

PredictSNP: Robust and Accurate Consensus Classifier for Mutations

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Sex Hormone Binding Globulin Deficiency Due to a Homozygous Missense Mutation. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014, 99, E1798-E1802.	1.8	34
2	The road from next-generation sequencing to personalized medicine. <i>Personalized Medicine</i> , 2014, 11, 523-544.	0.8	40
3	Majority Vote and Other Problems when using Computational Tools. <i>Human Mutation</i> , 2014, 35, 912-914.	1.1	15
4	Use of Contemporary Genetics in Cardiovascular Diagnosis. <i>Circulation</i> , 2014, 130, 1971-1980.	1.6	7
5	Compound heterozygous PNPLA6 mutations cause Boucher's "Neuhäuser syndrome with late-onset ataxia. <i>Journal of Neurology</i> , 2014, 261, 2411-2423.	1.8	30
6	Comparative genomics of closely related <i>Salmonella enterica</i> serovar Typhi strains reveals genome dynamics and the acquisition of novel pathogenic elements. <i>BMC Genomics</i> , 2014, 15, 1007.	1.2	18
7	ALK Mutations Confer Differential Oncogenic Activation and Sensitivity to ALK Inhibition Therapy in Neuroblastoma. <i>Cancer Cell</i> , 2014, 26, 682-694.	7.7	302
8	Feature-based multiple models improve classification of mutation-induced stability changes. <i>BMC Genomics</i> , 2014, 15, S6.	1.2	14
9	Beyond Ohdo syndrome: A familial missense mutation broadens the <i>MED12</i> spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3180-3185.	0.7	22
10	Better prediction of functional effects for sequence variants. <i>BMC Genomics</i> , 2015, 16, S1.	1.2	478
11	<i>In Silico</i> Prediction of the Effects of Mutations in the Human Mevalonate Kinase Gene: Towards a Predictive Framework for Mevalonate Kinase Deficiency. <i>Annals of Human Genetics</i> , 2015, 79, 451-459.	0.3	21
12	Retargeting of bile salt export pump and favorable outcome in children with progressive familial intrahepatic cholestasis type 2. <i>Hepatology</i> , 2015, 62, 198-206.	3.6	25
13	Bioinformatics Methods and Tools to Advance Clinical Care. <i>Yearbook of Medical Informatics</i> , 2015, 24, 170-173.	0.8	4
14	Diversity and impact of rare variants in genes encoding the platelet G protein-coupled receptors. <i>Thrombosis and Haemostasis</i> , 2015, 113, 826-837.	1.8	15
15	Systematic Mapping of Protein Mutational Space by Prolonged Drift Reveals the Deleterious Effects of Seemingly Neutral Mutations. <i>PLoS Computational Biology</i> , 2015, 11, e1004421.	1.5	79
16	Identification of novel drought-tolerant-associated SNPs in common bean (<i>Phaseolus vulgaris</i>). <i>Frontiers in Plant Science</i> , 2015, 6, 546.	1.7	51
17	Identification of Deleterious SNPs and Their Effects on Structural Level in <i>CHRNA3</i> Gene. <i>Biochemical Genetics</i> , 2015, 53, 159-168.	0.8	9
18	A novel mutation in <i>TFL1</i> homolog affecting determinacy in cowpea (<i>Vigna unguiculata</i>). <i>Molecular Genetics and Genomics</i> , 2015, 290, 55-65.	1.0	33

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19	Mechismo: predicting the mechanistic impact of mutations and modifications on molecular interactions. <i>Nucleic Acids Research</i> , 2015, 43, e10-e10.	6.5	95
20	Pathogenicity prediction of non-synonymous single nucleotide variants in dilated cardiomyopathy. <i>Briefings in Bioinformatics</i> , 2015, 16, 769-779.	3.2	10
21	Prediction of the pathogenicity of antithrombin sequence variations by in silico methods. <i>Thrombosis Research</i> , 2015, 135, 404-409.	0.8	16
22	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
23	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
24	Functional and in silico assessment of MAX variants of unknown significance. <i>Journal of Molecular Medicine</i> , 2015, 93, 1247-1255.	1.7	25
