

Disease variant prediction with deep generative models

Nature

599, 91-95

DOI: [10.1038/s41586-021-04043-8](https://doi.org/10.1038/s41586-021-04043-8)

Citation Report

#	ARTICLE	IF	CITATIONS
6	TP53_PROF: a machine learning model to predict impact of missense mutations in <i>TP53</i>. Briefings in Bioinformatics, 2022, 23, .	3.2	9
8	Extracting phylogenetic dimensions of coevolution reveals hidden functional signals. Scientific Reports, 2022, 12, 820.	1.6	12
9	Predicting and interpreting large-scale mutagenesis data using analyses of protein stability and conservation. Cell Reports, 2022, 38, 110207.	2.9	62
11	Genetic load: genomic estimates and applications in non-model animals. Nature Reviews Genetics, 2022, 23, 492-503.	7.7	82
12	The TKFC Ala185Thr variant, reported as "null"™ for fructose metabolism, is fully active as triokinase. FEBS Letters, 2022, . .	1.3	1
13	Exploring the relevance of NUP93 variants in steroid-resistant nephrotic syndrome using next generation sequencing and a fly kidney model. Pediatric Nephrology, 2022, 37, 2643-2656.	0.9	5
14	Dynamic disequilibrium-based pathogenicity model in mutated pyrin™s B30.2 domain™Casp1/p20 complex. Journal of Genetic Engineering and Biotechnology, 2022, 20, 31.	1.5	6
18	Machine-learning of complex evolutionary signals improves classification of SNVs. NAR Genomics and Bioinformatics, 2022, 4, lqac025.	1.5	4
20	Efficient Exploration of Sequence Space by Sequence-Guided Protein Engineering and Design. Biochemistry, 2023, 62, 210-220.	1.2	11
21	Case Report: Exome and RNA Sequencing Identify a Novel de novo Missense Variant in HNRNPK in a Chinese Patient With Au-Kline Syndrome. Frontiers in Genetics, 2022, 13, 853028.	1.1	1
23	Table Tennis Capture System Based on Image Recognition and Modeling. Scientific Programming, 2022, 2022, 1-11.	0.5	1
24	Democratizing the mapping of gene mutations to protein biophysics. Nature, 2022, 604, 47-48.	13.7	0
25	Learning meaningful representations of protein sequences. Nature Communications, 2022, 13, 1914.	5.8	55
26	Functional analysis of ATM variants in a high risk cohort provides insight into missing heritability. Cancer Genetics, 2022, 264-265, 40-49.	0.2	2
27	Predicting disease variants using biodiversity and machine learning. Nature Biotechnology, 2022, 40, 27-28.	9.4	3
30	Identification and characterization of two novel noncoding tyrosinase (TYR) gene variants leading to oculocutaneous albinism type 1. Journal of Biological Chemistry, 2022, 298, 101922.	1.6	2
31	Neural Networks for Classification and Image Generation of Aging in Genetic Syndromes. Frontiers in Genetics, 2022, 13, 864092.	1.1	14
32	Characterizing and explaining the impact of disease-associated mutations in proteins without known structures or structural homologs. Briefings in Bioinformatics, 2022, 23, .	3.2	18

