

REVEL: An Ensemble Method for Predicting the Pathogenicity of Genetic Variants

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Structural, Functional, and Clinical Characterization of a Novel <i>PTPN11</i> Mutation Cluster Underlying Noonan Syndrome. <i>Human Mutation</i> , 2017, 38, 451-459.	1.1	39
2	Sexual dimorphisms in genetic loci linked to body fat distribution. <i>Bioscience Reports</i> , 2017, 37, .	1.1	58
3	Next-generation sequencing of the monogenic obesity genes <i>LEP</i> , <i>LEPR</i> , <i>MC4R</i> , <i>PCSK1</i> and <i>POMC</i> in a Norwegian cohort of patients with morbid obesity and normal weight controls. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 51-56.	0.5	47
4	Robust and rapid algorithms facilitate large-scale whole genome sequencing downstream analysis in an integrative framework. <i>Nucleic Acids Research</i> , 2017, 45, gkx019.	6.5	36
5	Missense variant pathogenicity predictors generalize well across a range of function-specific prediction challenges. <i>Human Mutation</i> , 2017, 38, 1092-1108.	1.1	39
6	Identification of 8 Novel Mutations in Nephrogenesis-Related Genes in Chinese Han Patients with Unilateral Renal Agenesis. <i>American Journal of Nephrology</i> , 2017, 46, 55-63.	1.4	11
7	Ensemble variant interpretation methods to predict enzyme activity and assign pathogenicity in the <i>CAG14 NAGLU</i> (Human N-Acetylglucosaminidase) and <i>UBE2I</i> (Human SUMO ligase) challenges. <i>Human Mutation</i> , 2017, 38, 1109-1122.	1.1	14
8	<i>TLR3</i> Mutations in Adult Patients With Herpes Simplex Virus and Varicella-Zoster Virus Encephalitis. <i>Journal of Infectious Diseases</i> , 2017, 215, 1430-1434.	1.9	53
9	Nationwide genetic analysis for molecularly unresolved cystic fibrosis patients in a multiethnic society: implications for preconception carrier screening. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 223-236.	0.6	7
10	Mutations in two large pedigrees highlight the role of <i>ZNF711</i> in X-linked intellectual disability. <i>Gene</i> , 2017, 605, 92-98.	1.0	26
11	FIRE: functional inference of genetic variants that regulate gene expression. <i>Bioinformatics</i> , 2017, 33, 3895-3901.	1.8	30
12	Targeted sequencing of 36 known or putative colorectal cancer susceptibility genes. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 553-569.	0.6	32
13	Higher-than-expected population prevalence of potentially pathogenic germline <i>TP53</i> variants in individuals unselected for cancer history. <i>Human Mutation</i> , 2017, 38, 1723-1730.	1.1	40
14	Optimizing genomic medicine in epilepsy through a gene-customized approach to missense variant interpretation. <i>Genome Research</i> , 2017, 27, 1715-1729.	2.4	150
15	The prevalence of <i>DICER1</i> pathogenic variation in population databases. <i>International Journal of Cancer</i> , 2017, 141, 2030-2036.	2.3	75
16	<i>IDUA</i> mutational profile and genotype-phenotype relationships in UK patients with Mucopolysaccharidosis Type I. <i>Human Mutation</i> , 2017, 38, 1555-1568.	1.1	16
17	Settling the score: variant prioritization and Mendelian disease. <i>Nature Reviews Genetics</i> , 2017, 18, 599-612.	7.7	213
18	Exome Pool-Seq in neurodevelopmental disorders. <i>European Journal of Human Genetics</i> , 2017, 25, 1364-1376.	1.4	77

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19	Variant effect prediction tools assessed using independent, functional assay-based datasets: implications for discovery and diagnostics. <i>Human Genomics</i> , 2017, 11, 10.	1.4	68
20	Loss-of-Function and Gain-of-Function Mutations in <i>KCNQ5</i> Cause Intellectual Disability or Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2017, 101, 65-74.	2.6	99
