

Performance of mutation pathogenicity prediction met

Human Mutation

32, 358-368

DOI: [10.1002/humu.21445](https://doi.org/10.1002/humu.21445)

Citation Report

#	ARTICLE	IF	CITATIONS
1	EX-HOM (EXome HOMozygosity): A Proof of Principle. <i>Human Heredity</i> , 2011, 72, 45-53.	0.4	27
2	Functional studies of single-nucleotide polymorphic variants of human glutathione transferase T1-1 involving residues in the dimer interface. <i>Archives of Biochemistry and Biophysics</i> , 2011, 513, 87-93.	1.4	2
3	Bioinformatics challenges for personalized medicine. <i>Bioinformatics</i> , 2011, 27, 1741-1748.	1.8	223
4	A new disease-specific machine learning approach for the prediction of cancer-causing missense variants. <i>Genomics</i> , 2011, 98, 310-317.	1.3	68
5	Performance of mutation pathogenicity prediction methods on missense variants. <i>Human Mutation</i> , 2011, 32, 358-368.	1.1	468
6	Preliminary Analysis of the Nonsynonymous Polymorphism rs17563 in <i>BMP4</i> Gene in Brazilian Population Suggests Protection for Nonsyndromic Cleft Lip and Palate. <i>Plastic Surgery International</i> , 2012, 2012, 1-5.	0.7	13
7	Heuristic Methods for Finding Pathogenic Variants in Gene Coding Sequences. <i>Journal of the American Heart Association</i> , 2012, 1, e002642.	1.6	12
8	Strain-Dependent Airway Hyperresponsiveness and a Chromosome 7 Locus of Elevated Lymphocyte Numbers in Cystic Fibrosis Transmembrane Conductance Regulator-Deficient Mice. <i>Journal of Immunology</i> , 2012, 188, 2297-2304.	0.4	6
9	Role of rare variants in undetermined multiple adenomatous polyposis and early-onset colorectal cancer. <i>Journal of Human Genetics</i> , 2012, 57, 709-716.	1.1	9
10	KD4v: comprehensible knowledge discovery system for missense variant. <i>Nucleic Acids Research</i> , 2012, 40, W71-W75.	6.5	26
11	High-Throughput Sequencing and Rare Genetic Diseases. <i>Molecular Syndromology</i> , 2012, 3, 197-203.	0.3	6
12	MLPA and sequence analysis of DPY19L2 reveals point mutations causing globozoospermia. <i>Human Reproduction</i> , 2012, 27, 2549-2558.	0.4	62
13	Utility of gene-specific algorithms for predicting pathogenicity of uncertain gene variants. <i>Journal of the American Medical Informatics Association: JAMIA</i> , 2012, 19, 207-211.	2.2	25
14	Involvement of the WNT and FGF signaling pathways in non-isolated anorectal malformations: Sequencing analysis of WNT3A, WNT5A, WNT11, DACT1, FGF10, FGFR2 and the T gene. <i>International Journal of Molecular Medicine</i> , 2012, 30, 1459-1464.	1.8	24
15	The genetics and neuropathology of neurodegenerative disorders: perspectives and implications for research and clinical practice. <i>Acta Neuropathologica</i> , 2012, 124, 297-303.	3.9	12
16	In silico prediction of a disease-associated STIL mutant and its affect on the recruitment of centromere protein J (CENPJ). <i>FEBS Open Bio</i> , 2012, 2, 285-293.	1.0	53
17	Identifying disease mutations in genomic medicine settings: current challenges and how to accelerate progress. <i>Genome Medicine</i> , 2012, 4, 58.	3.6	68
18	SIFT web server: predicting effects of amino acid substitutions on proteins. <i>Nucleic Acids Research</i> , 2012, 40, W452-W457.	6.5	1,838

#	ARTICLE	IF	CITATIONS
19	Functional genomics based prioritization of potential nsSNPs in EPHX1, GSTT1, GSTM1 and GSTP1 genes for breast cancer susceptibility studies. <i>Genomics</i> , 2012, 99, 330-339.	1.3	17
20	TMEM127 Screening in a Large Cohort of Patients with Pheochromocytoma and/or Paraganglioma. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012, 97, E805-E809.	1.8	57
21	Identification of rare and frequent variants of the CASR gene by high-resolution melting. <i>Clinica Chimica Acta</i> , 2012, 413, 605-611.	0.5	16
22	Assessment of the potential pathogenicity of missense mutations identified in the GTPase-activating protein (GAP)-related domain of the neurofibromatosis type-1 (<i>NF1</i>) gene. <i>Human Mutation</i> , 2012, 33, 1687-1696.	1.1	21
23	Somatic mitochondrial DNA mutations in cancer escape purifying selection and high pathogenicity mutations lead to the oncogenic phenotype: pathogenicity analysis of reported somatic mtDNA mutations in tumors. <i>BMC Cancer</i> , 2012, 12, 53.	1.1	75
24	Patient-controlled encrypted genomic data: an approach to advance clinical genomics. <i>BMC Medical Genomics</i> , 2012, 5, 31.	0.7	9
25	An Excess of Deleterious Variants in VEGF-A Pathway Genes in Down-Syndrome-Associated Atrioventricular Septal Defects. <i>American Journal of Human Genetics</i> , 2012, 91, 646-659.	2.6	99
26	High-resolution melting analysis of 15 genes in 60 patients with cytochrome-c oxidase deficiency. <i>Journal of Human Genetics</i> , 2012, 57, 442-448.	1.1	18
27	Improving the prediction of the functional impact of cancer mutations by baseline tolerance transformation. <i>Genome Medicine</i> , 2012, 4, 89.	3.6	91
28	Harnessing Information Using Genomic Platforms. , 2012, , 727-744.		0
29	Bioinformatics Approaches to the Functional Profiling of Genetic Variants. , 0, , .		0
30	Bioinformatics for personal genome interpretation. <i>Briefings in Bioinformatics</i> , 2012, 13, 495-512.	3.2	62
31	Disease gene identification strategies for exome sequencing. <i>European Journal of Human Genetics</i> , 2012, 20, 490-497.	1.4	412
32	PON-P: Integrated predictor for pathogenicity of missense variants. <i>Human Mutation</i> , 2012, 33, 1166-1174.	1.1	88
33	The inherited ataxias: Genetic heterogeneity, mutation databases, and future directions in research and clinical diagnostics. <i>Human Mutation</i> , 2012, 33, 1324-1332.	1.1	86
34	Exome sequencing and the genetic basis of complex traits. <i>Nature Genetics</i> , 2012, 44, 623-630.	9.4	340
35	Exploring the Implications of INDELS in Neuropsychiatric Genetics: Challenges and Perspectives. <i>Journal of Molecular Neuroscience</i> , 2012, 47, 419-424.	1.1	1
36	Established and Emerging Trends in Computational Drug Discovery in the Structural Genomics Era. <i>Chemistry and Biology</i> , 2012, 19, 29-41.	6.2	57

#	ARTICLE	IF	CITATIONS
37	How to evaluate performance of prediction methods? Measures and their interpretation in variation effect analysis. BMC Genomics, 2012, 13, S2.	1.2	223
38	Predicting cancer-associated germline variations in proteins. BMC Genomics, 2012, 13, S8.	1.2	28
39	Novel mitochondrial DNA mutations responsible for maternally inherited nonsyndromic hearing loss. Human Mutation, 2012, 33, 681-689.	1.1	34
40	Classification of mismatch repair gene missense variants with PON-MMR. Human Mutation, 2012, 33, 642-650.	1.1	27
41	DDIG-in: discriminating between disease-associated and neutral non-frameshifting micro-indels. Genome Biology, 2013, 14, R23.	13.9	63
42	The Effects of Non-Synonymous Single Nucleotide Polymorphisms (nsSNPs) on Protein-Protein Interactions. Journal of Molecular Biology, 2013, 425, 3949-3963.	2.0	184
43	Collective judgment predicts disease-associated single nucleotide variants. BMC Genomics, 2013, 14, S2.	1.2	213
44	WS-SNPs&GO: a web server for predicting the deleterious effect of human protein variants using functional annotation. BMC Genomics, 2013, 14, S6.	1.2	248
45	Computational approaches to identify functional genetic variants in cancer genomes. Nature Methods, 2013, 10, 723-729.	9.0	161
46	Identification of COL6A2 mutations in progressive myoclonus epilepsy syndrome. Human Genetics, 2013, 132, 275-283.	1.8	21
47	Novel CYP2B6 Enzyme Variants in a Rwandese Population: Functional Characterization and Assessment of In Silico Prediction Tools. Human Mutation, 2013, 34, 725-734.	1.1	28
48	Molecular Mechanisms of Disease-Causing Missense Mutations. Journal of Molecular Biology, 2013, 425, 3919-3936.	2.0	242
49	Extrapolating the effect of deleterious nsSNPs in the binding adaptability of flavopiridol with CDK7 protein: a molecular dynamics approach. Human Genomics, 2013, 7, 10.	1.4	47
50	Mortalin mutations are not a frequent cause of early-onset Parkinson disease. Neurobiology of Aging, 2013, 34, 2694.e19-2694.e20.	1.5	15
51	The Sac1 Domain of <i>SYNJ1</i> Identified Mutated in a Family with Early-Onset Progressive Parkinsonism with Generalized Seizures. Human Mutation, 2013, 34, 1200-1207.	1.1	302
52	Status quo of annotation of human disease variants. BMC Bioinformatics, 2013, 14, 352.	1.2	3
53	Towards Precision Medicine: Advances in Computational Approaches for the Analysis of Human Variants. Journal of Molecular Biology, 2013, 425, 4047-4063.	2.0	122
54	<i>RNF43</i> is a tumour suppressor gene mutated in mucinous tumours of the ovary. Journal of Pathology, 2013, 229, 469-476.	2.1	102

