Predicting the Functional, Molecular, and Phenotypic C Substitutions using Hidden Markov Models

Human Mutation

34, 57-65

DOI: 10.1002/humu.22225

Citation Report

#	Article	IF	Citations
1	dbNSFP v2.0: A Database of Human Non-synonymous SNVs and Their Functional Predictions and Annotations. Human Mutation, 2013, 34, E2393-E2402.	1.1	546
2	Genetic analyses of bone morphogenetic protein 2, 4 and 7 in congenital combined pituitary hormone deficiency. BMC Endocrine Disorders, 2013, 13, 56.	0.9	9
3	Status quo of annotation of human disease variants. BMC Bioinformatics, 2013, 14, 352.	1.2	3
4	Predicting the Functional, Molecular, and Phenotypic Consequences of Amino Acid Substitutions using Hidden Markov Models. Human Mutation, 2013, 34, 57-65.	1.1	1,057
5	De novo mutations in hereditary diffuse leukoencephalopathy with axonal spheroids (HDLS). Neurology, 2013, 81, 2039-2044.	1.5	62
6	Predicting the functional consequences of cancer-associated amino acid substitutions. Bioinformatics, 2013, 29, 1504-1510.	1.8	208
7	Regression Modeling and Meta-Analysis of Diagnostic Accuracy of SNP-Based Pathogenicity Detection Tools for UGT1A1 Gene Mutation. Genetics Research International, 2013, 2013, 1-7.	2.0	3
8	PMD patient mutations reveal a long-distance intronic interaction that regulates PLP1/DM20 alternative splicing. Human Molecular Genetics, 2014, 23, 5464-5478.	1.4	32
9	FH4= STAP1 . Another Gene for Familial Hypercholesterolemia?. Circulation Research, 2014, 115, 534-536.	2.0	4
10	Germline Mutations in MAP3K6 Are Associated with Familial Gastric Cancer. PLoS Genetics, 2014, 10, e1004669.	1.5	57
11	Evolution- and Structure-Based Computational Strategy Reveals the Impact of Deleterious Missense Mutations on MODY 2 (Maturity-Onset Diabetes of the Young, Type 2). Theranostics, 2014, 4, 366-385.	4.6	48
12	Genetic Variations and Diseases in UniProtKB/Swissâ€Prot: The Ins and Outs of Expert Manual Curation. Human Mutation, 2014, 35, 927-935.	1.1	51
13	Kin-Driver: a database of driver mutations in protein kinases. Database: the Journal of Biological Databases and Curation, 2014, 2014, bau104-bau104.	1.4	23
14	Benchmarking mutation effect prediction algorithms using functionally validated cancer-related missense mutations. Genome Biology, 2014, 15, 484.	3.8	117
15	Computational approaches to interpreting genomic sequence variation. Genome Medicine, 2014, 6, 87.	3.6	33
16	The Functional Significance of Common Polymorphisms in Zinc Finger Transcription Factors. G3: Genes, Genomes, Genetics, 2014, 4, 1647-1655.	0.8	9
17	Adenylate cyclase 1 (ADCY1) mutations cause recessive hearing impairment in humans and defects in hair cell function and hearing in zebrafish. Human Molecular Genetics, 2014, 23, 3289-3298.	1.4	48
18	Hoyeraal-Hreidarsson syndrome caused by a germline mutation in the TEL patch of the telomere protein TPP1. Genes and Development, 2014, 28, 2090-2102.	2.7	106

#	Article	IF	Citations
19	First ComprehensiveIn SilicoAnalysis of the Functional and Structural Consequences of SNPs in HumanGalNAc-T1Gene. Computational and Mathematical Methods in Medicine, 2014, 2014, 1-15.	0.7	31
20	SuSPect: Enhanced Prediction of Single Amino Acid Variant (SAV) Phenotype Using Network Features. Journal of Molecular Biology, 2014, 426, 2692-2701.	2.0	189
21	Prediction of pathological mutations in proteins: the challenge of integrating sequence conservation and structure stability principles. Wiley Interdisciplinary Reviews: Computational Molecular Science, 2014, 4, 249-268.	6.2	19
22	Computational Approaches and Resources in Single Amino Acid Substitutions Analysis Toward Clinical Research. Advances in Protein Chemistry and Structural Biology, 2014, 94, 365-423.	1.0	22
23	Exome sequencing identified new mutations in a Marfan syndrome family. Diagnostic Pathology, 2014, 9, 25.	0.9	13
24	Activating Mutations Cluster in the â€œMolecular Brakeâ€•Regions of Protein Kinases and Do Not Associate with Conserved or Catalytic Residues. Human Mutation, 2014, 35, 318-328.	1.1	20
25	Targeted and Genomewide NGS Data Disqualify Mutations inMYO1A, the â€œDFNA48Geneâ€• as a Cause of Deafness. Human Mutation, 2014, 35, 565-570.	1.1	38
26	The Human Gene Mutation Database: building a comprehensive mutation repository for clinical and molecular genetics, diagnostic testing and personalized genomic medicine. Human Genetics, 2014, 133, 1-9.	1.8	1,153
27	Mutation Update and Genotype-Phenotype Correlations of Novel and Previously Described Mutations inTPM2andTPM3Causing Congenital Myopathies. Human Mutation, 2014, 35, 779-790.	1.1	92
28	Somatic aberrations of mismatch repair genes as a cause of microsatelliteâ€unstable cancers. Journal of Pathology, 2014, 234, 548-559.	2.1	134
29	Ranking non-synonymous single nucleotide polymorphisms based on disease concepts. Human Genomics, 2014, 8, 11.	1.4	163
30	Hotspot activating PRKD1 somatic mutations in polymorphous low-grade adenocarcinomas of the salivary glands. Nature Genetics, 2014, 46, 1166-1169.	9.4	188
31	Integrative genomic and transcriptomic characterization of papillary carcinomas of the breast. Molecular Oncology, 2014, 8, 1588-1602.	2.1	49
32	Novel pathogenic variants and genes for myopathies identified by whole exome sequencing. Molecular Genetics & Genomic Medicine, 2015, 3, 283-301.	0.6	43
33	A Novel de novo Mutation in CEACAM16 Associated with Postlingual Hearing Impairment. Molecular Syndromology, 2015, 6, 156-163.	0.3	14
34	A patient with PMP22-related hereditary neuropathy and DBH-gene-related dysautonomia. Journal of Neurology, 2015, 262, 2373-2381.	1.8	8
35	CoagVDb: a comprehensive database for coagulation factors and their associated SAPs. Biological Research, 2015, 48, 35.	1.5	4
36	Targeting tumor suppressor genes for cancer therapy. BioEssays, 2015, 37, 1277-1286.	1.2	65

#	Article	IF	Citations
37	Performance of In Silico Tools for the Evaluation ofUGT1A1Missense Variants. Human Mutation, 2015, 36, 1215-1225.	1.1	21
38	Whole exome sequencing identifies rare protein-coding variants in BehÃ§et's disease. Arthritis and Rheumatology, 2015, 68, n/a-n/a.	2.9	9
39	Analysis of genetic variation and potential applications in genome-scale metabolic modeling. Frontiers in Bioengineering and Biotechnology, 2015, 3, 13.	2.0	30
40	Anatomy of protein disorder, flexibility and disease-related mutations. Frontiers in Molecular Biosciences, 2015, 2, 47.	1.6	16
41	Comprehensive Molecular Diagnosis of a Large Chinese Leber Congenital Amaurosis Cohort. , 2015, 56, 3642.		82
42	Insight into Neutral and Disease-Associated Human Genetic Variants through Interpretable Predictors. PLoS ONE, 2015, 10, e0120729.	1.1	2
43	Proxy Molecular Diagnosis from Whole-Exome Sequencing Reveals Papillon-Lefevre Syndrome Caused by a Missense Mutation in CTSC. PLoS ONE, 2015, 10, e0121351.	1.1	4
44	Identifying Highly Penetrant Disease Causal Mutations Using Next Generation Sequencing: Guide to Whole Process. BioMed Research International, 2015, 2015, 1-16.	0.9	7
45	EpilepsyGene: a genetic resource for genes and mutations related to epilepsy. Nucleic Acids Research, 2015, 43, D893-D899.	6.5	71
46	Oncotator: Cancer Variant Annotation Tool. Human Mutation, 2015, 36, E2423-E2429.	1.1	448
47	Juvenile myelomonocytic leukemia due to a germline CBL Y371C mutation: 35-year follow-up of a large family. Human Genetics, 2015, 134, 775-787.	1.8	21
48	Sequential data selection for predicting the pathogenic effects of sequence variation. , 2015, , .		1
49	EvoTol: a protein-sequence based evolutionary intolerance framework for disease-gene prioritization. Nucleic Acids Research, 2015, 43, e33-e33.	6.5	33
50	Predicting survival in head and neck squamous cell carcinoma from TP53 mutation. Human Genetics, 2015, 134, 497-507.	1.8	31
51	Next-generation sequencing-based molecular diagnosis of 82 retinitis pigmentosa probands from Northern Ireland. Human Genetics, 2015, 134, 217-230.	1.8	85
52	mirTrios: an integrated pipeline for detection of de novo and rare inherited mutations from trios-based next-generation sequencing. Journal of Medical Genetics, 2015, 52, 275-281.	1.5	35
53	Variability in pathogenicity prediction programs: impact on clinical diagnostics. Molecular Genetics & Genomic Medicine, 2015, 3, 99-110.	0.6	44
54	MitImpact: an Exhaustive Collection of Pre-computed Pathogenicity Predictions of Human Mitochondrial Non-synonymous Variants. Human Mutation, 2015, 36, E2413-E2422.	1.1	61

#	Article	IF	Citations
55	The Evaluation of Tools Used to Predict the Impact of Missense Variants Is Hindered by Two Types of Circularity. Human Mutation, 2015, 36, 513-523.	1.1	283
56	Missense variants in CFTR nucleotide-binding domains predict quantitative phenotypes associated with cystic fibrosis disease severity. Human Molecular Genetics, 2015, 24, 1908-1917.	1.4	11
57	Integrated Genomic Analysis Suggests MLL3 Is a Novel Candidate Susceptibility Gene for Familial Nasopharyngeal Carcinoma. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 1222-1228.	1.1	17
58	Origin and dynamics of admixture in Brazilians and its effect on the pattern of deleterious mutations. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 8696-8701.	3.3	206
59	Molecular damage in <scp>F</scp>abry disease: Characterization and prediction of alphaâ€galactosidase <scp>A</scp> pathological mutations. Proteins: Structure, Function and Bioinformatics, 2015, 83, 91-104.	1.5	23
60	Neuronal Activity Promotes Glioma Growth through Neuroligin-3 Secretion. Cell, 2015, 161, 803-816.	13.5	550
61	Aortic Disease Presentation and Outcome Associated With ACTA2 Mutations. Circulation: Cardiovascular Genetics, 2015, 8, 457-464.	5.1	117
62	An integrative approach to predicting the functional effects of non-coding and coding sequence variation. Bioinformatics, 2015, 31, 1536-1543.	1.8	524
63	Mutations in the MYO15A Gene Are a Significant Cause of Nonsyndromic Hearing Loss. Annals of Otology, Rhinology and Laryngology, 2015, 124, 158S-168S.	0.6	42
64	PaPI: pseudo amino acid composition to score human protein-coding variants. BMC Bioinformatics, 2015, 16, 123.	1.2	44
65	On Human Disease-Causing Amino Acid Variants: Statistical Study of Sequence and Structural Patterns. Human Mutation, 2015, 36, 524-534.	1.1	122
66	Systematic Validation of RNF213 Coding Variants in Japanese Patients With Moyamoya Disease. Journal of the American Heart Association, 2015, 4, .	1.6	59
67	Vanno: A Visualization-Aided Variant Annotation Tool. Human Mutation, 2015, 36, 167-174.	1.1	6
68	Bioinformatic Analysis of GJB2 Gene Missense Mutations. Cell Biochemistry and Biophysics, 2015, 71, 1623-1642.	0.9	16
69	Ensembl 2015. Nucleic Acids Research, 2015, 43, D662-D669.	6.5	1,145
70	Molecular Methods for Diagnosis of Genetic Diseases Involving the Immune System. , 2016, , 5-18.		0
71	Practical aspects of NGS-based pathways analysis for personalized cancer science and medicine. Oncotarget, 2016, 7, 52493-52516.	0.8	15
72	The New Immortalized Uroepithelial Cell Line HBLAK Contains Defined Genetic Aberrations Typical of Early Stage Urothelial Tumors. Bladder Cancer, 2016, 2, 449-463.	0.2	34

#	Article	IF	Citations
73	A Survey of Computational Tools to Analyze and Interpret Whole Exome Sequencing Data. International Journal of Genomics, 2016, 2016, 1-16.	0.8	37
74	Impacts of Nonsynonymous Single Nucleotide Polymorphisms of Adiponectin Receptor 1 Gene on Corresponding Protein Stability: A Computational Approach. BioMed Research International, 2016, 2016, 1-12.	0.9	15
75	Eight Mutations of Three Genes (EDA, EDAR, and WNT10A) Identified in Seven Hypohidrotic Ectodermal Dysplasia Patients. Genes, 2016, 7, 65.	1.0	25
76	Alpha Helices Are More Robust to Mutations than Beta Strands. PLoS Computational Biology, 2016, 12, e1005242.	1.5	85
77	A profile-based method for identifying functional divergence of orthologous genes in bacterial genomes. Bioinformatics, 2016, 32, 3566-3574.	1.8	25
78	Massively parallel sequencing of phyllodes tumours of the breast reveals actionable mutations, and <scp>TERT</scp> promoter hotspot mutations and TERT gene amplification as likely drivers of progression. Journal of Pathology, 2016, 238, 508-518.	2.1	102
79	Homozygous nonsense mutation inSGCAis a common cause of limb-girdle muscular dystrophy in Assiut, Egypt. Muscle and Nerve, 2016, 54, 690-695.	1.0	12
80	Complex mode of inheritance in holoprosencephaly revealed by whole exome sequencing. Clinical Genetics, 2016, 89, 659-668.	1.0	36
81	A new tool for prioritization of sequence variants from whole exome sequencing data. Source Code for Biology and Medicine, 2016, 11, 10.	1.7	9
82	Whole exome sequencing reveals a C-terminal germline variant in CEBPA-associated acute myeloid leukemia: 45-year follow up of a large family. Haematologica, 2016, 101, 846-852.	1.7	42
83	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
84	Pathogenic FBN1 variants in familial thoracic aortic aneurysms and dissections. Clinical Genetics, 2016, 89, 719-723.	1.0	26
85	dbWGFP: a database and web server of human whole-genome single nucleotide variants and their functional predictions. Database: the Journal of Biological Databases and Curation, 2016, 2016, baw024.	1.4	27
86	Natural resistance to Meningococcal Disease related to CFH loci: Meta-analysis of genome-wide association studies. Scientific Reports, 2016, 6, 35842.	1.6	33
87	<scp>IGSF</scp> 10 mutations dysregulate gonadotropinâ€releasing hormone neuronal migration resulting in delayed puberty. EMBO Molecular Medicine, 2016, 8, 626-642.	3.3	109
88	iCAGES: integrated CAncer GEnome Score for comprehensively prioritizing driver genes in personal cancer genomes. Genome Medicine, 2016, 8, 135.	3.6	45
89	iFish: predicting the pathogenicity of human nonsynonymous variants using gene-specific/family-specific attributes and classifiers. Scientific Reports, 2016, 6, 31321.	1.6	24
90	Wbp2 is required for normal glutamatergic synapses in the cochlea and is crucial for hearing. EMBO Molecular Medicine, 2016, 8, 191-207.	3.3	41