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26	Computational analyses and prediction of guanylin deleterious SNPs. <i>Peptides</i> , 2015, 69, 92-102.	1.2	31
27	Assessment of the predictive accuracy of five in silico prediction tools, alone or in combination, and two metaservers to classify long QT syndrome gene mutations. <i>BMC Medical Genetics</i> , 2015, 16, 34.	2.1	73
28	Complementation of Yeast Genes with Human Genes as an Experimental Platform for Functional Testing of Human Genetic Variants. <i>Genetics</i> , 2015, 201, 1263-1274.	1.2	77
29	Computational approaches to study the effects of small genomic variations. <i>Journal of Molecular Modeling</i> , 2015, 21, 251.	0.8	21
30	New approaches to establish genetic causality. <i>Trends in Cardiovascular Medicine</i> , 2015, 25, 646-652.	2.3	11
31	Case-only exome sequencing and complex disease susceptibility gene discovery: study design considerations. <i>Journal of Medical Genetics</i> , 2015, 52, 10-16.	1.5	23
32	Living laboratory: whole-genome sequencing as a learning healthcare enterprise. <i>Clinical Genetics</i> , 2015, 87, 311-318.	1.0	20
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34	Prediction of the impact of coding missense and nonsense single nucleotide polymorphisms on HD5 and HBD1 antibacterial activity against <i>Escherichia coli</i> . <i>Biopolymers</i> , 2016, 106, 633-644.	1.2	28
35	Variation Interpretation Predictors: Principles, Types, Performance, and Choice. <i>Human Mutation</i> , 2016, 37, 579-597.	1.1	109
36	New Mutations Associated with Rasopathies in a Central European Population and Genotype-Phenotype Correlations. <i>Annals of Human Genetics</i> , 2016, 80, 50-62.	0.3	23

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38	ATM mutations in major stereotyped subsets of chronic lymphocytic leukemia: enrichment in subset #2 is associated with markedly short telomeres. <i>Haematologica</i> , 2016, 101, e369-e373.	1.7	16
39	UMDâ€Predictor: A Highâ€Throughput Sequencing Compliant System for Pathogenicity Prediction of any Human cDNA Substitution. <i>Human Mutation</i> , 2016, 37, 439-446.	1.1	104
40	Computational approaches for predicting mutant protein stability. <i>Journal of Computer-Aided Molecular Design</i> , 2016, 30, 401-412.	1.3	60
41	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
42	Comprehensive mutational profiling of core binding factor acute myeloid leukemia. <i>Blood</i> , 2016, 127, 2451-2459.	0.6	198
43	Identification of <i>Brucella melitensis</i> Rev.1 vaccine-strain genetic markers: Towards understanding the molecular mechanism behind virulence attenuation. <i>Vaccine</i> , 2016, 34, 4884-4891.	1.7	8
44	Dextromethorphan and debrisoquine metabolism and polymorphism of the gene for cytochrome P450 isozyme 2D50 in Thoroughbreds. <i>American Journal of Veterinary Research</i> , 2016, 77, 1029-1035.	0.3	6
45	Investigating regulatory signatures of human autophagy related gene 5 (ATG5) through functional in silico analysis. <i>Meta Gene</i> , 2016, 9, 237-248.	0.3	16
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48	Computational assessment of feature combinations for pathogenic variant prediction. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 431-446.	0.6	13
49	Analysis of a cohort of 101 <sc>CDAll</sc> patients: description of 24 new molecular variants and genotypeâ€phenotype correlations. <i>British Journal of Haematology</i> , 2016, 175, 696-704.	1.2	25
50	Acute Intermittent Porphyria: Predicted Pathogenicity of <i>HMBS</i> Variants Indicates Extremely Low Penetrance of the Autosomal Dominant Disease. <i>Human Mutation</i> , 2016, 37, 1215-1222.	1.1	129
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56	The Complementarity Between Protein-Specific and General Pathogenicity Predictors for Amino Acid Substitutions. <i>Human Mutation</i> , 2016, 37, 1013-1024.	1.1	42
