Comparison and integration of deleteriousness predicti SNVs in whole exome sequencing studies

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Citation Report

#	Article	IF	Citations
2	Rare and low-frequency variants and their association with plasma levels of fibrinogen, FVII, FVIII, and vWF. Blood, 2015, 126, e19-e29.	0.6	55
3	Secondary findings and carrier test frequencies in a large multiethnic sample. Genome Medicine, 2015, 7, 54.	3.6	47
4	<scp>SLC1A4</scp> mutations cause a novel disorder of intellectual disability, progressive microcephaly, spasticity and thin corpus callosum. Clinical Genetics, 2015, 88, 327-335.	1.0	49
5	Increased burden ofde novopredicted deleterious variants in complex congenital diaphragmatic hernia. Human Molecular Genetics, 2015, 24, 4764-4773.	1.4	65
6	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	6.0	646
7	A Note on the Evaluation of Mutation Prioritization Algorithms. , 2015, , .		0
8	ClinLabGeneticist: a tool for clinical management of genetic variants from whole exome sequencing in clinical genetic laboratories. Genome Medicine, 2015, 7, 77.	3.6	5
9	Standardizing Variant Interpretation in Genomic Sequencing: Implications for Genetic Counseling Practice. Current Genetic Medicine Reports, 2015, 3, 137-142.	1.9	1
10	Genomic variant annotation and prioritization with ANNOVAR and wANNOVAR. Nature Protocols, 2015, 10, 1556-1566.	5.5	727
11	The role of functional data in interpreting the effects of genetic variation. Molecular Biology of the Cell, 2015, 26, 3904-3908.	0.9	13
12	A Survey of Computational Tools to Analyze and Interpret Whole Exome Sequencing Data. International Journal of Genomics, 2016, 2016, 1-16.	0.8	37
13	Unravelling the Complexity of Inherited Retinal Dystrophies Molecular Testing: Added Value of Targeted Next-Generation Sequencing. BioMed Research International, 2016, 2016, 1-14.	0.9	47
14	Functional Studies and In Silico Analyses to Evaluate Non-Coding Variants in Inherited Cardiomyopathies. International Journal of Molecular Sciences, 2016, 17, 1883.	1.8	25
15	Autism Linked to Increased Oncogene Mutations but Decreased Cancer Rate. PLoS ONE, 2016, 11, e0149041.	1.1	25
16	Cystinuria Associated with Different SLC7A9 Gene Variants in the Cat. PLoS ONE, 2016, 11, e0159247.	1.1	13
17	Complex mode of inheritance in holoprosencephaly revealed by whole exome sequencing. Clinical Genetics, 2016, 89, 659-668.	1.0	36
18	dbNSFP v3.0: A One-Stop Database of Functional Predictions and Annotations for Human Nonsynonymous and Splice-Site SNVs. Human Mutation, 2016, 37, 235-241.	1.1	845
19	Understanding rare and common diseases in the context of human evolution. Genome Biology, 2016, 17, 225.	3.8	76

#	Article	IF	Citations
20	iCAGES: integrated CAncer GEnome Score for comprehensively prioritizing driver genes in personal cancer genomes. Genome Medicine, 2016, 8, 135.	3.6	45
21	Digenic mutations of human OCRL paralogs in Dentâ€™s disease type 2 associated with Chiari I malformation. Human Genome Variation, 2016, 3, 16042.	0.4	8
22	Analyses of more than 60,000 exomes questions the role of numerous genes previously associated with dilated cardiomyopathy. Molecular Genetics & Genomic Medicine, 2016, 4, 617-623.	0.6	29
23	Whole-exome identifies RXRG and TH germline variants in familial isolated prolactinoma. Cancer Genetics, 2016, 209, 251-257.	0.2	7
24	Evaluating the impact of missenses mutations in CYP2D6*7 and CYP2D6*14A: does it compromise tamoxifen metabolism?. Pharmacogenomics, 2016, 17, 561-570.	0.6	13
25	A Broad Overview of Computational Methods for Predicting the Pathophysiological Effects of Non-synonymous Variants. Methods in Molecular Biology, 2016, 1415, 423-440.	0.4	7
26	G23D: Online tool for mapping and visualization of genomic variants on 3D protein structures. BMC Genomics, 2016, 17, 681.	1.2	18
27	How to Identify Pathogenic Mutations among All Those Variations: Variant Annotation and Filtration in the Genome Sequencing Era. Human Mutation, 2016, 37, 1272-1282.	1.1	28
28	Advantages and pitfalls of an extended gene panel for investigating complex neurometabolic phenotypes. Brain, 2016, 139, 2844-2854.	3.7	35
29	Bioinformatics and Orphan Diseases. Translational Bioinformatics, 2016, , 313-338.	0.0	0
31	The role of next generation sequencing in understanding male and female sexual development: clinical implications. Expert Review of Endocrinology and Metabolism, 2016, 11, 433-443.	1.2	3
33	REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. American Journal of Human Genetics, 2016, 99, 877-885.	2.6	1,555
34	Biallelic Mutations in TBCD , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 962-973.	2.6	66
35	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. American Journal of Human Genetics, 2016, 99, 974-983.	2.6	49
36	A Cost-Effective Mutation Screening Strategy for Inherited Retinal Dystrophies. Ophthalmic Research, 2016, 56, 123-131.	1.0	2
37	Implementation of next-generation sequencing for molecular diagnosis of hereditary breast and ovarian cancer highlights its genetic heterogeneity. Breast Cancer Research and Treatment, 2016, 159, 245-256.	1.1	23
38	Deep Genetic Connection Between Cancer and Developmental Disorders. Human Mutation, 2016, 37, 1042-1050.	1.1	24
39	Mutational Spectrum in Holoprosencephaly Shows That FGF is a New Major Signaling Pathway. Human Mutation, 2016, 37, 1329-1339.	1.1	56

#	Article	IF	Citations
40	Multiple rare variants in high-risk pancreatic cancer-related genes may increase risk for pancreatic cancer in a subset of patients with and without germline CDKN2A mutations. Human Genetics, 2016, 135, 1241-1249.	1.8	24
41	Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients. Molecular Genetics & Genomic Medicine, 2016, 4, 568-580.	0.6	83
42	IMHOTEPâ€"a composite score integrating popular tools for predicting the functional consequences of non-synonymous sequence variants. Nucleic Acids Research, 2017, 45, gkw886.	6.5	10
43	Panel-Based Population Next-Generation Sequencing for Inherited Retinal Degenerations. Scientific Reports, 2016, 6, 33248.	1.6	49
44	Six Germline Genetic Variations Impair the Translesion Synthesis Activity of Human DNA Polymerase Î¹. Chemical Research in Toxicology, 2016, 29, 1741-1754.	1.7	9
45	A Biobank of Breast Cancer Explants with Preserved Intra-tumor Heterogeneity to Screen Anticancer Compounds. Cell, 2016, 167, 260-274.e22.	13.5	376
46	Combined variants in factor VIII and prostaglandin synthase-1 amplify hemorrhage severity across three generations of descendants. Journal of Thrombosis and Haemostasis, 2016, 14, 2230-2240.	1.9	6
47	ACOX2 deficiency: A disorder of bile acid synthesis with transaminase elevation, liver fibrosis, ataxia, and cognitive impairment. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 11289-11293.	3.3	75
48	InteractoMIX: a suite of computational tools to exploit interactomes in biological and clinical research. Biochemical Society Transactions, 2016, 44, 917-924.	1.6	3
49	Protein function in precision medicine: deep understanding with machine learning. FEBS Letters, 2016, 590, 2327-2341.	1.3	43
50	The population genomics of rhesus macaques (Macaca mulatta) based on whole-genome sequences. Genome Research, 2016, 26, 1651-1662.	2.4	101
51	Genetics of Combined Pituitary Hormone Deficiency: Roadmap into the Genome Era. Endocrine Reviews, 2016, 37, 636-675.	8.9	147
52	M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity. Nature Genetics, 2016, 48, 1581-1586.	9.4	654
53	Big Data Analytics in Genomics. , 2016, , .		7
54	Challenges and disparities in the application of personalized genomic medicine to populations with African ancestry. Nature Communications, 2016, 7, 12521.	5.8	68
55	Long-read sequencing and de novo assembly of a Chinese genome. Nature Communications, 2016, 7, 12065.	5.8	242
56	Testing the Complex Child: CGH Array, WES, Clinical Exome, WGS. Current Pediatrics Reports, 2016, 4, 155-163.	1.7	0
57	OncoMiner: A Pipeline for Bioinformatics Analysis of Exonic Sequence Variants in Cancer. , 2016, , 373-396.		2

#	Article	IF	Citations
58	Incorporating ENCODE information into association analysis of whole genome sequencing data. BMC Proceedings, 2016, 10, 257-261.	1.8	9
59	Inferring Crohnâ€™s disease association from exome sequences by integrating biological knowledge. BMC Medical Genomics, 2016, 9, 35.	0.7	8
60	The Rise and Rise of Exome Sequencing. Public Health Genomics, 2016, 19, 315-324.	0.6	15
61	Tools for Predicting the Functional Impact of Nonsynonymous Genetic Variation. Genetics, 2016, 203, 635-647.	1.2	84
62	Predicting regulatory variants with composite statistic. Bioinformatics, 2016, 32, 2729-2736.	1.8	40
63	The two-pore domain potassium channel, TWIK-1, has a role in the regulation of heart rate and atrial size. Journal of Molecular and Cellular Cardiology, 2016, 97, 24-35.	0.9	28
64	Multilevel biological characterization of exomic variants at the protein level significantly improves the identification of their deleterious effects. Bioinformatics, 2016, 32, 1797-1804.	1.8	32
65	RET and EDNRB mutation screening in patients with Hirschsprung disease: Functional studies and its implications for genetic counseling. European Journal of Human Genetics, 2016, 24, 823-829.	1.4	20
66	Utility of whole-genome sequencing for detection of newborn screening disorders in a population cohort of 1,696 neonates. Genetics in Medicine, 2016, 18, 221-230.	1.1	101
67	Polygenic inheritance of cryptorchidism susceptibility in the LE/orl rat. Molecular Human Reproduction, 2016, 22, 18-34.	1.3	9
68	RNF43 germline and somatic mutation in serrated neoplasia pathway and its association with BRAF mutation. Gut, 2017, 66, 1645-1656.	6.1	157
69	Intraflagellar transport 88 (IFT88) is crucial for craniofacial development in mice and is a candidate gene for human cleft lip and palate. Human Molecular Genetics, 2017, 26, ddx002.	1.4	41
71	Structural, Functional, and Clinical Characterization of a NovelPTPN11Mutation Cluster Underlying Noonan Syndrome. Human Mutation, 2017, 38, 451-459.	1.1	39
72	Practical Approaches for Whole-Genome Sequence Analysis of Heart- and Blood-Related Traits. American Journal of Human Genetics, 2017, 100, 205-215.	2.6	50
73	Predicting Severity of Disease-Causing Variants. Human Mutation, 2017, 38, 357-364.	1.1	33
74	InterVar: Clinical Interpretation of Genetic Variants by the 2015 ACMG-AMP Guidelines. American Journal of Human Genetics, 2017, 100, 267-280.	2.6	717
75	Remodelling of myocardial intercalated disc protein connexin 43 causes increased susceptibility to malignant arrhythmias in ARVC/D patients. Forensic Science International, 2017, 275, 14-22.	1.3	15
76	Germline Mutations in CDH23, Encoding Cadherin-Related 23, Are Associated with Both Familial and Sporadic Pituitary Adenomas. American Journal of Human Genetics, 2017, 100, 817-823.	2.6	57

#	Article	IF	Citations
77	Mutational landscape of Bâ€cell postâ€transplant lymphoproliferative disorders. British Journal of Haematology, 2017, 178, 48-56.	1.2	58
78	Are Double Mutations Double Trouble?. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	3
79	Atomic envoy enables molecular control. Nature, 2017, 545, 164-165.	13.7	0
80	An ion-transport enzyme that rocks. Nature, 2017, 545, 162-164.	13.7	5
81	A novel molecular diagnostics platform for somatic and germline precision oncology. Molecular Genetics & Genomic Medicine, 2017, 5, 336-359.	0.6	12
82	The Effect of an Extreme and Prolonged Population Bottleneck on Patterns of Deleterious Variation: Insights from the Greenlandic Inuit. Genetics, 2017, 205, 787-801.	1.2	54
83	Carrier frequency of Wilsonâ€™s disease in the Korean population: a DNA-based approach. Journal of Human Genetics, 2017, 62, 815-818.	1.1	40
84	Missense variant pathogenicity predictors generalize well across a range of functionâ€specific prediction challenges. Human Mutation, 2017, 38, 1092-1108.	1.1	39
85	Identification of 8 Novel Mutations in Nephrogenesis-Related Genes in Chinese Han Patients with Unilateral Renal Agenesis. American Journal of Nephrology, 2017, 46, 55-63.	1.4	11
86	The evolving genetic risk for sporadic ALS. Neurology, 2017, 89, 226-233.	1.5	41
87	Differential analysis of mutations in the Jewish population and their implications for diseases. Genetical Research, 2017, 99, e3.	0.3	7
88	PMut: a web-based tool for the annotation of pathological variants on proteins, 2017 update. Nucleic Acids Research, 2017, 45, W222-W228.	6.5	184
89	Is Rosai-Dorfman disease a reactve process? Detection of a MAP2K1 L115V mutation in a case of Rosai-Dorfman disease. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2017, 471, 545-547.	1.4	39
90	TLR3 Mutations in Adult Patients With Herpes Simplex Virus and Varicella-Zoster Virus Encephalitis. Journal of Infectious Diseases, 2017, 215, 1430-1434.	1.9	53
91	Computational predictors fail to identify amino acid substitution effects at rheostat positions. Scientific Reports, 2017, 7, 41329.	1.6	47
92	A molecular analysis of the <scp>GBA</scp> gene in Caucasian South Africans with Parkinson's disease. Molecular Genetics & Genomic Medicine, 2017, 5, 147-156.	0.6	22
93	In silico analysis of nonsynonymous single nucleotide polymorphisms of the human adiponectin receptor 2 (ADIPOR2) gene. Computational Biology and Chemistry, 2017, 68, 175-185.	1.1	20
94	Genetics and Genomics of Congenital Heart Disease. Circulation Research, 2017, 120, 923-940.	2.0	349