#	ARTICLE	IF	CITATIONS
55	From the periphery to centre stage: de novo single nucleotide variants play a key role in human genetic disease. <i>Journal of Medical Genetics</i> , 2013, 50, 203-211.	1.5	33
56	VariBench: A Benchmark Database for Variations. <i>Human Mutation</i> , 2013, 34, 42-49.	1.1	129
57	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. <i>Human Mutation</i> , 2013, 34, 255-265.	1.1	80
58	Predicting the Functional, Molecular, and Phenotypic Consequences of Amino Acid Substitutions using Hidden Markov Models. <i>Human Mutation</i> , 2013, 34, 57-65.	1.1	1,057
59	Prediction of phenotypes of missense mutations in human proteins from biological assemblies. <i>Proteins: Structure, Function and Bioinformatics</i> , 2013, 81, 199-213.	1.5	20
60	Predicting the functional consequences of non-synonymous DNA sequence variants – evaluation of bioinformatics tools and development of a consensus strategy. <i>Genomics</i> , 2013, 102, 223-228.	1.3	89
61	Exome Sequencing Identifies GNB4 Mutations as a Cause of Dominant Intermediate Charcot-Marie-Tooth Disease. <i>American Journal of Human Genetics</i> , 2013, 92, 422-430.	2.6	46
62	News from the Protein Mutability Landscape. <i>Journal of Molecular Biology</i> , 2013, 425, 3937-3948.	2.0	72
63	Congruency in the prediction of pathogenic missense mutations: state-of-the-art web-based tools. <i>Briefings in Bioinformatics</i> , 2013, 14, 448-459.	3.2	79
64	Molecular diagnostic testing for congenital disorders of glycosylation (CDG): Detection rate for single gene testing and next generation sequencing panel testing. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 78-85.	0.5	37
65	Simultaneous Sequencing of 24 Genes Associated with Steroid-Resistant Nephrotic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2013, 8, 637-648.	2.2	152
66	Predicting the Impact of Single-Nucleotide Polymorphisms in CDK2-Flavopiridol Complex by Molecular Dynamics Analysis. <i>Cell Biochemistry and Biophysics</i> , 2013, 66, 681-695.	0.9	14
67	Protein Structural Based Analysis for Interpretation of Missense Variants at the Genomics Era: Using MNGIE Disease as an Example. , 2013, , 79-96.		1
68	Understanding the immunological impact of the human mutation explosion. <i>Trends in Immunology</i> , 2013, 34, 99-106.	2.9	13
69	Identification of a novel IVD mutation in a consanguineous family with isovaleric acidemia. <i>Gene</i> , 2013, 513, 297-300.	1.0	5
70	The ACMSD gene, involved in tryptophan metabolism, is mutated in a family with cortical myoclonus, epilepsy, and parkinsonism. <i>Journal of Molecular Medicine</i> , 2013, 91, 1399-1406.	1.7	111
71	Functional characterization of novel genotypes and cellular oxidative stress studies in propionic acidemia. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 731-740.	1.7	44
72	Predicting the functional consequences of cancer-associated amino acid substitutions. <i>Bioinformatics</i> , 2013, 29, 1504-1510.	1.8	208

#	ARTICLE	IF	CITATIONS
73	In Vivo&/em> Modeling of the Morbid Human Genome using Danio rerio&/em>. Journal of Visualized Experiments, 2013, , e50338.	0.2	49
74	Prevalence of rare mitochondrial DNA mutations in mitochondrial disorders. Journal of Medical Genetics, 2013, 50, 704-714.	1.5	95
75	Functional Recurrent Mutations in the Human Mitochondrial Phylogeny: Dual Roles in Evolution and Disease. Genome Biology and Evolution, 2013, 5, 876-890.	1.1	60
76	Knowledge Discovery in Variant Databases Using Inductive Logic Programming. Bioinformatics and Biology Insights, 2013, 7, BBI.S11184.	1.0	6
77	Novel <i>OPA1</i> missense mutation in a family with optic atrophy and severe widespread neurological disorder. Acta Ophthalmologica, 2013, 91, e225-31.	0.6	39
78	Lessons from postgenome-wide association studies: functional analysis of cancer predisposition loci. Journal of Internal Medicine, 2013, 274, 414-424.	2.7	24
79	Comparative analysis of IRF6 variants in families with Van der Woude syndrome and popliteal pterygium syndrome using public whole-exome databases. Genetics in Medicine, 2013, 15, 338-344.	1.1	47
80	Lessons from next-generation sequencing analysis in hematological malignancies. Blood Cancer Journal, 2013, 3, e127-e127.	2.8	50
81	Functional and Structural Analysis of C-Terminal BRCA1 Missense Variants. PLoS ONE, 2013, 8, e61302.	1.1	16
82	Structural and Population-Based Evaluations of TBC1D1 p.Arg125Trp. PLoS ONE, 2013, 8, e63897.	1.1	8
83	Novel Missense Variants of ZFPM2/FOG2 Identified in Conotruncal Heart Defect Patients Do Not Impair Interaction with GATA4. PLoS ONE, 2014, 9, e102379.	1.1	21
85	Next Generation Sequencing for Disorders of Sex Development. Endocrine Development, 2014, 27, 53-62.	1.3	14
86	Identification of Splicing Defects Caused by Mutations in the Dysferlin Gene. Human Mutation, 2014, 35, 1532-1541.	1.1	22
87	Genetic testing for nephrotic syndrome and FSGS in the era of next-generation sequencing. Kidney International, 2014, 85, 1030-1038.	2.6	61
88	Corneal Endothelial Findings in a Czech Patient with Compound Heterozygous Mutations in KERA. Ophthalmic Genetics, 2014, 35, 252-254.	0.5	8
89	Hematogones in the Peripheral Blood of a 5½-Month-Old Boy with Cyclic Neutropenia Due to Heterozygous, Novel ELANE Gene Mutation p.Q97P, c.290 A>C. Pediatric and Developmental Pathology, 2014, 17, 393-399.	0.5	3
90	EYA4 is inactivated biallelically at a high frequency in sporadic lung cancer and is associated with familial lung cancer risk. Oncogene, 2014, 33, 4464-4473.	2.6	41
91	Integrating <i>In Silico</i> Prediction Methods, Molecular Docking, and Molecular Dynamics Simulation to Predict the Impact of ALK Missense Mutations in Structural Perspective. BioMed Research International, 2014, 2014, 1-14.	0.9	40

#	ARTICLE	IF	CITATIONS
92	PredictSNP: Robust and Accurate Consensus Classifier for Prediction of Disease-Related Mutations. PLoS Computational Biology, 2014, 10, e1003440.	1.5	593
93	The road from next-generation sequencing to personalized medicine. Personalized Medicine, 2014, 11, 523-544.	0.8	40
94	Genetic Variations and Diseases in UniProtKB/SwissProt: The Ins and Outs of Expert Manual Curation. Human Mutation, 2014, 35, 927-935.	1.1	51
95	Majority Vote and Other Problems when using Computational Tools. Human Mutation, 2014, 35, 912-914.	1.1	15
96	Use of Contemporary Genetics in Cardiovascular Diagnosis. Circulation, 2014, 130, 1971-1980.	1.6	7
97	Functionally compromisedCHD7alleles in patients with isolated GnRH deficiency. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 17953-17958.	3.3	74
98	Enrichment of LOVD-USHbases with 152 <i>USH2A</i> Genotypes Defines an Extensive Mutational Spectrum and Highlights Missense Hotspots. Human Mutation, 2014, 35, 1179-1186.	1.1	55
99	The identification of mitochondrial DNA variants in glioblastoma multiforme. Acta Neuropathologica Communications, 2014, 2, 1.	2.4	143
100	Predicting virus mutations through statistical relational learning. BMC Bioinformatics, 2014, 15, 309.	1.2	6
101	SuSPect: Enhanced Prediction of Single Amino Acid Variant (SAV) Phenotype Using Network Features. Journal of Molecular Biology, 2014, 426, 2692-2701.	2.0	189
102	Mutations in VPS26A are not a frequent cause of Parkinson's disease. Neurobiology of Aging, 2014, 35, 1512.e1-1512.e2.	1.5	7
103	Systematic investigation of predicted effect of nonsynonymous SNPs in human prion protein gene: a molecular modeling and molecular dynamics study. Journal of Biomolecular Structure and Dynamics, 2014, 32, 289-300.	2.0	6
104	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
105	Computational SNP Analysis: Current Approaches and Future Prospects. Cell Biochemistry and Biophysics, 2014, 68, 233-239.	0.9	42
106	Recessive dystonia-ataxia syndrome in a Turkish family caused by a COX20 (FAM36A) mutation. Journal of Neurology, 2014, 261, 207-212.	1.8	40
107	Mutation p.Leu128Pro in the 1A domain of K16 causes pachyonychia congenita with focal palmoplantar keratoderma in a Chinese family. European Journal of Pediatrics, 2014, 173, 737-741.	1.3	1
108	Molecular genetic epidemiology of human diseases: from patterns to predictions. Human Genetics, 2014, 133, 425-430.	1.8	10
109	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