57	Structural insights and functional implications of inter-individual variability in β 2-adrenergic receptor. <i>Scientific Reports</i> , 2016, 6, 24379.	1.6	15
58	Hybridization-Based Enrichment and Next Generation Sequencing to Explore Genetic Diversity in Plants. , 2016, , 117-136.		2
59	KinMutRF: a random forest classifier of sequence variants in the human protein kinase superfamily. <i>BMC Genomics</i> , 2016, 17, 396.	1.2	11
60	Genetic Association Analysis Reveals Differences in the Contribution of NOD2 Variants to the Clinical Phenotypes of Orofacial Granulomatosis. <i>Inflammatory Bowel Diseases</i> , 2016, 22, 1552-1558.	0.9	13
61	Tools for Predicting the Functional Impact of Nonsynonymous Genetic Variation. <i>Genetics</i> , 2016, 203, 635-647.	1.2	84
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65	Mitonuclear Epistasis for Development Time and Its Modification by Diet in <i>Drosophila</i> . <i>Genetics</i> , 2016, 203, 463-484.	1.2	86
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67	Evidence for polymorphism in the cytochrome P450 2D50 gene in horses. <i>Journal of Veterinary Pharmacology and Therapeutics</i> , 2016, 39, 245-254.	0.6	18
68	Unlike <i>ASXL1</i> and <i>ASXL2</i> mutations, <i>ASXL3</i> mutations are rare events in acute myeloid leukemia with t(8;21). <i>Leukemia and Lymphoma</i> , 2016, 57, 199-200.	0.6	19
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70	PON α P and PON α P2 predictor performance in CAGI challenges: Lessons learned. <i>Human Mutation</i> , 2017, 38, 1085-1091.	1.1	5
71	Elevation of glycosaminoglycans in the amniotic fluid of a fetus with mucopolysaccharidosis VII. <i>Prenatal Diagnosis</i> , 2017, 37, 435-439.	1.1	20
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77	Genotype-phenotype correlation in patients with isovaleric acidaemia: comparative structural modelling and computational analysis of novel variants. <i>Human Molecular Genetics</i> , 2017, 26, 3105-3115.	1.4	32
78	Computational Investigation of Growth Hormone Receptor Trp169Arg Heterozygous Mutation in a Child With Short Stature. <i>Journal of Cellular Biochemistry</i> , 2017, 118, 4762-4771.	1.2	8
79	Smart health: Big data enabled health paradigm within smart cities. <i>Expert Systems With Applications</i> , 2017, 87, 370-383.	4.4	249
80	Computational investigation of the human SOD1 mutant, Cys146Arg, that directs familial amyotrophic lateral sclerosis. <i>Molecular BioSystems</i> , 2017, 13, 1495-1503.	2.9	14
81	A theoretical study on Zn binding loop mutants instigating destabilization and metal binding loss in human SOD1 protein. <i>Journal of Molecular Modeling</i> , 2017, 23, 103.	0.8	5
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87	Hemorheological alterations in sickle cell anemia and their clinical consequences - The role of genetic modulators. <i>Clinical Hemorheology and Microcirculation</i> , 2017, 64, 859-866.	0.9	5
88	The novel homozygous KCNJ10 c.986T>C (p.(Leu329Pro)) variant is pathogenic for the SeSAME/EAST homologue in Malinois dogs. <i>European Journal of Human Genetics</i> , 2017, 25, 222-226.	1.4	16
89	A nonsynonymous mutation in the WFS1 gene in a Finnish family with age-related hearing impairment. <i>Hearing Research</i> , 2017, 355, 97-101.	0.9	9
90	Genetic risk factors in Finnish patients with Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2017, 45, 39-43.	1.1	19

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91	Characterizing clinically relevant natural variants of GPCRs using computational approaches. <i>Methods in Cell Biology</i> , 2017, 142, 187-204.	0.5	7