#	Article	IF	Citations
95	Mutations of CREBBP and SOCS1 are independent prognostic factors in diffuse large B cell lymphoma: mutational analysis of the SAKK 38/07 prospective clinical trial cohort. Journal of Hematology and Oncology, 2017, 10, 70.	6.9	66
96	Genome-wide enrichment of damaging de novo variants in patients with isolated and complex congenital diaphragmatic hernia. Human Genetics, 2017, 136, 679-691.	1.8	53
97	PERCH: A Unified Framework for Disease Gene Prioritization. Human Mutation, 2017, 38, 243-251.	1.1	119
98	Spectrum of germline mutations in smokers and non-smokers in Brazilian non-small-cell lung cancer (NSCLC) patients. Carcinogenesis, 2017, 38, 1112-1118.	1.3	21
99	DOMINO: Using Machine Learning to Predict Genes Associated with Dominant Disorders. American Journal of Human Genetics, 2017, 101, 623-629.	2.6	90
100	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	9.4	624
101	Rare germline variants in known melanoma susceptibility genes in familial melanoma. Human Molecular Genetics, 2017, 26, 4886-4895.	1.4	37
102	Characterisation of the novel deleterious RAD51C p.Arg312Trp variant and prioritisation criteria for functional analysis of RAD51C missense changes. British Journal of Cancer, 2017, 117, 1048-1062.	2.9	12
103	Whole Exome Sequencing Identified a Novel IGFBP6 Variant in a Disc Degeneration Pedigree. Genetic Testing and Molecular Biomarkers, 2017, 21, 580-585.	0.3	6
104	Whole exome sequencing and <scp>DNA</scp> methylation analysis in a clinical amyotrophic lateral sclerosis cohort. Molecular Genetics & Genomic Medicine, 2017, 5, 418-428.	0.6	14
105	Iterative Sequencing and Variant Screening (ISVS) as a novel pathogenic mutations search strategy - application for TMPRSS3 mutations screen. Scientific Reports, 2017, 7, 2543.	1.6	10
106	Predicting the functional consequences of non-synonymous single nucleotide polymorphisms in IL8 gene. Scientific Reports, 2017, 7, 6525.	1.6	75
107	Nâ€ethylâ€Nâ€nitrosoureaâ€“Induced Adaptor Protein 2 Sigma Subunit 1 (Ap2s1) Mutations Establish Ap2s1 Lossâ€ofâ€Function Mice. JBMR Plus, 2017, 1, 3-15.	1.3	16
108	De novo mutations in inhibitors of Wnt, BMP, and Ras/ERK signaling pathways in non-syndromic midline craniosynostosis. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E7341-E7347.	3.3	73
109	Human CRMP4 mutation and disrupted Crmp4 expression in mice are associated with ASD characteristics and sexual dimorphism. Scientific Reports, 2017, 7, 16812.	1.6	18
110	Germline Loss-of-Function Mutations in EPHB4 Cause a Second Form of Capillary Malformation-Arteriovenous Malformation (CM-AVM2) Deregulating RAS-MAPK Signaling. Circulation, 2017, 136, 1037-1048.	1.6	204
111	Exome sequencing identifies targets in the treatment-resistant ophthalmoplegic subphenotype of myasthenia gravis. Neuromuscular Disorders, 2017, 27, 816-825.	0.3	12
112	GTB â€“ an online genome tolerance browser. BMC Bioinformatics, 2017, 18, 20.	1.2	3

#	Article	IF	Citations
113	Not only dominant, not only optic atrophy: expanding the clinical spectrum associated with OPA1 mutations. Orphanet Journal of Rare Diseases, 2017, 12, 89.	1.2	39
114	A pipeline combining multiple strategies for prioritizing heterozygous variants for the identification of candidate genes in exome datasets. Human Genomics, 2017, 11, 11.	1.4	20
115	DEOGEN2: prediction and interactive visualization of single amino acid variant deleteriousness in human proteins. Nucleic Acids Research, 2017, 45, W201-W206.	6.5	114
116	A Cell Type-Specific Expression Signature Predicts Haploinsufficient Autism-Susceptibility Genes. Human Mutation, 2017, 38, 204-215.	1.1	38
117	In Silico Prediction of Deleteriousness for Nonsynonymous and Splice-Altering Single Nucleotide Variants in the Human Genome. Methods in Molecular Biology, 2017, 1498, 191-197.	0.4	17
118	Investigating the Molecular Mechanisms Behind Uncharacterized Cysteine Losses from Prediction of Their Oxidation State. Human Mutation, 2017, 38, 86-94.	1.1	4
119	A deep learning based scoring system for prioritizing susceptibility variants for mental disorders. , 2017, , .		2
120	Identification of potential genetic causal variants for rheumatoid arthritis by whole-exome sequencing. Oncotarget, 2017, 8, 111119-111129.	0.8	20
121	Spatial distribution of disease-associated variants in three-dimensional structures of protein complexes. Oncogenesis, 2017, 6, e380-e380.	2.1	20
123	High-throughput sequencing of the B-cell receptor in African Burkitt lymphoma reveals clues to pathogenesis. Blood Advances, 2017, 1, 535-544.	2.5	27
124	Target 5000: Target Capture Sequencing for Inherited Retinal Degenerations. Genes, 2017, 8, 304.	1.0	46
125	NOTCH1 Mutations in Aortic Stenosis: Association with Osteoprotegerin/RANK/RANKL. BioMed Research International, 2017, 2017, 1-10.	0.9	20
126	Benchmarking distributed data warehouse solutions for storing genomic variant information. Database: the Journal of Biological Databases and Curation, 2017, 2017, .	1.4	8
127	Whole-exome sequencing in amyotrophic lateral sclerosis suggests NEK1 is a risk gene in Chinese. Genome Medicine, 2017, 9, 97.	3.6	23
128	Evaluation of in silico algorithms for use with ACMG/AMP clinical variant interpretation guidelines. Genome Biology, 2017, 18, 225.	3.8	185
129	Variant Ranker: a web-tool to rank genomic data according to functional significance. BMC Bioinformatics, 2017, 18, 341.	1.2	21
130	A practical guide to filtering and prioritizing genetic variants. BioTechniques, 2017, 62, 18-30.	0.8	57
131	Genetic profiling of a rare condition: co-occurrence of albinism and multiple primary melanoma in a caucasian family. Oncotarget, 2017, 8, 29751-29759.	0.8	8

#	Article	IF	Citations
132	Isolated polycystic liver disease genes define effectors of polycystin-1 function. Journal of Clinical Investigation, 2017, 127, 1772-1785.	3.9	137
133	Genetic Variation in Genes Underlying Diverse Dementias May Explain a Small Proportion of Cases in the Alzheimerâ€™s Disease Sequencing Project. Dementia and Geriatric Cognitive Disorders, 2018, 45, 1-17.	0.7	22
134	Uncovering the heterogeneous genetic variations in two insulin-expressing tumors in a patient with MEN1. Oncology Letters, 2018, 15, 7123-7131.	0.8	0
135	Novel valosin-containing protein mutations associated with multisystem proteinopathy. Neuromuscular Disorders, 2018, 28, 491-501.	0.3	20
136	VarCards: an integrated genetic and clinical database for coding variants in the human genome. Nucleic Acids Research, 2018, 46, D1039-D1048.	6.5	148
137	A Review of Matched-pairs Feature Selection Methods for Gene Expression Data Analysis. Computational and Structural Biotechnology Journal, 2018, 16, 88-97.	1.9	49
138	KoVariome: Korean National Standard Reference Variome database of whole genomes with comprehensive SNV, indel, CNV, and SV analyses. Scientific Reports, 2018, 8, 5677.	1.6	39
139	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. Circulation Genomic and Precision Medicine, 2018, 11, e001887.	1.6	104
140	Inferring the effect of genomic variation in the new era of genomics. Human Mutation, 2018, 39, 756-773.	1.1	24
141	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. Neuron, 2018, 97, 488-493.	3.8	265
142	A phenotype centric benchmark of variant prioritisation tools. Npj Genomic Medicine, 2018, 3, 5.	1.7	39
143	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	2.6	138
144	Bioinformatics analysis of non-synonymous variants in the KLF genes related to cardiac diseases. Gene, 2018, 650, 68-76.	1.0	5
145	CLCN2 chloride channel mutations in familial hyperaldosteronism type II. Nature Genetics, 2018, 50, 349-354.	9.4	188
146	Tools for protein science. Protein Science, 2018, 27, 6-9.	3.1	2
147	Punctuated evolution of canonical genomic aberrations in uveal melanoma. Nature Communications, 2018, 9, 116.	5.8	144
148	Rare loss of function mutations in N-methyl-d-aspartate glutamate receptors and their contributions to schizophrenia susceptibility. Translational Psychiatry, 2018, 8, 12.	2.4	41
149	DNA methylation-based reclassification of olfactory neuroblastoma. Acta Neuropathologica, 2018, 136, 255-271.	3.9	59

#	Article	IF	Citations
150	Linked homozygous BMPR1B and PDHA2 variants in a consanguineous family with complex digit malformation and male infertility. European Journal of Human Genetics, 2018, 26, 876-885.	1.4	10
151	Mutation screening of 10 cancer susceptibility genes in unselected breast cancer patients. Clinical Genetics, 2018, 93, 41-51.	1.0	15
152	Comprehensive BRCA mutation analysis in the Greek population. Experience from a single clinical diagnostic center. Cancer Genetics, 2018, 220, 1-12.	0.2	11
153	Full-gene haplotypes refine CYP2D6 metabolizer phenotype inferences. International Journal of Legal Medicine, 2018, 132, 1007-1024.	1.2	11
154	Insights From Molecular Characterization of Adult Patients of Families With Multigenerational Diabetes. Diabetes, 2018, 67, 137-145.	0.3	23
155	Reâ€evaluatingÂpathogenicity of variants associated with the long QT syndrome. Journal of Cardiovascular Electrophysiology, 2018, 29, 98-104.	0.8	9
156	Biallelic mutations in <scp>DYNC2LI1</scp> are a rare cause of Ellisâ€van Creveld syndrome. Clinical Genetics, 2018, 93, 632-639.	1.0	23
157	Wholeâ€exome sequencing of sickle cell disease patients with hyperhemolysis syndrome suggests a role for rare variation in disease predisposition. Transfusion, 2018, 58, 726-735.	0.8	17
158	Predicted activity of UGT2B7, ABCB1, OPRM1, and COMT using full-gene haplotypes and their association with the CYP2D6-inferred metabolizer phenotype. Forensic Science International: Genetics, 2018, 33, 48-58.	1.6	4
159	MDS1 and EVI1 complex locus (MECOM): a novel candidate gene for hereditary hematological malignancies. Haematologica, 2018, 103, e55-e58.	1.7	41
160	Genetic Costs of Domestication and Improvement. Journal of Heredity, 2018, 109, 103-116.	1.0	149
161	iMEGES: integrated mental-disorder GEnome score by deep neural network for prioritizing the susceptibility genes for mental disorders in personal genomes. BMC Bioinformatics, 2018, 19, 501.	1.2	10
162	Representativeness of variation benchmark datasets. BMC Bioinformatics, 2018, 19, 461.	1.2	18
163	Whole genome sequencing and 6-year follow-up of a mother and daughter with frontometaphyseal dysplasia associated with keratitis, xerosis, poikiloderma, and acro-osteolysis. Medicine (United) Tj ETQq1 1 0.784314 rgBT /Overlock	1.4	9
164	DNAJC3 mutation in Thai familial type 2 diabetes mellitus. International Journal of Molecular Medicine, 2018, 42, 1064-1073.	1.8	6
165	De novo variants in congenital diaphragmatic hernia identify MYRF as a new syndrome and reveal genetic overlaps with other developmental disorders. PLoS Genetics, 2018, 14, e1007822.	1.5	79
166	Large-scale in-silico statistical mutagenesis analysis sheds light on the deleteriousness landscape of the human proteome. Scientific Reports, 2018, 8, 16980.	1.6	7
167	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

#	Article	IF	Citations
168	KIF5A and ALS2 Variants in a Family With Hereditary Spastic Paraplegia and Amyotrophic Lateral Sclerosis. Frontiers in Neurology, 2018, 9, 1078.	1.1	12
169	Use of Germline Genetic Variability for Prediction of Chemoresistance and Prognosis of Breast Cancer Patients. Cancers, 2018, 10, 511.	1.7	14
170	Challenging popular tools for the annotation of genetic variations with a real case, pathogenic mutations of lysosomal alpha-galactosidase. BMC Bioinformatics, 2018, 19, 433.	1.2	8
171	Multimodal imaging in a pedigree of X-linked Retinoschisis with a novel RS1 variant. BMC Medical Genetics, 2018, 19, 195.	2.1	5
172	Computational Methods for the Pharmacogenetic Interpretation of Next Generation Sequencing Data. Frontiers in Pharmacology, 2018, 9, 1437.	1.6	62
173	In vitrofunctional characterization of the novelDHHmutations p.(Asn337Lysfs*24) and p.(Glu212Lys) associated with gonadal dysgenesis. Human Mutation, 2018, 39, 2097-2109.	1.1	12
174	Exhaustive non-synonymous variants functionality prediction enables high resolution characterization of the neurofibromin architecture. EBioMedicine, 2018, 36, 508-516.	2.7	1
175	9p24 triplication in syndromic hydrocephalus with diffuse villous hyperplasia of the choroid plexus. Journal of Physical Education and Sports Management, 2018, 4, a003145.	0.5	8
176	Machine Learning Classification and Structureâ€“Functional Analysis of Cancer Mutations Reveal Unique Dynamic and Network Signatures of Driver Sites in Oncogenes and Tumor Suppressor Genes. Journal of Chemical Information and Modeling, 2018, 58, 2131-2150.	2.5	20
177	AutismKB 2.0: a knowledgebase for the genetic evidence of autism spectrum disorder. Database: the Journal of Biological Databases and Curation, 2018, 2018, .	1.4	37
178	BRCA-analyzer: Automatic workflow for processing NGS reads of BRCA1 and BRCA2 genes. Computational Biology and Chemistry, 2018, 77, 297-306.	1.1	10
179	NRG1 variant effects in patients with Hirschsprung disease. BMC Pediatrics, 2018, 18, 292.	0.7	12
180	Characterization of genetic alterations in brain metastases from nonâ€small cell lung cancer. FEBS Open Bio, 2018, 8, 1544-1552.	1.0	28
181	Comparative Genomics Approaches Accurately Predict Deleterious Variants in Plants. G3: Genes, Genomes, Genetics, 2018, 8, 3321-3329.	0.8	36
182	ClinPred: Prediction Tool to Identify Disease-Relevant Nonsynonymous Single-Nucleotide Variants. American Journal of Human Genetics, 2018, 103, 474-483.	2.6	149
183	Case Report: Identification of an HNF1B p.Arg527Gln mutation in a Maltese patient with atypical early onset diabetes and diabetic nephropathy. BMC Endocrine Disorders, 2018, 18, 28.	0.9	11
184	Cancer genetics, precision prevention and a call to action. Nature Genetics, 2018, 50, 1212-1218.	9.4	94
185	A novel association of campomelic dysplasia and hydrocephalus with an unbalanced chromosomal translocation upstream of SOX9. Journal of Physical Education and Sports Management, 2018, 4, a002766.	0.5	8

