Mario Ezquerra Trabalon

List of Publications by Year
in descending order

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109 papers	5,593 citations	117571 34 h-index	95218 68 g-index
111 all docs	111 docs citations	111 times ranked	7310 citing authors

#	Article	IF	Citations
1	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.	4.9	1,414
2	Diseaseâ€specific phenotypes in dopamine neurons from human iPSâ€based models of genetic and sporadic Parkinson's disease. EMBO Molecular Medicine, 2012, 4, 380-395.	3.3	501
3	Cerebrospinal tau, phosphoâ€tau, and betaâ€amyloid and neuropsychological functions in Parkinson's disease. Movement Disorders, 2009, 24, 2203-2210.	2.2	163
4	Significant association between the tau gene A0/A0 genotype and Parkinson's disease. Annals of Neurology, 2000, 47, 242-245.	2.8	129
5	LRRK2 Mutations in Spanish Patients With Parkinson Disease. Archives of Neurology, 2006, 63, 377.	4.9	127
6	Identification of blood serum microâ€RNAs associated with idiopathic and LRRK2 Parkinson's disease. Journal of Neuroscience Research, 2014, 92, 1071-1077.	1.3	122
7	Plasma miR-34a-5p and miR-545-3p as Early Biomarkers of Alzheimerâ€™s Disease: Potential and Limitations. Molecular Neurobiology, 2017, 54, 5550-5562.	1.9	119
8	Aberrant epigenome in <scp>iPSC</scp> â€derived dopaminergic neurons from Parkinson's disease patients. EMBO Molecular Medicine, 2015, 7, 1529-1546.	3.3	117
9	Frequency of Mutations in the Presenilin and Amyloid Precursor Protein Genes in Early-Onset Alzheimer Disease in Spain. Archives of Neurology, 2002, 59, 1759.	4.9	103
10	Mitochondria function associated genes contribute to Parkinsonâ€™s Disease risk and later age at onset. Npj Parkinson's Disease, 2019, 5, 8.	2.5	95
11	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. JAMA Neurology, 2021, 78, 464.	4.5	95
12	G2019S LRRK2 mutation causing Parkinson's disease without Lewy bodies. Journal of Neurology, Neurosurgery and Psychiatry, 2007, 78, 626-628.	0.9	90
13	Significant Changes in the Tau A0 and A3 Alleles in Progressive Supranuclear Palsy and Improved Genotyping by Silver Detection. Archives of Neurology, 1998, 55, 1122.	4.9	85
14	Moving beyond neurons: the role of cell type-specific gene regulation in Parkinsonâ€™s disease heritability. Npj Parkinson's Disease, 2019, 5, 6.	2.5	83
15	Nonmotor Symptoms in LRRK2 G2019S Associated Parkinsonâ€™s Disease. PLoS ONE, 2014, 9, e108982.	1.1	79
16	Novel haplotypes in 17q21 are associated with progressive supranuclear palsy. Annals of Neurology, 2004, 56, 249-258.	2.8	71
17	Primary progressive aphasia as the initial manifestation of corticobasal degeneration and unusual tauopathies. Acta Neuropathologica, 2003, 106, 419-435.	3.9	67
18	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. Movement Disorders, 2019, 34, 460-468.	2.2	66

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19	Further extension of the H1 haplotype associated with progressive supranuclear palsy. Movement Disorders, 2002, 17, 550-556.	2.2	61
20	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. Movement Disorders, 2020, 35, 774-780.	2.2	57
21	Identification of a novel polymorphism in the promoter region of the tau gene highly associated to progressive supranuclear palsy in humans. Neuroscience Letters, 1999, 275, 183-186.	1.0	56
22	Rapidly progressive diffuse Lewy body disease. Movement Disorders, 2011, 26, 1316-1323.	2.2	56
23	MicroRNA alterations in iPSC-derived dopaminergic neurons from Parkinson disease patients. Neurobiology of Aging, 2018, 69, 283-291.	1.5	55
24	HLA and microtubule-associated protein tau H1 haplotype associations in anti-IgLON5 disease. Neurology: Neuroimmunology and NeuroInflammation, 2019, 6, .	3.1	55
25	Lack of association of APOE and tau polymorphisms with dementia in Parkinsonâ€™s disease. Neuroscience Letters, 2008, 448, 20-23.	1.0	54
26	The <scp>MC1R</scp> melanoma risk variant p.<scp>R160W</scp> is associated with <scp>P</scp>arkinson disease. Annals of Neurology, 2015, 77, 889-894.	2.8	52
27	A novel mutation (V89L) in the presenilin 1 gene in a family with early onset Alzheimer's disease and marked behavioural disturbances. Journal of Neurology, Neurosurgery and Psychiatry, 2002, 72, 266-269.	0.9	51
28	Micro<scp>RNA</scp> association with synucleinopathy conversion in rapid eye movement behavior disorder. Annals of Neurology, 2015, 77, 895-901.	2.8	50
29	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.	2.2	47
30	A Novel Mutation in the PSEN2 Gene (T430M) Associated With Variable Expression in a Family With Early-Onset Alzheimer Disease. Archives of Neurology, 2003, 60, 1149.	4.9	46
31	CSF microRNA Profiling in Alzheimerâ€™s Disease: a Screening and Validation Study. Molecular Neurobiology, 2017, 54, 6647-6654.	1.9	45
32	Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342.	5.8	44
33	Clinicopathological and genetic correlates of frontotemporal lobar degeneration and corticobasal degeneration. Journal of Neurology, 2008, 255, 488-494.	1.8	40
34	Parkin loss of function contributes to RTP801 elevation and neurodegeneration in Parkinsonâ€™s disease. Cell Death and Disease, 2014, 5, e1364-e1364.	2.7	40
35	The Small GTPase RAC1/CED-10 Is Essential in Maintaining Dopaminergic Neuron Function and Survival Against Î±-Synuclein-Induced Toxicity. Molecular Neurobiology, 2018, 55, 7533-7552.	1.9	40
36	Screening for the LRRK2 G2019S and codon-1441 mutations in a pathological series of parkinsonian syndromes and frontotemporal lobar degeneration. Journal of the Neurological Sciences, 2008, 270, 94-98.	0.3	35