#	Article	IF	Citations
91	IMPACT: a whole-exome sequencing analysis pipeline for integrating molecular profiles with actionable therapeutics in clinical samples. Journal of the American Medical Informatics Association: JAMIA, 2016, 23, 721-730.	2.2	38
92	Common and Rare Genetic Variation in CCR2 , CCR5 , or CX3CR1 and Risk of Atherosclerotic Coronary Heart Disease and Glucometabolic Traits. Circulation: Cardiovascular Genetics, 2016, 9, 250-258.	5.1	20
93	Predicting pathogenic single nucleotide variants through a comprehensive analysis on multiple level features. Chemometrics and Intelligent Laboratory Systems, 2016, 156, 224-230.	1.8	0
94	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178.	0.7	32
95	Expanding the mutation spectrum in 130 probands with ARPKD: identification of 62 novel PKHD1 mutations by sanger sequencing and MLPA analysis. Journal of Human Genetics, 2016, 61, 811-821.	1.1	27
96	Mutational screening in patients with profound sensorineural hearing loss and neurodevelopmental delay: Description of a novel m.3861AÂ>ÂC mitochondrial mutation in the MT-ND1 gene. Biochemical and Biophysical Research Communications, 2016, 474, 702-708.	1.0	4
97	Genetic alterations of triple negative breast cancer by targeted next-generation sequencing and correlation with tumor morphology. Modern Pathology, 2016, 29, 476-488.	2.9	95
98	Mutations inPOMGNT1cause non-syndromic retinitis pigmentosa. Human Molecular Genetics, 2016, 25, 1479-1488.	1.4	42
99	Hoyeraal-Hreidarsson Syndrome due to PARN Mutations: Fourteen Years of Follow-Up. Pediatric Neurology, 2016, 56, 62-68.e1.	1.0	29
100	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
101	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
102	Increased burden of ultra-rare protein-altering variants among 4,877 individuals with schizophrenia. Nature Neuroscience, 2016, 19, 1433-1441.	7.1	427
103	Mutation analysis by direct and whole exome sequencing in familial and sporadic tooth agenesis. International Journal of Molecular Medicine, 2016, 38, 1338-1348.	1.8	22
104	The Ensembl Variant Effect Predictor. Genome Biology, 2016, 17, 122.	3.8	5,181
105	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
106	Computational assessment of feature combinations for pathogenic variant prediction. Molecular Genetics & Genomic Medicine, 2016, 4, 431-446.	0.6	13
107	Importance of Genetic Studies in Consanguineous Populations for the Characterization of Novel Human Gene Functions. Annals of Human Genetics, 2016, 80, 187-196.	0.3	41
108	Disrupted lymphocyte homeostasis in hepatitisâ€associated acquired aplastic anemia is associated with short telomeres. American Journal of Hematology, 2016, 91, 243-247.	2.0	11

#	Article	IF	Citations
109	Mutational Spectrum in Holoprosencephaly Shows That FGF is a New Major Signaling Pathway. Human Mutation, 2016, 37, 1329-1339.	1.1	56
110	Threeâ€dimensional modelling identifies novel genetic dependencies associated with breast cancer progression in the isogenic <scp>MCF10</scp> model. Journal of Pathology, 2016, 240, 315-328.	2.1	35
111	Inheritance-mode specific pathogenicity prioritization (ISPP) for human protein coding genes. Bioinformatics, 2016, 32, 3065-3071.	1.8	11
112	A genetic network that suppresses genome rearrangements in Saccharomyces cerevisiae and contains defects in cancers. Nature Communications, 2016, 7, 11256.	5.8	36
113	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
114	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
115	Mutations in Human Accelerated Regions Disrupt Cognition and Social Behavior. Cell, 2016, 167, 341-354.e12.	13.5	280
116	Genetic Factors of the Disease Course After Sepsis: Rare Deleterious Variants Are Predictive. EBioMedicine, 2016, 12, 227-238.	2.7	34
117	Association of Exome Sequences With Cardiovascular Traits Among Blacks in the Jackson Heart Study. Circulation: Cardiovascular Genetics, 2016, 9, 368-374.	5.1	8
118	DNA Mismatch Repair Deficiency in Rectal Cancer: Benchmarking Its Impact on Prognosis, Neoadjuvant Response Prediction, and Clinical Cancer Genetics. Journal of Clinical Oncology, 2016, 34, 3039-3046.	0.8	86
119	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
120	Truncating and missense PPM1D mutations in earlyâ€onset and/or familial/hereditary prostate cancer patients. Genes Chromosomes and Cancer, 2016, 55, 954-961.	1.5	15
121	Targeted exome sequencing resolves allelic and the genetic heterogeneity in the genetic diagnosis of nephronophthisis-related ciliopathy. Experimental and Molecular Medicine, 2016, 48, e251-e251.	3.2	26
122	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
123	ApoCanD: Database of human apoptotic proteins in the context of cancer. Scientific Reports, 2016, 6, 20797.	1.6	9
124	KinMutRF: a random forest classifier of sequence variants in the human protein kinase superfamily. BMC Genomics, 2016, 17, 396.	1.2	11
125	Self-regulation of functional pathways by motifs inside the disordered tails of beta-catenin. BMC Genomics, 2016, 17, 484.	1.2	10
126	Exome sequencing reveals a novel CWF19L1 mutation associated with intellectual disability and cerebellar atrophy. American Journal of Medical Genetics, Part A, 2016, 170, 1502-1509.	0.7	13

#	Article	IF	Citations
127	Tools for Predicting the Functional Impact of Nonsynonymous Genetic Variation. Genetics, 2016, 203, 635-647.	1.2	84
128	Molecular characterization, homology modeling and docking studies of the R2787H missense variation in BRCA2 gene: Association with breast cancer. Journal of Theoretical Biology, 2016, 403, 188-196.	0.8	5
129	The structural effects of mutations can aid in differential phenotype prediction of beta-myosin heavy chain (Myosin-7) missense variants. Bioinformatics, 2016, 32, 2947-2955.	1.8	9
130	Delineating the relationship between amyotrophic lateral sclerosis and frontotemporal dementia: Sequence and structure-based predictions. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2016, 1862, 1742-1754.	1.8	22
131	Uterine adenosarcomas are mesenchymal neoplasms. Journal of Pathology, 2016, 238, 381-388.	2.1	94
132	CerealsDB 3.0: expansion of resources and data integration. BMC Bioinformatics, 2016, 17, 256.	1.2	42
133	Mutation analysis of Swedish haemophilia B families â€“ high frequency of unique mutations. Haemophilia, 2016, 22, 440-445.	1.0	17
134	Web-based Gene Pathogenicity Analysis (WGPA): a web platform to interpret gene pathogenicity from personal genome data. Bioinformatics, 2016, 32, 635-637.	1.8	1
135	First independent replication of the involvement of LARS2 in Perrault syndrome by whole-exome sequencing of an Italian family. Journal of Human Genetics, 2016, 61, 295-300.	1.1	34
136	Developing maps of fitness consequences for plant genomes. Current Opinion in Plant Biology, 2016, 30, 101-107.	3.5	13
137	Perspectives in Polycystic Ovary Syndrome: From Hair to Eternity. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 759-768.	1.8	71
138	Assessing the Pathogenicity of Insertion and Deletion Variants with the Variant Effect Scoring Tool (VESTâ€indel). Human Mutation, 2016, 37, 28-35.	1.1	101
139	wKinMut-2: Identification and Interpretation of Pathogenic Variants in Human Protein Kinases. Human Mutation, 2016, 37, 36-42.	1.1	10
140	Targeted capture massively parallel sequencing analysis of LCIS and invasive lobular cancer: Repertoire of somatic genetic alterations and clonal relationships. Molecular Oncology, 2016, 10, 360-370.	2.1	41
141	TECPR2 mutations cause a new subtype of familial dysautonomia like hereditary sensory autonomic neuropathy with intellectual disability. European Journal of Paediatric Neurology, 2016, 20, 69-79.	0.7	45
142	The Genomic Landscape of Male Breast Cancers. Clinical Cancer Research, 2016, 22, 4045-4056.	3.2	119
143	CDH1/E-cadherin and solid tumors. An updated gene-disease association analysis using bioinformatics tools. Computational Biology and Chemistry, 2016, 60, 9-20.	1.1	7
144	Machine Learning in Genomic Medicine: A Review of Computational Problems and Data Sets. Proceedings of the IEEE, 2016, 104, 176-197.	16.4	186

#	Article	IF	Citations
145	Genes with de novo mutations are shared by four neuropsychiatric disorders discovered from NPdenovo database. Molecular Psychiatry, 2016, 21, 290-297.	4.1	167
146	Functional analysis reveals that RBM10 mutations contribute to lung adenocarcinoma pathogenesis by deregulating splicing. Scientific Reports, 2017, 7, 40488.	1.6	58
147	Ecological genomics of tropical trees: how local population size and allelic diversity of resistance genes relate to immune responses, cosusceptibility to pathogens, and negative density dependence. Molecular Ecology, 2017, 26, 2498-2513.	2.0	50
148	Comprehensive Computational Analysis of GWAS Loci Identifies CCR2 as a Candidate Gene for Celiac Disease Pathogenesis. Journal of Cellular Biochemistry, 2017, 118, 2193-2207.	1.2	17
150	Exome sequencing identifies SLC26A4, GJB2, SCARB2 and DUOX2 mutations in 2 siblings with Pendred syndrome in a Malaysian family. Orphanet Journal of Rare Diseases, 2017, 12, 40.	1.2	7
151	Annotating Mutational Effects on Proteins and Protein Interactions: Designing Novel and Revisiting Existing Protocols. Methods in Molecular Biology, 2017, 1550, 235-260.	0.4	18
152	Drug Response Prediction as a Link Prediction Problem. Scientific Reports, 2017, 7, 40321.	1.6	64
153	Functional annotation of sixty-five type-2 diabetes risk SNPs and its application in risk prediction. Scientific Reports, 2017, 7, 43709.	1.6	4
154	The Landscape of Somatic Genetic Alterations in Metaplastic Breast Carcinomas. Clinical Cancer Research, 2017, 23, 3859-3870.	3.2	129
155	Clinical, Molecular, and Computational Analysis in Patients With a Novel Double Mutation and a New Synonymous Variant in MeCP2: Report of the First Missense Mutation Within the AT-hook1 Cluster in Rett Syndrome. Journal of Child Neurology, 2017, 32, 694-703.	0.7	8
156	Diagnosis of CoPAN by whole exome sequencing: Waking up a sleeping tiger's eye. American Journal of Medical Genetics, Part A, 2017, 173, 1878-1886.	0.7	40
157	Computational prediction and analysis of deleterious cancer associated missense mutations in DYNC1H1. Molecular and Cellular Probes, 2017, 34, 21-29.	0.9	16
158	Predicting the effect of non synonymous SNPs in bovine TLR4 gene. Gene Reports, 2017, 6, 32-35.	0.4	4
159	Exploring the global landscape of genetic variation in coagulation factor XI deficiency. Blood, 2017, 130, e1-e6.	0.6	41
160	HIPred: an integrative approach to predicting haploinsufficient genes. Bioinformatics, 2017, 33, 1751-1757.	1.8	36
161	Myxoid fibroadenomas differ from conventional fibroadenomas: a hypothesisâ€generating study. Histopathology, 2017, 71, 626-634.	1.6	26
162	Missense variant pathogenicity predictors generalize well across a range of functionâ€specific prediction challenges. Human Mutation, 2017, 38, 1092-1108.	1.1	39
163	Comparison and optimization of in silico algorithms for predicting the pathogenicity of sodium channel variants in epilepsy. Epilepsia, 2017, 58, 1190-1198.	2.6	14

#	Article	IF	Citations
164	Bioinformatics in translational drug discovery. Bioscience Reports, 2017, 37, .	1.1	68
165	Matching phenotypes to whole genomes: Lessons learned from four iterations of the personal genome project community challenges. Human Mutation, 2017, 38, 1266-1276.	1.1	14
166	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
167	Compound heterozygous KCNQ1 mutations (A300T/P535T) in a child with sudden unexplained death: Insights into possible molecular mechanisms based on protein modeling. Gene, 2017, 627, 40-48.	1.0	7
168	A Computational Approach to Identify a Potential Alternative Drug With Its Positive Impact Toward PMP22. Journal of Cellular Biochemistry, 2017, 118, 3730-3743.	1.2	9
169	Data resources for the identification and interpretation of actionable mutations by clinicians. Annals of Oncology, 2017, 28, 946-957.	0.6	20
170	Whole exome sequencing with genomic triangulation implicatesCDH2-encoded N-cadherin as a novel pathogenic substrate for arrhythmogenic cardiomyopathy. Congenital Heart Disease, 2017, 12, 226-235.	0.0	46
171	The Human Gene Mutation Database: towards a comprehensive repository of inherited mutation data for medical research, genetic diagnosis and next-generation sequencing studies. Human Genetics, 2017, 136, 665-677.	1.8	1,106
172	Short stature and hypoparathyroidism in a child with Kenny-Caffey syndrome type 2 due to a novel mutation in FAM111A gene. International Journal of Pediatric Endocrinology (Springer), 2017, 2017, 1.	1.6	27
173	Genetic Heterogeneity in Therapy-NaÃ¯ve Synchronous Primary Breast Cancers and Their Metastases. Clinical Cancer Research, 2017, 23, 4402-4415.	3.2	91
174	Advances and challenges in targeting FGFR signalling in cancer. Nature Reviews Cancer, 2017, 17, 318-332.	12.8	523
175	Compromised BRCA1â€“PALB2 interaction is associated with breast cancer risk. Oncogene, 2017, 36, 4161-4170.	2.6	71
176	PERCH: A Unified Framework for Disease Gene Prioritization. Human Mutation, 2017, 38, 243-251.	1.1	119
177	The Rare-Variant Generalized Disequilibrium Test for Association Analysis of Nuclear and Extended Pedigrees with Application to Alzheimer Disease WGS Data. American Journal of Human Genetics, 2017, 100, 193-204.	2.6	26
178	Enrichment of deleterious variants of mitochondrial DNA polymerase gene (POLG1) in bipolar disorder. Psychiatry and Clinical Neurosciences, 2017, 71, 518-529.	1.0	29
179	Overexpressed somatic alleles are enriched in functional elements in Breast Cancer. Scientific Reports, 2017, 7, 8287.	1.6	3
180	Phyllodes tumors with and without fibroadenoma-like areas display distinct genomic features and may evolve through distinct pathways. Npj Breast Cancer, 2017, 3, 40.	2.3	52
181	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	13.7	1,099

#	Article	IF	Citations
182	CRIMEtoYHU: a new web tool to develop yeast-based functional assays for characterizing cancer-associated missense variants. FEMS Yeast Research, 2017, 17, .	1.1	4
183	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
184	Identification of ASAH1 as a susceptibility gene for familial keloids. European Journal of Human Genetics, 2017, 25, 1155-1161.	1.4	19
185	Annotating pathogenic non-coding variants in genic regions. Nature Communications, 2017, 8, 236.	5.8	122
186	Decoding disease-causing mechanisms of missense mutations from supramolecular structures. Scientific Reports, 2017, 7, 8541.	1.6	26
187	Genomic landscape of human erythroleukemia K562 cell line, as determined by next-generation sequencing and cytogenetics. Acta Haematologica Polonica, 2017, 48, 343-349.	0.1	3
188	Identification and characterization of a novel recessive KCNQ1 mutation associated with Romano-Ward Long-QT syndrome in two Iranian families. Journal of Electrocardiology, 2017, 50, 912-918.	0.4	6
189	In Silico Systems Biology Analysis of Variants of Uncertain Significance in Lynch Syndrome Supports the Prioritization of Functional Molecular Validation. Cancer Prevention Research, 2017, 10, 580-587.	0.7	9
190	A Functional Assay for Sick Sinus Syndrome Genetic Variants. Cellular Physiology and Biochemistry, 2017, 42, 2021-2029.	1.1	12
191	Mild case of congenital ichthyosiform erythroderma with periodic exacerbation: Novel mutations in <scp>ABCA</scp>12 and upregulation of calprotectin in the epidermis. Journal of Dermatology, 2017, 44, e282-e283.	0.6	3
192	Settling the score: variant prioritization and Mendelian disease. Nature Reviews Genetics, 2017, 18, 599-612.	7.7	213
193	NSD1- and NSD2-damaging mutations define a subset of laryngeal tumors with favorable prognosis. Nature Communications, 2017, 8, 1772.	5.8	40
194	Accuracy of Next-Generation Sequencing for Molecular Diagnosis in Patients With Infantile Nystagmus Syndrome. JAMA Ophthalmology, 2017, 135, 1376.	1.4	43
195	Functional analyses of a novel missense and other mutations of the vitamin D receptor in association with alopecia. Scientific Reports, 2017, 7, 5102.	1.6	14
196	Exome sequencing identifies targets in the treatment-resistant ophthalmoplegic subphenotype of myasthenia gravis. Neuromuscular Disorders, 2017, 27, 816-825.	0.3	12
197	GTB â€“ an online genome tolerance browser. BMC Bioinformatics, 2017, 18, 20.	1.2	3
198	Sequence variations of the EGR4 gene in Korean men with spermatogenesis impairment. BMC Medical Genetics, 2017, 18, 47.	2.1	4
199	Variant effect prediction tools assessed using independent, functional assay-based datasets: implications for discovery and diagnostics. Human Genomics, 2017, 11, 10.	1.4	68

