Javier SimÃ³n-SÃ¡nchez

List of Publications by Year
in descending order

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52 papers	12,894 citations	101543 36 h-index	175258 52 g-index
56 all docs	56 docs citations	56 times ranked	16040 citing authors

#	Article	IF	Citations
1	Biallelic variants in TSPOAP1, encoding the active-zone protein RIMBP1, cause autosomal recessive dystonia. Journal of Clinical Investigation, 2021, 131, .	8.2	18
2	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. JAMA Neurology, 2021, 78, 464.	9.0	95
3	Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342.	12.8	44
4	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2019, 18, 1091-1102.	10.2	1,414
5	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Populationâ€Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. Movement Disorders, 2019, 34, 1851-1863.	3.9	47
6	A nonsynonymous mutation in PLCG2 reduces the risk of Alzheimerâ€™s disease, dementia with Lewy bodies and frontotemporal dementia, and increases the likelihood of longevity. Acta Neuropathologica, 2019, 138, 237-250.	7.7	87
7	Parkinson's disease age at onset genomeâ€wide association study: Defining heritability, genetic loci, and Î±â€synuclein mechanisms. Movement Disorders, 2019, 34, 866-875.	3.9	258
8	Genome-wide analyses as part of the international FTLD-TDP whole-genome sequencing consortium reveals novel disease risk factors and increases support for immune dysfunction in FTLD. Acta Neuropathologica, 2019, 137, 879-899.	7.7	90
9	The wide genetic landscape of clinical frontotemporal dementia: systematic combined sequencing of 121 consecutive subjects. Genetics in Medicine, 2018, 20, 240-249.	2.4	60
10	HPCA confirmed as a genetic cause of DYT2â€like dystonia phenotype. Movement Disorders, 2018, 33, 1354-1358.	3.9	31
11	No supportive evidence for TIA1 gene mutations in a European cohort of ALS-FTD spectrum patients. Neurobiology of Aging, 2018, 69, 293.e9-293.e11.	3.1	15
12	Discovery and functional prioritization of Parkinsonâ€™s disease candidate genes from large-scale whole exome sequencing. Genome Biology, 2017, 18, 22.	8.8	96
13	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. Neurobiology of Aging, 2017, 57, 247.e9-247.e13.	3.1	108
14	Lack of evidence for a role of genetic variation in TMEM230 in the risk for Parkinson's disease in the Caucasian population. Neurobiology of Aging, 2017, 50, 167.e11-167.e13.	3.1	24
15	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	7.6	323
16	Comprehensive promoter level expression quantitative trait loci analysis of the human frontal lobe. Genome Medicine, 2016, 8, 65.	8.2	20
17	C9orf72 is differentially expressed in the central nervous system and myeloid cells and consistently reduced in C9orf72, MAPT and GRN mutation carriers. Acta Neuropathologica Communications, 2016, 4, 37.	5.2	58
18	Deletions at 22q11.2 in idiopathic Parkinson's disease: a combined analysis of genome-wide association data. Lancet Neurology, The, 2016, 15, 585-596.	10.2	77