#	Article	IF	Citations
186	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884.	1.1	23
187	A single-center study on 140 patients with cerebral cavernous malformations: 28 new pathogenic variants and functional characterization of a PDCD10 large deletion. Human Mutation, 2018, 39, 1885-1900.	1.1	16
188	Functional Assays Are Essential for Interpretation of Missense Variants Associated with Variable Expressivity. American Journal of Human Genetics, 2018, 102, 1062-1077.	2.6	69
189	Wholeâ€exome sequencing for variant discovery in blepharospasm. Molecular Genetics & Genomic Medicine, 2018, 6, 601-626.	0.6	20
190	Computational analysis of non-synonymous SNPs in bovine Mx1 gene. Gene Reports, 2018, 11, 294-298.	0.4	0
191	De novo mutations in FLNC leading to early-onset restrictive cardiomyopathy and congenital myopathy. Human Mutation, 2018, 39, 1161-1172.	1.1	49
192	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. Neuron, 2018, 99, 302-314.e4.	3.8	112
193	UGT1A1 Genetic Variations and a Haplotype Associated with Neonatal Hyperbilirubinemia in Indonesian Population. BioMed Research International, 2018, 2018, 1-11.	0.9	8
194	Protein phenotype diagnosis of autosomal dominant calmodulin mutations causing irregular heart rhythms. Journal of Cellular Biochemistry, 2018, 119, 8233-8248.	1.2	14
195	Somatic mutations in specific and connected subpathways are associated with short neuroblastoma patientsâ€™ survival and indicate proteins targetable at onset of disease. International Journal of Cancer, 2018, 143, 2525-2536.	2.3	27
196	Exome sequencing identifies novel dysferlin mutation in a family with pauci-symptomatic heterozygous carriers. BMC Medical Genetics, 2018, 19, 95.	2.1	2
197	Targeted next-generation sequencing as a comprehensive test for Mendelian diseases: a cohort diagnostic study. Scientific Reports, 2018, 8, 11646.	1.6	17
198	Predicting the clinical impact of human mutation with deep neural networks. Nature Genetics, 2018, 50, 1161-1170.	9.4	288
199	Performance evaluation of pathogenicity-computation methods for missense variants. Nucleic Acids Research, 2018, 46, 7793-7804.	6.5	168
200	The Analysis of Variants in the General Population Reveals That PMM2 Is Extremely Tolerant to Missense Mutations and That Diagnosis of PMM2-CDG Can Benefit from the Identification of Modifiers. International Journal of Molecular Sciences, 2018, 19, 2218.	1.8	32
201	Whole-Exome Sequencing in Searching for New Variants Associated With the Development of Parkinsonâ€™s Disease. Frontiers in Aging Neuroscience, 2018, 10, 136.	1.7	17
202	Systematic discovery of germline cancer predisposition genes through the identification of somatic second hits. Nature Communications, 2018, 9, 2601.	5.8	47
203	Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. American Journal of Human Genetics, 2018, 103, 100-114.	2.6	34

#	Article	IF	Citations
204	Comprehensive genomic diagnosis of non-syndromic and syndromic hereditary hearing loss in Spanish patients. BMC Medical Genomics, 2018, 11, 58.	0.7	65
205	Exome sequencing of 85 Williamsâ€“Beuren syndrome cases rules out coding variation as a major contributor to remaining variance in social behavior. Molecular Genetics & Genomic Medicine, 2018, 6, 749-765.	0.6	9
206	Molecular Deconvolution Platform to Establish Disease Mechanisms by Surveying GPCR Signaling. Cell Reports, 2018, 24, 557-568.e5.	2.9	12
207	Cardiomyopathy and Preeclampsia. Circulation, 2018, 138, 2359-2366.	1.6	60
208	Frequency, impact and a preclinical study of novel ERBB gene family mutations in HER2-positive breast cancer. Therapeutic Advances in Medical Oncology, 2018, 10, 175883591877829.	1.4	11
209	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. Genome Medicine, 2018, 10, 56.	3.6	112
210	Complex phenotype of dyskeratosis congenita and mood dysregulation with novel homozygous RTEL1 and TPH1 variants. American Journal of Medical Genetics, Part A, 2018, 176, 1432-1437.	0.7	7
211	Evidence for GALNT12 as a moderate penetrance gene for colorectal cancer. Human Mutation, 2018, 39, 1092-1101.	1.1	20
212	RheoScale: A tool to aggregate and quantify experimentally determined substitution outcomes for multiple variants at individual protein positions. Human Mutation, 2018, 39, 1814-1826.	1.1	23
213	Deep-coverage whole genome sequences and blood lipids among 16,324 individuals. Nature Communications, 2018, 9, 3391.	5.8	140
214	Mouse models in the era of large human tumour sequencing studies. Open Biology, 2018, 8, .	1.5	7
215	Breast cancer patients suggestive of Li-Fraumeni syndrome: mutational spectrum, candidate genes, and unexplained heredity. Breast Cancer Research, 2018, 20, 87.	2.2	9
216	De novo MYH9 mutation in congenital scalp hemangioma. Journal of Physical Education and Sports Management, 2018, 4, a002998.	0.5	9
217	A gene-centric strategy for identifying disease-causing rare variants in dilated cardiomyopathy. Genetics in Medicine, 2019, 21, 133-143.	1.1	25
218	Evaluation of computational techniques for predicting non-synonymous single nucleotide variants pathogenicity. Genomics, 2019, 111, 869-882.	1.3	36
219	Implementing precision cancer medicine in the genomic era. Seminars in Cancer Biology, 2019, 55, 16-27.	4.3	24
220	A founder homozygous DSG2 variant in East Asia results in ARVC with full penetrance and heart failure phenotype. International Journal of Cardiology, 2019, 274, 263-270.	0.8	32
221	Predicting Non-Synonymous Single Nucleotide Variants Pathogenic Effects in Human Diseases. , 2019, , 400-409.		1

#	Article	IF	Citations
222	Computational resources associating diseases with genotypes, phenotypes and exposures. Briefings in Bioinformatics, 2019, 20, 2098-2115.	3.2	27
223	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
224	ALG9 Mutation Carriers Develop Kidney and Liver Cysts. Journal of the American Society of Nephrology: JASN, 2019, 30, 2091-2102.	3.0	91
225	Whole genome sequencing and rare variant analysis in essential tremor families. PLoS ONE, 2019, 14, e0220512.	1.1	28
226	Benchmarking subcellular localization and variant tolerance predictors on membrane proteins. BMC Genomics, 2019, 20, 547.	1.2	14
227	Mutations in RHOT1 Disrupt Endoplasmic Reticulumâ€“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
228	Dissecting in silico Mutation Prediction of Variants in African Genomes: Challenges and Perspectives. Frontiers in Genetics, 2019, 10, 601.	1.1	25
229	Assessing computational predictions of the phenotypic effect of cystathionineâ€betaâ€synthase variants. Human Mutation, 2019, 40, 1530-1545.	1.1	5
230	Computational assessment of somatic and germline mutations of p16INK4a: Structural insights and implications in disease. Informatics in Medicine Unlocked, 2019, 17, 100208.	1.9	2
231	A recurrent missense mutation in the EDAR gene causes severe autosomal recessive hypohidrotic ectodermal dysplasia in two consanguineous Kashmiri families. Journal of Gene Medicine, 2019, 21, e3113.	1.4	2
232	VIPdb, a genetic Variant Impact Predictor Database. Human Mutation, 2019, 40, 1202-1214.	1.1	24
233	Quantifying gene selection in cancer through protein functional alteration bias. Nucleic Acids Research, 2019, 47, 6642-6655.	6.5	21
234	Actionable Pharmacogenetic Variation in the Slovenian Genomic Database. Frontiers in Pharmacology, 2019, 10, 240.	1.6	10
235	Characterization of the c.793-1Gâ€‰>â€‰A splicing variant in CHEK2 gene as pathogenic: a case report. BMC Medical Genetics, 2019, 20, 131.	2.1	6
236	Pharmacogenomics of statin-related myopathy: Meta-analysis of rare variants from whole-exome sequencing. PLoS ONE, 2019, 14, e0218115.	1.1	18
237	Molecular diagnostic workflow, clinical interpretation of sequence variants, and data repository procedures in 140 individuals with familial cerebral cavernous malformations. Human Mutation, 2019, 40, e24-e36.	1.1	3
238	Genetic Mutations Underlying Phenotypic Plasticity in Basosquamous Carcinoma. Journal of Investigative Dermatology, 2019, 139, 2263-2271.e5.	0.3	24
239	Recessive Inheritance of Congenital Hydrocephalus With Other Structural Brain Abnormalities Caused by Compound Heterozygous Mutations in ATP1A3. Frontiers in Cellular Neuroscience, 2019, 13, 425.	1.8	14

#	Article	IF	Citations
240	Essentials of Bioinformatics, Volume II. , 2019, , .		1
241	Variation in Actionable Pharmacogenetic Markers in Natives and Mestizos From Mexico. Frontiers in Pharmacology, 2019, 10, 1169.	1.6	15
242	Gene4Denovo: an integrated database and analytic platform for de novo mutations in humans. Nucleic Acids Research, 2020, 48, D913-D926.	6.5	41
243	De novo Mutations From Whole Exome Sequencing in Neurodevelopmental and Psychiatric Disorders: From Discovery to Application. Frontiers in Genetics, 2019, 10, 258.	1.1	49
244	The genetic landscape of the human solute carrier (SLC) transporter superfamily. Human Genetics, 2019, 138, 1359-1377.	1.8	79
245	FLNC Missense Variants in Familial Noncompaction Cardiomyopathy. Neurology International, 2019, 9, 8181.	0.2	3
246	SLC12A ion transporter mutations in sporadic and familial human congenital hydrocephalus. Molecular Genetics & Genomic Medicine, 2019, 7, e892.	0.6	22
247	Insights into pathological mutations in insulin-like growth factor I through in silico screening and molecular dynamics simulation. Journal of Molecular Modeling, 2019, 25, 276.	0.8	1
249	REVEL and BayesDel outperform other in silico meta-predictors for clinical variant classification. Scientific Reports, 2019, 9, 12752.	1.6	52
250	Pathway analysis of genomic pathology tests for prognostic cancer subtyping. Journal of Biomedical Informatics, 2019, 98, 103286.	2.5	3
251	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. Nature Genetics, 2019, 51, 1459-1474.	9.4	251
252	Intraventricular meningiomas frequently harbor NF2 mutations but lack common genetic alterations in TRAF7, AKT1, SMO, KLF4, PIK3CA, and TERT. Acta Neuropathologica Communications, 2019, 7, 140.	2.4	26
253	funtrp: identifying protein positions for variation driven functional tuning. Nucleic Acids Research, 2019, 47, e142-e142.	6.5	29
254	Quantitative approaches to variant classification increase the yield and precision of genetic testing in Mendelian diseases: the case of hypertrophic cardiomyopathy. Genome Medicine, 2019, 11, 5.	3.6	90
255	Innovative strategies for annotating the â€œrelationSNPâ€•between variants and molecular phenotypes. BioData Mining, 2019, 12, 10.	2.2	6
256	Characterization of intellectual disability and autism comorbidity through gene panel sequencing. Human Mutation, 2019, 40, 1346-1363.	1.1	54
257	Geneâ€specific features enhance interpretation of mutational impact on acid Î±â€glucosidase enzyme activity. Human Mutation, 2019, 40, 1507-1518.	1.1	8
258	Exome Sequencing in Clinical Hepatology. Hepatology, 2019, 70, 2185-2192.	3.6	19

#	Article	IF	Citations
259	Integration of Random Forest Classifiers and Deep Convolutional Neural Networks for Classification and Biomolecular Modeling of Cancer Driver Mutations. Frontiers in Molecular Biosciences, 2019, 6, 44.	1.6	51
260	Increased diagnostic and new genes identification outcome using research reanalysis of singleton exome sequencing. European Journal of Human Genetics, 2019, 27, 1519-1531.	1.4	43
261	The Kipoi repository accelerates community exchange and reuse of predictive models for genomics. Nature Biotechnology, 2019, 37, 592-600.	9.4	118
262	Comprehensive Analysis of Rare Variants of 101 Autism-Linked Genes in a Hungarian Cohort of Autism Spectrum Disorder Patients. Frontiers in Genetics, 2019, 10, 434.	1.1	14
263	Assessing concordance among human, in silico predictions and functional assays on genetic variant classification. Bioinformatics, 2019, 35, 5163-5170.	1.8	4
264	A catalog of genetic loci associated with kidney function from analyses of a million individuals. Nature Genetics, 2019, 51, 957-972.	9.4	549
265	Analysis of hereditary cancer syndromes by using a panel of genes: novel and multiple pathogenic mutations. BMC Cancer, 2019, 19, 535.	1.1	77
266	Recessive Mutations in KIF12 Cause High Gammaâ€Glutamyltransferase Cholestasis. Hepatology Communications, 2019, 3, 471-477.	2.0	21
267	Metabolic reprogramming toward oxidative phosphorylation identifies a therapeutic target for mantle cell lymphoma. Science Translational Medicine, 2019, 11, .	5.8	161
268	Challenges and Considerations in Sequence Variant Interpretation for Mendelian Disorders. Annals of Laboratory Medicine, 2019, 39, 421-429.	1.2	31
269	GenePy - a score for estimating gene pathogenicity in individuals using next-generation sequencing data. BMC Bioinformatics, 2019, 20, 254.	1.2	21
270	Study of chromatin remodeling genes implicates SMARCA4 as a putative player in oncogenesis in neuroblastoma. International Journal of Cancer, 2019, 145, 2781-2791.	2.3	16
271	Visualizing flow in an intact CSF network using optical coherence tomography: implications for human congenital hydrocephalus. Scientific Reports, 2019, 9, 6196.	1.6	27
272	Functional characterization of 3D protein structures informed by human genetic diversity. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 8960-8965.	3.3	33
273	Identification of a novel ANO5 missense mutation in a Chinese family with familial florid osseous dysplasia. Journal of Human Genetics, 2019, 64, 599-607.	1.1	6
274	Comparison of PredictiveIn SilicoTools on Missense Variants inGJB2,GJB6, andGJB3Genes Associated with Autosomal Recessive Deafness 1A (DFNB1A). Scientific World Journal, The, 2019, 2019, 1-9.	0.8	26
275	Clinical utility of genomic analysis in adults with idiopathic liver disease. Journal of Hepatology, 2019, 70, 1214-1221.	1.8	47
276	Improved measures for evolutionary conservation that exploit taxonomy distances. Nature Communications, 2019, 10, 1556.	5.8	21

