# Mario Ezquerra Trabalon

List of Publications by Year
in descending order

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Version: 2024-02-01

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|---|---|---|---|
| 109 papers | 5,593 citations | 117625 34 h-index | 95266 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. | 10.2 | 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. | 6.9 | 501 |
| 3 | Cerebrospinal tau, phosphoâ€tau, and betaâ€amyloid and neuropsychological functions in Parkinson's disease. Movement Disorders, 2009, 24, 2203-2210. | 3.9 | 163 |
| 4 | Significant association between the tau gene A0/A0 genotype and Parkinson's disease. Annals of Neurology, 2000, 47, 242-245. | 5.3 | 129 |
| 5 | LRRK2 Mutations in Spanish Patients With Parkinson Disease. Archives of Neurology, 2006, 63, 377. | 4.5 | 127 |
| 6 | Identification of blood serum microâ€RNAs associated with idiopathic and LRRK2 Parkinson's disease. Journal of Neuroscience Research, 2014, 92, 1071-1077. | 2.9 | 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. | 4.0 | 119 |
| 8 | Aberrant epigenome in <scp>iPSC</scp> â€derived dopaminergic neurons from Parkinson's disease patients. EMBO Molecular Medicine, 2015, 7, 1529-1546. | 6.9 | 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. | 4.5 | 103 |
| 10 | Mitochondria function associated genes contribute to Parkinsonâ€™s Disease risk and later age at onset. Npj Parkinson's Disease, 2019, 5, 8. | 5.3 | 95 |
| 11 | 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 |
| 12 | G2019S LRRK2 mutation causing Parkinson's disease without Lewy bodies. Journal of Neurology, Neurosurgery and Psychiatry, 2007, 78, 626-628. | 1.9 | 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. | 4.5 | 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. | 5.3 | 83 |
| 15 | Nonmotor Symptoms in LRRK2 G2019S Associated Parkinsonâ€™s Disease. PLoS ONE, 2014, 9, e108982. | 2.5 | 79 |
| 16 | Novel haplotypes in 17q21 are associated with progressive supranuclear palsy. Annals of Neurology, 2004, 56, 249-258. | 5.3 | 71 |
| 17 | Primary progressive aphasia as the initial manifestation of corticobasal degeneration and unusual tauopathies. Acta Neuropathologica, 2003, 106, 419-435. | 7.7 | 67 |
| 18 | The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. Movement Disorders, 2019, 34, 460-468. | 3.9 | 66 |