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19	A novel homozygous DJ1 mutation causes parkinsonism and ALS in a Turkish family. Parkinsonism and Related Disorders, 2016, 29, 117-120.	2.2	23
20	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. American Journal of Human Genetics, 2016, 98, 500-513.	6.2	333
21	Pilot whole-exome sequencing of a German early-onset Alzheimer's disease cohort reveals a substantial frequency of PSEN2 variants. Neurobiology of Aging, 2016, 37, 208.e11-208.e17.	3.1	38
22	Variation in PARK10 is not associated with risk and age at onset ofÂParkinson's disease in large clinical cohorts. Neurobiology of Aging, 2015, 36, 2907.e13-2907.e17.	3.1	5
23	Parkinson disease GWAS. Neurology, 2015, 84, 966-967.	1.1	7
24	Accurate prediction of a minimal region around a genetic association signal that contains the causal variant. European Journal of Human Genetics, 2014, 22, 238-242.	2.8	7
25	Analysis of Genome-Wide Association Studies of Alzheimer Disease and of Parkinson Disease to Determine If These 2 Diseases Share a Common Genetic Risk. JAMA Neurology, 2013, 70, 1268-76.	9.0	51
26	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. Human Molecular Genetics, 2013, 22, 1039-1049.	2.9	122
27	The Val158Met COMT polymorphism is a modifier of the age at onset in Parkinson's disease with a sexual dimorphism. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 666-673.	1.9	43
28	Fine-Mapping, Gene Expression and Splicing Analysis of the Disease Associated LRRK2 Locus. PLoS ONE, 2013, 8, e70724.	2.5	45
29	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	2.9	176
30	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. Brain, 2012, 135, 2875-2882.	7.6	114
31	Use of support vector machines for disease risk prediction in genome-wide association studies: Concerns and opportunities. Human Mutation, 2012, 33, 1708-1718.	2.5	42
32	The chromosome 9 ALS and FTD locus is probably derived from a single founder. Neurobiology of Aging, 2012, 33, 209.e3-209.e8.	3.1	115
33	The clinical and pathological phenotype of C9ORF72 hexanucleotide repeat expansions. Brain, 2012, 135, 723-735.	7.6	249
34	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Lancet Neurology, The, 2012, 11, 323-330.	10.2	1,039
35	Cooperative Genome-Wide Analysis Shows Increased Homozygosity in Early Onset Parkinson's Disease. PLoS ONE, 2012, 7, e28787.	2.5	21
36	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	8.1	3,833

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37	Genome-wide association study confirms extant PD risk loci among the Dutch. European Journal of Human Genetics, 2011, 19, 655-661.	2.8	164
38	Measures of Autozygosity in Decline: Globalization, Urbanization, and Its Implications for Medical Genetics. PLoS Genetics, 2009, 5, e1000415.	3.5	76
39	SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614.	5.3	257
40	Genome-wide association study reveals genetic risk underlying Parkinson's disease. Nature Genetics, 2009, 41, 1308-1312.	21.4	1,745
41	Variation at GRN 3â€²-UTR rs5848 Is Not Associated with a Risk of Frontotemporal Lobar Degeneration in Dutch Population. PLoS ONE, 2009, 4, e7494.	2.5	23
42	Structural genomic variation in ischemic stroke. Neurogenetics, 2008, 9, 101-108.	1.4	32
43	Genomewide SNP assay reveals mutations underlying Parkinson disease. Human Mutation, 2008, 29, 315-322.	2.5	46
44	Genome-wide association studies in neurological disorders. Lancet Neurology, The, 2008, 7, 1067-1072.	10.2	49
45	Sequencing analysis of OMI/HTRA2 shows previously reported pathogenic mutations in neurologically normal controls. Human Molecular Genetics, 2008, 17, 1988-1993.	2.9	106
46	Lack of replication of association between GIGYF2 variants and Parkinson disease. Human Molecular Genetics, 2008, 18, 341-346.	2.9	55
47	Deletion at ITPR1 Underlies Ataxia in Mice and Spinocerebellar Ataxia 15 in Humans. PLoS Genetics, 2007, 3, e108.	3.5	269
48	Genome-wide SNP assay reveals structural genomic variation, extended homozygosity and cell-line induced alterations in normal individuals. Human Molecular Genetics, 2007, 16, 1-14.	2.9	211
49	A genome-wide genotyping study in patients with ischaemic stroke: initial analysis and data release. Lancet Neurology, The, 2007, 6, 414-420.	10.2	175
50	LRRK2 is expressed in areas affected by Parkinson's disease in the adult mouse brain. European Journal of Neuroscience, 2006, 23, 659-666.	2.6	77
51	Genome-wide genotyping in Parkinson's disease and neurologically normal controls: first stage analysis and public release of data. Lancet Neurology, The, 2006, 5, 911-916.	10.2	360
52	Analysis of SCA-2 and SCA-3 repeats in Parkinsonism: Evidence of SCA-2 expansion in a family with autosomal dominant Parkinson's disease. Neuroscience Letters, 2005, 382, 191-194.	2.1	33