#	Article	IF	Citations
200	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
201	DEOGEN2: prediction and interactive visualization of single amino acid variant deleteriousness in human proteins. Nucleic Acids Research, 2017, 45, W201-W206.	6.5	114
202	Impact of clinical exomes in neurodevelopmental and neurometabolic disorders. Molecular Genetics and Metabolism, 2017, 121, 297-307.	0.5	50
203	Centroid tracking based dynamic hand gesture recognition using discrete Hidden Markov Models. Neurocomputing, 2017, 228, 79-83.	3.5	22
204	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
205	EFHC1 variants in juvenile myoclonic epilepsy: reanalysis according to NHGRI and ACMG guidelines for assigning disease causality. Genetics in Medicine, 2017, 19, 144-156.	1.1	34
206	Role of E542 and E545 missense mutations of PIK3CA in breast cancer: a comparative computational approach. Journal of Biomolecular Structure and Dynamics, 2017, 35, 2745-2757.	2.0	33
207	Investigating the Molecular Mechanisms Behind Uncharacterized Cysteine Losses from Prediction of Their Oxidation State. Human Mutation, 2017, 38, 86-94.	1.1	4
208	Phenotypic Variability and mTOR Pathway Gene Aberrations in Familial Tuberous Sclerosis. Journal of Pediatric Neurology, 2017, 15, 316-324.	0.0	0
209	High Prevalence of Diabetes-Predisposing Variants in MODY Genes Among Danish Women With Gestational Diabetes Mellitus. Journal of the Endocrine Society, 2017, 1, 681-690.	0.1	32
210	Spatial distribution of disease-associated variants in three-dimensional structures of protein complexes. Oncogenesis, 2017, 6, e380-e380.	2.1	20
211	Development of pathogenicity predictors specific for variants that do not comply with clinical guidelines for the use of computational evidence. BMC Genomics, 2017, 18, 569.	1.2	14
212	TSC1 Mutations in Keratoconus Patients With or Without Tuberous Sclerosis. , 2017, 58, 6462.		10
213	Associating mutations causing cystinuria with disease severity with the aim of providing precision medicine. BMC Genomics, 2017, 18, 550.	1.2	16
214	Fibrinogen as a Pleiotropic Protein Causing Human Diseases: The Mutational Burden of AÎ±, BÎ², and Î³ Chains. International Journal of Molecular Sciences, 2017, 18, 2711.	1.8	36
215	Exome Sequencing Identifies a Novel MAP3K14 Mutation in Recessive Atypical Combined Immunodeficiency. Frontiers in Immunology, 2017, 8, 1624.	2.2	16
216	Benchmarking distributed data warehouse solutions for storing genomic variant information. Database: the Journal of Biological Databases and Curation, 2017, 2017, .	1.4	8
217	Impact of genetic variation on three dimensional structure and function of proteins. PLoS ONE, 2017, 12, e0171355.	1.1	55

#	Article	IF	Citations
218	Determining the role of missense mutations in the POU domain of HNF1A that reduce the DNA-binding affinity: A computational approach. PLoS ONE, 2017, 12, e0174953.	1.1	43
219	Alport syndrome cold cases: Missing mutations identified by exome sequencing and functional analysis. PLoS ONE, 2017, 12, e0178630.	1.1	16
220	High proportion of genetic cases in patients with advanced cardiomyopathy including a novel homozygous Plakophilin 2-gene mutation. PLoS ONE, 2017, 12, e0189489.	1.1	33
221	Functional significance of rare neuroligin 1 variants found in autism. PLoS Genetics, 2017, 13, e1006940.	1.5	76
222	Identification of GAA variants through whole exome sequencing targeted to a cohort of 606 patients with unexplained limb-girdle muscle weakness. Orphanet Journal of Rare Diseases, 2017, 12, 173.	1.2	21
223	Evaluation of in silico algorithms for use with ACMG/AMP clinical variant interpretation guidelines. Genome Biology, 2017, 18, 225.	3.8	185
224	Variant Ranker: a web-tool to rank genomic data according to functional significance. BMC Bioinformatics, 2017, 18, 341.	1.2	21
225	An integrative approach to predicting the functional effects of small indels in non-coding regions of the human genome. BMC Bioinformatics, 2017, 18, 442.	1.2	34
226	Elucidation of MRAS-mediated Noonan syndrome with cardiac hypertrophy. JCI Insight, 2017, 2, e91225.	2.3	66
227	Sequencing of Lynch syndrome tumors reveals the importance of epigenetic alterations. Oncotarget, 2017, 8, 108020-108030.	0.8	18
228	Genetic Biopsy for Prediction of Surveillance Intervals after Endoscopic Resection of Colonic Polyps: Results of the GENESIS Study. United European Gastroenterology Journal, 2018, 6, 290-299.	1.6	8
229	Structural dynamics is a determinant of the functional significance of missense variants. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 4164-4169.	3.3	76
230	Comprehensive Characterization of Cancer Driver Genes and Mutations. Cell, 2018, 173, 371-385.e18.	13.5	1,670
231	In silicocharacterization of functional single nucleotide polymorphisms of folate pathway genes. Annals of Human Genetics, 2018, 82, 186-199.	0.3	6
232	m6ASNP: a tool for annotating genetic variants by m6A function. GigaScience, 2018, 7, .	3.3	36
233	VarCards: an integrated genetic and clinical database for coding variants in the human genome. Nucleic Acids Research, 2018, 46, D1039-D1048.	6.5	148
234	Germline but not somatic de novo mutations are common in human congenital diaphragmatic hernia. Birth Defects Research, 2018, 110, 610-617.	0.8	12
235	Allele-specific SHAPE-MaP assessment of the effects of somatic variation and protein binding on mRNA structure. Rna, 2018, 24, 513-528.	1.6	20

#	Article	IF	Citations
236	A phenotype centric benchmark of variant prioritisation tools. Npj Genomic Medicine, 2018, 3, 5.	1.7	39
237	Bioinformatics analysis of non-synonymous variants in the KLF genes related to cardiac diseases. Gene, 2018, 650, 68-76.	1.0	5
238	First report of an unusual novel double mutation affecting the transcription repression domain of MeCP2 and causing a severe phenotype of Rett syndrome: Molecular analyses and computational investigation. Biochemical and Biophysical Research Communications, 2018, 497, 93-101.	1.0	2
239	ConsensusDriver Improves upon Individual Algorithms for Predicting Driver Alterations in Different Cancer Types and Individual Patients. Cancer Research, 2018, 78, 290-301.	0.4	20
240	â€œLowe syndrome: A particularly severe phenotype without clinical kidney involvementâ€• American Journal of Medical Genetics, Part A, 2018, 176, 460-464.	0.7	4
241	Tools for protein science. Protein Science, 2018, 27, 6-9.	3.1	2
242	Geneâ€Specific Variant Classifier (DPYDâ€Varifier) to Identify Deleterious Alleles of Dihydropyrimidine Dehydrogenase. Clinical Pharmacology and Therapeutics, 2018, 104, 709-718.	2.3	43
243	The use of whole exome sequencing and murine patient derived xenografts as a method of chemosensitivity testing in sarcoma. Clinical Sarcoma Research, 2018, 8, 4.	2.3	4
244	A novel, homozygous mutation in desert hedgehog (DHH) in a 46, XY patient with dysgenetic testes presenting with primary amenorrhoea: a case report. International Journal of Pediatric Endocrinology (Springer), 2018, 2018, 2.	1.6	14
245	Germline variants in pancreatic cancer patients with a personal or family history of cancer fulfilling the revised Bethesda guidelines. Journal of Gastroenterology, 2018, 53, 1159-1167.	2.3	7
246	MoBiDiC Prioritization Algorithm, a Free, Accessible, and Efficient Pipeline for Single-Nucleotide Variant Annotation and Prioritization for Next-Generation Sequencing Routine Molecular Diagnosis. Journal of Molecular Diagnostics, 2018, 20, 465-473.	1.2	13
247	Novel biallelic <scp>SZT2</scp> mutations in 3 cases of earlyâ€onset epileptic encephalopathy. Clinical Genetics, 2018, 93, 266-274.	1.0	25
248	m6AVar: a database of functional variants involved in m6A modification. Nucleic Acids Research, 2018, 46, D139-D145.	6.5	181
249	HUMA: A platform for the analysis of genetic variation in humans. Human Mutation, 2018, 39, 40-51.	1.1	8
250	Mutations in AIFM1 cause an X-linked childhood cerebellar ataxia partially responsive to riboflavin. European Journal of Paediatric Neurology, 2018, 22, 93-101.	0.7	37
251	Analysis of clinically relevant somatic mutations in high-risk head and neck cutaneous squamous cell carcinoma. Modern Pathology, 2018, 31, 275-287.	2.9	37
252	Insights From Molecular Characterization of Adult Patients of Families With Multigenerational Diabetes. Diabetes, 2018, 67, 137-145.	0.3	23
253	In Silico Approach to Investigate the Structural and Functional Attributes of Familial Hypercholesterolemia Variants Reported in the Saudi Population. Journal of Computational Biology, 2018, 25, 170-181.	0.8	4

#	Article	IF	Citations
254	Combined Small Cell Carcinoma of the Lung: Is It aÂSingle Entity?. Journal of Thoracic Oncology, 2018, 13, 237-245.	0.5	47
255	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
256	Phenotypic expression of a novel desmin gene mutation: hypertrophic cardiomyopathy followed by systemic myopathy. Journal of Human Genetics, 2018, 63, 249-254.	1.1	24
257	Mismatch Repair Deficiency in High-Grade Meningioma: A Rare but Recurrent Event Associated With Dramatic Immune Activation and Clinical Response to PD-1 Blockade. JCO Precision Oncology, 2018, 2018, 1-12.	1.5	35
258	dbCPM: a manually curated database for exploring the cancer passenger mutations. Briefings in Bioinformatics, 2018, , .	3.2	10
259	Personalized Clinical Decision Making Through Implementation of a Molecular Tumor Board: A German Single-Center Experience. JCO Precision Oncology, 2018, 2, 1-16.	1.5	41
260	Detection and benchmarking of somatic mutations in cancer genomes using RNA-seq data. PeerJ, 2018, 6, e5362.	0.9	42
261	Somatic Sequencing Identifies Trametinib-Responsive Myelodysplastic Syndrome and Finds Acquired Clonal Hematopoiesis of Indeterminate Potential. JCO Precision Oncology, 2018, 2, 0-0.	1.5	0
262	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
263	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
264	Molecular Analysis of KCNQ1, KCNH2 and SCN5A Genes in Iranian Patients with Long QT Syndrome. Journal of Molecular and Genetic Medicine: an International Journal of Biomedical Research, 2018, 12, .	0.1	0
265	Personal Genome Project UK (PGP-UK): a research and citizen science hybrid project in support of personalized medicine. BMC Medical Genomics, 2018, 11, 108.	0.7	34
266	Genetic heterogeneity and actionable mutations in HER2-positive primary breast cancers and their brain metastases. Oncotarget, 2018, 9, 20617-20630.	0.8	36
267	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
268	Identification of novel compound heterozygous SPG7 mutations-related hereditary spastic paraplegia in a Chinese family: a case report. BMC Neurology, 2018, 18, 196.	0.8	1
269	Computational Methods for the Pharmacogenetic Interpretation of Next Generation Sequencing Data. Frontiers in Pharmacology, 2018, 9, 1437.	1.6	62
270	Loss of the FAT1 Tumor Suppressor Promotes Resistance to CDK4/6 Inhibitors via the Hippo Pathway. Cancer Cell, 2018, 34, 893-905.e8.	7.7	307
271	Exhaustive non-synonymous variants functionality prediction enables high resolution characterization of the neurofibromin architecture. EBioMedicine, 2018, 36, 508-516.	2.7	1

#	Article	IF	Citations
272	ENTPRISE-X: Predicting disease-associated frameshift and nonsense mutations. PLoS ONE, 2018, 13, e0196849.	1.1	20
273	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
274	Prioritization of Variants Detected by Next Generation Sequencing According to the Mutation Tolerance and Mutational Architecture of the Corresponding Genes. International Journal of Molecular Sciences, 2018, 19, 1584.	1.8	16
275	Defining the molecular signatures of Achilles tendinopathy and anterior cruciate ligament ruptures: A whole-exome sequencing approach. PLoS ONE, 2018, 13, e0205860.	1.1	16
276	Novel sequence variants in the TLR6 gene associated with advanced breast cancer risk in the Saudi Arabian population. PLoS ONE, 2018, 13, e0203376.	1.1	16
277	BRCA-analyzer: Automatic workflow for processing NGS reads of BRCA1 and BRCA2 genes. Computational Biology and Chemistry, 2018, 77, 297-306.	1.1	10
278	Syndromic hearing loss molecular diagnosis: Application of massive parallel sequencing. Hearing Research, 2018, 370, 181-188.	0.9	6
279	Identification of therapeutic targets in chordoma through comprehensive genomic and transcriptomic analyses. Journal of Physical Education and Sports Management, 2018, 4, a003418.	0.5	13
280	A Bayesian framework for efficient and accurate variant prediction. PLoS ONE, 2018, 13, e0203553.	1.1	12
281	Differences in clinical characteristics and mutational pattern between synchronous and metachronous colorectal liver metastases. Cancer Management and Research, 2018, Volume 10, 2871-2881.	0.9	11
282	In silico Prioritization of Transporterâ€“Drug Relationships From Drug Sensitivity Screens. Frontiers in Pharmacology, 2018, 9, 1011.	1.6	23
283	Positive selection drives the evolution of endocrine regulatory bone morphogenetic protein system in mammals. Oncotarget, 2018, 9, 18435-18445.	0.8	15
284	Genomic and expression profiling reveal molecular heterogeneity of disseminated tumor cells in bone marrow of early breast cancer. Npj Breast Cancer, 2018, 4, 31.	2.3	23
285	Correlation of genomic alterations between tumor tissue and circulating tumor DNA by next-generation sequencing. Journal of Cancer Research and Clinical Oncology, 2018, 144, 2167-2175.	1.2	9
286	The investigation for potential modifier genes in patients with neurofibromatosis type 1 based on next-generation sequencing. OncoTargets and Therapy, 2018, Volume 11, 919-932.	1.0	5
287	Novel missense and 3â€²-UTR splice site variants in LHFPL5 cause autosomal recessive nonsyndromic hearing impairment. Journal of Human Genetics, 2018, 63, 1099-1107.	1.1	3
288	A Multi-Model Based Approach for Driver Missense Identification. , 2018, , .		0
289	Computational insights of K1444N substitution in GAP-related domain of NF1 gene associated with neurofibromatosis type 1 disease: a molecular modeling and dynamics approach. Metabolic Brain Disease, 2018, 33, 1443-1457.	1.4	24

