

Javier Simón-Sánchez

List of Publications by Year in descending order

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Version: 2024-02-01

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	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. <i>Neuron</i> , 2011, 72, 257-268.	8.1	3,833
2	Genome-wide association study reveals genetic risk underlying Parkinson's disease. <i>Nature Genetics</i> , 2009, 41, 1308-1312.	21.4	1,745
3	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. <i>Lancet Neurology</i> , The, 2019, 18, 1091-1102.	10.2	1,414
4	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. <i>Lancet Neurology</i> , The, 2012, 11, 323-330.	10.2	1,039
5	Genome-wide genotyping in Parkinson's disease and neurologically normal controls: first stage analysis and public release of data. <i>Lancet Neurology</i> , The, 2006, 5, 911-916.	10.2	360
6	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. <i>American Journal of Human Genetics</i> , 2016, 98, 500-513.	6.2	333
7	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. <i>Brain</i> , 2017, 140, 3191-3203.	7.6	323
8	Deletion at ITPR1 Underlies Ataxia in Mice and Spinocerebellar Ataxia 15 in Humans. <i>PLoS Genetics</i> , 2007, 3, e108.	3.5	269
9	Parkinson's disease age at onset genome-wide association study: Defining heritability, genetic loci, and LRRK2/synuclein mechanisms. <i>Movement Disorders</i> , 2019, 34, 866-875.	3.9	258
10	SNCA variants are associated with increased risk for multiple system atrophy. <i>Annals of Neurology</i> , 2009, 65, 610-614.	5.3	257
11	The clinical and pathological phenotype of C9ORF72 hexanucleotide repeat expansions. <i>Brain</i> , 2012, 135, 723-735.	7.6	249
12	Genome-wide SNP assay reveals structural genomic variation, extended homozygosity and cell-line induced alterations in normal individuals. <i>Human Molecular Genetics</i> , 2007, 16, 1-14.	2.9	211
13	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. <i>Human Molecular Genetics</i> , 2012, 21, 4996-5009.	2.9	176
14	A genome-wide genotyping study in patients with ischaemic stroke: initial analysis and data release. <i>Lancet Neurology</i> , The, 2007, 6, 414-420.	10.2	175
15	Genome-wide association study confirms extant PD risk loci among the Dutch. <i>European Journal of Human Genetics</i> , 2011, 19, 655-661.	2.8	164
16	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. <i>Human Molecular Genetics</i> , 2013, 22, 1039-1049.	2.9	122
17	The chromosome 9 ALS and FTD locus is probably derived from a single founder. <i>Neurobiology of Aging</i> , 2012, 33, 209.e3-209.e8.	3.1	115
18	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. <i>Brain</i> , 2012, 135, 2875-2882.	7.6	114

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19	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. <i>Neurobiology of Aging</i> , 2017, 57, 247.e9-247.e13.	3.1	108
20	Sequencing analysis of OMI/HTRA2 shows previously reported pathogenic mutations in neurologically normal controls. <i>Human Molecular Genetics</i> , 2008, 17, 1988-1993.	2.9	106
21	Discovery and functional prioritization of Parkinson's disease candidate genes from large-scale whole exome sequencing. <i>Genome Biology</i> , 2017, 18, 22.	8.8	96
22	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. <i>JAMA Neurology</i> , 2021, 78, 464.	9.0	95
23	Genome-wide analyses as part of the international FTL-D-TDP whole-genome sequencing consortium reveals novel disease risk factors and increases support for immune dysfunction in FTL-D. <i>Acta Neuropathologica</i> , 2019, 137, 879-899.	7.7	90
24	A nonsynonymous mutation in PLCG2 reduces the risk of Alzheimer's disease, dementia with Lewy bodies and frontotemporal dementia, and increases the likelihood of longevity. <i>Acta Neuropathologica</i> , 2019, 138, 237-250.	7.7	87
25	LRRK2 is expressed in areas affected by Parkinson's disease in the adult mouse brain. <i>European Journal of Neuroscience</i> , 2006, 23, 659-666.	2.6	77
26	Deletions at 22q11.2 in idiopathic Parkinson's disease: a combined analysis of genome-wide association data. <i>Lancet Neurology</i> , The, 2016, 15, 585-596.	10.2	77
27	Measures of Autozygosity in Decline: Globalization, Urbanization, and Its Implications for Medical Genetics. <i>PLoS Genetics</i> , 2009, 5, e1000415.	3.5	76
28	The wide genetic landscape of clinical frontotemporal dementia: systematic combined sequencing of 121 consecutive subjects. <i>Genetics in Medicine</i> , 2018, 20, 240-249.	2.4	60
29	C9orf72 is differentially expressed in the central nervous system and myeloid cells and consistently reduced in C9orf72, MAPT and GRN mutation carriers. <i>Acta Neuropathologica Communications</i> , 2016, 4, 37.	5.2	58
30	Lack of replication of association between GIGYF2 variants and Parkinson disease. <i>Human Molecular Genetics</i> , 2008, 18, 341-346.	2.9	55
31	Analysis of Genome-Wide Association Studies of Alzheimer Disease and of Parkinson Disease to Determine If These 2 Diseases Share a Common Genetic Risk. <i>JAMA Neurology</i> , 2013, 70, 1268-76.	9.0	51
32	Genome-wide association studies in neurological disorders. <i>Lancet Neurology</i> , The, 2008, 7, 1067-1072.	10.2	49
33	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. <i>Movement Disorders</i> , 2019, 34, 1851-1863.	3.9	47
34	Genomewide SNP assay reveals mutations underlying Parkinson disease. <i>Human Mutation</i> , 2008, 29, 315-322.	2.5	46
35	Fine-Mapping, Gene Expression and Splicing Analysis of the Disease Associated LRRK2 Locus. <i>PLoS ONE</i> , 2013, 8, e70724.	2.5	45
36	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome. <i>Nature Communications</i> , 2021, 12, 7342.	12.8	44