21	Patterns of Novel Alleles and Genotype/Phenotype Correlations Resulting from the Analysis of 108 Previously Undetected Mutations in Patients Affected by Neurofibromatosis Type I. <i>International Journal of Molecular Sciences</i> , 2017, 18, 2071.	1.8	11
22	Exome Sequencing Identified a Novel <i>FBN2</i> Mutation in a Chinese Family with Congenital Contractural Arachnodactyly. <i>International Journal of Molecular Sciences</i> , 2017, 18, 626.	1.8	8
23	Syntool: A Novel Region-Based Intolerance Score to Single Nucleotide Substitution for Synonymous Mutations Predictions Based on 123,136 Individuals. <i>BioMed Research International</i> , 2017, 2017, 1-5.	0.9	7
24	Evaluation of in silico algorithms for use with ACMG/AMP clinical variant interpretation guidelines. <i>Genome Biology</i> , 2017, 18, 225.	3.8	185
25	Analysis of <i>DICER1</i> in familial and sporadic cases of transposition of the great arteries. <i>Congenital Heart Disease</i> , 2018, 13, 401-406.	0.0	2
26	De novo mutations of the <i>ATP6V1A</i> gene cause developmental encephalopathy with epilepsy. <i>Brain</i> , 2018, 141, 1703-1718.	3.7	69
27	VarCards: an integrated genetic and clinical database for coding variants in the human genome. <i>Nucleic Acids Research</i> , 2018, 46, D1039-D1048.	6.5	148
28	Inferring the effect of genomic variation in the new era of genomics. <i>Human Mutation</i> , 2018, 39, 756-773.	1.1	24
29	<i>COQ2</i> variants in Parkinson's disease and multiple system atrophy. <i>Journal of Neural Transmission</i> , 2018, 125, 937-944.	1.4	10
30	Functional Dysregulation of <i>CDC42</i> Causes Diverse Developmental Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 102, 309-320.	2.6	138
31	Bioinformatics analysis of non-synonymous variants in the <i>KLF</i> genes related to cardiac diseases. <i>Gene</i> , 2018, 650, 68-76.	1.0	5
32	Cancer genetics meets biomolecular mechanism bridging an age-old gulf. <i>FEBS Letters</i> , 2018, 592, 463-474.	1.3	9
33	Rare germline mutations in African American men diagnosed with early-onset prostate cancer. <i>Prostate</i> , 2018, 78, 321-326.	1.2	20
34	Tools for protein science. <i>Protein Science</i> , 2018, 27, 6-9.	3.1	2
35	Evaluating the breast cancer predisposition role of rare variants in genes associated with low-penetrance breast cancer risk SNPs. <i>Breast Cancer Research</i> , 2018, 20, 3.	2.2	19
36	Somatic mutation landscape of a meningioma and its pulmonary metastasis. <i>Cancer Communications</i> , 2018, 38, 1-7.	3.7	7

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37	Phenotype risk scores identify patients with unrecognized Mendelian disease patterns. <i>Science</i> , 2018, 359, 1233-1239.	6.0	164
38	Exome sequencing confirms molecular diagnoses in 38 Chinese families with hereditary spherocytosis. <i>Science China Life Sciences</i> , 2018, 61, 947-953.	2.3	38
39	A Distinct Phenotype of Eyes Shut Homolog (EYS)-Retinitis Pigmentosa Is Associated With Variants Near the C-Terminus. <i>American Journal of Ophthalmology</i> , 2018, 190, 99-112.	1.7	23
40	Primary mediastinal paraganglioma associated with a familial variant in the succinate dehydrogenase B subunit gene. <i>Journal of Surgical Oncology</i> , 2018, 117, 160-162.	0.8	6
41	A targeted sequencing panel identifies rare damaging variants in multiple genes in the cranial neural tube defect, anencephaly. <i>Clinical Genetics</i> , 2018, 93, 870-879.	1.0	29
42	GRIP: a novel case-control analysis method for Mendelian disease gene discovery. <i>Genome Biology</i> , 2018, 19, 203.	3.8	3
43	Assessment of coding region variants in Kuwaiti population: implications for medical genetics and population genomics. <i>Scientific Reports</i> , 2018, 8, 16583.	1.6	26
44	Functional characterization of TRPM4 variants identified in sudden unexpected natural death. <i>Forensic Science International</i> , 2018, 293, 37-46.	1.3	11