#	ARTICLE	IF	CITATIONS
33	Computational and experimental methods for classifying variants of unknown clinical significance.. Cold Spring Harbor Molecular Case Studies, 2022, 8, .	0.7	7
34	Computed Tomography Texture Features and Risk Factor Analysis of Postoperative Recurrence of Patients with Advanced Gastric Cancer after Radical Treatment under Artificial Intelligence Algorithm. Computational Intelligence and Neuroscience, 2022, 2022, 1-9.	1.1	1
35	Analysis of 6.4 million SARS-CoV-2 genomes identifies mutations associated with fitness. Science, 2022, 376, 1327-1332.	6.0	172
39	Determinants of trafficking, conduction, and disease within a K ⁺ channel revealed through multiparametric deep mutational scanning. ELife, 0, 11, .	2.8	23
42	Interpreting protein variant effects with computational predictors and deep mutational scanning. DMM Disease Models and Mechanisms, 2022, 15, .	1.2	25
43	Interpretable pairwise distillations for generative protein sequence models. PLoS Computational Biology, 2022, 18, e1010219.	1.5	1
44	The impact of rare germline variants on human somatic mutation processes. Nature Communications, 2022, 13, .	5.8	13
45	Revision of <i>RUNX1</i> variant curation rules. Blood Advances, 2022, 6, 4726-4730.	2.5	7
46	The Location of Missense Variants in the Human GIP Gene Is Indicative for Natural Selection. Frontiers in Endocrinology, 0, 13, .	1.5	1
47	Proactive functional classification of all possible missense single-nucleotide variants in <i>KCNQ4</i> . Genome Research, 2022, 32, 1573-1584.	2.4	8
48	A Chromosome-Length Assembly of the Hawaiian Monk Seal (<i>Neomonachus schauinslandi</i>): A History of Genetic Purgings and Genomic Stability. Genes, 2022, 13, 1270.	1.0	1
52	How Functional Genomics Can Keep Pace With VUS Identification. Frontiers in Cardiovascular Medicine, 0, 9, .	1.1	8
53	Observation of Clinical Efficacy of Anisodamine and Chlorpromazine in the Treatment of Intractable Hiccup after Stroke. BioMed Research International, 2022, 2022, 1-10.	0.9	1
55	Approach to the patient with a variant of uncertain significance on genetic testing. Clinical Endocrinology, 0, , .	1.2	3
57	Heterozygous variants in MYH10 associated with neurodevelopmental disorders and congenital anomalies with evidence for primary cilia-dependent defects in Hedgehog signaling. Genetics in Medicine, 2022, 24, 2065-2078.	1.1	2
60	mvPPT: A Highly Efficient and Sensitive Pathogenicity Prediction Tool for Missense Variants. Genomics, Proteomics and Bioinformatics, 2023, 21, 414-426.	3.0	2
61	Clinical significance of genetic variation in hypertrophic cardiomyopathy: comparison of computational tools to prioritize missense variants. Frontiers in Cardiovascular Medicine, 0, 9, .	1.1	5
62	Challenges and opportunities associated with rare-variant pharmacogenomics. Trends in Pharmacological Sciences, 2022, 43, 852-865.	4.0	14

#	ARTICLE	IF	CITATIONS
63	Germline Missense Variants in <i>CDC20</i> Result in Aberrant Mitotic Progression and Familial Cancer. <i>Cancer Research</i> , 2022, 82, 3499-3515.	0.4	4
64	Neuromuscular disorders: finding the missing genetic diagnoses. <i>Trends in Genetics</i> , 2022, 38, 956-971.	2.9	4
65	Generating depth images of preterm infants in given poses using GANs. <i>Computer Methods and Programs in Biomedicine</i> , 2022, 225, 107057.	2.6	1
66	Vasor: Accurate prediction of variant effects for amino acid substitutions in multidrug resistance protein 3. <i>Hepatology Communications</i> , 2022, 6, 3098-3111.	2.0	3
67	Treating Superhard Materials as Anomalies. <i>Journal of the American Chemical Society</i> , 2022, 144, 18075-18080.	6.6	8
68	Computational approaches for predicting variant impact: An overview from resources, principles to applications. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	11
69	Artificial Intelligence Applied to Cardiomyopathies: Is It Time for Clinical Application?. <i>Current Cardiology Reports</i> , 2022, 24, 1547-1555.	1.3	1
72	Artificial intelligence in hematological diagnostics: Game changer or gadget?. <i>Blood Reviews</i> , 2023, 58, 101019.	2.8	19
74	The CancerMuts software package for the prioritization of missense cancer variants: a case study of <i>AMBRA1</i> in melanoma. <i>Cell Death and Disease</i> , 2022, 13, .	2.7	7
75	Shared Cancer Dataset Analysis Identifies and Predicts the Quantitative Effects of Pan-Cancer Somatic Driver Variants. <i>Cancer Research</i> , 2023, 83, 74-88.	0.4	2
76	Evolutionary coupling analysis guides identification of mistrafficking-sensitive variants in cardiac K ⁺ channels: Validation with hERG. <i>Frontiers in Pharmacology</i> , 0, 13, .	1.6	0
79	Intragenic compensation through the lens of deep mutational scanning. <i>Biophysical Reviews</i> , 2022, 14, 1161-1182.	1.5	4
80	Comprehensive In Silico Functional Prediction Analysis of CDKL5 by Single Amino Acid Substitution in the Catalytic Domain. <i>International Journal of Molecular Sciences</i> , 2022, 23, 12281.	1.8	0
81	Refactored genetic codes enable bidirectional genetic isolation. <i>Science</i> , 2022, 378, 516-523.	6.0	22
82	Ensembl 2023. <i>Nucleic Acids Research</i> , 2023, 51, D933-D941.	6.5	153
84	<i>In silico</i> versus functional characterization of genetic variants: lessons from muscle channelopathies. <i>Brain</i> , 2023, 146, 1316-1321.	3.7	1
85	End-to-end learning of multiple sequence alignments with differentiable Smith-Waterman. <i>Bioinformatics</i> , 2023, 39, .	1.8	11
86	Functional genomics of OCTN2 variants informs protein-specific variant effect predictor for Carnitine Transporter Deficiency. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	6