#	ARTICLE	IF	CITATIONS
110	Novel genetic abnormalities in Bernard-Soulier syndrome in India. <i>Annals of Hematology</i> , 2014, 93, 381-384.	0.8	9
111	Application of Evolutionary Based in Silico Methods to Predict the Impact of Single Amino Acid Substitutions in Vitelliform Macular Dystrophy. <i>Advances in Protein Chemistry and Structural Biology</i> , 2014, 94, 177-267.	1.0	10
113	Novel variants in the SOHLH2 gene are implicated in human premature ovarian failure. <i>Fertility and Sterility</i> , 2014, 101, 1104-1109.e6.	0.5	50
114	Computational Identification of Pathogenic Associated nsSNPs and its Structural Impact in UROD Gene: A Molecular Dynamics Approach. <i>Cell Biochemistry and Biophysics</i> , 2014, 70, 735-746.	0.9	3
115	An Integrated in Silico Approach to Analyze the Involvement of Single Amino Acid Polymorphisms in FANCD1/BRCA2-PALB2 and FANCD1/BRCA2-RAD51 Complex. <i>Cell Biochemistry and Biophysics</i> , 2014, 70, 939-956.	0.9	8
116	Performance of Protein Disorder Prediction Programs on Amino Acid Substitutions. <i>Human Mutation</i> , 2014, 35, 794-804.	1.1	20
117	Pathological Unfoldomics of Uncontrolled Chaos: Intrinsically Disordered Proteins and Human Diseases. <i>Chemical Reviews</i> , 2014, 114, 6844-6879.	23.0	231
119	EnsembleGASVR: a novel ensemble method for classifying missense single nucleotide polymorphisms. <i>Bioinformatics</i> , 2014, 30, 2324-2333.	1.8	17
120	Lessons learned from genetic testing for channelopathies. <i>Lancet Neurology</i> , The, 2014, 13, 1068-1070.	4.9	4
121	Influence of the SNPs on the structural stability of CBS protein: Insight from molecular dynamics simulations. <i>Frontiers in Biology</i> , 2014, 9, 504-518.	0.7	5
122	Clinical utility of genetic testing in pediatric drug-resistant epilepsy: A pilot study. <i>Epilepsy and Behavior</i> , 2014, 37, 241-248.	0.9	47
123	Proper reporting of predictor performance. <i>Nature Methods</i> , 2014, 11, 781-781.	9.0	7
124	Next Generation Sequencing and the Future of Genetic Diagnosis. <i>Neurotherapeutics</i> , 2014, 11, 699-707.	2.1	126
125	Ranking non-synonymous single nucleotide polymorphisms based on disease concepts. <i>Human Genomics</i> , 2014, 8, 11.	1.4	163
126	Integrative visual analysis of protein sequence mutations. <i>BMC Proceedings</i> , 2014, 8, S2.	1.8	13
127	MutaCYP: Classification of missense mutations in human cytochromes P450. <i>BMC Medical Genomics</i> , 2014, 7, 47.	0.7	8
128	Cardiac Potassium Channel Subtypes: New Roles in Repolarization and Arrhythmia. <i>Physiological Reviews</i> , 2014, 94, 609-653.	13.1	181
129	Combined sequence and sequence-structure-based methods for analyzing RAAS gene SNPs: a computational approach. <i>Journal of Receptor and Signal Transduction Research</i> , 2014, 34, 513-526.	1.3	10

#	ARTICLE	IF	CITATIONS
131	Novel mutations of PKD genes in the Czech population with autosomal dominant polycystic kidney disease. BMC Medical Genetics, 2014, 15, 41.	2.1	23
132	PhenoVar: a phenotype-driven approach in clinical genomics for the diagnosis of polymalformative syndromes. BMC Medical Genetics, 2014, 7, 22.	0.7	22
133	Unraveling Cellular Phenotypes of Novel <i>TorsinA/TOR1A</i> Mutations. Human Mutation, 2014, 35, 1114-1122.	1.1	34
134	Translating Genetic Association Signals for Diabetes and Metabolic Traits into Molecular Mechanisms for Disease. Frontiers in Diabetes, 2014, , 133-145.	0.4	0
135	Response to Cederbaum. Genetics in Medicine, 2015, 17, 1013-1014.	1.1	0
136	A rigorous approach for selection of optimal variant sets for carrier screening with demonstration of clinical utility. Molecular Genetics & Genomic Medicine, 2015, 3, 363-373.	0.6	7
137	Identification of novel PKD1 and PKD2 mutations in a Chinese population with autosomal dominant polycystic kidney disease. Scientific Reports, 2015, 5, 17468.	1.6	19
138	A Novel <i>UMOD</i> Gene Mutation Associated with Uromodulin-associated Kidney Disease in a Young Woman with Moderate Kidney Dysfunction. Internal Medicine, 2015, 54, 631-635.	0.3	6
139	CoagVDb: a comprehensive database for coagulation factors and their associated SAPs. Biological Research, 2015, 48, 35.	1.5	4
140	Characterization of All Possible Single-Nucleotide Change Caused Amino Acid Substitutions in the Kinase Domain of Bruton Tyrosine Kinase. Human Mutation, 2015, 36, 638-647.	1.1	39
141	BALL-SNP: combining genetic and structural information to identify candidate non-synonymous single nucleotide polymorphisms. Genome Medicine, 2015, 7, 65.	3.6	9
142	Performance of In Silico Tools for the Evaluation of <i>UGT1A1</i> Missense Variants. Human Mutation, 2015, 36, 1215-1225.	1.1	21
143	Analysis of genetic variation and potential applications in genome-scale metabolic modeling. Frontiers in Bioengineering and Biotechnology, 2015, 3, 13.	2.0	30
144	Whole-Exome Sequencing in the Differential Diagnosis of Primary Adrenal Insufficiency in Children. Frontiers in Endocrinology, 2015, 6, 113.	1.5	44
145	Insight into Neutral and Disease-Associated Human Genetic Variants through Interpretable Predictors. PLoS ONE, 2015, 10, e0120729.	1.1	2
146	Lessons Learned from Whole Exome Sequencing in Multiplex Families Affected by a Complex Genetic Disorder, Intracranial Aneurysm. PLoS ONE, 2015, 10, e0121104.	1.1	32
147	Increased Aggregation Is More Frequently Associated to Human Disease-Associated Mutations Than to Neutral Polymorphisms. PLoS Computational Biology, 2015, 11, e1004374.	1.5	38
148	Identifying Highly Penetrant Disease Causal Mutations Using Next Generation Sequencing: Guide to Whole Process. BioMed Research International, 2015, 2015, 1-16.	0.9	7

#	ARTICLE	IF	CITATIONS
149	Integration of structural dynamics and molecular evolution via protein interaction networks: a new era in genomic medicine. <i>Current Opinion in Structural Biology</i> , 2015, 35, 135-142.	2.6	29
150	Comparison of predicted and actual consequences of missense mutations. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, E5189-98.	3.3	200
151	NSD1 mutations generate a genome-wide DNA methylation signature. <i>Nature Communications</i> , 2015, 6, 10207.	5.8	170
152	Rational laboratory diagnostics of primary immunodeficiency disorders. <i>Laboratoriums Medizin</i> , 2015, 39, 343-354.	0.1	3
153	Comparison and integration of deleteriousness prediction methods for nonsynonymous SNVs in whole exome sequencing studies. <i>Human Molecular Genetics</i> , 2015, 24, 2125-2137.	1.4	892
154	Insights into genotype-phenotype correlations from <i>CREBBP</i> point mutation screening in a cohort of 46 Rubinstein-Taybi syndrome patients. <i>Clinical Genetics</i> , 2015, 88, 431-440.	1.0	51
155	Predicting survival in head and neck squamous cell carcinoma from TP53 mutation. <i>Human Genetics</i> , 2015, 134, 497-507.	1.8	31
156	Pathogenicity prediction of non-synonymous single nucleotide variants in dilated cardiomyopathy. <i>Briefings in Bioinformatics</i> , 2015, 16, 769-779.	3.2	10
157	Prediction of the pathogenicity of antithrombin sequence variations by in silico methods. <i>Thrombosis Research</i> , 2015, 135, 404-409.	0.8	16
158	Systems Pharmacology and Pharmacogenomics for Drug Discovery and Development. , 2015, , 173-193.		0
159	DDIG-in: detecting disease-causing genetic variations due to frameshifting indels and nonsense mutations employing sequence and structural properties at nucleotide and protein levels. <i>Bioinformatics</i> , 2015, 31, 1599-1606.	1.8	52
160	Variability in pathogenicity prediction programs: impact on clinical diagnostics. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 99-110.	0.6	44
161	MitImpact: an Exhaustive Collection of Pre-computed Pathogenicity Predictions of Human Mitochondrial Non-synonymous Variants. <i>Human Mutation</i> , 2015, 36, E2413-E2422.	1.1	61
162	<i>DNAJC13</i> genetic variants in parkinsonism. <i>Movement Disorders</i> , 2015, 30, 273-278.	2.2	42
163	Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. <i>Genetics in Medicine</i> , 2015, 17, 405-424.	1.1	20,455
164	The Evaluation of Tools Used to Predict the Impact of Missense Variants Is Hindered by Two Types of Circularity. <i>Human Mutation</i> , 2015, 36, 513-523.	1.1	283
165	Novel GCH1 variant in Dopa-responsive dystonia and Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2015, 21, 394-397.	1.1	29
167	The Contribution of Missense Mutations in Core and Rim Residues of Protein-Protein Interfaces to Human Disease. <i>Journal of Molecular Biology</i> , 2015, 427, 2886-2898.	2.0	105