45	Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium. <i>Scientific Reports</i> , 2018, 8, 17201.	1.6	70
46	Multimodal imaging in a pedigree of X-linked Retinoschisis with a novel RS1 variant. <i>BMC Medical Genetics</i> , 2018, 19, 195.	2.1	5
47	Computational Methods for the Pharmacogenetic Interpretation of Next Generation Sequencing Data. <i>Frontiers in Pharmacology</i> , 2018, 9, 1437.	1.6	62
48	Description of 22 new alpha-1 antitrypsin genetic variants. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 161.	1.2	19
49	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. <i>Journal of Chemical Information and Modeling</i> , 2018, 58, 2131-2150.	2.5	20
50	<i>RYR1</i> and <i>CACNA1S</i> genetic variants identified with statin-associated muscle symptoms. <i>Pharmacogenomics</i> , 2018, 19, 1235-1249.	0.6	20
51	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. <i>Human Mutation</i> , 2018, 39, 1593-1613.	1.1	312
52	Bioinformatics in Clinical Genomic Sequencing. <i>Advances in Molecular Pathology</i> , 2018, 1, 9-26.	0.2	1
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57	From genome-wide associations to candidate causal variants by statistical fine-mapping. <i>Nature Reviews Genetics</i> , 2018, 19, 491-504.	7.7	611
58	Improved, ACMG-compliant, in silico prediction of pathogenicity for missense substitutions encoded by <i>TP53</i> variants. <i>Human Mutation</i> , 2018, 39, 1061-1069.	1.1	29
59	Functional Assays Are Essential for Interpretation of Missense Variants Associated with Variable Expressivity. <i>American Journal of Human Genetics</i> , 2018, 102, 1062-1077.	2.6	69
60	Whole-exome sequencing for variant discovery in blepharospasm. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 601-626.	0.6	20
61	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. <i>Neuron</i> , 2018, 99, 302-314.e4.	3.8	112
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67	Accurate prediction of functional, structural, and stability changes in <i>PITX2</i> mutations using in silico bioinformatics algorithms. <i>PLoS ONE</i> , 2018, 13, e0195971.	1.1	35
68	Predicting the clinical impact of human mutation with deep neural networks. <i>Nature Genetics</i> , 2018, 50, 1161-1170.	9.4	288
69	Genetic polymorphisms of <i>CYP2S1</i> , <i>CYP2J2</i> and <i>CYP2R1</i> genes in three Chinese populations: Han, Tibetan and Uighur. <i>Pharmacogenomics</i> , 2018, 19, 961-977.	0.6	5
70	Performance evaluation of pathogenicity-computation methods for missense variants. <i>Nucleic Acids Research</i> , 2018, 46, 7793-7804.	6.5	168
71	Deep Scleral Exposure: A Degenerative Outcome of End-Stage Stargardt Disease. <i>American Journal of Ophthalmology</i> , 2018, 195, 16-25.	1.7	10
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78	Novel DCC variants in congenital mirror movements and evaluation of disease-associated missense variants. <i>European Journal of Medical Genetics</i> , 2018, 61, 329-334.	0.7	7
79	Evidence for <i>GALNT12</i> as a moderate penetrance gene for colorectal cancer. <i>Human Mutation</i> , 2018, 39, 1092-1101.	1.1	20
80	Diagnostics of rare disorders: whole-exome sequencing deciphering locus heterogeneity in telomere biology disorders. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 139.	1.2	8
81	Identification of pathways and genes associated with cerebral palsy. <i>Genes and Genomics</i> , 2018, 40, 1339-1349.	0.5	8
82	Real-world clinical applicability of pathogenicity predictors assessed on <i>SERPINA1</i> mutations in alpha-1-antitrypsin deficiency. <i>Human Mutation</i> , 2018, 39, 1203-1213.	1.1	36
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89	Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. <i>Npj Genomic Medicine</i> , 2019, 4, 18.	1.7	29