#	Article	IF	Citations
290	Systematic pan-cancer analysis of somatic allele frequency. Scientific Reports, 2018, 8, 7735.	1.6	21
291	Computational analysis of non-synonymous SNPs in bovine Mx1 gene. Gene Reports, 2018, 11, 294-298.	0.4	0
292	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. Neuron, 2018, 99, 302-314.e4.	3.8	112
293	Genetic Mutations in a Patient with Chronic Myeloid Leukemia Showing Blast Crisis 10 Years After Presentation. Anticancer Research, 2018, 38, 3961-3966.	0.5	5
294	Accurate prediction of functional, structural, and stability changes in PITX2 mutations using in silico bioinformatics algorithms. PLoS ONE, 2018, 13, e0195971.	1.1	35
295	Whole exome sequencing analysis in severe chronic obstructive pulmonary disease. Human Molecular Genetics, 2018, 27, 3801-3812.	1.4	32
296	Organoid Models of Human Liver Cancers Derived from Tumor Needle Biopsies. Cell Reports, 2018, 24, 1363-1376.	2.9	288
297	Pan-Cancer Analysis Reveals the Functional Importance of Protein Lysine Modification in Cancer Development. Frontiers in Genetics, 2018, 9, 254.	1.1	39
298	Targeted next-generation sequencing as a comprehensive test for Mendelian diseases: a cohort diagnostic study. Scientific Reports, 2018, 8, 11646.	1.6	17
299	Detection of variants in dystroglycanopathy-associated genes through the application of targeted whole-exome sequencing analysis to a large cohort of patients with unexplained limb-girdle muscle weakness. Skeletal Muscle, 2018, 8, 23.	1.9	40
300	Predicting the clinical impact of human mutation with deep neural networks. Nature Genetics, 2018, 50, 1161-1170.	9.4	288
301	MaxMIF: A New Method for Identifying Cancer Driver Genes through Effective Data Integration. Advanced Science, 2018, 5, 1800640.	5.6	37
302	Performance evaluation of pathogenicity-computation methods for missense variants. Nucleic Acids Research, 2018, 46, 7793-7804.	6.5	168
303	Association Between LTF Polymorphism and Risk of HIV-1 Transmission Among Zambian Seropositive Mothers. Current HIV Research, 2018, 16, 52-57.	0.2	1
304	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
305	In silico analysis of SLC3A1 and SLC7A9 mutations in Iranian patients with Cystinuria. Molecular Biology Reports, 2018, 45, 1165-1173.	1.0	14
306	Effects of genetic variants in the TSPO gene on protein structure and stability. PLoS ONE, 2018, 13, e0195627.	1.1	19
307	A New Integrated Newborn Screening Workflow Can Provide a Shortcut to Differential Diagnosis and Confirmation of Inherited Metabolic Diseases. Yonsei Medical Journal, 2018, 59, 652.	0.9	9

#	Article	IF	Citations
308	Prioritization and functional assessment of noncoding variants associated with complex diseases. Genome Medicine, 2018, 10, 53.	3.6	33
309	Cardiomyopathy and Preeclampsia. Circulation, 2018, 138, 2359-2366.	1.6	60
310	VAReporter: variant reporter for cancer research of massive parallel sequencing. BMC Genomics, 2018, 19, 86.	1.2	2
311	Morpholino Antisense Oligomers as a Potential Therapeutic Option for the Correction of Alternative Splicing in PMD, SPG2, and HEMS. Molecular Therapy - Nucleic Acids, 2018, 12, 420-432.	2.3	13
312	New findings on SNP variants of human protein L-isoaspartyl methyltransferase that affect catalytic activity, thermal stability, and aggregation. PLoS ONE, 2018, 13, e0198266.	1.1	6
313	IDGenetics: a comprehensive database for genes and mutations of intellectual disability related disorders. Neuroscience Letters, 2018, 685, 96-101.	1.0	10
314	Clinical diversity in patients with Schnyder corneal dystrophyâ€"a novel and known UBIAD1 pathogenic variants. Graefe's Archive for Clinical and Experimental Ophthalmology, 2018, 256, 2127-2134.	1.0	6
315	KMT2C Mutations in Diffuse-Type Gastric Adenocarcinoma Promote Epithelial-to-Mesenchymal Transition. Clinical Cancer Research, 2018, 24, 6556-6569.	3.2	70
316	A putative human infertility allele of the meiotic recombinase DMC1 does not affect fertility in mice. Human Molecular Genetics, 2018, 27, 3911-3918.	1.4	14
317	Familial Cancer Variant Prioritization Pipeline version 2 (FCVPPv2) applied to a papillary thyroid cancer family. Scientific Reports, 2018, 8, 11635.	1.6	30
318	Phosphorylation of the RSRSP stretch is critical for splicing regulation by RNA-Binding Motif Protein 20 (RBM20) through nuclear localization. Scientific Reports, 2018, 8, 8970.	1.6	58
319	Genetic background of Japanese patients with pediatric hypertrophic and restrictive cardiomyopathy. Journal of Human Genetics, 2018, 63, 989-996.	1.1	26
320	Evaluation of computational techniques for predicting non-synonymous single nucleotide variants pathogenicity. Genomics, 2019, 111, 869-882.	1.3	36
321	Investigating regime shifts and the factors controlling Total Inorganic Nitrogen concentrations in treated wastewater using non-homogeneous Hidden Markov and multinomial logistic regression models. Science of the Total Environment, 2019, 646, 625-633.	3.9	21
322	Implementing precision cancer medicine in the genomic era. Seminars in Cancer Biology, 2019, 55, 16-27.	4.3	24
323	Predicting Non-Synonymous Single Nucleotide Variants Pathogenic Effects in Human Diseases. , 2019, , 400-409.		1
324	Genome Informatics. , 2019, , 178-194.		0
325	Computational resources associating diseases with genotypes, phenotypes and exposures. Briefings in Bioinformatics, 2019, 20, 2098-2115.	3.2	27

#	Article	IF	Citations
326	Detecting and Annotating Rare Variants. , 2019, , 388-399.		4
327	Evolutionary history of human colitis-associated colorectal cancer. Gut, 2019, 68, 985-995.	6.1	97
328	Bioinformatics classification of mutations in patients with Mucopolysaccharidosis IIIA. Metabolic Brain Disease, 2019, 34, 1577-1594.	1.4	21
329	The impact of SOCS1 mutations in diffuse large Bâ€cell lymphoma. British Journal of Haematology, 2019, 187, 627-637.	1.2	15
330	Whole genome sequencing and rare variant analysis in essential tremor families. PLoS ONE, 2019, 14, e0220512.	1.1	28
331	Benchmarking subcellular localization and variant tolerance predictors on membrane proteins. BMC Genomics, 2019, 20, 547.	1.2	14
332	Concurrent chromothripsis events in a case of TP53 depleted acute myeloid leukemia with myelodysplasia-related changes. Cancer Genetics, 2019, 237, 63-68.	0.2	5
333	Prediction of disease-associated mutations in the transmembrane regions of proteins with known 3D structure. PLoS ONE, 2019, 14, e0219452.	1.1	19
334	Pan-Genomic and Polymorphic Driven Prediction of Antibiotic Resistance in Elizabethkingia. Frontiers in Microbiology, 2019, 10, 1446.	1.5	14
335	Estimation of allele-specific fitness effects across human protein-coding sequences and implications for disease. Genome Research, 2019, 29, 1310-1321.	2.4	24
336	First description of an unusual novel double mutation in MECP 2 coâ€occurring with the m.827A>G mutation in the MTâ€RNR1 gene associated with angelmanâ€like syndrome. International Journal of Developmental Neuroscience, 2019, 79, 37-44.	0.7	3
337	Gene4Denovo: an integrated database and analytic platform for de novo mutations in humans. Nucleic Acids Research, 2020, 48, D913-D926.	6.5	41
338	Whole exome sequencing identifies a rare variant in DAAM2 as a potential candidate in idiopathic pulmonary ossification. Annals of Translational Medicine, 2019, 7, 327-327.	0.7	3
339	Mechanism of Action of Non-Synonymous Single Nucleotide Variations Associated with Î±-Carbonic Anhydrase II Deficiency. Molecules, 2019, 24, 3987.	1.7	18
340	The genetic landscape of the human solute carrier (SLC) transporter superfamily. Human Genetics, 2019, 138, 1359-1377.	1.8	79
341	Detecting TP53 mutations in diagnostic and archival liquid-based Pap samples from ovarian cancer patients using an ultra-sensitive ddPCR method. Scientific Reports, 2019, 9, 15506.	1.6	10
342	Low mutation rate in the TTN gene in paediatric patients with dilated cardiomyopathy â€“ a pilot study. Scientific Reports, 2019, 9, 16409.	1.6	11
343	Visible-Light-Controlled Reaction-Separation for Asymmetric Sulfoxidation in Water with Photoresponsive Metallomicelles. ACS Sustainable Chemistry and Engineering, 2019, 7, 17967-17978.	3.2	17

#	Article	IF	Citations
344	Structure-based Method for Predicting Deleterious Missense SNPs. , 2019, 2019, .		2
345	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
346	Variant Interpretation for Cancer (VIC): a computational tool for assessing clinical impacts of somatic variants. Genome Medicine, 2019, 11, 53.	3.6	36
347	Bioinformatics Workflows for Genomic Variant Discovery, Interpretation and Prioritization. , 0, , .		3
348	A Combined Targeted and Whole Exome Sequencing Approach Identified Novel Candidate Genes Involved in Heritable Pulmonary Arterial Hypertension. Scientific Reports, 2019, 9, 753.	1.6	24
349	Computing the Pathogenicity of Alzheimerâ€™s Disease Presenilin 1 Mutations. Journal of Chemical Information and Modeling, 2019, 59, 858-870.	2.5	19
350	Advancing Personalized Medicine Through the Application of Whole Exome Sequencing and Big Data Analytics. Frontiers in Genetics, 2019, 10, 49.	1.1	140
351	Missense variants in NOX1 and p22phox in a case of very-early-onset inflammatory bowel disease are functionally linked to NOD2. Journal of Physical Education and Sports Management, 2019, 5, a002428.	0.5	13
352	Innovative strategies for annotating the â€œrelationSNPâ€•between variants and molecular phenotypes. BioData Mining, 2019, 12, 10.	2.2	6
353	Laying the foundation for genomically-based risk assessment in chronic myeloid leukemia. Leukemia, 2019, 33, 1835-1850.	3.3	97
354	Characterization of intellectual disability and autism comorbidity through gene panel sequencing. Human Mutation, 2019, 40, 1346-1363.	1.1	54
355	Decreased ACKR3 (CXCR7) function causes oculomotor synkinesis in mice and humans. Human Molecular Genetics, 2019, 28, 3113-3125.	1.4	8
356	Unique Mutational Spectrum of the GJB2 Gene and Its Pathogenic Contribution to Deafness in Tuvinians (Southern Siberia, Russia): A High Prevalence of Rare Variant c.516G>C (p.Trp172Cys). Genes, 2019, 10, 429.	1.0	13
357	Genetic variant pathogenicity prediction trained using disease-specific clinical sequencing data sets. Genome Research, 2019, 29, 1144-1151.	2.4	19
358	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
359	Computational and artificial neural network based study of functional SNPs of human LEPR protein associated with reproductive function. Journal of Cellular Biochemistry, 2019, 120, 18910-18926.	1.2	5
360	RNA sequence analysis reveals macroscopic somatic clonal expansion across normal tissues. Science, 2019, 364, .	6.0	369
361	Unravelling the covalent binding of zampanolide and taccalonolide AJ to a minimalist representation of a human microtubule. Journal of Computer-Aided Molecular Design, 2019, 33, 627-644.	1.3	11

#	Article	IF	Citations
362	Somatic mutations and promotor methylation of the ryanodine receptor 2 is a common event in the pathogenesis of head and neck cancer. International Journal of Cancer, 2019, 145, 3299-3310.	2.3	34
363	Oligogenic inheritance of a human heart disease involving a genetic modifier. Science, 2019, 364, 865-870.	6.0	142
364	Identification of a Novel NOG Missense Mutation in a Chinese Family With Symphalangism and Tarsal Coalitions. Frontiers in Genetics, 2019, 10, 353.	1.1	7
365	Integration of multiple epigenomic marks improves prediction of variant impact in saturation mutagenesis reporter assay. Human Mutation, 2019, 40, 1280-1291.	1.1	46
366	Functional analysis of new variants at the Low Density Lipoprotein Receptor associated with familial hypercholesterolemia. Human Mutation, 2019, 40, 1181-1190.	1.1	10
367	Whole-exome sequencing indicates FLG2 variant associated with leg ulcers in Brazilian sickle cell anemia patients. Experimental Biology and Medicine, 2019, 244, 932-939.	1.1	7
368	GenePy - a score for estimating gene pathogenicity in individuals using next-generation sequencing data. BMC Bioinformatics, 2019, 20, 254.	1.2	21
369	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
370	A novel <scp>CDKN</scp>2A variant (p16^{L117P}) in a patient with familial and multiple primary melanomas. Pigment Cell and Melanoma Research, 2019, 32, 734-738.	1.5	7
371	Finding driver mutations in cancer: Elucidating the role of background mutational processes. PLoS Computational Biology, 2019, 15, e1006981.	1.5	61
372	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
373	Improved measures for evolutionary conservation that exploit taxonomy distances. Nature Communications, 2019, 10, 1556.	5.8	21
374	UniProt genomic mapping for deciphering functional effects of missense variants. Human Mutation, 2019, 40, 694-705.	1.1	29
375	Data Science Driven Drug Repurposing for Metabolic Disorders. , 2019, , 191-227.		7
376	Computational Molecular Phenotypic Analysis of PTPN22 (W620R), IL6R (D358A), and TYK2 (P1104A) Gene Mutations of Rheumatoid Arthritis. Frontiers in Genetics, 2019, 10, 168.	1.1	18
377	New insights into the pathogenicity of non-synonymous variants through multi-level analysis. Scientific Reports, 2019, 9, 1667.	1.6	40
378	How good are pathogenicity predictors in detecting benign variants?. PLoS Computational Biology, 2019, 15, e1006481.	1.5	79
379	The Frog Xenopus as a Model to Study Joubert Syndrome: The Case of a Human Patient With Compound Heterozygous Variants in PIBF1. Frontiers in Physiology, 2019, 10, 134.	1.3	13

#	Article	IF	Citations
380	A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. American Journal of Human Genetics, 2019, 104, 596-610.	2.6	32
381	Whole-exome sequencing detects mutations in pediatric patients with atypical hemolytic uremic syndrome in Taiwan. Clinica Chimica Acta, 2019, 494, 143-150.	0.5	8
382	Evaluation of two approaches to lysosomal acid lipase deficiency patient identification: An observational retrospective study. Atherosclerosis, 2019, 285, 49-54.	0.4	3
383	Approaches to functionally validate candidate genetic variants involved in colorectal cancer predisposition. Molecular Aspects of Medicine, 2019, 69, 27-40.	2.7	5
384	Massively parallel sequencing analysis of benign melanocytic naevi. Histopathology, 2019, 75, 29-38.	1.6	12
385	DIAPH2 alterations increase cellular motility and may contribute to the metastatic potential of laryngeal squamous cell carcinoma. Carcinogenesis, 2019, 40, 1251-1259.	1.3	12
386	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
387	Understanding the structure-function relationship of HPRT1 missense mutations in association with Leschâ€“Nyhan disease and HPRT1-related gout by in silico mutational analysis. Computers in Biology and Medicine, 2019, 107, 161-171.	3.9	20
388	New genetic signals for lung function highlight pathways and chronic obstructive pulmonary disease associations across multiple ancestries. Nature Genetics, 2019, 51, 481-493.	9.4	350
389	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
390	Novel mutations in the RS1 gene in Japanese patients with X-linked congenital retinoschisis. Human Genome Variation, 2019, 6, 3.	0.4	18
391	Integrative Modeling and Novel Technologies in Human Genomics. , 2019, , 155-189.		0
392	Advances in identifying coding variants of common complex diseases. Journal of Bio-X Research, 2019, 2, 153-158.	0.3	0
393	Genetics of Congenital Isolated TSH Deficiency: Mutation Screening of the Known Causative Genes and a Literature Review. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 6229-6237.	1.8	15
394	Application of Computational Biology and Artificial Intelligence Technologies in Cancer Precision Drug Discovery. BioMed Research International, 2019, 2019, 1-15.	0.9	42
395	Ontology-based prediction of cancer driver genes. Scientific Reports, 2019, 9, 17405.	1.6	16
396	Contribution of rare coding mutations in CD36 to type 2 diabetes and cardio-metabolic complications. Scientific Reports, 2019, 9, 17123.	1.6	8
397	Germline variants in cancer genes in high-risk non-BRCA patients from Puerto Rico. Scientific Reports, 2019, 9, 17769.	1.6	12

