype. <i>Journal of Neurology</i> , 2021, 268, 1453-1460.	1.8	2
4375	A combined theoretical and numerical modeling study of cyclic nonlinear response of sandy seabed. <i>Ocean Engineering</i> , 2021, 219, 108348.	1.9	4
4376	First report on the microbial communities of the wild and planted raspberry rhizosphere – A statement on the taxa, processes and a new indicator of functional diversity. <i>Ecological Indicators</i> , 2021, 121, 107117.	2.6	10
4377	The FNS-based analysis of precursors and cross-correlations in EEG signal related to an imaginary motor task. <i>Biomedical Signal Processing and Control</i> , 2021, 64, 102315.	3.5	1
4378	An experimental and kinetic modeling study of cyclopentane and dimethyl ether blends. <i>Combustion and Flame</i> , 2021, 225, 255-271.	2.8	19
4379	Improved desorption temperature of magnesium hydride via multi-layering Mg/Fe thin film. <i>International Journal of Hydrogen Energy</i> , 2021, 46, 4181-4187.	3.8	7
4380	Country institutional environments and international strategy: A review and analysis of the research. <i>Journal of International Management</i> , 2021, 27, 100811.	2.4	35
4381	OncoVar: an integrated database and analysis platform for oncogenic driver variants in cancers. <i>Nucleic Acids Research</i> , 2021, 49, D1289-D1301.	6.5	64
4382	Failure mechanism of geopolymer composite lightweight sandwich panel under flexural and edgewise compressive loads. <i>Construction and Building Materials</i> , 2021, 270, 121496.	3.2	8
4383	Dynamic model to predict the association between air quality, COVID-19 cases, and level of lockdown. <i>Environmental Pollution</i> , 2021, 268, 115920.	3.7	27
4384	Research trends: Tropical dry forests: The neglected research agenda?. <i>Forest Policy and Economics</i> , 2021, 122, 102333.	1.5	18
4385	Thermally induced spin transition in Fe(pyrazine)[Fe(CN)5NO]. <i>Journal of Physics and Chemistry of Solids</i> , 2021, 150, 109843.	1.9	14
4386	Morphological modulation to improve thermoelectric performances of PEDOT:PSS films by DMSO vapor post-treatment. <i>Synthetic Metals</i> , 2021, 271, 116628.	2.1	18
4387	Analysis of 50,000 exome-sequenced UK Biobank subjects fails to identify genes influencing probability of developing a mood disorder resulting in psychiatric referral. <i>Journal of Affective Disorders</i> , 2021, 281, 216-219.	2.0	8
4388	Genetic consequences of long-term small effective population size in the critically endangered pygmy hog. <i>Evolutionary Applications</i> , 2021, 14, 710-720.	1.5	19
4389	Clinical, Biochemical, and Molecular Characterization of Metachromatic Leukodystrophy Among Egyptian Pediatric Patients: Expansion of the ARSA Mutational Spectrum. <i>Journal of Molecular Neuroscience</i> , 2021, 71, 1112-1130.	1.1	1
4390	Red and blond Mangalitza pigs display a signature of divergent directional selection in the <i>SLC45A2</i> gene. <i>Animal Genetics</i> , 2021, 52, 66-77.	0.6	3
4391	Spectrum-frequency and genotype-phenotype analysis of rhodopsin variants. <i>Experimental Eye Research</i> , 2021, 203, 108405.	1.2	14

#	ARTICLE	IF	CITATIONS
4392	Novel disease-causing variants in a cohort of Iranian patients with metachromatic leukodystrophy and in silico analysis of their pathogenicity. <i>Clinical Neurology and Neurosurgery</i> , 2021, 201, 106448.	0.6	6
4393	Clinical and Genetic Analysis of 63 Families Demonstrating Early and Advanced Characteristic Fundus as the Signature of CRB1 Mutations. <i>American Journal of Ophthalmology</i> , 2021, 223, 160-168.	1.7	10
4394	Genetic characterization of the Albanian Gaucher disease patient population. <i>JIMD Reports</i> , 2021, 57, 52-57.	0.7	2
4395	Early truncation of the N-terminal variable region of <i>EYA4</i> gene causes dominant hearing loss without cardiac phenotype. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1569.	0.6	7
4396	Nitrous oxide emissions in response to long-term application of the nitrification inhibitor DMPP in an acidic luvisol. <i>Applied Soil Ecology</i> , 2021, 159, 103861.	2.1	9
4397	CeO ₂ /Co ₃ O ₄ porous nanosheet prepared using rose petal as biotemplate for photo-catalytic degradation of organic contaminants. <i>Applied Surface Science</i> , 2021, 543, 148677.	3.1	40
4398	Interoceptive Awareness and Suicidal Ideation in a Clinical Eating Disorder Sample: The Role of Body Trust. <i>Behavior Therapy</i> , 2021, 52, 1105-1113.	1.3	14
4399	Crayfish shell biochar for the mitigation of Pb contaminated water and soil: Characteristics, mechanisms, and applications. <i>Environmental Pollution</i> , 2021, 271, 116308.	3.7	52
4400	A novel method to estimate cellular internalization of nanoparticles into gram-negative bacteria: Non-lytic removal of outer membrane and cell wall. <i>NanoImpact</i> , 2021, 21, 100283.	2.4	5
4401	Paleostress variation during the same regional deformation in the Eastern Dharwar Craton (southern India). <i>Journal of Structural Geology</i> , 2021, 143, 104268.	1.0	2
4402	Associations between brain amyloid accumulation and the use of angiotensin-converting enzyme inhibitors versus angiotensin receptor blockers. <i>Neurobiology of Aging</i> , 2021, 100, 22-31.	1.5	22
4403	Monitoring time-varying terrestrial water storage changes using daily GNSS measurements in Yunnan, southwest China. <i>Remote Sensing of Environment</i> , 2021, 254, 112249.	4.6	43
4404	<i>Biallelic ZNF335</i> mutations cause basal ganglia abnormality with progressive cerebral/cerebellar atrophy. <i>Journal of Neurogenetics</i> , 2021, 35, 23-28.	0.6	2
4405	Recurrent <i>PROC</i> and novel <i>PROS1</i> mutations in Vietnamese patients diagnosed with idiopathic deep venous thrombosis. <i>International Journal of Laboratory Hematology</i> , 2021, 43, 266-272.	0.7	10
4406	Pseudo-LiDAR point cloud magnification. <i>Neurocomputing</i> , 2021, 422, 129-138.	3.5	6
4407	Novel mutations in ATP13A2 associated with mixed neurological presentations and iron toxicity due to nonsense-mediated decay. <i>Brain Research</i> , 2021, 1750, 147167.	1.1	7
4408	Clinical, biochemical and molecular findings of 24 Brazilian patients with glutaric acidemia type 1: 4 novel mutations in the GCDH gene. <i>Metabolic Brain Disease</i> , 2021, 36, 205-212.	1.4	7
4409	Air pollution and cause-specific mortality: A comparative study of urban and rural areas in China. <i>Chemosphere</i> , 2021, 262, 127884.	4.2	52

#	ARTICLE	IF	CITATIONS
4410	Beyond monogenetic rare variants: tackling the low rate of genetic diagnoses in predominantly antibody deficiency. <i>Cellular and Molecular Immunology</i> , 2021, 18, 588-603.	4.8	17
4411	Genomic analysis of low-grade serous ovarian carcinoma to identify key drivers and therapeutic vulnerabilities. <i>Journal of Pathology</i> , 2021, 253, 41-54.	2.1	54
4412	Molecular characteristics of varicocele: integration of whole-exome and transcriptome sequencing. <i>Fertility and Sterility</i> , 2021, 115, 363-372.	0.5	8
4413	Comprehensive functional annotation of susceptibility variants identifies genetic heterogeneity between lung adenocarcinoma and squamous cell carcinoma. <i>Frontiers of Medicine</i> , 2021, 15, 275-291.	1.5	21
4414	Molecular dynamics simulations for genetic interpretation in protein coding regions: where we are, where to go and when. <i>Briefings in Bioinformatics</i> , 2021, 22, 3-19.	3.2	30
4415	Structural and functional alterations of nitric oxide synthase 3 due to missense variants associate with high-altitude pulmonary edema through dynamic study. <i>Journal of Biomolecular Structure and Dynamics</i> , 2021, 39, 294-309.	2.0	10
4416	Phenotypic continuum between Waardenburg syndrome and idiopathic hypogonadotropic hypogonadism in humans with SOX10 variants. <i>Genetics in Medicine</i> , 2021, 23, 629-636.	1.1	9
4417	Detection of genes positively selected in Cuban Anolis lizards that naturally inhabit hot and open areas and currently thrive in urban areas. <i>Ecology and Evolution</i> , 2021, 11, 1719-1728.	0.8	7
4418	Whole exome sequencing and trio analysis to broaden the variant spectrum of genes in idiopathic hypogonadotropic hypogonadism. <i>Asian Journal of Andrology</i> , 2021, 23, 288.	0.8	5
4419	Comprehensive Mutation Analysis and Report of 12 Novel Mutations in a Cohort of Patients with Spinal Muscular Atrophy in Iran. <i>Journal of Molecular Neuroscience</i> , 2021, 71, 2281-2298.	1.1	9
4421	Investigating the causal role of MRE11A p.E506* in breast and ovarian cancer. <i>Scientific Reports</i> , 2021, 11, 2409.	1.6	5
4422	Mining Grapevine Downy Mildew Susceptibility Genes: A Resource for Genomics-Based Breeding and Tailored Gene Editing. <i>Biomolecules</i> , 2021, 11, 181.	1.8	15
4423	Analysis of 200,000 Exome-Sequenced UK Biobank Subjects Implicates Genes Involved in Increased and Decreased Risk of Hypertension. <i>Pulse</i> , 2021, 9, 17-29.	0.9	10
4424	GuidePro: a multi-source ensemble predictor for prioritizing sgRNAs in CRISPR/Cas9 protein knockouts. <i>Bioinformatics</i> , 2021, 37, 134-136.	1.8	7
4425	Two-stage-vote ensemble framework based on integration of mutation data and gene interaction network for uncovering driver genes. <i>Briefings in Bioinformatics</i> , 2022, 23, .	3.2	5
4426	Sharing genetic variants with the NGS pipeline is essential for effective genomic data sharing and reproducibility in health information exchange. <i>Scientific Reports</i> , 2021, 11, 2268.	1.6	1
4427	Genetic Variants and Impact in PDE6B Rod-Cone Dystrophy. <i>Essentials in Ophthalmology</i> , 2021, , 197-206.	0.0	1
4429	Oncogenic potential of ATAD2 in stomach cancer and insights into the protein-protein interactions at its AAA-ATPase domain and bromodomain. <i>Journal of Biomolecular Structure and Dynamics</i> , 2022, 40, 5606-5622.	2.0	5

#	ARTICLE	IF	CITATIONS
4431	Novel Compound Heterozygous Mutation c.3955_3958dup and c.5825C>#x3e;T in the <i>ATM</i> Gene: Clinical Evidence of Ataxia-Telangiectasia and Cancer in a Peruvian Family. <i>Molecular Syndromology</i> , 2021, 12, 289-293.	0.3	1
4432	Solving Image Processing Critical Problems Using Machine Learning. <i>Studies in Big Data</i> , 2021, , 213-248.	0.8	1
4433	The genetic landscape of polycystic kidney disease in Ireland. <i>European Journal of Human Genetics</i> , 2021, 29, 827-838.	1.4	11
4434	Mechanisms for Development of Ciprofloxacin Resistance in a Clinical Isolate of <i>Pseudomonas aeruginosa</i> . <i>Frontiers in Microbiology</i> , 2020, 11, 598291.	1.5	16
4435	De Novo Mutation and Rapid Protein (Co-)evolution during Meiotic Adaptation in <i>Arabidopsis arenosa</i>. <i>Molecular Biology and Evolution</i> , 2021, 38, 1980-1994.	3.5	18
4436	Putative second hit rare genetic variants in families with seemingly GBA-associated Parkinsonâ€™s disease. <i>Npj Genomic Medicine</i> , 2021, 6, 2.	1.7	11
4437	Association of mutation in PTPN14 gene and gingival fibromatosis with distinctive facies: a novel finding in whole exome sequencing. <i>Clinical Dysmorphology</i> , 2021, 30, 93-99.	0.1	2
4438	Identification of a novel <i>SUOX</i> pathogenic variants as the cause of isolated sulfite oxidase deficiency in a Chinese pedigree. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1590.	0.6	3
4439	MVP predicts theÂpathogenicity of missense variants by deep learning. <i>Nature Communications</i> , 2021, 12, 510.	5.8	85
4440	The computational approach to variant interpretation. , 2021, , 89-119.		6
4441	Evaluation of clinical findings and neurofibromatosis type 1 bright objects on brain magnetic resonance images of 60 Turkish patients with NF1 gene variants. <i>Neurological Sciences</i> , 2021, 42, 2045-2057.	0.9	1
4442	Epidermolysis bullosa simplex due to biâ€allelic <i>DST</i> mutations: Case series and review of the literature. <i>Pediatric Dermatology</i> , 2021, 38, 436-441.	0.5	9
4443	Text Analytics and Mixed Feature Extraction in Ovarian Cancer Clinical and Genetic Data. <i>IEEE Access</i> , 2021, 9, 58034-58051.	2.6	2
4444	Genetics of Cellular Immunodeficiencies. <i>Rare Diseases of the Immune System</i> , 2021, , 5-24.	0.1	0
4445	Molecular Genetics in the Next Generation Sequencing Era. , 2021, , 215-230.		0
4446	Novel hereditary angioedema linked with a heparan sulfate 3-O-sulfotransferase 6 gene mutation. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 1041-1048.	1.5	65
4447	Expanding the clinical and genetic spectrum of FDXR deficiency by functional validation of variants of uncertain significance. <i>Human Mutation</i> , 2021, 42, 310-319.	1.1	11
4448	Describing variability in the tyrosinase (<i>TYR</i>) gene, the <i>albino</i> coat colour locus, in domestic and wild European rabbits. <i>Italian Journal of Animal Science</i> , 2021, 20, 181-187.	0.8	7

#	ARTICLE	IF	CITATIONS
4449	Genetic variant effect prediction by supervised nonnegative matrix tri-factorization. <i>Molecular Omics</i> , 2021, 17, 740-751.	1.4	1
4450	Vitamin D Metabolism or Action. , 2021, , 335-372.		0
4451	Gene spectrum and clinical traits of 10 patients with primary carnitine deficiency. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1583.	0.6	1
4454	Relevance of pathogenicity prediction tools in human RYR1 variants of unknown significance. <i>Scientific Reports</i> , 2021, 11, 3445.	1.6	4
4455	Clinical and Molecular Characterization of Achromatopsia Patients: A Longitudinal Study. <i>International Journal of Molecular Sciences</i> , 2021, 22, 1681.	1.8	19
4456	Deleterious variants in X-linked CFAP47 induce asthenoteratozoospermia and primary male infertility. <i>American Journal of Human Genetics</i> , 2021, 108, 309-323.	2.6	74
4457	Prognostic value of integrated cytogenetic, somatic variation, and copy number variation analyses in Korean patients with newly diagnosed multiple myeloma. <i>PLoS ONE</i> , 2021, 16, e0246322.	1.1	5
4458	<i>In silico</i> screening and molecular dynamics simulation of deleterious PAH mutations responsible for phenylketonuria genetic disorder. <i>Proteins: Structure, Function and Bioinformatics</i> , 2021, 89, 683-696.	1.5	3
4459	Clinical characterization and identification of rare genetic variants in atypical hemolytic uremic syndrome: A Swedish retrospective observational study. <i>Therapeutic Apheresis and Dialysis</i> , 2021, 25, 988-1000.	0.4	4
4460	A founder mutation in the <i>PLPBP</i> gene in families from Saguenay-Lac-St-Jean region affected by a pyridoxine-dependent epilepsy. <i>JIMD Reports</i> , 2021, 69, 32-41.		7
4461	Measurable residual disease in elderly acute myeloid leukemia: results from the PETHEMA-FLUGAZA phase 3 clinical trial. <i>Blood Advances</i> , 2021, 5, 760-770.	2.5	18
4464	Describing variability in pig genes involved in coronavirus infections for a One Health perspective in conservation of animal genetic resources. <i>Scientific Reports</i> , 2021, 11, 3359.	1.6	9
4465	Whole genome sequencing of 45 Japanese patients with intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1468-1480.	0.7	13
4466	Polymorphisms in canine immunoglobulin heavy chain gene cluster: a double-edged sword for diabetes mellitus in the dog. <i>Animal Genetics</i> , 2021, 52, 333-341.	0.6	2
4467	Novel SCN5A variants identified in a group of Iranian Brugada syndrome patients. <i>Functional and Integrative Genomics</i> , 2021, 21, 331-340.	1.4	1
4468	Optic Atrophy and Inner Retinal Thinning in CACNA1F-Related Congenital Stationary Night Blindness. <i>Genes</i> , 2021, 12, 330.	1.0	6
4470	<i>In silico</i> screening and identification of deleterious missense SNPs along with their effects on CD-209 gene: An insight to CD-209 related-diseases. <i>PLoS ONE</i> , 2021, 16, e0247249.	1.1	6
4471	Whole Locus Sequencing Identifies a Prevalent Founder Deep Intronic RPRIP1 Pathologic Variant in the French Leber Congenital Amaurosis Cohort. <i>Genes</i> , 2021, 12, 287.	1.0	3

#	ARTICLE	IF	CITATIONS
4472	<i>CHM</i> mutation spectrum and disease: An update at the time of human therapeutic trials. Human Mutation, 2021, 42, 323-341.	1.1	8
4473	A unique missense variant in the E1A-binding protein P400 gene is implicated in schizophrenia by whole-exome sequencing and mutant mouse models. Translational Psychiatry, 2021, 11, 132.	2.4	0
4474	Identification of novel, clonally stable, somatic mutations targeting transcription factors PAX5 and NKX2-3, the epigenetic regulator LRIF1, and BRAF in a case of atypical B-cell chronic lymphocytic leukemia harboring a t(14;18)(q32;q21). Journal of Physical Education and Sports Management, 2021, 7, a005934.	0.5	1
4477	A novel mutation in the COL12A1 gene. Gene, 2021, 768, 145266.	1.0	6
4478	Intrafamilial Phenotypic Variability Associated with the I1739V Mutation in the SCN9A Gene. Case Reports in Neurology, 2021, 13, 135-139.	0.3	2
4480	ABCA3 gene mutations shape the clinical profiles of severe unexplained respiratory distress syndrome in late preterm and term infants. Translational Pediatrics, 2021, 10, 350-358.	0.5	6
4482	hiPSC-Derived Epidermal Keratinocytes from Ichthyosis Patients Show Altered Expression of Cornification Markers. International Journal of Molecular Sciences, 2021, 22, 1785.	1.8	4
4483	Mutation spectrum of NOD2 reveals recessive inheritance as a main driver of Early Onset Crohn's Disease. Scientific Reports, 2021, 11, 5595.	1.6	29
4484	Next-generation sequencing for inborn errors of immunity. Human Immunology, 2021, 82, 871-882.	1.2	12
4485	Gene diagnosis and pedigree analysis of two Han ethnicity families with propionic acidemia in Fujian. Medicine (United States), 2021, 100, e24161.	0.4	2
4486	Multi-modal meta-analysis of cancer cell line omics profiles identifies ECHDC1 as a novel breast tumor suppressor. Molecular Systems Biology, 2021, 17, e9526.	3.2	8
4487	Candidate Markers of Olaparib Response from Genomic Data Analyses of Human Cancer Cell Lines. Cancers, 2021, 13, 1296.	1.7	3
4488	Functional Studies of Novel FOXL2 Variants in Chinese Families With Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome. Frontiers in Genetics, 2021, 12, 616112.	1.1	5
4489	Identification of candidate genes and pathways in retinopathy of prematurity by whole exome sequencing of preterm infants enriched in phenotypic extremes. Scientific Reports, 2021, 11, 4966.	1.6	7
4491	ETV4 plays a role on the primary events during the adenoma-adenocarcinoma progression in colorectal cancer. BMC Cancer, 2021, 21, 207.	1.1	10
4492	Nonframeshifting indel variations in polyalanine repeat of <sc><i>HOXD13</i></sc> gene underlies hereditary limb malformation for two Chinese families. Developmental Dynamics, 2021, 250, 1220-1228.	0.8	4
4493	Dravet syndrome-associated mutations in <i>GABRA1</i>, <i>GABRB2</i> and <i>GABRG2</i> define the genetic landscape of defects of GABAA receptors. Brain Communications, 2021, 3, fcab033.	1.5	21
4494	Understanding the Origins of Loss of Protein Function by Analyzing the Effects of Thousands of Variants on Activity and Abundance. Molecular Biology and Evolution, 2021, 38, 3235-3246.	3.5	65

#	ARTICLE	IF	CITATIONS
4495	Clinical and molecular findings in children and young adults with persistent low alkaline phosphatase concentrations. <i>Annals of Clinical Biochemistry</i> , 2021, 58, 000456322110001.	0.8	2
4496	Autosomal Recessive Rod-Cone Dystrophy Associated With Compound Heterozygous Variants in ARL3 Gene. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 635424.	1.8	18
4497	Co-occurrence of orofacial clefts and clubfoot phenotypes in a sub-Saharan African cohort: Whole-exome sequencing implicates multiple syndromes and genes. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1655.	0.6	3
4498	Evidence of synergism among three genetic variants in a patient with LMNA-related lipodystrophy and amyotrophic lateral sclerosis leading to a remarkable nuclear phenotype. <i>Molecular and Cellular Biochemistry</i> , 2021, 476, 2633-2650.	1.4	4
4499	Vitamin C and Vitamin D3 show strong binding with the amyloidogenic region of G555F mutant of Fibrinogen A alpha-chain associated with renal amyloidosis: proposed possible therapeutic intervention. <i>Molecular Diversity</i> , 2022, 26, 939-949.	2.1	6
4500	Novel splicing (c.6529-1G>T) and missense (c.1667G>A) mutations causing factor V deficiency. <i>Blood Coagulation and Fibrinolysis</i> , 2021, 32, 344-348.	0.5	0
4501	Classifying molecular phenotypes of <i>G6PC</i> variants for pathogenic properties and to guide therapeutic development. <i>JIMD Reports</i> , 2021, 60, 56-66.	0.7	2
4502	PARKIN, PINK1, and DJ1 analysis in early-onset Parkinson's disease in Ireland. <i>Irish Journal of Medical Science</i> , 2022, 191, 901-907.	0.8	6
4503	Significant impact of circulating tumour DNA mutations on survival in metastatic breast cancer patients. <i>Scientific Reports</i> , 2021, 11, 6761.	1.6	16
4505	A Missense Mutation in the KLF7 Gene Is a Potential Candidate Variant for Congenital Deafness in Australian Stumpy Tail Cattle Dogs. <i>Genes</i> , 2021, 12, 467.	1.0	5
4506	Missense Variant of Endoplasmic Reticulum Region of WFS1 Gene Causes Autosomal Dominant Hearing Loss without Syndromic Phenotype. <i>BioMed Research International</i> , 2021, 2021, 1-9.	0.9	4
4507	Variant Spectrum of Formin Homology 2 Domain-Containing 3 Gene in Chinese Patients With Hypertrophic Cardiomyopathy. <i>Journal of the American Heart Association</i> , 2021, 10, e018236.	1.6	10
4508	Polymorphisms of the matrix metalloproteinase genes are associated with essential hypertension in a Caucasian population of Central Russia. <i>Scientific Reports</i> , 2021, 11, 5224.	1.6	34
4509	Cardiovascular manifestations of intermediate and major hyperhomocysteinemia due to vitamin B12 and folate deficiency and/or inherited disorders of one-carbon metabolism: a 3.5-year retrospective cross-sectional study of consecutive patients. <i>American Journal of Clinical Nutrition</i> , 2021, 113, 1157-1167.	2.2	17
4510	Insights into adaption and growth evolution: a comparative genomics study on two distinct cattle breeds from Northern and Southern China. <i>Molecular Therapy - Nucleic Acids</i> , 2021, 23, 959-967.	2.3	9
4511	Mutational profiles of marker genes of cervical carcinoma in Bangladeshi patients. <i>BMC Cancer</i> , 2021, 21, 289.	1.1	4
4512	Low Diversity of Human Variation Despite Mostly Mild Functional Impact of De Novo Variants. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 635382.	1.6	2
4513	Mutation profile and immunoscore signature in thymic carcinomas: An exploratory study and review of the literature. <i>Thoracic Cancer</i> , 2021, 12, 1271-1278.	0.8	8

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4514	Computational prediction of protein subdomain stability in MYBPC3 enables clinical risk stratification in hypertrophic cardiomyopathy and enhances variant interpretation. <i>Genetics in Medicine</i> , 2021, 23, 1281-1287.	1.1	11
4515	Prediction of Functional Consequences of Missense Mutations in ANO4 Gene. <i>International Journal of Molecular Sciences</i> , 2021, 22, 2732.	1.8	3
4516	Identifying the causes of recurrent pregnancy loss in consanguineous couples using whole exome sequencing on the products of miscarriage with no chromosomal abnormalities. <i>Scientific Reports</i> , 2021, 11, 6952.	1.6	15
4517	A novel homozygous splice-site mutation in SCARB2 is associated with progressive myoclonic epilepsy with renal failure. <i>Neurological Sciences</i> , 2021, 42, 5077-5085.	0.9	3
4519	Whole-exome sequencing in 168 Korean patients with inherited retinal degeneration. <i>BMC Medical Genomics</i> , 2021, 14, 74.	0.7	24
4520	Rare and novel GNAS gene mutations in Chinese patients with thyroid cancer. <i>Precision Medical Sciences</i> , 2021, 10, 83-85.	0.1	1
4521	A recurring NFS1 pathogenic variant causes a mitochondrial disorder with variable intra-familial patient outcomes. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 26, 100699.	0.4	5
4522	A knockout mutation associated with juvenile paroxysmal dyskinesia in Markiesje dogs indicates SOD1 pleiotropy. <i>Human Genetics</i> , 2021, 140, 1547-1552.	1.8	9
4523	Contribution of coding/non-coding variants in NUS1 to late-onset sporadic Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2021, 84, 29-34.	1.1	2
4524	A rare case of an NLRP12-associated autoinflammatory disease. <i>European Journal of Medical Genetics</i> , 2021, 64, 104168.	0.7	5
4525	Genetic Regulation of Biomarkers as Stress Proxies in Dairy Cows. <i>Genes</i> , 2021, 12, 534.	1.0	3
4526	The Positivity of Phosphorylated STAT3 Is a Novel Marker for Favorable Prognosis in Germinal Center B-Cell Type of Diffuse Large B-Cell Lymphoma. <i>American Journal of Surgical Pathology</i> , 2021, 45, 832-840.	2.1	0
4527	Structural insight into mutations at 155 position of valosin containing protein (VCP) linked to inclusion body myopathy with Paget disease of bone and frontotemporal Dementia. <i>Saudi Journal of Biological Sciences</i> , 2021, 28, 2128-2138.	1.8	0
4528	Molecular epidemiology of non-syndromic autosomal recessive congenital ichthyosis in a Middle-Eastern population. <i>Experimental Dermatology</i> , 2021, 30, 1290-1297.	1.4	10
4529	The distinct morphological phenotypes of Southeast Asian aborigines are shaped by novel mechanisms for adaptation to tropical rainforests. <i>National Science Review</i> , 2022, 9, nwab072.	4.6	3
4530	Overexpression of a Cytochrome P450 Monooxygenase Involved in Orobanchol Biosynthesis Increases Susceptibility to Fusarium Head Blight. <i>Frontiers in Plant Science</i> , 2021, 12, 662025.	1.7	6
4531	Exome-wide scan identifies significant association of rs4788084 in IL27 promoter with increase in hepatic fat content among Indians. <i>Gene</i> , 2021, 775, 145431.	1.0	13
4533	Massive parallel sequencing in a family with rectal cancer. <i>Hereditary Cancer in Clinical Practice</i> , 2021, 19, 23.	0.6	3

