Jordi PÃ©rez Tur

List of Publications by Year
in descending order

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121 papers	13,784 citations	36303 51 h-index	21540 114 g-index
134 all docs	134 docs citations	134 times ranked	11765 citing authors

#	Article	IF	Citations
1	CRIDECO Anticholinergic Load Scale: An Updated Anticholinergic Burden Scale. Comparison with the ACB Scale in Spanish Individuals with Subjective Memory Complaints. Journal of Personalized Medicine, 2022, 12, 207.	2.5	11
2	New insights into the genetic etiology of Alzheimerâ€™s disease and related dementias. Nature Genetics, 2022, 54, 412-436.	21.4	700
3	Association of Rare APOE Missense Variants V236E and R251G With Risk of Alzheimer Disease. JAMA Neurology, 2022, 79, 652.	9.0	31
4	Epigenome-wide association study of COVID-19 severity with respiratory failure. EBioMedicine, 2021, 66, 103339.	6.1	90
5	Pharmacist-Physician Interprofessional Collaboration to Promote Early Detection of Cognitive Impairment: Increasing Diagnosis Rate. Frontiers in Pharmacology, 2021, 12, 579489.	3.5	11
6	Common variants in Alzheimerâ€™s disease and risk stratification by polygenic risk scores. Nature Communications, 2021, 12, 3417.	12.8	140
7	Presenilin-1 Mutations Are a Cause of Primary Lateral Sclerosis-Like Syndrome. Frontiers in Molecular Neuroscience, 2021, 14, 721047.	2.9	3
8	SARS-CoV-2 susceptibility and COVID-19 disease severity are associated with genetic variants affecting gene expression in a variety of tissues. Cell Reports, 2021, 37, 110020.	6.4	25
9	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	28.9	185
10	The Discovery of the Dardarin Gene 15â€‰Years Later: A Globalized Local History. Movement Disorders, 2020, 35, 708-708.	3.9	3
11	Metabolic alterations in plasma from patients with familial and idiopathic Parkinsonâ€™s disease. Aging, 2020, 12, 16690-16708.	3.1	32
12	The width of the third ventricle associates with cognition and behaviour in motor neuron disease. Acta Neurologica Scandinavica, 2019, 139, 118-127.	2.1	5
13	Brain signal intensity changes as biomarkers in amyotrophic lateral sclerosis. Acta Neurologica Scandinavica, 2018, 137, 262-271.	2.1	27
14	Epigenetic Study in Parkinsonâ€™s Disease: A Pilot Analysis of DNA Methylation in Candidate Genes in Brain. Cells, 2018, 7, 150.	4.1	25
15	PM20D1 is aÂquantitative trait locus associated with Alzheimerâ€™s disease. Nature Medicine, 2018, 24, 598-603.	30.7	73
16	Proyecto CRIDECO: Cribado de deterioro cognitivo en farmacia comunitaria a partir de la queja subjetiva de memoria. FarmacÃ‰uticos Comunitarios, 2018, 10, 20-26.	0.0	1
17	Clinical profile of motor neuron disease patients with lower urinary tract symptoms and neurogenic bladder. Journal of the Neurological Sciences, 2017, 378, 130-136.	0.6	17
18	Genetic and constitutional factors are major contributors to substantia nigra hyperechogenicity. Scientific Reports, 2017, 7, 7119.	3.3	6

