

# 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	Mendelian Randomisation Study of Smoking, Alcohol, and Coffee Drinking in Relation to Parkinson's Disease. <i>Journal of Parkinson's Disease</i> , 2022, 12, 267-282.	1.5	21
2	Differential Phospho-Signatures in Blood Cells Identify LRRK2 G2019S Carriers in Parkinson's Disease. <i>Movement Disorders</i> , 2022, 37, 1004-1015.	2.2	9
3	Dairy Intake and Parkinson's Disease: A Mendelian Randomization Study. <i>Movement Disorders</i> , 2022, 37, 857-864.	2.2	15
4	Altered expression of the immunoregulatory ligand-receptor pair CD200-CD200R1 in the brain of Parkinson's disease patients. <i>Npj Parkinson's Disease</i> , 2022, 8, 27.	2.5	8
5	Smoking is associated with age at disease onset in Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2022, 97, 79-83.	1.1	2
6	The Interaction between HLA-DRB1 and Smoking in Parkinson's Disease Revisited. <i>Movement Disorders</i> , 2022, 37, 1929-1937.	2.2	4
7	Analysis of DNMT3 and VAMP4 as genetic modifiers of LRRK2 Parkinson's disease. <i>Neurobiology of Aging</i> , 2021, 97, 148.e17-148.e24.	1.5	16
8	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. <i>JAMA Neurology</i> , 2021, 78, 464.	4.5	95
9	Regulatory rare variants of the dopaminergic gene ANKK1 as potential risk factors for Parkinson's disease. <i>Scientific Reports</i> , 2021, 11, 9879.	1.6	4
10	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. <i>Annals of Neurology</i> , 2021, 90, 35-42.	2.8	29
11	Transcriptome analysis in LRRK2 and idiopathic Parkinson's disease at different glucose levels. <i>Npj Parkinson's Disease</i> , 2021, 7, 109.	2.5	1
12	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome. <i>Nature Communications</i> , 2021, 12, 7342.	5.8	44
13	CCAAT/enhancer binding protein $\beta$ is a transcriptional repressor of $\alpha$ -synuclein. <i>Cell Death and Differentiation</i> , 2020, 27, 509-524.	5.0	14
14	MicroRNA Deregulation in Blood Serum Identifies Multiple System Atrophy Altered Pathways. <i>Movement Disorders</i> , 2020, 35, 1873-1879.	2.2	15
15	Peripheral insulin and amylin levels in Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2020, 79, 91-96.	1.1	20
16	Disrupted Mitochondrial and Metabolic Plasticity Underlie Comorbidity between Age-Related and Degenerative Disorders as Parkinson Disease and Type 2 Diabetes Mellitus. <i>Antioxidants</i> , 2020, 9, 1063.	2.2	8
17	Transcriptomic differences in MSA clinical variants. <i>Scientific Reports</i> , 2020, 10, 10310.	1.6	7
18	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. <i>Movement Disorders</i> , 2020, 35, 774-780.	2.2	57

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19	MTOR Pathway-Based Discovery of Genetic Susceptibility to L-DOPA-Induced Dyskinesia in Parkinson's Disease Patients. <i>Molecular Neurobiology</i> , 2019, 56, 2092-2100.	1.9	17
20	Whole-genome DNA hyper-methylation in iPSC-derived dopaminergic neurons from Parkinson's disease patients. <i>Clinical Epigenetics</i> , 2019, 11, 108.	1.8	16
21	Accumulation of mitochondrial 7S DNA in idiopathic and LRRK2 associated Parkinson's disease. <i>EBioMedicine</i> , 2019, 48, 554-567.	2.7	28
22	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.	4.9	1,414
23	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.	2.2	47
24	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. <i>Movement Disorders</i> , 2019, 34, 460-468.	2.2	66