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37	Age at Onset in LRRK2-Associated PD is Modified by SNCA Variants. Journal of Molecular Neuroscience, 2012, 48, 245-247.	1.1	34
38	Microarray expression analysis in idiopathic and LRRK2-associated Parkinson's disease. Neurobiology of Disease, 2012, 45, 462-468.	2.1	33
39	Different MAPT haplotypes are associated with Parkinson's disease and progressive supranuclear palsy. Neurobiology of Aging, 2011, 32, 547.e11-547.e16.	1.5	32
40	Discovering the 3â€² UTR-mediated regulation of alpha-synuclein. Nucleic Acids Research, 2017, 45, 12888-12903.	6.5	32
41	Cerebrospinal fluid levels of coenzyme Q10 are reduced in multiple system atrophy. Parkinsonism and Related Disorders, 2018, 46, 16-23.	1.1	32
42	123I-MIBG cardiac uptake and smell identification in parkinsonian patients with LRRK2 mutations. Journal of Neurology, 2011, 258, 1126-1132.	1.8	31
43	A Novel Intronic Mutation in the DDP1 Gene in a Family With X-linked Dystonia-Deafness Syndrome. Archives of Neurology, 2005, 62, 306.	4.9	30
44	Brain transcriptomic profiling in idiopathic and LRRK2-associated Parkinson's disease. Brain Research, 2012, 1466, 152-157.	1.1	30
45	Absence of LRRK2 mutations in a cohort of patients with idiopathic REM sleep behavior disorder. Neurology, 2016, 86, 1072-1073.	1.5	30
46	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42.	2.8	29
47	TREM2 R47H variant and risk of essential tremor: A cross-sectional international multicenter study. Parkinsonism and Related Disorders, 2015, 21, 306-309.	1.1	28
48	Accumulation of mitochondrial 7S DNA in idiopathic and LRRK2 associated Parkinson's disease. EBioMedicine, 2019, 48, 554-567.	2.7	28
49	The genotype 2/2 of the presenilin-1 polymorphism is decreased in Spanish early-onset Alzheimer's disease. Neuroscience Letters, 1997, 227, 201-204.	1.0	26
50	Î±-Antichymotrypsin gene polymorphism and risk for Alzheimer's disease in the Spanish population. Neuroscience Letters, 1998, 240, 107-109.	1.0	26
51	Cerebrospinal fluid cytokines in multiple system atrophy: A cross-sectional Catalan MSA registry study. Parkinsonism and Related Disorders, 2019, 65, 3-12.	1.1	26
52	Î±â€synuclein (SNCA) but not dynamin 3 (DNM3) influences age at onset of leucineâ€rich repeat kinase 2 (LRRK2) Parkinson's disease in Spain. Movement Disorders, 2018, 33, 637-641.	2.2	25
53	Mitochondrial and autophagic alterations in skin fibroblasts from Parkinson disease patients with Parkin mutations. Aging, 2019, 11, 3750-3767.	1.4	25
54	5â€²-upstream variants of CRHR1 and MAPT genes associated with age at onset in progressive supranuclear palsy and cortical basal degeneration. Neurobiology of Disease, 2009, 33, 164-170.	2.1	24