#	Article	IF	Citations
398	Computing the Pathogenicity of Wilsonâ€™s Disease ATP7B Mutations: Implications for Disease Prevalence. Journal of Chemical Information and Modeling, 2019, 59, 5230-5243.	2.5	10
399	VPOT: A Customizable Variant Prioritization Ordering Tool for Annotated Variants. Genomics, Proteomics and Bioinformatics, 2019, 17, 540-545.	3.0	10
400	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
401	From Clinical Phenotype to Genotypic Modelling: Incidence and Prevalence of Recessive Dystrophic Epidermolysis Bullosa (RDEB) . Clinical, Cosmetic and Investigational Dermatology, 2019, Volume 12, 933-942.	0.8	15
402	Comparison of somatic variant detection algorithms using Ion Torrent targeted deep sequencing data. BMC Medical Genomics, 2019, 12, 181.	0.7	9
403	Non-Redundant and Overlapping Oncogenic Readouts of Non-Canonical and Novel Colorectal Cancer KRAS and NRAS Mutants. Cells, 2019, 8, 1557.	1.8	7
404	Variations in maternal adenylate cyclase genes are associated with congenital Zika syndrome in a cohort from Northeast, Brazil. Journal of Internal Medicine, 2019, 285, 215-222.	2.7	18
405	Sudden death in mild hypertrophic cardiomyopathy with compound DSG2/DSC2/MYH6 mutations: Revisiting phenotype after genetic assessment in a master runner athlete. Journal of Electrocardiology, 2019, 53, 95-99.	0.4	10
406	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
407	Mitochondrial DNA Mutations are Associated with Ulcerative Colitis Preneoplasia but Tend to be Negatively Selected in Cancer. Molecular Cancer Research, 2019, 17, 488-498.	1.5	25
408	FactorialHMM: fast and exact inference in factorial hidden Markov models. Bioinformatics, 2019, 35, 2162-2164.	1.8	1
409	Exploring the spatiotemporal genetic heterogeneity in metastatic lung adenocarcinoma using a nuclei flowâ€sorting approach. Journal of Pathology, 2019, 247, 199-213.	2.1	8
410	Next Generation Sequencing Analysis in Early Onset Dementia Patients. Journal of Alzheimer's Disease, 2019, 67, 243-256.	1.2	29
411	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. Human Mutation, 2019, 40, 53-72.	1.1	48
412	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
413	An optimized prediction framework to assess the functional impact of pharmacogenetic variants. Pharmacogenomics Journal, 2019, 19, 115-126.	0.9	109
414	Frequently used bioinformatics tools overestimate the damaging effect of allelic variants. Genes and Immunity, 2019, 20, 10-22.	2.2	12
415	Targeted deep sequencing of the PEAR1 locus for platelet aggregation in European and African American families. Platelets, 2019, 30, 380-386.	1.1	19

#	Article	IF	Citations
416	Structure-Based Analysis of Single Nucleotide Variants in the Renin-Angiotensinogen Complex. Global Heart, 2017, 12, 121.	0.9	31
417	Role of Structural Bioinformatics in Drug Discovery by Computational SNP Analysis. Global Heart, 2017, 12, 151.	0.9	38
418	Development of Bioinformatics Infrastructure for Genomics Research. Global Heart, 2017, 12, 91.	0.9	47
419	Meta-analysis of up to 622,409 individuals identifies 40 novel smoking behaviour associated genetic loci. Molecular Psychiatry, 2020, 25, 2392-2409.	4.1	83
420	Molecular modelling and dynamics of CA2 missense mutations causative to carbonic anhydrase 2 deficiency syndrome. Journal of Biomolecular Structure and Dynamics, 2020, 38, 4067-4080.	2.0	20
421	Novel HARS2 missense variants identified in individuals with sensorineural hearing impairment and Perrault syndrome. European Journal of Medical Genetics, 2020, 63, 103733.	0.7	9
422	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
423	Next generation sequencing panel in undifferentiated autoinflammatory diseases identifies patients with colchicine-responder recurrent fevers. Rheumatology, 2020, 59, 344-360.	0.9	36
424	Sorting nexin 27 (SNX27) variants associated with seizures, developmental delay, behavioral disturbance, and subcortical brain abnormalities. Clinical Genetics, 2020, 97, 437-446.	1.0	10
425	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
426	Whole-Exome Sequencing of Matched Primary and Metastatic Papillary Thyroid Cancer. Thyroid, 2020, 30, 42-56.	2.4	31
427	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
428	Pathogenicity Reclassification of RPE65 Missense Variants Related to Leber Congenital Amaurosis and Early-Onset Retinal Dystrophy. Genes, 2020, 11, 24.	1.0	14
429	Identification of altered biological processes in heterogeneous RNA-sequencing data by discretization of expression profiles. Nucleic Acids Research, 2020, 48, 1730-1747.	6.5	8
430	Prediction of mutation effects using a deep temporal convolutional network. Bioinformatics, 2020, 36, 2047-2052.	1.8	13
431	The Melanocortin 4 Receptor p.Ile269Asn Mutation Is Associated with Childhood and Adult Obesity in Mexicans. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e1468-e1477.	1.8	9
432	Identification of a homozygous VRK1 mutation in two patients with adultâ€onset distal hereditary motor neuropathy. Muscle and Nerve, 2020, 61, 395-400.	1.0	10
433	Predâ€MutHTP: Prediction of diseaseâ€causing and neutral mutations in human transmembrane proteins. Human Mutation, 2020, 41, 581-590.	1.1	21

#	Article	IF	Citations
434	Identification of pathogenic variant enriched regions across genes and gene families. Genome Research, 2020, 30, 62-71.	2.4	47
435	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
436	Identification of the First PAX4-MODY Family Reported in Brazil . Diabetes, Metabolic Syndrome and Obesity: Targets and Therapy, 2020, Volume 13, 2623-2631.	1.1	10
437	Analysis of the Spectrum of ACE2 Variation Suggests a Possible Influence of Rare and Common Variants on Susceptibility to COVID-19 and Severity of Outcome. Frontiers in Genetics, 2020, 11, 551220.	1.1	32
438	Cancer Predisposition Genes in Cancer-Free Families. Cancers, 2020, 12, 2770.	1.7	2
439	Whole genome, transcriptome and methylome profiling enhances actionable target discovery in high-risk pediatric cancer. Nature Medicine, 2020, 26, 1742-1753.	15.2	185
440	CerealsDBâ€"new tools for the analysis of the wheat genome: update 2020. Database: the Journal of Biological Databases and Curation, 2020, 2020, .	1.4	16
441	Estimation of varicocele associated human ARG2 and NOS1 proteins and computational analysis on the effect of its nsSNPs. International Journal of Biological Macromolecules, 2020, 164, 735-747.	3.6	3
442	SLC12A2 variants cause a neurodevelopmental disorder or cochleovestibular defect. Brain, 2020, 143, 2380-2387.	3.7	34
443	Genetic Spectrum of Syndromic and Non-Syndromic Hearing Loss in Pakistani Families. Genes, 2020, 11, 1329.	1.0	7
444	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. Nature Communications, 2020, 11, 5918.	5.8	305
445	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
446	Comprehensive Characterization of Stage IIIA Non-Small Cell Lung Carcinoma . Cancer Management and Research, 2020, Volume 12, 11973-11988.	0.9	2
447	Structural and conformational changes induced by missense variants in the zinc finger domains of GATA3 involved in breast cancer. RSC Advances, 2020, 10, 39640-39653.	1.7	8
448	SUCLG1 mutations and mitochondrial encephalomyopathy: a case study and review of the literature. Molecular Biology Reports, 2020, 47, 9699-9714.	1.0	4
449	Prediction of driver variants in the cancer genome via machine learning methodologies. Briefings in Bioinformatics, 2021, 22, .	3.2	13
450	A pan-cancer analysis reveals nonstop extension mutations causing SMAD4 tumour suppressor degradation. Nature Cell Biology, 2020, 22, 999-1010.	4.6	12
451	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

#	Article	IF	Citations
452	The First Report of Biallelic Missense Mutations in the SFRP4 Gene Causing Pyle Disease in Two Siblings. Frontiers in Genetics, 2020, 11, 593407.	1.1	8
453	Multi-omic studies on missense PLG variants in families with otitis media. Scientific Reports, 2020, 10, 15035.	1.6	4
454	The genetic architecture of appendicular lean mass characterized by association analysis in the UK Biobank study. Communications Biology, 2020, 3, 608.	2.0	83
455	Computational Modeling of NLRP3 Identifies Enhanced ATP Binding and Multimerization in Cryopyrin-Associated Periodic Syndromes. Frontiers in Immunology, 2020, 11, 584364.	2.2	9
456	A DNAH17 missense variant causes flagella destabilization and asthenozoospermia. Journal of Experimental Medicine, 2020, 217, .	4.2	88
457	m⁶A RNA modification modulates PI3K/Akt/mTOR signal pathway in Gastrointestinal Cancer. Theranostics, 2020, 10, 9528-9543.	4.6	62
458	The single nucleotideÎ²-arrestin2 variant, A248T, resembles dynamical properties of activated arrestin. Turkish Journal of Chemistry, 2020, 44, 409-420.	0.5	2
459	Assessing Lysosomal Disorders in the NGS Era: Identification of Novel Rare Variants. International Journal of Molecular Sciences, 2020, 21, 6355.	1.8	8
460	Genotypeâ€“phenotype correlation of 33 patients with maple syrup urine disease. American Journal of Medical Genetics, Part A, 2020, 182, 2486-2500.	0.7	6
461	A novel nonsense mutation in TNNT2 in a Chinese pedigree with hypertrophic cardiomyopathy. Medicine (United States), 2020, 99, e21843.	0.4	2
462	Assessing performance of pathogenicity predictors using clinically relevant variant datasets. Journal of Medical Genetics, 2021, 58, 547-555.	1.5	57
463	Characterization of rare ABCC8 variants identified in Spanish pulmonary arterial hypertension patients. Scientific Reports, 2020, 10, 15135.	1.6	19
464	Predicting the pathogenicity of protein coding mutations using Natural Language Processing. , 2020, 2020, 5842-5846.		2
465	IDRMutPred: predicting disease-associated germline nonsynonymous single nucleotide variants (nsSNVs) in intrinsically disordered regions. Bioinformatics, 2020, 36, 4977-4983.	1.8	5
466	MosaicBase: A Knowledgebase of Postzygotic Mosaic Variants in Noncancer Disease-related and Healthy Human Individuals. Genomics, Proteomics and Bioinformatics, 2020, 18, 140-149.	3.0	10
467	Investigating the structural impacts of a novel missense variant identified with whole exome sequencing in an Egyptian patient with propionic acidemia. Molecular Genetics and Metabolism Reports, 2020, 25, 100645.	0.4	3
468	Structural insights and evaluation of the potential impact of missense variants on the interactions of SLIT2 with ROBO1/4 in cancer progression. Scientific Reports, 2020, 10, 21909.	1.6	1
469	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

#	Article	IF	Citations
470	Whole-exome sequencing in patients with protein aggregate myopathies reveals causative mutations associated with novel atypical phenotypes. Neurological Sciences, 2021, 42, 2819-2827.	0.9	2
471	Variant discovery using next-generation sequencing and its future role in pharmacogenetics. Pharmacogenomics, 2020, 21, 471-486.	0.6	9
472	Mutation-Associated Phenotypic Heterogeneity in Novel and Canonical PIK3CA Helical and Kinase Domain Mutants. Cells, 2020, 9, 1116.	1.8	6
473	Genomically Aided Diagnosis of Severe Developmental Disorders. Annual Review of Genomics and Human Genetics, 2020, 21, 327-349.	2.5	3
474	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
475	TTRMDB: A database for structural and functional analysis on the impact of SNPs over transthyretin (TTR) using bioinformatic tools. Computational Biology and Chemistry, 2020, 87, 107290.	1.1	8
476	Family-specific analysis of variant pathogenicity prediction tools. NAR Genomics and Bioinformatics, 2020, 2, lqaa014.	1.5	8
477	Novel frameshift variant in MYL2 reveals molecular differences between dominant and recessive forms of hypertrophic cardiomyopathy. PLoS Genetics, 2020, 16, e1008639.	1.5	16
478	Driverâ€™s intention recognition algorithm based on recessive Markoff model. Journal of Intelligent and Fuzzy Systems, 2020, 38, 1603-1614.	0.8	8
479	Computational Approaches for Unraveling the Effects of Variation in the Human Genome and Microbiome. Annual Review of Biomedical Data Science, 2020, 3, 411-432.	2.8	5
480	GJB4andGJC3variants in non-syndromic hearing impairment in Ghana. Experimental Biology and Medicine, 2020, 245, 1355-1367.	1.1	4
481	DHH pathogenic variants involved in 46,XY disorders of sex development differentially impact protein self-cleavage and structural conformation. Human Genetics, 2020, 139, 1455-1470.	1.8	2
482	SEMA3A and IGSF10 Are Novel Contributors to Combined Pituitary Hormone Deficiency (CPHD). Frontiers in Endocrinology, 2020, 11, 368.	1.5	13
483	Adultâ€onset veryâ€longâ€chain acylâ€CoA dehydrogenase deficiency (VLCADD). European Journal of Neurology, 2020, 27, 2257-2266.	1.7	6
484	Identification of a Pathogenic TGFBR2 Variant in a Patient With Loeysâ€“Dietz Syndrome. Frontiers in Genetics, 2020, 11, 479.	1.1	6
485	Mapping the TYR gene reveals novel and previously reported variants in Eastern Indian patients highlighting preponderance of the same changes in multiple unrelated ethnicities. Annals of Human Genetics, 2020, 84, 303-312.	0.3	5
486	Benchmarking analysis of deleterious SNP prediction tools on CYP2D6 enzyme. Chemical Biology and Drug Design, 2020, 96, 984-994.	1.5	6
487	Expansion of the genetic landscape of ERLIN2â€related disorders. Annals of Clinical and Translational Neurology, 2020, 7, 573-578.	1.7	12

#	Article	IF	Citations
488	A Novel System for Functional Determination of Variants of Uncertain Significance using Deep Convolutional Neural Networks. Scientific Reports, 2020, 10, 4192.	1.6	5
489	m7GHub: deciphering the location, regulation and pathogenesis of internal mRNA N7-methylguanosine (m7G) sites in human. Bioinformatics, 2020, 36, 3528-3536.	1.8	85
490	Emerging strategies to bridge the gap between pharmacogenomic research and its clinical implementation. Npj Genomic Medicine, 2020, 5, 9.	1.7	42
491	Clinical implications of experimental analyses of AID function on predictive computational tools: Challenge of missense variants. Clinical Genetics, 2020, 97, 844-856.	1.0	0
492	Identification of Somatic Mutations in Thirty-year-old Serum Cell-free DNA From Patients With Breast Cancer: A Feasibility Study. Clinical Breast Cancer, 2020, 20, 413-421.e1.	1.1	2
493	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
494	Adrenal Medullary Hyperplasia: An Under the Radar Cause of Endocrine Hypertension. American Journal of the Medical Sciences, 2022, 363, 64-68.	0.4	3
495	Survival of the cheapest: how proteome cost minimization drives evolution. Quarterly Reviews of Biophysics, 2020, 53, e7.	2.4	12
496	Clinical and Molecular Investigation of Familial Multiple Lipomatosis: Variants in the HMGA2 Gene . Clinical, Cosmetic and Investigational Dermatology, 2020, Volume 13, 1-10.	0.8	6
497	Mechanistic insights into the deleterious roles of Nasu-Hakola disease associated TREM2 variants. Scientific Reports, 2020, 10, 3663.	1.6	24
498	Comprehensive assessment of computational algorithms in predicting cancer driver mutations. Genome Biology, 2020, 21, 43.	3.8	47
499	In-silico analysis of deleterious missense SNPs of human TYR gene associated with oculocutaneous albinism type 1 (OCA1). Meta Gene, 2020, 24, 100674.	0.3	5
500	A novel compound heterozygous mutation in DGKE in a Chinese patient causes atypical hemolytic uremic syndrome. Hematology, 2020, 25, 101-107.	0.7	4
501	Computational analyses prioritize and reveal the deleterious nsSNPs in human angiotensinogen gene. Computational Biology and Chemistry, 2020, 84, 107199.	1.1	8
502	CYP2R1 and CYP27A1 genes: An in silico approach to identify the deleterious mutations, impact on structure and their differential expression in disease conditions. Genomics, 2020, 112, 3677-3686.	1.3	5
503	Next generation sequencing exome data analysis aids in the discovery of SNP and INDEL patterns in Parkinson's disease. Genomics, 2020, 112, 3722-3728.	1.3	5
504	Using an integrative machine learning approach utilising homology modelling to clinically interpret genetic variants: CACNA1F as an exemplar. European Journal of Human Genetics, 2020, 28, 1274-1282.	1.4	11
505	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