#	Article	IF	Citations
277	eDiVAâ€"Classification and prioritization of pathogenic variants for clinical diagnostics. Human Mutation, 2019, 40, 865-878.	1.1	19
278	PKD1 Duplicated regions limit clinical Utility of Whole Exome Sequencing for Genetic Diagnosis of Autosomal Dominant Polycystic Kidney Disease. Scientific Reports, 2019, 9, 4141.	1.6	44
279	Mutational spectrum and clinical signatures in 114 families with hereditary multiple osteochondromas: insights into molecular properties of selected exostosin variants. Human Molecular Genetics, 2019, 28, 2133-2142.	1.4	12
280	Role of protein structure in variant annotation: structural insight of mutations causing 6-pyruvoyl-tetrahydropterin synthase deficiency. Pathology, 2019, 51, 274-280.	0.3	7
281	New insights into the pathogenicity of non-synonymous variants through multi-level analysis. Scientific Reports, 2019, 9, 1667.	1.6	40
282	Comparison of different functional prediction scores using a gene-based permutation model for identifying cancer driver genes. BMC Medical Genomics, 2019, 12, 22.	0.7	12
283	How good are pathogenicity predictors in detecting benign variants?. PLoS Computational Biology, 2019, 15, e1006481.	1.5	79
284	Methods for the Analysis and Interpretation for Rare Variants Associated with Complex Traits. Current Protocols in Human Genetics, 2019, 101, e83.	3.5	11
285	SeqSQC: A Bioconductor Package for Evaluating the Sample Quality of Next-generation Sequencing Data. Genomics, Proteomics and Bioinformatics, 2019, 17, 211-218.	3.0	6
286	Mutational load in carotid body tumor. BMC Medical Genomics, 2019, 12, 39.	0.7	12
287	New Chondrosarcoma Cell Lines with Preserved Stem Cell Properties to Study the Genomic Drift During In Vitro/In Vivo Growth. Journal of Clinical Medicine, 2019, 8, 455.	1.0	18
288	The preliminary efficacy evaluation of the CTLA-4-Ig treatment against Lupus nephritis through in-silico analyses. Journal of Theoretical Biology, 2019, 471, 74-81.	0.8	5
289	Genome sequencing for rightward hemispheric language dominance. Genes, Brain and Behavior, 2019, 18, e12572.	1.1	14
290	Generalized Cytokine Increase in the Setting of a Multisystem Clinical Disorder and Carcinoid Syndrome Associated with a Novel NLRP12 Variant. Digestive Diseases and Sciences, 2019, 64, 2140-2146.	1.1	5
291	The underacknowledged PPA-ALS. Neurology, 2019, 92, e1354-e1366.	1.5	29
292	Biallelic Variants in the Nuclear Pore Complex Protein NUP93 Are Associated with Non-progressive Congenital Ataxia. Cerebellum, 2019, 18, 422-432.	1.4	10
293	Janus kinase 2 variants associated with the transformation of myeloproliferative neoplasms into acute myeloid leukemia. Cancer, 2019, 125, 1855-1866.	2.0	21
294	Comprehensive sequencing of the myocilin gene in a selected cohort of severe primary open-angle glaucoma patients. Scientific Reports, 2019, 9, 3100.	1.6	8

#	Article	IF	Citations
295	Integrative Modeling and Novel Technologies in Human Genomics. , 2019, , 155-189.		0
296	Application of Computational Biology and Artificial Intelligence Technologies in Cancer Precision Drug Discovery. BioMed Research International, 2019, 2019, 1-15.	0.9	42
297	Zazz: Variant Annotation and Exploration of Next Generation Sequencing Variants. , 2019, , .		0
298	Case report: Novel GJB2 variant c.113T>C associated with autosomal recessive non-syndromic hearing loss (ARNSHL) in a Han family. Medicine (United States), 2019, 98, e18253.	0.4	1
299	Re-analysis of whole-exome sequencing data uncovers novel diagnostic variants and improves molecular diagnostic yields for sudden death and idiopathic diseases. Genome Medicine, 2019, 11, 83.	3.6	54
300	Personalised analytics for rare disease diagnostics. Nature Communications, 2019, 10, 5274.	5.8	15
301	VarSight: prioritizing clinically reported variants with binary classification algorithms. BMC Bioinformatics, 2019, 20, 496.	1.2	14
302	Future Preventive Gene Therapy of Polygenic Diseases from a Population Genetics Perspective. International Journal of Molecular Sciences, 2019, 20, 5013.	1.8	3
303	VPOT: A Customizable Variant Prioritization Ordering Tool for Annotated Variants. Genomics, Proteomics and Bioinformatics, 2019, 17, 540-545.	3.0	10
304	Computational Analysis of nsSNPs of ADA Gene in Severe Combined Immunodeficiency Using Molecular Modeling and Dynamics Simulation. Journal of Immunology Research, 2019, 2019, 1-14.	0.9	11
305	ATG7 and ATG9A loss-of-function variants trigger autophagy impairment and ovarian failure. Genetics in Medicine, 2019, 21, 930-938.	1.1	55
306	Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation. Neuron, 2019, 101, 429-443.e4.	3.8	56
307	Widening the landscape of heritable pulmonary hypertension mutations in paediatric and adult cases. European Respiratory Journal, 2019, 53, 1801371.	3.1	72
308	Trans-ethnic kidney function association study reveals putative causal genes and effects on kidney-specific disease aetiologies. Nature Communications, 2019, 10, 29.	5.8	113
309	The Burden of Candidate Pathogenic Variants for Kidney and Genitourinary Disorders Emerging From Exome Sequencing. Annals of Internal Medicine, 2019, 170, 11.	2.0	60
310	Recessive Rare Variants in Deoxyhypusine Synthase, an Enzyme Involved in the Synthesis of Hypusine, Are Associated with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 287-298.	2.6	38
311	Evidence for an autosomal recessive pattern of inheritance in Keratitis-ichthyosis-deafness (KID) syndrome: Exome sequencing reveals a novel homozygous GJB2 mutation. Meta Gene, 2019, 19, 15-22.	0.3	1
312	Characterization of Tumor-Suppressor Gene Inactivation Events in 33 Cancer Types. Cell Reports, 2019, 26, 496-506.e3.	2.9	21

#	Article	IF	Citations
313	Detecting genetic modifiers of spondyloepimetaphyseal dysplasia with joint laxity in the Caucasian Afrikaner community. Human Molecular Genetics, 2019, 28, 1053-1063.	1.4	1
314	Exome sequencing of sporadic childhoodâ€onset schizophrenia suggests the contribution of Xâ€linked genes in males. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 335-340.	1.1	8
315	A study in a Polish ataxia cohort indicates genetic heterogeneity and points to MTCL1 as a novel candidate gene. Clinical Genetics, 2019, 95, 415-419.	1.0	8
316	Ensembl 2019. Nucleic Acids Research, 2019, 47, D745-D751.	6.5	879
317	A review study: Computational techniques for expecting the impact of non-synonymous single nucleotide variants in human diseases. Gene, 2019, 680, 20-33.	1.0	47
318	Leveraging the power of new molecular technologies in the clinical setting requires unprecedented awareness of limitations and drawbacks: experience of one diagnostic laboratory. Journal of Medical Genetics, 2019, 56, 408-412.	1.5	3
319	An optimized prediction framework to assess the functional impact of pharmacogenetic variants. Pharmacogenomics Journal, 2019, 19, 115-126.	0.9	109
320	Frequently used bioinformatics tools overestimate the damaging effect of allelic variants. Genes and Immunity, 2019, 20, 10-22.	2.2	12
321	Diagnostic interpretation of genetic studies in patients with primary immunodeficiency diseases: AÂworking group report of the Primary Immunodeficiency Diseases Committee of the American Academy of Allergy, Asthma & Immunology. Journal of Allergy and Clinical Immunology, 2020, 145, 46-69.	1.5	54
322	Mutations in MTHFR and POLG impaired activity of the mitochondrial respiratory chain in 46-year-old twins with spastic paraparesis. Journal of Human Genetics, 2020, 65, 91-98.	1.1	5
323	Next generation sequencing panel in undifferentiated autoinflammatory diseases identifies patients with colchicine-responder recurrent fevers. Rheumatology, 2020, 59, 344-360.	0.9	36
324	Predicting novel disease mutations in the cardiac sodium channel. Biochemical and Biophysical Research Communications, 2020, 521, 603-611.	1.0	8
325	New Technologies in Pre- and Postnatal Diagnosis. , 2020, , 941-969.		0
326	The CYSMA web server: An example of integrative tool for in silico analysis of missense variants identified in Mendelian disorders. Human Mutation, 2020, 41, 375-386.	1.1	6
327	Prediction of impacts of mutations on protein structure and interactions: SDM, a statistical approach, and mCSM, using machine learning. Protein Science, 2020, 29, 247-257.	3.1	58
328	A contiguous microdeletion syndrome at Xp23.13 with non-obstructive azoospermia and congenital cataracts. Journal of Assisted Reproduction and Genetics, 2020, 37, 471-475.	1.2	8
329	Assessing genomic diversity and signatures of selection in Original Braunvieh cattle using whole-genome sequencing data. BMC Genomics, 2020, 21, 27.	1.2	47
330	Pathogenicity Reclassification of RPE65 Missense Variants Related to Leber Congenital Amaurosis and Early-Onset Retinal Dystrophy. Genes, 2020, 11, 24.	1.0	14

#	Article	IF	Citations
331	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. American Journal of Human Genetics, 2020, 106, 26-40.	2.6	42
332	A Germline Mutation in the C2 Domain of PLCÎ³2 Associated with Gain-of-Function Expands the Phenotype for PLCG2-Related Diseases. Journal of Clinical Immunology, 2020, 40, 267-276.	2.0	31
333	Characterization of a TP53 Somatic Variant of Unknown Function From an Ovarian Cancer Patient Using Organoid Culture and Computational Modeling. Clinical Obstetrics and Gynecology, 2020, 63, 109-119.	0.6	7
334	Germline MUTYH Mutation in a Pediatric Cancer Survivor Developing a Secondary Malignancy. Journal of Pediatric Hematology/Oncology, 2020, 42, e647-e654.	0.3	2
335	InMeRF: prediction of pathogenicity of missense variants by individual modeling for each amino acid substitution. NAR Genomics and Bioinformatics, 2020, 2, lqaa038.	1.5	16
336	Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. Nature Medicine, 2020, 26, 1754-1765.	15.2	84
337	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	9.4	96
338	Exome Sequencing Implicates Impaired GABA Signaling and Neuronal Ion Transport in Trigeminal Neuralgia. IScience, 2020, 23, 101552.	1.9	32
339	A Recurrent Pathogenic Variant of INPP5K Underlies Autosomal Recessive Congenital Muscular Dystrophy With Cataracts and Intellectual Disability: Evidence for a Founder Effect in Southern Italy. Frontiers in Genetics, 2020, 11, 565868.	1.1	8
340	Gene Panel Tumor Testing in Ovarian Cancer Patients Significantly Increases the Yield of Clinically Actionable Germline Variants beyond BRCA1/BRCA2. Cancers, 2020, 12, 2834.	1.7	6
341	Whole genome, transcriptome and methylome profiling enhances actionable target discovery in high-risk pediatric cancer. Nature Medicine, 2020, 26, 1742-1753.	15.2	185
342	The DNA methylation landscape of advanced prostate cancer. Nature Genetics, 2020, 52, 778-789.	9.4	198
343	Family-specific genetic variants: Principles, detection, and clinical interpretation. , 2020, , 73-104.		0
344	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. Nature Communications, 2020, 11, 5918.	5.8	305
345	PTMsnp: A Web Server for the Identification of Driver Mutations That Affect Protein Post-translational Modification. Frontiers in Cell and Developmental Biology, 2020, 8, 593661.	1.8	7
346	Lack of pathogenic germline DICER1 variants in males with testicular germ-cell tumors. Cancer Genetics, 2020, 248-249, 49-56.	0.2	0
347	Genetic landscape of common venous malformations in the head and neck. Journal of Vascular Surgery: Venous and Lymphatic Disorders, 2020, 9, 1007-1016.e7.	0.9	7
348	Comprehensive Genetic Analysis of 128 Candidate Genes in a Cohort With Idiopathic, Severe, or Familial Osteoporosis. Journal of the Endocrine Society, 2020, 4, bvaa148.	0.1	11