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55	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.	1.5	24
56	Analysis of the coding and the 5â€² flanking regions of the Î±-synuclein gene in patients with Parkinson's disease. Movement Disorders, 2001, 16, 1115-1119.	2.2	23
57	A novel mutation in the PSEN1 gene (L286P) associated with familial earlyâ€onset dementia of Alzheimer type and lobar haematomas. European Journal of Neurology, 2007, 14, 1409-1412.	1.7	23
58	Fused in Sarcoma (FUS) gene mutations are not a frequent cause of essential tremor in Europeans. Neurobiology of Aging, 2013, 34, 2441.e9-2441.e11.	1.5	22
59	Exhaustion of mitochondrial and autophagic reserve may contribute to the development of LRRK2 G2019S -Parkinsonâ€™s disease. Journal of Translational Medicine, 2018, 16, 160.	1.8	22
60	SNCA and mTOR Pathway Single Nucleotide Polymorphisms Interact to Modulate the Age at Onset of Parkinson's Disease. Movement Disorders, 2019, 34, 1333-1344.	2.2	21
61	Mendelian Randomisation Study of Smoking, Alcohol, and Coffee Drinking in Relation to Parkinsonâ€™s Disease. Journal of Parkinson's Disease, 2022, 12, 267-282.	1.5	21
62	An exome study of Parkinsonâ€™s disease in Sardinia, a Mediterranean genetic isolate. Neurogenetics, 2015, 16, 55-64.	0.7	20
63	Peripheral insulin and amylin levels in Parkinson's disease. Parkinsonism and Related Disorders, 2020, 79, 91-96.	1.1	20
64	A Novel Presenilin 1 Mutation (Leu166Arg) Associated With Early-Onset Alzheimer Disease. Archives of Neurology, 2000, 57, 485.	4.9	19
65	LRRK2 haplotypeâ€sharing analysis in Parkinson's disease reveals a novel p.S1761R mutation. Movement Disorders, 2012, 27, 146-150.	2.2	19
66	High cerebrospinal tau levels are associated with the rs242557 tau gene variant and low cerebrospinal Î²-amyloid in Parkinson disease. Neuroscience Letters, 2011, 487, 169-173.	1.0	18
67	Phenotypic Variability Within the Inclusion Body Spectrum of Basophilic Inclusion Body Disease and Neuronal Intermediate Filament Inclusion Disease in Frontotemporal Lobar Degenerations With FUS-Positive Inclusions. Journal of Neuropathology and Experimental Neurology, 2012, 71, 795-805.	0.9	18
68	Parkinsonism, dysautonomia, REM behaviour disorder and visual hallucinations mimicking synucleinopathy in a patient with progressive supranuclear palsy. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 578-579.	0.9	17
69	LINGO1 gene analysis in Parkinson's disease phenotypes. Movement Disorders, 2011, 26, 722-727.	2.2	17
70	MTOR Pathway-Based Discovery of Genetic Susceptibility to L-DOPA-Induced Dyskinesia in Parkinsonâ€™s Disease Patients. Molecular Neurobiology, 2019, 56, 2092-2100.	1.9	17
71	Alzheimer disease is not associated with polymorphisms in the angiotensinogen and renin genes. American Journal of Medical Genetics Part A, 2001, 105, 761-764.	2.4	16
72	Ubiquitin-negative mini-pick-like bodies in the dentate gyrus in p301l tauopathy. Journal of Alzheimer's Disease, 2004, 5, 445-454.	1.2	16