#	Article	IF	Citations
506	Tumour characteristics provide evidence for germline mismatch repair missense variant pathogenicity. Journal of Medical Genetics, 2020, 57, 62-69.	1.5	11
507	A novel F11 mutation in a Chinese paediatric patient with severe factor XI deficiency. Thrombosis Research, 2020, 190, 89-90.	0.8	0
508	CScape-somatic: distinguishing driver and passenger point mutations in the cancer genome. Bioinformatics, 2020, 36, 3637-3644.	1.8	19
509	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
510	PredCID: prediction of driver frameshift indels in human cancer. Briefings in Bioinformatics, 2021, 22, .	3.2	29
511	Variant Calling in Next Generation Sequencing Data. , 2021, , 129-140.		0
512	Comprehensive in silico mutational-sensitivity analysis of PTEN establishes signature regions implicated in pathogenesis of Autism Spectrum Disorders. Genomics, 2021, 113, 999-1017.	1.3	3
513	MobiDetails: online DNA variants interpretation. European Journal of Human Genetics, 2021, 29, 356-360.	1.4	34
514	Whole-Exome Sequencing of Patients With Posterior Segment Uveitis. American Journal of Ophthalmology, 2021, 221, 246-259.	1.7	10
515	RMDisease: a database of genetic variants that affect RNA modifications, with implications for epitranscriptome pathogenesis. Nucleic Acids Research, 2021, 49, D1396-D1404.	6.5	65
516	Comprehensive in-silico analysis of damage associated SNPs in hOCT1 affecting Imatinib response in chronic myeloid leukemia. Genomics, 2021, 113, 755-766.	1.3	6
517	In Silico Tools and Approaches for the Prediction of Functional and Structural Effects of Single-Nucleotide Polymorphisms on Proteins: An Expert Review. OMICS A Journal of Integrative Biology, 2021, 25, 23-37.	1.0	22
518	A role for the MEGF6 gene in predisposition to osteoporosis. Annals of Human Genetics, 2021, 85, 58-72.	0.3	15
519	Predictive significance of selected gene mutations in relapsed and refractory chronic lymphocytic leukemia patients treated with ibrutinib. European Journal of Haematology, 2021, 106, 320-326.	1.1	2
520	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
521	OncoVar: an integrated database and analysis platform for oncogenic driver variants in cancers. Nucleic Acids Research, 2021, 49, D1289-D1301.	6.5	64
522	Computational Tools for Causal Inference in Genetics. Cold Spring Harbor Perspectives in Medicine, 2021, 11, a039248.	2.9	3
523	Biallelic ZNF335 mutations cause basal ganglia abnormality with progressive cerebral/cerebellar atrophy. Journal of Neurogenetics, 2021, 35, 23-28.	0.6	2

#	Article	IF	Citations
524	Comprehensive functional annotation of susceptibility variants identifies genetic heterogeneity between lung adenocarcinoma and squamous cell carcinoma. Frontiers of Medicine, 2021, 15, 275-291.	1.5	21
525	Molecular dynamics simulations for genetic interpretation in protein coding regions: where we are, where to go and when. Briefings in Bioinformatics, 2021, 22, 3-19.	3.2	30
526	Kabuki Syndrome: Identification of Two Novel Variants in <i>KMT2D</i> and <i>KDM6A</i>. Molecular Syndromology, 2021, 12, 118-126.	0.3	4
527	GPCards: An integrated database of genotypeâ€“phenotype correlations in human genetic diseases. Computational and Structural Biotechnology Journal, 2021, 19, 1603-1611.	1.9	5
528	Classification of genetic variants in hereditary cancer genes. , 2021, , 349-387.		0
529	An Overview of Bioinformatics Resources for SNP Analysis. , 2021, , 113-135.		1
530	Hunting for the perfect test: Neuromuscular diagnosis in the age of genomic bounty. Muscle and Nerve, 2021, 63, 282-284.	1.0	1
531	Putative second hit rare genetic variants in families with seemingly GBA-associated Parkinsonâ€™s disease. Npj Genomic Medicine, 2021, 6, 2.	1.7	11
532	Performance of mutation pathogenicity prediction tools on missense variants associated with 46,XY differences of sex development. Clinics, 2021, 76, e2052.	0.6	10
533	Genetic variant effect prediction by supervised nonnegative matrix tri-factorization. Molecular Omics, 2021, 17, 740-751.	1.4	1
534	Integrating Evolutionary Genetics to Medical Genomics: Evolutionary Approaches to Investigate Disease-Causing Variants. , 0, , .		0
535	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
537	Novel SCN5A variants identified in a group of Iranian Brugada syndrome patients. Functional and Integrative Genomics, 2021, 21, 331-340.	1.4	1
538	Hypermutated phenotype in gliosarcoma of the spinal cord. Npj Precision Oncology, 2021, 5, 8.	2.3	5
539	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
541	Oculocutaneous albinism type 1B associated with a functionally significant tyrosinase gene polymorphism detected with Whole Exome Sequencing. Ophthalmic Genetics, 2021, 42, 291-295.	0.5	3
542	Complement Factor I Mutation May Contribute to Development of Thrombotic Microangiopathy in Lupus Nephritis. Frontiers in Medicine, 2020, 7, 621609.	1.2	3
543	New Insights Into Mitochondrial DNA Reconstruction and Variant Detection in Ancient Samples. Frontiers in Genetics, 2021, 12, 619950.	1.1	6

#	Article	IF	Citations
544	A recurrent ZSWIM7 mutation causes male infertility resulting from decreased meiotic recombination. Human Reproduction, 2021, 36, 1436-1445.	0.4	18
545	Identification of candidate genes and pathways in retinopathy of prematurity by whole exome sequencing of preterm infants enriched in phenotypic extremes. Scientific Reports, 2021, 11, 4966.	1.6	7
547	A homozygous loss-of-function mutation in GP1BB causing variable clinical phenotypes in a family with Bernardâ€“Soulier syndrome. Blood Coagulation and Fibrinolysis, 2021, 32, 352-355.	0.5	0
548	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
549	Low Diversity of Human Variation Despite Mostly Mild Functional Impact of De Novo Variants. Frontiers in Molecular Biosciences, 2021, 8, 635382.	1.6	2
550	Type IV Collagen Variants in CKD: Performance of Computational Predictions for Identifying Pathogenic Variants. Kidney Medicine, 2021, 3, 257-266.	1.0	9
551	Case Report: A Novel PAX3 Mutation Associated With Waardenburg Syndrome Type 1. Frontiers in Genetics, 2021, 12, 609040.	1.1	1
552	Whole-exome sequencing in 168 Korean patients with inherited retinal degeneration. BMC Medical Genomics, 2021, 14, 74.	0.7	24
553	Identification of Four Novel Variants and Determination of Genotypeâ€“Phenotype Correlations for ABCA4 Variants Associated With Inherited Retinal Degenerations. Frontiers in Cell and Developmental Biology, 2021, 9, 634843.	1.8	8
555	Identification of Lapatinib Derivatives and Analogs to Control Metastatic Breast Cancer-specific to South Asian Population-a Pharmacogenomic Approach. WSEAS Transactions on Biology and Biomedicine, 2021, 18, 51-62.	0.3	4
556	Multi-system neurological disorder associated with a CRYAB variant. Neurogenetics, 2021, 22, 117-125.	0.7	1
557	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
558	Case Report: Compound Heterozygous Variants in MOCS3 Identified in a Chinese Infant With Molybdenum Cofactor Deficiency. Frontiers in Genetics, 2021, 12, 651878.	1.1	5
559	Comprehensive analysis of germline mutations in northern Brazil: a panel of 16 genes for hereditary cancer-predisposing syndrome investigation. BMC Cancer, 2021, 21, 363.	1.1	7
560	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
561	In Silico Predictions of KCNQ Variant Pathogenicity in Epilepsy. Pediatric Neurology, 2021, 118, 48-54.	1.0	2
562	Cancer-causingBRCA2missense mutations disrupt an intracellular protein assembly mechanism to disable genome maintenance. Nucleic Acids Research, 2021, 49, 5588-5604.	6.5	20
563	Identifying and elucidating the roles of Y198N and Y204F mutations in the PAH enzyme through molecular dynamic simulations. Journal of Biomolecular Structure and Dynamics, 2021, , 1-12.	2.0	3

#	Article	IF	Citations
564	Identification of driver genes based on gene mutational effects and network centrality. BMC Bioinformatics, 2021, 22, 457.	1.2	3
565	driveR: a novel method for prioritizing cancer driver genes using somatic genomics data. BMC Bioinformatics, 2021, 22, 263.	1.2	9
567	ConsRM: collection and large-scale prediction of the evolutionarily conserved RNA methylation sites, with implications for the functional epitranscriptome. Briefings in Bioinformatics, 2021, 22, .	3.2	34
568	A family with Milroy disease caused by the FLT4/VEGFR3 gene variant c.2774ÂTâ€‰›â€‰A. BMC Medical Genomics, 2021, 14, 151.	0.7	1
569	A clinical case of multiple primary cancers in a carrier of rare SDK2 and NOTCH2 gene mutations. Egyptian Journal of Medical Human Genetics, 2021, 22, .	0.5	0
570	Towards a New, Endophenotype-Based Strategy for Pathogenicity Prediction in BRCA1 and BRCA2: In Silico Modeling of the Outcome of HDR/SGE Assays for Missense Variants. International Journal of Molecular Sciences, 2021, 22, 6226.	1.8	0
571	Genetic Characterization of Cancer of Unknown Primary Using Liquid Biopsy Approaches. Frontiers in Cell and Developmental Biology, 2021, 9, 666156.	1.8	12
572	Leveraging supervised learning for functionallyÂinformed fine-mapping of cis-eQTLs identifies an additional 20,913 putative causal eQTLs. Nature Communications, 2021, 12, 3394.	5.8	44
574	APP, PSEN1, and PSEN2 Variants in Alzheimerâ€™s Disease: Systematic Re-evaluation According to ACMG Guidelines. Frontiers in Aging Neuroscience, 2021, 13, 695808.	1.7	33
575	Evolutionary and functional lessons from human-specific amino acid substitution matrices. NAR Genomics and Bioinformatics, 2021, 3, lqab079.	1.5	1
576	Novel homozygous mutations in Pakistani families with Charcotâ€“Marieâ€“Tooth disease. BMC Medical Genomics, 2021, 14, 174.	0.7	4
577	Pittâ€“Hopkins syndrome: phenotypic and genotypic description of four unrelated patients and structural analysis of corresponding missense mutations. Neurogenetics, 2021, 22, 161-169.	0.7	4
578	A Curriculum for Genomic Education of Molecular Genetic Pathology Fellows. Journal of Molecular Diagnostics, 2021, 23, 1218-1240.	1.2	4
579	Germ-line mutations in WDR77 predispose to familial papillary thyroid cancer. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	13
580	3Cnet: pathogenicity prediction of human variants using multitask learning with evolutionary constraints. Bioinformatics, 2021, 37, 4626-4634.	1.8	17
581	Emergence and maintenance of actionable genetic drivers at medulloblastoma relapse. Neuro-Oncology, 2022, 24, 153-165.	0.6	28
582	Hearing Impairment with Monoallelic GJB2 Variants. Journal of Molecular Diagnostics, 2021, 23, 1279-1291.	1.2	10
583	PON-Sol2: Prediction of Effects of Variants on Protein Solubility. International Journal of Molecular Sciences, 2021, 22, 8027.	1.8	10

#	Article	IF	Citations
585	The Spectrum and Novel Mutations in RS1 Gene in a Russian Cohort of Patients with X-Linked Retinoschisis. Russian Journal of Genetics, 2021, 57, 847-855.	0.2	1
586	RNA editing affects cisâ€regulatory elements and predicts adverse cancer survival. Cancer Medicine, 2021, 10, 6114-6127.	1.3	5
587	Efficacy of computational predictions of the functional effect of idiosyncratic pharmacogenetic variants. PeerJ, 2021, 9, e11774.	0.9	2
588	The protective effects of the methylenetetrahydrofolate reductase rs1801131 variant among Saudi smokers. Saudi Journal of Biological Sciences, 2021, 28, 3972-3980.	1.8	2
589	CDON gene contributes to pituitary stalk interruption syndrome associated with unilateral facial and abducens nerve palsy. Journal of Applied Genetics, 2021, 62, 621-629.	1.0	3
590	Genomics pipelines to investigate susceptibility in whole genome and exome sequenced data for variant discovery, annotation, prediction and genotyping. PeerJ, 2021, 9, e11724.	0.9	12
591	In silico saturation mutagenesis of cancer genes. Nature, 2021, 596, 428-432.	13.7	61
592	The structural, functional, and dynamic effect of Tau tubulin kinase1 upon a mutation: A neuroâ€degenerative hotspot. Journal of Cellular Biochemistry, 2021, 122, 1653-1664.	1.2	11
593	Whole-Genome Sequencing Improves the Diagnosis of DFNB1 Monoallelic Patients. Genes, 2021, 12, 1267.	1.0	4
594	Case Report: Identification of a Novel Homozygous Mutation in GPD1 Gene of a Chinese Child With Transient Infantile Hypertriglyceridemia. Frontiers in Genetics, 2021, 12, 726116.	1.1	4
595	Structural effects driven by rare point mutations in amylin hormone, the type II diabetes-associated peptide. Biochimica Et Biophysica Acta - General Subjects, 2021, 1865, 129935.	1.1	2
596	New Missense Mutation Gly238Ala in the TBX5 Gene and Its Phenotypical Characteristics. Russian Journal of Genetics, 2021, 57, 949-954.	0.2	0
597	Allele-specific RT-PCR for the rapid detection of recurrent SLC12A3 mutations for Gitelman syndrome. Npj Genomic Medicine, 2021, 6, 68.	1.7	2
598	A novel machine learning-based approach for the computational functional assessment of pharmacogenomic variants. Human Genomics, 2021, 15, 51.	1.4	14
600	X-CNV: genome-wide prediction of the pathogenicity of copy number variations. Genome Medicine, 2021, 13, 132.	3.6	24
601	A domain damage index to prioritizing the pathogenicity of missense variants. Human Mutation, 2021, 42, 1503-1517.	1.1	0
602	Duo: A Signature Based Method to Batch-Analyze Functional Similarities of Proteins. Frontiers in Microbiology, 2021, 12, 698322.	1.5	1
603	Identification of cancer-related mutations in human pluripotent stem cells using RNA-seq analysis. Nature Protocols, 2021, 16, 4522-4537.	5.5	8