#	ARTICLE	IF	CITATIONS
4534	Analysis of 200 000 exome-sequenced UK Biobank subjects illustrates the contribution of rare genetic variants to hyperlipidaemia. <i>Journal of Medical Genetics</i> , 2022, 59, 597-604.	1.5	12
4535	Accessing Livestock Resources in Ensembl. <i>Frontiers in Genetics</i> , 2021, 12, 650228.	1.1	3
4536	Phenotypic spectrum and genomics of undiagnosed arthrogryposis multiplex congenita. <i>Journal of Medical Genetics</i> , 2022, 59, 559-567.	1.5	25
4537	Identification of mutations in the ATP7B gene in 14 Wilson disease children. <i>Medicine (United States)</i> , 2021, 100, e25463.	0.4	1
4538	Comprehensive Study of Germline Mutations and Double-Hit Events in Esophageal Squamous Cell Cancer. <i>Frontiers in Oncology</i> , 2021, 11, 637431.	1.3	5
4539	Familial Psychosis Associated With a Missense Mutation at MACF1 Gene Combined With the Rare Duplications DUP3p26.3 and DUP16q23.3, Affecting the CNTN6 and CDH13 Genes. <i>Frontiers in Genetics</i> , 2021, 12, 622886.	1.1	3
4540	Analysis of whole genome sequenced cases and controls shows that the association of variants in <i>TOMM40</i> , <i>BCAM</i> , <i>NECTIN2</i> and <i>APOC1</i> with late onset Alzheimer's disease is driven by linkage disequilibrium with <i>APOE</i> $\mu_2/\mu_3/\mu_4$ alleles. <i>Journal of Neurogenetics</i> , 2021, 35, 59-66.	0.6	3
4541	Identification of homozygous mutations for hearing loss. <i>Gene</i> , 2021, 778, 145464.	1.0	2
4542	Investigation of Association of Rare, Functional Genetic Variants With Heavy Drinking and Problem Drinking in Exome Sequenced UK Biobank Participants. <i>Alcohol and Alcoholism</i> , 2021, , .	0.9	1
4543	Variants associated with urea cycle disorders in Japanese patients: Nationwide study and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2026-2036.	0.7	7
4545	Whole exome sequencing, in silico and functional studies confirm the association of the GJB2 mutation p.Cys169Tyr with deafness and suggest a role for the TMEM59 gene in the hearing process. <i>Saudi Journal of Biological Sciences</i> , 2021, 28, 4421-4429.	1.8	5
4546	A homozygous variant in <i>TBPL2</i> was identified in women with oocyte maturation defects and infertility. <i>Human Reproduction</i> , 2021, 36, 2011-2019.	0.4	14
4547	Screening for extremely rare pathogenic variants of monogenic diabetes using targeted panel sequencing. <i>Endocrine</i> , 2021, 73, 752-757.	1.1	3
4548	Incorporation of exome-based CNV analysis makes trio-WES a more powerful tool for clinical diagnosis in neurodevelopmental disorders: A retrospective study. <i>Human Mutation</i> , 2021, 42, 990-1004.	1.1	25
4549	Rare Missense Functional Variants at <i>COL4A1</i> and <i>COL4A2</i> in Sporadic Intracerebral Hemorrhage. <i>Neurology</i> , 2021, 97, .	1.5	6
4550	Molecular genetics with clinical characteristics of Leber congenital amaurosis in the Han population of western China. <i>Ophthalmic Genetics</i> , 2021, 42, 392-401.	0.5	6
4551	Imprecise Medicine: BRCA2 Variants of Uncertain Significance (VUS), the Challenges and Benefits to Integrate a Functional Assay Workflow with Clinical Decision Rules. <i>Genes</i> , 2021, 12, 780.	1.0	15
4552	ADDRESS: A Database of Disease-associated Human Variants Incorporating Protein Structure and Folding Stabilities. <i>Journal of Molecular Biology</i> , 2021, 433, 166840.	2.0	15

#	ARTICLE	IF	CITATIONS
4554	In Silico Predictions of KCNQ Variant Pathogenicity in Epilepsy. <i>Pediatric Neurology</i> , 2021, 118, 48-54.	1.0	2
4555	Whole-Exome Sequencing Reveals Rare Germline Mutations in Patients With Hemifacial Microsomia. <i>Frontiers in Genetics</i> , 2021, 12, 580761.	1.1	6
4556	Sequence Neighborhoods Enable Reliable Prediction of Pathogenic Mutations in Cancer Genomes. <i>Cancers</i> , 2021, 13, 2366.	1.7	6
4557	Single-cell transcriptomics trajectory and molecular convergence of clinically relevant mutations in Brugada syndrome. <i>American Journal of Physiology - Heart and Circulatory Physiology</i> , 2021, 320, H1935-H1948.	1.5	6
4558	Role of RNA in Molecular Diagnosis of MADD Patients. <i>Biomedicines</i> , 2021, 9, 507.	1.4	4
4559	Characterization of <i>PROM1</i> p.Arg373Cys Variant in a Cohort of Chinese Patients: Macular Dystrophy Plus Peripheral Bone-Spicule Degeneration. , 2021, 62, 19.		6
4560	Sudden Death without a Clear Cause after Comprehensive Investigation: An Example of Forensic Approach to Atypical/Uncertain Findings. <i>Diagnostics</i> , 2021, 11, 886.	1.3	8
4561	Identifying Susceptibility Loci for Cutaneous Squamous Cell Carcinoma Using a Fast Sequence Kernel Association Test. <i>Frontiers in Genetics</i> , 2021, 12, 657499.	1.1	2
4562	Expanding the Phenotypic and Genotypic Spectrum of Bietti Crystalline Dystrophy. <i>Genes</i> , 2021, 12, 713.	1.0	7
4563	Analysis of 200,000 exome-sequenced UK Biobank subjects fails to identify genes influencing probability of developing a mood disorder resulting in psychiatric referral. <i>Psychiatric Genetics</i> , 2021, 31, 194-198.	0.6	1
4565	Novel missense mutations in <i>PTCHD1</i> alter its plasma membrane subcellular localization and cause intellectual disability and autism spectrum disorder. <i>Human Mutation</i> , 2021, 42, 848-861.	1.1	8
4566	Whole-Exome Sequencing Reveals a Rare Variant of <i>OTOF</i> Gene Causing Congenital Non-syndromic Hearing Loss Among Large Muslim Families Favoring Consanguinity. <i>Frontiers in Genetics</i> , 2021, 12, 641925.	1.1	5
4567	Genetic Screening Revealed Latent Keratoconus in Asymptomatic Individuals. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 650344.	1.8	6
4568	Structural genomics approach to investigate deleterious impact of nsSNPs in conserved telomere maintenance component 1. <i>Scientific Reports</i> , 2021, 11, 10202.	1.6	18
4570	Mutation pattern and genotype-phenotype correlations of <i>SETD2</i> in neurodevelopmental disorders. <i>European Journal of Medical Genetics</i> , 2021, 64, 104200.	0.7	5
4571	Mutations in <i>MT-ATP6</i> are a frequent cause of adult-onset spinocerebellar ataxia. <i>Journal of Neurology</i> , 2021, 268, 4866-4873.	1.8	8
4572	Refinement of the clinical variant interpretation framework by statistical evidence and machine learning. <i>Med</i> , 2021, 2, 611-632.e9.	2.2	1
4574	A novel <i>RLIM/RNF12</i> variant disrupts protein stability and function to cause severe Tonne-Kalscheuer syndrome. <i>Scientific Reports</i> , 2021, 11, 9560.	1.6	5

#	ARTICLE	IF	CITATIONS
4575	Variants in the degenon of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. <i>American Journal of Human Genetics</i> , 2021, 108, 857-873.	2.6	19
4576	Genetic and immunologic findings in children with recurrent aphthous stomatitis with systemic inflammation. <i>Pediatric Rheumatology</i> , 2021, 19, 70.	0.9	12
4578	ConsRM: collection and large-scale prediction of the evolutionarily conserved RNA methylation sites, with implications for the functional epitranscriptome. <i>Briefings in Bioinformatics</i> , 2021, 22, .	3.2	34
4579	Young and early-onset dilated cardiomyopathy with malignant ventricular arrhythmia and sudden cardiac death induced by the heterozygous LDB3, MYH6, and SYNE1 missense mutations. <i>Annals of Noninvasive Electrocardiology</i> , 2021, 26, e12840.	0.5	11
4580	TBC1D24 emerges as an important contributor to progressive postlingual dominant hearing loss. <i>Scientific Reports</i> , 2021, 11, 10300.	1.6	4
4581	Adult diagnosis of congenital serine biosynthesis defect: A treatable cause of progressive neuropathy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2102-2107.	0.7	9
4582	Haploinsufficiency of the HIRA gene may not always produce severe neurodevelopmental consequences. <i>Psychiatric Genetics</i> , 2021, Publish Ahead of Print, 140-142.	0.6	2
4583	A novel homozygous missense variant in the NAXE gene in an Iranian family with progressive encephalopathy with brain edema and leukoencephalopathy. <i>Acta Neurologica Belgica</i> , 2022, 122, 1201-1210.	0.5	10
4584	Bi-allelic variants in MTMR5/SBF1 cause Charcot-Marie-Tooth type 4B3 featuring mitochondrial dysfunction. <i>BMC Medical Genomics</i> , 2021, 14, 157.	0.7	2
4586	A Homozygous Deep Intronic Mutation Alters the Splicing of Nebulin Gene in a Patient With Nemaline Myopathy. <i>Frontiers in Neurology</i> , 2021, 12, 660113.	1.1	6
4587	Mutational Landscape of Pirin and Elucidation of the Impact of Most Detrimental Missense Variants That Accelerate the Breast Cancer Pathways: A Computational Modelling Study. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 692835.	1.6	27
4589	Computational prediction of the effects of non-synonymous single nucleotide polymorphisms on the GPI-anchor transamidase subunit GPI8p of <i>Plasmodium falciparum</i> . <i>Computational Biology and Chemistry</i> , 2021, 92, 107461.	1.1	1
4590	An Axin2 mutation and perinatal risk factors contribute to sagittal craniosynostosis: evidence from a Chinese female monozygotic diamniotic twin family. <i>Hereditas</i> , 2021, 158, 20.	0.5	1
4591	Monogenic Childhood Diabetes: Dissecting Clinical Heterogeneity by Next-Generation Sequencing in Maturity-Onset Diabetes of the Young. <i>OMICS A Journal of Integrative Biology</i> , 2021, 25, 431-449.	1.0	12
4592	Broadening the phenotypic spectrum of Beta3GalT6-associated phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3153-3160.	0.7	3
4593	Severe neurodevelopmental disorder with intractable seizures due to a novel SLC1A4 homozygous variant. <i>European Journal of Medical Genetics</i> , 2021, 64, 104263.	0.7	8
4594	Towards a New, Endophenotype-Based Strategy for Pathogenicity Prediction in BRCA1 and BRCA2: In Silico Modeling of the Outcome of HDR/SGE Assays for Missense Variants. <i>International Journal of Molecular Sciences</i> , 2021, 22, 6226.	1.8	0
4595	In silico prediction of the functional consequences of nsSNPs in human beta-catenin gene. <i>Gene Reports</i> , 2021, 23, 101066.	0.4	2

#	ARTICLE	IF	CITATIONS
4596	Novel AQP2 Mutations and Clinical Characteristics in Seven Chinese Families With Congenital Nephrogenic Diabetes Insipidus. <i>Frontiers in Endocrinology</i> , 2021, 12, 686818.	1.5	2
4597	Causal Relationship and Shared Genetic Loci between Psoriasis and Type 2 Diabetes through Trans-Disease Meta-Analysis. <i>Journal of Investigative Dermatology</i> , 2021, 141, 1493-1502.	0.3	29
4598	Functional genomics of GPR126 in airway smooth muscle and bronchial epithelial cells. <i>FASEB Journal</i> , 2021, 35, e21300.	0.2	7
4599	Predicting the Most Deleterious Missense Nonsynonymous Single-Nucleotide Polymorphisms of Hennekam Syndrome-Causing CCBE1 Gene, In Silico Analysis. <i>Scientific World Journal</i> , The, 2021, 2021, 1-19.	0.8	6
4600	Functionally significant polymorphisms of the MMP-9 gene are associated with peptic ulcer disease in the Caucasian population of Central Russia. <i>Scientific Reports</i> , 2021, 11, 13515.	1.6	27
4602	New Insights of Phospholipase A2 Associated Neurodegeneration Phenotype Based on the Long-Term Follow-Up of a Large Hungarian Family. <i>Frontiers in Genetics</i> , 2021, 12, 628904.	1.1	2
4603	Characterization of Rheumatoid Arthritis Risk-Associated SNPs and Identification of Novel Therapeutic Sites Using an In-Silico Approach. <i>Biology</i> , 2021, 10, 501.	1.3	11
4605	A trans locus causes a ribosomopathy in hypertrophic hearts that affects mRNA translation in a protein length-dependent fashion. <i>Genome Biology</i> , 2021, 22, 191.	3.8	4
4606	Whole-Genome Sequencing Identifies Two Novel Rare Mutations in BMP5 and BMP2 in Monozygotic Twins With Microtia. <i>Journal of Craniofacial Surgery</i> , 2022, 33, e212-e217.	0.3	6
4607	Broadening INPP5E phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. <i>Npj Genomic Medicine</i> , 2021, 6, 53.	1.7	8
4608	Characterization of a novel mutation V136L in bone morphogenetic protein 15 identified in a woman affected by POI. <i>Journal of Ovarian Research</i> , 2021, 14, 85.	1.3	9
4609	Current status and relevance of single nucleotide polymorphisms in IL-6-/IL-12-type cytokine receptors. <i>Cytokine</i> , 2021, 148, 155550.	1.4	8
4610	Identification of Novel Mutations in Colorectal Cancer Patients Using AmpliSeq Comprehensive Cancer Panel. <i>Journal of Personalized Medicine</i> , 2021, 11, 535.	1.1	3
4612	In silico analysis of deleterious SNPs of human MTUS1 gene and their impacts on subsequent protein structure and function. <i>PLoS ONE</i> , 2021, 16, e0252932.	1.1	18
4613	Novel AVPR2 mutations and clinical characteristics in 28 Chinese families with congenital nephrogenic diabetes insipidus. <i>Journal of Endocrinological Investigation</i> , 2021, 44, 2777-2783.	1.8	5
4614	Identification of a novel point mutation in DAX-1 gene in a patient with adrenal hypoplasia congenita. <i>Annals of Pediatric Endocrinology and Metabolism</i> , 2021, 26, 126-129.	0.8	2
4615	Evolutionary and functional lessons from human-specific amino acid substitution matrices. <i>NAR Genomics and Bioinformatics</i> , 2021, 3, lqab079.	1.5	1
4616	Medical Big Data Analysis System to Discover Associations between Genetic Variants and Diseases. , 2021, , .		0

#	ARTICLE	IF	CITATIONS
4617	Whole-Exome Sequencing in a Cohort of High Myopia Patients in Northwest China. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 645501.	1.8	7
4618	BRCA1 and BRCA2 Variation in Taiwanese General Population and the Cancer Cohort. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 685174.	1.6	9
4619	Pittâ€“Hopkins syndrome: phenotypic and genotypic description of four unrelated patients and structural analysis of corresponding missense mutations. <i>Neurogenetics</i> , 2021, 22, 161-169.	0.7	4
4620	GDAP1 mutations are frequent among Brazilian patients with autosomal recessive axonal Charcot-Marie-Tooth disease. <i>Neuromuscular Disorders</i> , 2021, 31, 505-511.	0.3	5
4621	Population diversity of cassava mosaic begomoviruses increases over the course of serial vegetative propagation. <i>Journal of General Virology</i> , 2021, 102, .	1.3	14
4622	The first insight into the genetic structure of the population of modern Serbia. <i>Scientific Reports</i> , 2021, 11, 13995.	1.6	1
4623	Contribution of HCN1 variant to sinus bradycardia: A case report. <i>Journal of Arrhythmia</i> , 2021, 37, 1337-1347.	0.5	2
4624	Genes affecting ionizing radiation survival identified through combined exome sequencing and functional screening. <i>Human Mutation</i> , 2021, 42, 1124-1138.	1.1	0
4625	3Cnet: pathogenicity prediction of human variants using multitask learning with evolutionary constraints. <i>Bioinformatics</i> , 2021, 37, 4626-4634.	1.8	17
4626	<scp><i>CARMNâ€“NOTCH2</i></scp> fusion transcript drives high <scp>NOTCH2</scp> expression in glomus tumors of the upper digestive tract. <i>Genes Chromosomes and Cancer</i> , 2021, 60, 723-732.	1.5	11
4627	Myopia in African Americans Is Significantly Linked to Chromosome 7p15.2-14.2. , 2021, 62, 16.		2
4628	Methodology in phenome-wide association studies: a systematic review. <i>Journal of Medical Genetics</i> , 2021, 58, 720-728.	1.5	10
4629	Genetic analysis of four consanguineous multiplex families with inflammatory bowel disease. <i>Gastroenterology Report</i> , 2021, 9, 521-532.	0.6	5
4630	Novel variants in GUCY2D causing retinopathy and the genotype-phenotype correlation. <i>Experimental Eye Research</i> , 2021, 208, 108637.	1.2	7
4631	Mutation analysis of SOD1, C9orf72, TARDBP and FUS genes in ethnically-diverse Malaysian patients with amyotrophic lateral sclerosis (ALS). <i>Neurobiology of Aging</i> , 2021, 108, 200-206.	1.5	7
4632	Discrepancies of RET gene and risk of differentiated thyroid carcinoma. <i>Cancer Biomarkers</i> , 2022, 33, 111-121.	0.8	0
4633	Analysis of rare coding variants in 200,000 exomeâ€“sequenced subjects reveals novel genetic risk factors for type 2 diabetes. <i>Diabetes/Metabolism Research and Reviews</i> , 2022, 38, e3482.	1.7	11
4634	A systematic view of computational methods for identifying driver genes based on somatic mutation data. <i>Briefings in Functional Genomics</i> , 2021, 20, 333-343.	1.3	4

#	ARTICLE	IF	CITATIONS
4635	Genetic susceptibility to acute graft versus host disease in pediatric patients undergoing HSCT. Bone Marrow Transplantation, 2021, 56, 2697-2704.	1.3	2
4636	Comparative analysis of clonal evolution among patients with right- and left-sided colon and rectal cancer. IScience, 2021, 24, 102718.	1.9	9
4637	Hearing Impairment with Monoallelic GJB2 Variants. Journal of Molecular Diagnostics, 2021, 23, 1279-1291.	1.2	10
4638	Sequencing of 640,000 exomes identifies <i>GPR75</i> variants associated with protection from obesity. Science, 2021, 373, .	6.0	130
4639	Variation in the Evolution and Sequences of Proglucagon and the Receptors for Proglucagon-Derived Peptides in Mammals. Frontiers in Endocrinology, 2021, 12, 700066.	1.5	6
4641	Determination of genetic effects and functional SNPs of bovine HTR1B gene on milk fatty acid traits. BMC Genomics, 2021, 22, 575.	1.2	4
4642	The Second Oncogenic Hit Determines the Cell Fate of ETV6-RUNX1 Positive Leukemia. Frontiers in Cell and Developmental Biology, 2021, 9, 704591.	1.8	19
4643	The protective effects of the methylenetetrahydrofolate reductase rs1801131 variant among Saudi smokers. Saudi Journal of Biological Sciences, 2021, 28, 3972-3980.	1.8	2
4644	Clinical characteristics and genetic spectrum of 26 individuals of Chinese origin with primary ciliary dyskinesia. Orphanet Journal of Rare Diseases, 2021, 16, 293.	1.2	11
4645	LDLR variants functional characterization: Contribution to variant classification. Atherosclerosis, 2021, 329, 14-21.	0.4	11
4646	Computational Insights into the Deleterious Impacts of Missense Variants on N-Acetyl-d-glucosamine Kinase Structure and Function. International Journal of Molecular Sciences, 2021, 22, 8048.	1.8	6
4647	CDON gene contributes to pituitary stalk interruption syndrome associated with unilateral facial and abducens nerve palsy. Journal of Applied Genetics, 2021, 62, 621-629.	1.0	3
4648	Rare Gain-of-Function KCND3 Variant Associated with Cerebellar Ataxia, Parkinsonism, Cognitive Dysfunction, and Brain Iron Accumulation. International Journal of Molecular Sciences, 2021, 22, 8247.	1.8	7
4649	The conserved ASTN2/BRINP1 locus at 9q33.1â€“33.2 is associated with major psychiatric disorders in a large pedigree from Southern Spain. Scientific Reports, 2021, 11, 14529.	1.6	3
4650	Identification of Novel Variants in Cleft Palate-Associated Genes in Brazilian Patients With Non-syndromic Cleft Palate Only. Frontiers in Cell and Developmental Biology, 2021, 9, 638522.	1.8	5
4651	An FGA Frameshift Variant Associated with Afibrinogenemia in Dachshunds. Genes, 2021, 12, 1065.	1.0	0
4652	DPYD Exome, mRNA Expression and Uracil Levels in Early Severe Toxicity to Fluoropyrimidines: An Extreme Phenotype Approach. Journal of Personalized Medicine, 2021, 11, 792.	1.1	2
4653	Feature Point Extraction and Motion Tracking of Cardiac Color Ultrasound under Improved Lucas-Kanade Algorithm. Journal of Healthcare Engineering, 2021, 2021, 1-10.	1.1	0

#	ARTICLE	IF	CITATIONS
4654	Genetic factors contributing to late adverse musculoskeletal effects in childhood acute lymphoblastic leukemia survivors. <i>Pharmacogenomics Journal</i> , 2022, 22, 19-24.	0.9	2
4655	A global map of associations between types of protein posttranslational modifications and human genetic diseases. <i>IScience</i> , 2021, 24, 102917.	1.9	7
4656	Structural effects driven by rare point mutations in amylin hormone, the type II diabetes-associated peptide. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 2021, 1865, 129935.	1.1	2
4657	Novel GRHL3 Variants in a South African Cohort With Cleft Lip and Palate. <i>Cleft Palate-Craniofacial Journal</i> , 2022, 59, 1125-1130.	0.5	1
4658	New Missense Mutation Gly238Ala in the TBX5 Gene and Its Phenotypical Characteristics. <i>Russian Journal of Genetics</i> , 2021, 57, 949-954.	0.2	0
4659	Fucosidosis in Tunisian patients: mutational analysis and homology-based modeling of FUCA1 enzyme. <i>BMC Medical Genomics</i> , 2021, 14, 208.	0.7	4
4660	Strategies to Identify Genetic Variants Causing Infertility. <i>Trends in Molecular Medicine</i> , 2021, 27, 792-806.	3.5	9
4661	<i>PNPT1</i> , <i>MYO15A</i> , <i>PTPRQ</i> , and <i>SLC12A2</i> associated genetic and phenotypic heterogeneity among hearing impaired assortative mating families in Southern India. <i>Annals of Human Genetics</i> , 2022, 86, 1-13.	0.3	5
4662	Whole genome sequences of two Trichophyton indotineae clinical isolates from India emerging as threats during therapeutic treatment of dermatophytosis. <i>3 Biotech</i> , 2021, 11, 402.	1.1	9
4663	Genetic evaluation supports differential diagnosis in adolescent patients with delayed puberty. <i>European Journal of Endocrinology</i> , 2021, 185, 617-627.	1.9	15
4664	Novel FERMT3 and PTPRQ Mutations Associated with Leukocyte Adhesion Deficiency-III and Sensorineural Hearing Loss. <i>Journal of Pediatric Genetics</i> , 2023, 12, 348-351.	0.3	0
4665	Hereditary sensory and autonomic neuropathy in a family of mixed breed dogs associated with a novel <i>RETREG1</i> variant. <i>Journal of Veterinary Internal Medicine</i> , 2021, 35, 2306-2314.	0.6	8
4666	<i>Lens orientalis</i> Contributes Quantitative Trait Loci and Candidate Genes Associated With Ascochyta Blight Resistance in Lentil. <i>Frontiers in Plant Science</i> , 2021, 12, 703283.	1.7	11
4667	A Missense Mutation in the MYBPH Gene Is Associated With Abdominal Fat Traits in Meat-Type Chickens. <i>Frontiers in Genetics</i> , 2021, 12, 698163.	1.1	4
4668	Homozygous mutations in <i>CCDC34</i> cause male infertility with oligoasthenoteratozoospermia in humans and mice. <i>Journal of Medical Genetics</i> , 2022, 59, 710-718.	1.5	20
4669	A domain damage index to prioritizing the pathogenicity of missense variants. <i>Human Mutation</i> , 2021, 42, 1503-1517.	1.1	0
4670	Comprehensive characterization of 536 patient-derived xenograft models prioritizes candidates for targeted treatment. <i>Nature Communications</i> , 2021, 12, 5086.	5.8	58
4671	Mutational Analysis of Mucopolysaccharidosis in Iranian Patients. <i>Zahedan Journal of Researches in Medical Sciences</i> , 2021, 23, .	0.1	0

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4672	Somatic NF1 mutations in pituitary adenomas: Report of two cases. <i>Cancer Genetics</i> , 2021, 256-257, 26-30.	0.2	1
4673	Bi-allelic mutations of DNAH10 cause primary male infertility with asthenoteratozoospermia in humans and mice. <i>American Journal of Human Genetics</i> , 2021, 108, 1466-1477.	2.6	50
4674	Germline CDH1 G212E Missense Variant: Combining Clinical, In Vitro and In Vivo Strategies to Unravel Disease Burden. <i>Cancers</i> , 2021, 13, 4359.	1.7	9
4676	Mapping gene and gene pathways associated with coronary artery disease: a CARDIoGRAM exome and multi-ancestry UK biobank analysis. <i>Scientific Reports</i> , 2021, 11, 16461.	1.6	4
4677	Functional and structural analysis of cytokine-selective IL6ST defects that cause recessive hyper-IgE syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 585-598.	1.5	20
4678	Clinical and Molecular Update on the Fourth Reported Family with Hamamy Syndrome. <i>Molecular Syndromology</i> , 2021, 12, 342-350.	0.3	1
4679	Phenotypic and immune functional profiling of patients with suspected Mendelian Susceptibility to Mycobacterial Disease in South Africa. <i>BMC Immunology</i> , 2021, 22, 62.	0.9	4
4680	Improved pathogenicity prediction for rare human missense variants. <i>American Journal of Human Genetics</i> , 2021, 108, 1891-1906.	2.6	51
4681	Yield of clinically reportable genetic variants in unselected cerebral palsy by whole genome sequencing. <i>Npj Genomic Medicine</i> , 2021, 6, 74.	1.7	16
4682	Restoring fertility in yeast hybrids: Breeding and quantitative genetics of beneficial traits. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	21
4683	A thrombophilia family with protein S deficiency due to protein translation disorders caused by a Leu607Ser heterozygous mutation in PROS1. <i>Thrombosis Journal</i> , 2021, 19, 64.	0.9	2
4684	Epidemiological aspects of hereditary fructose intolerance: A database study. <i>Human Mutation</i> , 2021, 42, 1548-1566.	1.1	12
4685	A systematic review and meta-analysis of the association of ABCC2/ABCG2 polymorphisms with antiepileptic drug responses in epileptic patients. <i>Epilepsy Research</i> , 2021, 175, 106678.	0.8	8
4686	CDH1 germline mutations in a Chinese cohort with hereditary diffuse gastric cancer. <i>Journal of Cancer Research and Clinical Oncology</i> , 2022, 148, 2145-2151.	1.2	6
4687	Characterization of Two Variants at Met 1 of the Human LDLR Gene Encoding the Same Amino Acid but Causing Different Functional Phenotypes. <i>Biomedicines</i> , 2021, 9, 1219.	1.4	5
4688	Autosomal dominant Shwachmanâ€™Diamond syndrome with a novel heterozygous missense variant in the SRP54 gene causing severe phenotypic features. <i>British Journal of Haematology</i> , 2021, , .	1.2	2
4689	Incorporating genetic counseling into the evaluation of pediatric bone marrow failure. <i>Journal of Genetic Counseling</i> , 2022, 31, 433-446.	0.9	1
4690	Genomic and epigenomic evolution of acquired resistance to combination therapy in esophageal squamous cell carcinoma. <i>JCI Insight</i> , 2021, 6, .	2.3	7

