# Eduardo Prez-Palma

## List of Publications by Year in Descending Order



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|---|---|---|---|
| 20 papers | 324 citations | 11 h-index | 17 g-index |
| 23 ext. papers | 521 ext. citations | 9.2 avg, IF | 2.98 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 20 | Development and Validation of a Prediction Model for Early Diagnosis of -Related Epilepsies.. *Neurology*, **2022**, | 6.5 | 2 |
| 19 | Genome-Wide Analysis of Copy Number Variation in Latin American Parkinsonâs Disease Patients. *Movement Disorders*, **2021**, 36, 434-441 | 7 | 4 |
| 18 | Identification of pathogenic variant enriched regions across genes and gene families. *Genome Research*, **2020**, 30, 62-71 | 9.7 | 14 |
| 17 | Current knowledge of SLC6A1-related neurodevelopmental disorders. *Brain Communications*, **2020**, 2, fcaa170 | 4.5 | 11 |
| 16 | Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. *Science Translational Medicine*, **2020**, 12, | 17.5 | 27 |
| 15 | Copy number variants in lipid metabolism genes are associated with gallstones disease in men. *European Journal of Human Genetics*, **2020**, 28, 264-273 | 5.3 | 2 |
| 14 | Variants in ABCG8 and TRAF3 genes confer risk for gallstone disease in admixed Latinos with Mapuche Native American ancestry. *Scientific Reports*, **2019**, 9, 772 | 4.9 | 16 |
| 13 | Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphate-binding region. *Epilepsia*, **2019**, 60, 406-418 | 6.4 | 26 |
| 12 | 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 | 20.1 | 21 |
| 11 | Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy. *New England Journal of Medicine*, **2019**, 380, e24 | 59.2 | 4 |
| 10 | Variant Score Ranker-a web application for intuitive missense variant prioritization. *Bioinformatics*, **2019**, 35, 4478-4479 | 7.2 | 5 |
| 9 | Assessment of genetic variant burden in epilepsy-associated brain lesions. *European Journal of Human Genetics*, **2019**, 27, 1738-1744 | 5.3 | 4 |
| 8 | Whole Genome Sequence, Variant Discovery and Annotation in Mapuche-Huilliche Native South Americans. *Scientific Reports*, **2019**, 9, 2132 | 4.9 | 6 |
| 7 | Spectrum of GABAA receptor variants in epilepsy. *Current Opinion in Neurology*, **2019**, 32, 183-190 | 7.1 | 24 |
| 6 | Duplications at 19q13.33 in patients with neurodevelopmental disorders. *Neurology: Genetics*, **2018**, 4, e210 | 3.8 | 1 |
| 5 | Heterogeneous contribution of microdeletions in the development of common generalised and focal epilepsies. *Journal of Medical Genetics*, **2017**, 54, 598-606 | 5.8 | 14 |
| 4 | Early Transcriptional Changes Induced by Wnt/-Catenin Signaling in Hippocampal Neurons. *Neural Plasticity*, **2016**, 2016, 4672841 | 3.3 | 16 |

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|---|---|---|---|
| 3 | Overrepresentation of glutamate signaling in Alzheimerâs disease: network-based pathway enrichment using meta-analysis of genome-wide association studies. *PLoS ONE*, **2014**, 9, e95413 | 3.7 | 42 |
| 2 | Wnt/Ecatenin signaling in Alzheimerâs disease. *CNS and Neurological Disorders - Drug Targets*, **2014**, 13, 745-54 | 2.6 | 58 |
| 1 | A novel functional low-density lipoprotein receptor-related protein 6 gene alternative splice variant is associated with Alzheimerâs disease. *Neurobiology of Aging*, **2013**, 34, 1709.e9-18 | 5.6 | 27 |