#	Article	IF	Citations
605	Whole-exome sequencing of consanguineous families with infertile men and women identifies homologous mutations in SPATA22 and MEIOB. Human Reproduction, 2021, 36, 2793-2804.	0.4	17
608	Clinical, Biochemical, and Genetic Heterogeneity in Glutaric Aciduria Type II Patients. Genes, 2021, 12, 1334.	1.0	1
609	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
610	Genome sequencing data analysis for rare disease gene discovery. Briefings in Bioinformatics, 2022, 23, .	3.2	6
611	Structural and functional analysis of disease-associated mutations in GOT1 gene: An in silico study. Computers in Biology and Medicine, 2021, 136, 104695.	3.9	12
612	Clinical Impact of Detecting Low-Frequency Variants in Cell-Free DNA on Treatment of Castration-Resistant Prostate Cancer. Clinical Cancer Research, 2021, 27, 6164-6173.	3.2	10
613	Developmental and temporal characteristics of clonal sperm mosaicism. Cell, 2021, 184, 4772-4783.e15.	13.5	27
614	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
615	Molecular dynamics approach to identification of new OGG1 cancer-associated somatic variants with impaired activity. Journal of Biological Chemistry, 2021, 296, 100229.	1.6	7
616	Genome-wide association study of febrile seizures implicates fever response and neuronal excitability genes. Brain, 2022, 145, 555-568.	3.7	29
617	Pan-cancer analysis of transcripts encoding novel open-reading frames (nORFs) and their potential biological functions. Npj Genomic Medicine, 2021, 6, 4.	1.7	20
618	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
619	Untangling a complex web: Computational analyses of tumor molecular profiles to decode driver mechanisms. Journal of Genetics and Genomics, 2020, 47, 595-609.	1.7	5
620	A Novel Locus and Candidate Gene for Familial Developmental Dyslexia on Chromosome 4q. Zeitschrift FÃœr Kinder- Und Jugendpsychiatrie Und Psychotherapie, 2020, 48, 478-489.	0.4	5
622	iFish: predicting the pathogenicity of human nonsynonymous variants using gene-specific/family-specific attributes and classifiers. , 0, .		1
623	AI-Driver: an ensemble method for identifying driver mutations in personal cancer genomes. NAR Genomics and Bioinformatics, 2020, 2, lqaa084.	1.5	19
645	IRS2 mutations linked to invasion in pleomorphic invasive lobular carcinoma. JCI Insight, 2018, 3, .	2.3	18
646	Whole-exome sequencing uncovers oxidoreductases DHTKD1 and OGDHL as linkers between mitochondrial dysfunction and eosinophilic esophagitis. JCI Insight, 2018, 3, .	2.3	39

#	Article	IF	Citations
648	Mutation profiling of anaplastic ependymoma grade III by Ion Proton next generation DNA sequencing. F1000Research, 2019, 8, 613.	0.8	5
649	Towards Increasing the Clinical Relevance of In Silico Methods to Predict Pathogenic Missense Variants. PLoS Computational Biology, 2016, 12, e1004725.	1.5	34
650	High-confidence assessment of functional impact of human mitochondrial non-synonymous genome variations by APOGEE. PLoS Computational Biology, 2017, 13, e1005628.	1.5	54
651	Validation of Next-Generation Sequencing of Entire Mitochondrial Genomes and the Diversity of Mitochondrial DNA Mutations in Oral Squamous Cell Carcinoma. PLoS ONE, 2015, 10, e0135643.	1.1	41
652	ENTPRISE: An Algorithm for Predicting Human Disease-Associated Amino Acid Substitutions from Sequence Entropy and Predicted Protein Structures. PLoS ONE, 2016, 11, e0150965.	1.1	23
653	The D519G Polymorphism of Glyceronephosphate O-Acyltransferase Is a Risk Factor for Familial Porphyria Cutanea Tarda. PLoS ONE, 2016, 11, e0163322.	1.1	7
654	POU4F3 mutation screening in Japanese hearing loss patients: Massively parallel DNA sequencing-based analysis identified novel variants associated with autosomal dominant hearing loss. PLoS ONE, 2017, 12, e0177636.	1.1	31
655	Whole gene sequencing identifies deep-intronic variants with potential functional impact in patients with hypertrophic cardiomyopathy. PLoS ONE, 2017, 12, e0182946.	1.1	41
656	Generalising better: Applying deep learning to integrate deleteriousness prediction scores for whole-exome SNV studies. PLoS ONE, 2018, 13, e0192829.	1.1	14
657	WFS1 mutation screening in a large series of Japanese hearing loss patients: Massively parallel DNA sequencing-based analysis. PLoS ONE, 2018, 13, e0193359.	1.1	33
658	Recurrent, low-frequency coding variants contributing to colorectal cancer in the Swedish population. PLoS ONE, 2018, 13, e0193547.	1.1	10
659	Novel putative drivers revealed by targeted exome sequencing of advanced solid tumors. PLoS ONE, 2018, 13, e0194790.	1.1	3
660	Using deep mutational scanning to benchmark variant effect predictors and identify disease mutations. Molecular Systems Biology, 2020, 16, e9380.	3.2	120
661	Next-generation sequencing refines the genetic architecture of Greek GnRH-deficient patients. Endocrine Connections, 2019, 8, 468-480.	0.8	16
662	Systematic alanine scanning of PAX8 paired domain reveals functional importance of the N-subdomain. Journal of Molecular Endocrinology, 2019, 62, 129-135.	1.1	11
663	Acquired somatic TP53 or PIK3CA mutations are potential predictors of when polyps evolve into colorectal cancer. Oncotarget, 2017, 8, 72352-72362.	0.8	17
664	Somatic mutations in early onset luminal breast cancer. Oncotarget, 2018, 9, 22460-22479.	0.8	25
665	NRAS germline variant G138R and multiple rare somatic mutations on APC in colorectal cancer patients in Taiwan by next generation sequencing. Oncotarget, 2016, 7, 37566-37580.	0.8	5

#	Article	IF	Citations
666	Reliability of Whole-Exome Sequencing for Assessing Intratumor Genetic Heterogeneity. SSRN Electronic Journal, 0, , .	0.4	2
667	In silico analysis for determining the deleterious nonsynonymous single nucleotide polymorphisms of genes. Molecular Biology Research Communications, 2019, 8, 141-150.	0.2	9
668	Identification of osteosarcoma driver genes using a network method. Oncology Letters, 2020, 19, 1215-1222.	0.8	1
669	Structured Genome-Scale Variant and Clinical Data Reporting for Meta-Analysis in an Era of Genomic Medicine. Journal of Genomes and Exomes, 0, 2, 31-42.	0.0	2
670	Comparative functional characterization of novel non-syndromicGJB2gene variant p.Gly45Arg and lethal syndromic variant p.Gly45Glu. PeerJ, 2016, 4, e2494.	0.9	7
671	Rare variant association study of veteran twin whole-genomes links severe depression with a nonsynonymous change in the neuronal gene BHLHE22. World Journal of Biological Psychiatry, 2022, 23, 295-306.	1.3	1
673	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
674	Prevalence and Clinical Characteristics of Hearing Loss Caused by MYH14 Variants. Genes, 2021, 12, 1623.	1.0	5
675	An Investigation of the Role of Common and Rare Variants in a Large Italian Multiplex Family of Multiple Sclerosis Patients. Genes, 2021, 12, 1607.	1.0	4
677	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
678	Computational algorithmic and molecular dynamics study of functional and structural impacts of non-synonymous single nucleotide polymorphisms in human DHFR gene. Computational Biology and Chemistry, 2021, 95, 107587.	1.1	2
683	Mutations of the CHEK2 gene in patients with cancer and their presence in the Latin American population. F1000Research, 0, 5, 2791.	0.8	2
689	Structural Impact of Single Nucleotide Variations (SNVs). , 2018, , 1-6.		0
697	Distinguishing Driver Missense Mutations from Benign Polymorphisms in Breast Cancer. Lecture Notes in Computer Science, 2019, , 294-302.	1.0	0
712	Identification of nsSNPs of transcription factor E2F1 predisposing individuals to lung cancer and head and neck cancer. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2020, 821, 111704.	0.4	7
713	Mutation profiling of anaplastic ependymoma grade III by Ion Proton next generation DNA sequencing. F1000Research, 2019, 8, 613.	0.8	7
715	<scp>Mouseâ€NtraDuctal</scp> (<scp>MIND</scp>): an in vivo model for studying the underlying mechanisms of <scp>DCIS</scp> malignancy. Journal of Pathology, 2022, 256, 186-201.	2.1	12
717	Integrative genomic analysis focused on cell cycle genes for MYC-driven aggressive mature B-cell lymphoma. Journal of Clinical and Experimental Hematopathology: JCEH, 2020, 60, 87-96.	0.3	1

#	Article	IF	Citations
721	Computational and structural based approach to identify malignant nonsynonymous single nucleotide polymorphisms associated with CDK4 gene. PLoS ONE, 2021, 16, e0259691.	1.1	7
722	Synonymous and non-synonymous polymorphisms in toll-like receptor 2 (TLR2) gene among complicated measles cases at a tertiary care hospital, Peshawar, Pakistan. Journal of King Abdulaziz University, Islamic Economics, 2021, 42, 1229-1236.	0.5	1
723	VPMBench: a test bench for variant prioritization methods. BMC Bioinformatics, 2021, 22, 543.	1.2	0
724	Impact of Deleterious Mutations on Structure, Function and Stability of Serum/Glucocorticoid Regulated Kinase 1: A Gene to Diseases Correlation. Frontiers in Molecular Biosciences, 2021, 8, 780284.	1.6	12
726	Profile of genetic variations in severely calcified carotid plaques by whole-exome sequencing. , 2020, 11, 286.		2
730	Meta-analysis diagnostic accuracy of SNP-based pathogenicity detection tools: a case of UTG1A1 gene mutations. International Journal of Molecular Epidemiology and Genetics, 2013, 4, 77-85.	0.4	7
731	Mutation near the binding interfaces at Î±-hemoglobin stabilizing protein is highly pathogenic. American Journal of Translational Research (discontinued), 2016, 8, 4224-4232.	0.0	12
732	Identification of a Novel KCNQ1 Frameshift Mutation and Review of the Literature among Iranian Long QT Families. Iranian Biomedical Journal, 2019, 23, 228-34.	0.4	0
733	A Novel Mutation of the KLK6 Gene in a Family With Knee Osteoarthritis. Frontiers in Genetics, 2021, 12, 784176.	1.1	3
734	Genomic analysis to screen potential genes and mutations in children with non-syndromic early onset severe obesity: a multicentre study in Turkey. Molecular Biology Reports, 2021, , 1.	1.0	3
735	Understanding the impact of missense mutations on the structure and function of the EDA gene in Xâ€linked hypohidrotic ectodermal dysplasia: A bioinformatics approach. Journal of Cellular Biochemistry, 2022, 123, 431-449.	1.2	5
736	MutTMPredictor: Robust and accurate cascade XGBoost classifier for prediction of mutations in transmembrane proteins. Computational and Structural Biotechnology Journal, 2021, 19, 6400-6416.	1.9	16
737	NOTCH3 mutations in a cohort of Portuguese patients within CADASIL spectrum phenotype. Neurogenetics, 2022, 23, 1-9.	0.7	6
738	What makes a good prediction? Feature importance and beginning to open the black box of machine learning in genetics. Human Genetics, 2022, 141, 1515-1528.	1.8	18
739	Computational Methods and Approaches in Pharmacogenomic Research. , 2022, , 53-83.		1
740	Pathogenicity Prediction of Single Amino Acid Variants with Machine Learning Model Based on Protein Structural Energies. IEEE/ACM Transactions on Computational Biology and Bioinformatics, 2021, PP, 1-1.	1.9	2
741	Genomic variants reducing expression of two endocytic receptors in 46,XY differences of sex development. Human Mutation, 2022, , .	1.1	0
742	Cytochrome b lineages of Haemoproteus tinniculi in the endangered saker falcon (Falco cherrug) in Saudi Arabia. Journal of King Saud University - Science, 2022, 34, 101755.	1.6	1

#	Article	IF	Citations
743	PPSNV: A Novel Predictor for Pathogenicity of Nonsynonymous SNV based on Ensemble Learning. , 2021, , .		0
744	Identification of a novel de novo mutation in the CTNNB1 gene in an Iranian patient with intellectual disability. Neurological Sciences, 2022, 43, 2859.	0.9	4
745	Comparative assessment of genes driving cancer and somatic evolution in non-cancer tissues: an update of the Network of Cancer Genes (NCG) resource. Genome Biology, 2022, 23, 35.	3.8	38
746	CerealsDB: A Whistle-Stop Tour of an Open Access SNP Resource. Methods in Molecular Biology, 2022, 2443, 133-146.	0.4	1
747	Analysis of coding variants in the human FTO gene from the gnomAD database. PLoS ONE, 2022, 17, e0248610.	1.1	1
748	Learning protein fitness models from evolutionary and assay-labeled data. Nature Biotechnology, 2022, 40, 1114-1122.	9.4	90
750	Machine learning techniques for pathogenicity prediction of non-synonymous single nucleotide polymorphisms in human body. Journal of Ambient Intelligence and Humanized Computing, 0, , 1.	3.3	0
751	Potential Involvement of NSD1, KRT24 and ACACA in the Genetic Predisposition to Colorectal Cancer. Cancers, 2022, 14, 699.	1.7	0
752	Mitochondrial Dynamics and Mitochondria-Lysosome Contacts in Neurogenetic Diseases. Frontiers in Neuroscience, 2022, 16, 784880.	1.4	8
753	Comprehensive Analysis of Co-Mutations Identifies Cooperating Mechanisms of Tumorigenesis. Cancers, 2022, 14, 415.	1.7	8
755	Germline sequence variants contributing to cancer susceptibility in South African breast cancer patients of African ancestry. Scientific Reports, 2022, 12, 802.	1.6	4
756	Case Report: Reinterpretation and Reclassification of ARSB:p.Arg159Cys Variant Identified in an Emirati Patient With Hearing Loss Caused by a Pathogenic Variant in the CDH23 Gene. Frontiers in Pediatrics, 2021, 9, 803732.	0.9	1
757	Genetic findings in patients with primary fibrotic atrial cardiomyopathy. European Journal of Medical Genetics, 2022, 65, 104429.	0.7	4
758	Machine learning methods for prediction of cancer driver genes: a survey paper. Briefings in Bioinformatics, 2022, 23, .	3.2	15
759	Genome Nexus: A Comprehensive Resource for the Annotation and Interpretation of Genomic Variants in Cancer. JCO Clinical Cancer Informatics, 2022, 6, e2100144.	1.0	4
760	Answer ALS, a large-scale resource for sporadic and familial ALS combining clinical and multi-omics data from induced pluripotent cell lines. Nature Neuroscience, 2022, 25, 226-237.	7.1	66
761	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
762	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

#	Article	IF	Citations
763	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
764	PirePred. Journal of Molecular Diagnostics, 2022, 24, 406-425.	1.2	1
765	Variant Identification in BARD1, PRDM9, RCC1, and RECQL in Patients with Ovarian Cancer by Targeted Next-generation Sequencing of DNA Pools. Cancer Prevention Research, 2022, 15, 151-160.	0.7	2
767	A phase 2 evaluation of pembrolizumab for recurrent Lynchâ€like versus sporadic endometrial cancers with microsatellite instability. Cancer, 2022, 128, 1206-1218.	2.0	28
768	An Engineering Approach Towards Multi-site Virtual Molecular Tumor Board Software. Communications in Computer and Information Science, 2021, , 156-170.	0.4	1
769	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
770	Computational Resources for the Interpretation of Variations in Cancer. Advances in Experimental Medicine and Biology, 2022, 1361, 177-198.	0.8	2
771	Novel RAB3GAP1 Mutation in the First Tunisian Family With Warburg Micro Syndrome. Journal		