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19	10. Genetics of Alzheimer's disease. , 2017, , 117-132.		0
20	Proteomic Alterations by Mutations Involved in Parkinson's Disease and Related Disorders. Current Protein and Peptide Science, 2017, 18, 654-655.	1.4	0
21	Other Proteins Involved in Parkinson's Disease and Related Disorders. Current Protein and Peptide Science, 2017, 18, 765-778.	1.4	5
22	Assessing the role of TUBA4A gene in frontotemporal degeneration. Neurobiology of Aging, 2016, 38, 215.e13-215.e14.	3.1	9
23	Clinical and neuroimaging characterization of two C9orf72-positive siblings with amyotrophic lateral sclerosis and schizophrenia. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 297-300.	1.7	3
24	Analysis of theCHCHD10gene in patients with frontotemporal dementia and amyotrophic lateral sclerosis from Spain. Brain, 2015, 138, e400-e400.	7.6	56
25	Parkin and <i>LRRK2</i>/Dardarin Mutations in Early Onset Parkinsonâ€™s Disease in the Basque Country (Spain). Journal of Behavioral and Brain Science, 2015, 05, 101-108.	0.5	0
26	Recurrence of carbamoyl phosphate synthetase 1 (CPS1) deficiency in Turkish patients: Characterization of a founder mutation by use of recombinant CPS1 from insect cells expression. Molecular Genetics and Metabolism, 2014, 113, 267-273.	1.1	8
27	Assessing the role of the TREM2 p.R47H variant as a risk factor for Alzheimer's disease and frontotemporal dementia. Neurobiology of Aging, 2014, 35, 444.e1-444.e4.	3.1	92
28	Structural and functional in silico analysis of LRRK2 missense substitutions. Molecular Biology Reports, 2014, 41, 2529-2542.	2.3	19
29	Analysis of theC9orf72Gene in Patients with Amyotrophic Lateral Sclerosis in Spain and Different Populations Worldwide. Human Mutation, 2013, 34, 79-82.	2.5	85
30	Benign hereditary chorea: Clinical features and long-term follow-up in a Spanish family. Parkinsonism and Related Disorders, 2013, 19, 394-396.	2.2	9
31	Rare Variants in Calcium Homeostasis Modulator 1 (CALHM1) Found in Early Onset Alzheimerâ€™s Disease Patients Alter Calcium Homeostasis. PLoS ONE, 2013, 8, e74203.	2.5	26
32	LRRK2 haplotypeâ€sharing analysis in Parkinson's disease reveals a novel p.S1761R mutation. Movement Disorders, 2012, 27, 146-150.	3.9	19
33	Transcriptional profile of Parkinson blood mononuclear cells with LRRK2 mutation. Neurobiology of Aging, 2011, 32, 1839-1848.	3.1	83
34	Phylogenetic and in silico structural analysis of the Parkinson diseaseâ€related kinase PINK1. Human Mutation, 2011, 32, 369-378.	2.5	32
35	Functional characterization of three singleâ€nucleotide polymorphisms present in the human APOE promoter sequence: Differential effects in neuronal cells and on DNAâ€“protein interactions. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 185-201.	1.7	32
36	Regional distribution of the leucine-rich glioma inactivated (LGI) gene family transcripts in the adult mouse brain. Brain Research, 2010, 1307, 177-194.	2.2	59

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37	LGI1 Is a Nogo Receptor 1 Ligand that Antagonizes Myelin-Based Growth Inhibition. Journal of Neuroscience, 2010, 30, 6607-6612.	3.6	71
38	Genetic screening of Alzheimer's disease genes in Iberian and African samples yields novel mutations in presenilins and APP. Neurobiology of Aging, 2010, 31, 725-731.	3.1	196
39	Homocysteine and cognitive impairment in Parkinson's disease: A biochemical, neuroimaging, and genetic study. Movement Disorders, 2009, 24, 1437-1444.	3.9	82
40	Mutations in Progranulin Gene: Clinical, Pathological, and Ribonucleic Acid Expression Findings. Biological Psychiatry, 2008, 63, 946-952.	1.3	62
41	Molecular Characterization of Putative Modulatory Factors in Two Spanish Families with A1555G Deafness. Audiology and Neuro-Otology, 2008, 13, 320-327.	1.3	2
42	Mechanistic insight into the dominant mode of the Parkinson's disease-associated G2019S LRRK2 mutation. Human Molecular Genetics, 2007, 16, 2031-2039.	2.9	132
43	ABCA1 polymorphisms and Alzheimer's disease. Neuroscience Letters, 2007, 416, 180-183.	2.1	29
44	Association between FOXP2 polymorphisms and schizophrenia with auditory hallucinations. Psychiatric Genetics, 2006, 16, 67-72.	1.1	116
45	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
46	The LGI1/Epitempin gene encodes two protein isoforms differentially expressed in human brain. Journal of Neurochemistry, 2006, 98, 985-991.	3.9	24
47	Genetic analysis of the LGI/Epitempin gene family in sporadic and familial lateral temporal lobe epilepsy. Epilepsy Research, 2006, 70, 118-126.	1.6	6
48	Parkinson's disease genetics: a complex disease comes to the clinic. Lancet Neurology, The, 2006, 5, 896-897.	10.2	4
49	Parkinson's disease due to the R1441G mutation in Dardarin: A founder effect in the basques. Movement Disorders, 2006, 21, 1954-1959.	3.9	84
50	The epilepsy gene LGI1 encodes a secreted glycoprotein that binds to the cell surface. Human Molecular Genetics, 2006, 15, 3436-3445.	2.9	86
51	Genetic Screening for Two LRRK2 Mutations in French Patients with Idiopathic Parkinson's Disease. Genetic Testing and Molecular Biomarkers, 2006, 10, 290-293.	1.7	15
52	Familial Parkinson's disease: Clinical and genetic analysis of four Basque families. Annals of Neurology, 2005, 57, 365-372.	5.3	56
53	Genetic linkage of autosomal dominant progressive supranuclear palsy to 1q31.1. Annals of Neurology, 2005, 57, 634-641.	5.3	48
54	A novel mutation (K317M) in the MAPT gene causes FTDP and motor neuron disease. Neurology, 2005, 64, 1578-1585.	1.1	97