#	Article	IF	Citations
349	Identification of candidate genetic variants and altered protein expression in neural stem and mature neural cells support altered microtubule function to be an essential component in bipolar disorder. Translational Psychiatry, 2020, 10, 390.	2.4	18
350	The Novel Desmin Variant p.Leu115Ile Is Associated With a Unique Form of Biventricular Arrhythmogenic Cardiomyopathy. Canadian Journal of Cardiology, 2021, 37, 857-866.	0.8	28
351	SUCLG1 mutations and mitochondrial encephalomyopathy: a case study and review of the literature. Molecular Biology Reports, 2020, 47, 9699-9714.	1.0	4
352	EAAT1 variants associated with glaucoma. Biochemical and Biophysical Research Communications, 2020, 529, 943-949.	1.0	11
353	Evidence of the milder phenotypic spectrum of c. 1582G >A PIGT variant: Delineation based on seven novel Polish patients. Clinical Genetics, 2020, 98, 468-476.	1.0	7
354	MISTIC: A prediction tool to reveal disease-relevant deleterious missense variants. PLoS ONE, 2020, 15, e0236962.	1.1	26
355	A novel SNCA E83Q mutation in a case of dementia with Lewy bodies and atypical frontotemporal lobar degeneration. Neuropathology, 2020, 40, 620-626.	0.7	27
356	The E3 ubiquitin ligase UBR5 interacts with TTC7A and may be associated with very early onset inflammatory bowel disease. Scientific Reports, 2020, 10, 18648.	1.6	4
357	Using whole-exome sequencing and protein interaction networks to prioritize candidate genes for germline cutaneous melanoma susceptibility. Scientific Reports, 2020, 10, 17198.	1.6	8
358	A DNAH17 missense variant causes flagella destabilization and asthenozoospermia. Journal of Experimental Medicine, 2020, 217, .	4.2	88
359	Identification of Missense ADGRV1 Mutation as a Candidate Genetic Cause of Familial Febrile Seizure 4. Children, 2020, 7, 144.	0.6	10
360	Annotation of Human Exome Gene Variants with Consensus Pathogenicity. Genes, 2020, 11, 1076.	1.0	4
361	CAPICE: a computational method for Consequence-Agnostic Pathogenicity Interpretation of Clinical Exome variations. Genome Medicine, 2020, 12, 75.	3.6	30
362	Integrated Genomic Characterization of the Human Immunome in Cancer. Cancer Research, 2020, 80, 4854-4867.	0.4	11
363	Assessing performance of pathogenicity predictors using clinically relevant variant datasets. Journal of Medical Genetics, 2021, 58, 547-555.	1.5	57
364	Report of a germline double heterozygote in MSH2 and PALB2. Molecular Genetics & Genomic Medicine, 2020, 8, e1242.	0.6	2
365	IDRMutPred: predicting disease-associated germline nonsynonymous single nucleotide variants (nsSNVs) in intrinsically disordered regions. Bioinformatics, 2020, 36, 4977-4983.	1.8	5
366	Dynamic incorporation of multiple in silico functional annotations empowers rare variant association analysis of large whole-genome sequencing studies at scale. Nature Genetics, 2020, 52, 969-983.	9.4	146

#	Article	IF	Citations
367	Improving the informativeness of Mendelian disease-derived pathogenicity scores for common disease. Nature Communications, 2020, 11, 6258.	5.8	8
368	Mutation Frequency in Main Susceptibility Genes Among Patients With Head and Neck Paragangliomas. Frontiers in Genetics, 2020, 11, 614908.	1.1	16
369	dbNSFP v4: a comprehensive database of transcript-specific functional predictions and annotations for human nonsynonymous and splice-site SNVs. Genome Medicine, 2020, 12, 103.	3.6	300
370	Transcriptomics in Alzheimerâ€™s Disease: Aspects and Challenges. International Journal of Molecular Sciences, 2020, 21, 3517.	1.8	37
371	Whole Exome Sequencing with Comprehensive Gene Set Analysis Identified a Biparental-Origin Homozygous c.509G>A Mutation in PPIB Gene Clustered in Two Taiwanese Families Exhibiting Fetal Skeletal Dysplasia during Prenatal Ultrasound. Diagnostics, 2020, 10, 286.	1.3	7
372	Identification of rare missense mutations in NOTCH2 and HERC2 associated with familial central precocious puberty via whole-exome sequencing. Gynecological Endocrinology, 2020, 36, 682-686.	0.7	11
373	Mutations in heat shock protein beta-1 (HSPB1) are associated with a range of clinical phenotypes related to different patterns of motor neuron dysfunction: A case series. Journal of the Neurological Sciences, 2020, 413, 116809.	0.3	14
374	Mutation severity spectrum of rare alleles in the human genome is predictive of disease type. PLoS Computational Biology, 2020, 16, e1007775.	1.5	11
375	Revisiting the complex architecture of ALS in Turkey: Expanding genotypes, shared phenotypes, molecular networks, and a public variant database. Human Mutation, 2020, 41, e7-e45.	1.1	10
376	Rare variant association testing in the non-coding genome. Human Genetics, 2020, 139, 1345-1362.	1.8	21
377	Rheostat positions: A new classification of protein positions relevant to pharmacogenomics. Medicinal Chemistry Research, 2020, 29, 1133-1146.	1.1	16
378	A Novel Pathogenic Variant in CARMIL2 (RLTPR) Causing CARMIL2 Deficiency and EBV-Associated Smooth Muscle Tumors. Frontiers in Immunology, 2020, 11, 884.	2.2	26
379	Identifying genetic variants and pathways associated with extreme levels of fetal hemoglobin in sickle cell disease in Tanzania. BMC Medical Genetics, 2020, 21, 125.	2.1	9
380	LEAP: Using machine learning to support variant classification in a clinical setting. Human Mutation, 2020, 41, 1079-1090.	1.1	23
381	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. Journal of Neuromuscular Diseases, 2020, 7, 153-166.	1.1	18
382	Protein Subdomain Enrichment of NUP155 Variants Identify a Novel Predicted Pathogenic Hotspot. Frontiers in Cardiovascular Medicine, 2020, 7, 8.	1.1	4
383	Evolution of approaches to identify melanoma missing heritability. Expert Review of Molecular Diagnostics, 2020, 20, 523-531.	1.5	16
384	High mutation burden of circulating cellâ€free DNA in earlyâ€stage breast cancer patients is associated with a poor relapseâ€free survival. Cancer Medicine, 2020, 9, 5922-5931.	1.3	9

#	Article	IF	Citations
385	In-silico analysis to identify the role of MEN1 missense mutations in breast cancer. Journal of Theoretical and Computational Chemistry, 2020, 19, 2041002.	1.8	3
386	Targeted next-generation sequencing study in familial ALS-FTD Portuguese patients negative for C9orf72 HRE. Journal of Neurology, 2020, 267, 3578-3592.	1.8	2
387	Results of targeted next-generation sequencing in children with cystic kidney diseases often change the clinical diagnosis. PLoS ONE, 2020, 15, e0235071.	1.1	12
388	Germline Mutations in Familial Papillary Thyroid Cancer. Endocrine Pathology, 2020, 31, 14-20.	5.2	14
389	EEF1A2 mutations in epileptic encephalopathy/intellectual disability: Understanding the potential mechanism of phenotypic variation. Epilepsy and Behavior, 2020, 105, 106955.	0.9	11
390	Comprehensive assessment of computational algorithms in predicting cancer driver mutations. Genome Biology, 2020, 21, 43.	3.8	47
391	The genetics of situs inversus without primary ciliary dyskinesia. Scientific Reports, 2020, 10, 3677.	1.6	37
392	Genetic Modifiers and Rare Mendelian Disease. Genes, 2020, 11, 239.	1.0	96
393	The Rare IL22RA2 Signal Peptide Coding Variant rs28385692 Decreases Secretion of IL-22BP Isoform-1, -2 and -3 and Is Associated with Risk for Multiple Sclerosis. Cells, 2020, 9, 175.	1.8	1
394	De novo variants in the Helicase-C domain of CHD8 are associated with severe phenotypes including autism, language disability and overgrowth. Human Genetics, 2020, 139, 499-512.	1.8	32
395	De novo variants in exomes of congenital heart disease patients identify risk genes and pathways. Genome Medicine, 2020, 12, 9.	3.6	43
396	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing. Eye and Vision (London, England), 2020, 7, 3.	1.4	5
397	Autosomal Dominant Tubulointerstitial Kidney Diseaseâ€"Uromodulin Misclassified as Focal Segmental Glomerulosclerosis or Hereditary Glomerular Disease. Kidney International Reports, 2020, 5, 519-529.	0.4	14
398	Variation benchmark datasets: update, criteria, quality and applications. Database: the Journal of Biological Databases and Curation, 2020, 2020, .	1.4	27
399	DSNetwork: An Integrative Approach to Visualize Predictions of Variantsâ€™ Deleteriousness. Frontiers in Genetics, 2019, 10, 1349.	1.1	5
400	An Improved Phenotype-Driven Tool for Rare Mendelian Variant Prioritization: Benchmarking Exomiser on Real Patient Whole-Exome Data. Genes, 2020, 11, 460.	1.0	42
401	Biâ€allelic mutations in HARS1 severely impair histidylâ€tRNA synthetase expression and enzymatic activity causing a novel multisystem ataxic syndrome. Human Mutation, 2020, 41, 1232-1237.	1.1	15
402	A high definition picture of somatic mutations in chronic lymphoproliferative disorder of natural killer cells. Blood Cancer Journal, 2020, 10, 42.	2.8	22

#	Article	IF	Citations
403	A research-driven approach to the identification of novel natural killer cell deficiencies affecting cytotoxic function. Blood, 2020, 135, 629-637.	0.6	4
404	Is Gene-Size an Issue for the Diagnosis of Skeletal Muscle Disorders?. Journal of Neuromuscular Diseases, 2020, 7, 203-216.	1.1	9
405	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. Haematologica, 2021, 106, 1693-1704.	1.7	40
406	Variant Calling in Next Generation Sequencing Data. , 2021, , 129-140.		0
407	MobiDetails: online DNA variants interpretation. European Journal of Human Genetics, 2021, 29, 356-360.	1.4	34
408	Triâ€Allelic Haplotypes Determine and Differentiate Functionally Normal Allele CYP2D6*2 and Impaired Allele CYP2D6*41. Clinical Pharmacology and Therapeutics, 2021, 109, 1256-1264.	2.3	7
409	Mutation analysis of MFSD8 in an amyotrophic lateral sclerosis cohort from mainland China. European Journal of Neuroscience, 2021, 53, 1197-1206.	1.2	2
410	Presynaptic congenital myasthenic syndrome due to three novel mutations in SLC5A7 encoding the sodium-dependant high-affinity choline transporter. Neuromuscular Disorders, 2021, 31, 21-28.	0.3	11
411	OncoVar: an integrated database and analysis platform for oncogenic driver variants in cancers. Nucleic Acids Research, 2021, 49, D1289-D1301.	6.5	64
412	High-Throughput Affinity Measurements of Transcription Factor and DNA Mutations Reveal Affinity and Specificity Determinants. Cell Systems, 2021, 12, 112-127.e11.	2.9	22
413	Functional and clinical implications of genetic structure in 1686 Italian exomes. Human Mutation, 2021, 42, 272-289.	1.1	5
414	Whole exome sequencing reveals pathogenic variants in MYO3A, MYO15A and COL9A3 and differential frequencies in ancestral alleles in hearing impairment genes among individuals from Cameroon. Human Molecular Genetics, 2021, 29, 3729-3743.	1.4	9
415	Identification of the genetic basis of sporadic polydactyly in China by targeted sequencing. Computational and Structural Biotechnology Journal, 2021, 19, 3482-3490.	1.9	0
416	GPCards: An integrated database of genotypeâ€“phenotype correlations in human genetic diseases. Computational and Structural Biotechnology Journal, 2021, 19, 1603-1611.	1.9	5
417	Clinical Utility of Functional RNA Analysis for the Reclassification of Splicing Gene Variants in Hereditary Cancer. Cancer Genomics and Proteomics, 2021, 18, 285-294.	1.0	6
418	Classification of genetic variants in hereditary cancer genes. , 2021, , 349-387.		0
419	Genetic analysis of ALS cases in the isolated island population of Malta. European Journal of Human Genetics, 2021, 29, 604-614.	1.4	18
420	Whole Exome Sequencing in Coloboma/Microphthalmia: Identification of Novel and Recurrent Variants in Seven Genes. Genes, 2021, 12, 65.	1.0	16

#	Article	IF	Citations
421	AIM in Genomic Basis of Medicine: Applications. , 2021, , 1-10.		0
422	Putative second hit rare genetic variants in families with seemingly GBA-associated Parkinsonâ€™s disease. Npj Genomic Medicine, 2021, 6, 2.	1.7	11
423	Identification of cancer related genes using feature selection and association rule mining. Informatics in Medicine Unlocked, 2021, 24, 100595.	1.9	15
424	MVP predicts theÂpathogenicity of missense variants by deep learning. Nature Communications, 2021, 12, 510.	5.8	85
425	Performance of mutation pathogenicity prediction tools on missense variants associated with 46,XY differences of sex development. Clinics, 2021, 76, e2052.	0.6	10
426	Rare missense variant inMSH4associated with primary gonadal failure in both 46, XX and 46, XY individuals. Human Reproduction, 2021, 36, 1134-1145.	0.4	18
427	Genetic variant effect prediction by supervised nonnegative matrix tri-factorization. Molecular Omics, 2021, 17, 740-751.	1.4	1
428	Integrating Evolutionary Genetics to Medical Genomics: Evolutionary Approaches to Investigate Disease-Causing Variants. , 0, , .		0
429	Rare Recurrent Variants in Noncoding Regions Impact Attention-Deficit Hyperactivity Disorder (ADHD) Gene Networks in Children of both African American and European American Ancestry. Genes, 2021, 12, 310.	1.0	10
430	Measurable residual disease in elderly acute myeloid leukemia: results from the PETHEMA-FLUGAZA phase 3 clinical trial. Blood Advances, 2021, 5, 760-770.	2.5	18
431	A Genome-First Approach to Characterize DICER1 Pathogenic Variant Prevalence, Penetrance, and Phenotype. JAMA Network Open, 2021, 4, e210112.	2.8	25
432	ALOX12 mutation in a family with dominantly inherited bleeding diathesis. Journal of Human Genetics, 2021, 66, 753-759.	1.1	5
433	Prioritizing variants of uncertain significance for reclassification using a rule-based algorithm in inherited retinal dystrophies. Npj Genomic Medicine, 2021, 6, 18.	1.7	20
434	Hypermutated phenotype in gliosarcoma of the spinal cord. Npj Precision Oncology, 2021, 5, 8.	2.3	5
435	Next Generation Exome Sequencing of Pediatric Asthma Identifies Rare and Novel Variants in Candidate Genes. Disease Markers, 2021, 2021, 1-10.	0.6	6
436	Genetic variants of small airways and interstitial pulmonary disease in children. Scientific Reports, 2021, 11, 2715.	1.6	4
437	Structure-Based Approaches to Classify the Functional Impact of ZBTB18 Missense Variants in Health and Disease. ACS Chemical Neuroscience, 2021, 12, 979-989.	1.7	4
439	Predicting the functional consequences of genetic variants in co-stimulatory ligand B7-1 using in-silico approaches. Human Immunology, 2021, 82, 103-120.	1.2	1