25	SNCA and mTOR Pathway Single Nucleotide Polymorphisms Interact to Modulate the Age at Onset of Parkinson's Disease. <i>Movement Disorders</i> , 2019, 34, 1333-1344.	2.2	21
26	Cerebrospinal fluid cytokines in multiple system atrophy: A cross-sectional Catalan MSA registry study. <i>Parkinsonism and Related Disorders</i> , 2019, 65, 3-12.	1.1	26
27	Mitochondria function associated genes contribute to Parkinson's Disease risk and later age at onset. <i>Npj Parkinson's Disease</i> , 2019, 5, 8.	2.5	95
28	Moving beyond neurons: the role of cell type-specific gene regulation in Parkinson's disease heritability. <i>Npj Parkinson's Disease</i> , 2019, 5, 6.	2.5	83
29	HLA and microtubule-associated protein tau H1 haplotype associations in anti-IgLON5 disease. <i>Neurology: Neuroimmunology and NeuroInflammation</i> , 2019, 6, .	3.1	55
30	Parkinson's disease as a systemic pathology. <i>Aging</i> , 2019, 11, 1081-1082.	1.4	3
31	Mitochondrial and autophagic alterations in skin fibroblasts from Parkinson disease patients with Parkin mutations. <i>Aging</i> , 2019, 11, 3750-3767.	1.4	25
32	±-Synuclein ( <i>SNCA</i> ) but not dynamin 3 ( <i>DNM3</i> ) influences age at onset of leucine-rich repeat kinase 2 ( <i>LRRK2</i> ) Parkinson's disease in Spain. <i>Movement Disorders</i> , 2018, 33, 637-641.	2.2	25
33	Target-enriched sequencing of chromosome 17q21.31 in sporadic tauopathies reveals no candidate variants. <i>Neurobiology of Aging</i> , 2018, 66, 177.e7-177.e10.	1.5	1
34	Transcriptional alterations in skin fibroblasts from Parkinson's disease patients with parkin mutations. <i>Neurobiology of Aging</i> , 2018, 65, 206-216.	1.5	13
35	The Small GTPase RAC1/CED-10 Is Essential in Maintaining Dopaminergic Neuron Function and Survival Against ±-Synuclein-Induced Toxicity. <i>Molecular Neurobiology</i> , 2018, 55, 7533-7552.	1.9	40
36	Cerebrospinal fluid levels of coenzyme Q10 are reduced in multiple system atrophy. <i>Parkinsonism and Related Disorders</i> , 2018, 46, 16-23.	1.1	32

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37	LRP10 in $\alpha$ -synucleinopathies. <i>Lancet Neurology</i> , The, 2018, 17, 1032.	4.9	15
38	Exhaustion of mitochondrial and autophagic reserve may contribute to the development of LRRK2 G2019S -Parkinson's disease. <i>Journal of Translational Medicine</i> , 2018, 16, 160.	1.8	22
39	MicroRNA alterations in iPSC-derived dopaminergic neurons from Parkinson disease patients. <i>Neurobiology of Aging</i> , 2018, 69, 283-291.	1.5	55
40	<i>MAPT</i> association with REM sleep behavior disorder. <i>Neurology: Genetics</i> , 2017, 3, e131.	0.9	10
41	CSF microRNA Profiling in Alzheimer's Disease: a Screening and Validation Study. <i>Molecular Neurobiology</i> , 2017, 54, 6647-6654.	1.9	45
42	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.	1.5	24
43	Plasma miR-34a-5p and miR-545-3p as Early Biomarkers of Alzheimer's Disease: Potential and Limitations. <i>Molecular Neurobiology</i> , 2017, 54, 5550-5562.	1.9	119
44	Discovering the 3' UTR-mediated regulation of alpha-synuclein. <i>Nucleic Acids Research</i> , 2017, 45, 12888-12903.	6.5	32
45	Epigenetic Research of Neurodegenerative Disorders Using Patient iPSC-Based Models. <i>Stem Cells International</i> , 2016, 2016, 1-16.	1.2	13
46	Reply. <i>Annals of Neurology</i> , 2016, 79, 161-163.	2.8	3