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55	Genome-Wide Analysis of the Parkinsonism-Dementia Complex of Guam. Archives of Neurology, 2004, 61, 1889-97.	4.5	44
56	Cloning of the Gene Containing Mutations that Cause PARK8-Linked Parkinson's Disease. Neuron, 2004, 44, 595-600.	8.1	2,183
57	Mitochondrial polymporphisms in Parkinson's Disease. Neuroscience Letters, 2004, 370, 171-174.	2.1	37
58	Expression of the LGI1 gene product in astrocytic gliomas: downregulation with malignant progression. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2003, 443, 561-564.	2.8	28
59	A genomic screen of Spanish multiple sclerosis patients reveals multiple loci associated with the disease. Journal of Neuroimmunology, 2003, 143, 124-128.	2.3	35
60	Apolipoprotein E Pittsburgh variant is not associated with the risk of late-onset Alzheimer's disease in a Spanish population. American Journal of Medical Genetics Part A, 2003, 120B, 121-124.	2.4	5
61	Autosomal Dominant Lateral Temporal Epilepsy: Clinical Spectrum, New Epitempin Mutations, and Genetic Heterogeneity in Seven European Families. Epilepsia, 2003, 44, 1289-1297.	5.1	134
62	Mutations in the LGI1/Epitempin gene on 10q24 cause autosomal dominant lateral temporal epilepsy. Human Molecular Genetics, 2002, 11, 1119-1128.	2.9	289
63	Contribution of APOE promoter polymorphisms to Alzheimerâ€™s disease risk. Neurology, 2002, 59, 59-66.	1.1	102
64	Identification and characterization of a novel human brain-specific gene, homologous to S. scrofa tmp83.5 , in the chromosome 10q24 critical region for temporal lobe epilepsy and spastic paraplegia. Gene, 2002, 282, 87-94.	2.2	8
65	The novel EPTP repeat defines a superfamily of proteins implicated in epileptic disorders. Trends in Biochemical Sciences, 2002, 27, 441-444.	7.5	109
66	ApoE Ïµ3â€haplotype modulates Alzheimer betaâ€amyloid deposition in the brain. American Journal of Medical Genetics Part A, 2002, 114, 288-291.	2.4	28
67	Association of lipoprotein base Ser447Ter polymorphism with brain infarction: a population-based neuropathological study. Annals of Medicine, 2001, 33, 486-492.	3.8	36
68	A clinical and pathological study of motor neurone disease on Guam. Brain, 2001, 124, 2215-2222.	7.6	39
69	Prevalence of Alzheimerâ€™s disease in very elderly people. Neurology, 2001, 56, 1690-1696.	1.1	195
70	The Genetic and Pathological Classification of Familial Frontotemporal Dementia. Archives of Neurology, 2001, 58, 1813.	4.5	114
71	The Presenilin 1 C92S Mutation Increases AÎ² 42 Production. Biochemical and Biophysical Research Communications, 2000, 277, 261-263.	2.1	19
72	No association between TAU haplotype and Alzheimer's disease in population or clinic based series or in familial disease. Neuroscience Letters, 2000, 285, 147-149.	2.1	40