#	ARTICLE	IF	CITATIONS
4691	Detecting Rewiring Events in Protein-Protein Interaction Networks Based on Transcriptomic Data. <i>Frontiers in Bioinformatics</i> , 2021, 1, .	1.0	4
4692	Diagnostic yield of clinical exome sequencing as a first-tier genetic test for the diagnosis of genetic disorders in pediatric patients: results from a referral center study. <i>Human Genetics</i> , 2022, 141, 1269-1278.	1.8	10
4693	TNFRSF13B genotypes control immune-mediated pathology by regulating the functions of innate B cells. <i>JCI Insight</i> , 2021, 6, .	2.3	4
4694	Association of the functionally significant polymorphisms of the MMP9 gene with H. pylori-positive gastric ulcer in the Caucasian population of Central Russia. <i>PLoS ONE</i> , 2021, 16, e0257060.	1.1	13
4695	Proteomics-derived basal biomarker DNA-PKcs is associated with intrinsic subtype and long-term clinical outcomes in breast cancer. <i>Npj Breast Cancer</i> , 2021, 7, 114.	2.3	3
4696	Genetic factors in treatment-related cardiovascular complications in survivors of childhood acute lymphoblastic leukemia. <i>Pharmacogenomics</i> , 2021, 22, 885-901.	0.6	1
4697	Contribution of rare variant associations to neurodegenerative disease presentation. <i>Npj Genomic Medicine</i> , 2021, 6, 80.	1.7	14
4698	Adapting the ACMG/AMP variant classification framework: A perspective from the ClinGen Hemoglobinopathy Variant Curation Expert Panel. <i>Human Mutation</i> , 2022, 43, 1089-1096.	1.1	20
4699	Identification of Two de novo Variants of CACNA1A in Pediatric Chinese Patients With Paroxysmal Tonic Upgaze. <i>Frontiers in Pediatrics</i> , 2021, 9, 722105.	0.9	3
4701	Characterization of a Compound Heterozygous SLC2A9 Mutation That Causes Hypouricemia. <i>Biomedicines</i> , 2021, 9, 1172.	1.4	2
4703	Identifying digenic disease genes via machine learning in the Undiagnosed Diseases Network. <i>American Journal of Human Genetics</i> , 2021, 108, 1946-1963.	2.6	25
4704	<i>PRPH2</i> mutation update: In silico assessment of 245 reported and 7 novel variants in patients with retinal disease. <i>Human Mutation</i> , 2021, 42, 1521-1547.	1.1	13
4705	Deleterious Rare Desmosomal Variants Contribute to Hypertrophic Cardiomyopathy and Are Associated With Distinctive Clinical Features. <i>Canadian Journal of Cardiology</i> , 2022, 38, 41-48.	0.8	6
4706	Whole-exome sequencing identification of a recurrent CRYBB2 variant in a four-generation Chinese family with congenital nuclear cataracts. <i>Experimental and Therapeutic Medicine</i> , 2021, 22, 1375.	0.8	1
4707	Exonic SNP in MHC-DMB2 is associated with gene expression and humoral immunity in Japanese quails. <i>Veterinary Immunology and Immunopathology</i> , 2021, 239, 110302.	0.5	4
4708	Multiple Sources of Introduction of North American <i>Arabidopsis thaliana</i> from across Eurasia. <i>Molecular Biology and Evolution</i> , 2021, 38, 5328-5344.	3.5	6
4709	Whole-Genome Sequencing Reveals Large ATP8B1 Deletion/Duplications as Second Mutations Missed by Exome-Based Sequencing. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 1491-1499.	1.2	2
4710	A genome-first approach to rare variants in hypertrophic cardiomyopathy genes <i>MYBPC3</i> and <i>MYH7</i> in a medical biobank. <i>Human Molecular Genetics</i> , 2022, 31, 827-837.	1.4	4

#	ARTICLE	IF	CITATIONS
4711	Dominant <i>KPNA3</i> Mutations Cause Infantile-Onset Hereditary Spastic Paraplegia. <i>Annals of Neurology</i> , 2021, 90, 738-750.	2.8	5
4712	Developmental disabilities across the world: A scientometric review from 1936 to 2020. <i>Research in Developmental Disabilities</i> , 2021, 117, 104031.	1.2	20
4713	Multiple variants in <i>XDH</i> and <i>MOCOS</i> underlie xanthine urolithiasis in dogs. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 29, 100792.	0.4	3
4714	De novo mutation in <i>COL2A1</i> leads to lethal foetal skeletal dysplasia. <i>Bone</i> , 2021, 153, 116169.	1.4	6
4716	Genetic profile of adult T-cell leukemia/lymphoma in Okinawa: Association with prognosis, ethnicity, and HTLV-1 strains. <i>Cancer Science</i> , 2021, 112, 1300-1309.	1.7	14
4717	Molecular dynamics approach to identification of new <i>OGG1</i> cancer-associated somatic variants with impaired activity. <i>Journal of Biological Chemistry</i> , 2021, 296, 100229.	1.6	7
4718	Genome-wide association study of febrile seizures implicates fever response and neuronal excitability genes. <i>Brain</i> , 2022, 145, 555-568.	3.7	29
4720	Multiple Linear Regression Allows Weighted Burden Analysis of Rare Coding Variants in an Ethnically Heterogeneous Population. <i>Human Heredity</i> , 2020, 85, 1-10.	0.4	23
4722	Genomic sequencing of rare diseases. , 2021, , 61-95.		6
4723	Hypothalamic Norepinephrine Concentration and Heart Mass in Hypertensive ISIAH Rats Are Associated with a Genetic Locus on Chromosome 18. <i>Journal of Personalized Medicine</i> , 2021, 11, 67.	1.1	7
4724	DiaDeL: An Accurate Deep Learning-Based Model With Mutational Signatures for Predicting Metastasis Stage and Cancer Types. <i>IEEE/ACM Transactions on Computational Biology and Bioinformatics</i> , 2022, 19, 1336-1343.	1.9	3
4725	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. <i>Human Mutation</i> , 2018, 39, 1246-1261.	1.1	31
4726	The expanding <i>LARS2</i> phenotypic spectrum: HLASA, Perrault syndrome with leukodystrophy, and mitochondrial myopathy. <i>Human Mutation</i> , 2020, 41, 1425-1434.	1.1	15
4727	Next Generation Sequencing (NGS) Strategies for Genetic Testing of Cerebral Cavernous Malformation (CCM) Disease. <i>Methods in Molecular Biology</i> , 2020, 2152, 59-75.	0.4	2
4728	Bioinformatics Tools in Clinical Genomics. , 2015, , 177-196.		2
4729	Approaches for Classifying DNA Variants Found by Sanger Sequencing in a Medical Genetics Laboratory. <i>Methods in Molecular Biology</i> , 2014, 1168, 227-250.	0.4	3
4730	Studying Cancer Genomics Through Next-Generation DNA Sequencing and Bioinformatics. <i>Methods in Molecular Biology</i> , 2014, 1168, 83-98.	0.4	12
4731	Computational Tools for Designing Smart Libraries. <i>Methods in Molecular Biology</i> , 2014, 1179, 291-314.	0.4	21

#	ARTICLE	IF	CITATIONS
4732	Identification and Analysis of Genes Associated with Inherited Retinal Diseases. <i>Methods in Molecular Biology</i> , 2019, 1834, 3-27.	0.4	12
4733	Determination of the Clinical Significance of an Unclassified Variant. <i>Methods in Molecular Biology</i> , 2012, 837, 337-348.	0.4	10
4734	Fibroblast Growth Factor Receptor and Related Skeletal Disorders. , 2016, , 177-187.		1
4735	TILLING in <i>Lotus japonicus</i> . <i>Compendium of Plant Genomes</i> , 2014, , 229-243.	0.3	1
4736	Identifying Driver Mutations in Cancer. <i>Translational Bioinformatics</i> , 2013, , 33-56.	0.0	4
4737	Systems Biology and Integrated Computational Methods for Cancer-Associated Mutation Analysis. , 2020, , 335-362.		3
4738	Pattern dystrophy-like changes and coquille dâ€™oeuf atrophy in elderly patients affected by pseudoxanthoma elasticum. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2020, 258, 1881-1892.	1.0	7
4739	Comparison of Pathogenicity Prediction Tools on Somatic Variants. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 1383-1392.	1.2	19
4740	Maternal mosaicism underlies the inheritance of a rare germline AKT3 variant which is responsible for megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome in two Roma half-siblings. <i>Experimental and Molecular Pathology</i> , 2020, 115, 104471.	0.9	5
4741	A novel PAK1 variant causative of neurodevelopmental disorder with postnatal macrocephaly. <i>Journal of Human Genetics</i> , 2020, 65, 481-485.	1.1	14
4742	Multiancestry genome-wide association study of 520,000 subjects identifies 32 loci associated with stroke and stroke subtypes. <i>Nature Genetics</i> , 2018, 50, 524-537.	9.4	1,124
4743	The Mount Sinai cohort of large-scale genomic, transcriptomic and proteomic data in Alzheimer's disease. <i>Scientific Data</i> , 2018, 5, 180185.	2.4	320
4744	Systematic analysis of genetic variants in Han Chinese patients with sporadic Parkinsonâ€™s disease. <i>Scientific Reports</i> , 2016, 6, 33850.	1.6	12
4745	DeMaSk: a deep mutational scanning substitution matrix and its use for variant impact prediction. <i>Bioinformatics</i> , 2021, 36, 5322-5329.	1.8	24
4746	AI-Driver: an ensemble method for identifying driver mutations in personal cancer genomes. <i>NAR Genomics and Bioinformatics</i> , 2020, 2, lqaa084.	1.5	19
4747	Whole exome sequencing in unclassified autoinflammatory diseases: more monogenic diseases in the pipeline?. <i>Rheumatology</i> , 2021, 60, 607-616.	0.9	13
4748	SOX11-related syndrome: report on a new case and review. <i>Clinical Dysmorphology</i> , 2021, 30, 44-49.	0.1	7
4811	Weighted burden analysis of exomeâ€sequenced lateâ€onset Alzheimer's cases and controls provides further evidence for a role for <i>PSEN1</i> and suggests involvement of the PI3K/Akt/GSKâ€3â€ β and WNT signalling pathways. <i>Annals of Human Genetics</i> , 2020, 84, 291-302.	0.3	24

#	ARTICLE	IF	CITATIONS
4812	Mutational landscape of severe combined immunodeficiency patients from Turkey. <i>International Journal of Immunogenetics</i> , 2020, 47, 529-538.	0.8	14
4813	Mining the Human Metabolome for Precision Oncology Research. , 2020, , .		1
4814	ALDH3B2 Polymorphism Is Associated with Colorectal Cancer Susceptibility. <i>Journal of Oncology</i> , 2020, 2020, 1-5.	0.6	7
4815	Variants in <i>ACE2</i> and <i>TMPRSS2</i> Genes Are Not Major Determinants of COVID-19 Severity in UK Biobank Subjects. <i>Human Heredity</i> , 2020, 85, 66-68.	0.4	10
4816	Sialic acid catabolism by N-acetylneuraminase pyruvate lyase is essential for muscle function. <i>JCI Insight</i> , 2018, 3, .	2.3	36
4817	LGR4 deficiency results in delayed puberty through impaired Wnt/ β -catenin signaling. <i>JCI Insight</i> , 2020, 5, .	2.3	25
4818	MPEG1/perforin-2 mutations in human pulmonary nontuberculous mycobacterial infections. <i>JCI Insight</i> , 2017, 2, .	2.3	30
4819	Whole-exome sequencing uncovers oxidoreductases DHTKD1 and OGDHL as linkers between mitochondrial dysfunction and eosinophilic esophagitis. <i>JCI Insight</i> , 2018, 3, .	2.3	39
4820	Ankyrin-B dysfunction predisposes to arrhythmogenic cardiomyopathy and is amenable to therapy. <i>Journal of Clinical Investigation</i> , 2019, 129, 3171-3184.	3.9	42
4821	Genetic hallmarks of recurrent/metastatic adenoid cystic carcinoma. <i>Journal of Clinical Investigation</i> , 2019, 129, 4276-4289.	3.9	134
4822	Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. <i>Journal of Clinical Investigation</i> , 2015, 125, 636-651.	3.9	136
4823	Dysfunctional SEMA3E signaling underlies gonadotropin-releasing hormone neuron deficiency in Kallmann syndrome. <i>Journal of Clinical Investigation</i> , 2015, 125, 2413-2428.	3.9	97
4824	A mutation in the nucleoporin-107 gene causes XX gonadal dysgenesis. <i>Journal of Clinical Investigation</i> , 2015, 125, 4295-4304.	3.9	77
4825	Dominant-negative NFKBIA mutation promotes IL-1 β production causing hepatic disease with severe immunodeficiency. <i>Journal of Clinical Investigation</i> , 2020, 130, 5817-5832.	3.9	17
4826	Biological and clinical significance of dysplastic hematopoiesis in patients with newly diagnosed multiple myeloma. <i>Blood</i> , 2020, 135, 2375-2387.	0.6	24
4828	Whole exome sequencing of a single osteosarcoma case; integrative analysis with whole transcriptome RNA-seq data. <i>Human Genomics</i> , 2014, 8, 20.	1.4	6
4829	Multiple Endocrine Tumors Associated with Germline <i>MAX</i> Mutations: Multiple Endocrine Neoplasia Type 5?. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e1163-e1182.	1.8	43
4830	Recent advances in predicting gene-disease associations. <i>F1000Research</i> , 2017, 6, 578.	0.8	36

#	ARTICLE	IF	CITATIONS
4831	Meta-analysis of exome array data identifies six novel genetic loci for lung function. Wellcome Open Research, 0, 3, 4.	0.9	11
4832	Meta-analysis of exome array data identifies six novel genetic loci for lung function. Wellcome Open Research, 0, 3, 4.	0.9	1
4833	Meta-analysis of exome array data identifies six novel genetic loci for lung function. Wellcome Open Research, 2018, 3, 4.	0.9	19
4834	Perturbation of PALB2 function by the T413S mutation found in small cell lung cancer. Wellcome Open Research, 0, 2, 110.	0.9	5
4835	Perturbation of PALB2 function by the T413S mutation found in small cell lung cancer. Wellcome Open Research, 2017, 2, 110.	0.9	5
4836	Interpretation of Genomic Variants Using a Unified Biological Network Approach. PLoS Computational Biology, 2013, 9, e1002886.	1.5	162
4837	PredictSNP2: A Unified Platform for Accurately Evaluating SNP Effects by Exploiting the Different Characteristics of Variants in Distinct Genomic Regions. PLoS Computational Biology, 2016, 12, e1004962.	1.5	149
4838	Predicted Molecular Effects of Sequence Variants Link to System Level of Disease. PLoS Computational Biology, 2016, 12, e1005047.	1.5	14
4839	Oncodomains: A protein domain-centric framework for analyzing rare variants in tumor samples. PLoS Computational Biology, 2017, 13, e1005428.	1.5	25
4840	Whole-Exome Re-Sequencing in a Family Quartet Identifies POP1 Mutations As the Cause of a Novel Skeletal Dysplasia. PLoS Genetics, 2011, 7, e1002027.	1.5	72
4841	Heterogeneity of Human Neutrophil CD177 Expression Results from CD177P1 Pseudogene Conversion. PLoS Genetics, 2016, 12, e1006067.	1.5	36
4842	Mutations of RagA GTPase in mTORC1 Pathway Are Associated with Autosomal Dominant Cataracts. PLoS Genetics, 2016, 12, e1006090.	1.5	23
4843	Whole Exome Sequencing in Atrial Fibrillation. PLoS Genetics, 2016, 12, e1006284.	1.5	35
4844	Rare Functional Variant in TM2D3 is Associated with Late-Onset Alzheimer's Disease. PLoS Genetics, 2016, 12, e1006327.	1.5	47
4845	Mutations in TSPEAR, Encoding a Regulator of Notch Signaling, Affect Tooth and Hair Follicle Morphogenesis. PLoS Genetics, 2016, 12, e1006369.	1.5	32
4846	Predicting the impact of Lynch syndrome-causing missense mutations from structural calculations. PLoS Genetics, 2017, 13, e1006739.	1.5	90
4847	Recurrent promoter mutations in melanoma are defined by an extended context-specific mutational signature. PLoS Genetics, 2017, 13, e1006773.	1.5	67
4848	Characterisation of canine KCNIP4: A novel gene for cerebellar ataxia identified by whole-genome sequencing two affected Norwegian Buhund dogs. PLoS Genetics, 2020, 16, e1008527.	1.5	10

#	ARTICLE	IF	CITATIONS
4849	Whole Genome Sequencing of <i>Mycobacterium africanum</i> Strains from Mali Provides Insights into the Mechanisms of Geographic Restriction. <i>PLoS Neglected Tropical Diseases</i> , 2016, 10, e0004332.	1.3	41
4850	Trans-Species Polymorphism and Selection in the MHC Class II DRA Genes of Domestic Sheep. <i>PLoS ONE</i> , 2010, 5, e11402.	1.1	28
4851	Prediction of Deleterious Non-Synonymous SNPs Based on Protein Interaction Network and Hybrid Properties. <i>PLoS ONE</i> , 2010, 5, e11900.	1.1	70
4852	Bioinformatic Analysis of Pathogenic Missense Mutations of Activin Receptor Like Kinase 1 Ectodomain. <i>PLoS ONE</i> , 2011, 6, e26431.	1.1	14
4853	Evolution of the Bovine TLR Gene Family and Member Associations with <i>Mycobacterium avium</i> Subspecies Paratuberculosis Infection. <i>PLoS ONE</i> , 2011, 6, e27744.	1.1	48
4854	Statistical Guidance for Experimental Design and Data Analysis of Mutation Detection in Rare Monogenic Mendelian Diseases by Exome Sequencing. <i>PLoS ONE</i> , 2012, 7, e31358.	1.1	24
4855	Screening of MAMLD1 Mutations in 70 Children with 46,XY DSD: Identification and Functional Analysis of Two New Mutations. <i>PLoS ONE</i> , 2012, 7, e32505.	1.1	41
4856	Evolution of <i>Burkholderia pseudomallei</i> in Recurrent Melioidosis. <i>PLoS ONE</i> , 2012, 7, e36507.	1.1	96
4857	The Cumulative Effects of Polymorphisms in the DNA Mismatch Repair Genes and Tobacco Smoking in Oesophageal Cancer Risk. <i>PLoS ONE</i> , 2012, 7, e36962.	1.1	47
4858	Association of Estimated Glomerular Filtration Rate and Urinary Uromodulin Concentrations with Rare Variants Identified by UMOD Gene Region Sequencing. <i>PLoS ONE</i> , 2012, 7, e38311.	1.1	24
4859	Two Rare Human Mitofusin 2 Mutations Alter Mitochondrial Dynamics and Induce Retinal and Cardiac Pathology in <i>Drosophila</i> . <i>PLoS ONE</i> , 2012, 7, e44296.	1.1	28
4860	Characterization of LEDGF/p75 Genetic Variants and Association with HIV-1 Disease Progression. <i>PLoS ONE</i> , 2012, 7, e50204.	1.1	10
4861	Functional Consequences of a Novel Variant of PCSK1. <i>PLoS ONE</i> , 2013, 8, e55065.	1.1	24
4862	Application of Massively Parallel Sequencing to Genetic Diagnosis in Multiplex Families with Idiopathic Sensorineural Hearing Impairment. <i>PLoS ONE</i> , 2013, 8, e57369.	1.1	32
4863	Few Single Nucleotide Variations in Exomes of Human Cord Blood Induced Pluripotent Stem Cells. <i>PLoS ONE</i> , 2013, 8, e59908.	1.1	31
4864	Interplay between DMD Point Mutations and Splicing Signals in Dystrophinopathy Phenotypes. <i>PLoS ONE</i> , 2013, 8, e59916.	1.1	42
4865	Comprehensive Functional Annotation of Seventy-One Breast Cancer Risk Loci. <i>PLoS ONE</i> , 2013, 8, e63925.	1.1	41
4866	Interaction between $\hat{1}^3$ -Aminobutyric Acid A Receptor Genes: New Evidence in Migraine Susceptibility. <i>PLoS ONE</i> , 2013, 8, e74087.	1.1	18

#	ARTICLE	IF	CITATIONS
4867	Genetic Alterations within the DENND1A Gene in Patients with Polycystic Ovary Syndrome (PCOS). PLoS ONE, 2013, 8, e77186.	1.1	34
4868	Massively Parallel Sequencing Reveals an Accumulation of De Novo Mutations and an Activating Mutation of LPAR1 in a Patient with Metastatic Neuroblastoma. PLoS ONE, 2013, 8, e77731.	1.1	24
4869	Novel HCN2 Mutation Contributes to Febrile Seizures by Shifting the Channel's Kinetics in a Temperature-Dependent Manner. PLoS ONE, 2013, 8, e80376.	1.1	49
4870	A Novel Null Homozygous Mutation Confirms CACNA2D2 as a Gene Mutated in Epileptic Encephalopathy. PLoS ONE, 2013, 8, e82154.	1.1	67
4871	Whole-Exome Sequencing to Identify a Novel LMNA Gene Mutation Associated with Inherited Cardiac Conduction Disease. PLoS ONE, 2013, 8, e83322.	1.1	12
4872	MET Genetic Abnormalities Unreliable for Patient Selection for Therapeutic Intervention in Oropharyngeal Squamous Cell Carcinoma. PLoS ONE, 2014, 9, e84319.	1.1	17
4873	Deriving a Mutation Index of Carcinogenicity Using Protein Structure and Protein Interfaces. PLoS ONE, 2014, 9, e84598.	1.1	22
4874	Identification and Glycerol-Induced Correction of Misfolding Mutations in the X-Linked Mental Retardation Gene CASK. PLoS ONE, 2014, 9, e88276.	1.1	20
4875	Analysis of Rare Variants in the C3 Gene in Patients with Age-Related Macular Degeneration. PLoS ONE, 2014, 9, e94165.	1.1	34
4876	Direct Evidence on the Contribution of a Missense Mutation in GDF9 to Variation in Ovulation Rate of Finnsheep. PLoS ONE, 2014, 9, e95251.	1.1	41
4877	Genome at Juncture of Early Human Migration: A Systematic Analysis of Two Whole Genomes and Thirteen Exomes from Kuwaiti Population Subgroup of Inferred Saudi Arabian Tribe Ancestry. PLoS ONE, 2014, 9, e99069.	1.1	41
4878	In Silico Identification of New Putative Pathogenic Variants in the Neu1 Sialidase Gene Affecting Enzyme Function and Subcellular Localization. PLoS ONE, 2014, 9, e104229.	1.1	6
4879	Computational Analysis of Functional Single Nucleotide Polymorphisms Associated with the CYP11B2 Gene. PLoS ONE, 2014, 9, e104311.	1.1	39
4880	Sorghum Phytochrome B Inhibits Flowering in Long Days by Activating Expression of SbPRR37 and SbGHD7, Repressors of SbEHD1, SbCN8 and SbCN12. PLoS ONE, 2014, 9, e105352.	1.1	97
4881	Molecular Genetics of the Usher Syndrome in Lebanon: Identification of 11 Novel Protein Truncating Mutations by Whole Exome Sequencing. PLoS ONE, 2014, 9, e107326.	1.1	10
4882	A Novel Familial Mutation in the PCSK1 Gene That Alters the Oxyanion Hole Residue of Proprotein Convertase 1/3 and Impairs Its Enzymatic Activity. PLoS ONE, 2014, 9, e108878.	1.1	19
4883	Identification of a PRPF4 Loss-of-Function Variant That Abrogates U4/U6.U5 Tri-snRNP Integration and Is Associated with Retinitis Pigmentosa. PLoS ONE, 2014, 9, e111754.	1.1	36
4884	Fine Mapping and Candidate Gene Search of Quantitative Trait Loci for Growth and Obesity Using Mouse Intersubspecific Subcongenic Intercrosses and Exome Sequencing. PLoS ONE, 2014, 9, e113233.	1.1	10

#	ARTICLE	IF	CITATIONS
4885	Frequency of COL4A3/COL4A4 Mutations amongst Families Segregating Glomerular Microscopic Hematuria and Evidence for Activation of the Unfolded Protein Response. Focal and Segmental Glomerulosclerosis Is a Frequent Development during Ageing. PLoS ONE, 2014, 9, e115015.	1.1	53
4886	Novel ENAM and LAMB3 Mutations in Chinese Families with Hypoplastic Amelogenesis Imperfecta. PLoS ONE, 2015, 10, e0116514.	1.1	24
4887	Identification of Aortic Arch-Specific Quantitative Trait Loci for Atherosclerosis by an Intercross of DBA/2J and 129S6 Apolipoprotein E-Deficient Mice. PLoS ONE, 2015, 10, e0117478.	1.1	19
4888	A Deep Catalog of Autosomal Single Nucleotide Variation in the Pig. PLoS ONE, 2015, 10, e0118867.	1.1	22
4889	BAP1 Missense Mutation c.2054 A>T (p.E685V) Completely Disrupts Normal Splicing through Creation of a Novel 5â€™ Splice Site in a Human Mesothelioma Cell Line. PLoS ONE, 2015, 10, e0119224.	1.1	9
4890	Evaluation of the Contribution of the EYA4 and GRHL2 Genes in Korean Patients with Autosomal Dominant Non-Syndromic Hearing Loss. PLoS ONE, 2015, 10, e0119443.	1.1	19
4891	TYK2 Protein-Coding Variants Protect against Rheumatoid Arthritis and Autoimmunity, with No Evidence of Major Pleiotropic Effects on Non-Autoimmune Complex Traits. PLoS ONE, 2015, 10, e0122271.	1.1	120
4892	Using the MCF10A/MCF10CA1a Breast Cancer Progression Cell Line Model to Investigate the Effect of Active, Mutant Forms of EGFR in Breast Cancer Development and Treatment Using Gefitinib. PLoS ONE, 2015, 10, e0125232.	1.1	27
4893	Leveraging the Power of High Performance Computing for Next Generation Sequencing Data Analysis: Tricks and Twists from a High Throughput Exome Workflow. PLoS ONE, 2015, 10, e0126321.	1.1	37
4894	Combining GWAS and RNA-Seq Approaches for Detection of the Causal Mutation for Hereditary Junctional Epidermolysis Bullosa in Sheep. PLoS ONE, 2015, 10, e0126416.	1.1	15
4895	A Novel Alpha Cardiac Actin (ACTC1) Mutation Mapping to a Domain in Close Contact with Myosin Heavy Chain Leads to a Variety of Congenital Heart Defects, Arrhythmia and Possibly Midline Defects. PLoS ONE, 2015, 10, e0127903.	1.1	14
4896	Targeted Resequencing of the Pericentromere of Chromosome 2 Linked to Constitutional Delay of Growth and Puberty. PLoS ONE, 2015, 10, e0128524.	1.1	10
4897	Comprehensive Analysis of Disease-Related Genes in Chronic Lymphocytic Leukemia by Multiplex PCR-Based Next Generation Sequencing. PLoS ONE, 2015, 10, e0129544.	1.1	23
4898	Screening for THAP1 Mutations in Polish Patients with Dystonia Shows Known and Novel Substitutions. PLoS ONE, 2015, 10, e0129656.	1.1	2
4899	Exome Analyses of Long QT Syndrome Reveal Candidate Pathogenic Mutations in Calmodulin-Interacting Genes. PLoS ONE, 2015, 10, e0130329.	1.1	30
4900	Truncating Homozygous Mutation of Carboxypeptidase E (CPE) in a Morbidly Obese Female with Type 2 Diabetes Mellitus, Intellectual Disability and Hypogonadotrophic Hypogonadism. PLoS ONE, 2015, 10, e0131417.	1.1	72
4901	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families. PLoS ONE, 2015, 10, e0131679.	1.1	36
4902	A Novel WRN Frameshift Mutation Identified by Multiplex Genetic Testing in a Family with Multiple Cases of Cancer. PLoS ONE, 2015, 10, e0133020.	1.1	11