#	Article	IF	Citations
440	Ptosis as Clinical Presentation in a Patient With Emeryâ€“Dreifuss Muscular Dystrophy Type 5. Journal of Neuro-Ophthalmology, 2021, 41, e333-e334.	0.4	0
441	Discordant clinical features of identical hypertrophic cardiomyopathy twins. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	19
442	Reconstructing the Lineage Histories and Differentiation Trajectories of Individual Cancer Cells in Myeloproliferative Neoplasms. Cell Stem Cell, 2021, 28, 514-523.e9.	5.2	130
443	Candidate Markers of Olaparib Response from Genomic Data Analyses of Human Cancer Cell Lines. Cancers, 2021, 13, 1296.	1.7	3
444	Refining Genotypes and Phenotypes in KCNA2-Related Neurological Disorders. International Journal of Molecular Sciences, 2021, 22, 2824.	1.8	20
445	Expanding the Genotypic Spectrum of Congenital Sensory and Autonomic Neuropathies Using Whole-Exome Sequencing. Neurology: Genetics, 2021, 7, e568.	0.9	6
446	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. American Journal of Clinical Nutrition, 2021, 113, 1157-1167.	2.2	17
447	Combined Genome, Transcriptome and Metabolome Analysis in the Diagnosis of Childhood Cerebellar Ataxia. International Journal of Molecular Sciences, 2021, 22, 2990.	1.8	3
448	Exploring dementia and neuronal ceroid lipofuscinosis genes in 100 FTD-like patients from 6 towns and rural villages on the Adriatic Sea cost of Apulia. Scientific Reports, 2021, 11, 6353.	1.6	7
449	Type IV Collagen Variants in CKD: Performance of Computational Predictions for Identifying Pathogenic Variants. Kidney Medicine, 2021, 3, 257-266.	1.0	9
450	The Genetic Germline Background of Single and Multiple Primary Melanomas. Frontiers in Molecular Biosciences, 2020, 7, 555630.	1.6	6
451	Integrated mutational landscape analysis of uterine leiomyosarcomas. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	48
453	Machine learning-based reclassification of germline variants of unknown significance: The RENOVO algorithm. American Journal of Human Genetics, 2021, 108, 682-695.	2.6	13
454	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. Frontiers in Genetics, 2021, 12, 622886.	1.1	3
455	Genetic Defects in DNAH2 Underlie Male Infertility With Multiple Morphological Abnormalities of the Sperm Flagella in Humans and Mice. Frontiers in Cell and Developmental Biology, 2021, 9, 662903.	1.8	22
456	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. Circulation Research, 2021, 128, 1156-1169.	2.0	27
458	Exome-Wide Association Study on Alanine Aminotransferase Identifies Sequence Variants in the GPAM and APOE Associated With Fatty Liver Disease. Gastroenterology, 2021, 160, 1634-1646.e7.	0.6	82
459	Rare Germline Variants in Chordoma-Related Genes and Chordoma Susceptibility. Cancers, 2021, 13, 2704.	1.7	5

#	Article	IF	Citations
460	PPFIA4 mutation: A second hit in POLG related disease?. Epilepsy and Behavior Reports, 2021, 16, 100455.	0.5	2
461	The DBSAV Database: Predicting Deleteriousness of Single Amino Acid Variations in the Human Proteome. Journal of Molecular Biology, 2021, 433, 166915.	2.0	15
462	Neurodevelopmental phenotypes associated with pathogenic variants in SLC6A1. Journal of Medical Genetics, 2022, 59, 536-543.	1.5	18
463	Rho-GTPase Activating Protein myosin MYO9A identified as a novel candidate gene for monogenic focal segmental glomerulosclerosis. Kidney International, 2021, 99, 1102-1117.	2.6	12
464	A novel variant in the COX15 gene causing a fatal infantile cardioencephalomyopathy: A case report with clinical and molecular review. European Journal of Medical Genetics, 2021, 64, 104195.	0.7	1
466	Identification of COL3A1 variants associated with sporadic thoracic aortic dissection: a case-control study. Frontiers of Medicine, 2021, 15, 438-447.	1.5	3
467	Phenotypic diversity, disease progression, and pathogenicity of <scp>MVK</scp> missense variants in mevalonic aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 1272-1287.	1.7	17
468	How Machine Learning and Statistical Models Advance Molecular Diagnostics of Rare Disorders Via Analysis of RNA Sequencing Data. Frontiers in Molecular Biosciences, 2021, 8, 647277.	1.6	12
469	Evolutionary and functional lessons from human-specific amino acid substitution matrices. NAR Genomics and Bioinformatics, 2021, 3, lqab079.	1.5	1
471	The Use of Whole Genome and Exome Sequencing for Newborn Screening: Challenges and Opportunities for Population Health. Frontiers in Pediatrics, 2021, 9, 663752.	0.9	47
472	RBM20 Is a Candidate Gene for Hypertrophic Cardiomyopathy. Canadian Journal of Cardiology, 2021, 37, 1751-1759.	0.8	10
474	Radiation Necrosis with Proton Therapy in a Patient with Aarskog-Scott Syndrome and Medulloblastoma. International Journal of Particle Therapy, 2022, 8, 58-65.	0.9	2
476	Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	1.1	9
477	Screening for monogenic subtypes of gestational diabetes in a high prevalence island population â€“ A whole exome sequencing study. Diabetes/Metabolism Research and Reviews, 2022, 38, e3486.	1.7	2
478	Hearing Impairment with Monoallelic GJB2 Variants. Journal of Molecular Diagnostics, 2021, 23, 1279-1291.	1.2	10
480	PON-Sol2: Prediction of Effects of Variants on Protein Solubility. International Journal of Molecular Sciences, 2021, 22, 8027.	1.8	10
481	A genetic variant conferred high expression of CAV2 promotes pancreatic cancer progression and associates with poor prognosis. European Journal of Cancer, 2021, 151, 94-105.	1.3	10
482	RNA editing affects cisâ€regulatory elements and predicts adverse cancer survival. Cancer Medicine, 2021, 10, 6114-6127.	1.3	5

#	Article	IF	Citations
483	Functional Characterization of 21 Rare Allelic CYP1A2 Variants Identified in a Population of 4773 Japanese Individuals by Assessing Phenacetin O-Deethylation. Journal of Personalized Medicine, 2021, 11, 690.	1.1	5
484	Uniting biobank resources reveals novel genetic pathways modulating susceptibility for atopic dermatitis. Journal of Allergy and Clinical Immunology, 2022, 149, 1105-1112.e9.	1.5	41
486	A recurrent de novo variant supports <scp>KCNC2</scp> involvement in the pathogenesis of developmental and epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2021, 185, 3384-3389.	0.7	15
487	Case Report: Blepharophimosis and Ptosis as Leading Dysmorphic Features of Rare Congenital Malformation Syndrome With Developmental Delay â€“ New Cases With TRAF7 Variants. Frontiers in Medicine, 2021, 8, 708717.	1.2	4
488	A novel machine learning-based approach for the computational functional assessment of pharmacogenomic variants. Human Genomics, 2021, 15, 51.	1.4	14
489	Assigning function to SNPs: Considerations when interpreting genetic variation. Seminars in Cell and Developmental Biology, 2022, 121, 135-142.	2.3	13
490	Genetic evaluation supports differential diagnosis in adolescent patients with delayed puberty. European Journal of Endocrinology, 2021, 185, 617-627.	1.9	15
491	Multiregional genetic evolution of metastatic uveal melanoma. Npj Genomic Medicine, 2021, 6, 70.	1.7	9
493	L-Type Calcium Channel: Predicting Pathogenic/Likely Pathogenic Status for Variants of Uncertain Clinical Significance. Membranes, 2021, 11, 599.	1.4	3
494	X-CNV: genome-wide prediction of the pathogenicity of copy number variations. Genome Medicine, 2021, 13, 132.	3.6	24
495	A domain damage index to prioritizing the pathogenicity of missense variants. Human Mutation, 2021, 42, 1503-1517.	1.1	0
496	Natural Mutations Affect Structure and Function of gC1q Domain of Otolin-1. International Journal of Molecular Sciences, 2021, 22, 9085.	1.8	5
497	A clinically applicable integrative molecular classification of meningiomas. Nature, 2021, 597, 119-125.	13.7	180
498	Monogenic Diabetes in Youth With Presumed Type 2 Diabetes: Results From the Progress in Diabetes Genetics in Youth (ProDiGY) Collaboration. Diabetes Care, 2021, 44, 2312-2319.	4.3	21
499	Predicting functional consequences of mutations using molecular interaction network features. Human Genetics, 2022, 141, 1195-1210.	1.8	9
500	A convergent molecular network underlying autism and congenital heart disease. Cell Systems, 2021, 12, 1094-1107.e6.	2.9	19
501	DIAPH1 Variants in Nonâ€“East Asian Patients With Sporadic Moyamoya Disease. JAMA Neurology, 2021, 78, 993.	4.5	33
502	Genome sequencing data analysis for rare disease gene discovery. Briefings in Bioinformatics, 2022, 23, .	3.2	6

#	Article	IF	Citations
503	Improved pathogenicity prediction for rare human missense variants. American Journal of Human Genetics, 2021, 108, 1891-1906.	2.6	51
504	A Rare Case of Brachyolmia with Amelogenesis Imperfecta Caused by a New Pathogenic Splicing Variant in LTBP3. Genes, 2021, 12, 1406.	1.0	2
505	Yield of clinically reportable genetic variants in unselected cerebral palsy by whole genome sequencing. Npj Genomic Medicine, 2021, 6, 74.	1.7	16
506	Biallelic variants of ATP13A3 cause dose-dependent childhood-onset pulmonary arterial hypertension characterised by extreme morbidity and mortality. Journal of Medical Genetics, 2022, 59, 906-911.	1.5	22
507	First characterization of congenital myasthenic syndrome type 5 in North Africa. Molecular Biology Reports, 2021, 48, 6999-7006.	1.0	4
508	Next Generation Sequencing of Tumor and Matched Plasma Samples: Identification of Somatic Variants in ctDNA From Ovarian Cancer Patients. Frontiers in Oncology, 2021, 11, 754094.	1.3	5
509	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. Human Genetics, 2022, 141, 1269-1278.	1.8	10
510	Identifying digenic disease genes via machine learning in the Undiagnosed Diseases Network. American Journal of Human Genetics, 2021, 108, 1946-1963.	2.6	25
511	Whole genome sequence analysis of platelet traits in the NHLBI Trans-Omics for Precision Medicine (TOPMed) initiative. Human Molecular Genetics, 2022, 31, 347-361.	1.4	9
512	Early-Onset Atrial Fibrillation and the Prevalence of Rare Variants in Cardiomyopathy and Arrhythmia Genes. JAMA Cardiology, 2021, 6, 1371.	3.0	66
513	Rare, Damaging DNA Variants in CORIN and Risk of Coronary Artery Disease: Insights From Functional Genomics and Large-Scale Sequencing Analyses. Circulation Genomic and Precision Medicine, 2021, 14, e003399.	1.6	10
514	The ethnogeographic variability of genetic factors underlying G6PD deficiency. Pharmacological Research, 2021, 173, 105904.	3.1	14
515	Study of rare genetic variants in TM4SF20, NFXL1, CNTNAP2, and ATP2C2 in Pakistani probands and families with language impairment. Meta Gene, 2021, 30, 100966.	0.3	6
516	Rare and de novo coding variants in chromodomain genes in Chiari I malformation. American Journal of Human Genetics, 2021, 108, 100-114.	2.6	17
517	A catalogue of 863 Rett-syndrome-causing MECP2 mutations and lessons learned from data integration. Scientific Data, 2021, 8, 10.	2.4	12
518	Current cancer driver variant predictors learn to recognize driver genes instead of functional variants. BMC Biology, 2021, 19, 3.	1.7	14
519	SBSA: an online service for somatic binding sequence annotation. Nucleic Acids Research, 2022, 50, e4-e4.	6.5	8
520	Wide spectrum of NR5A1â€related phenotypes in 46,XY and 46,XX individuals. Birth Defects Research Part C: Embryo Today Reviews, 2016, 108, 309-320.	3.6	76

#	Article	IF	Citations
521	Finding a Needle in a Haystack: Variant Effect Predictor (VEP) Prioritizes Disease Causative Variants from Millions of Neutral Ones. , 2019, , 85-104.		1
522	Novel polymorphisms associated with hyperalphalipoproteinemia and apparent cardioprotection. Journal of Clinical Lipidology, 2018, 12, 110-115.	0.6	8
523	Comparison of Pathogenicity Prediction Tools on Somatic Variants. Journal of Molecular Diagnostics, 2020, 22, 1383-1392.	1.2	19
524	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	5.8	35
525	AI-Driver: an ensemble method for identifying driver mutations in personal cancer genomes. NAR Genomics and Bioinformatics, 2020, 2, lqaa084.	1.5	19
549	Deficient LRRC8A-dependent volume-regulated anion channel activity is associated with male infertility in mice. JCI Insight, 2018, 3, .	2.3	29
550	A nonhuman primate model of inherited retinal disease. Journal of Clinical Investigation, 2019, 129, 863-874.	3.9	78
551	Human CRY1 variants associate with attention deficit/hyperactivity disorder. Journal of Clinical Investigation, 2020, 130, 3885-3900.	3.9	35
552	Biological and clinical significance of dysplastic hematopoiesis in patients with newly diagnosed multiple myeloma. Blood, 2020, 135, 2375-2387.	0.6	24
553	Towards Increasing the Clinical Relevance of In Silico Methods to Predict Pathogenic Missense Variants. PLoS Computational Biology, 2016, 12, e1004725.	1.5	34
554	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
555	Germline Variants of Prostate Cancer in Japanese Families. PLoS ONE, 2016, 11, e0164233.	1.1	21
556	Molecular investigation by whole exome sequencing revealed a high proportion of pathogenic variants among Thai victims of sudden unexpected death syndrome. PLoS ONE, 2017, 12, e0180056.	1.1	17
557	Generalising better: Applying deep learning to integrate deleteriousness prediction scores for whole-exome SNV studies. PLoS ONE, 2018, 13, e0192829.	1.1	14
558	Hypogonadotropic hypogonadism and pituitary hypoplasia as recurrent features in Ulnar-Mammary syndrome. Endocrine Connections, 2018, 7, 1432-1441.	0.8	9
559	Status Dystonicus, Oculogyric Crisis and Paroxysmal Dyskinesia in a 25 Year-Old Woman with a Novel KCNMA1 Variant, K457E. Tremor and Other Hyperkinetic Movements, 2020, 10, 49.	1.1	7
560	Population Prevalence of Deleterious <i>SGCE</i> Variants. Tremor and Other Hyperkinetic Movements, 2020, 10, 50.	1.1	4
561	Assembling and Validating Bioinformatic Pipelines for Next-Generation Sequencing Clinical Assays. Archives of Pathology and Laboratory Medicine, 2020, 144, 1118-1130.	1.2	13

