

Eduardo PÃ©rez-Palma

List of Publications by Year in descending order

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Version: 2024-02-01

20
papers

663
citations

687220

13
h-index

752573

20
g-index

23
all docs

23
docs citations

23
times ranked

1583
citing authors

#	ARTICLE	IF	CITATIONS
1	Development and Validation of a Prediction Model for Early Diagnosis of <i>SCN1A</i> -Related Epilepsies. <i>Neurology</i> , 2022, 98, .	1.5	24
2	Genome-Wide Analysis of Copy Number Variation in Latin American Parkinson's Disease Patients. <i>Movement Disorders</i> , 2021, 36, 434-441.	2.2	12
3	Copy number variants in lipid metabolism genes are associated with gallstones disease in men. <i>European Journal of Human Genetics</i> , 2020, 28, 264-273.	1.4	6
4	Identification of pathogenic variant enriched regions across genes and gene families. <i>Genome Research</i> , 2020, 30, 62-71.	2.4	47
5	Current knowledge of SLC6A1-related neurodevelopmental disorders. <i>Brain Communications</i> , 2020, 2, fcaa170.	1.5	44
6	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. <i>Science Translational Medicine</i> , 2020, 12, .	5.8	84
7	Assessment of genetic variant burden in epilepsy-associated brain lesions. <i>European Journal of Human Genetics</i> , 2019, 27, 1738-1744.	1.4	12
8	Variants in <i>ABCG8</i> and <i>TRAF3</i> genes confer risk for gallstone disease in admixed Latinos with Mapuche Native American ancestry. <i>Scientific Reports</i> , 2019, 9, 772.	1.6	30
9	Spectrum of neurodevelopmental disease associated with the <i>GNAO1</i> guanosine triphosphate-binding region. <i>Epilepsia</i> , 2019, 60, 406-418.	2.6	53
10	Simple ClinVar: an interactive web server to explore and retrieve gene and disease variants aggregated in ClinVar database. <i>Nucleic Acids Research</i> , 2019, 47, W99-W105.	6.5	51
11	Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy. <i>New England Journal of Medicine</i> , 2019, 380, e24.	13.9	4
12	Variant Score Ranker—a web application for intuitive missense variant prioritization. <i>Bioinformatics</i> , 2019, 35, 4478-4479.	1.8	5
13	Whole Genome Sequence, Variant Discovery and Annotation in Mapuche-Huilliche Native South Americans. <i>Scientific Reports</i> , 2019, 9, 2132.	1.6	12
14	Spectrum of GABAA receptor variants in epilepsy. <i>Current Opinion in Neurology</i> , 2019, 32, 183-190.	1.8	59
15	Duplications at 19q13.33 in patients with neurodevelopmental disorders. <i>Neurology: Genetics</i> , 2018, 4, e210.	0.9	4
16	Heterogeneous contribution of microdeletions in the development of common generalised and focal epilepsies. <i>Journal of Medical Genetics</i> , 2017, 54, 598-606.	1.5	22
17	Early Transcriptional Changes Induced by Wnt/ β -Catenin Signaling in Hippocampal Neurons. <i>Neural Plasticity</i> , 2016, 2016, 1-13.	1.0	19
18	Overrepresentation of Glutamate Signaling in Alzheimer's Disease: Network-Based Pathway Enrichment Using Meta-Analysis of Genome-Wide Association Studies. <i>PLoS ONE</i> , 2014, 9, e95413.	1.1	52

#	ARTICLE	IF	CITATIONS
19	Wnt/β-Catenin Signaling in Alzheimer's Disease. CNS and Neurological Disorders - Drug Targets, 2014, 13, 745-754.	0.8	82
20	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