#	ARTICLE	IF	CITATIONS
168	Missense variants in CFTR nucleotide-binding domains predict quantitative phenotypes associated with cystic fibrosis disease severity. <i>Human Molecular Genetics</i> , 2015, 24, 1908-1917.	1.4	11
169	Functional and in silico assessment of MAX variants of unknown significance. <i>Journal of Molecular Medicine</i> , 2015, 93, 1247-1255.	1.7	25
170	Accumulation of rare variants in the arylsulfatase G (ARSG) gene in task-specific dystonia. <i>Journal of Neurology</i> , 2015, 262, 1340-1343.	1.8	14
171	An integrative approach to predicting the functional effects of non-coding and coding sequence variation. <i>Bioinformatics</i> , 2015, 31, 1536-1543.	1.8	524
172	Types and effects of protein variations. <i>Human Genetics</i> , 2015, 134, 405-421.	1.8	29
173	Whole-Exome Sequencing in the Clinic: Lessons from Six Consecutive Cases from the Clinician's Perspective. <i>Molecular Syndromology</i> , 2015, 6, 23-31.	0.3	33
174	PaPI: pseudo amino acid composition to score human protein-coding variants. <i>BMC Bioinformatics</i> , 2015, 16, 123.	1.2	44
175	Arrhythmogenic Cardiomyopathy in a Patient With a Rare Loss-of-Function <i>KCNQ1</i> Mutation. <i>Journal of the American Heart Association</i> , 2015, 4, e001526.	1.6	23
176	Personalized Biochemistry and Biophysics. <i>Biochemistry</i> , 2015, 54, 2551-2559.	1.2	31
177	On Human Disease-Causing Amino Acid Variants: Statistical Study of Sequence and Structural Patterns. <i>Human Mutation</i> , 2015, 36, 524-534.	1.1	122
179	Mining the coding and non-coding genome for cancer drivers. <i>Cancer Letters</i> , 2015, 369, 307-315.	3.2	15
180	Building the foundation for genomics in precision medicine. <i>Nature</i> , 2015, 526, 336-342.	13.7	376
181	Structural modeling and in silico analysis of non-synonymous single nucleotide polymorphisms of human 3 β -hydroxysteroid dehydrogenase type 2. <i>Meta Gene</i> , 2015, 5, 162-172.	0.3	37
182	Computational approaches to study the effects of small genomic variations. <i>Journal of Molecular Modeling</i> , 2015, 21, 251.	0.8	21
183	New approaches to establish genetic causality. <i>Trends in Cardiovascular Medicine</i> , 2015, 25, 646-652.	2.3	11
184	Systems and Synthetic Biology. , 2015, , .		7
185	VariSNP, A Benchmark Database for Variations From dbSNP. <i>Human Mutation</i> , 2015, 36, 161-166.	1.1	50
186	Bioinformatics for Clinical Next Generation Sequencing. <i>Clinical Chemistry</i> , 2015, 61, 124-135.	1.5	114

#	ARTICLE	IF	CITATIONS
187	Amyotrophic lateral sclerosis onset is influenced by the burden of rare variants in known amyotrophic lateral sclerosis genes. <i>Annals of Neurology</i> , 2015, 77, 100-113.	2.8	171
188	Atlas of the clinical genetics of human dilated cardiomyopathy. <i>European Heart Journal</i> , 2015, 36, 1123-1135.	1.0	456
189	fabp4 is central to eight obesity associated genes: A functional gene network-based polymorphic study. <i>Journal of Theoretical Biology</i> , 2015, 364, 344-354.	0.8	33
190	TP53 mutations and protein immunopositivity may predict for poor outcome but also for trastuzumab benefit in patients with early breast cancer treated in the adjuvant setting. <i>Oncotarget</i> , 2016, 7, 32731-32753.	0.8	30
191	Diagnostic Genomics and Clinical Bioinformatics. , 2016, , 37-50.		2
192	SAAFEC: Predicting the Effect of Single Point Mutations on Protein Folding Free Energy Using a Knowledge-Modified MM/PBSA Approach. <i>International Journal of Molecular Sciences</i> , 2016, 17, 512.	1.8	72
193	Tumor Infiltrating Lymphocytes Affect the Outcome of Patients with Operable Triple-Negative Breast Cancer in Combination with Mutated Amino Acid Classes. <i>PLoS ONE</i> , 2016, 11, e0163138.	1.1	8
194	Clinical exome sequencing in neurogenetic and neuropsychiatric disorders. <i>Annals of the New York Academy of Sciences</i> , 2016, 1366, 49-60.	1.8	23
195	Genetics of movement disorders in the next generation sequencing era. <i>Movement Disorders</i> , 2016, 31, 458-470.	2.2	34
196	Variation Interpretation Predictors: Principles, Types, Performance, and Choice. <i>Human Mutation</i> , 2016, 37, 579-597.	1.1	109
197	Assessment of pathogenicity of natural IGFALS gene variants by in silico bioinformatics tools and in vitro functional studies. <i>Molecular and Cellular Endocrinology</i> , 2016, 429, 19-28.	1.6	11
198	Predicting pathogenic single nucleotide variants through a comprehensive analysis on multiple level features. <i>Chemometrics and Intelligent Laboratory Systems</i> , 2016, 156, 224-230.	1.8	0
199	An extended set of yeast-based functional assays accurately identifies human disease mutations. <i>Genome Research</i> , 2016, 26, 670-680.	2.4	116
200	Comparison of pathogenicity prediction tools on missense variants in RYR1 and CACNA1S associated with malignant hyperthermia. <i>British Journal of Anaesthesia</i> , 2016, 117, 124-128.	1.5	34
201	A Broad Overview of Computational Methods for Predicting the Pathophysiological Effects of Non-synonymous Variants. <i>Methods in Molecular Biology</i> , 2016, 1415, 423-440.	0.4	7
202	Machine learning approaches for discrimination of Extracellular Matrix proteins using hybrid feature space. <i>Journal of Theoretical Biology</i> , 2016, 403, 30-37.	0.8	40
203	Computational assessment of feature combinations for pathogenic variant prediction. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 431-446.	0.6	13
204	IMHOTEP—a composite score integrating popular tools for predicting the functional consequences of non-synonymous sequence variants. <i>Nucleic Acids Research</i> , 2017, 45, gkw886.	6.5	10