#	Article	IF	Citations
562	Two locus inheritance of non-syndromic midline craniosynostosis via rare SMAD6 and common BMP2 alleles. ELife, 2016, 5, .	2.8	168
563	Rare missense variants in the human cytosolic antibody receptor preserve antiviral function. ELife, 2019, 8, .	2.8	9
564	GATA6 mutations in hiPSCs inform mechanisms for maldevelopment of the heart, pancreas, and diaphragm. ELife, 2020, 9, .	2.8	31
565	Sequence variant analysis of RNA sequences in severe equine asthma. PeerJ, 2018, 6, e5759.	0.9	8
566	Evaluation of performance of leading algorithms for variant pathogenicity predictions and designing a combinatory predictor method: application to Rett syndrome variants. PeerJ, 2019, 7, e8106.	0.9	10
567	Genetic characteristics of non-familial epilepsy. PeerJ, 2019, 7, e8278.	0.9	15
568	From genotype to phenotype inArabidopsis thaliana:in-silicogenome interpretation predicts 288 phenotypes from sequencing data. Nucleic Acids Research, 2022, 50, e16-e16.	6.5	6
569	Component of oligomeric Golgi complex 1 deficiency leads to hypoglycemia: a case report and literature review. BMC Pediatrics, 2021, 21, 442.	0.7	1
570	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
572	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
588	Brief Guidelines on Preparation of Manuscripts Containing Information on the Results of Molecular Genetic Research. Voprosy Sovremennoi Pediatrii - Current Pediatrics, 2018, 17, 364-366.	0.1	0
596	PILOT RESEARCH OF A GENETIC PREDISPOSITION FOR CLINICAL MANIFESTATIONS OF ACUTE INTERMITTENT PORPHYRIA. Gematologiya I Transfuziologiya, 2019, 64, 123-137.	0.1	1
598	Exploration of the genomic landscape of a long-term surviving stage III colorectal cancer patient identifies recurrent and rare mutations: a case report. Translational Cancer Research, 2020, 9, 2992-2998.	0.4	0
601	Integrated analysis of whole genome and transcriptome sequencing in a young patient with gastric cancer provides insights for precision therapy. Oncology Letters, 2020, 20, 1-1.	0.8	0
607	Burden of rare coding variants in an Italian cohort of familial multiple sclerosis. Journal of Neuroimmunology, 2021, 362, 577760.	1.1	3
608	VPMBench: a test bench for variant prioritization methods. BMC Bioinformatics, 2021, 22, 543.	1.2	0
609	Deep Learning of the Retina Enables Phenome- and Genome-Wide Analyses of the Microvasculature. Circulation, 2022, 145, 134-150.	1.6	57
610	Exome sequencing identifies mutations in three cases diagnosed with Retinitis Pigmentosa and hearing impairment. Molecular Vision, 2020, 26, 216-225.	1.1	2

#	Article	IF	Citations
611	Identification of novel mutations by targeted NGS in Moroccan families clinically diagnosed with a neuromuscular disorder. Clinica Chimica Acta, 2022, 524, 51-58.	0.5	2
612	Desmoplakin and periplakin genetically and functionally contribute to eosinophilic esophagitis. Nature Communications, 2021, 12, 6795.	5.8	23
613	MLb-LDLr. JACC Basic To Translational Science, 2021, 6, 815-827.	1.9	10
615	The Somatic Mutation Paradigm in Congenital Malformations: Hirschsprung Disease as a Model. International Journal of Molecular Sciences, 2021, 22, 12354.	1.8	3
616	Overlap phenotypes of the left ventricular noncompaction and hypertrophic cardiomyopathy with complex arrhythmias and heart failure induced by the novel truncated DSC2 mutation. Orphanet Journal of Rare Diseases, 2021, 16, 496.	1.2	11
617	Computational Methods and Approaches in Pharmacogenomic Research. , 2022, , 53-83.		1
618	Association of Protein Function-Altering Variants With Cardiometabolic Traits: The Strong Heart Study. SSRN Electronic Journal, 0, , .	0.4	0
619	NOVEL HOMOZYGOUS VARIANT OF TBC1 DOMAIN FAMILY MEMBER 8 GENE IN FOUR LIBYAN SIBLINGS WITH AUTISTIC SPECTRUM DISORDER AND INTELLECTUAL DISABILITY WITHOUT EPILEPSY. Innovare Journal of Medical Sciences, 0, , 1-4.	0.2	0
620	Preneoplastic somatic mutations including MYD88 ^{L265P} in lymphoplasmacytic lymphoma. Science Advances, 2022, 8, eabl4644.	4.7	21
621	A comparison on predicting functional impact of genomic variants. NAR Genomics and Bioinformatics, 2022, 4, lqab122.	1.5	12
622	Artecanin of Laurus nobilis is a novel inhibitor of SARS-CoV-2 main protease with highly desirable druglikeness. Journal of Biomolecular Structure and Dynamics, 2023, 41, 2355-2367.	2.0	5
623	Evaluation of phenotype-driven gene prioritization methods for Mendelian diseases. Briefings in Bioinformatics, 2022, 23, .	3.2	17
625	Sequencing of a Chinese tetralogy of Fallot cohort reveals clustering mutations in myogenic heart progenitors. JCI Insight, 2022, 7, .	2.3	9
626	Whole Genome Sequencing Unravels New Genetic Determinants of Early-Onset Familial Osteoporosis and Low BMD in Malta. Genes, 2022, 13, 204.	1.0	2
627	Potential Involvement of NSD1, KRT24 and ACACA in the Genetic Predisposition to Colorectal Cancer. Cancers, 2022, 14, 699.	1.7	0
628	Comprehensive Analysis of Co-Mutations Identifies Cooperating Mechanisms of Tumorigenesis. Cancers, 2022, 14, 415.	1.7	8
629	Fitness Effects of Mutations: An Assessment of PROVEAN Predictions Using Mutation Accumulation Data. Genome Biology and Evolution, 2022, 14, .	1.1	19
630	Rare coding variants in 35 genes associate with circulating lipid levelsâ€"A multi-ancestry analysis of 170,000 exomes. American Journal of Human Genetics, 2022, 109, 81-96.	2.6	24

#	Article	IF	Citations
631	Machine learning methods for prediction of cancer driver genes: a survey paper. Briefings in Bioinformatics, 2022, 23, .	3.2	15
632	Next-generation sequencing of the whole mitochondrial genome identifies functionally deleterious mutations in patients with multiple sclerosis. PLoS ONE, 2022, 17, e0263606.	1.1	10
633	Analysis of missense variants in the human genome reveals widespread gene-specific clustering and improves prediction of pathogenicity. American Journal of Human Genetics, 2022, 109, 457-470.	2.6	29
634	The Diverse Phenotype of Intestinal Dysmotility Secondary to ACTG2â€related Disorders. Journal of Pediatric Gastroenterology and Nutrition, 2022, 74, 575-581.	0.9	4
635	Identifying Actionable Variants Using Capture-Based Targeted Sequencing in 563 Patients With Non-Small Cell Lung Carcinoma. Frontiers in Oncology, 2021, 11, 812433.	1.3	0
636	Further delineation of familial polycystic ovary syndrome (PCOS) via <scp>wholeâ€exome</scp> sequencing: <scp>PCOS</scp>â€related rare <scp>FBN3</scp> and <scp>FN1</scp> gene variants are identified. Journal of Obstetrics and Gynaecology Research, 2022, 48, 1202-1211.	0.6	9
637	Influence of PRKCE non-synonymous variants on protein dynamics and functionality. Human Molecular Genetics, 2022, 31, 2236-2261.	1.4	12
638	PirePred. Journal of Molecular Diagnostics, 2022, 24, 406-425.	1.2	1
639	Whole-Exome Sequencing Revealed a Pathogenic Nonsense Variant in theSLC19A2Gene in an Iranian Family with Thiamine-Responsive Megaloblastic Anemia. Laboratory Medicine, 2022, 53, 640-650.	0.8	0
640	Computational Resources for the Interpretation of Variations in Cancer. Advances in Experimental Medicine and Biology, 2022, 1361, 177-198.	0.8	2
641	AIM in Genomic Basis of Medicine: Applications. , 2022, , 1087-1096.		0
642	DVPred: a disease-specific prediction tool for variant pathogenicity classification for hearing loss. Human Genetics, 2022, 141, 401-411.	1.8	6
643	The Role of Natural Polymorphic Variants of DNA Polymerase Î² in DNA Repair. International Journal of Molecular Sciences, 2022, 23, 2390.	1.8	8
646	Transcriptome-wide association study of coronary artery disease identifies novel susceptibility genes. Basic Research in Cardiology, 2022, 117, 6.	2.5	22
647	Integrated Analysis of Coexpression and Exome Sequencing to Prioritize Susceptibility Genes for Familial Cutaneous Melanoma. Journal of Investigative Dermatology, 2022, 142, 2464-2475.e5.	0.3	4
648	Scalable Dual-Fluorescence Assay for Functional Interpretation of HNF-4Î± Missense Variants. Frontiers in Endocrinology, 2022, 13, 812747.	1.5	0
649	Filamin A Is a Potential Driver of Breast Cancer Metastasis via Regulation of MMP-1. Frontiers in Oncology, 2022, 12, 836126.	1.3	5
650	Computational Analysis of the Potential Impact of MTC Complex Missenses SNPs Associated with Male Infertility. BioMed Research International, 2022, 2022, 1-18.	0.9	3

#	Article	IF	Citations
651	AmazonForest: In Silico Metaprediction of Pathogenic Variants. Biology, 2022, 11, 538.	1.3	0
652	The role of SPAG1 in the assembly of axonemal dyneins in human airway epithelia. Journal of Cell Science, 2022, 135, .	1.2	5
653	GENESIS: Gene-Specific Machine Learning Models for Variants of Uncertain Significance Found in Catecholaminergic Polymorphic Ventricular Tachycardia and Long QT Syndrome-Associated Genes. Circulation: Arrhythmia and Electrophysiology, 2022, 15, 101161CIRCEP121010326.	2.1	17
654	Venus: Elucidating the Impact of Amino Acid Variants on Protein Function Beyond Structure Destabilisation. Journal of Molecular Biology, 2022, 434, 167567.	2.0	13
655	Fibrillar Collagen Variants in Spontaneous Coronary Artery Dissection. JAMA Cardiology, 2022, 7, 396.	3.0	19
656	OGDHL Variant rs2293239: A Potential Genetic Driver of Chinese Familial Depressive Disorder. Frontiers in Psychiatry, 2022, 13, 771950.	1.3	2
657	An expanded phenotype centric benchmark of variant prioritisation tools. Human Mutation, 2022, 43, 539-546.	1.1	9
658	Structural Consequence of Non-Synonymous Single-Nucleotide Variants in the N-Terminal Domain of LIS1. International Journal of Molecular Sciences, 2022, 23, 3109.	1.8	3
659	A comprehensive WGS-based pipeline for the identification of new candidate genes in inherited retinal dystrophies. Npj Genomic Medicine, 2022, 7, 17.	1.7	7
660	Whole genome sequencing delineates regulatory, copy number, and cryptic splice variants in early onset cardiomyopathy. Npj Genomic Medicine, 2022, 7, 18.	1.7	14
661	Novel Variants of ANO5 in Two Patients With Limb Girdle Muscular Dystrophy: Case Report. Frontiers in Neurology, 2022, 13, 868655.	1.1	0
662	Predictive Markers of Response to Neoadjuvant Durvalumab with Nab-Paclitaxel and Dose-Dense Doxorubicin/Cyclophosphamide in Basal-Like Triple-Negative Breast Cancer. Clinical Cancer Research, 2022, 28, 2587-2597.	3.2	16
663	Impaired neurogenesis alters brain biomechanics in a neuroprogenitor-based genetic subtype of congenital hydrocephalus. Nature Neuroscience, 2022, 25, 458-473.	7.1	46
664	Assessment of 13 in silico pathogenicity methods on cancer-related variants. Computers in Biology and Medicine, 2022, 145, 105434.	3.9	2
665	Aicardi-GoutiÃ¨res Syndrome due to a SAMHD1 Mutation Presenting with Deep White Matter Cysts. Molecular Syndromology, 2022, 13, 132-138.	0.3	3
667	Predicting deleterious missense genetic variants via integrative supervised nonnegative matrix tri-factorization. Scientific Reports, 2021, 11, 23747.	1.6	0
668	Leveraging cell-type-specific regulatory networks to interpret genetic variants in abdominal aortic aneurysm. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	8
669	Identification of deleterious single nucleotide polymorphism (SNP)s in the human TBX5 gene & prediction of their structural & functional consequences: An in silico approach. Biochemistry and Biophysics Reports, 2021, 28, 101179.	0.7	2

#	Article	IF	Citations
670	A functionally impaired missense variant identified in French Canadian families implicates FANCI as a candidate ovarian cancer-predisposing gene. Genome Medicine, 2021, 13, 186.	3.6	12
671	A rare CTSC mutation in Papillon-LefÃ¨vre Syndrome results in abolished serine protease activity and reduced NET formation but otherwise normal neutrophil function. PLoS ONE, 2021, 16, e0261724.	1.1	4
672	Whole-exome sequencing reveals damaging gene variants associated with hypoalphalipoproteinemia. Journal of Lipid Research, 2022, 63, 100209.	2.0	2
674	Diagnostic yield of whole exome data in fetuses aborted for conotruncal malformations. Prenatal Diagnosis, 2022, 42, 852-861.	1.1	1
707	Genome interpretation using in silico predictors of variant impact. Human Genetics, 2022, 141, 1549-1577.	1.8	26
708	The Genetic and Molecular Analyses of RAD51C and RAD51D Identifies Rare Variants Implicated in Hereditary Ovarian Cancer from a Genetically Unique Population. Cancers, 2022, 14, 2251.	1.7	4
709	Mutation spectrum of congenital heart disease in a consanguineous Turkish population. Molecular Genetics & Genomic Medicine, 2022, 10, e1944.	0.6	4
710	Rare and potential pathogenic mutations of LMNA and LAMA4 associated with familial arrhythmogenic right ventricular cardiomyopathy/dysplasia with right ventricular heart failure, cerebral thromboembolism and hereditary electrocardiogram abnormality. Orphanet Journal of Rare Diseases, 2022, 17, 183.	1.2	5
711	HPMPdb: a machine learning-ready database of protein molecular phenotypes associated to human missense variants. Current Research in Structural Biology, 2022, , .	1.1	1
712	Oxytocin and vasotocin receptor variation and the evolution of human prosociality. Comprehensive Psychoneuroendocrinology, 2022, 11, 100139.	0.7	6
713	Computational and experimental methods for classifying variants of unknown clinical significance.. Cold Spring Harbor Molecular Case Studies, 2022, 8, .	0.7	7
715	PHACT: Phylogeny-Aware Computing of Tolerance for Missense Mutations. Molecular Biology and Evolution, 2022, 39, .	3.5	3
716	Association of protein function-altering variants with cardiometabolic traits: the strong heart study. Scientific Reports, 2022, 12, .	1.6	0
718	PON-All: Amino Acid Substitution Tolerance Predictor for All Organisms. Frontiers in Molecular Biosciences, 0, 9, .	1.6	7
719	Presence of rare potential pathogenic variants in subjects under 65Âyears old with very severe or fatal COVID-19. Scientific Reports, 2022, 12, .	1.6	6
720	A diseaseâ€associated missense mutation in CYP4F3 affects the metabolism of leukotriene B4 via disruption of electron transfer. Journal of Cachexia, Sarcopenia and Muscle, 2022, 13, 2242-2253.	2.9	5
721	Exome sequencing identifies genetic variants in anophthalmia and microphthalmia. American Journal of Medical Genetics, Part A, 2022, 188, 2376-2388.	0.7	2
722	Calculating genetic risk for dysfunction in pleiotropic biological processes using whole exome sequencing data. Journal of Neurodevelopmental Disorders, 2022, 14, .	1.5	0

