Predicting functional effects of missense variants in vol channels

Science Translational Medicine

12,

DOI: 10.1126/scitranslmed.aay6848

Citation Report

#	Article	IF	Citations
1	Comprehensive characterization of amino acid positions in protein structures reveals molecular effect of missense variants. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 28201-28211.	3.3	68
2	<scp>MYT1L</scp>: A systematic review of genetic variation encompassing schizophrenia and autism. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 227-233.	1.1	19
3	Artificial intelligence and machine learningâ€aided drug discovery in central nervous system diseases: Stateâ€ofâ€theâ€arts and future directions. Medicinal Research Reviews, 2021, 41, 1427-1473.	5.0	120
4	Clinical spectrum and treatment outcome of 95 children with continuous spikes and waves during sleep (CSWS). European Journal of Paediatric Neurology, 2021, 30, 121-127.	0.7	17
5	The study of sodium and potassium channel gene single-nucleotide variation significance in non-mechanical forms of epilepsy. Egyptian Journal of Medical Human Genetics, 2021, 22, .	0.5	1
8	MVP predicts theÂpathogenicity of missense variants by deep learning. Nature Communications, 2021, 12, 510.	5.8	85
9	Assessing the impact of pain-linked Nav1.7 variants: An example of two variants with no biophysical effect. Channels, 2021, 15, 208-228.	1.5	5
11	Non-extensitivity and criticality of atomic hydropathicity around a voltage-gated sodium channelâ€™s pore: a modeling study. Journal of Biological Physics, 2021, 47, 61-77.	0.7	3
12	Computational analysis of 10,860 phenotypic annotations in individuals with SCN2A-related disorders. Genetics in Medicine, 2021, 23, 1263-1272.	1.1	38
13	Hydropathicity-based prediction of pain-causing NaV1.7 variants. BMC Bioinformatics, 2021, 22, 212.	1.2	5
14	SCN8A Encephalopathy: Case Report and Literature Review. Neurology International, 2021, 13, 143-150.	1.3	7
16	Opportunities and challenges for the computational interpretation of rare variation in clinically important genes. American Journal of Human Genetics, 2021, 108, 535-548.	2.6	40
17	Autism-associated SHANK3 missense point mutations impact conformational fluctuations and protein turnover at synapses. ELife, 2021, 10, .	2.8	14
18	Computational Tools to Assess the Functional Consequences of Rare and Noncoding Pharmacogenetic Variability. Clinical Pharmacology and Therapeutics, 2021, 110, 626-636.	2.3	16
19	Advances in genotype-phenotype associations for CACNA1A-related epilepsies. European Journal of Paediatric Neurology, 2021, 33, A2.	0.7	1
20	The suitability of high throughput automated patch clamp for physiological applications. Journal of Physiology, 2022, 600, 277-297.	1.3	18
21	Heterozygous KCNH2 variant phenotyping using Flp-In HEK293 and high-throughput automated patch clamp electrophysiology. Biology Methods and Protocols, 2021, 6, bpab003.	1.0	12
22	Persistent sodium currents in SCN1A developmental and degenerative epileptic dyskinetic encephalopathy. Brain Communications, 2021, 3, fcab235.	1.5	12