#	ARTICLE	IF	CITATIONS
205	A dominant negative mutation at the ATP binding domain of <i>AMHR2</i> is associated with a defective anti-Müllerian hormone signaling pathway. <i>Molecular Human Reproduction</i> , 2016, 22, 669-678.	1.3	28
206	Protein function in precision medicine: deep understanding with machine learning. <i>FEBS Letters</i> , 2016, 590, 2327-2341.	1.3	43
207	Rapid Proteasomal Degradation of Mutant Proteins Is the Primary Mechanism Leading to Tumorigenesis in Patients With Missense <i>AIP</i> Mutations. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 3144-3154.	1.8	47
208	Overview of Laboratory Testing and Clinical Presentations of Complement Deficiencies and Dysregulation. <i>Advances in Clinical Chemistry</i> , 2016, 77, 1-75.	1.8	36
209	Large scale analysis of protein stability in OMIM disease related human protein variants. <i>BMC Genomics</i> , 2016, 17, 397.	1.2	37
210	Tools for Predicting the Functional Impact of Nonsynonymous Genetic Variation. <i>Genetics</i> , 2016, 203, 635-647.	1.2	84
211	A knowledge-based approach for predicting gene-disease associations. <i>Bioinformatics</i> , 2016, 32, 2831-2838.	1.8	56
212	Combined sequence and sequence-structure based methods for analyzing <i>FGF23</i> , <i>CYP24A1</i> and <i>VDR</i> genes. <i>Meta Gene</i> , 2016, 9, 26-36.	0.3	7
213	Diagnosis of complement alternative pathway disorders. <i>Kidney International</i> , 2016, 89, 278-288.	2.6	74
214	Clinical exome sequencing in neurologic disease. <i>Neurology: Clinical Practice</i> , 2016, 6, 164-176.	0.8	56
215	Assessing the Pathogenicity of Insertion and Deletion Variants with the Variant Effect Scoring Tool (VEST-Indel). <i>Human Mutation</i> , 2016, 37, 28-35.	1.1	101
216	Integrating population variation and protein structural analysis to improve clinical interpretation of missense variation: application to the WD40 domain. <i>Human Molecular Genetics</i> , 2016, 25, 927-935.	1.4	26
217	BALL-SNP from genetic variants toward computational diagnostics. <i>Bioinformatics</i> , 2016, 32, 1888-1890.	1.8	0
218	Computational Analysis of Missense Variants of G Protein-Coupled Receptors Involved in the Neuroendocrine Regulation of Reproduction. <i>Neuroendocrinology</i> , 2016, 103, 230-239.	1.2	16
219	A novel mutation in <i>GP1BA</i> gene leads to mono-allelic Bernard Soulier syndrome form of macrothrombocytopenia. <i>Blood Coagulation and Fibrinolysis</i> , 2017, 28, 94-95.	0.5	10
220	Blind prediction of deleterious amino acid variations with SNPs&GO. <i>Human Mutation</i> , 2017, 38, 1064-1071.	1.1	24
221	Comprehensive Computational Analysis of GWAS Loci Identifies <i>CCR2</i> as a Candidate Gene for Celiac Disease Pathogenesis. <i>Journal of Cellular Biochemistry</i> , 2017, 118, 2193-2207.	1.2	17
222	Assessment of in silico protein sequence analysis in the clinical classification of variants in cancer risk genes. <i>Journal of Community Genetics</i> , 2017, 8, 87-95.	0.5	20

#	ARTICLE	IF	CITATIONS
223	Crohn disease risk prediction“Best practices and pitfalls with exome data. Human Mutation, 2017, 38, 1193-1200.	1.1	12
224	InterVar: Clinical Interpretation of Genetic Variants by the 2015 ACMG-AMP Guidelines. American Journal of Human Genetics, 2017, 100, 267-280.	2.6	717
225	PON“P and PON“P2 predictor performance in CAGI challenges: Lessons learned. Human Mutation, 2017, 38, 1085-1091.	1.1	5
226	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
227	A comprehensive computational study on pathogenic mis-sense mutations spanning the RING2 and REP domains of Parkin protein. Gene, 2017, 610, 49-58.	1.0	3
228	Are Double Mutations Double Trouble?. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	3
229	Next Generation Sequencing Based Clinical Molecular Diagnosis of Human Genetic Disorders. , 2017, , .		2
230	Large differences in proportions of harmful and benign amino acid substitutions between proteins and diseases. Human Mutation, 2017, 38, 839-848.	1.1	17
231	Common sequence variants affect molecular function more than rare variants?. Scientific Reports, 2017, 7, 1608.	1.6	20
232	Next Generation of Carrier Screening. , 2017, , 339-354.		0
233	Comparison of in silico prediction and experimental assessment of ABCB4 variants identified in patients with biliary diseases. International Journal of Biochemistry and Cell Biology, 2017, 89, 101-109.	1.2	12
234	<i>In silico</i> analysis of non-synonymous single nucleotide polymorphisms in human DAZL gene associated with male infertility. Systems Biology in Reproductive Medicine, 2017, 63, 248-258.	1.0	23
235	In-depth Characterization of the Homodimerization Domain of the Transcription Factor THAP1 and Dystonia-Causing Mutations Therein. Journal of Molecular Neuroscience, 2017, 62, 11-16.	1.1	14
236	Using MutPred derived mtDNA load scores to evaluate mtDNA variation in hypertension and diabetes in a two-population cohort: The SABPA study. Journal of Genetics and Genomics, 2017, 44, 139-149.	1.7	20
237	Standards and Guidelines for the Interpretation and Reporting of Sequence Variants in Cancer. Journal of Molecular Diagnostics, 2017, 19, 4-23.	1.2	1,267
238	Potential and pitfalls in the genetic diagnosis of kidney diseases. CKJ: Clinical Kidney Journal, 2017, 10, 581-585.	1.4	7
239	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
240	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

#	ARTICLE	IF	CITATIONS
241	Genetic diagnosis of polycystic kidney disease, Alport syndrome, and thalassemia minor in a large Chinese family. <i>Clinical Science</i> , 2017, 131, 2427-2438.	1.8	3
242	Comprehensive Analysis of Tissue-wide Gene Expression and Phenotype Data Reveals Tissues Affected in Rare Genetic Disorders. <i>Cell Systems</i> , 2017, 5, 140-148.e2.	2.9	18
243	Prevalence of Rare Genetic Variations and Their Implications in NGS-data Interpretation. <i>Scientific Reports</i> , 2017, 7, 9810.	1.6	16
244	Prediction of harmful variants on mitochondrial genes: Test of habitat-dependent and demographic effects in a euryhaline fish. <i>Ecology and Evolution</i> , 2017, 7, 3826-3835.	0.8	5
245	Application of whole-exome sequencing to direct the specific functional testing and diagnosis of rare inherited bleeding disorders in patients from the Åresund Region, Scandinavia. <i>British Journal of Haematology</i> , 2017, 179, 308-322.	1.2	49
246	BRCA1/2 missense mutations and the value of in-silico analyses. <i>European Journal of Medical Genetics</i> , 2017, 60, 572-577.	0.7	7
247	GTB – an online genome tolerance browser. <i>BMC Bioinformatics</i> , 2017, 18, 20.	1.2	3
248	Genetic diagnosis in hemophilia and von Willebrand disease. <i>Blood Reviews</i> , 2017, 31, 47-56.	2.8	47
249	Computational analysis unravels novel destructive single nucleotide polymorphisms in the non-synonymous region of human caveolin gene. <i>Gene Reports</i> , 2017, 6, 142-157.	0.4	7
250	Investigating the Molecular Mechanisms Behind Uncharacterized Cysteine Losses from Prediction of Their Oxidation State. <i>Human Mutation</i> , 2017, 38, 86-94.	1.1	4
251	How to Define Pathogenicity, Health, and Disease?. <i>Human Mutation</i> , 2017, 38, 129-136.	1.1	21
252	Spatial distribution of disease-associated variants in three-dimensional structures of protein complexes. <i>Oncogenesis</i> , 2017, 6, e380-e380.	2.1	20
253	Development of pathogenicity predictors specific for variants that do not comply with clinical guidelines for the use of computational evidence. <i>BMC Genomics</i> , 2017, 18, 569.	1.2	14
254	Single-Nucleotide Polymorphism of PPAR γ 3, a Protein at the Crossroads of Physiological and Pathological Processes. <i>International Journal of Molecular Sciences</i> , 2017, 18, 361.	1.8	11
255	Disease-associated mitochondrial mutations and the evolution of primate mitogenomes. <i>PLoS ONE</i> , 2017, 12, e0177403.	1.1	7
256	Novel genes and mutations in patients affected by recurrent pregnancy loss. <i>PLoS ONE</i> , 2017, 12, e0186149.	1.1	55
257	Rapid functional analysis of computationally complex rare human IRF6 gene variants using a novel zebrafish model. <i>PLoS Genetics</i> , 2017, 13, e1007009.	1.5	28
258	Evaluation of in silico algorithms for use with ACMG/AMP clinical variant interpretation guidelines. <i>Genome Biology</i> , 2017, 18, 225.	3.8	185

