Eduardo PÃ©rez-Palma

List of Publications by Year
in descending order

20 papers	663 citations	686830 13 h-index	752256 20 g-index
23 all docs	23 docs citations	23 times ranked	1583 citing authors

#	Article	IF	Citations
1	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. Science Translational Medicine, 2020, 12, .	5.8	84
2	Wnt/β-Catenin Signaling in Alzheimerâ€™s Disease. CNS and Neurological Disorders - Drug Targets, 2014, 13, 745-754.	0.8	82
3	Spectrum of GABAA receptor variants in epilepsy. Current Opinion in Neurology, 2019, 32, 183-190.	1.8	59
4	Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphateâ€“binding region. Epilepsia, 2019, 60, 406-418.	2.6	53
5	Overrepresentation of Glutamate Signaling in Alzheimer's Disease: Network-Based Pathway Enrichment Using Meta-Analysis of Genome-Wide Association Studies. PLoS ONE, 2014, 9, e95413.	1.1	52
6	Simple ClinVar: an interactive web server to explore and retrieve gene and disease variants aggregated in ClinVar database. Nucleic Acids Research, 2019, 47, W99-W105.	6.5	51
7	Identification of pathogenic variant enriched regions across genes and gene families. Genome Research, 2020, 30, 62-71.	2.4	47
8	Current knowledge of SLC6A1-related neurodevelopmental disorders. Brain Communications, 2020, 2, fcaa170.	1.5	44
9	A novel functional low-density lipoprotein receptor-related protein 6 gene alternative splice variant is associated with Alzheimer's disease. Neurobiology of Aging, 2013, 34, 1709.e9-1709.e18.	1.5	39
10	Variants in ABCG8 and TRAF3 genes confer risk for gallstone disease in admixed Latinos with Mapuche Native American ancestry. Scientific Reports, 2019, 9, 772.	1.6	30
11	Development and Validation of a Prediction Model for Early Diagnosis of SCN1A -Related Epilepsies. Neurology, 2022, 98, .	1.5	24
12	Heterogeneous contribution of microdeletions in the development of common generalised and focal epilepsies. Journal of Medical Genetics, 2017, 54, 598-606.	1.5	22
13	Early Transcriptional Changes Induced by Wnt/Î²-Catenin Signaling in Hippocampal Neurons. Neural Plasticity, 2016, 2016, 1-13.	1.0	19
14	Assessment of genetic variant burden in epilepsy-associated brain lesions. European Journal of Human Genetics, 2019, 27, 1738-1744.	1.4	12
15	Whole Genome Sequence, Variant Discovery and Annotation in Mapuche-Huilliche Native South Americans. Scientific Reports, 2019, 9, 2132.	1.6	12
16	Genomeâ€Wide Analysis of Copy Number Variation in Latin American Parkinson's Disease Patients. Movement Disorders, 2021, 36, 434-441.	2.2	12
17	Copy number variants in lipid metabolism genes are associated with gallstones disease in men. European Journal of Human Genetics, 2020, 28, 264-273.	1.4	6
18	Variant Score Rankerâ€”a web application for intuitive missense variant prioritization. Bioinformatics, 2019, 35, 4478-4479.	1.8	5

#	Article	IF	Citations
19	Duplications at 19q13.33 in patients with neurodevelopmental disorders. Neurology: Genetics, 2018, 4, e210.	0.9	4
20	Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy. New England Journal of Medicine, 2019, 380, e24.	13.9	4