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73	Cardiovascular risk factors and Alzheimer's disease: a genetic association study in a population aged 85 or over. Neuroscience Letters, 2000, 292, 195-198.	2.1	48
74	Susceptibility Locus for Alzheimer's Disease on Chromosome 10. Science, 2000, 290, 2304-2305.	12.6	372
75	A chromosome 4p haplotype segregating with Parkinson's disease and postural tremor. Human Molecular Genetics, 1999, 8, 81-85.	2.9	229
76	A full genome scan for late onset Alzheimer's disease. Human Molecular Genetics, 1999, 8, 237-245.	2.9	334
77	Association of an Extended Haplotype in the Tau Gene with Progressive Supranuclear Palsy. Human Molecular Genetics, 1999, 8, 711-715.	2.9	749
78	Alzheimer disease PS-1 exon 9 deletion defined. Nature Medicine, 1999, 5, 1090-1090.	30.7	50
79	Î±-2 macroglobulin gene and Alzheimer disease. Nature Genetics, 1999, 22, 17-19.	21.4	91
80	Association between coding variability in the LRP gene and the risk of late-onset Alzheimer's disease. Human Genetics, 1999, 104, 432-434.	3.8	53
81	Mutation in thetau exon 10 splice site region in familial frontotemporal dementia. Annals of Neurology, 1999, 45, 270-271.	5.3	43
82	Genetic association of ?2-macroglobulin with Alzheimer's disease in a Finnish elderly population. Annals of Neurology, 1999, 46, 382-390.	5.3	98
83	Apolipoprotein E genotype does not affect the age of onset of dementia in families with defined tau mutations. Neuroscience Letters, 1999, 260, 193-195.	2.1	27
84	No association between the alpha-2 macroglobulin I1000V polymorphism and Alzheimer's disease. Neuroscience Letters, 1999, 262, 137-139.	2.1	48
85	A novel mutation in the apolipoprotein E gene (APOE*4 Pittsburgh) is associated with the risk of late-onset Alzheimer's disease. Neuroscience Letters, 1999, 263, 129-132.	2.1	32
86	Genetic variability at the amyloid-Î² precursor protein locus may contribute to the risk of late-onset Alzheimer's disease. Neuroscience Letters, 1999, 269, 67-70.	2.1	43
87	Alzheimer's disease presenilin-1 exon 9 deletion and L250S mutations sensitiZe SH-SY5Y neuroblastoma cells to hyperosmotic stress-induced apoptosis. Neuroscience, 1999, 95, 593-601.	2.3	39
88	Atypical parkinsonism in the French West Indies. Lancet, The, 1999, 354, 1474.	13.7	4
89	Neurodegenerative diseases of Guam: Analysis of TAU. Neurology, 1999, 53, 411-411.	1.1	40
90	The tau gene A0 allele and progressive supranuclear palsy. Neurology, 1999, 53, 1219-1219.	1.1	45