#	ARTICLE	IF	CITATIONS
4903	Rare Circulating Cells in Familial Waldenström Macroglobulinemia Displaying the MYD88 L265P Mutation Are Enriched by Epstein-Barr Virus Immortalization. PLoS ONE, 2015, 10, e0136505.	1.1	6
4904	Association of Germline CHEK2 Gene Variants with Risk and Prognosis of Non-Hodgkin Lymphoma. PLoS ONE, 2015, 10, e0140819.	1.1	31
4905	Comprehensive Analysis of Deafness Genes in Families with Autosomal Recessive Nonsyndromic Hearing Loss. PLoS ONE, 2015, 10, e0142154.	1.1	52
4906	Whole Exome Sequencing of Rapid Autopsy Tumors and Xenograft Models Reveals Possible Driver Mutations Underlying Tumor Progression. PLoS ONE, 2015, 10, e0142631.	1.1	28
4907	Influence of Familial Renal Glycosuria Due to Mutations in the SLC5A2 Gene on Changes in Glucose Tolerance over Time. PLoS ONE, 2016, 11, e0146114.	1.1	22
4908	Clinical and Molecular Characterization of BSCL2 Mutations in a Taiwanese Cohort with Hereditary Neuropathy. PLoS ONE, 2016, 11, e0147677.	1.1	18
4909	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. PLoS ONE, 2016, 11, e0150555.	1.1	32
4910	ENTPRISE: An Algorithm for Predicting Human Disease-Associated Amino Acid Substitutions from Sequence Entropy and Predicted Protein Structures. PLoS ONE, 2016, 11, e0150965.	1.1	23
4911	Alterations in Fibronectin Type III Domain Containing 1 Protein Gene Are Associated with Hypertension. PLoS ONE, 2016, 11, e0151399.	1.1	15
4912	Î±1-Syntrophin Variant Identified in Drug-Induced Long QT Syndrome Increases Late Sodium Current. PLoS ONE, 2016, 11, e0152355.	1.1	10
4913	Investigation of Rare Single-Nucleotide PCDH15 Variants in Schizophrenia and Autism Spectrum Disorders. PLoS ONE, 2016, 11, e0153224.	1.1	15
4914	Exome Sequencing of a Pedigree Reveals S339L Mutation in the TLN2 Gene as a Cause of Fifth Finger Camptodactyly. PLoS ONE, 2016, 11, e0155180.	1.1	9
4915	Genomic Characteristics of Genetic Creutzfeldt-Jakob Disease Patients with V180I Mutation and Associations with Other Neurodegenerative Disorders. PLoS ONE, 2016, 11, e0157540.	1.1	7
4916	Identification of Novel and Recurrent Disease-Causing Mutations in Retinal Dystrophies Using Whole Exome Sequencing (WES): Benefits and Limitations. PLoS ONE, 2016, 11, e0158692.	1.1	20
4917	Plasmid Complement of Lactococcus lactis NCDO712 Reveals a Novel Pilus Gene Cluster. PLoS ONE, 2016, 11, e0167970.	1.1	34
4918	Single Nucleotide Variants of Candidate Genes in Aggrecan Metabolic Pathway Are Associated with Lumbar Disc Degeneration and Modic Changes. PLoS ONE, 2017, 12, e0169835.	1.1	11
4919	Molecular modeling and molecular dynamic simulation of the effects of variants in the TGFBR2 kinase domain as a paradigm for interpretation of variants obtained by next generation sequencing. PLoS ONE, 2017, 12, e0170822.	1.1	19
4920	Whole exome sequencing implicates eye development, the unfolded protein response and plasma membrane homeostasis in primary open-angle glaucoma. PLoS ONE, 2017, 12, e0172427.	1.1	8

#	ARTICLE	IF	CITATIONS
4921	Novel compound heterozygous MYO7A mutations in Moroccan families with autosomal recessive non-syndromic hearing loss. PLoS ONE, 2017, 12, e0176516.	1.1	8
4922	Whole exome sequencing of a consanguineous family identifies the possible modifying effect of a globally rare AK5 allelic variant in celiac disease development among Saudi patients. PLoS ONE, 2017, 12, e0176664.	1.1	14
4923	Biotinidase deficiency: Genotype-biochemical phenotype association in Brazilian patients. PLoS ONE, 2017, 12, e0177503.	1.1	19
4924	POU4F3 mutation screening in Japanese hearing loss patients: Massively parallel DNA sequencing-based analysis identified novel variants associated with autosomal dominant hearing loss. PLoS ONE, 2017, 12, e0177636.	1.1	31
4925	Development of next generation sequencing panel for UMOD and association with kidney disease. PLoS ONE, 2017, 12, e0178321.	1.1	4
4926	A case-control study on association of proteasome subunit beta 8 (PSMB8) and transporter associated with antigen processing 1 (TAP1) polymorphisms and their transcript levels in vitiligo from Gujarat. PLoS ONE, 2017, 12, e0180958.	1.1	26
4927	Variants of ACAN are associated with severity of lumbar disc herniation in patients with chronic low back pain. PLoS ONE, 2017, 12, e0181580.	1.1	11
4928	Whole gene sequencing identifies deep-intronic variants with potential functional impact in patients with hypertrophic cardiomyopathy. PLoS ONE, 2017, 12, e0182946.	1.1	41
4929	Whole exome sequencing as a diagnostic tool for patients with ciliopathy-like phenotypes. PLoS ONE, 2017, 12, e0183081.	1.1	8
4930	Telomere biology and telomerase mutations in cirrhotic patients with hepatocellular carcinoma. PLoS ONE, 2017, 12, e0183287.	1.1	20
4931	Whole-genome sequencing reveals mutational landscape underlying phenotypic differences between two widespread Chinese cattle breeds. PLoS ONE, 2017, 12, e0183921.	1.1	33
4932	Identification of a novel frameshift mutation in the ILDR1 gene in a UAE family, mutations review and phenotype genotype correlation. PLoS ONE, 2017, 12, e0185281.	1.1	9
4933	The frequency of cancer predisposition gene mutations in hereditary breast and ovarian cancer patients in Taiwan: From BRCA1/2 to multi-gene panels. PLoS ONE, 2017, 12, e0185615.	1.1	22
4934	Pea Marker Database (PMD) – A new online database combining known pea (Pisum sativum L.) gene-based markers. PLoS ONE, 2017, 12, e0186713.	1.1	30
4935	Mutational analysis of ITPR1 in a Taiwanese cohort with cerebellar ataxias. PLoS ONE, 2017, 12, e0187503.	1.1	13
4936	Comprehensive analysis of three TYK2 gene variants in the susceptibility to Chagas disease infection and cardiomyopathy. PLoS ONE, 2018, 13, e0190591.	1.1	4
4937	A homozygous missense variant in VWA2, encoding an interactor of the Fraser-complex, in a patient with vesicoureteral reflux. PLoS ONE, 2018, 13, e0191224.	1.1	5
4938	Airway ciliary dysfunction and respiratory symptoms in patients with transposition of the great arteries. PLoS ONE, 2018, 13, e0191605.	1.1	17

#	ARTICLE	IF	CITATIONS
4939	Genetic variation in the C-type lectin receptor CLEC4M in type 1 von Willebrand Disease patients. PLoS ONE, 2018, 13, e0192024.	1.1	8
4940	WFS1 mutation screening in a large series of Japanese hearing loss patients: Massively parallel DNA sequencing-based analysis. PLoS ONE, 2018, 13, e0193359.	1.1	33
4941	Germline mutations in candidate predisposition genes in individuals with cutaneous melanoma and at least two independent additional primary cancers. PLoS ONE, 2018, 13, e0194098.	1.1	16
4942	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants. PLoS ONE, 2020, 15, e0238529.	1.1	12
4943	Effects of germline and somatic events in candidate BRCA-like genes on breast-tumor signatures. PLoS ONE, 2020, 15, e0239197.	1.1	13
4944	Hepatic lipase (LIPC) sequencing in individuals with extremely high and low high-density lipoprotein cholesterol levels. PLoS ONE, 2020, 15, e0243919.	1.1	3
4945	Evolutionary analysis of TSP-1 gene in Plateau zokor (<i>MyospalaxBaileyi</i>) and its expression pattern under hypoxia. Cellular and Molecular Biology, 2019, 65, 48.	0.3	3
4946	Glycogen storage disease type VI can progress to cirrhosis: ten Chinese patients with GSD VI and a literature review. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1321-1333.	0.4	7
4947	A comprehensive in silico analysis, distribution and frequency of human Nkx2-5 mutations; A critical gene in congenital heart disease. Journal of Cardiovascular and Thoracic Research, 2019, 11, 287-299.	0.3	8
4948	Bi-allelic pathogenic variants in <i>NDUFC2</i> cause early-onset Leigh syndrome and stalled biogenesis of complex I. EMBO Molecular Medicine, 2020, 12, e12619.	3.3	17
4949	Novel SLCO2A1 mutations cause gender-differentiated pachydermoperiostosis. Endocrine Connections, 2018, 7, 1116-1128.	0.8	6
4950	Next-generation sequencing refines the genetic architecture of Greek GnRH-deficient patients. Endocrine Connections, 2019, 8, 468-480.	0.8	16
4951	Intratumor heterogeneity of prognostic DNA-based molecular markers in adrenocortical carcinoma. Endocrine Connections, 2020, 9, 705-714.	0.8	10
4952	Systematic alanine scanning of PAX8 paired domain reveals functional importance of the N-subdomain. Journal of Molecular Endocrinology, 2019, 62, 129-135.	1.1	11
4953	Adrenocortical cancer cell line mutational profile reveals aggressive genetic background. Journal of Molecular Endocrinology, 2019, 62, 179-186.	1.1	7
4955	A Novel Mutation in the BCKDHB Gene Causes in an Iranian Child Classic Maple Syrup Urine Disease. Zahedan Journal of Researches in Medical Sciences, 2016, In Press, .	0.1	2
4956	Screening for Novel LOX and SOD1 Variants in Keratoconus Patients from Brazil. Journal of Ophthalmic and Vision Research, 2020, 15, 138-148.	0.7	10
4957	A novel autosomal recessive TERT T1129P mutation in a dyskeratosis congenita family leads to cellular senescence and loss of CD34+ hematopoietic stem cells not reversible by mTOR-inhibition. Aging, 2015, 7, 911-927.	1.4	13

#	ARTICLE	IF	CITATIONS
4958	Identification of polymorphisms in cancer patients that differentially affect survival with age. <i>Aging</i> , 2017, 9, 2117-2136.	1.4	8
4959	Mutation profiling of 19 candidate genes in acute myeloid leukemia suggests significance of <i>DNMT3A</i> mutations. <i>Oncotarget</i> , 2016, 7, 54825-54837.	0.8	22
4960	Association mining of mutated cancer genes in different clinical stages across 11 cancer types. <i>Oncotarget</i> , 2016, 7, 68270-68277.	0.8	9
4961	Recurrent mutations in NF- κ B pathway components, KMT2D, and NOTCH1/2 in ocular adnexal MALT-type marginal zone lymphomas. <i>Oncotarget</i> , 2016, 7, 62627-62639.	0.8	52
4962	Genomic characterization of pediatric T-cell acute lymphoblastic leukemia reveals novel recurrent driver mutations. <i>Oncotarget</i> , 2016, 7, 65485-65503.	0.8	54
4963	Germline mutations in Japanese familial pancreatic cancer patients. <i>Oncotarget</i> , 2016, 7, 74227-74235.	0.8	62
4964	High throughput estimation of functional cell activities reveals disease mechanisms and predicts relevant clinical outcomes. <i>Oncotarget</i> , 2017, 8, 5160-5178.	0.8	66
4965	Intratumoral heterogeneity of intrahepatic cholangiocarcinoma. <i>Oncotarget</i> , 2017, 8, 14957-14968.	0.8	34
4966	Synthetic lethal interaction between the tumour suppressor <i>STAG2</i> and its paralog <i>STAG1</i> . <i>Oncotarget</i> , 2017, 8, 37619-37632.	0.8	61
4967	Whole-exome sequencing identified genetic risk factors for asparaginase-related complications in childhood ALL patients. <i>Oncotarget</i> , 2017, 8, 43752-43767.	0.8	33
4968	A novel mutation R190H in the AT-hook 1 domain of MeCP2 identified in an atypical Rett syndrome. <i>Oncotarget</i> , 2017, 8, 82156-82164.	0.8	7
4969	A comprehensive genomic meta-analysis identifies confirmatory role of <i>OBSCN</i> gene in breast tumorigenesis. <i>Oncotarget</i> , 2017, 8, 102263-102276.	0.8	23
4970	Exome sequencing identified a novel missense mutation c.464G>A (p.G155D) in Ca ²⁺ -binding protein 4 (<i>CABP4</i>) in a Chinese pedigree with autosomal dominant nocturnal frontal lobe epilepsy. <i>Oncotarget</i> , 2017, 8, 78940-78947.	0.8	17
4971	Mutational analysis of primary central nervous system lymphoma. <i>Oncotarget</i> , 2014, 5, 5065-5075.	0.8	149
4972	Targeted ultra-deep sequencing reveals recurrent and mutually exclusive mutations of cancer genes in blastic plasmacytoid dendritic cell neoplasm. <i>Oncotarget</i> , 2014, 5, 6404-6413.	0.8	82
4973	Genetic identification and molecular modeling characterization reveal a novel <i>PROM1</i> mutation in Stargardt4-like macular dystrophy. <i>Oncotarget</i> , 2018, 9, 122-141.	0.8	32
4974	Identification of the genetic and clinical characteristics of neuroblastomas using genome-wide analysis. <i>Oncotarget</i> , 2017, 8, 107513-107529.	0.8	23
4975	Fanconi Anemia germline variants as susceptibility factors in aplastic anemia, MDS and AML. <i>Oncotarget</i> , 2018, 9, 2050-2057.	0.8	16

#	ARTICLE	IF	CITATIONS
4976	Catalytic mTOR inhibitors can overcome intrinsic and acquired resistance to allosteric mTOR inhibitors. <i>Oncotarget</i> , 2014, 5, 8544-8557.	0.8	56
4977	Single cell whole genome sequencing reveals that NFKB1 mutation affects radiotherapy sensitivity in cervical cancer. <i>Oncotarget</i> , 2018, 9, 7332-7340.	0.8	14
4978	The transcriptional and mutational landscapes of lipid metabolism-related genes in colon cancer. <i>Oncotarget</i> , 2018, 9, 5919-5930.	0.8	28
4979	Mutational analysis of genes coding for cell surface proteins in colorectal cancer cell lines reveal novel altered pathways, druggable mutations and mutated epitopes for targeted therapy. <i>Oncotarget</i> , 2014, 5, 9199-9213.	0.8	31
4980	Mutational landscape of radiation-associated angiosarcoma of the breast. <i>Oncotarget</i> , 2018, 9, 10042-10053.	0.8	21
4981	Identification of different mutational profiles in cancers arising in specific colon segments by next generation sequencing. <i>Oncotarget</i> , 2018, 9, 23960-23974.	0.8	13
4982	Deep sequencing of a recurrent oligodendroglioma and the derived xenografts reveals new insights into the evolution of human oligodendroglioma and candidate driver genes. <i>Oncotarget</i> , 2019, 10, 3641-3653.	0.8	1
4983	Germline mutations in cancer-predisposition genes in patients with biliary tract cancer. <i>Oncotarget</i> , 2019, 10, 5949-5957.	0.8	9
4984	Clonal expansion and linear genome evolution through breast cancer progression from pre-invasive stages to asynchronous metastasis. <i>Oncotarget</i> , 2015, 6, 5634-5649.	0.8	42
4985	Immunohistochemical and genomic profiles of diffuse large B-cell lymphomas: Implications for targeted EZH2 inhibitor therapy?. <i>Oncotarget</i> , 2015, 6, 16712-16724.	0.8	32
4986	Cytoplasmic accumulation of NCoR in malignant melanoma: consequences of altered gene repression and prognostic significance. <i>Oncotarget</i> , 2015, 6, 9284-9294.	0.8	8
4987	Multiple mechanisms of MYCN dysregulation in Wilms tumour. <i>Oncotarget</i> , 2015, 6, 7232-7243.	0.8	85
4988	Integrated genomic analyses identify frequent gene fusion events and <i>VHL</i> inactivation in gastrointestinal stromal tumors. <i>Oncotarget</i> , 2016, 7, 6538-6551.	0.8	29
4989	Identification of GALNT14 as a novel neuroblastoma predisposition gene. <i>Oncotarget</i> , 2015, 6, 26335-26346.	0.8	43
4990	Involvement of <i>MBD4</i> inactivation in mismatch repair-deficient tumorigenesis. <i>Oncotarget</i> , 2015, 6, 42892-42904.	0.8	43
4991	Elucidating the cancer-specific genetic alteration spectrum of glioblastoma derived cell lines from whole exome and RNA sequencing. <i>Oncotarget</i> , 2015, 6, 43452-43471.	0.8	62
4992	Polo-like kinase 1 mediates BRCA1 phosphorylation and recruitment at DNA double-strand breaks. <i>Oncotarget</i> , 2016, 7, 2269-2283.	0.8	27
4993	Clinical implications of <i>TP53</i> mutations in myelodysplastic syndromes treated with hypomethylating agents. <i>Oncotarget</i> , 2016, 7, 14172-14187.	0.8	86

#	ARTICLE	IF	CITATIONS
4994	Dynamic epigenetic changes to <i>VHL</i> occur with sunitinib in metastatic clear cell renal cancer. <i>Oncotarget</i> , 2016, 7, 25241-25250.	0.8	14
4995	The degree of intratumor mutational heterogeneity varies by primary tumor sub-site. <i>Oncotarget</i> , 2016, 7, 27185-27198.	0.8	37
4996	Targeted sequencing identifies genetic alterations that confer primary resistance to EGFR tyrosine kinase inhibitor (Korean Lung Cancer Consortium). <i>Oncotarget</i> , 2016, 7, 36311-36320.	0.8	44
4997	MAPK activation and <i>HRAS</i> mutation identified in pituitary spindle cell oncocytoma. <i>Oncotarget</i> , 2016, 7, 37054-37063.	0.8	27
4998	Two novel mutations of PEX6 in one Chinese Zellweger spectrum disorder and their clinical characteristics. <i>Annals of Translational Medicine</i> , 2019, 7, 368-368.	0.7	15
4999	TILLING in European Rice: Hunting Mutations for Crop Improvement. <i>Crop Science</i> , 2013, 53, 2550-2562.	0.8	11
5000	Wild Mice As Bountiful Resources of Novel Genetic Variants for Quantitative Traits. <i>Current Genomics</i> , 2013, 14, 225-229.	0.7	6
5001	Probable Novel PSEN1 Gln222Leu Mutation in a Chinese Family with Early-Onset Alzheimer's Disease. <i>Current Alzheimer Research</i> , 2019, 16, 764-769.	0.7	2
5002	Monogenic Diabetes: Genetics and Relevance on Diabetes Mellitus Personalized Medicine. <i>Current Diabetes Reviews</i> , 2020, 16, 807-819.	0.6	14
5003	Identifying Extreme Observations, Outliers and Noise in Clinical and Genetic Data. <i>Current Bioinformatics</i> , 2017, 12, 101-117.	0.7	5
5004	GTF2I Mutations Are Common in Thymic Epithelial Tumors But Not in Hematological Malignancies. , 2017, 37, 5459-5462.		9
5005	Association of -c.894G>T transversion with susceptibility to metabolic syndrome in Azar-cohort population: A case-control study and analysis of the SNP molecular effects. <i>Iranian Journal of Basic Medical Sciences</i> , 2021, 24, 408-419.	1.0	4
5006	Functional investigation of the Val1714Gly and Asp1733Gly variants by computational tools and yeast transcription activation assay. <i>Molecular Biology Research Communications</i> , 2019, 8, 113-118.	0.2	3
5007	In silico analysis for determining the deleterious nonsynonymous single nucleotide polymorphisms of genes. <i>Molecular Biology Research Communications</i> , 2019, 8, 141-150.	0.2	9
5008	Moyamoya vasculopathy shows a genetic mutational gradient decreasing from East to West. <i>Journal of Neurosurgical Sciences</i> , 2020, 64, 165-172.	0.3	17
5009	TRAP1 chaperone protein mutations and autoinflammation. <i>Life Science Alliance</i> , 2020, 3, e201900376.	1.3	9
5010	Whole Exome Sequencing Reveals a XPNPEP3 Novel Mutation Causing Nephronophthisis in a Pediatric Patient. <i>Iranian Biomedical Journal</i> , 2020, 24, 400-403.	0.4	4
5011	Molecular Mechanism of Cancer Susceptibility Associated with FokI Single Nucleotide Polymorphism of VDR in Relation to Breast Cancer. <i>Asian Pacific Journal of Cancer Prevention</i> , 2019, 20, 199-206.	0.5	10

#	ARTICLE	IF	CITATIONS
5012	The Nudix Hydrolase 15 (NUDT15) Gene Variants among Jordanian Arab Population. <i>Asian Pacific Journal of Cancer Prevention</i> , 2019, 20, 801-808.	0.5	10
5013	Somatic Mitochondrial Mutations in Oral Cavity Cancers among Senegalese Patients. <i>Asian Pacific Journal of Cancer Prevention</i> , 2019, 20, 2203-2208.	0.5	1
5014	Dent's disease: Identification of seven new pathogenic mutations in the CLCN5 gene. <i>Journal of Pediatric Genetics</i> , 2013, 2, 133-40.	0.3	4
5015	Molecular Genetic Background of an Autosomal Dominant Hypercholesterolemia in the Czech Republic. <i>Physiological Research</i> , 2017, 66, S47-S54.	0.4	6
5016	HER2 Status in High-Risk Endometrial Cancers (PORTEC-3): Relationship with Histotype, Molecular Classification, and Clinical Outcomes. <i>Cancers</i> , 2021, 13, 44.	1.7	40
5017	IL-12 and IL-23 "Close Relatives with Structural Homologies but Distinct Immunological Functions. <i>Cells</i> , 2020, 9, 2184.	1.8	31
5018	Whole exome sequencing identifies genomic alterations in proximal and distal colorectal cancer. <i>Bulletin of the Geological Society of Malaysia</i> , 2019, 2, .	0.5	1
5020	A recurrent mutation in bone morphogenetic proteins-2-inducible kinase gene is associated with developmental dysplasia of the hip. <i>Experimental and Therapeutic Medicine</i> , 2017, 13, 1773-1778.	0.8	16
5021	Biallelic mutations in carbamoyl phosphate synthetase 1 induced hyperammonemia in a neonate: A case report. <i>Experimental and Therapeutic Medicine</i> , 2020, 20, 623-629.	0.8	2
5022	ALDH1L1 variant rs2276724 and mRNA expression predict post-operative clinical outcomes and are associated with TP53 expression in HBV-related hepatocellular carcinoma. <i>Oncology Reports</i> , 2017, 38, 1451-1463.	1.2	13
5023	Retinal dystrophies, genomic applications in diagnosis and prospects for therapy. <i>Translational Pediatrics</i> , 2015, 4, 139-63.	0.5	62
5024	Novel mutations of the arylsulphatase B (ARSB) gene in Indian patients with mucopolysaccharidosis type VI. <i>Indian Journal of Medical Research</i> , 2015, 142, 414.	0.4	9
5025	Exploration of structural stability in deleterious nsSNPs of the XPA gene: A molecular dynamics approach. <i>Journal of Carcinogenesis</i> , 2011, 10, 26.	2.5	3
5026	A Rare Missense Mutation and a Polymorphism with High Frequency in LDLR Gene among Iranian Patients with Familial Hypercholesterolemia. <i>Advanced Biomedical Research</i> , 2018, 7, 37.	0.2	3
5027	Investigation of hub genes and their nonsynonymous single nucleotide polymorphism analysis in <i>Plasmodium falciparum</i> for designing therapeutic methodologies using next-generation sequencing approach. <i>Indian Journal of Pharmacology</i> , 2019, 51, 389.	0.4	1
5028	Discovery of DiPeptidyl Peptidase-4 Gene Variants and the Associations with Efficacy of Vildagliptin in Patients with Type 2 Diabetes - A Pilot Study. <i>Journal of Diabetes & Metabolism</i> , 2013, 01, .	0.2	2
5029	Comprehensive Analysis of rsSNPs Associated with Hypertension Using In-Silico Bioinformatics Tools. <i>Open Access Library Journal (oalib)</i> , 2016, 03, 1-24.	0.1	6
5030	Targeted Next-Generation Sequencing of MLH1, MSH2, and MSH6 Genes in Patients with Endometrial Carcinoma under 50 Years of Age. <i>Balkan Medical Journal</i> , 2019, 36, 37-42.	0.3	4

#	ARTICLE	IF	CITATIONS
5031	Genetic and Clinical Characteristics of Patients with Vitamin D Dependent Rickets Type 1A. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 34-40.	0.4	18
5032	Three Siblings with Idiopathic Hypogonadotropic Hypogonadism in a Nonconsanguineous Family: A Novel <i><i>KISS1R/GPR54</i></i> Loss-of-Function Mutation. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 444-448.	0.4	5
5033	Xanthomas Can Be Misdiagnosed and Mistreated in Homozygous Familial Hypercholesterolemia Patients: A Call for Increased Awareness Among Dermatologists and Health Care Practitioners. Global Heart, 2020, 15, 19.	0.9	10
5034	Value of predictive bioinformatics in inherited metabolic diseases. World Journal of Medical Genetics, 2015, 5, 46.	1.0	5
5035	Whole Exome Sequencing Reveals Multiple Mutations in Uncommon Genes of Familial Hypercholesterolaemia. Journal of Cardiovascular Disease Research (discontinued), 2019, 10, 09-15.	0.1	3
5036	Deep Sequencing Data Analysis: Challenges and Solutions. , 0, , .		4
5037	A Follow-up Association Study of Genetic Variants for Bone Mineral Density in a Korean Population. Genomics and Informatics, 2014, 12, 114.	0.4	3
5038	Prevalence and Phenotypic Expression of Mutations in the MYH7, MYBPC3 and TNNT2 Genes in Families with Hypertrophic Cardiomyopathy in the South of Brazil: A Cross-Sectional Study. Arquivos Brasileiros De Cardiologia, 2016, 107, 257-265.	0.3	6
5039	Germline Variations of Apurinic/Apyrimidinic Endonuclease 1 (APEX1) Detected in Female Breast Cancer Patients. Asian Pacific Journal of Cancer Prevention, 2014, 15, 7589-7595.	0.5	8
5040	Massive Parallel Sequencing for Diagnostic Genetic Testing of BRCA Genes - a Single Center Experience. Asian Pacific Journal of Cancer Prevention, 2015, 16, 7935-7941.	0.5	12
5041	Prediction and Analysis of Breast Cancer Related Deleterious Non-Synonymous Single Nucleotide Polymorphisms in the PTEN Gene. Asian Pacific Journal of Cancer Prevention, 2016, 17, 2199-2203.	0.5	4
5042	Perusal of Mbl2 Gene -Susceptibility to Tuberculosis in Different Indian Populations. Journal of Applied Pharmaceutical Science, 0, , 097-099.	0.7	1
5043	Predicting mutational routes to new adaptive phenotypes. ELife, 2019, 8, .	2.8	55
5044	Genetic diversity of CHC22 clathrin impacts its function in glucose metabolism. ELife, 2019, 8, .	2.8	22
5045	Sequence variant analysis of RNA sequences in severe equine asthma. PeerJ, 2018, 6, e5759.	0.9	8
5046	VaRank: a simple and powerful tool for ranking genetic variants. PeerJ, 2015, 3, e796.	0.9	80
5047	Accumulation of sequence variants in genes of Wnt signaling and focal adhesion pathways in human corneas further explains their involvement in keratoconus. PeerJ, 2020, 8, e8982.	0.9	12
5048	Global RNA editome landscape discovers reduced RNA editing in glioma: loss of editing of gamma-amino butyric acid receptor alpha subunit 3 (GABRA3) favors glioma migration and invasion. PeerJ, 2020, 8, e9755.	0.9	13