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|---|---|---|---|
| 19 | Further extension of the H1 haplotype associated with progressive supranuclear palsy. Movement Disorders, 2002, 17, 550-556. | 3.9 | 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. | 3.9 | 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. | 2.1 | 56 |
| 22 | Rapidly progressive diffuse Lewy body disease. Movement Disorders, 2011, 26, 1316-1323. | 3.9 | 56 |
| 23 | MicroRNA alterations in iPSC-derived dopaminergic neurons from Parkinson disease patients. Neurobiology of Aging, 2018, 69, 283-291. | 3.1 | 55 |
| 24 | HLA and microtubule-associated protein tau H1 haplotype associations in anti-IgLON5 disease. Neurology: Neuroimmunology and NeuroInflammation, 2019, 6, . | 6.0 | 55 |
| 25 | Lack of association of APOE and tau polymorphisms with dementia in Parkinsonâ€™s disease. Neuroscience Letters, 2008, 448, 20-23. | 2.1 | 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. | 5.3 | 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. | 1.9 | 51 |
| 28 | Micro<scp>RNA</scp> association with synucleinopathy conversion in rapid eye movement behavior disorder. Annals of Neurology, 2015, 77, 895-901. | 5.3 | 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. | 3.9 | 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. | 4.5 | 46 |
| 31 | CSF microRNA Profiling in Alzheimerâ€™s Disease: a Screening and Validation Study. Molecular Neurobiology, 2017, 54, 6647-6654. | 4.0 | 45 |
| 32 | Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342. | 12.8 | 44 |
| 33 | Clinicopathological and genetic correlates of frontotemporal lobar degeneration and corticobasal degeneration. Journal of Neurology, 2008, 255, 488-494. | 3.6 | 40 |
| 34 | Parkin loss of function contributes to RTP801 elevation and neurodegeneration in Parkinsonâ€™s disease. Cell Death and Disease, 2014, 5, e1364-e1364. | 6.3 | 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. | 4.0 | 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. | 0.6 | 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. | 2.3 | 34 |
| 38 | Microarray expression analysis in idiopathic and LRRK2-associated Parkinson's disease. Neurobiology of Disease, 2012, 45, 462-468. | 4.4 | 33 |
| 39 | Different MAPT haplotypes are associated with Parkinson's disease and progressive supranuclear palsy. Neurobiology of Aging, 2011, 32, 547.e11-547.e16. | 3.1 | 32 |
| 40 | Discovering the 3â€² UTR-mediated regulation of alpha-synuclein. Nucleic Acids Research, 2017, 45, 12888-12903. | 14.5 | 32 |
| 41 | Cerebrospinal fluid levels of coenzyme Q10 are reduced in multiple system atrophy. Parkinsonism and Related Disorders, 2018, 46, 16-23. | 2.2 | 32 |
| 42 | 123I-MIBG cardiac uptake and smell identification in parkinsonian patients with LRRK2 mutations. Journal of Neurology, 2011, 258, 1126-1132. | 3.6 | 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. | 4.5 | 30 |
| 44 | Brain transcriptomic profiling in idiopathic and LRRK2-associated Parkinson's disease. Brain Research, 2012, 1466, 152-157. | 2.2 | 30 |
| 45 | Absence of LRRK2 mutations in a cohort of patients with idiopathic REM sleep behavior disorder. Neurology, 2016, 86, 1072-1073. | 1.1 | 30 |
| 46 | Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42. | 5.3 | 29 |
| 47 | TREM2 R47H variant and risk of essential tremor: A cross-sectional international multicenter study. Parkinsonism and Related Disorders, 2015, 21, 306-309. | 2.2 | 28 |
| 48 | Accumulation of mitochondrial 7S DNA in idiopathic and LRRK2 associated Parkinson's disease. EBioMedicine, 2019, 48, 554-567. | 6.1 | 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. | 2.1 | 26 |
| 50 | Î±-Antichymotrypsin gene polymorphism and risk for Alzheimer's disease in the Spanish population. Neuroscience Letters, 1998, 240, 107-109. | 2.1 | 26 |
| 51 | Cerebrospinal fluid cytokines in multiple system atrophy: A cross-sectional Catalan MSA registry study. Parkinsonism and Related Disorders, 2019, 65, 3-12. | 2.2 | 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. | 3.9 | 25 |
| 53 | Mitochondrial and autophagic alterations in skin fibroblasts from Parkinson disease patients with Parkin mutations. Aging, 2019, 11, 3750-3767. | 3.1 | 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. | 4.4 | 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. | 3.1 | 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. | 3.9 | 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. | 3.3 | 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. | 3.1 | 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. | 4.4 | 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. | 3.9 | 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. | 2.8 | 21 |
| 62 | An exome study of Parkinsonâ€™s disease in Sardinia, a Mediterranean genetic isolate. Neurogenetics, 2015, 16, 55-64. | 1.4 | 20 |
| 63 | Peripheral insulin and amylin levels in Parkinson's disease. Parkinsonism and Related Disorders, 2020, 79, 91-96. | 2.2 | 20 |
| 64 | A Novel Presenilin 1 Mutation (Leu166Arg) Associated With Early-Onset Alzheimer Disease. Archives of Neurology, 2000, 57, 485. | 4.5 | 19 |
| 65 | LRRK2 haplotypeâ€sharing analysis in Parkinson's disease reveals a novel p.S1761R mutation. Movement Disorders, 2012, 27, 146-150. | 3.9 | 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. | 2.1 | 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. | 1.7 | 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. | 1.9 | 17 |
| 69 | LINGO1 gene analysis in Parkinson's disease phenotypes. Movement Disorders, 2011, 26, 722-727. | 3.9 | 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. | 4.0 | 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. | 2.6 | 16 |