47	Reply. <i>Annals of Neurology</i> , 2016, 79, 868-868.	2.8	0
48	Absence of <i>LRRK2</i> mutations in a cohort of patients with idiopathic REM sleep behavior disorder. <i>Neurology</i> , 2016, 86, 1072-1073.	1.5	30
49	Aberrant epigenome in iPSC-derived dopaminergic neurons from Parkinson's disease patients. <i>EMBO Molecular Medicine</i> , 2015, 7, 1529-1546.	3.3	117
50	Reply. <i>Annals of Neurology</i> , 2015, 78, 153-154.	2.8	1
51	The <i>MC1R</i> melanoma risk variant p.R160W is associated with Parkinson disease. <i>Annals of Neurology</i> , 2015, 77, 889-894.	2.8	52
52	TREM2 R47H variant and risk of essential tremor: A cross-sectional international multicenter study. <i>Parkinsonism and Related Disorders</i> , 2015, 21, 306-309.	1.1	28
53	MicroRNA association with synucleinopathy conversion in rapid eye movement behavior disorder. <i>Annals of Neurology</i> , 2015, 77, 895-901.	2.8	50
54	An exome study of Parkinson's disease in Sardinia, a Mediterranean genetic isolate. <i>Neurogenetics</i> , 2015, 16, 55-64.	0.7	20

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55	Nonmotor Symptoms in LRRK2 G2019S Associated Parkinson's Disease. <i>PLoS ONE</i> , 2014, 9, e108982.	1.1	79
56	Parkin loss of function contributes to RTP801 elevation and neurodegeneration in Parkinson's disease. <i>Cell Death and Disease</i> , 2014, 5, e1364-e1364.	2.7	40
57	Identification of blood serum microRNAs associated with idiopathic and LRRK2 Parkinson's disease. <i>Journal of Neuroscience Research</i> , 2014, 92, 1071-1077.	1.3	122
58	Fused in Sarcoma (FUS) gene mutations are not a frequent cause of essential tremor in Europeans. <i>Neurobiology of Aging</i> , 2013, 34, 2441.e9-2441.e11.	1.5	22
59	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. <i>Journal of Neuropathology and Experimental Neurology</i> , 2012, 71, 795-805.	0.9	18
60	Age at Onset in LRRK2-Associated PD is Modified by SNCA Variants. <i>Journal of Molecular Neuroscience</i> , 2012, 48, 245-247.	1.1	34
61	Disease-specific phenotypes in dopamine neurons from human iPSC-based models of genetic and sporadic Parkinson's disease. <i>EMBO Molecular Medicine</i> , 2012, 4, 380-395.	3.3	501
62	Brain transcriptomic profiling in idiopathic and LRRK2-associated Parkinson's disease. <i>Brain Research</i> , 2012, 1466, 152-157.	1.1	30
63	Microarray expression analysis in idiopathic and LRRK2-associated Parkinson's disease. <i>Neurobiology of Disease</i> , 2012, 45, 462-468.	2.1	33
64	LRRK2 haplotype-sharing analysis in Parkinson's disease reveals a novel p.S1761R mutation. <i>Movement Disorders</i> , 2012, 27, 146-150.	2.2	19
65	High cerebrospinal tau levels are associated with the rs242557 tau gene variant and low cerebrospinal $\beta$ -amyloid in Parkinson disease. <i>Neuroscience Letters</i> , 2011, 487, 169-173.	1.0	18
66	Different MAPT haplotypes are associated with Parkinson's disease and progressive supranuclear palsy. <i>Neurobiology of Aging</i> , 2011, 32, 547.e11-547.e16.	1.5	32
67	Identifying the genetic components underlying the pathophysiology of movement disorders. <i>The Application of Clinical Genetics</i> , 2011, 4, 81.	1.4	1
68	Lack of interaction of SNCA and MAPT genotypes in Parkinson's disease. <i>European Journal of Neurology</i> , 2011, 18, e32-e32.	1.7	12
69	123I-MIBG cardiac uptake and smell identification in parkinsonian patients with LRRK2 mutations. <i>Journal of Neurology</i> , 2011, 258, 1126-1132.	1.8	31