#	Article	IF	Citations
723	Interpreting protein variant effects with computational predictors and deep mutational scanning. DMM Disease Models and Mechanisms, 2022, 15, .	1.2	25
724	Quantifying concordant genetic effects of de novo mutations on multiple disorders. ELife, 0, 11, .	2.8	3
725	Dissecting Generalizability and Actionability of Disease-Associated Genes From 20 Worldwide Ethnolinguistic Cultural Groups. Frontiers in Genetics, 0, 13, .	1.1	3
726	Network assisted analysis of de novo variants using protein-protein interaction information identified 46 candidate genes for congenital heart disease. PLoS Genetics, 2022, 18, e1010252.	1.5	3
727	A calibrated functional patch-clamp assay to enhance clinical variant interpretation in KCNH2-related long QT syndrome. American Journal of Human Genetics, 2022, 109, 1199-1207.	2.6	16
728	RAD51B Harbors Germline Mutations Associated With Pancreatic Ductal Adenocarcinoma. JCO Precision Oncology, 2022, , .	1.5	1
730	Analysis of the entire mitochondrial genome reveals Leberâ€™s hereditary optic neuropathy mitochondrial DNA mutations in an Arab cohort with multiple sclerosis. Scientific Reports, 2022, 12, .	1.6	1
731	CaMKII Inhibition Attenuates Distinct Gain-of-Function Effects Produced by Mutant Nav1.6 Channels and Reduces Neuronal Excitability. Cells, 2022, 11, 2108.	1.8	2
732	Diagnostics of BAP1-Tumor Predisposition Syndrome by a Multitesting Approach: A Ten-Year-Long Experience. Diagnostics, 2022, 12, 1710.	1.3	4
733	Whole-Genome Sequencing Identified KCNJ12 and SLC25A5 Mutations in Port-Wine Stains. Frontiers in Medicine, 0, 9, .	1.2	0
734	A Comprehensive Evaluation of the Performance of Prediction Algorithms on Clinically Relevant Missense Variants. International Journal of Molecular Sciences, 2022, 23, 7946.	1.8	5
735	Using Machine Learning for Predicting the Effect of Mutations in the Initiation Codon. IEEE Journal of Biomedical and Health Informatics, 2022, 26, 5750-5756.	3.9	1
736	Aggregated Genomic Data as Cohort-Specific Allelic Frequencies can Boost Variants and Genes Prioritization in Non-Solved Cases of Inherited Retinal Dystrophies. International Journal of Molecular Sciences, 2022, 23, 8431.	1.8	4
737	Cutting-Edge AI Technologies Meet Precision Medicine to Improve Cancer Care. Biomolecules, 2022, 12, 1133.	1.8	1
738	Predictive Modelling in Clinical Bioinformatics: Key Concepts for Startups. BioTech, 2022, 11, 35.	1.3	3
740	Rare germline ATM variants of uncertain significance in chronic lymphocytic leukaemia and other cancers. British Journal of Haematology, 0, , .	1.2	1
741	mvPPT: A Highly Efficient and Sensitive Pathogenicity Prediction Tool for Missense Variants. Genomics, Proteomics and Bioinformatics, 2023, 21, 414-426.	3.0	2
742	RB1 loss triggers dependence on ESRRG in retinoblastoma. Science Advances, 2022, 8, .	4.7	5

#	Article	IF	Citations
743	Clinical significance of genetic variation in hypertrophic cardiomyopathy: comparison of computational tools to prioritize missense variants. Frontiers in Cardiovascular Medicine, 0, 9, .	1.1	5
744	From bugs to bedside: functional annotation of human genetic variation for neurological disorders using invertebrate models. Human Molecular Genetics, 2022, 31, R37-R46.	1.4	8
745	Statistical models of the genetic etiology of congenital heart disease. Current Opinion in Genetics and Development, 2022, 76, 101967.	1.5	0
746	Effect of an autism-associated KCNMB2 variant, G124R, on BK channel properties. Current Research in Physiology, 2022, 5, 404-413.	0.8	3
748	Endophenotype effect sizes support variant pathogenicity in monogenic disease susceptibility genes. Nature Communications, 2022, 13, .	5.8	3
749	Computational approaches for predicting variant impact: An overview from resources, principles to applications. Frontiers in Genetics, 0, 13, .	1.1	11
750	Diagnosis of a Single-Nucleotide Variant in Whole-Exome Sequencing Data for Patients With Inherited Diseases: Machine Learning Study Using Artificial Intelligence Variant Prioritization. JMIR Bioinformatics and Biotechnology, 2022, 3, e37701.	0.4	0
751	In silico analyses of Wnt1 nsSNPs reveal structurally destabilizing variants, altered interactions with Frizzled receptors and its deregulation in tumorigenesis. Scientific Reports, 2022, 12, .	1.6	0
752	Assessment of genetic susceptibility to multiple primary cancers through whole-exome sequencing in two large multi-ancestry studies. BMC Medicine, 2022, 20, .	2.3	3
753	Screening of candidate genes at GLC3B and GLC3C loci in Chinese primary congenital glaucoma patients with targeted next generation sequencing. Ophthalmic Genetics, 2023, 44, 133-138.	0.5	1
754	A whole genome sequencing approach to anterior cruciate ligament ruptureâ€“a twin study in two unrelated families. PLoS ONE, 2022, 17, e0274354.	1.1	1
755	MetaRNN: differentiating rare pathogenic and rare benign missense SNVs and InDels using deep learning. Genome Medicine, 2022, 14, .	3.6	28
756	Whole exome sequencing in Brugada and long QT syndromes revealed novel rare and potential pathogenic mutations related to the dysfunction of the cardiac sodium channel. Orphanet Journal of Rare Diseases, 2022, 17, .	1.2	5
757	A framework for detecting noncoding rare-variant associations of large-scale whole-genome sequencing studies. Nature Methods, 2022, 19, 1599-1611.	9.0	36
758	MARGINAL: An Automatic Classification of Variants in BRCA1 and BRCA2 Genes Using a Machine Learning Model. Biomolecules, 2022, 12, 1552.	1.8	3
759	Association between peroxisome proliferator activated receptor gamma coactivator 1 gene with overweight and obesity risk: Caseâ€“control study and meta-analysis. , 2022, 34, 201123.		2
760	In silico assessment of missense point mutations on human cathelicidin LL-37. Journal of Molecular Graphics and Modelling, 2023, 118, 108368.	1.3	1
761	Predicting functional effect of missense variants using graph attention neural networks. Nature Machine Intelligence, 2022, 4, 1017-1028.	8.3	15

#	Article	IF	Citations
762	In silico versus functional characterization of genetic variants: lessons from muscle channelopathies. Brain, 2023, 146, 1316-1321.	3.7	1
763	SWI/SNF complex gene variations are associated with a higher tumor mutational burden and a better response to immune checkpoint inhibitor treatment: a pan-cancer analysis of next-generation sequencing data corresponding to 4591 cases. Cancer Cell International, 2022, 22, .	1.8	7
765	Using reported pathogenic variants to identify therapeutic opportunities for genetic diseases. Molecular Genetics & Genomic Medicine, 0, , .	0.6	1
766	Next-generation sequencing of postmortem molecular markers to support for medicolegal autopsy. Forensic Science International: Reports, 2022, 6, 100300.	0.4	0
767	Congenital Stationary Night Blindness: Clinical and Genetic Features. International Journal of Molecular Sciences, 2022, 23, 14965.	1.8	4
768	Insights on variant analysis in silico tools for pathogenicity prediction. Frontiers in Genetics, 0, 13, .	1.1	17
770	Editorial: Towards genome interpretation: Computational methods to model the genotype-phenotype relationship. Frontiers in Bioinformatics, 0, 2, .	1.0	1
771	Evaluation of in silico predictors on short nucleotide variants in HBA1, HBA2, and HBB associated with haemoglobinopathies. ELife, 0, 11, .	2.8	6
772	Opportunities and Challenges with Artificial Intelligence in Genomics. Clinics in Laboratory Medicine, 2023, 43, 87-97.	0.7	2
773	Powerful, scalable and resource-efficient meta-analysis of rare variant associations in large whole genome sequencing studies. Nature Genetics, 2023, 55, 154-164.	9.4	12
774	Brain metastatic outgrowth and osimertinib resistance are potentiated by RhoA in EGFR-mutant lung cancer. Nature Communications, 2022, 13, .	5.8	6
775	Structural Evaluation and Conformational Dynamics of ZNF141T474I Mutation Provoking Postaxial Polydactyly Type A. Bioengineering, 2022, 9, 749.	1.6	5
776	Whole exome sequencing in dense families suggests genetic pleiotropy amongst Mendelian and complex neuropsychiatric syndromes. Scientific Reports, 2022, 12, .	1.6	2
777	Case report: Adult-onset limb girdle muscular dystrophy in sibling pair due to novel homozygous LAMA2 missense variant. Frontiers in Neurology, 0, 14, .	1.1	0
778	Molecular Genetic Characteristics of FANCI, a Proposed New Ovarian Cancer Predisposing Gene. Genes, 2023, 14, 277.	1.0	3
779	SLC26A1 is a major determinant of sulfate homeostasis in humans. Journal of Clinical Investigation, 2023, 133, .	3.9	5
780	Exploring genotypeâ€“phenotype correlations in glutaric aciduria type 1. Journal of Inherited Metabolic Disease, 2023, 46, 371-390.	1.7	8
781	Abnormal expression of lysosomal glycoproteins in patients with congenital disorders of glycosylation. BMC Research Notes, 2023, 16, .	0.6	0

#	Article	IF	Citations
782	Integration of deep learning with Ramachandran plot molecular dynamics simulation for genetic variant classification. IScience, 2023, 26, 106122.	1.9	2
783	Case report: Novel compound heterozygosity for pathogenic variants in MED23 in a syndromic patient with postnatal microcephaly. Frontiers in Neurology, 0, 14, .	1.1	1
784	Model performance and interpretability of semi-supervised generative adversarial networks to predict oncogenic variants with unlabeled data. BMC Bioinformatics, 2023, 24, .	1.2	5
785	Explainable AI for Estimating Pathogenicity of Genetic Variants Using Large-Scale Knowledge Graphs. Cancers, 2023, 15, 1118.	1.7	1
786	Whole-Exome Sequencing Analyses Support a Role of Vitamin D Metabolism in Ischemic Stroke. Stroke, 2023, 54, 800-809.	1.0	1
787	Extensive set of African ancestry-informative markers (AIMs) to study ancestry and population health. Frontiers in Genetics, 0, 14, .	1.1	0
789	A Comprehensive Genetic Analysis of Slovenian Families with Multiple Cases of Orofacial Clefts Reveals Novel Variants in the Genes IRF6, GRHL3, and TBX22. International Journal of Molecular Sciences, 2023, 24, 4262.	1.8	0
790	CACNA1S mutationâ€associated dental anomalies: A calcium channelopathy. Oral Diseases, 0, , .	1.5	3
792	Genetic analyses of DNA repair pathway associated genes implicate new candidate cancer predisposing genes in ancestrally defined ovarian cancer cases. Frontiers in Oncology, 0, 13, .	1.3	1
793	In Silico-Based Structural Evaluation to Categorize the Pathogenicity of Mutations Identified in the RAD Class of Proteins. ACS Omega, 2023, 8, 10266-10277.	1.6	0
794	Imputation-powered whole-exome analysis identifies genes associated with kidney function and disease in the UK Biobank. Nature Communications, 2023, 14, .	5.8	3
795	Whole exome sequencing and rare variant association study to identify genetic modifiers, KLF1 mutations, and a novel double mutation in Thai patients with hemoglobin E/beta-thalassemia. Hematology, 2023, 28, .	0.7	0
798	Whole Genome Analysis of Venous Thromboembolism: the Trans-Omics for Precision Medicine Program. Circulation Genomic and Precision Medicine, 2023, 16, .	1.6	2
799	GNA11 Variants Identified in Patients with Hypercalcemia or Hypocalcemia. Journal of Bone and Mineral Research, 2020, 38, 907-917.	3.1	2
800	Pathogenicity predicting algorithm of single amino acid variants. , 2022, , .		0
801	A Novel Variant in VPS13B Underlying Cohen Syndrome. BioMed Research International, 2023, 2023, 1-7.	0.9	0
802	Prevalence of Monogenic Bone Disorders in a Dutch Cohort of Atypical Femur Fracture Patients. Journal of Bone and Mineral Research, 2020, 38, 896-906.	3.1	1
803	Phenotypic prediction in glutaric aciduria type 1 combining in silico and in vitro modeling with realâ€world data. Journal of Inherited Metabolic Disease, 2023, 46, 391-405.	1.7	5

#	Article	IF	Citations
804	De novo variants implicate chromatin modification, transcriptional regulation, and retinoic acid signaling in syndromic craniosynostosis. American Journal of Human Genetics, 2023, 110, 846-862.	2.6	2
877	PMMVar: Leveraging Multi-level Protein Structures for Enhanced Coding Variant Pathogenicity Prediction. , 2023, , .		0