92	Targeted Exome Sequencing of Krebs Cycle Genes Reveals Candidate Cancer-Associated Predisposing Mutations in Pheochromocytomas and Paragangliomas. <i>Clinical Cancer Research</i> , 2017, 23, 6315-6324.	3.2	73
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98	TRPM4 non-selective cation channel variants in long QT syndrome. <i>BMC Medical Genetics</i> , 2017, 18, 31.	2.1	26
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105	Impact of genetic variation on three dimensional structure and function of proteins. <i>PLoS ONE</i> , 2017, 12, e0171355.	1.1	55
106	Molecular analysis of TSC1 and TSC2 genes and phenotypic correlations in Brazilian families with tuberous sclerosis. <i>PLoS ONE</i> , 2017, 12, e0185713.	1.1	24
107	Molecular dynamic simulations reveal suboptimal binding of salbutamol in T164I variant of β 2 adrenergic receptor. <i>PLoS ONE</i> , 2017, 12, e0186666.	1.1	34
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109	Identification and in silico characterization of p.G380R substitution in FGFR3, associated with achondroplasia in a non-consanguineous Pakistani family. <i>Diagnostic Pathology</i> , 2017, 12, 47.	0.9	7
110	Effects of Type 1 Diabetes Risk Alleles on Immune Cell Gene Expression. <i>Genes</i> , 2017, 8, 167.	1.0	17
111	Structural dynamics is a determinant of the functional significance of missense variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 4164-4169.	3.3	76
112	Genetics of Iranian Alpha-Thalassemia Patients: A Comprehensive Original Study. <i>Biochemical Genetics</i> , 2018, 56, 506-521.	0.8	11
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118	Spectrum of GALNS mutations and haplotype study in Brazilian patients with Mucopolysaccharidosis type IVA. <i>Meta Gene</i> , 2018, 16, 77-84.	0.3	6
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121	Impact of a CD4 gene haplotype on the immune response in minipigs. <i>Immunogenetics</i> , 2018, 70, 209-222.	1.2	6
122	Relations of mitochondrial genetic variants to measures of vascular function. <i>Mitochondrion</i> , 2018, 40, 51-57.	1.6	7
123	Exome array analysis identifies ETFB as a novel susceptibility gene for anthracycline-induced cardiotoxicity in cancer patients. <i>Breast Cancer Research and Treatment</i> , 2018, 167, 249-256.	1.1	23
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141	Computational modelling approaches as a potential platform to understand the molecular genetics association between Parkinson's and Gaucher diseases. Metabolic Brain Disease, 2018, 33, 1835-1847.	1.4	31
142	Gain-of-function mutations in DNMT3A in patients with paraganglioma. Genetics in Medicine, 2018, 20, 1644-1651.	1.1	73
143	Genetic Regulation of PCSK9 (Proprotein Convertase Subtilisin/Kexin Type 9) Plasma Levels and Its Impact on Atherosclerotic Vascular Disease Phenotypes. Circulation Genomic and Precision Medicine, 2018, 11, e001992.	1.6	37
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145	Establishing the role of PLVAP in protein-losing enteropathy: a homozygous missense variant leads to an attenuated phenotype. <i>Journal of Medical Genetics</i> , 2018, 55, 779-784.	1.5	14
146	Structural analysis of missense mutations in galactokinase 1 (GALK1) leading to galactosemia type 2. <i>Journal of Cellular Biochemistry</i> , 2018, 119, 7585-7598.	1.2	35
147	The Possible Role of Gene Variant Coding Nonfunctional Toll-Like Receptor 2 in the Pathogenesis of Mastocytosis. <i>International Archives of Allergy and Immunology</i> , 2018, 177, 80-86.	0.9	6
148	Investigating the Influence of Hotspot Mutations in Protein-Protein Interaction of IDH1 Homodimer Protein: A Computational Approach. <i>Advances in Protein Chemistry and Structural Biology</i> , 2018, 111, 243-261.	1.0	11
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