#	ARTICLE	IF	CITATIONS
5049	PLA2G6 Mutations Related to Distinct Phenotypes: A New Case with Early-onset Parkinsonism. Tremor and Other Hyperkinetic Movements, 2016, 6, 363.	1.1	11
5050	Intermediate Phenotypes of ATP1A3 Mutations: Phenotype-Genotype Correlations. Tremor and Other Hyperkinetic Movements, 2015, 5, 336.	1.1	18
5051	KCTD7-related progressive myoclonic epilepsy: report of three Indian families and review of literature. Clinical Dysmorphology, 2022, 31, 6-10.	0.1	4
5052	A Novel Variant of <i>ATP5MC3</i> Associated with Both Dystonia and Spastic Paraplegia. Movement Disorders, 2022, 37, 375-383.	2.2	10
5053	Prenatal diagnosis of a nonsense mutation in the L1CAM gene resulting in congenital hydrocephalus: A case report and literature review. Experimental and Therapeutic Medicine, 2021, 22, 1416.	0.8	4
5054	Clinical Characteristics and Mutation Spectrum of Neurofibromatosis Type 1 in 27 Turkish Families. , 2021, 38, 365-373.		4
5056	Personalized treatment with retigabine for pharmacoresistant epilepsy arising from a pathogenic variant in the KCNQ2 selectivity filter. Epileptic Disorders, 2021, 23, 695-705.	0.7	14
5057	Which Is the Best In Silico Program for the Missense Variations in IDUA Gene? A Comparison of 33 Programs Plus a Conservation Score and Evaluation of 586 Missense Variants. Frontiers in Molecular Biosciences, 2021, 8, 752797.	1.6	5
5058	Prevalence and Clinical Characteristics of Hearing Loss Caused by MYH14 Variants. Genes, 2021, 12, 1623.	1.0	5
5059	A DNA Damage Response Gene Panel for Different Histologic Types of Epithelial Ovarian Carcinomas and Their Outcomes. Biomedicines, 2021, 9, 1384.	1.4	4
5060	An Investigation of the Role of Common and Rare Variants in a Large Italian Multiplex Family of Multiple Sclerosis Patients. Genes, 2021, 12, 1607.	1.0	4
5061	Exploration of weighting schemes based on allele frequency and annotation for weighted burden association analysis of complex phenotypes. Gene, 2022, 809, 146039.	1.0	5
5062	Primary antibody deficiencies in Turkey: molecular and clinical aspects. Immunologic Research, 2021, , 1.	1.3	2
5063	Detection and analysis of common pathogenic germline mutations in Peutz-Jeghers syndrome. World Journal of Gastroenterology, 2021, 27, 6631-6646.	1.4	6
5064	Whole-exome sequencing identifies an RS1 variant in a Chinese family with X-linked retinoschisis. Experimental and Therapeutic Medicine, 2021, 22, 1406.	0.8	1
5065	Comprehensive Identification of Deleterious TP53 Missense VUS Variants Based on Their Impact on TP53 Structural Stability. International Journal of Molecular Sciences, 2021, 22, 11345.	1.8	5
5066	Improving Prediction Accuracy via Subspace Modeling in a Statistical Geometry Based Computational Protein Mutagenesis. International Journal of Knowledge Discovery in Bioinformatics, 2010, 1, 54-68.	0.8	0
5068	Genetic Instability in Gastric Cancer. , 0, , .		1

#	ARTICLE	IF	CITATIONS
5069	Disease Gene Prioritization. , 0, , .		0
5070	Analytical Approaches for Exome Sequence Data. Translational Bioinformatics, 2012, , 105-120.	0.0	0
5072	Mutational Analysis. , 2013, , 540-541.		0
5073	Analysis pipeline for the detection of mutations causative of rare diseases on whole exome sequencing data. EMBnet Journal, 2013, 19, 79.	0.2	0
5074	A computational approach to analyze the missense mutations in human angiogenin variants leading to amyotrophic lateral sclerosis. Bangladesh Journal of Pharmacology, 2013, 8, .	0.1	1
5078	Intelligent Integrative Knowledge Bases: Bridging Genomics, Integrative Biology and Translational Medicine. Lecture Notes in Computer Science, 2014, , 255-270.	1.0	0
5079	The 2013 Incentive Award of the Okayama Medical Association in General Medical Science (2013 Yuuki) Tj ETQq0 0,0 rgBT /Overlock 10	0.0	0
5080	Application of Targeted Next-Generation Sequencing in Patients with Autosomal Recessive Inherited Retinal Dystrophies: Improvement of Genetic and Clinical Diagnosis. Translational Medicine (Sunnyvale, Calif), 2014, 04, .	0.4	0
5081	Designing Algorithms for Determining Significance of DNA Missense Changes. Methods in Molecular Biology, 2014, 1168, 251-262.	0.4	0
5082	Collagen gene interactions and endurance running performance. SA Sports Medicine, 2014, 26, 9-14.	0.1	1
5084	In-Silico Prediction of Candidate SNPs in TRIOBP, TMC1 and EYA4 Genes Causing Hereditary Deafness in Three Sudanese Patients Using Next Generation Sequencing. Nova Journal of Medical and Biological Sciences, 2014, 03, 1-10.	0.0	0
5085	Next Generation Sequencing in Cancer Research and Clinical Application. , 2014, , 71-94.		1
5087	MICO: A meta-tool for prediction of the effects of non-synonymous mutations. Bioinformatics, 2014, 10, 469-471.	0.2	0
5088	Current Massively Parallel Sequencing Technologies: Platforms and Reporting Considerations. , 2015, , 3-17.		0
5089	Genomic Applications in Pharmacogenomics. , 2015, , 553-562.		0
5093	Natural Selection at Rare Variants. , 2015, , 123-133.		0
5101	Translational Research Methods: Basics of Renal Molecular Biology. , 2016, , 425-445.		0
5103	An Improved Algorithm Based on SURF for MR Infant Brain Image Registration. Lecture Notes in Computer Science, 2016, , 458-470.	1.0	0

#	ARTICLE	IF	CITATIONS
5104	Genomic Analysis. , 2016, , 83-106.		0
5108	A pilot exome-wide association study of age-related cataract in Koreans. Journal of Biomedical Research, 2016, 30, 186-90.	0.7	4
5111	Evaluation of the RYR1 gene genetic diversity in the Latvian White pig breed. Biopolymers and Cell, 2016, 32, 184-189.	0.1	1
5117	Genetic and Genomic Approaches to Acute Lung Injury. Respiratory Medicine, 2017, , 133-159.	0.1	0
5120	Gerbode Ventricular Septal Defect â€“A Rare Cardiac Anomaly Associated with Genetic Variants in Indian Population- A Case Series. Journal of Clinical and Diagnostic Research JCDR, 2017, 11, GR01-GR04.	0.8	3
5122	Cancer Specific Non-Synonymous Single Nucleotide Polymorphism Prediction in the Context of Haplotype and Protein Interacting Sites. Journal of Biomedical Science and Engineering, 2017, 10, 28-44.	0.2	0
5125	Familial Eosinophilic Granulomatosis with Polyangiitis. Open Journal of Rheumatology and Autoimmune Diseases, 2017, 07, 137-146.	0.1	1
5126	Detection of Somatic Mutations in Gastroenteropancreatic Neuroendocrine Tumors Using Targeted Deep Sequencing. Anticancer Research, 2017, 37, 705-712.	0.5	3
5131	Genetic analysis of single-minded 1 gene in early-onset severely obese children and adolescents. PLoS ONE, 2017, 12, e0177222.	1.1	4
5135	Acute lymphoblastic leukemia and genetic variations in BHMT gene: Case-control study and computational characterization. Cancer Biomarkers, 2017, 19, 393-401.	0.8	3
5139	Genomic Variant Classifier Tool. Lecture Notes in Networks and Systems, 2018, , 453-456.	0.5	0
5141	Next-Generation Sequencing Reveals One Novel Missense Mutation in COL1A2 Gene in an Iranian Family with Osteogenesis imperfecta. Iranian Biomedical Journal, 2017, 21, 338-342.	0.4	1
5142	Characterizing the polymorphism K232A of the diacylglycerol-acyltransferase-1 lipogenic enzyme of bovine Bos taurus using in silico comparative protein prediction analyses. Brazilian Journal of Biology, 2018, 78, 389-390.	0.4	1
5147	Whole-exome sequencing for detecting inborn errors of immunity: overview and perspectives. F1000Research, 2017, 6, 2056.	0.8	3
5150	Fully Automatic Synaptic Cleft Detection and Segmentation from EM Images Based on Deep Learning. Lecture Notes in Computer Science, 2018, , 64-74.	1.0	4
5151	Germline Genetic IKZF1 Variation and Predisposition to Childhood Acute Lymphoblastic Leukemia. SSRN Electronic Journal, 0, , .	0.4	0
5152	Identification of Acquired Notch3 Dependency in Metastatic Head and Neck Cancer. SSRN Electronic Journal, 0, , .	0.4	0
5154	Computational Prediction of Driver Missense Mutations in Melanoma. Lecture Notes in Computer Science, 2018, , 438-447.	1.0	0

#	ARTICLE	IF	CITATIONS
5155	Single-Cell Whole-Genome Sequencing Reveals B Lymphocyte Mutational Landscapes Across the Human Lifespan. SSRN Electronic Journal, 0, , .	0.4	0
5158	A novel γ 160c mutation of keratin 10 gene in a chinese male infant with epidermolytic hyperkeratosis. Turkish Journal of Pediatrics, 2018, 60, 426.	0.3	0
5161	Pathway-based analysis of genome-wide association study of circadian phenotypes. Journal of Biomedical Research, 2018, 32, 361-370.	0.7	0
5163	POSTLINGUAL SENSORINEURAL HEARING LOSS DUE TO A VERY RARE COCH PATHOGENIC VARIANT. Journal of Hearing Science, 2018, 8, 31-37.	0.1	1
5179	In silico approaches to identify the functional and structural effects of non-synonymous SNPs in selective sweeps of the Berkshire pig genome. Asian-Australasian Journal of Animal Sciences, 2018, 31, 1150-1159.	2.4	2
5192	Implementation of Genome Sequencing Assays. , 2019, , 219-236.		0
5193	Current Massively Parallel Sequencing Technologies: Platforms and Reporting Considerations. , 2019, , 11-21.		0
5196	Genomic Revolution-Driven Cancer Research. , 2019, , 39-60.		0
5197	Simultaneous detection of target CNVs and SNVs of thalassemia by multiplex PCR and nextâ€¢generation sequencing. Molecular Medicine Reports, 2019, 19, 2837-2848.	1.1	7
5198	A Method of Penicillin Bottle Defect Inspection Based on BP Neural Network. Communications in Computer and Information Science, 2019, , 31-40.	0.4	0
5199	Development of Flax (<i>Linum usitatissimum</i> L.) Mutant Populations for Forward and Reverse Genetics. Plant Genetics and Genomics: Crops and Models, 2019, , 145-165.	0.3	0
5201	Probabilistic Models for Predicting Mutational Routes to New Adaptive Phenotypes. Bio-protocol, 2019, 9, e3407.	0.2	1
5202	PGRN and Neurodegenerative Diseases Other Than FTL. , 2019, , 71-84.		0
5207	In Silico Analysis of the Novel Variant Q375R in the Phenylalanine Hydroxylase Gene. Gene, Cell and Tissue, 2019, In Press, .	0.2	0
5215	The identification of SNPs in THCA synthase gene from Pakistani Cannabis. Asia-Pacific Journal of Molecular Biology and Biotechnology, 0, , 1-9.	0.2	1
5217	in silico Single Nucleotide Polymorphism Prediction and Design for Targeting Amyloid Precursor Protein in Alzheimers Disease. Asian Journal of Chemistry, 2019, 31, 1959-1965.	0.1	0
5218	TMCO1 Gen Sekans VaryanlatlarÄ±nÄ±n Fonksiyonel Ä–zelliklerinin In Silico Analizlerle DeÄ–Yerlendirilmesi. DÄ–zce Äœniversitesi Bilim Ve Teknoloji Dergisi, 2019, 7, 1931-1946.	0.2	0
5230	Effect of non synonymous SNP on JAK1 protein structure and subsequent function. Bioinformation, 2019, 15, 723-729.	0.2	1

#	ARTICLE	IF	CITATIONS
5232	PILOT RESEARCH OF A GENETIC PREDISPOSITION FOR CLINICAL MANIFESTATIONS OF ACUTE INTERMITTENT PORPHYRIA. <i>Gematologiya I Transfuziologiya</i> , 2019, 64, 123-137.	0.1	1
5236	Epidemiologic, clinical and pathogenesis features of achromatopsia in the Russian population. <i>Rossiiskii Oftal'mologicheskii Zhurnal</i> , 2020, 13, 12-22.	0.1	2
5237	Exomes of Ductal Luminal Breast Cancer Patients from Southwest Colombia: Gene Mutational Profile and Related Expression Alterations. <i>Biomolecules</i> , 2020, 10, 698.	1.8	4
5239	Once in a Blue Moon, a Very Rare Coexistence of Glutaric Acidemia Type I and Mucopolysaccharidosis Type IIIB in a Patient. <i>Iranian Biomedical Journal</i> , 2020, 24, 201-205.	0.4	2
5240	Identification of nsSNPs of transcription factor E2F1 predisposing individuals to lung cancer and head and neck cancer. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2020, 821, 111704.	0.4	7
5242	Identification of a novel titin-cap/telethonin mutation in a Portuguese family with hypertrophic cardiomyopathy. <i>Revista Portuguesa De Cardiologia (English Edition)</i> , 2020, 39, 317-327.	0.2	0
5246	Massive parallel sequencing in individuals with multiple primary tumours reveals the benefit of re-analysis. <i>Hereditary Cancer in Clinical Practice</i> , 2021, 19, 46.	0.6	3
5247	De novo mutation in KITLG gene causes a variant of Familial Progressive Hyper- and Hypo-pigmentation (FPHH). <i>Molecular Genetics & Genomic Medicine</i> , 2021, , e1841.	0.6	3
5248	Integrative assessment of CIP2A overexpression and mutational effects in human malignancies identifies possible deleterious variants. <i>Computers in Biology and Medicine</i> , 2021, 139, 104986.	3.9	6
5249	A novel synonymous ABCA3 variant identified in a Chinese family with lethal neonatal respiratory failure. <i>BMC Medical Genomics</i> , 2021, 14, 256.	0.7	2
5250	Molecular evolution of PCSK family: Analysis of natural selection rate and gene loss. <i>PLoS ONE</i> , 2021, 16, e0259085.	1.1	2
5251	Clinical and molecular spectrum of tuberous sclerosis complex patients: identification of three novel mutations. <i>Erciyes Medical Journal</i> , 2020, , .	0.0	0
5252	FGFR4 c.1162G>A (p.Gly388Arg) Polymorphism Analysis in Turkish Patients with Retinoblastoma. <i>Journal of Oncology</i> , 2020, 2020, 1-8.	0.6	2
5253	Marfan syndrome: whole-exome sequencing reveals de novo mutations, second gene and genotype-phenotype correlations in the Chinese population. <i>Bioscience Reports</i> , 2020, 40, .	1.1	8
5254	Automated Calculation of Fundamental Matrix from Stereo Images from a Different Point of View. , 2021, , 105-118.		0
5256	Vertical transmission of a large calvarial ossification defect due to heterozygous variants of ALX4 and TWIST1. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 916-922.	0.7	1
5258	TargetMM: Accurate Missense Mutation Prediction by Utilizing Local and Global Sequence Information with Classifier Ensemble. <i>Combinatorial Chemistry and High Throughput Screening</i> , 2021, 25, 38-52.	0.6	5
5260	Sequence-Based Structural and Evolution of Polymorphisms in Bovine Toll-Like Receptor2 Gene in Dhanni and Jersey Cattle Breeds. <i>Genetic polymorphisms in bovine TLR2</i> . <i>Russian Journal of Genetics</i> , 2020, 56, 1484-1495.	0.2	1

#	ARTICLE	IF	CITATIONS
5261	Phenotypic differences of mutation-negative cases in Gitelman syndrome clinically diagnosed in adulthood. <i>Human Mutation</i> , 2021, 42, 300-309.	1.1	4
5262	<i>De novo FZR1</i> loss-of-function variants cause developmental and epileptic encephalopathies. <i>Brain</i> , 2022, 145, 1684-1697.	3.7	5
5264	Clinical and molecular characterization of four patients with NTCP deficiency from two unrelated families harboring the novel SLC10A1 variant c.595A>C (p.Ser199Arg). <i>Molecular Medicine Reports</i> , 2019, 20, 4915-4924.	1.1	9
5267	Diagnostische Verfahren. , 2020, , 551-631.		0
5268	Novel mutations in breast cancer patients from southwestern Colombia. <i>Genetics and Molecular Biology</i> , 2020, 43, e20190359.	0.6	1
5269	Factor 8 Gene Mutation Spectrum of 270 Patients with Haemophilia A: Identification of 36 Novel Mutations. <i>Turkish Journal of Haematology</i> , 2020, 37, 145-153.	0.2	9
5273	Hypophosphatasia: A Novel Mutation Associated with an Atypical Newborn Presentation. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , 2020, 12, 104-108.	0.4	4
5277	Heterogeneity of Axenfeld-Rieger Syndrome: Molecular and Clinical Findings in Chinese Patients. <i>Frontiers in Genetics</i> , 2021, 12, 732170.	1.1	2
5279	Computational Analysis of non-synonymous Single Nucleotide Polymorphism in UROD Gene linked with Familial Porphyria Cutanea Tarda. <i>Gene Reports</i> , 2021, 25, 101413.	0.4	0
5281	Synonymous and non-synonymous polymorphisms in toll-like receptor 2 (TLR2) gene among complicated measles cases at a tertiary care hospital, Peshawar, Pakistan. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2021, 42, 1229-1236.	0.5	1
5282	Mutation spectrum and genotype-phenotype correlations in 157 Korean CADASIL patients: a multicenter study. <i>Neurogenetics</i> , 2022, 23, 45-58.	0.7	12
5283	A Missense Variant in the Bardet-Biedl Syndrome 2 Gene (BBS2) Leads to a Novel Syndromic Retinal Degeneration in the Shetland Sheepdog. <i>Genes</i> , 2021, 12, 1771.	1.0	4
5285	Impact of Deleterious Mutations on Structure, Function and Stability of Serum/Glucocorticoid Regulated Kinase 1: A Gene to Diseases Correlation. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 780284.	1.6	12
5289	Improving Prediction Accuracy via Subspace Modeling. , 0, , 33-48.		0
5290	Improving Prediction Accuracy via Subspace Modeling in a Statistical Geometry Based Computational Protein Mutagenesis. , 0, , 1010-1024.		0
5294	A weighted empirical Bayes risk prediction model using multiple traits. <i>Statistical Applications in Genetics and Molecular Biology</i> , 2020, 19, .	0.2	0
5299	The design of image processing software based on UAV remote sensing platform. , 2020, , .		0
5300	Introducing, OncoTarget. <i>Oncotarget</i> , 2010, 1, 2-2.	0.8	0

#	ARTICLE	IF	CITATIONS
5301	Introducing, OncoTarget. Oncotarget, 2010, 1, 2-2.	0.8	0
5302	Introducing, OncoTarget. Oncotarget, 2010, 1, 2-2.	0.8	0
5303	Introducing, OncoTarget. Oncotarget, 2010, 1, 2-2.	0.8	0
5304	Introducing, OncoTarget. Oncotarget, 2010, 1, 2-2.	0.8	0
5305	Introducing, OncoTarget. Oncotarget, 2010, 1, 2-2.	0.8	0
5306	Introducing, OncoTarget. Oncotarget, 2010, 1, 2-2.	0.8	0
5309	Mutations Within the Transcription Factor <i>PROP1</i> in a Cohort of Turkish Patients with Combined Pituitary Hormone Deficiency. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2020, 12, 261-268.	0.4	4
5310	Identification of four novel cytochrome P4501B1 mutations (p.I94X, p.H279D, p.Q340H, and p.K433K) in primary congenital glaucoma patients. Molecular Vision, 2009, 15, 2926-37.	1.1	18
5311	Mitochondrial DNA analysis in primary congenital glaucoma. Molecular Vision, 2010, 16, 518-33.	1.1	35
5312	Screening of CYP1B1 and MYOC in Moroccan families with primary congenital glaucoma: three novel mutations in CYP1B1. Molecular Vision, 2010, 16, 1215-26.	1.1	23
5313	MYOC and FOXC1 gene analysis in primary congenital glaucoma. Molecular Vision, 2010, 16, 1996-2006.	1.1	15
5314	VSX1 gene analysis in keratoconus. Molecular Vision, 2010, 16, 2395-401.	1.1	47
5315	Assessment of canine BEST1 variations identifies new mutations and establishes an independent bestrophinopathy model (cmr3). Molecular Vision, 2010, 16, 2791-804.	1.1	39
5316	Mitochondrial complex 1 gene analysis in keratoconus. Molecular Vision, 2011, 17, 1514-25.	1.1	33
5317	Mutations in a novel serine protease PRSS56 in families with nanophthalmos. Molecular Vision, 2011, 17, 1850-61.	1.1	43
5318	A recurrent missense mutation in GJA3 associated with autosomal dominant cataract linked to chromosome 13q. Molecular Vision, 2011, 17, 2255-62.	1.1	13
5319	Methods, challenges, and promise of next-generation sequencing in cancer biology. Yale Journal of Biology and Medicine, 2011, 84, 439-46.	0.2	13
5320	BEST1 sequence variants in Italian patients with vitelliform macular dystrophy. Molecular Vision, 2012, 18, 2736-48.	1.1	21

#	ARTICLE	IF	CITATIONS
5321	Mitochondrial DNA nucleotide changes in primary congenital glaucoma patients. <i>Molecular Vision</i> , 2013, 19, 220-30.	1.1	16
5322	Genetic variants of FZD4 and LRP5 genes in patients with advanced retinopathy of prematurity. <i>Molecular Vision</i> , 2013, 19, 476-85.	1.1	44
5323	Identification of a novel p.R1443W mutation in RP1 gene associated with retinitis pigmentosa sine pigmento. <i>International Journal of Ophthalmology</i> , 2013, 6, 430-5.	0.5	7
5324	Genetic screen of African Americans with Fuchs endothelial corneal dystrophy. <i>Molecular Vision</i> , 2013, 19, 2508-16.	1.1	13
5325	AIP1L1 implicated in the pathogenesis of two cases of autosomal recessive retinal degeneration. <i>Molecular Vision</i> , 2014, 20, 1-14.	1.1	6
5326	Integrative analysis of two cell lines derived from a non-small-lung cancer patient—a panomics approach. <i>Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing</i> , 2014, , 75-86.	0.7	4
5327	Molecular profiling of complete congenital stationary night blindness: a pilot study on an Indian cohort. <i>Molecular Vision</i> , 2014, 20, 341-51.	1.1	14
5328	PATH-SCAN: a reporting tool for identifying clinically actionable variants. <i>Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing</i> , 2014, , 229-40.	0.7	10
5329	Mutation analysis of pre-mRNA splicing genes in Chinese families with retinitis pigmentosa. <i>Molecular Vision</i> , 2014, 20, 770-9.	1.1	17
5330	Analysis of rare variants in the CFH gene in patients with the cuticular drusen subtype of age-related macular degeneration. <i>Molecular Vision</i> , 2015, 21, 285-92.	1.1	19
5331	In silico Evaluation of Nonsynonymous Single Nucleotide Polymorphisms in the ADIPOQ Gene Associated with Diabetes, Obesity, and Inflammation. <i>Avicenna Journal of Medical Biotechnology</i> , 2015, 7, 121-7.	0.2	5
5332	Exome sequencing on malignant meningiomas identified mutations in neurofibromatosis type 2 (NF2) and meningioma 1 (MN1) genes. <i>Discovery Medicine</i> , 2014, 18, 301-311.	0.5	20
5333	SPO11-C631T Gene Polymorphism: Association With Male Infertility and an in Silico-Analysis. <i>Journal of Family & Reproductive Health</i> , 2015, 9, 155-63.	0.4	12
5334	Mutation spectrum of the Norrie disease pseudoglioma (NDP) gene in Indian patients with FEVR. <i>Molecular Vision</i> , 2016, 22, 491-502.	1.1	14
5335	Targeted next-generation sequencing extends the phenotypic and mutational spectrums for EYS mutations. <i>Molecular Vision</i> , 2016, 22, 646-57.	1.1	14
5336	A Bioinformatics Approach to Prioritize Single Nucleotide Polymorphisms in TLRs Signaling Pathway Genes. <i>International Journal of Molecular and Cellular Medicine</i> , 2016, 5, 65-79.	1.1	5
5338	X-linked heterozygous mutations in cause female-limited early onset high myopia. <i>Molecular Vision</i> , 2016, 22, 1257-1266.	1.1	29
5339	Mutation near the binding interfaces at Î±-hemoglobin stabilizing protein is highly pathogenic. <i>American Journal of Translational Research (discontinued)</i> , 2016, 8, 4224-4232.	0.0	12