70	LINGO1 gene analysis in Parkinson's disease phenotypes. <i>Movement Disorders</i> , 2011, 26, 722-727.	2.2	17
71	Rapidly progressive diffuse Lewy body disease. <i>Movement Disorders</i> , 2011, 26, 1316-1323.	2.2	56
72	Reply: Rapidly progressing diffuse Lewy body disease. <i>Movement Disorders</i> , 2011, 26, 2585-2585.	2.2	0

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73	Parkinsonism, dysautonomia, REM behaviour disorder and visual hallucinations mimicking synucleinopathy in a patient with progressive supranuclear palsy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2009, 80, 578-579.	0.9	17
74	5â€²-upstream variants of CRHR1 and MAPT genes associated with age at onset in progressive supranuclear palsy and cortical basal degeneration. <i>Neurobiology of Disease</i> , 2009, 33, 164-170.	2.1	24
75	Cerebrospinal tau, phosphoâ€²tau, and betaâ€²amyloid and neuropsychological functions in Parkinson's disease. <i>Movement Disorders</i> , 2009, 24, 2203-2210.	2.2	163
76	G2019S LRRK2 mutation causing Parkinson's disease without Lewy bodies. <i>BMJ Case Reports</i> , 2009, 2009, bcr0820080632-bcr0820080632.	0.2	11
77	Clinicopathological and genetic correlates of frontotemporal lobar degeneration and corticobasal degeneration. <i>Journal of Neurology</i> , 2008, 255, 488-494.	1.8	40
78	Screening for the LRRK2 G2019S and codon-1441 mutations in a pathological series of parkinsonian syndromes and frontotemporal lobar degeneration. <i>Journal of the Neurological Sciences</i> , 2008, 270, 94-98.	0.3	35
79	Lack of association of APOE and tau polymorphisms with dementia in Parkinsonâ€™s disease. <i>Neuroscience Letters</i> , 2008, 448, 20-23.	1.0	54
80	G2019S LRRK2 mutation causing Parkinson's disease without Lewy bodies. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2007, 78, 626-628.	0.9	90
81	A novel <i>MAPT</i> mutation (P301T) associated with familial frontotemporal dementia. <i>European Journal of Neurology</i> , 2007, 14, e9-10.	1.7	16
82	A novel mutation in the <i>PSEN1</i> gene (L286P) associated with familial early-onset dementia of Alzheimer type and lobar haematomas. <i>European Journal of Neurology</i> , 2007, 14, 1409-1412.	1.7	23
83	Increased cerebral activity in Parkinson's disease patients carrying the DRD2 TaqIA A1 allele during a demanding motor task: a compensatory mechanism?. <i>Genes, Brain and Behavior</i> , 2007, 6, 588-592.	1.1	14
84	Tau and saitoihin gene expression pattern in progressive supranuclear palsy. <i>Brain Research</i> , 2007, 1145, 168-176.	1.1	10
85	Late-onset frontotemporal dementia associated with a novel PGRN mutation. <i>Journal of Neural Transmission</i> , 2007, 114, 1051-1054.	1.4	13
86	Brain tau expression and correlation with the H1/H1 tau genotype in frontotemporal lobar degeneration patients. <i>Journal of Neural Transmission</i> , 2007, 114, 1585-1588.	1.4	6
87	No evidence of CRHR1 gene involvement in progressive supranuclear palsy. <i>Neuroscience Letters</i> , 2006, 409, 61-64.	1.0	2
88	LRRK2 Mutations in Spanish Patients With Parkinson Disease. <i>Archives of Neurology</i> , 2006, 63, 377.	4.9	127
89	A Novel Intronic Mutation in the DDP1 Gene in a Family With X-linked Dystonia-Deafness Syndrome. <i>Archives of Neurology</i> , 2005, 62, 306.	4.9	30