#	ARTICLE	IF	CITATIONS
87	Genetics of congenital arrhythmia syndromes: the challenge of variant interpretation. <i>Current Opinion in Genetics and Development</i> , 2022, 77, 102004.	1.5	4
89	From sequence to function through structure: Deep learning for protein design. <i>Computational and Structural Biotechnology Journal</i> , 2023, 21, 238-250.	1.9	29
91	Artificial intelligence-based recognition for variant pathogenicity of BRCA1 using AlphaFold2-predicted structures. <i>Theranostics</i> , 2023, 13, 391-402.	4.6	2
94	Lynch syndrome, molecular mechanisms and variant classification. <i>British Journal of Cancer</i> , 2023, 128, 726-734.	2.9	8
95	Effect of naturally-occurring mutations on the stability and function of cancer-associated NQO1: Comparison of experiments and computation. <i>Frontiers in Molecular Biosciences</i> , 0, 9, .	1.6	4
96	Data-driven historical characterization of epilepsy-associated genes. <i>European Journal of Paediatric Neurology</i> , 2023, 42, 82-87.	0.7	11
97	Opportunities and Challenges with Artificial Intelligence in Genomics. <i>Clinics in Laboratory Medicine</i> , 2023, 43, 87-97.	0.7	2
98	A robust pipeline for ranking carrier frequencies of autosomal recessive and X-linked Mendelian disorders. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	3
100	Predicting pathogenicity from non-coding mutations. <i>Nature Biomedical Engineering</i> , 0, , .	11.6	0
105	Is artificial intelligence capable of generating hospital discharge summaries from inpatient records?. , 2022, 1, e0000158.		3
106	Multiple-model machine learning identifies potential functional genes in dilated cardiomyopathy. <i>Frontiers in Cardiovascular Medicine</i> , 0, 9, .	1.1	1
107	Bayesian Inference and Dynamic Neural Feedback Promote the Clinical Application of Intelligent Congenital Heart Disease Diagnosis. <i>Engineering</i> , 2023, 23, 90-102.	3.2	3
108	Discovery of drug-omics associations in type 2 diabetes with generative deep-learning models. <i>Nature Biotechnology</i> , 2023, 41, 399-408.	9.4	18
109	Deep mutational scanning: A versatile tool in systematically mapping genotypes to phenotypes. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	6
111	Interpreting the molecular mechanisms of disease variants in human transmembrane proteins. <i>Biophysical Journal</i> , 2023, 122, 2176-2191.	0.2	7
112	Comparison of In Vitro and In Silico Assessments of Human Galactose-1-Phosphate Uridyltransferase Coding Variants. <i>Cureus</i> , 2023, , .	0.2	0
113	The global prevalence and ethnic heterogeneity of iron-refractory iron deficiency anaemia. <i>Orphanet Journal of Rare Diseases</i> , 2023, 18, .	1.2	2
114	Mutually beneficial confluence of structure-based modeling of protein dynamics and machine learning methods. <i>Current Opinion in Structural Biology</i> , 2023, 78, 102517.	2.6	11