#	ARTICLE	IF	CITATIONS
259	<i>In vivo</i> bioassay to test the pathogenicity of missense human AIP variants. <i>Journal of Medical Genetics</i> , 2018, 55, 522-529.	1.5	15
260	A phenotype centric benchmark of variant prioritisation tools. <i>Npj Genomic Medicine</i> , 2018, 3, 5.	1.7	39
261	Frequent mutations of RetNet genes in eoHM: Further confirmation in 325 probands and comparison with late-onset high myopia based on exome sequencing. <i>Experimental Eye Research</i> , 2018, 171, 76-91.	1.2	36
262	Prediction of factor VIII inhibitor development in the SIPPET cohort by mutational analysis and factor VIII antigen measurement. <i>Journal of Thrombosis and Haemostasis</i> , 2018, 16, 778-790.	1.9	23
263	Vitamin D receptor (VDR) non-synonymous single nucleotide polymorphisms (nsSNPs) affect the calcitriol drug response - A theoretical insight. <i>Journal of Molecular Graphics and Modelling</i> , 2018, 81, 14-24.	1.3	4
264	MERRF Classification: Implications for Diagnosis and Clinical Trials. <i>Pediatric Neurology</i> , 2018, 80, 8-23.	1.0	58
265	Gene-Specific Variant Classifier (DPYD-Classifier) to Identify Deleterious Alleles of Dihydropyrimidine Dehydrogenase. <i>Clinical Pharmacology and Therapeutics</i> , 2018, 104, 709-718.	2.3	43
266	Modeling mutant/wild-type interactions to ascertain pathogenicity of PROKR2 missense variants in patients with isolated GnRH deficiency. <i>Human Molecular Genetics</i> , 2018, 27, 338-350.	1.4	21
267	Prevalence of cholesteryl ester storage disease among hypercholesterolemic subjects and functional characterization of mutations in the lysosomal acid lipase gene. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 169-176.	0.5	15
268	An Integrated Computational Framework to Assess the Mutational Landscape of Î±-L-iduronidase (IDUA) Gene. <i>Journal of Cellular Biochemistry</i> , 2018, 119, 555-565.	1.2	10
269	A Clinical and Molecular Genetic Study of 50 Families with Autosomal Recessive Parkinsonism Revealed Known and Novel Gene Mutations. <i>Molecular Neurobiology</i> , 2018, 55, 3477-3489.	1.9	67
270	Risk of colorectal cancer for carriers of a germ-line mutation in POLE or POLD1. <i>Genetics in Medicine</i> , 2018, 20, 890-895.	1.1	49
271	Systematics for types and effects of DNA variations. <i>BMC Genomics</i> , 2018, 19, 974.	1.2	8
272	Representativeness of variation benchmark datasets. <i>BMC Bioinformatics</i> , 2018, 19, 461.	1.2	18
273	High-frequency actionable pathogenic exome variants in an average-risk cohort. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003178.	0.5	23
274	Identification of 34 novel mutations in propionic acidemia: Functional characterization of missense variants and phenotype associations. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 266-275.	0.5	19
275	Comparative Genomics Approaches Accurately Predict Deleterious Variants in Plants. <i>G3: Genes, Genomes, Genetics</i> , 2018, 8, 3321-3329.	0.8	36
276	Introduction to Genetics of Skeletal and Mineral Metabolic Diseases. , 2018, , 1-21.		2

#	ARTICLE	IF	CITATIONS
277	Performance evaluation of pathogenicity-computation methods for missense variants. <i>Nucleic Acids Research</i> , 2018, 46, 7793-7804.	6.5	168
278	Accurate Classification of NF1 Gene Variants in 84 Italian Patients with Neurofibromatosis Type 1. <i>Genes</i> , 2018, 9, 216.	1.0	22
279	Pharmacogenomics of CYP2C9: Functional and Clinical Considerations. <i>Journal of Personalized Medicine</i> , 2018, 8, 1.	1.1	136
280	Classification of Genetic Variants. , 2018, , 257-280.		0
281	Three-dimensional spatial analysis of missense variants in RTEL1 identifies pathogenic variants in patients with Familial Interstitial Pneumonia. <i>BMC Bioinformatics</i> , 2018, 19, 18.	1.2	18
282	A Hotspot for Disease-Associated Variants of Human PGM1 Is Associated with Impaired Ligand Binding and Loop Dynamics. <i>Structure</i> , 2018, 26, 1337-1345.e3.	1.6	17
283	Î±-Adducin nsSNPs affect mRNA secondary structure, protein modification and stability. <i>Meta Gene</i> , 2018, 17, 153-162.	0.3	7
284	Genetic and genomic testing for neurologic disease in clinical practice. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 147, 11-22.	1.0	20
285	A Bioinformatics Toolkit: In Silico Tools and Online Resources for Investigating Genetic Variation. <i>Seminars in Thrombosis and Hemostasis</i> , 2019, 45, 674-684.	1.5	1
286	Computational analysis of high-risk SNPs in human CHK2 gene responsible for hereditary breast cancer: A functional and structural impact. <i>PLoS ONE</i> , 2019, 14, e0220711.	1.1	14
287	Benchmarking subcellular localization and variant tolerance predictors on membrane proteins. <i>BMC Genomics</i> , 2019, 20, 547.	1.2	14
288	What went wrong with variant effect predictor performance for the PCM1 challenge. <i>Human Mutation</i> , 2019, 40, 1486-1494.	1.1	8
289	Epigenetic signatures in overgrowth syndromes: Translational opportunities. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019, 181, 491-501.	0.7	10
290	Computational analysis of Alzheimer-causing mutations in amyloid precursor protein and presenilin 1. <i>Archives of Biochemistry and Biophysics</i> , 2019, 678, 108168.	1.4	13
291	Fanconi-BRCA pathway mutations in childhood T-cell acute lymphoblastic leukemia. <i>PLoS ONE</i> , 2019, 14, e0221288.	1.1	16
292	Decoding of novel missense TSC2 gene variants using in-silico methods. <i>BMC Medical Genetics</i> , 2019, 20, 164.	2.1	4
293	Insights into pathological mutations in insulin-like growth factor I through in silico screening and molecular dynamics simulation. <i>Journal of Molecular Modeling</i> , 2019, 25, 276.	0.8	1
294	Determining the pathogenicity of CFTR missense variants: Multiple comparisons of in silico predictors and variant annotation databases. <i>Genetics and Molecular Biology</i> , 2019, 42, 560-570.	0.6	6

#	ARTICLE	IF	CITATIONS
295	Innovative strategies for annotating the "relationSNP" between variants and molecular phenotypes. <i>BioData Mining</i> , 2019, 12, 10.	2.2	6
296	Structural and Functional Impact of Seven Missense Variants of Phenylalanine Hydroxylase. <i>Genes</i> , 2019, 10, 459.	1.0	2
297	Challenges and Considerations in Sequence Variant Interpretation for Mendelian Disorders. <i>Annals of Laboratory Medicine</i> , 2019, 39, 421-429.	1.2	31
298	Comparison of Predictive <i>In Silico</i> Tools on Missense Variants in <i>GJB2</i> , <i>GJB6</i> , and <i>GJB3</i> Genes Associated with Autosomal Recessive Deafness 1A (DFNB1A). <i>Scientific World Journal, The</i> , 2019, 2019, 1-9.	0.8	26
299	New insights into the pathogenicity of non-synonymous variants through multi-level analysis. <i>Scientific Reports</i> , 2019, 9, 1667.	1.6	40
300	Hereditary xanthinuria in a goat. <i>Journal of Veterinary Internal Medicine</i> , 2019, 33, 1009-1014.	0.6	3
301	How good are pathogenicity predictors in detecting benign variants?. <i>PLoS Computational Biology</i> , 2019, 15, e1006481.	1.5	79
302	Characterizing variants of unknown significance in rhodopsin: A functional genomics approach. <i>Human Mutation</i> , 2019, 40, 1127-1144.	1.1	22
303	Clinical application of next-generation sequencing to the practice of neurology. <i>Lancet Neurology, The</i> , 2019, 18, 492-503.	4.9	76
304	Structural and Computational Characterization of Disease-Related Mutations Involved in Protein-Protein Interfaces. <i>International Journal of Molecular Sciences</i> , 2019, 20, 1583.	1.8	17
305	The Oculome Panel Test. <i>Ophthalmology</i> , 2019, 126, 888-907.	2.5	77
306	Application of Computational Biology and Artificial Intelligence Technologies in Cancer Precision Drug Discovery. <i>BioMed Research International</i> , 2019, 2019, 1-15.	0.9	42
307	Association of functional variants and protein-to-protein physical interactions of human MutY homolog linked with familial adenomatous polyposis and colorectal cancer syndrome. <i>Non-coding RNA Research</i> , 2019, 4, 155-173.	2.4	1
308	Two Novel Variants in the ATRX Gene Associated with Variable Phenotypes. <i>Case Reports in Genetics</i> , 2019, 2019, 1-5.	0.1	1
309	Predicting disease-associated mutation of metal-binding sites in proteins using a deep learning approach. <i>Nature Machine Intelligence</i> , 2019, 1, 561-567.	8.3	48
310	Mutation analysis and pathogenicity identification of Mucopolysaccharidosis type IVA in 8 south China families. <i>Gene</i> , 2019, 686, 261-269.	1.0	3
311	A review study: Computational techniques for expecting the impact of non-synonymous single nucleotide variants in human diseases. <i>Gene</i> , 2019, 680, 20-33.	1.0	47
312	An optimized prediction framework to assess the functional impact of pharmacogenetic variants. <i>Pharmacogenomics Journal</i> , 2019, 19, 115-126.	0.9	109