90	Association study of the G258S transferrin gene polymorphism and Parkinson's disease in the Spanish population. <i>Journal of Neurology</i> , 2005, 252, 1269-1270.	1.8	10

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91	Novel haplotypes in 17q21 are associated with progressive supranuclear palsy. <i>Annals of Neurology</i> , 2004, 56, 249-258.	2.8	71
92	Ubiquitin-negative mini-pick-like bodies in the dentate gyrus in p301l tauopathy. <i>Journal of Alzheimer's Disease</i> , 2004, 5, 445-454.	1.2	16
93	Primary progressive aphasia as the initial manifestation of corticobasal degeneration and unusual tauopathies. <i>Acta Neuropathologica</i> , 2003, 106, 419-435.	3.9	67
94	Mutational study of the nuclear factor kappa B inducing kinase gene in patients with progressive supranuclear palsy. <i>Neuroscience Letters</i> , 2003, 340, 158-160.	1.0	1
95	A Novel Mutation in the PSEN2 Gene (T430M) Associated With Variable Expression in a Family With Early-Onset Alzheimer Disease. <i>Archives of Neurology</i> , 2003, 60, 1149.	4.9	46
96	Frequency of Mutations in the Presenilin and Amyloid Precursor Protein Genes in Early-Onset Alzheimer Disease in Spain. <i>Archives of Neurology</i> , 2002, 59, 1759.	4.9	103
97	A novel mutation (V89L) in the presenilin 1 gene in a family with early onset Alzheimer's disease and marked behavioural disturbances. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2002, 72, 266-269.	0.9	51
98	Further extension of the H1 haplotype associated with progressive supranuclear palsy. <i>Movement Disorders</i> , 2002, 17, 550-556.	2.2	61
99	Detection of the presenilin 1 gene mutation (M139T) in early-onset familial Alzheimer disease in Spain. <i>Neuroscience Letters</i> , 2001, 299, 239-241.	1.0	12
100	Analysis of the coding and the 5' flanking regions of the $\alpha$ -synuclein gene in patients with Parkinson's disease. <i>Movement Disorders</i> , 2001, 16, 1115-1119.	2.2	23
101	Alzheimer disease is not associated with polymorphisms in the angiotensinogen and renin genes. <i>American Journal of Medical Genetics Part A</i> , 2001, 105, 761-764.	2.4	16
102	Significant association between the tau gene A0/A0 genotype and Parkinson's disease. <i>Annals of Neurology</i> , 2000, 47, 242-245.	2.8	129
103	A Novel Presenilin 1 Mutation (Leu166Arg) Associated With Early-Onset Alzheimer Disease. <i>Archives of Neurology</i> , 2000, 57, 485.	4.9	19
104	Identification of a novel polymorphism in the promoter region of the tau gene highly associated to progressive supranuclear palsy in humans. <i>Neuroscience Letters</i> , 1999, 275, 183-186.	1.0	56
105	$\epsilon$ -Antichymotrypsin gene polymorphism and risk for Alzheimer's disease in the Spanish population. <i>Neuroscience Letters</i> , 1998, 240, 107-109.	1.0	26
106	Apolipoprotein E $\epsilon$ 4 alleles and meiotic origin of non-disjunction in Down syndrome children and in their corresponding fathers and mothers. <i>Neuroscience Letters</i> , 1998, 248, 1-4.	1.0	11
107	Significant Changes in the Tau A0 and A3 Alleles in Progressive Supranuclear Palsy and Improved Genotyping by Silver Detection. <i>Archives of Neurology</i> , 1998, 55, 1122.	4.9	85
108	The genotype 2/2 of the presenilin-1 polymorphism is decreased in Spanish early-onset Alzheimer's disease. <i>Neuroscience Letters</i> , 1997, 227, 201-204.	1.0	26

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109	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