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93	Using MARRVEL v1.2 for Bioinformatics Analysis of Human Genes and Variant Pathogenicity. <i>Current Protocols in Bioinformatics</i> , 2019, 67, e85.	25.8	14
94	CAGI5: Objective performance assessments of predictions based on the Evolutionary Action equation. <i>Human Mutation</i> , 2019, 40, 1436-1454.	1.1	26
95	Genotype-phenotype associations in Fanconi anemia: A literature review. <i>Blood Reviews</i> , 2019, 37, 100589.	2.8	116
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97	Assessing computational predictions of the phenotypic effect of cystathionine β -synthase variants. <i>Human Mutation</i> , 2019, 40, 1530-1545.	1.1	5
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100	DNA Damage Response and Repair Pathway Alteration and Its Association With Tumor Mutation Burden and Platinum-Based Chemotherapy in SCLC. <i>Journal of Thoracic Oncology</i> , 2019, 14, 1640-1650.	0.5	64
101	VIPdb, a genetic Variant Impact Predictor Database. <i>Human Mutation</i> , 2019, 40, 1202-1214.	1.1	24
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103	Assessment of predicted enzymatic activity of \pm N-acetylglucosaminidase variants of unknown significance for CAGI 2016. <i>Human Mutation</i> , 2019, 40, 1519-1529.	1.1	10
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106	Genetic diagnosis in first or second trimester pregnancy loss using exome sequencing: a systematic review of human essential genes. <i>Journal of Assisted Reproduction and Genetics</i> , 2019, 36, 1539-1548.	1.2	25
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115	Disease modeling of a mutation in <i>ACTN1</i> guides clinical therapy in hypertrophic cardiomyopathy. <i>EMBO Molecular Medicine</i> , 2019, 11, e11115.	3.3	88
116	Biallelic <i>DMXL2</i> mutations impair autophagy and cause Ohtahara syndrome with progressive course. <i>Brain</i> , 2019, 142, 3876-3891.	3.7	23
117	In Vivo Functional Study of Disease-associated Rare Human Variants Using <i>Drosophila</i> . <i>Journal of Visualized Experiments</i> , 2019, , .	0.2	34
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129	Germline variation in O6-methylguanine-DNA methyltransferase (MGMT) as cause of hereditary colorectal cancer. <i>Cancer Letters</i> , 2019, 447, 86-92.	3.2	12
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133	Improved Pathogenic Variant Localization via a Hierarchical Model of Sub-regional Intolerance. <i>American Journal of Human Genetics</i> , 2019, 104, 299-309.	2.6	29
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138	The Relevance of Variants With Unknown Significance for Autism Spectrum Disorder Considering the Genotype-Phenotype Interrelationship. <i>Frontiers in Psychiatry</i> , 2019, 10, 409.	1.3	8
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143	Assessment of patient clinical descriptions and pathogenic variants from gene panel sequences in the CAGI5 intellectual disability challenge. <i>Human Mutation</i> , 2019, 40, 1330-1345.	1.1	11
144	Gene pathogenicity prediction of Mendelian diseases via the random forest algorithm. <i>Human Genetics</i> , 2019, 138, 673-679.	1.8	4
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147	Challenges and Considerations in Sequence Variant Interpretation for Mendelian Disorders. <i>Annals of Laboratory Medicine</i> , 2019, 39, 421-429.	1.2	31
148	Excessive Seizure Clusters in an Otherwise Well-Controlled Epilepsy as a Possible Hallmark of Untreated Vitamin B6-Responsive Epilepsy due to a Homozygous PLPBP Missense Variant. <i>Journal of Pediatric Genetics</i> , 2019, 08, 222-225.	0.3	9
149	A segregating human allele of <i>SPO11</i> modeled in mice disrupts timing and amounts of meiotic recombination, causing oligospermia and a decreased ovarian reserve. <i>Biology of Reproduction</i> , 2019, 101, 347-359.	1.2	10
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