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37	The Val158Met COMT polymorphism is a modifier of the age at onset in Parkinson's disease with a sexual dimorphism. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 666-673.	1.9	43
38	Use of support vector machines for disease risk prediction in genome-wide association studies: Concerns and opportunities. <i>Human Mutation</i> , 2012, 33, 1708-1718.	2.5	42
39	Pilot whole-exome sequencing of a German early-onset Alzheimer's disease cohort reveals a substantial frequency of PSEN2 variants. <i>Neurobiology of Aging</i> , 2016, 37, 208.e11-208.e17.	3.1	38
40	Analysis of SCA-2 and SCA-3 repeats in Parkinsonism: Evidence of SCA-2 expansion in a family with autosomal dominant Parkinson's disease. <i>Neuroscience Letters</i> , 2005, 382, 191-194.	2.1	33
41	Structural genomic variation in ischemic stroke. <i>Neurogenetics</i> , 2008, 9, 101-108.	1.4	32
42	<i>HPCA</i> confirmed as a genetic cause of DYT2-like dystonia phenotype. <i>Movement Disorders</i> , 2018, 33, 1354-1358.	3.9	31
43	Lack of evidence for a role of genetic variation in TMEM230 in the risk for Parkinson's disease in the Caucasian population. <i>Neurobiology of Aging</i> , 2017, 50, 167.e11-167.e13.	3.1	24
44	A novel homozygous DJ1 mutation causes parkinsonism and ALS in a Turkish family. <i>Parkinsonism and Related Disorders</i> , 2016, 29, 117-120.	2.2	23
45	Variation at GRN 3' UTR rs5848 Is Not Associated with a Risk of Frontotemporal Lobar Degeneration in Dutch Population. <i>PLoS ONE</i> , 2009, 4, e7494.	2.5	23
46	Cooperative Genome-Wide Analysis Shows Increased Homozygosity in Early Onset Parkinson's Disease. <i>PLoS ONE</i> , 2012, 7, e28787.	2.5	21
47	Comprehensive promoter level expression quantitative trait loci analysis of the human frontal lobe. <i>Genome Medicine</i> , 2016, 8, 65.	8.2	20
48	Biallelic variants in TSPOAP1, encoding the active-zone protein RIMBP1, cause autosomal recessive dystonia. <i>Journal of Clinical Investigation</i> , 2021, 131, .	8.2	18
49	No supportive evidence for TIA1 gene mutations in a European cohort of ALS-FTD spectrum patients. <i>Neurobiology of Aging</i> , 2018, 69, 293.e9-293.e11.	3.1	15
50	Accurate prediction of a minimal region around a genetic association signal that contains the causal variant. <i>European Journal of Human Genetics</i> , 2014, 22, 238-242.	2.8	7
51	Parkinson disease GWAS. <i>Neurology</i> , 2015, 84, 966-967.	1.1	7
52	Variation in PARK10 is not associated with risk and age at onset of Parkinson's disease in large clinical cohorts. <i>Neurobiology of Aging</i> , 2015, 36, 2907.e13-2907.e17.	3.1	5