#	Article	IF	Citations
23	Pathogenic in-Frame Variants in SCN8A: Expanding the Genetic Landscape of SCN8A-Associated Disease. Frontiers in Pharmacology, 2021, 12, 748415.	1.6	1
24	Identification of discriminative gene-level and protein-level features associated with pathogenic gain-of-function and loss-of-function variants. American Journal of Human Genetics, 2021, 108, 2301-2318.	2.6	21
27	Gene variant effects across sodium channelopathies predict function and guide precision therapy. Brain, 2022, 145, 4275-4286.	3.7	43
28	Novel Fluorescence-Based High-Throughput FLIPR Assay Utilizing Membrane-Tethered Genetic Calcium Sensors to Identify T-Type Calcium Channel Modulators. ACS Pharmacology and Translational Science, 2022, 5, 156-168.	2.5	1
29	Genetic Landscape of <i>SCN1A</i> Variants in a Turkish Cohort with GEFS+ Spectrum and Dravet Syndrome. Molecular Syndromology, 0, , 1-12.	0.3	2
32	Further delineation of phenotypic spectrum of <scp>SCN2A</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2022, 188, 867-877.	0.7	3
33	Analysing an allelic series of rare missense variants of CACNA1I in a Swedish schizophrenia cohort. Brain, 2022, 145, 1839-1853.	3.7	18
34	Integration of Protein Structure and Population-Scale DNA Sequence Data for Disease Gene Discovery and Variant Interpretation. Annual Review of Biomedical Data Science, 2022, 5, .	2.8	0
35	Functional correlates of clinical phenotype and severity in recurrent SCN2A variants. Communications Biology, 2022, 5, .	2.0	13
36	Genetic pain loss disorders. Nature Reviews Disease Primers, 2022, 8, .	18.1	18
37	Clinical and genetic characterization of <scp>CACNA1A</scp>â€related disease. Clinical Genetics, 2022, 102, 288-295.	1.0	9
38	The gain of function SCN1A disorder spectrum: novel epilepsy phenotypes and therapeutic implications. Brain, 2022, 145, 3816-3831.	3.7	43
39	Predicting the functional effects of voltage-gated potassium channel missense variants with multi-task learning. EBioMedicine, 2022, 81, 104115.	2.7	8
41	Current practice in diagnostic genetic testing of the epilepsies. Epileptic Disorders, 2022, 24, 765-786.	0.7	37
42	Investigation of CACNA1I Cav3.3 Dysfunction in Hemiplegic Migraine. Frontiers in Molecular Neuroscience, 0, 15, .	1.4	7
43	Technical Applications of Microelectrode Array and Patch Clamp Recordings on Human Induced Pluripotent Stem Cell-Derived Cardiomyocytes. Journal of Visualized Experiments, 2022, , .	0.2	1
44	Case report: Novel SCN4A variant associated with a severe congenital myasthenic syndrome/myopathy phenotype. Frontiers in Pediatrics, 0, 10, .	0.9	3
45	A cross-disorder dosage sensitivity map of the human genome. Cell, 2022, 185, 3041-3055.e25.	13.5	117

#	Article	IF	Citations
46	Severe epilepsy phenotype with SCN1A missense variants located outside the sodium channel core region: Relationship between functional results and clinical phenotype. Seizure: the Journal of the British Epilepsy Association, 2022, 101, 109-116.	0.9	2
47	De novo SCN3A missense variant associated with self-limiting generalized epilepsy with fever sensitivity. European Journal of Medical Genetics, 2022, 65, 104577.	0.7	0
48	SCN1A and Dravet syndrome. , 2023, , 43-63.		1
49	Conserved patterns across ion channels correlate with variant pathogenicity and clinical phenotypes. Brain, 2023, 146, 923-934.	3.7	11
51	Delineation of functionally essential protein regions for 242 neurodevelopmental genes. Brain, 2023, 146, 519-533.	3.7	6
52	Evolutionary coupling analysis guides identification of mistrafficking-sensitive variants in cardiac K+ channels: Validation with hERG. Frontiers in Pharmacology, 0, 13, .	1.6	0
53	Predictive precision medicine efforts for voltage-gated sodium channel genetic variants. Brain, 2022, 145, 4148-4150.	3.7	1
54	Predicting functional effect of missense variants using graph attention neural networks. Nature Machine Intelligence, 2022, 4, 1017-1028.	8.3	15
55	In silico versus functional characterization of genetic variants: lessons from muscle channelopathies. Brain, 2023, 146, 1316-1321.	3.7	1
56	Genetics of congenital arrhythmia syndromes: the challenge of variant interpretation. Current Opinion in Genetics and Development, 2022, 77, 102004.	1.5	4
57	Data-driven historical characterization of epilepsy-associated genes. European Journal of Paediatric Neurology, 2023, 42, 82-87.	0.7	11
58	Continuous Bayesian variant interpretation accounts for incomplete penetrance among Mendelian cardiac channelopathies. Genetics in Medicine, 2023, 25, 100355.	1.1	4
59	De novo mutation hotspots in homologous protein domains identify function-altering mutations in neurodevelopmental disorders. American Journal of Human Genetics, 2023, 110, 92-104.	2.6	3
60	Automated Patch-Clamp and Induced Pluripotent Stem Cell-Derived Cardiomyocytes: A Synergistic Approach in the Study of Brugada Syndrome. International Journal of Molecular Sciences, 2023, 24, 6687.	1.8	2
61	KCNA1 gainâ€ofâ€function epileptic encephalopathy treated with 4âaminopyridine. Annals of Clinical and Translational Neurology, 2023, 10, 656-663.	1.7	8
62	Investigating genotypeâ€“phenotype relationship of extreme neuropathic pain disorders in a UK national cohort. Brain Communications, 2023, 5, .	1.5	3
63	Mendelian inheritance revisited: dominance and recessiveness in medical genetics. Nature Reviews Genetics, 2023, 24, 442-463.	7.7	16
64	Predicting functional effects of ion channel variants using new phenotypic machine learning methods. PLoS Computational Biology, 2023, 19, e1010959.	1.5	7

#	Article	IF	Citations