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73	A novel MAPT mutation (P301T) associated with familial frontotemporal dementia. European Journal of Neurology, 2007, 14, e9-10.	1.7	16
74	Whole-genome DNA hyper-methylation in iPSC-derived dopaminergic neurons from Parkinsonâ€™s disease patients. Clinical Epigenetics, 2019, 11, 108.	1.8	16
75	Analysis of DNM3 and VAMP4 as genetic modifiers of LRRK2 Parkinsonâ€™s disease. Neurobiology of Aging, 2021, 97, 148.e17-148.e24.	1.5	16
76	LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032.	4.9	15
77	<scp>MicroRNA</scp> Deregulation in Blood Serum Identifies Multiple System Atrophy Altered Pathways. Movement Disorders, 2020, 35, 1873-1879.	2.2	15
78	Dairy Intake and Parkinson's Disease: A Mendelian Randomization Study. Movement Disorders, 2022, 37, 857-864.	2.2	15
79	Increased cerebral activity in Parkinson?s disease patients carrying the DRD2 TaqIA A1 allele during a demanding motor task: a compensatory mechanism?. Genes, Brain and Behavior, 2007, 6, 588-592.	1.1	14
80	CCAAT/enhancer binding protein Î´ is a transcriptional repressor of Î±-synuclein. Cell Death and Differentiation, 2020, 27, 509-524.	5.0	14
81	Late-onset frontotemporal dementia associated with a novel PGRN mutation. Journal of Neural Transmission, 2007, 114, 1051-1054.	1.4	13
82	Epigenetic Research of Neurodegenerative Disorders Using Patient iPSC-Based Models. Stem Cells International, 2016, 2016, 1-16.	1.2	13
83	Transcriptional alterations in skin fibroblasts from Parkinson's disease patients with parkin mutations. Neurobiology of Aging, 2018, 65, 206-216.	1.5	13
84	Detection of the presenilin 1 gene mutation (M139T) in early-onset familial Alzheimer disease in Spain. Neuroscience Letters, 2001, 299, 239-241.	1.0	12
85	Lack of interaction of SNCA and MAPT genotypes in Parkinson's disease. European Journal of Neurology, 2011, 18, e32-e32.	1.7	12
86	Apolipoprotein E Ïµ4 alleles and meiotic origin of non-disjunction in Down syndrome children and in their corresponding fathers and mothers. Neuroscience Letters, 1998, 248, 1-4.	1.0	11
87	G2019S LRRK2 mutation causing Parkinson's disease without Lewy bodies. BMJ Case Reports, 2009, 2009, bcr0820080632-bcr0820080632.	0.2	11
88	Association study of the G258S transferrin gene polymorphism and Parkinson's disease in the Spanish population. Journal of Neurology, 2005, 252, 1269-1270.	1.8	10
89	Tau and saitohin gene expression pattern in progressive supranuclear palsy. Brain Research, 2007, 1145, 168-176.	1.1	10
90	MAPT association with REM sleep behavior disorder. Neurology: Genetics, 2017, 3, e131.	0.9	10

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91	Low apolipoprotein E epsilon4 allele frequency in the population of Catalonia (Spain) determined by PCR-RFLP and Laser fluorescent sequencer. European Journal of Epidemiology, 1997, 13, 841-843.	2.5	9
92	Differential Phosphoâ€Signatures in Blood Cells Identify <scp>LRRK2 G2019S</scp> Carriers in Parkinson's Disease. Movement Disorders, 2022, 37, 1004-1015.	2.2	9
93	Disrupted Mitochondrial and Metabolic Plasticity Underlie Comorbidity between Age-Related and Degenerative Disorders as Parkinson Disease and Type 2 Diabetes Mellitus. Antioxidants, 2020, 9, 1063.	2.2	8
94	Altered expression of the immunoregulatory ligand-receptor pair CD200-CD200R1 in the brain of Parkinsonâ€™s disease patients. Npj Parkinson's Disease, 2022, 8, 27.	2.5	8
95	Transcriptomic differences in MSA clinical variants. Scientific Reports, 2020, 10, 10310.	1.6	7
96	Brain tau expression and correlation with the H1/H1 tau genotype in frontotemporal lobar degeneration patients. Journal of Neural Transmission, 2007, 114, 1585-1588.	1.4	6
97	Regulatory rare variants of the dopaminergic gene ANKK1 as potential risk factors for Parkinsonâ€™s disease. Scientific Reports, 2021, 11, 9879.	1.6	4
98	The Interaction between <scp>HLAâ€ÐRB1</scp> and Smoking in Parkinson's Disease Revisited. Movement Disorders, 2022, 37, 1929-1937.	2.2	4
99	Reply. Annals of Neurology, 2016, 79, 161-163.	2.8	3
100	Parkinsonâ€™s disease as a systemic pathology. Aging, 2019, 11, 1081-1082.	1.4	3
101	No evidence of CRHR1 gene involvement in progressive supranuclear palsy. Neuroscience Letters, 2006, 409, 61-64.	1.0	2
102	Smoking is associated with age at disease onset in Parkinson's disease. Parkinsonism and Related Disorders, 2022, 97, 79-83.	1.1	2
103	Mutational study of the nuclear factor kappa B inducing kinase gene in patients with progressive supranuclear palsy. Neuroscience Letters, 2003, 340, 158-160.	1.0	1
104	Identifying the genetic components underlying the pathophysiology of movement disorders. The Application of Clinical Genetics, 2011, 4, 81.	1.4	1
105	Reply. Annals of Neurology, 2015, 78, 153-154.	2.8	1
106	Target-enriched sequencing of chromosome 17q21.31 in sporadic tauopathies reveals no candidate variants. Neurobiology of Aging, 2018, 66, 177.e7-177.e10.	1.5	1
107	Transcriptome analysis in LRRK2 and idiopathic Parkinsonâ€™s disease at different glucose levels. Npj Parkinson's Disease, 2021, 7, 109.	2.5	1
108	Reply: Rapidly progressing diffuse Lewy body disease. Movement Disorders, 2011, 26, 2585-2585.	2.2	0

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109	Reply. Annals of Neurology, 2016, 79, 868-868.	2.8	0