#	Article	IF	Citations
784	AmazonForest: In Silico Metaprediction of Pathogenic Variants. Biology, 2022, 11, 538.	1.3	0
785	Case Report: Next-Generation Sequencing Identified a Novel Pair of Compound-Heterozygous Mutations of LPL Gene Causing Lipoprotein Lipase Deficiency. Frontiers in Genetics, 2022, 13, 831133.	1.1	2
786	Screening of OTULIN gene mutation with targeted next generation sequencing in Turkish populations and in silico analysis of these mutations. Molecular Biology Reports, 2022, 49, 4643-4652.	1.0	2
787	A CTNNB1â€altered medulloblastoma shows the immunophenotypic, DNA methylation and transcriptomic profiles of SHHâ€activated, and not WNTâ€activated, medulloblastoma. Neuropathology and Applied Neurobiology, 2022, 48, e12815.	1.8	0
788	Insights into the structure and dynamics of SARS-CoV-2 spike glycoprotein double mutant L452R-E484Q. 3 Biotech, 2022, 12, 87.	1.1	5
790	In vivo anti-tumor effect of PARP inhibition in IDH1/2 mutant MDS/AML resistant to targeted inhibitors of mutant IDH1/2. Leukemia, 2022, 36, 1313-1323.	3.3	11
791	OGDHL Variant rs2293239: A Potential Genetic Driver of Chinese Familial Depressive Disorder. Frontiers in Psychiatry, 2022, 13, 771950.	1.3	2
792	Molecular Characterization of Portuguese Patients with Hereditary Cerebellar Ataxia. Cells, 2022, 11, 981.	1.8	6
793	An expanded phenotype centric benchmark of variant prioritisation tools. Human Mutation, 2022, 43, 539-546.	1.1	9
795	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
796	Immunopeptidomic Analyses of Colorectal Cancers With and Without Microsatellite Instability. Molecular and Cellular Proteomics, 2022, 21, 100228.	2.5	20
797	Evaluating the impact of in silico predictors on clinical variant classification. Genetics in Medicine, 2022, 24, 924-930.	1.1	20
798	Recent Advances in Machine Learning Variant Effect Prediction Tools for Protein Engineering. Industrial & Engineering Chemistry Research, 2022, 61, 6235-6245.	1.8	15
799	MEFV gene allele frequency and genotype distribution in 3230 patientsâ€™ analyses by next generation sequencing methods. Gene, 2022, 827, 146447.	1.0	6
800	Assessment of 13 in silico pathogenicity methods on cancer-related variants. Computers in Biology and Medicine, 2022, 145, 105434.	3.9	2
801	Genetic landscape of human mitochondrial genome using whole-genome sequencing. Human Molecular Genetics, 2022, 31, 1747-1761.	1.4	4
802	Molecular-based precision oncology clinical decision making augmented by artificial intelligence. Emerging Topics in Life Sciences, 2021, 5, 757-764.	1.1	4
803	A Novel GEMIN4 Variant in a Consanguineous Family Leads to Neurodevelopmental Impairment with Severe Microcephaly, Spastic Quadriplegia, Epilepsy, and Cataracts. Genes, 2022, 13, 92.	1.0	6

#	Article	IF	Citations
804	Screening and Functional Analysis of TEK Mutations in Chinese Children With Primary Congenital Glaucoma. Frontiers in Genetics, 2021, 12, 764509.	1.1	7
805	In silico identification and characterization of small-molecule inhibitors specific to RhoG/Rac1 signaling pathway. Journal of Biomolecular Structure and Dynamics, 2023, 41, 560-580.	2.0	8
807	Simulation-Supported Engineering of Self-Adaptive Software Systems. , 2021, , .		2
808	The Novel Phosphatase Domain Mutations Q171R and Y65S Switch PTEN from Tumor Suppressor to Oncogene. Cells, 2021, 10, 3423.	1.8	1
809	Characterization of SLC34A2 as a Potential Prognostic Marker of Oncological Diseases. Biomolecules, 2021, 11, 1878.	1.8	4
810	LYRUS: a machine learning model for predicting the pathogenicity of missense variants. Bioinformatics Advances, 2022, 2, vbab045.	0.9	4
812	Whole mitochondrial genome sequencing of Malaysian patients with cardiomyopathy. PeerJ, 2022, 10, e13265.	0.9	1
846	Novel Mutation in the First Tunisian Family With Warburg Micro Syndrome.. Journal of Clinical		

#	Article	IF	Citations
859	Using Long-Term Follow-Up Data to Classify Genetic Variants in Newborn Screened Conditions. Frontiers in Genetics, 0, 13, .	1.1	2
860	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
861	Protein structural bioinformatics: An overview. Computers in Biology and Medicine, 2022, 147, 105695.	3.9	15
862	Interpreting protein variant effects with computational predictors and deep mutational scanning. DMM Disease Models and Mechanisms, 2022, 15, .	1.2	25
863	Dynamic insights into the effects of nonsynonymous polymorphisms (nsSNPs) on loss of TREM2 function. Scientific Reports, 2022, 12, .	1.6	5
865	Multiple primary malignances managed with surgical excision: a case report with next generation sequencing analysis. Molecular Biology Reports, 0, , .	1.0	1
866	PTBP2 â€“ a gene with relevance for both Anorexia nervosa and body weight regulation. Translational Psychiatry, 2022, 12, .	2.4	4
868	Functional and clinical analysis of five EDA variants associated with ectodermal dysplasia but with a hard-to-predict significance. Frontiers in Genetics, 0, 13, .	1.1	3
869	In-silico analysis of deleterious single nucleotide polymorphisms of PNMT gene. Molecular Simulation, 2022, 48, 1411-1425.	0.9	6
870	Novel <scp>MSH6</scp> mutation predicted metastasis in eyelid and periocular squamous cell carcinoma. Journal of the European Academy of Dermatology and Venereology, 2022, 36, 2331-2342.	1.3	2
871	Investigation of CACNA1I Cav3.3 Dysfunction in Hemiplegic Migraine. Frontiers in Molecular Neuroscience, 0, 15, .	1.4	7
872	ASXL1 mutations predict inferior molecular response to nilotinib treatment in chronic myeloid leukemia. Leukemia, 2022, 36, 2242-2249.	3.3	14
873	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
874	Computational Approaches for Investigating Disease-causing Mutations in Membrane Proteins: Database Development, Analysis and Prediction. Current Topics in Medicinal Chemistry, 2022, 22, 1766-1775.	1.0	3
875	Early-onset diabetes involving three consecutive generations had different clinical features from age-matched type 2 diabetes without a family history in China. Endocrine, 0, , .	1.1	0
876	Comprehensive in Silico Analyses of Single Nucleotide Variants of the Human Orthologues of 171 Murine Loci to Seek Novel Insights into the Genetics of Human Pigmentation. Proceedings of the Zoological Society, 0, , .	0.4	0
878	Definitive Chemoradiation and Durvalumab Consolidation for Locally Advanced, Unresectable KRAS-mutated Non-Small Cell Lung Cancer. Clinical Lung Cancer, 2022, 23, 620-629.	1.1	6
879	Identification of potential targets of the curcumin analog CCA-1.1 for glioblastoma treatment : integrated computational analysis and in vitro study. Scientific Reports, 2022, 12, .	1.6	4

#	Article	IF	Citations
880	Late-Onset Autosomal Dominant Macular Degeneration Caused by Deletion of the CRX Gene. Ophthalmology, 2023, 130, 68-76.	2.5	7
881	Medulloblastoma group 3 and 4 tumors comprise a clinically and biologically significant expression continuum reflecting human cerebellar development. Cell Reports, 2022, 40, 111162.	2.9	21
882	In silico mutational analysis to identify the role and pathogenicity of BCL-w missense variants. Journal of Genetic Engineering and Biotechnology, 2022, 20, 120.	1.5	2
883	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
884	Prevalence estimates of putatively pathogenic leptin variants in the gnomAD database. PLoS ONE, 2022, 17, e0266642.	1.1	2
886	m6A-TSHub: Unveiling the Context-Specific m6A Methylation and m6A-Affecting Mutations in 23 Human Tissues. Genomics, Proteomics and Bioinformatics, 2023, 21, 678-694.	3.0	21
888	Exploring Plausible Therapeutic Targets for Alzheimer's Disease using Multi-omics Approach, Machine Learning and Docking. Current Topics in Medicinal Chemistry, 2022, 22, 1868-1879.	1.0	4
889	Comprehensive analysis predicting effects of deleterious SNPs of human progesterone receptor gene on its structure and functions: a computational approach. Journal of Biomolecular Structure and Dynamics, 2023, 41, 8002-8017.	2.0	0
890	RMDisease V2.0: an updated database of genetic variants that affect RNA modifications with disease and trait implication. Nucleic Acids Research, 2023, 51, D1388-D1396.	6.5	23
891	Computational approaches for predicting variant impact: An overview from resources, principles to applications. Frontiers in Genetics, 0, 13, .	1.1	11
892	Roles of neuroligins in central nervous system development: focus on glial neuroligins and neuron neuroligins. Journal of Translational Medicine, 2022, 20, .	1.8	9
893	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
895	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
896	Variant predictions in congenital adrenal hyperplasia caused by mutations in CYP21A2. Frontiers in Pharmacology, 0, 13, .	1.6	2
897	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
898	Structural impact of pathogenic SNPs on Î²-tubulin using molecular dynamics study. Journal of Biomolecular Structure and Dynamics, 0, , 1-11.	2.0	0
899	An additive destabilising effect of compound T60I and V122I substitutions in ATTRv amyloidosis. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 0, , 1-12.	1.4	1
900	Specifications of the ACMG/AMP variant curation guidelines for myocilin: Recommendations from the clingen glaucoma expert panel. Human Mutation, 2022, 43, 2170-2186.	1.1	11

#	Article	IF	Citations
901	Mutational analysis of phospholipase C epsilon 1 gene in Egyptian children with steroid-resistant nephrotic syndrome. Egyptian Journal of Medical Human Genetics, 2022, 23, .	0.5	1
902	A bioinformatics approach to the identification of novel deleterious mutations of human TPMT through validated screening and molecular dynamics. Scientific Reports, 2022, 12, .	1.6	6
904	Biological network topology features predict gene dependencies in cancer cell-lines. Bioinformatics Advances, 0, , .	0.9	0
905	Genetic and clinical characteristics of ALS patients with NEK1 gene variants. Neurobiology of Aging, 2022, , .	1.5	0
906	The predictive role of ERBB2 point mutations in metastatic colorectal cancer: A systematic review. Cancer Treatment Reviews, 2023, 112, 102488.	3.4	6
907	Next-generation sequencing of postmortem molecular markers to support for medicolegal autopsy. Forensic Science International: Reports, 2022, 6, 100300.	0.4	0
908	Phenotypic screening models for rapid diagnosis of genetic variants and discovery of personalized therapeutics. Molecular Aspects of Medicine, 2023, 91, 101153.	2.7	7
909	Isolation, identification and application of Aspergillus oryzae BL18 with high protease activity as starter culture in doubanjiang (broad bean paste) fermentation. Food Bioscience, 2023, 51, 102225.	2.0	4
910	Reporting Two Novel Mutations in Two Iranian Families with Cystic Fibrosis, Molecular and Bioinformatic Analysis. Iranian Biomedical Journal, 2022, 26, 398-405.	0.4	1
912	Insights on variant analysis in silico tools for pathogenicity prediction. Frontiers in Genetics, 0, 13, .	1.1	17
914	Evaluation of in silico predictors on short nucleotide variants in HBA1, HBA2, and HBB associated with haemoglobinopathies. ELife, 0, 11, .	2.8	6
915	Integrative Meta-Analysis of Huntingtonâ€™s Disease Transcriptome Landscape. Genes, 2022, 13, 2385.	1.0	3
916	Diverse monogenic subforms of human spermatogenic failure. Nature Communications, 2022, 13, .	5.8	17
917	A novel homozygous nonsense <scp> NDNF </scp> variant in Kallmann syndrome. American Journal of Medical Genetics, Part A, 0, , .	0.7	0
918	MUG: A mutation overview of GPCR subfamily A17 receptors. Computational and Structural Biotechnology Journal, 2023, 21, 586-600.	1.9	0
920	Calibration of computational tools for missense variant pathogenicity classification and ClinGen recommendations for PP3/BP4 criteria. American Journal of Human Genetics, 2022, 109, 2163-2177.	2.6	124
921	Rapid Targeted Sequencing Using Dried Blood Spot Samples for Patients With Suspected Actionable Genetic Diseases. Annals of Laboratory Medicine, 2023, 43, 280-289.	1.2	4
924	Identification of a Novel Non-Canonical Splice-Site Variant in ABCD1. Journal of Clinical Medicine, 2023, 12, 473.	1.0	1

#	Article	IF	Citations
925	Rare variant analyses across multiethnic cohorts identify novel genes for refractive error. Communications Biology, 2023, 6, .	2.0	2
926	A case report of maturity-onset diabetes of the young (MODY12) in a Chinese Han patient with a novel ABCC8 gene mutation. Medicine (United States), 2022, 101, e32139.	0.4	1
927	Application of an in silico approach identifies a genetic locus within ITGB2, and its interactions with HSPG2 and FGF9, to be associated with anterior cruciate ligament rupture risk. European Journal of Sport Science, 2023, 23, 2098-2108.	1.4	0
928	Computational analysis of structural and functional evaluation of the deleterious missense variants in the human CTLA4 gene. Journal of Biomolecular Structure and Dynamics, 2023, 41, 14179-14196.	2.0	0
930	Exploring genotypeâ€“phenotype correlations in glutaric aciduria type 1. Journal of Inherited Metabolic Disease, 2023, 46, 371-390.	1.7	8
931	Identification of a novel homozygous mutation of the BCKDHB gene in an Iranian patient with maple syrup disease using next-generation sequencing. , 2023, 36, 201173.		0
932	A case of neonatal osteofibrous dysplasia with novel CDK12 and DDR2 mutations. Bone Reports, 2023, 18, 101666.	0.2	0
933	Comprehensive in-silico analysis of deleterious SNPs in APOC2 and APOA5 and their differential expression in cancer and cardiovascular diseases conditions. Genomics, 2023, 115, 110567.	1.3	1
934	Deep genomic analysis of malignant peripheral nerve sheath tumor cell lines challenges current malignant peripheral nerve sheath tumor diagnosis. IScience, 2023, 26, 106096.	1.9	5
935	De Novo Mutations Contributes Approximately 7% of Pathogenicity in Inherited Eye Diseases. , 2023, 64, 5.		3
936	Integration of deep learning with Ramachandran plot molecular dynamics simulation for genetic variant classification. IScience, 2023, 26, 106122.	1.9	2
937	Common and rare variant associations with latent traits underlying depression, bipolar disorder, and schizophrenia. Translational Psychiatry, 2023, 13, .	2.4	2
938	Molecular Dynamic Simulation Analysis of a Novel Missense Variant in CYB5R3 Gene in Patients with Methemoglobinemia. Medicina (Lithuania), 2023, 59, 379.	0.8	5
939	N6-methyladenosine modification in 18S rRNA promotes tumorigenesis and chemoresistance via HSF4b/HSP90B1/mutant p53 axis. Cell Chemical Biology, 2023, 30, 144-158.e10.	2.5	2
940	Hypothesis-free phenotype prediction within a genetics-first framework. Nature Communications, 2023, 14, .	5.8	1
941	Genomic Alterations, Gene Expression Profiles and Functional Enrichment of Normal-Karyotype Acute Myeloid Leukaemia Based on Targeted Next-Generation Sequencing. Cancers, 2023, 15, 1386.	1.7	0
943	Computational and mitochondrial functional studies of novel compound heterozygous variants in SPATA5 gene support a causal link with epileptogenic encephalopathy. Human Genomics, 2023, 17, .	1.4	1
944	Iroquois Family Genes in Gastric Carcinogenesis: A Comprehensive Review. Genes, 2023, 14, 621.	1.0	0

#	Article	IF	Citations
945	Whole exome sequencing in unexplained recurrent miscarriage families identified novel pathogenic genetic causes of euploid miscarriage. Human Reproduction, 2023, 38, 1003-1018.	0.4	3
946	Identification of Genetic Alterations in Rapid Progressive Glioblastoma by Use of Whole Exome Sequencing. Diagnostics, 2023, 13, 1017.	1.3	3
947	Hypercholesterolemia in the Malaysian Cohort Participants: Genetic and Non-Genetic Risk Factors. Genes, 2023, 14, 721.	1.0	1
948	Pharmacological Chaperones and Protein Conformational Diseases: Approaches of Computational Structural Biology. International Journal of Molecular Sciences, 2023, 24, 5819.	1.8	3
949	Next-Generation Sequencing (NGS) Analysis Illustrates the Phenotypic Variability of Collagen Type IV Nephropathies. Genes, 2023, 14, 764.	1.0	0
951	Identification of GLI1 and KIAA0825 Variants in Two Families with Postaxial Polydactyly. Genes, 2023, 14, 869.	1.0	0
952	Molecular and Sociodemographic Colorectal Cancer Disparities in Latinos Living in Puerto Rico. Genes, 2023, 14, 894.	1.0	0
954	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
955	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
984	AI in Genomics and Epigenomics. Healthy Ageing and Longevity, 2023, , 217-243.	0.2	0
1001	Machine Learning for Protein Engineering. Challenges and Advances in Computational Chemistry and Physics, 2023, , 277-311.	0.6	1
1037	Computational approaches for identifying disease-causing mutations in proteins. Advances in Protein Chemistry and Structural Biology, 2024, , 141-171.	1.0	0