#	ARTICLE	IF	CITATIONS
115	Third-generation computational approaches for genetic variant interpretation. <i>Brain</i> , 2023, 146, 411-412.	3.7	5
116	Machine learning in computational modelling of membrane protein sequences and structures: From methodologies to applications. <i>Computational and Structural Biotechnology Journal</i> , 2023, 21, 1205-1226.	1.9	3
117	Predicting mutational function using machine learning. <i>Mutation Research - Reviews in Mutation Research</i> , 2023, 791, 108457.	2.4	0
119	Towards the accurate alignment of over a million protein sequences: Current state of the art. <i>Current Opinion in Structural Biology</i> , 2023, 80, 102577.	2.6	1
120	SESNet: sequence-structure feature-integrated deep learning method for data-efficient protein engineering. <i>Journal of Cheminformatics</i> , 2023, 15, .	2.8	4
121	Challenging gold standard hematology diagnostics through the introduction of whole genome sequencing and artificial intelligence. <i>International Journal of Laboratory Hematology</i> , 2023, 45, 156-162.	0.7	1
122	Persistent spectral theory-guided protein engineering. <i>Nature Computational Science</i> , 2023, 3, 149-163.	3.8	17
124	ProtelInfer, deep neural networks for protein functional inference. <i>ELife</i> , 0, 12, .	2.8	31
125	Inferring protein fitness landscapes from laboratory evolution experiments. <i>PLoS Computational Biology</i> , 2023, 19, e1010956.	1.5	7
128	Comprehensive laboratory diagnosis of Fanconi anaemia: comparison of cellular and molecular analysis. <i>Journal of Medical Genetics</i> , 2023, 60, 801-809.	1.5	1
129	SMPD1 expression profile and mutation landscape help decipher genotypeâ€“phenotype association and precision diagnosis for acid sphingomyelinase deficiency. <i>Hereditas</i> , 2023, 160, .	0.5	2
130	Mutations in IFN- γ signaling genes sensitize tumors to immune checkpoint blockade. <i>Cancer Cell</i> , 2023, 41, 651-652.	7.7	7
131	An Assessment of Quaternary Structure Functionality in Homomer Protein Complexes. <i>Molecular Biology and Evolution</i> , 2023, 40, .	3.5	2
132	Application of machine learning in prediction of bone cement leakage during single-level thoracolumbar percutaneous vertebroplasty. <i>BMC Surgery</i> , 2023, 23, .	0.6	1
133	Enzyme function prediction using contrastive learning. <i>Science</i> , 2023, 379, 1358-1363.	6.0	59
134	Pathogenicity predicting algorithm of single amino acid variants. , 2022, , .		0
135	Clinical profiling of MRD48 and functional characterization of two novel pathogenic RAC1 variants. <i>European Journal of Human Genetics</i> , 0, , .	1.4	3
136	Latent generative landscapes as maps of functional diversity in protein sequence space. <i>Nature Communications</i> , 2023, 14, .	5.8	7

#	ARTICLE	IF	CITATIONS
137	DeMAG predicts the effects of variants in clinically actionable genes by integrating structural and evolutionary epistatic features. Nature Communications, 2023, 14, .	5.8	2
138	Predicting the pathogenicity of missense variants using features derived from AlphaFold2. Bioinformatics, 2023, 39, .	1.8	13
141	From Genomes to Variant Interpretations Through Protein Structures. SpringerBriefs in Applied Sciences and Technology, 2023, , 41-50.	0.2	0
166	How protein fold: Insights from nuclear magnetic resonance spectroscopy. , 2024, , 619-635.		0
190	Learn from the past to predict viral pandemics. Nature, 2023, 622, 700-702.	13.7	0
192	Machine Learning for Protein Engineering. Challenges and Advances in Computational Chemistry and Physics, 2023, , 277-311.	0.6	1
198	Advancing variant effect prediction using protein language models. Nature Genetics, 2023, 55, 1426-1427.	9.4	1
264	Heterogeneities in Hereditary Cancer Genes as Revealed by a Large-Scale Genome Analysis. , 2023, , 59-78.		0
290	Machine learning for functional protein design. Nature Biotechnology, 2024, 42, 216-228.	9.4	1