#	ARTICLE	IF	CITATIONS
5340	Diagnostic genetic testing for patients with bilateral optic neuropathy and comparison of clinical features according to mutation status. <i>Molecular Vision</i> , 2017, 23, 548-560.	1.1	11
5341	Computational Analysis of High Risk Missense Variant in Human UTY Gene: A Candidate Gene of AZFa Sub-region. <i>Journal of Reproduction and Infertility</i> , 2017, 18, 298-306.	1.0	6
5342	Identification of Novel PTPRQ and MYO1A Mutations in An Iranian Pedigree with Autosomal Recessive Hearing Loss. <i>Cell Journal</i> , 2018, 20, 127-131.	0.2	5
5343	Myopia in Chinese families shows linkage to 10q26.13. <i>Molecular Vision</i> , 2018, 24, 29-42.	1.1	3
5344	Optic nerve regeneration in the mouse is a complex trait modulated by genetic background. <i>Molecular Vision</i> , 2018, 24, 174-186.	1.1	12
5345	Ofatumumab Monoclonal Antibody Affinity Maturation Through in silico Modeling. <i>Iranian Biomedical Journal</i> , 2018, 22, 180-92.	0.4	13
5346	Variants in the gene in a Brazilian population with Stargardt disease. <i>Molecular Vision</i> , 2018, 24, 546-559.	1.1	12
5347	A study of the pathogenicity of variants in familial heart disease. The value of cosegregation. <i>American Journal of Translational Research (discontinued)</i> , 2019, 11, 1724-1735.	0.0	2
5348	Identification of a Novel KCNQ1 Frameshift Mutation and Review of the Literature among Iranian Long QT Families. <i>Iranian Biomedical Journal</i> , 2019, 23, 228-34.	0.4	0
5349	Identification of novel mutations in patients with fibrinogen disorders and genotype/phenotype correlations. <i>Blood Transfusion</i> , 2019, 17, 247-254.	0.3	3
5350	Targeted next-generation sequencing extends the mutational spectrums for mutations in Chinese families with optic atrophy. <i>Molecular Vision</i> , 2019, 25, 912-920.	1.1	3
5351	Comprehensive Computational Analysis of Protein Phenotype Changes Due to Plausible Deleterious Variants of Human SPTLC1 Gene. <i>International Journal of Molecular and Cellular Medicine</i> , 2019, 8, 67-84.	1.1	0
5352	Analysis of variants in Chinese individuals with primary open-angle glaucoma using molecular inversion probe (MIP)-based panel sequencing. <i>Molecular Vision</i> , 2020, 26, 378-391.	1.1	3
5353	Biallelic -associated retinal dystrophies: Expanding the mutational and clinical spectrum. <i>Molecular Vision</i> , 2020, 26, 423-433.	1.1	4
5355	L51P, a novel mutation in the PAS domain of hERG channel, confers long QT syndrome by impairing channel activation. <i>American Journal of Translational Research (discontinued)</i> , 2020, 12, 8040-8049.	0.0	0
5356	Genotype phenotype analysis in a family carrying truncating mutations in the titin gene. <i>Acta Myologica</i> , 2021, 40, 61-65.	1.5	0
5357	Analysis of glucocerebrosidase (GBA) gene mutations in Iranian patients with Gaucher disease. <i>Iranian Journal of Child Neurology</i> , 2021, 15, 139-166.	0.2	0
5358	Identification of a Novel Stop Loss Mutation in P2RX2 Gene in an Iranian Family with Autosomal Nonsyndromic Hearing Loss. <i>Iranian Biomedical Journal</i> , 2021, 25, 368-73.	0.4	0

#	ARTICLE	IF	CITATIONS
5359	Analysis of mutations in the Chinese Uyghur population. American Journal of Translational Research (discontinued), 2021, 13, 10871-10881.	0.0	0
5360	Homozygous R136S mutation in PRNP gene causes inherited early onset prion disease. Alzheimer's Research and Therapy, 2021, 13, 176.	3.0	1
5361	Pattern Recognition Molecules of Lectin Complement Pathway in Ischemic Stroke. Pharmacogenomics and Personalized Medicine, 2021, 14, 1347-1368.	0.4	1
5362	Common Genomic Aberrations in Mouse and Human Breast Cancers with Concurrent P53 Deficiency and Activated PTEN-PI3K-AKT Pathway. International Journal of Biological Sciences, 2022, 18, 229-241.	2.6	2
5363	Pattern Recognition Molecules of Lectin Complement Pathway in Ischemic Stroke. Pharmacogenomics and Personalized Medicine, 2021, Volume 14, 1347-1368.	0.4	1
5364	PIK3CA hotspot mutations p. H1047R and p. H1047L sensitize breast cancer cells to thymoquinone treatment by regulating the PI3K/Akt1 pathway. Molecular Biology Reports, 2021, , 1.	1.0	14
5365	Bi-allelic variants in human WDR63 cause male infertility via abnormal inner dynein arms assembly. Cell Discovery, 2021, 7, 110.	3.1	19
5368	Two novel CD40LG gene mutations causing X-linked hyper IgM syndrome in Vietnamese patients. Clinical and Experimental Medicine, 2021, , 1.	1.9	1
5369	Digenic heterozygous mutations of KCNH2 and SCN5A induced young and early-onset long QT syndrome and sinoatrial node dysfunction. Annals of Noninvasive Electrocardiology, 2022, 27, e12889.	0.5	6
5370	Diagnostic Yield of Targeted Hearing Loss Gene Panel Sequencing in a Large German Cohort With a Balanced Age Distribution from a Single Diagnostic Center: An Eight-year Study. Ear and Hearing, 2022, 43, 1049-1066.	1.0	13
5371	Identification of Pathogenic CNVs in Unexplained Developmental Disabilities Using Exome Sequencing: A Family Trio Study. Russian Journal of Genetics, 2021, 57, 1351-1355.	0.2	2
5372	Genetic Cancer Susceptibility in Adolescents and Adults 25 Years or Younger With Colorectal Cancer. Gastroenterology, 2022, 162, 969-974.e6.	0.6	2
5373	Genetic Analysis of Acid β -Glucosidase in Patients with Multiple Myeloma from Central Taiwan: A Small-Cohort Case-Control Study. Biomedicine Hub, 2021, 6, 138-144.	0.4	0
5374	Identification and in silico Characterization of Deleterious Single Nucleotide Variations in Human ZP2 Gene. Frontiers in Cell and Developmental Biology, 2021, 9, 763166.	1.8	2
5375	Clinical features and genetic spectrum of NMNAT1-associated retinal degeneration. Eye, 2022, 36, 2279-2285.	1.1	3
5376	Molecular analysis of ABCA4 gene in an Iranian cohort with Stargardt disease. Gene Reports, 2021, , 101450.	0.4	0
5377	Novel Likely Pathogenic Variants Identified by Panel-Based Exome Sequencing in Congenital Cataract Patients. Journal of Ophthalmology, 2021, 2021, 1-10.	0.6	0
5379	A chickpea genetic variation map based on the sequencing of 3,366 genomes. Nature, 2021, 599, 622-627.	13.7	106

#	ARTICLE	IF	CITATIONS
5380	Comprehensive Study of Human FBXW7 Deleterious nsSNPs Functional Inference and Susceptibility to Gynaecological Cancer. Applied Biochemistry and Biotechnology, 2022, 194, 407-433.	1.4	2
5381	Benign and Intermediate-grade Melanocytic Tumors With BRAF Mutations and Spitzoid Morphology. American Journal of Surgical Pathology, 2022, 46, 476-485.	2.1	11
5382	Identification of discriminative gene-level and protein-level features associated with pathogenic gain-of-function and loss-of-function variants. American Journal of Human Genetics, 2021, 108, 2301-2318.	2.6	21
5383	Glycoprotein 2 in health and disease: lifting the veil. Genes and Environment, 2021, 43, 53.	0.9	3
5384	NOTCH3 mutations in a cohort of Portuguese patients within CADASIL spectrum phenotype. Neurogenetics, 2022, 23, 1-9.	0.7	6
5386	Assessment of POLE and POLD1 mutations as prognosis and immunotherapy biomarkers for stomach adenocarcinoma. Translational Cancer Research, 2022, 11, 193-205.	0.4	10
5387	Genotype-phenotype correlation of Parkinson's disease with PRKN variants. Neurobiology of Aging, 2022, 114, 117-128.	1.5	13
5388	Evolution of fluoroquinolone-resistant Escherichia coli in the gut after ciprofloxacin treatment. International Journal of Medical Microbiology, 2022, 312, 151548.	1.5	1
5389	Homozygous R136S mutation in PRNP gene causes inherited early onset prion disease. Alzheimer's Research and Therapy, 2021, 13, 176.	3.0	8
5390	Molecular analysis and genotype-phenotype correlations in patients with classical congenital adrenal hyperplasia due to 21-hydroxylase deficiency from southern Poland – experience of a clinical center. Hormones, 2022, , 1.	0.9	2
5391	In Silico Analysis of Collagens Missense SNPs and Human Abnormalities. Biochemical Genetics, 2022, 60, 1630-1656.	0.8	1
5392	Rare variants in TP73 in a frontotemporal dementia cohort link this gene with primary progressive aphasia phenotypes. European Journal of Neurology, 2022, , .	1.7	1
5393	Cohort-based association study of germline genetic variants with acute and chronic health complications of childhood cancer and its treatment: Genetic Risks for Childhood Cancer Complications Switzerland (GECCOS) study protocol. BMJ Open, 2022, 12, e052131.	0.8	1
5394	New mutation of the TP53 gene associated with the hereditary breast cancer in a young Tuvian woman. Siberian Journal of Oncology, 2022, 20, 164-170.	0.1	0
5395	Analysis of coding variants in the human FTO gene from the gnomAD database. PLoS ONE, 2022, 17, e0248610.	1.1	1
5396	Weighted burden analysis in 200,000 exome-sequenced subjects characterises rare variant effects on BMI. International Journal of Obesity, 2022, , .	1.6	3
5397	Novel RCBTB1 variants causing later-onset non-syndromic retinal dystrophy with macular chorioretinal atrophy. Ophthalmic Genetics, 2022, , 1-8.	0.5	2
5398	Whole exome sequencing identifies potential candidate genes for spina bifida derived from mouse models. American Journal of Medical Genetics, Part A, 2022, , .	0.7	2

#	ARTICLE	IF	CITATIONS
5399	Identification of the most damaging nsSNPs in the human CFL1 gene and their functional and structural impacts on cofilin-1 protein. <i>Gene</i> , 2022, 819, 146206.	1.0	9
5400	Computational design of a cutinase for plastic biodegradation by mining molecular dynamics simulations trajectories. <i>Computational and Structural Biotechnology Journal</i> , 2022, 20, 459-470.	1.9	27
5401	Preneoplastic somatic mutations including <i>MYD88</i> ^{L265P} in lymphoplasmacytic lymphoma. <i>Science Advances</i> , 2022, 8, eabl4644.	4.7	21
5402	Predicting and interpreting large-scale mutagenesis data using analyses of protein stability and conservation. <i>Cell Reports</i> , 2022, 38, 110207.	2.9	62
5403	Targeted next-generation sequencing in a large series of fetuses with severe renal diseases. <i>Human Mutation</i> , 2022, 43, 347-361.	1.1	12
5404	A comparison on predicting functional impact of genomic variants. <i>NAR Genomics and Bioinformatics</i> , 2022, 4, lqab122.	1.5	12
5405	In-silico analysis unravels the structural and functional consequences of non-synonymous SNPs in the human IL-10 gene. <i>Egyptian Journal of Medical Human Genetics</i> , 2022, 23, .	0.5	2
5406	Evaluation of phenotype-driven gene prioritization methods for Mendelian diseases. <i>Briefings in Bioinformatics</i> , 2022, 23, .	3.2	17
5407	Molecular characteristics of young-onset colorectal cancer in Vietnamese patients. <i>Asia-Pacific Journal of Clinical Oncology</i> , 2022, , .	0.7	3
5409	Gene Network of Susceptibility to Atypical Femoral Fractures Related to Bisphosphonate Treatment. <i>Genes</i> , 2022, 13, 146.	1.0	3
5411	Variant interpretation in molecular autopsy: a useful dilemma. <i>International Journal of Legal Medicine</i> , 2022, 136, 475-482.	1.2	9
5412	SorghumBase: a web-based portal for sorghum genetic information and community advancement. <i>Planta</i> , 2022, 255, 35.	1.6	12
5414	Novel Mutations of TSPY1 Gene Associate Spermatogenic Failure Among Men. <i>Reproductive Sciences</i> , 2022, 29, 1241-1261.	1.1	3
5415	Amelioration of a neurodevelopmental disorder by carbamazepine in a case having a gain-of-function <i>GRIA3</i> variant. <i>Human Genetics</i> , 2022, 141, 283-293.	1.8	6
5416	Hybridization underlies localized trait evolution in cavefish. <i>IScience</i> , 2022, 25, 103778.	1.9	17
5418	Identification of the somatic mutations of <i>SMAD4</i> among HCC Egyptian patients using NGS. <i>Gene Reports</i> , 2022, 26, 101478.	0.4	0
5419	In-silico analysis of nonsynonymous genomic variants within <i>CCM2</i> gene reaffirm the existence of dual cores within typical PTB domain. <i>Biochemistry and Biophysics Reports</i> , 2022, 29, 101218.	0.7	1
5420	Polymorphisms of the filaggrin gene are associated with atopic dermatitis in the Caucasian population of Central Russia. <i>Gene</i> , 2022, 818, 146219.	1.0	0

#	ARTICLE	IF	CITATIONS
5421	Investigation of Putative Functional SNPs of Human HAT1 Protein: A Comprehensive <i>in silico</i> Study. <i>Cytology and Genetics</i> , 2022, 56, 98-107.	0.2	0
5422	Genome Nexus: A Comprehensive Resource for the Annotation and Interpretation of Genomic Variants in Cancer. <i>JCO Clinical Cancer Informatics</i> , 2022, 6, e2100144.	1.0	4
5423	Contribution of Common and Rare Genetic Variants in CEP72 on Vincristine-Induced Peripheral Neuropathy in Brain Tumor Patients. <i>British Journal of Clinical Pharmacology</i> , 2022, , .	1.1	2
5424	Genetic load: genomic estimates and applications in non-model animals. <i>Nature Reviews Genetics</i> , 2022, 23, 492-503.	7.7	82
5425	Somatic mutation analyses of stem-like cells in gingivobuccal oral squamous cell carcinoma reveals DNA damage response genes. <i>Genomics</i> , 2022, 114, 110308.	1.3	1
5426	Further delineation of familial polycystic ovary syndrome (PCOS) via whole-exome sequencing: PCOS-related rare <i>FBN3</i> and <i>FN1</i> gene variants are identified. <i>Journal of Obstetrics and Gynaecology Research</i> , 2022, 48, 1202-1211.	0.6	9
5427	Conservation genetics as a management tool: The five best-supported paradigms to assist the management of threatened species. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	92
5428	Good Tumor Response to Chemoradioimmunotherapy in dMMR/MSI-H Advanced Colorectal Cancer: A Case Series. <i>Frontiers in Immunology</i> , 2021, 12, 784336.	2.2	12
5430	Whole-exome sequencing, amplification and infiltration patterns in human glioblastoma. <i>American Journal of Cancer Research</i> , 2021, 11, 5543-5558.	1.4	0
5432	Scaling up oligogenic diseases research with OLIDA: the Oligogenic Diseases Database. <i>Database: the Journal of Biological Databases and Curation</i> , 2022, 2022, .	1.4	10
5433	Whole-Exome Sequencing Revealed a Pathogenic Nonsense Variant in the <i>SLC19A2</i> Gene in an Iranian Family with Thiamine-Responsive Megaloblastic Anemia. <i>Laboratory Medicine</i> , 2022, 53, 640-650.	0.8	0
5434	Identification and <i>in silico</i> Analysis of Nonsense SNPs of Human Colorectal Cancer Protein. <i>Journal of Oleo Science</i> , 2022, 71, 363-370.	0.6	2
5435	Novel <i>ABCB4</i> mutations in an infertile female with progressive familial intrahepatic cholestasis type 3: A case report. <i>World Journal of Clinical Cases</i> , 2022, 10, 1998-2006.	0.3	2
5437	Domestication reprogrammed the budding yeast life cycle. <i>Nature Ecology and Evolution</i> , 2022, 6, 448-460.	3.4	32
5438	DSDatlas: disorders of sex development atlas for reproductive endocrinological-related gene discovery in integrative omics platforms. <i>F&S Science</i> , 2022, 3, 108-117.	0.5	1
5439	mTOR pathway repressing expression of FoxO3 is a potential mechanism involved in neonatal white matter dysplasia. <i>Human Molecular Genetics</i> , 2022, 31, 2508-2520.	1.4	2
5440	DVPred: a disease-specific prediction tool for variant pathogenicity classification for hearing loss. <i>Human Genetics</i> , 2022, 141, 401-411.	1.8	6
5441	Clonal Evolution in Patients with Hormone Receptor Positive, HER-2 Negative Breast Cancer Treated with Chemotherapy or CDK4/6 Inhibitors. <i>Oncology Research and Treatment</i> , 2022, 45, 248-253.	0.8	3

#	ARTICLE	IF	CITATIONS
5442	Clinical and genetic characterization of a large cohort of patients with Wilson's disease in China. <i>Translational Neurodegeneration</i> , 2022, 11, 13.	3.6	15
5443	Two rare variants reveal the significance of Grainyhead-like 3 Arginine 391 underlying non-syndromic cleft palate only. <i>Oral Diseases</i> , 2023, 29, 1632-1643.	1.5	4
5444	Identification of ultra-rare disruptive variants in voltage-gated calcium channel-encoding genes in Japanese samples of schizophrenia and autism spectrum disorder. <i>Translational Psychiatry</i> , 2022, 12, 84.	2.4	4
5445	Clonal haematopoiesis of indeterminate potential and cardiovascular events in systemic lupus erythematosus (HEMATOPLUS study). <i>Rheumatology</i> , 2022, 61, 4355-4363.	0.9	14
5447	Landscape of TP53 Alterations in Chronic Lymphocytic Leukemia via Data Mining Mutation Databases. <i>Frontiers in Oncology</i> , 2022, 12, 808886.	1.3	5
5448	Homozygous variants in <i>AKAP3</i> induce asthenoteratozoospermia and male infertility. <i>Journal of Medical Genetics</i> , 2023, 60, 137-143.	1.5	9
5449	Harmonizing model organism data in the Alliance of Genome Resources. <i>Genetics</i> , 2022, 220, .	1.2	52
5450	The S100A7 nuclear interactors in autoimmune diseases: a coevolutionary study in mammals. <i>Immunogenetics</i> , 2022, , 1.	1.2	0
5451	Survival analysis of clinical and genetic factors in an amyotrophic lateral sclerosis cohort from China. <i>Neurological Research</i> , 2022, 44, 651-658.	0.6	1
5452	One Health and Cattle Genetic Resources: Mining More than 500 Cattle Genomes to Identify Variants in Candidate Genes Potentially Affecting Coronavirus Infections. <i>Animals</i> , 2022, 12, 838.	1.0	1
5453	Computational Analysis of the Potential Impact of MTC Complex Missenses SNPs Associated with Male Infertility. <i>BioMed Research International</i> , 2022, 2022, 1-18.	0.9	3
5454	Machine-learning of complex evolutionary signals improves classification of SNVs. <i>NAR Genomics and Bioinformatics</i> , 2022, 4, lqac025.	1.5	4
5455	SOD-1 Variants in Amyotrophic Lateral Sclerosis: Systematic Re-Evaluation According to ACMG-AMP Guidelines. <i>Genes</i> , 2022, 13, 537.	1.0	7
5456	Design of a targeted next-generation DNA sequencing panel for pediatric T-cell lymphoblastic lymphoma to unravel biology and optimize treatment. <i>Genes Chromosomes and Cancer</i> , 2022, 61, 459-470.	1.5	2
5457	Gene Variants Involved in Nonsense-Mediated mRNA Decay Suggest a Role in Autism Spectrum Disorder. <i>Biomedicines</i> , 2022, 10, 665.	1.4	6
5458	AmazonForest: In Silico Metaprediction of Pathogenic Variants. <i>Biology</i> , 2022, 11, 538.	1.3	0
5459	Image encryption algorithm based on lattice hash function and privacy protection. <i>Multimedia Tools and Applications</i> , 2022, 81, 18251-18277.	2.6	6
5460	Comprehensive analysis of recessive carrier status using exome and genome sequencing data in 1543 Southern Chinese. <i>Npj Genomic Medicine</i> , 2022, 7, 23.	1.7	6

#	ARTICLE	IF	CITATIONS
5461	A <i>CTNNB1</i> altered medulloblastoma shows the immunophenotypic, DNA methylation and transcriptomic profiles of SHH-activated, and not WNT-activated, medulloblastoma. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, e12815.	1.8	0
5462	Clinical Characteristics and Pathogenic Gene Identification in Chinese Patients With Paget's Disease of Bone. <i>Frontiers in Endocrinology</i> , 2022, 13, 850462.	1.5	6
5464	Functionally significant polymorphisms of the <i>MMP9</i> gene are associated with primary open-angle glaucoma in the population of Russia. <i>European Journal of Ophthalmology</i> , 2022, 32, 3208-3219.	0.7	2
5465	Targeted Exome Sequencing of Genes Involved in Rare CNVs in Early-Onset Severe Obesity. <i>Frontiers in Genetics</i> , 2022, 13, 839349.	1.1	3
5466	Identification of Novel and Known LDLR Variants Triggering Severe Familial Hypercholesterolemia in Saudi Families. <i>Current Vascular Pharmacology</i> , 2022, 20, 361-369.	0.8	1
5467	Implementation of preemptive DNA sequence-based pharmacogenomics testing across a large academic medical center: The Mayo-Baylor RIGHT 10K Study. <i>Genetics in Medicine</i> , 2022, 24, 1062-1072.	1.1	28
5469	Update on CD164 and LMX1A genes to strengthen their causative role in autosomal dominant hearing loss. <i>Human Genetics</i> , 2022, 141, 445-453.	1.8	6
5470	CRISPR Detection and Research on Screening Mutant Gene TTN of Moyamoya Disease Family Based on Whole Exome Sequencing. <i>Frontiers in Molecular Biosciences</i> , 2022, 9, 846579.	1.6	2
5471	Identification of ENPP1 Haploinsufficiency in Patients With Diffuse Idiopathic Skeletal Hyperostosis and Early-Onset Osteoporosis. <i>Journal of Bone and Mineral Research</i> , 2020, 37, 1125-1135.	3.1	18
5472	Aberrant hippocampal transmission and behavior in mice with a stargazin mutation linked to intellectual disability. <i>Molecular Psychiatry</i> , 2022, 27, 2457-2469.	4.1	3
5473	Identification of Novel Mutations in Chinese Infants With Citrullinemia. <i>Frontiers in Genetics</i> , 2022, 13, 783799.	1.1	2
5474	Comprehensive variant spectrum of the <i>CNGA3</i> gene in patients affected by achromatopsia. <i>Human Mutation</i> , 2022, 43, 832-858.	1.1	8
5475	Investigating underlying human immunity genes, implicated diseases and their relationship to COVID-19. <i>Personalized Medicine</i> , 2022, , .	0.8	2
5476	Novel dominant and recessive variants in human <i>ROBO1</i> cause distinct neurodevelopmental defects through different mechanisms. <i>Human Molecular Genetics</i> , 2022, 31, 2751-2765.	1.4	3
5477	In-silico analysis of non-synonymous single nucleotide polymorphisms in human Î²-defensin type 1 gene reveals their impact on protein-ligand binding sites. <i>Computational Biology and Chemistry</i> , 2022, 98, 107669.	1.1	4
5478	Comparison of the clinical and genetic features of autosomal dominant optic atrophy and normal tension glaucoma in young Chinese adults. <i>Eye</i> , 2022, , .	1.1	0
5479	Severe combined immunodeficiencies: Expanding the mutation spectrum in Turkey and identification of 12 novel variants. <i>Scandinavian Journal of Immunology</i> , 2022, 95, e13163.	1.3	1
5480	Genotype-phenotype association of TARDBP mutations in Chinese patients with amyotrophic lateral sclerosis: a single-center study and systematic review of published literature. <i>Journal of Neurology</i> , 2022, 269, 4204-4212.	1.8	4

#	ARTICLE	IF	CITATIONS
5481	Germline mosaicism of a missense variant in <i>KCNC2</i> in a multiplex family with autism and epilepsy characterized by long-read sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2071-2081.	0.7	7
5482	Circuit topology predicts pathogenicity of missense mutations. <i>Proteins: Structure, Function and Bioinformatics</i> , 2022, 90, 1634-1644.	1.5	7
5483	Ciliary and immune dysfunctions and their genetic background in patients with non-cystic fibrosis bronchiectasis in Central Iran. <i>Irish Journal of Medical Science</i> , 2023, 192, 277-283.	0.8	1
5484	Re-evaluation of missense variant classifications in <i>NF2</i> . <i>Human Mutation</i> , 2022, 43, 643-654.	1.1	5
5485	Mutation screening of the <i>DNAJC7</i> gene in Japanese patients with sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2022, 113, 131-136.	1.5	6
5486	Identification of Rare Loss-of-Function Genetic Variation Regulating Body Fat Distribution. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 1065-1077.	1.8	12
5487	Molecular-based precision oncology clinical decision making augmented by artificial intelligence. <i>Emerging Topics in Life Sciences</i> , 2021, 5, 757-764.	1.1	4
5488	In-silico analysis reveals druggable single nucleotide polymorphisms in angiotensin 1 converting enzyme involved in the onset of blood pressure. <i>BMC Research Notes</i> , 2021, 14, 457.	0.6	2
5489	Worldwide diversity, association potential, and natural selection in the superimposed taste genes, <i>CD36</i> and <i>GNAT3</i> . <i>Chemical Senses</i> , 2022, 47, .	1.1	1
5490	Successful outcomes of intracytoplasmic sperm injection-embryo transfer using ejaculated spermatozoa from two Chinese asthenoteratozoospermic brothers with a compound heterozygous <i>FSIP2</i> mutation. <i>Andrologia</i> , 2022, 54, e14351.	1.0	7
5492	Whole exome sequencing identifies deleterious rare variants in <i>CCDC141</i> in familial self-limited delayed puberty. <i>Npj Genomic Medicine</i> , 2021, 6, 107.	1.7	4
5493	Design and performance of a bovine 200k SNP chip developed for endangered German Black Pied cattle (DSN). <i>BMC Genomics</i> , 2021, 22, 905.	1.2	9
5494	Discovery of a genetic module essential for assigning left-right asymmetry in humans and ancestral vertebrates. <i>Nature Genetics</i> , 2022, 54, 62-72.	9.4	16
5496	Functional and structural analysis of non-synonymous single nucleotide polymorphisms (nsSNPs) in the MYB oncoproteins associated with human cancer. <i>Scientific Reports</i> , 2021, 11, 24206.	1.6	7
5497	Analysis of <i>CYP27A1</i> mutations in Han Chinese women with intrahepatic cholestasis of pregnancy. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2021, , 1-9.	0.7	0
5498	The Novel Phosphatase Domain Mutations Q171R and Y65S Switch PTEN from Tumor Suppressor to Oncogene. <i>Cells</i> , 2021, 10, 3423.	1.8	1
5499	Prediction of disease-associated nsSNPs by integrating multi-scale ResNet models with deep feature fusion. <i>Briefings in Bioinformatics</i> , 2022, 23, .	3.2	13
5500	A Rare Mutation in <i>LMNB2</i> Associated with Lipodystrophy Drives Premature Cell Senescence. <i>Cells</i> , 2022, 11, 50.	1.8	3

#	ARTICLE	IF	CITATIONS
5501	Filaggrin gene polymorphisms are associated with atopic dermatitis in women but not in men in the Caucasian population of Central Russia. <i>PLoS ONE</i> , 2021, 16, e0261026.	1.1	9
5502	LYRUS: a machine learning model for predicting the pathogenicity of missense variants. <i>Bioinformatics Advances</i> , 2022, 2, vbab045.	0.9	4
5503	Annotating and prioritizing genomic variants using the Ensembl Variant Effect Predictor—A tutorial. <i>Human Mutation</i> , 2022, 43, 986-997.	1.1	30
5504	Ribosomal protein S6 kinase beta-1 gene variants cause hypertrophic cardiomyopathy. <i>Journal of Medical Genetics</i> , 2021, , jmedgenet-2021-107866.	1.5	1
5506	Identification of deleterious single nucleotide polymorphism (SNP)s in the human TBX5 gene & prediction of their structural & functional consequences: An in silico approach. <i>Biochemistry and Biophysics Reports</i> , 2021, 28, 101179.	0.7	2
5508	<i>Ectodysplasin</i> pathogenic variants affecting the furinâ€cleavage site and unusual clinical features define Xâ€linked hypohidrotic ectodermal dysplasia in India. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 788-805.	0.7	3
5509	Identification of ACOT13 and PTGER2 as novel candidate genes of autosomal dominant polycystic kidney disease through whole exome sequencing. <i>European Journal of Medical Research</i> , 2021, 26, 142.	0.9	4
5510	VIPPID: a gene-specific single nucleotide variant pathogenicity prediction tool for primary immunodeficiency diseases. <i>Briefings in Bioinformatics</i> , 2022, 23, .	3.2	7
5511	Deciphering Genetic Susceptibility to Tuberculous Meningitis. <i>Frontiers in Neurology</i> , 2022, 13, 820168.	1.1	2
5513	Predominant Founder Effect among Recurrent Pathogenic Variants for an X-Linked Disorder. <i>Genes</i> , 2022, 13, 675.	1.0	2
5514	A rare genetic variant in the cleavage site of prepro-orexin is associated with idiopathic hypersomnia. <i>Npj Genomic Medicine</i> , 2022, 7, 29.	1.7	5
5515	Predicting the functional impact of KCNQ1 variants with artificial neural networks. <i>PLoS Computational Biology</i> , 2022, 18, e1010038.	1.5	5
5516	PSEN1 G417S mutation in a Chinese pedigree causing early-onset parkinsonism with cognitive impairment. <i>Neurobiology of Aging</i> , 2022, 115, 70-76.	1.5	0
5517	Discovering single-cell eQTLs from scRNA-seq data only. <i>Gene</i> , 2022, 829, 146520.	1.0	9
5632	Computational screening and analysis of deleterious nsSNPs in human <i>p</i>14ARF (<i>CDKN2A</i>) Tj ETQq0 0 0 rgBT /Overlock 10 Dynamics, 2023, 41, 3964-3975.	2.0	8
5636	PDigenic Mutations in Junctional Epidermolysis Bullosa in An Iranian Family. <i>Cell Journal</i> , 2021, 23, 598-602.	0.2	1
5639	Novel mutations responsible for Î±-thalassemia in Iranian families. <i>Hemoglobin</i> , 2013, 37, 148-59.	0.4	12
5644	ÂBile salt export pump deficiency disease: two novel, late onset, ABCB11 mutations identified by next generation sequencing. <i>Annals of Hepatology</i> , 2016, 15, 795-800.	0.6	9