#	ARTICLE	IF	CITATIONS
313	The CYSMA web server: An example of integrative tool for in silico analysis of missense variants identified in Mendelian disorders. <i>Human Mutation</i> , 2020, 41, 375-386.	1.1	6
314	Spanish Guidelines for the use of targeted deep sequencing in myelodysplastic syndromes and chronic myelomonocytic leukaemia. <i>British Journal of Haematology</i> , 2020, 188, 605-622.	1.2	25
315	InMeRF: prediction of pathogenicity of missense variants by individual modeling for each amino acid substitution. <i>NAR Genomics and Bioinformatics</i> , 2020, 2, lqaa038.	1.5	16
316	Identification of pathogenic missense mutations using protein stability predictors. <i>Scientific Reports</i> , 2020, 10, 15387.	1.6	66
317	In silico analysis of the functional and structural consequences of SNPs in human ARX gene associated with EIEE1. <i>Informatics in Medicine Unlocked</i> , 2020, 21, 100447.	1.9	7
318	Whole genome sequencing for the investigation of canine mammary tumor inheritance - an initial assessment of high-risk breast cancer genes reveal BRCA2 and STK11 variants potentially associated with risk in purebred dogs. <i>Canine Medicine and Genetics</i> , 2020, 7, .	1.4	11
319	In silico analysis of likely pathogenic variants in human GGCX gene. <i>Informatics in Medicine Unlocked</i> , 2020, 19, 100337.	1.9	2
320	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. <i>Nature Communications</i> , 2020, 11, 5918.	5.8	305
321	MISTIC: A prediction tool to reveal disease-relevant deleterious missense variants. <i>PLoS ONE</i> , 2020, 15, e0236962.	1.1	26
322	Comprehensive characterization of amino acid positions in protein structures reveals molecular effect of missense variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 28201-28211.	3.3	68
323	Consistency of the Tools That Predict the Impact of Single Nucleotide Variants (SNVs) on Gene Functionality: The BRCA1 Gene. <i>Biomolecules</i> , 2020, 10, 475.	1.8	0
324	Assessing performance of pathogenicity predictors using clinically relevant variant datasets. <i>Journal of Medical Genetics</i> , 2021, 58, 547-555.	1.5	57
325	Secuenciación de nueva generación (NGS) de ADN: presente y futuro en la práctica clínica. <i>Revista Universitas Medica</i> , 2020, 61, .	0.0	7
326	A Yeast-Based Model for Hereditary Motor and Sensory Neuropathies: A Simple System for Complex, Heterogeneous Diseases. <i>International Journal of Molecular Sciences</i> , 2020, 21, 4277.	1.8	9
327	Problems in variation interpretation guidelines and in their implementation in computational tools. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1206.	0.6	17
328	Variants of uncertain significance in the era of high-throughput genome sequencing: a lesson from breast and ovary cancers. <i>Journal of Experimental and Clinical Cancer Research</i> , 2020, 39, 46.	3.5	108
329	Survival of the cheapest: how proteome cost minimization drives evolution. <i>Quarterly Reviews of Biophysics</i> , 2020, 53, e7.	2.4	12
330	Mutation m.3395A>G in MT-ND1 leads to variable pathologic manifestations. <i>Human Molecular Genetics</i> , 2020, 29, 980-989.	1.4	5

#	ARTICLE	IF	CITATIONS
331	Rhapsody: predicting the pathogenicity of human missense variants. <i>Bioinformatics</i> , 2020, 36, 3084-3092.	1.8	63
332	Phenotypic and genotypic characterization of families with complex intellectual disability identified pathogenic genetic variations in known and novel disease genes. <i>Scientific Reports</i> , 2020, 10, 968.	1.6	8
333	Variation benchmark datasets: update, criteria, quality and applications. <i>Database: the Journal of Biological Databases and Curation</i> , 2020, 2020, .	1.4	27
334	A multicriteria approach based on rough set theory for the incremental Periodic prediction. <i>European Journal of Operational Research</i> , 2020, 286, 282-298.	3.5	19
335	Functional effects of protein variants. <i>Biochimie</i> , 2021, 180, 104-120.	1.3	30
336	Mapping bull help canopy in northern California using Landsat to enable long-term monitoring. <i>Remote Sensing of Environment</i> , 2021, 254, 112243.	4.6	28
337	Alignment-free method for functional annotation of amino acid substitutions: Application on epigenetic factors involved in hematologic malignancies. <i>PLoS ONE</i> , 2021, 16, e0244948.	1.1	0
338	Performance of mutation pathogenicity prediction tools on missense variants associated with 46,XY differences of sex development. <i>Clinics</i> , 2021, 76, e2052.	0.6	10
339	Structure-Based Approaches to Classify the Functional Impact of ZBTB18 Missense Variants in Health and Disease. <i>ACS Chemical Neuroscience</i> , 2021, 12, 979-989.	1.7	4
340	Challenges in Clinicogenetic Correlations: One Phenotype “ Many Genes. <i>Movement Disorders Clinical Practice</i> , 2021, 8, 311-321.	0.8	12
342	Type IV Collagen Variants in CKD: Performance of Computational Predictions for Identifying Pathogenic Variants. <i>Kidney Medicine</i> , 2021, 3, 257-266.	1.0	9
343	The Interpretation of Sequence Variants in Myeloid Neoplasms. <i>American Journal of Clinical Pathology</i> , 2021, 156, 728-748.	0.4	1
344	Critical assessment of coiled-coil predictions based on protein structure data. <i>Scientific Reports</i> , 2021, 11, 12439.	1.6	14
346	Evolutionary and functional lessons from human-specific amino acid substitution matrices. <i>NAR Genomics and Bioinformatics</i> , 2021, 3, lqab079.	1.5	1
347	Characterization of a novel ABCC2 mutation in infantile Dubin Johnson syndrome. <i>Clinica Chimica Acta</i> , 2021, 518, 43-50.	0.5	4
348	Genomics pipelines to investigate susceptibility in whole genome and exome sequenced data for variant discovery, annotation, prediction and genotyping. <i>PeerJ</i> , 2021, 9, e11724.	0.9	12
349	Pathogenic nsSNPs that increase the risks of cancers among the Orang Asli and Malays. <i>Scientific Reports</i> , 2021, 11, 16158.	1.6	6
350	Mutation of c.244G>T in NR5A1 gene causing 46, XY DSD by affecting RNA splicing. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 370.	1.2	1

#	ARTICLE	IF	CITATIONS
352	Adapting the ACMG/AMP variant classification framework: A perspective from the ClinGen Hemoglobinopathy Variant Curation Expert Panel. <i>Human Mutation</i> , 2022, 43, 1089-1096.	1.1	20
353	An Empirical Pipeline for Personalized Diagnosis of Lafora Disease Mutations. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0
354	Approaches for Classifying DNA Variants Found by Sanger Sequencing in a Medical Genetics Laboratory. <i>Methods in Molecular Biology</i> , 2014, 1168, 227-250.	0.4	3
355	Computational Tools for Designing Smart Libraries. <i>Methods in Molecular Biology</i> , 2014, 1179, 291-314.	0.4	21
356	Functional Annotation of Rare Genetic Variants. , 2015, , 57-70.		2
357	Selection and Application of Machine Learning- Algorithms in Production Quality. <i>Technologien Fu'r Die Intelligente Automation</i> , 2019, , 46-57.	0.3	12
370	Towards Increasing the Clinical Relevance of In Silico Methods to Predict Pathogenic Missense Variants. <i>PLoS Computational Biology</i> , 2016, 12, e1004725.	1.5	34
371	PredictSNP2: A Unified Platform for Accurately Evaluating SNP Effects by Exploiting the Different Characteristics of Variants in Distinct Genomic Regions. <i>PLoS Computational Biology</i> , 2016, 12, e1004962.	1.5	149
372	High-confidence assessment of functional impact of human mitochondrial non-synonymous genome variations by APOGEE. <i>PLoS Computational Biology</i> , 2017, 13, e1005628.	1.5	54
373	High-Throughput Sequencing of mGluR Signaling Pathway Genes Reveals Enrichment of Rare Variants in Autism. <i>PLoS ONE</i> , 2012, 7, e35003.	1.1	96
374	Evaluating Purifying Selection in the Mitochondrial DNA of Various Mammalian Species. <i>PLoS ONE</i> , 2013, 8, e58993.	1.1	39
375	Gene-Specific Function Prediction for Non-Synonymous Mutations in Monogenic Diabetes Genes. <i>PLoS ONE</i> , 2014, 9, e104452.	1.1	23
376	PON-P2: Prediction Method for Fast and Reliable Identification of Harmful Variants. <i>PLoS ONE</i> , 2015, 10, e0117380.	1.1	180
377	Novel Carboxypeptidase A6 (CPA6) Mutations Identified in Patients with Juvenile Myoclonic and Generalized Epilepsy. <i>PLoS ONE</i> , 2015, 10, e0123180.	1.1	17
378	ENTPRISE: An Algorithm for Predicting Human Disease-Associated Amino Acid Substitutions from Sequence Entropy and Predicted Protein Structures. <i>PLoS ONE</i> , 2016, 11, e0150965.	1.1	23
379	Generalising better: Applying deep learning to integrate deleteriousness prediction scores for whole-exome SNV studies. <i>PLoS ONE</i> , 2018, 13, e0192829.	1.1	14
380	Novel putative drivers revealed by targeted exome sequencing of advanced solid tumors. <i>PLoS ONE</i> , 2018, 13, e0194790.	1.1	3
381	Systematic alanine scanning of PAX8 paired domain reveals functional importance of the N-subdomain. <i>Journal of Molecular Endocrinology</i> , 2019, 62, 129-135.	1.1	11