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91	Genetic dissection of Alzheimer's disease and related dementias: amyloid and its relationship to tau. Nature Neuroscience, 1998, 1, 355-358.	14.8	310
92	A variant of Alzheimer's disease with spastic paraparesis and unusual plaques due to deletion of exon 9 of presenilin 1. Nature Medicine, 1998, 4, 452-455.	30.7	347
93	Increased AÎ²42(43) from cell lines expressing presenilin 1 mutations. Annals of Neurology, 1998, 43, 256-258.	5.3	117
94	Low frequency of ?-synuclein mutations in familial Parkinson's disease. Annals of Neurology, 1998, 43, 394-397.	5.3	153
95	Exclusion of genetic linkage to 4q21-23 and 17q21 in a family with lewy body parkinsonism. , 1998, 81, 166-171.		5
96	Cloning and characterization of the presenilin-2 gene promoter. Molecular Brain Research, 1998, 56, 57-65.	2.3	19
97	Pronounced impact of Th1/E47cs mutation compared with -491 AT mutation on neural APOE gene expression and risk of developing Alzheimer's disease. Human Molecular Genetics, 1998, 7, 1511-1516.	2.9	127
98	Genetic Studies on Chromosome 12 in Late-Onset Alzheimer Disease. JAMA - Journal of the American Medical Association, 1998, 280, 619.	7.4	95
99	Genetics of Alzheimer's disease. Essays in Biochemistry, 1998, 33, 117-131.	4.7	76
100	Distortion of Allelic Expression of Apolipoprotein E in Alzheimer's Disease. Human Molecular Genetics, 1997, 6, 2151-2154.	2.9	86
101	A New Pathogenic Mutation in the APP Gene (I716V) Increases the Relative Proportion of AÂ42(43). Human Molecular Genetics, 1997, 6, 2087-2089.	2.9	209
102	Association between the low density lipoprotein receptor-related protein (LRP) and Alzheimer's disease. Neuroscience Letters, 1997, 227, 68-70.	2.1	92
103	Early-onset Alzheimer's disease with a presenilin-1 mutation at the site corresponding to the volga German presenilin-2 mutation. Annals of Neurology, 1997, 42, 124-128.	5.3	40
104	Apolipoprotein E and Alzheimer disease: genotype-specific risks by age and sex. American Journal of Human Genetics, 1997, 60, 439-46.	6.2	100
105	A Further Presenilin 1 Mutation in the Exon 8 Cluster in Familial Alzheimer's Disease. Experimental Neurology, 1996, 5, 207-212.	1.7	27
106	Presenilin-1 polymorphism and Alzheimer's disease. Lancet, The, 1996, 347, 1560-1561.	13.7	67
107	Analysis of the APOE alleles impact in Down's syndrome. Neuroscience Letters, 1996, 220, 57-60.	2.1	21
108	Structure and alternative splicing of the Presenilin-2 gene. NeuroReport, 1996, 7, 1680-1684.	1.2	50

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109	Complete analysis of the presenilin 1 gene in early onset AlzheimerÊ¼s disease. NeuroReport, 1996, 7, 801-805.	1.2	150
110	Apolipoprotein E in Guamanian amyotrophic lateral sclerosis/ parkinsonism-dementia complex: genotype analysis and relationships to neuropathological changes. Acta Neuropathologica, 1996, 91, 247-253.	7.7	23
111	Increased amyloid-Î²42(43) in brains of mice expressing mutant presenilin 1. Nature, 1996, 383, 710-713.	27.8	1,480
112	Evidence for apolipoprotein E Îµ4 association in early-onset Alzheimer's patients with late-onset relatives. American Journal of Medical Genetics Part A, 1995, 60, 550-553.	2.4	28
113	The structure of the presenilin 1 (S182) gene and identification of six novel mutations in early onset AD families. Nature Genetics, 1995, 11, 219-222.	21.4	461
114	Alzheimer's disease: AÃŸ or ApoE amyloidosis?. Lancet, The, 1995, 346, 59.	13.7	1
115	A mutation in AlzheimerÊ¼s disease destroying a splice acceptor site in the presenilin-1 gene. NeuroReport, 1995, 7, 297-301.	1.2	262
116	A mutation in Alzheimer's disease destroying a splice acceptor site in the presenilin-1 gene. NeuroReport, 1995, 7, 297-301.	1.2	50
117	A mutation in AlzheimerÊ¼s disease destroying a splice acceptor site in the presenilin-1 gene. NeuroReport, 1995, 7, 297-301.	1.2	5
118	Apolipoprotein E, Ã›4 allele as a major risk factor for sporadic early and late-onset forms of Alzheimer's disease: analysis of the 19q13.2 chromosomal region. Human Molecular Genetics, 1994, 3, 569-574.	2.9	400
119	Solubilization of asymmetric acetylcholinesterase by polyanions. Neuroscience Letters, 1991, 126, 172-174.	2.1	5
120	Chondroitinases release acetylcholinesterase from chick skeletal muscle. FEBS Letters, 1991, 286, 25-27.	2.8	8
121	Two types of asymmetric acetylcholinesterase in chick hindlimb muscle: Developmental profiles,in Vivo and in cell culture, and recovery after inactivation. Cellular and Molecular Neurobiology, 1991, 11, 191-201.	3.3	3