#	ARTICLE	IF	CITATIONS
5645	Identification of a Novel Homozygous Mutation in Gene in an Iranian Family with Bardet-Biedl Syndrome.. Avicenna Journal of Medical Biotechnology, 2021, 13, 230-233.	0.2	0
5646	Genome interpretation using in silico predictors of variant impact. Human Genetics, 2022, 141, 1549-1577.	1.8	26
5647	Functional profiling of LDLR variants: Important evidence for variant classification. Journal of Clinical Lipidology, 2022, 16, 516-524.	0.6	8
5648	Whole-exome sequencing identifies rare genetic variants associated with human plasma metabolites. American Journal of Human Genetics, 2022, 109, 1038-1054.	2.6	17
5649	Scrutinizing Deleterious Nonsynonymous SNPs and Their Effect on Human POLD1 Gene. Genetical Research, 2022, 2022, 1-12.	0.3	2
5650	Revisiting benchmark study for response to methodological critiques of "Evaluation of phenotype-driven gene prioritization methods for Mendelian diseases"™. Briefings in Bioinformatics, 2022, 23, .	3.2	4
5651	Case Report: Family Curse: An SCN5A Mutation, c.611C>A, p.A204E Associated With a Family History of Dilated Cardiomyopathy and Arrhythmia. Frontiers in Cardiovascular Medicine, 2022, 9, .	1.1	0
5652	Proposer: The web server that readily proposes protein stabilizing mutations with high PPV. Computational and Structural Biotechnology Journal, 2022, 20, 2415-2433.	1.9	3
5653	Genomic and transcriptomic somatic alterations of hepatocellular carcinoma in non-cirrhotic livers. Cancer Genetics, 2022, 264-265, 90-99.	0.2	3
5654	Oxytocin and vasotocin receptor variation and the evolution of human prosociality. Comprehensive Psychoneuroendocrinology, 2022, 11, 100139.	0.7	6
5655	Novel Missense and Splice Site Mutations in USH2A, CDH23, PCDH15, and ADGRV1 Are Associated With Usher Syndrome in Lebanon. Frontiers in Genetics, 2022, 13, .	1.1	2
5656	Autosomal Dominant Retinitis Pigmentosa"Associated <i>TOPORS</i> Protein Truncating Variants Are Exclusively Located in the Region of Amino Acid Residues 807 to 867. , 2022, 63, 19.		1
5657	Fine Mapping of a Major Backfat QTL Reveals a Causal Regulatory Variant Affecting the CCND2 Gene. Frontiers in Genetics, 2022, 13, .	1.1	9
5658	Genomic analyses of 10,376 individuals in the Westlake BioBank for Chinese (WBBC) pilot project. Nature Communications, 2022, 13, .	5.8	41
5659	A Study of the Genomic Variations Associated with Autistic Spectrum Disorders in a Russian Cohort of Patients Using Whole-Exome Sequencing. Genes, 2022, 13, 920.	1.0	3
5660	Clinical description and genetic analysis of a novel familial skeletal dysplasia characterized by high bone mass and lucent bone lesions. Bone, 2022, 161, 116450.	1.4	2
5661	Health Risks of Hypermutation in Mice on the International Space Station. SSRN Electronic Journal, 0, , .	0.4	0
5662	Immune Alterations in a Patient With Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome: A Case Report. Frontiers in Immunology, 2022, 13, .	2.2	1

#	ARTICLE	IF	CITATIONS
5663	Different Phenotypes Represent Advancing Stages of <i>ABCA4</i> -Associated Retinopathy: A Longitudinal Study of 212 Chinese Families From a Tertiary Center. , 2022, 63, 28.		6
5664	Genetic Alterations, DNA Methylation, Alloantibodies and Phenotypic Heterogeneity in Type III von Willebrand Disease. <i>Genes</i> , 2022, 13, 971.	1.0	0
5665	Eph and Ephrin Variants in Malaysian Neural Tube Defect Families. <i>Genes</i> , 2022, 13, 952.	1.0	1
5666	Analysis of Mutational Profile of Hypopharyngeal and Laryngeal Head and Neck Squamous Cell Carcinomas Identifies KMT2C as a Potential Tumor Suppressor. <i>Frontiers in Oncology</i> , 0, 12, .	1.3	3
5667	Evidence of Selection in the Ectodysplasin Pathway among Endangered Aquatic Mammals. <i>Integrative Organismal Biology</i> , 2022, 4, .	0.9	2
5668	PHACT: Phylogeny-Aware Computing of Tolerance for Missense Mutations. <i>Molecular Biology and Evolution</i> , 2022, 39, .	3.5	3
5669	Nonsynonymous SNPs within <i>C7H15orf39</i> and <i>NOS2</i> are associated with boar semen quality. <i>Animal Biotechnology</i> , 0, , 1-5.	0.7	0
5670	Polymorphisms of the matrix metalloproteinase 9 gene are associated with duodenal ulcer in a Caucasian population of Central Russia. <i>Journal of King Saud University - Science</i> , 2022, 34, 102142.	1.6	3
5671	<i>ALK</i> and <i>CD36</i> as new causing genes for hereditary gingival fibromatosis New evidences of genetic heterogeneity of causing hereditary gingival fibromatosis and <i>ALK</i> and <i>CD36</i> as new candidate genes. <i>Journal of Periodontology</i> , 0, , .	1.7	5
5672	Quantitative assessment of copy number alterations by liquid biopsy for neuroblastoma. <i>Genes Chromosomes and Cancer</i> , 2022, 61, 662-669.	1.5	6
5673	E2UbcH5B-derived peptide ligands target HECT E3-E2 binding site and block the Ub-dependent SARS-CoV-2 egression: A computational study. <i>Computers in Biology and Medicine</i> , 2022, 146, 105660.	3.9	3
5676	A Stress Response that Allows Highly Mutated Eukaryotic Cells to Survive and Proliferate. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0
5677	Exploring the Structural and Functional Effects of Nonsynonymous SNPs in the Human Serotonin Transporter Gene Through <i>In Silico</i> Approaches. <i>Bioinformatics and Biology Insights</i> , 2022, 16, 117793222211043.	1.0	1
5678	Broadening the phenotypic spectrum of <i>TUBA1A</i> tubulinopathy to syndromic arthrogyrosis multiplex congenita. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	0
5679	Presence of rare potential pathogenic variants in subjects under 65 years old with very severe or fatal COVID-19. <i>Scientific Reports</i> , 2022, 12, .	1.6	6
5680	Protein structural bioinformatics: An overview. <i>Computers in Biology and Medicine</i> , 2022, 147, 105695.	3.9	15
5681	MYO1H is a novel candidate gene for autosomal dominant pure hereditary spastic paraplegia. <i>Molecular Genetics and Genomics</i> , 0, , .	1.0	0
5682	Dynamic insights into the effects of nonsynonymous polymorphisms (nsSNPs) on loss of TREM2 function. <i>Scientific Reports</i> , 2022, 12, .	1.6	5

#	ARTICLE	IF	CITATIONS
5683	Rare KCND3 Loss-of-Function Mutation Associated With the SCA19/22. <i>Frontiers in Molecular Neuroscience</i> , 0, 15, .	1.4	5
5684	Novel mutations in unrelated Vietnamese patients with chronic granulomatous disease. <i>Clinica Chimica Acta</i> , 2022, 533, 114-121.	0.5	1
5685	Molecular Analysis and Conformational Dynamics of Human MC4R Disease-Causing Mutations. <i>Molecules</i> , 2022, 27, 4037.	1.7	1
5686	Functional Analysis of Wild-Type and 27 CYP3A4 Variants on Dronedaron Metabolism in Vitro. <i>Current Drug Metabolism</i> , 2022, 23, .	0.7	0
5687	Impaired Neurodevelopmental Genes in Slovenian Autistic Children Elucidate the Comorbidity of Autism With Other Developmental Disorders. <i>Frontiers in Molecular Neuroscience</i> , 0, 15, .	1.4	1
5688	Secondary Genomic findings in the 2020 China Neonatal Genomes Project participants. <i>World Journal of Pediatrics</i> , 0, , .	0.8	5
5689	Genetic determinants of lung cancer: Understanding the oncogenic potential of somatic missense mutations. <i>Genomics</i> , 2022, , 110401.	1.3	6
5690	A novel missense variant in the CASK gene causes intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia. <i>BMC Medical Genomics</i> , 2022, 15, .	0.7	1
5691	Pathogenicity Reclassification of Genetic Variants Related to Early-Onset Breast Cancer among Women of Mongoloid Origin. <i>Asian Pacific Journal of Cancer Prevention</i> , 2022, 23, 2027-2033.	0.5	0
5692	Disease relevance of rare VPS13B missense variants for neurodevelopmental Cohen syndrome. <i>Scientific Reports</i> , 2022, 12, .	1.6	9
5694	Testing for Dihydropyrimidine Dehydrogenase Deficiency to Individualize 5-Fluorouracil Therapy. <i>Cancers</i> , 2022, 14, 3207.	1.7	14
5695	Burden tests can be used to map causal genes for a simple metabolic trait in an exome-sequenced polyploid mutant population. <i>Plant Biotechnology Journal</i> , 0, , .	4.1	0
5696	Exome sequencing analysis of Japanese autism spectrum disorder case-control sample supports an increased burden of synaptic function-related genes. <i>Translational Psychiatry</i> , 2022, 12, .	2.4	4
5697	In-Silico Analysis of Deleterious SNPs of FGF4 Gene and Their Impacts on Protein Structure, Function and Bladder Cancer Prognosis. <i>Life</i> , 2022, 12, 1018.	1.1	5
5699	Identification of 12 OCA Cases in Chinese Population and Two Novel Variants. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1
5700	Exome Sequencing of a Portuguese Cohort of Frontotemporal Dementia Patients: Looking Into the ALS-FTD Continuum. <i>Frontiers in Neurology</i> , 0, 13, .	1.1	2
5701	Uncovering the Contribution of Moderate-Penetrance Susceptibility Genes to Breast Cancer by Whole-Exome Sequencing and Targeted Enrichment Sequencing of Candidate Genes in Women of European Ancestry. <i>Cancers</i> , 2022, 14, 3363.	1.7	2
5702	Functional and clinical analysis of five EDA variants associated with ectodermal dysplasia but with a hard-to-predict significance. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	3

#	ARTICLE	IF	CITATIONS
5703	Comparative genomic analyses of multiple backcross mouse populations suggest <i>SGCG</i> as a novel potential obesity-modifier gene. <i>Human Molecular Genetics</i> , 0, , .	1.4	3
5704	How Functional Genomics Can Keep Pace With VUS Identification. <i>Frontiers in Cardiovascular Medicine</i> , 0, 9, .	1.1	8
5707	The Phenotypic Continuum of <i>ATP1A3</i> -Related Disorders. <i>Neurology</i> , 2022, 99, .	1.5	16
5709	In-silico profiling of deleterious non-synonymous single nucleotide polymorphisms of ARSA (arylsulphatase A) for enhanced diagnosis of metachromatic leukodystrophy. , 2022, 34, 201079.		0
5710	Loss of FOCAD, operating via the SKI messenger RNA surveillance pathway, causes a pediatric syndrome with liver cirrhosis. <i>Nature Genetics</i> , 2022, 54, 1214-1226.	9.4	6
5711	Role of Mitochondrial Mutations in Ocular Aggregopathy. <i>Cureus</i> , 2022, , .	0.2	0
5712	Autosomal dominant optic atrophy caused by six novel pathogenic OPA1 variants and genotype-phenotype correlation analysis. <i>BMC Ophthalmology</i> , 2022, 22, .	0.6	1
5713	Genetic evidence of tri-genealogy hypothesis on the origin of ethnic minorities in Yunnan. <i>BMC Biology</i> , 2022, 20, .	1.7	5
5714	Whole-exome sequencing identified mutational profiles of urothelial carcinoma post kidney transplantation. <i>Journal of Translational Medicine</i> , 2022, 20, .	1.8	2
5715	ASXL1 mutations predict inferior molecular response to nilotinib treatment in chronic myeloid leukemia. <i>Leukemia</i> , 2022, 36, 2242-2249.	3.3	14
5716	Germline Mutations in <i>CIDEB</i> and Protection against Liver Disease. <i>New England Journal of Medicine</i> , 2022, 387, 332-344.	13.9	42
5717	PNPLA1-Mediated Acylceramide Biosynthesis and Autosomal Recessive Congenital Ichthyosis. <i>Metabolites</i> , 2022, 12, 685.	1.3	1
5718	Studies on the Indian catfish <i>Clarias magur</i> reveal <i>Insulin-Like Growth Factor II</i> to be the major type and its upregulation in high-growth performing fish. <i>Aquaculture Research</i> , 2022, 53, 5253-5260.	0.9	2
5719	Clinical, biological, radiological, and genetic study of LPAC syndrome in Tunisian patients. <i>Arab Journal of Gastroenterology</i> , 2022, 23, 210-217.	0.4	1
5720	In silico Prediction of Deleterious Single Nucleotide Polymorphism in S100A4 Metastatic Gene: Potential Early Diagnostic Marker. <i>Contrast Media and Molecular Imaging</i> , 2022, 2022, 1-12.	0.4	0
5721	Early-onset diabetes involving three consecutive generations had different clinical features from age-matched type 2 diabetes without a family history in China. <i>Endocrine</i> , 0, , .	1.1	0
5722	Case Report: Three novel pathogenic <i>ABCC2</i> mutations identified in two patients with Dubin-Johnson syndrome. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	3
5724	Clinical and Genetic Analysis of <i>RDH12</i> -Associated Retinopathy in 27 Chinese Families: A Hypomorphic Allele Leads to Cone-Rod Dystrophy. , 2022, 63, 24.		8

#	ARTICLE	IF	CITATIONS
5725	Novel compound heterozygous mutation in STAMBP causes a neurodevelopmental disorder by disrupting cortical proliferation. <i>Frontiers in Neuroscience</i> , 0, 16, .	1.4	1
5726	Novel compound heterozygous variants of <sc>DNAH17</sc> in a Chinese infertile man with multiple morphological abnormalities of sperm flagella. <i>Andrologia</i> , 2022, 54, .	1.0	2
5727	Prediction of post-translational modification cross-talk and mutation within proteins via imbalanced learning. <i>Expert Systems With Applications</i> , 2023, 211, 118593.	4.4	0
5729	De novo variants in genes regulating stress granule assembly associate with neurodevelopmental disorders. <i>Science Advances</i> , 2022, 8, .	4.7	16
5732	Humanized substitutions of Vmat1 in mice alter amygdala-dependent behaviors associated with the evolution of anxiety. <i>IScience</i> , 2022, 25, 104800.	1.9	1
5734	Genetic predisposition to portoâ€sinusoidal vascular disorder: A functional genomicâ€based, multigenerational family study. <i>Hepatology</i> , 2023, 77, 501-511.	3.6	7
5735	Whole Exome Sequencing Revealed Variants That Predict Pulmonary Artery Involvement in Patients with Takayasu Arteritis. <i>Journal of Inflammation Research</i> , 0, Volume 15, 4817-4831.	1.6	3
5736	Identification of novel genes by targeted exome sequencing in Retinoblastoma. <i>Ophthalmic Genetics</i> , 2022, 43, 771-788.	0.5	1
5737	mvPPT: A Highly Efficient and Sensitive Pathogenicity Prediction Tool for Missense Variants. <i>Genomics, Proteomics and Bioinformatics</i> , 2023, 21, 414-426.	3.0	2
5738	Diet X Gene Interactions Control Femoral Bone Adaptation to Low Dietary Calcium. <i>JBMR Plus</i> , 2022, 6, .	1.3	1
5739	Genotypeâ€phenotype characteristics of Vietnamese patients diagnosed with Charcotâ€Marieâ€Tooth disease. <i>Brain and Behavior</i> , 2022, 12, .	1.0	3
5740	Diverse mutations and structural variations contribute to Notch signaling deregulation in paediatric Tâ€cell lymphoblastic lymphoma. <i>Pediatric Blood and Cancer</i> , 2022, 69, .	0.8	6
5741	AutoCaSc: Prioritizing candidate genes for neurodevelopmental disorders. <i>Human Mutation</i> , 2022, 43, 1795-1807.	1.1	5
5742	Multiancestry exome sequencing reveals INHBE mutations associated with favorable fat distribution and protection from diabetes. <i>Nature Communications</i> , 2022, 13, .	5.8	18
5743	Clinical significance of genetic variation in hypertrophic cardiomyopathy: comparison of computational tools to prioritize missense variants. <i>Frontiers in Cardiovascular Medicine</i> , 0, 9, .	1.1	5
5744	Whole exome and genome sequencing in mendelian disorders: a diagnostic and health economic analysis. <i>European Journal of Human Genetics</i> , 2022, 30, 1121-1131.	1.4	34
5746	Prevalence estimates of putatively pathogenic leptin variants in the gnomAD database. <i>PLoS ONE</i> , 2022, 17, e0266642.	1.1	2
5747	Challenges in quantifying genome erosion for conservation. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	6

#	ARTICLE	IF	CITATIONS
5748	m6A-TSHub: Unveiling the Context-Specific m6A Methylation and m6A-Affecting Mutations in 23 Human Tissues. <i>Genomics, Proteomics and Bioinformatics</i> , 2023, 21, 678-694.	3.0	21
5749	Genetic architecture of tuberculosis susceptibility: A comprehensive research synopsis, meta-analyses, and epidemiological evidence. <i>Infection, Genetics and Evolution</i> , 2022, 104, 105352.	1.0	3
5750	Assessing the Relationship between the Predicted Deleteriousness of 97 Missense Mutations in 68 Lipid Genes and Phenotypic Variation of Complex Fatness Traits in Pigs. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0
5751	Screening of Pathogenic Missense Single Nucleotide Variants From <i>LHPP</i> Gene Associated With the Hepatocellular Carcinoma: An <i>In silico</i> Approach. <i>Bioinformatics and Biology Insights</i> , 2022, 16, 117793222211155.	1.0	1
5752	SNPs Classification and Terminology: dbSNP Reference SNP (rs) Gene and Consequence Annotation. , 2022, , 3-12.		0
5753	Oxa-376 and Oxa-530 variants of Î²-lactamase: computational study uncovers potential therapeutic targets of <i>Acinetobacter baumannii</i>. <i>RSC Advances</i> , 2022, 12, 24319-24338.	1.7	4
5754	Germline variants in cancer-predisposing genes in pancreatic cancer patients with a family history of cancer. <i>Japanese Journal of Clinical Oncology</i> , 0, , .	0.6	1
5755	A genetic model for central chondrosarcoma evolution correlates with patient outcome. <i>Genome Medicine</i> , 2022, 14, .	3.6	9
5756	Study of variants in genes implicated in rare familial migraine syndromes and their association with migraine in 200,000 exomeâ€sequenced UK Biobank participants. <i>Annals of Human Genetics</i> , 0, , .	0.3	4
5757	Ancestry-driven recalibration of tumor mutational burden and disparate clinical outcomes in response to immune checkpoint inhibitors. <i>Cancer Cell</i> , 2022, 40, 1161-1172.e5.	7.7	44
5758	Mutations in <i>BCOR</i> , a co-repressor of <i>CRX/OTX2</i> , are associated with early-onset retinal degeneration. <i>Science Advances</i> , 2022, 8, .	4.7	6
5759	A Gly684Ala substitution in the androgen receptor is the cause for azoospermia in a Chinese family with mild androgen insensitivity syndrome and normal hormone levels. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1
5760	Identification and in-silico characterization of splice-site variants from a large cardiogenetic national registry. <i>European Journal of Human Genetics</i> , 2023, 31, 512-520.	1.4	3
5761	Ultra-Rare BRD9 Loss-of-Function Variants Limit the Antiviral Action of Interferon. <i>Scientific Reports</i> , 2022, 12, .	1.6	1
5762	A heterozygous LAMA5 variant may contribute to slowly progressive, vinculin-enhanced familial FSGS and pulmonary defects. <i>JCI Insight</i> , 2022, 7, .	2.3	1
5763	Polymorphisms of hypertension susceptibility genes as a risk factors of preeclampsia in the Caucasian population of central Russia. <i>Placenta</i> , 2022, 129, 51-61.	0.7	10
5764	Characterization of <i>POR</i> haplotype distribution in African populations and comparison with other global populations. <i>Pharmacogenomics</i> , 2022, 23, 771-782.	0.6	1
5765	Tracing Bai-Yue Ancestry in Aboriginal Li People on Hainan Island. <i>Molecular Biology and Evolution</i> , 2022, 39, .	3.5	13

#	ARTICLE	IF	CITATIONS
5766	Novel mutations in GJA1 in two Brazilian families with oculodentodigital dysplasia. <i>Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology</i> , 2023, 135, 96-100.	0.2	0
5767	Lymphoma as an Exclusion Criteria for CVID Diagnosis Revisited. <i>Journal of Clinical Immunology</i> , 2023, 43, 181-191.	2.0	1
5768	Two Loci Contribute to Age-Related Hearing Loss Resistance in the Japanese Wild-Derived Inbred MSM/Ms Mice. <i>Biomedicines</i> , 2022, 10, 2221.	1.4	3
5769	A rapid turnaround gene panel for severe autoinflammation: Genetic results within 48 hours. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	1
5770	Characterization of <i>CYP2D6</i> Pharmacogenetic Variation in Sub-Saharan African Populations. <i>Clinical Pharmacology and Therapeutics</i> , 2023, 113, 643-659.	2.3	12
5771	Functional and Structural Impact of Deleterious Missense Single Nucleotide Polymorphisms in the NR3C1, CYP3A5, and TNF- α Genes: An In Silico Analysis. <i>Biomolecules</i> , 2022, 12, 1307.	1.8	3
5772	Genetic analyses of the electrocardiographic QT interval and its components identify additional loci and pathways. <i>Nature Communications</i> , 2022, 13, .	5.8	15
5773	Novel clinical, molecular and bioinformatics insights into the genetic background of autism. <i>Human Genomics</i> , 2022, 16, .	1.4	2
5774	Detection of a novel SETBP1 variant in a Chinese neonate with Schinzel-Giedion syndrome. <i>Frontiers in Pediatrics</i> , 0, 10, .	0.9	3
5775	A novel germline mutation of the <i>PALB</i> gene in a young Yakut breast cancer woman. <i>Siberian Journal of Oncology</i> , 2022, 21, 72-79.	0.1	2
5776	RMDisease V2.0: an updated database of genetic variants that affect RNA modifications with disease and trait implication. <i>Nucleic Acids Research</i> , 2023, 51, D1388-D1396.	6.5	23
5777	Prediction of evolutionary constraint by genomic annotations improves functional prioritization of genomic variants in maize. <i>Genome Biology</i> , 2022, 23, .	3.8	7
5778	Comparative Genomics Provides Etiologic and Biological Insight into Melanoma Subtypes. <i>Cancer Discovery</i> , 2022, 12, 2856-2879.	7.7	22
5779	In silico investigation of nonsynonymous single nucleotide polymorphisms in <i>BCL2</i> apoptosis regulator gene to design novel protein-based drugs against cancer. <i>Journal of Cellular Biochemistry</i> , 2022, 123, 2044-2056.	1.2	2
5780	Rapid genome sequencing for pediatrics. <i>Human Mutation</i> , 2022, 43, 1507-1518.	1.1	9
5781	SNP discovery and association study for growth, fatness and meat quality traits in Iberian crossbred pigs. <i>Scientific Reports</i> , 2022, 12, .	1.6	6
5782	Novel Variants and Phenotypes in <i>NEUROG3</i> -Associated Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 108, 52-58.	1.8	1
5784	Heterozygous LRP1 deficiency causes developmental dysplasia of the hip by impairing triradiate chondrocytes differentiation due to inhibition of autophagy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	6

#	ARTICLE	IF	CITATIONS
5785	Towards a Better Understanding of Genotype-Phenotype Correlations and Therapeutic Targets for Cardiocutaneous Genes: The Importance of Functional Studies above Prediction. <i>International Journal of Molecular Sciences</i> , 2022, 23, 10765.	1.8	4
5786	Pedigree-based study to identify GOLGB1 as a risk gene for bipolar disorder. <i>Translational Psychiatry</i> , 2022, 12, .	2.4	0
5787	Identification of a novel de novo pathogenic variant in GFAP in an Iranian family with Alexander disease by whole-exome sequencing. <i>European Journal of Medical Research</i> , 2022, 27, .	0.9	0
5788	Identification of variants in genes associated with autoinflammatory disorders in a cohort of patients with psoriatic arthritis. <i>RMD Open</i> , 2022, 8, e002561.	1.8	7
5789	Genomic insights into the evolutionary history and diversification of bulb traits in garlic. <i>Genome Biology</i> , 2022, 23, .	3.8	6
5790	In silico analyses of Wnt1 nsSNPs reveal structurally destabilizing variants, altered interactions with Frizzled receptors and its deregulation in tumorigenesis. <i>Scientific Reports</i> , 2022, 12, .	1.6	0
5791	P. Ala278Val mutation might cause a pathogenic defect in HEXB folding leading to the Sandhoff disease. <i>Metabolic Brain Disease</i> , 0, , .	1.4	0
5792	Genomic profiling with whole-exome sequencing revealed distinct mutations and novel pathways in Asian melanoma. <i>Journal of Dermatology</i> , 2022, 49, 1299-1309.	0.6	3
5793	Exome sequencing in a Swedish family with PMS2 mutation with varying penetrance of colorectal cancer: investigating the presence of genetic risk modifiers in colorectal cancer risk. <i>European Journal of Cancer Prevention</i> , 2023, 32, 113-118.	0.6	2
5794	Single Nucleotide Variants in KIF14 Gene May Have Prognostic Value in Breast Cancer. <i>Molecular Diagnosis and Therapy</i> , 2022, 26, 665-678.	1.6	1
5795	Phenotypic spectrum in recessive STING-associated vasculopathy with onset in infancy: Four novel cases and analysis of previously reported cases. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	5
5796	Enamel defects in Acp4R110C/R110C mice and human ACP4 mutations. <i>Scientific Reports</i> , 2022, 12, .	1.6	6
5797	Molecular Dynamic Simulation of Neurexin1± Mutations Associated with Mental Disorder. <i>Journal of Molecular Neuroscience</i> , 0, , .	1.1	0
5798	Novel Titin Gene Mutation Causing Autosomal Dominant Limb-Girdle Muscular Dystrophy. <i>Cureus</i> , 2022, , .	0.2	1
5799	Description of the molecular and phenotypic spectrum in Chinese patients with aggrecan deficiency: Novel ACAN heterozygous variants in eight Chinese children and a review of the literature. <i>Frontiers in Endocrinology</i> , 0, 13, .	1.5	0
5801	Pulmonary Hypertension Associated Genetic Variants in Sarcoidosis Associated Pulmonary Hypertension. <i>Diagnostics</i> , 2022, 12, 2564.	1.3	1
5802	Genotype/Phenotype Correlation of Cases with <i>PTPN11</i> Gene Mutation: Eastern Black Sea Experience. <i>Journal of Ankara University Faculty of Medicine</i> , 2022, 75, 368-372.	0.0	0
5803	Channel HCN4 mutation R666Q associated with sporadic arrhythmia decreases channel electrophysiological function and increases protein degradation. <i>Journal of Biological Chemistry</i> , 2022, 298, 102599.	1.6	1