#	ARTICLE	IF	CITATIONS
382	Recent Advances in Predicting Functional Impact of Single Amino Acid Polymorphisms: A Review of Useful Features, Computational Methods and Available Tools. <i>Current Bioinformatics</i> , 2013, 8, 161-176.	0.7	7
383	An improved understanding of cancer genomics through massively parallel sequencing. <i>Translational Cancer Research</i> , 2014, 3, 243-259.	0.4	10
384	Structured Genome-Scale Variant and Clinical Data Reporting for Meta-Analysis in an Era of Genomic Medicine. <i>Journal of Genomes and Exomes</i> , 0, 2, 31-42.	0.0	2
385	Deep Sequencing Data Analysis: Challenges and Solutions. , 0, , .		4
386	The aetiology of cardiovascular disease: a role for mitochondrial DNA?. <i>Cardiovascular Journal of Africa</i> , 2018, 29, 122-132.	0.2	14
387	Phenotype inference in an Escherichia coli strain panel. <i>ELife</i> , 2017, 6, .	2.8	38
388	An empirical pipeline for personalized diagnosis of Lafora disease mutations. <i>IScience</i> , 2021, 24, 103276.	1.9	7
389	MICO: A meta-tool for prediction of the effects of non-synonymous mutations. <i>Bioinformatics</i> , 2014, 10, 469-471.	0.2	0
391	SVI: A Simple Single-Nucleotide Human Variant Interpretation Tool for Clinical Use. <i>Lecture Notes in Computer Science</i> , 2015, , 180-194.	1.0	5
392	Introduction to Molecular Genetics. , 2016, , 3-24.		0
398	Testing the Mutation Accumulation Theory of Aging Using Bioinformatic Tools. <i>Advances in Aging Research</i> , 2018, 07, 17-28.	0.3	1
402	In Silico Analysis of the Novel Variant Q375R in the Phenylalanine Hydroxylase Gene. <i>Gene, Cell and Tissue</i> , 2019, In Press, .	0.2	0
403	TMCO1 Gen Sekans Varyanlatların Fonksiyonel Özelliklerinin In Silico Analizlerle Değerlendirilmesi. <i>Dicle Üniversitesi Bilim Ve Teknoloji Dergisi</i> , 2019, 7, 1931-1946.	0.2	0
405	A Preliminary Comparison of P-Tool Consistency. <i>IFMBE Proceedings</i> , 2020, , 731-735.	0.2	0
406	PILOT RESEARCH OF A GENETIC PREDISPOSITION FOR CLINICAL MANIFESTATIONS OF ACUTE INTERMITTENT PORPHYRIA. <i>Gematologiya I Transfuziologiya</i> , 2019, 64, 123-137.	0.1	1
407	miR-618 rs2682818 C>A polymorphism decreases Hirschsprung disease risk in Chinese children. <i>Bioscience Reports</i> , 2020, 40, .	1.1	5
410	The impact of post-alignment processing procedures on whole-exome sequencing data. <i>Genetics and Molecular Biology</i> , 2020, 43, e20200047.	0.6	0
411	Introduction to Molecular Genetics. , 2020, , 3-26.		0

#	ARTICLE	IF	CITATIONS
412	VPMBench: a test bench for variant prioritization methods. BMC Bioinformatics, 2021, 22, 543.	1.2	0
414	A study of the pathogenicity of variants in familial heart disease. The value of cosegregation. American Journal of Translational Research (discontinued), 2019, 11, 1724-1735.	0.0	2
415	MLb-LDLr. JACC Basic To Translational Science, 2021, 6, 815-827.	1.9	10
416	Comprehensive Characterization of the Coding and Non-Coding Single Nucleotide Polymorphisms in the Tumor Protein p63 (TP63) Gene Using In Silico Tools. Biomolecules, 2021, 11, 1733.	1.8	2
417	Rare and potentially pathogenic variants in hydroxycarboxylic acid receptor genes identified in breast cancer cases. BMC Medical Genomics, 2021, 14, 284.	0.7	2
418	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
419	Automated prediction of the clinical impact of structural copy number variations. Scientific Reports, 2022, 12, 555.	1.6	3
420	PPSNV: A Novel Predictor for Pathogenicity of Nonsynonymous SNV based on Ensemble Learning. , 2021, , .		0
422	Theranostic Interpolation of Genomic Instability in Breast Cancer. International Journal of Molecular Sciences, 2022, 23, 1861.	1.8	8
423	Current and Future Approaches to Classify VUSs in LGMD-Related Genes. Genes, 2022, 13, 382.	1.0	3
424	MutateX: an automated pipeline for <i>in silico</i> saturation mutagenesis of protein structures and structural ensembles. Briefings in Bioinformatics, 2022, 23, .	3.2	26
425	Assessment of 13 <i>in silico</i> pathogenicity methods on cancer-related variants. Computers in Biology and Medicine, 2022, 145, 105434.	3.9	2
426	Interpreting ciliopathy-associated missense variants of uncertain significance (VUS) in <i>Caenorhabditis elegans</i> . Human Molecular Genetics, 2022, 31, 1574-1587.	1.4	9
427	Computational refinement identifies functional destructive single nucleotide polymorphisms associated with human retinoid X receptor gene. Journal of Biomolecular Structure and Dynamics, 2023, 41, 1458-1478.	2.0	5
428	Variants in mitochondrial amidoxime reducing component 1 and hydroxysteroid 17 β dehydrogenase 13 reduce severity of nonalcoholic fatty liver disease in children and suppress fibrotic pathways through distinct mechanisms. Hepatology Communications, 2022, 6, 1934-1948.	2.0	18
429	Computational approaches toward single-nucleotide polymorphism discovery and its applications in plant breeding. , 2022, , 513-536.		0
430	Structural bioinformatics enhances the interpretation of somatic mutations in KDM6A found in human cancers. Computational and Structural Biotechnology Journal, 2022, 20, 2200-2211.	1.9	5
431	Scrutinizing Deleterious Nonsynonymous SNPs and Their Effect on Human POLD1 Gene. Genetical Research, 2022, 2022, 1-12.	0.3	2

#	ARTICLE	IF	CITATIONS
432	Interpreting protein variant effects with computational predictors and deep mutational scanning. <i>DMM Disease Models and Mechanisms</i> , 2022, 15, .	1.2	25
433	Personalized structural biology reveals the molecular mechanisms underlying heterogeneous epileptic phenotypes caused by de novo KCNC2 variants. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100131.	1.0	1
434	Benign SNPs in the Coding Region of <i>TP53</i> : Finding the Needles in a Haystack of Pathogenic Variants. <i>Cancer Research</i> , 2022, 82, 3420-3431.	0.4	6
435	A Comprehensive Evaluation of the Performance of Prediction Algorithms on Clinically Relevant Missense Variants. <i>International Journal of Molecular Sciences</i> , 2022, 23, 7946.	1.8	5
436	Computer-Aided (In Silico) Modeling of Cytochrome P450-Mediated Food-Drug Interactions (FDI). <i>International Journal of Molecular Sciences</i> , 2022, 23, 8498.	1.8	8
437	Clinical significance of genetic variation in hypertrophic cardiomyopathy: comparison of computational tools to prioritize missense variants. <i>Frontiers in Cardiovascular Medicine</i> , 0, 9, .	1.1	5
438	d-StructMAN: Containerized structural annotation on the scale from genetic variants to whole proteomes. <i>GigaScience</i> , 2022, 11, .	3.3	0
440	Addressing Noise and Estimating Uncertainty in Biomedical Data through the Exploration of Chemical Space. <i>International Journal of Molecular Sciences</i> , 2022, 23, 12975.	1.8	0
441	Elucidating the mutational impact in causing Niemann-Pick disease type C: an in silico approach. <i>Journal of Biomolecular Structure and Dynamics</i> , 2023, 41, 8561-8570.	2.0	2
443	Identification of prolactin receptor variants with diverse effects on receptor signalling. <i>Journal of Molecular Endocrinology</i> , 2023, 70, .	1.1	2
444	aRgus: Multilevel visualization of non-synonymous single nucleotide variants & advanced pathogenicity score modeling for genetic vulnerability assessment. <i>Computational and Structural Biotechnology Journal</i> , 2023, 21, 1077-1083.	1.9	7
445	In Silico-Based Structural Evaluation to Categorize the Pathogenicity of Mutations Identified in the RAD Class of Proteins. <i>ACS Omega</i> , 2023, 8, 10266-10277.	1.6	0
446	High risk genetic variants of human insulin receptor substrate 1 (IRS1) infer structural instability and functional interference. <i>Journal of Biomolecular Structure and Dynamics</i> , 2023, 41, 15150-15164.	2.0	0
447	Wilson's Disease—Genetic Puzzles with Diagnostic Implications. <i>Diagnostics</i> , 2023, 13, 1287.	1.3	3
448	Structural and Pathogenic Impacts of ABCA4 Variants in Retinal Degenerations—An In-Silico Study. <i>International Journal of Molecular Sciences</i> , 2023, 24, 7280.	1.8	1
452	Prediction of Functional Effects of Protein Amino Acid Mutations. <i>Lecture Notes in Computer Science</i> , 2023, , 59-71.	1.0	0
465	Genetic and Genomic Results and Management. , 2024, , 93-110.		0