#	ARTICLE	IF	CITATIONS
5804	Expanding the genetic spectrum for Chinese familial hypercholesterolemia population with six genetic mutations identified using a next-generation sequencing-based laboratory-developed screening test. <i>Molecular Genetics & Genomic Medicine</i> , 0, , .	0.6	2
5806	The fly homolog of <i>SUPT16H</i> , a gene associated with neurodevelopmental disorders, is required in a cell-autonomous fashion for cell survival. <i>Human Molecular Genetics</i> , 2023, 32, 984-997.	1.4	6
5808	MetaRNN: differentiating rare pathogenic and rare benign missense SNVs and InDels using deep learning. <i>Genome Medicine</i> , 2022, 14, .	3.6	28
5809	The Modifying Effect of Obesity on the Association of Matrix Metalloproteinase Gene Polymorphisms with Breast Cancer Risk. <i>Biomedicines</i> , 2022, 10, 2617.	1.4	11
5810	Matrix Metalloproteinase Gene Polymorphisms Are Associated with Breast Cancer in the Caucasian Women of Russia. <i>International Journal of Molecular Sciences</i> , 2022, 23, 12638.	1.8	11
5811	Novel pathogenic variant combination in LPL causing familial chylomicronemia syndrome in an Asian family and experimental validation in vitro: a case report. <i>Translational Pediatrics</i> , 2022, 11, 1717-1725.	0.5	2
5812	Eosinophilic Infiltration of the Sino-Atrial Node in Sudden Cardiac Death Caused by Long QT Syndrome. <i>International Journal of Molecular Sciences</i> , 2022, 23, 11666.	1.8	1
5813	Preliminary study on the molecular features of mutation in multiple primary oral cancer by whole exome sequencing. <i>Frontiers in Oncology</i> , 0, 12, .	1.3	3
5814	Case report: Functional characterization of a de novo c.145G>A p.Val49Met pathogenic variant in a case of PIGA-CDG with megacolon. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1
5816	Mutation of the MYH3 gene causes recessive cleft palate in Limousine cattle. <i>Genetics Selection Evolution</i> , 2022, 54, .	1.2	2
5817	Global population genetics and diversity in the TAS2R bitter taste receptor family. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	4
5818	Shared genetic risk factors and causal association between psoriasis and coronary artery disease. <i>Nature Communications</i> , 2022, 13, .	5.8	20
5819	In Silico Study of ULK1 Gene as a Susceptible Biomarker for Neurodegeneration. <i>Proceedings of the National Academy of Sciences India Section B - Biological Sciences</i> , 0, , .	0.4	0
5820	Characterization of Arabian Peninsula whole exomes: Contributing to the catalogue of human diversity. <i>IScience</i> , 2022, 25, 105336.	1.9	0
5821	Parkinson's disease gene prioritising using an efficient and biologically appropriate network-based consensus strategy. <i>Journal of Computational Science</i> , 2022, 65, 101879.	1.5	0
5822	Comprehensive in silico prioritization of pathogenic nsSNPs in human Î²-adducin gene towards finding its relation with cancer. , 2022, 34, 201119.		0
5823	In silico assessment of missense point mutations on human cathelicidin LL-37. <i>Journal of Molecular Graphics and Modelling</i> , 2023, 118, 108368.	1.3	1
5824	A missense variant in <i>NCF1</i> is associated with susceptibility to unexplained recurrent spontaneous abortion. <i>Open Life Sciences</i> , 2022, 17, 1443-1450.	0.6	0

#	ARTICLE	IF	CITATIONS
5825	Machine learning with in silico analysis markedly improves survival prediction modeling in colon cancer patients. <i>Cancer Medicine</i> , 2023, 12, 7603-7615.	1.3	4
5826	Deleterious Variation in Natural Populations and Implications for Conservation Genetics. <i>Annual Review of Animal Biosciences</i> , 2023, 11, 93-114.	3.6	32
5827	Analysis of 1276 Haplotype-Resolved Genomes Allows Characterization of Cis- and Trans-Abundant Genes. <i>Methods in Molecular Biology</i> , 2023, , 237-272.	0.4	0
5828	Population Genomics for Insect Conservation. <i>Annual Review of Animal Biosciences</i> , 2023, 11, 115-140.	3.6	13
5829	A novel missense mutation in GREB1L identified in a three-generation family with renal hypodysplasia/aplasia-3. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, .	1.2	4
5830	Identification and Characterization of Novel Mutations in Chronic Kidney Disease (CKD) and Autosomal Dominant Polycystic Kidney Disease (ADPKD) in Saudi Subjects by Whole-Exome Sequencing. <i>Medicina (Lithuania)</i> , 2022, 58, 1657.	0.8	3
5831	Bilateral Nonsyndromic Sensorineural Hearing Loss Caused by a NARS2 Mutation. <i>Cureus</i> , 2022, , .	0.2	2
5832	Identification of a novel ESR1 mutation in a Chinese PCOS woman with estrogen insensitivity in IVF treatment. <i>Reproductive Biology and Endocrinology</i> , 2022, 20, .	1.4	3
5833	Loci on chromosome 12q13.2 encompassing ERBB3, PA2G4 and RAB5B are associated with polycystic ovary syndrome. <i>Gene</i> , 2023, 852, 147062.	1.0	6
5834	Somatic Mutations Alter Interleukin Signaling Pathways in Grade II Invasive Breast Cancer Patients: An Egyptian Experience. <i>Current Issues in Molecular Biology</i> , 2022, 44, 5890-5901.	1.0	0
5835	Identification of prolactin receptor variants with diverse effects on receptor signalling. <i>Journal of Molecular Endocrinology</i> , 2023, 70, .	1.1	2
5837	Millennium-old pathogenic Mendelian mutation discovery for multiple osteochondromas from a Gaelic Medieval graveyard. <i>European Journal of Human Genetics</i> , 2023, 31, 248-251.	1.4	2
5838	African Suid Genomes Provide Insights into the Local Adaptation to Diverse African Environments. <i>Molecular Biology and Evolution</i> , 2022, 39, .	3.5	9
5839	Lynch syndrome, molecular mechanisms and variant classification. <i>British Journal of Cancer</i> , 2023, 128, 726-734.	2.9	8
5840	Novel and Founder Pathogenic Variants in X-Linked Alport Syndrome Families in Greece. <i>Genes</i> , 2022, 13, 2203.	1.0	3
5841	Somatic mutations in tumor and plasma of locoregional recurrent and/or metastatic head and neck cancer using a next-generation sequencing panel: A preliminary study. <i>Cancer Medicine</i> , 2023, 12, 6615-6622.	1.3	1
5843	Efficacy of low-dose hCG on FET cycle in patients with recurrent implantation failure. <i>Frontiers in Endocrinology</i> , 0, 13, .	1.5	1
5844	Genetic interaction between GABRA1 and ERBB4 variants in the pathogenesis of genetic generalized epilepsy. <i>Epilepsy Research</i> , 2023, 189, 107070.	0.8	1

#	ARTICLE	IF	CITATIONS
5845	A Hybrid Quantum Image-Matching Algorithm. <i>Entropy</i> , 2022, 24, 1816.	1.1	2
5846	Analysis of SLC26A4, FOXP1, and KCNJ10 Gene Variants in Patients with Incomplete Partition of the Cochlea and Enlarged Vestibular Aqueduct (EVA) Anomalies. <i>International Journal of Molecular Sciences</i> , 2022, 23, 15372.	1.8	2
5847	Incidence of Hereditary Gastric Cancer May Be Much Higher than Reported. <i>Cancers</i> , 2022, 14, 6125.	1.7	0
5848	Diverse monogenic subforms of human spermatogenic failure. <i>Nature Communications</i> , 2022, 13, .	5.8	17
5850	Genetic Factors Causing Thyroid Dysmorphogenesis as the Major Etiologies for Primary Congenital Hypothyroidism: Clinical and Genetic Characterization of 33 Patients. <i>Journal of Clinical Medicine</i> , 2022, 11, 7313.	1.0	1
5851	WDR87 interacts with CFAP47 protein in the middle piece of spermatozoa flagella to participate in sperm tail assembly. <i>Molecular Human Reproduction</i> , 2022, 29, .	1.3	2
5852	Effects of Pre-Pregnancy Overweight/Obesity on the Pattern of Association of Hypertension Susceptibility Genes with Preeclampsia. <i>Life</i> , 2022, 12, 2018.	1.1	6
5853	The Association of Variants within Types V and XI Collagen Genes with Knee Joint Laxity Measurements. <i>Genes</i> , 2022, 13, 2359.	1.0	1
5854	Genetic architecture of heart failure with preserved versus reduced ejection fraction. <i>Nature Communications</i> , 2022, 13, .	5.8	14
5856	TogoVar: A comprehensive Japanese genetic variation database. <i>Human Genome Variation</i> , 2022, 9, .	0.4	4
5857	Targeted deep sequencing analyses of long QT syndrome in a Japanese population. <i>PLoS ONE</i> , 2022, 17, e0277242.	1.1	0
5858	Whole exome sequencing in dense families suggests genetic pleiotropy amongst Mendelian and complex neuropsychiatric syndromes. <i>Scientific Reports</i> , 2022, 12, .	1.6	2
5859	From high masked to high realized genetic load in inbred Scandinavian wolves. <i>Molecular Ecology</i> , 2023, 32, 1567-1580.	2.0	11
5861	Intra- and Interspecies RNA-Seq Based Variants in the Lactation Process of Ruminants. <i>Animals</i> , 2022, 12, 3592.	1.0	0
5862	De novo mutation hotspots in homologous protein domains identify function-altering mutations in neurodevelopmental disorders. <i>American Journal of Human Genetics</i> , 2023, 110, 92-104.	2.6	3
5863	Insights from incorporating quantum computing into drug design workflows. <i>Bioinformatics</i> , 2023, 39, .	1.8	9
5864	Effects of the pathological E200K mutation on human prion protein: A computational screening and molecular dynamics approach. <i>Journal of Cellular Biochemistry</i> , 0, .	1.2	0
5865	Rapid Targeted Sequencing Using Dried Blood Spot Samples for Patients With Suspected Actionable Genetic Diseases. <i>Annals of Laboratory Medicine</i> , 2023, 43, 280-289.	1.2	4

#	ARTICLE	IF	CITATIONS
5866	VPatho: a deep learning-based two-stage approach for accurate prediction of gain-of-function and loss-of-function variants. <i>Briefings in Bioinformatics</i> , 2023, 24, .	3.2	5
5867	Identification of deleterious nsSNPs in human HGF gene: in silico approach. <i>Journal of Biomolecular Structure and Dynamics</i> , 2023, 41, 11889-11903.	2.0	2
5868	Whole-exome sequencing identifies FANC heterozygous germline mutation as an adverse factor for immunosuppressive therapy in Chinese aplastic anemia patients aged 40 or younger: a single-center retrospective study. <i>Annals of Hematology</i> , 0, , .	0.8	0
5869	Case report: KPTN gene-related syndrome associated with a spectrum of neurodevelopmental anomalies including severe epilepsy. <i>Frontiers in Neurology</i> , 0, 13, .	1.1	0
5870	Novel high-risk missense mutations identification in FAT4 gene causing Hennekam syndrome and Van Maldergem syndrome 2 through molecular dynamics simulation. <i>Informatics in Medicine Unlocked</i> , 2023, 37, 101160.	1.9	3
5871	Biallelic mutations in <i>CFAP54</i> cause male infertility with severe MMAF and NOA. <i>Journal of Medical Genetics</i> , 2023, 60, 827-834.	1.5	7
5872	Functional Characterization of a Spectrum of Novel Romano-Ward Syndrome KCNQ1 Variants. <i>International Journal of Molecular Sciences</i> , 2023, 24, 1350.	1.8	2
5873	A Case of Congenital Hypotonia and Developmental Delay in an Individual with a De Novo Variant Outside of the Canonical HX-Motif of ATN1. <i>Case Reports in Genetics</i> , 2023, 2023, 1-5.	0.1	0
5875	PDGFRB and NOTCH3 Mutations are Detectable in a Wider Range of Pericytic Tumors, Including Myopericytomas, Angioleiomyomas, Glomus Tumors, and Their Combined Tumors. <i>Modern Pathology</i> , 2023, 36, 100070.	2.9	4
5876	A novel frameshift mutation in TRPV6 is associated with hereditary pancreatitis. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1
5877	E96V Mutation in the Kdelr3 Gene Is Associated with Type 2 Diabetes Susceptibility in Obese NZO Mice. <i>International Journal of Molecular Sciences</i> , 2023, 24, 845.	1.8	0
5878	A Pathogenic Variant Reclassified to the Pseudogene PMS2P1 in a Patient with Suspected Hereditary Cancer. <i>International Journal of Molecular Sciences</i> , 2023, 24, 1398.	1.8	1
5879	Identification of the SIRT1 gene's most harmful non-synonymous SNPs and their effects on functional and structural features-an in silico analysis. <i>F1000Research</i> , 0, 12, 66.	0.8	0
5880	Dendritic Spine in Autism Genetics: Whole-Exome Sequencing Identifying De Novo Variant of CTTNBP2 in a Quad Family Affected by Autism Spectrum Disorder. <i>Children</i> , 2023, 10, 80.	0.6	3
5881	Frequency of Pathogenic Germline Mutations in Early and Late Onset Familial Breast Cancer Patients Using Multi-Gene Panel Sequencing: An Egyptian Study. <i>Genes</i> , 2023, 14, 106.	1.0	3
5882	Epigenomic charting and functional annotation of risk loci in renal cell carcinoma. <i>Nature Communications</i> , 2023, 14, .	5.8	10
5883	Bi-allelic human <i>TEKT3</i> mutations cause male infertility with oligoasthenoteratozoospermia owing to acrosomal hypoplasia and reduced progressive motility. <i>Human Molecular Genetics</i> , 2023, 32, 1730-1740.	1.4	4
5884	Identifying rare genetic variants in 21 highly multiplex autism families: the role of diagnosis and autistic traits. <i>Molecular Psychiatry</i> , 2023, 28, 2148-2157.	4.1	4

#	ARTICLE	IF	CITATIONS
5887	OUTER RETINAL TUBULATION IN BIETTI CRYSTALLINE DYSTROPHY ASSOCIATED WITH THE RETINAL PIGMENT EPITHELIUM ATROPHY. <i>Retina</i> , 2023, 43, 659-669.	1.0	2
5888	The PER3rs772027021 SNP induces pigmentation phenotypes of dyschromatosis universalis hereditaria. <i>Journal of Molecular Medicine</i> , 2023, 101, 279-294.	1.7	2
5889	Genetic variants of cancer-associated genes analyzed using next-generation sequencing in small sporadic vestibular schwannomas. <i>Oncology Letters</i> , 2023, 25, .	0.8	0
5890	Assessing the relationship between the in silico predicted consequences of 97 missense mutations mapping to 68 genes related to lipid metabolism and their association with porcine fatness traits. <i>Genomics</i> , 2023, 115, 110589.	1.3	0
5891	Rare variant analyses in large-scale cohorts identified SLC13A1 associated with chronic pain. <i>Pain</i> , 2023, 164, 1841-1851.	2.0	3
5893	Selective vitamins as potential options for dietary therapeutic interventions: In silico and In vitro insights from mutant C terminal fragment of FGA. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 2023, 230, 106290.	1.2	0
5894	Whole-exome sequencing and electrophysiological study reveal a novel loss-of-function mutation of KCNA10 in epinephrine provoked long QT syndrome with familial history of sudden cardiac death. <i>Legal Medicine</i> , 2023, 62, 102245.	0.6	0
5895	Genomic health is dependent on long-term population demographic history. <i>Molecular Ecology</i> , 2023, 32, 1943-1954.	2.0	5
5896	Intergenic Interactions of SBNO1, NFAT5 and GLT8D1 Determine the Susceptibility to Knee Osteoarthritis among Europeans of Russia. <i>Life</i> , 2023, 13, 405.	1.1	6
5897	The in-silico evaluation of important GLUT9 residue for uric acid transport based on renal hypouricemia type 2. <i>Chemico-Biological Interactions</i> , 2023, 373, 110378.	1.7	1
5898	Genetic Variant Overlap Analysis Identifies Established and Putative Genes Involved in Pulmonary Fibrosis. <i>International Journal of Molecular Sciences</i> , 2023, 24, 2790.	1.8	2
5899	Integration of deep learning with Ramachandran plot molecular dynamics simulation for genetic variant classification. <i>IScience</i> , 2023, 26, 106122.	1.9	2
5901	Novel ELAC2 Mutations in Individuals Presenting with Variably Severe Neurological Disease in the Presence or Absence of Cardiomyopathy. <i>Life</i> , 2023, 13, 445.	1.1	2
5902	Delineating the Spectrum of Genetic Variants Associated with Bardet-Biedl Syndrome in Consanguineous Pakistani Pedigrees. <i>Genes</i> , 2023, 14, 404.	1.0	5
5903	The Usher syndrome 1C protein harmonin regulates canonical Wnt signaling. <i>Frontiers in Cell and Developmental Biology</i> , 0, 11, .	1.8	3
5904	Genetic architecture of hippocampus subfields volumes in Alzheimer's disease. <i>CNS Neuroscience and Therapeutics</i> , 2024, 30, .	1.9	0
5905	Inherited mutations affecting the SRCAP complex are central in moderate-penetrance predisposition to uterine leiomyomas. <i>American Journal of Human Genetics</i> , 2023, 110, 460-474.	2.6	4
5906	Extensive set of African ancestry-informative markers (AIMs) to study ancestry and population health. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	0

#	ARTICLE	IF	CITATIONS
5907	Systematic Assessment of Protein C-Termini Mutated in Human Disorders. <i>Biomolecules</i> , 2023, 13, 355.	1.8	0
5908	GJB4 variants linked to skin disease exhibit a trafficking deficiency en route to gap junction formation that can be restored by co-expression of select connexins. <i>Frontiers in Cell and Developmental Biology</i> , 0, 11, .	1.8	1
5910	SNP based analysis depicts phenotypic variability in heme oxygenase-1 protein. <i>Biyokimya Dergisi</i> , 2023, .	0.1	0
5911	Deficiency of primate-specific SSX1 induced asthenoteratozoospermia in infertile men and cynomolgus monkey and tree shrew models. <i>American Journal of Human Genetics</i> , 2023, 110, 516-530.	2.6	5
5912	Whole Exome Sequencing Reveals Novel Candidate Genes in Familial Forms of Glaucomatous Neurodegeneration. <i>Genes</i> , 2023, 14, 495.	1.0	0
5913	Genetic background of idiopathic neurodevelopmental delay patients with significant brain deviation volume. <i>Chinese Medical Journal</i> , 0, Publish Ahead of Print, .	0.9	0
5914	GABRG2 Variants Associated with Febrile Seizures. <i>Biomolecules</i> , 2023, 13, 414.	1.8	4
5915	Novel <i>IRF6</i> variant in orofacial cleft patients from Durban, South Africa. <i>Molecular Genetics & Genomic Medicine</i> , 0, , .	0.6	1
5916	Whole-Genome Sequencing Data Reveal New Loci Affecting Milk Production in German Black Pied Cattle (DSN). <i>Genes</i> , 2023, 14, 581.	1.0	0
5917	Computational and mitochondrial functional studies of novel compound heterozygous variants in SPATA5 gene support a causal link with epileptogenic encephalopathy. <i>Human Genomics</i> , 2023, 17, .	1.4	1
5918	Unified views on variant impact across many diseases. <i>Trends in Genetics</i> , 2023, 39, 442-450.	2.9	3
5919	From gene to dose: Long-read sequencing and *-allele tools to refine phenotype predictions of CYP2C19. <i>Frontiers in Pharmacology</i> , 0, 14, .	1.6	2
5920	Identification of EPX Variants in Human Eosinophilic Granulomatosis With Polyangiitis (Churg-Strauss). <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2023, , .	2.0	0
5921	Minimal residual disease detection by mutation-specific droplet digital PCR for leukemia/lymphoma. <i>International Journal of Hematology</i> , 2023, 117, 910-918.	0.7	2
5922	Novel mutations in the ABCD1 gene caused adrenomyeloneuropathy in the Chinese population. <i>Frontiers in Neurology</i> , 0, 14, .	1.1	0
5923	Variant Landscape of 15 Genes Involved in Corneal Dystrophies: Report of 30 Families and Comprehensive Analysis of the Literature. <i>International Journal of Molecular Sciences</i> , 2023, 24, 5012.	1.8	0
5924	Evaluation of Human-Induced Pluripotent Stem Cells Derived from a Patient with Schwartzâ€™s Jampel Syndrome Revealed Distinct Hyperexcitability in the Skeletal Muscles. <i>Biomedicines</i> , 2023, 11, 814.	1.4	2
5925	Genetic architecture of spatial electrical biomarkers for cardiac arrhythmia and relationship with cardiovascular disease. <i>Nature Communications</i> , 2023, 14, .	5.8	1

#	ARTICLE	IF	CITATIONS
5926	Pseudotyped Viruses for Phlebovirus. <i>Advances in Experimental Medicine and Biology</i> , 2023, , 253-264.	0.8	0
5927	Whole exome sequencing study identifies candidate loss of function variants and locus heterogeneity in familial cholesteatoma. <i>PLoS ONE</i> , 2023, 18, e0272174.	1.1	3
5928	Hypercholesterolemia in the Malaysian Cohort Participants: Genetic and Non-Genetic Risk Factors. <i>Genes</i> , 2023, 14, 721.	1.0	1
5929	Revisiting the Biological and Clinical Impact of CDH1 Missense Variants. , 2023, , 79-97.		1
5930	Design of a low-density SNP panel for intramuscular fat content and fatty acid composition of backfat in free-range Iberian pigs. <i>Journal of Animal Science</i> , 2023, 101, .	0.2	4
5931	CLPP inhibition triggers apoptosis in human ovarian granulosa cells via COX5A abnormalityâ€“Mediated mitochondrial dysfunction. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	2
5932	Evaluation of SNPs from human IGFBP6 associated with gene expression: an <i>in-silico</i> study. <i>Journal of Biomolecular Structure and Dynamics</i> , 0, , 1-13.	2.0	1
5933	Global detection of human variants and isoforms by deep proteome sequencing. <i>Nature Biotechnology</i> , 2023, 41, 1776-1786.	9.4	43
5934	Computational Methods Summarizing Mutational Patterns in Cancer: Promise and Limitations for Clinical Applications. <i>Cancers</i> , 2023, 15, 1958.	1.7	1
5935	GNA11 Variants Identified in Patients with Hypercalcemia or Hypocalcemia. <i>Journal of Bone and Mineral Research</i> , 2020, 38, 907-917.	3.1	2
5936	A Novel Heterozygous Missense Variant in <i>Parathyroid Hormone 1</i> is Related to the Occurrence of Developmental Dysplasia of the Hip. <i>Genetic Testing and Molecular Biomarkers</i> , 2023, 27, 74-80.	0.3	0
5937	<i>In silico</i> exploration and molecular dynamics of deleterious SNPs on the human TERF1 protein triggering male infertility. <i>Journal of Biomolecular Structure and Dynamics</i> , 2023, 41, 14665-14688.	2.0	0
5938	Autoimmunity and immunodeficiency associated with monoallelic LIG4 mutations via haploinsufficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2023, , .	1.5	0
5940	Clinical and genetic analysis further delineates the phenotypic spectrum of ALDH1A3-related anophthalmia and microphthalmia. <i>European Journal of Human Genetics</i> , 0, , .	1.4	1
5941	Molecular Diagnosis of Genetic Diseases of the Kidney: Primer for Pediatric Nephrologists. , 2023, , 119-169.		0
5942	Comparison of two multi-trait association testing methods and sequence-based fine mapping of six additive QTL in Swiss Large White pigs. <i>BMC Genomics</i> , 2023, 24, .	1.2	2
5943	Surveillance of cfDNA Hot Spot Mutations in NSCLC Patients during Disease Progression. <i>International Journal of Molecular Sciences</i> , 2023, 24, 6958.	1.8	1
5944	Genomicâ€“transcriptomic evolution in lung cancer and metastasis. <i>Nature</i> , 2023, 616, 543-552.	13.7	44

#	ARTICLE	IF	CITATIONS
5945	Signal Transducer and Activator of Transcription 6 Changes and Protein Frustration by Single Amino Acid Substitutions: Implications for Cancer Progression. <i>OMICS A Journal of Integrative Biology</i> , 2023, 27, 171-179.	1.0	0
5946	The evolution of lung cancer and impact of subclonal selection in TRACERx. <i>Nature</i> , 2023, 616, 525-533.	13.7	62
5947	Whole-exome sequencing study of hypospadias. <i>IScience</i> , 2023, 26, 106663.	1.9	2
5948	Biallelic <i>NPR1</i> loss of function variants are responsible for neonatal systemic hypertension. <i>Journal of Medical Genetics</i> , 2023, 60, 993-998.	1.5	1
5949	<i>TP53</i> mutations predict poor response to immunotherapy in patients with metastatic solid tumors. <i>Cancer Medicine</i> , 2023, 12, 12438-12451.	1.3	2
5950	Research on computer vision technology based on BP-LSTM hybrid network. <i>Applied Mathematics and Nonlinear Sciences</i> , 2023, 8, 975-984.	0.9	1
5951	Contribution of LRP1 in Human Congenital Heart Disease Correlates with Its Roles in the Outflow Tract and Atrioventricular Cushion Development. <i>Genes</i> , 2023, 14, 947.	1.0	0
5952	Whole-exome sequencing in moyamoya patients of Northern-European origin identifies gene variants involved in Nitric Oxide metabolism: A pilot study. <i>Brain and Spine</i> , 2023, 3, 101745.	0.0	1
5975	Prediction of Functional Effects of Protein Amino Acid Mutations. <i>Lecture Notes in Computer Science</i> , 2023, , 59-71.	1.0	0
5994	AI in Genomics and Epigenomics. <i>Healthy Ageing and Longevity</i> , 2023, , 217-243.	0.2	0
5996	Advances in Eco-TILLING: In Search of Superior Natural Variants. , 2023, , 115-135.		0
6074	The role of genetic variation in CYP2R1, the principal vitamin D 25-hydroxylase, and CYP3A4 in vitamin D homeostasis. , 2024, , 341-357.		0
6100	Heterogeneities in Hereditary Cancer Genes as Revealed by a Large-Scale Genome Analysis. , 2023, , 59-78.		0
6107	Machine learning applications in cancer genomics. , 2024, , 41-72.		0
6111	Genetic and Genomic Results and Management. , 2024, , 93-110.		0
6124	Pan-cancer classification of multi-omics data based on machine learning models. <i>Network Modeling Analysis in Health Informatics and Bioinformatics</i> , 2024, 13, .	1.2	0