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|---|---|---|---|
| 73 | A novel MAPT mutation (P301T) associated with familial frontotemporal dementia. European Journal of Neurology, 2007, 14, e9-10. | 3.3 | 16 |
| 74 | Whole-genome DNA hyper-methylation in iPSC-derived dopaminergic neurons from Parkinsonâ€™s disease patients. Clinical Epigenetics, 2019, 11, 108. | 4.1 | 16 |
| 75 | Analysis of DNM3 and VAMP4 as genetic modifiers of LRRK2 Parkinsonâ€™s disease. Neurobiology of Aging, 2021, 97, 148.e17-148.e24. | 3.1 | 16 |
| 76 | LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032. | 10.2 | 15 |
| 77 | <scp>MicroRNA</scp> Deregulation in Blood Serum Identifies Multiple System Atrophy Altered Pathways. Movement Disorders, 2020, 35, 1873-1879. | 3.9 | 15 |
| 78 | Dairy Intake and Parkinson's Disease: A Mendelian Randomization Study. Movement Disorders, 2022, 37, 857-864. | 3.9 | 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. | 2.2 | 14 |
| 80 | CCAAT/enhancer binding protein Î´ is a transcriptional repressor of Î±-synuclein. Cell Death and Differentiation, 2020, 27, 509-524. | 11.2 | 14 |
| 81 | Late-onset frontotemporal dementia associated with a novel PGRN mutation. Journal of Neural Transmission, 2007, 114, 1051-1054. | 2.8 | 13 |
| 82 | Epigenetic Research of Neurodegenerative Disorders Using Patient iPSC-Based Models. Stem Cells International, 2016, 2016, 1-16. | 2.5 | 13 |
| 83 | Transcriptional alterations in skin fibroblasts from Parkinson's disease patients with parkin mutations. Neurobiology of Aging, 2018, 65, 206-216. | 3.1 | 13 |
| 84 | Detection of the presenilin 1 gene mutation (M139T) in early-onset familial Alzheimer disease in Spain. Neuroscience Letters, 2001, 299, 239-241. | 2.1 | 12 |
| 85 | Lack of interaction of SNCA and MAPT genotypes in Parkinson's disease. European Journal of Neurology, 2011, 18, e32-e32. | 3.3 | 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. | 2.1 | 11 |
| 87 | G2019S LRRK2 mutation causing Parkinson's disease without Lewy bodies. BMJ Case Reports, 2009, 2009, bcr0820080632-bcr0820080632. | 0.5 | 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. | 3.6 | 10 |
| 89 | Tau and saitohin gene expression pattern in progressive supranuclear palsy. Brain Research, 2007, 1145, 168-176. | 2.2 | 10 |
| 90 | MAPT association with REM sleep behavior disorder. Neurology: Genetics, 2017, 3, e131. | 1.9 | 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. | 5.7 | 9 |
| 92 | Differential Phosphoâ€Signatures in Blood Cells Identify <scp>LRRK2 G2019S</scp> Carriers in Parkinson's Disease. Movement Disorders, 2022, 37, 1004-1015. | 3.9 | 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. | 5.1 | 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. | 5.3 | 8 |
| 95 | Transcriptomic differences in MSA clinical variants. Scientific Reports, 2020, 10, 10310. | 3.3 | 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. | 2.8 | 6 |
| 97 | Regulatory rare variants of the dopaminergic gene ANKK1 as potential risk factors for Parkinsonâ€™s disease. Scientific Reports, 2021, 11, 9879. | 3.3 | 4 |
| 98 | The Interaction between <scp>HLAâ€ÐRB1</scp> and Smoking in Parkinson's Disease Revisited. Movement Disorders, 2022, 37, 1929-1937. | 3.9 | 4 |
| 99 | Reply. Annals of Neurology, 2016, 79, 161-163. | 5.3 | 3 |
| 100 | Parkinsonâ€™s disease as a systemic pathology. Aging, 2019, 11, 1081-1082. | 3.1 | 3 |
| 101 | No evidence of CRHR1 gene involvement in progressive supranuclear palsy. Neuroscience Letters, 2006, 409, 61-64. | 2.1 | 2 |
| 102 | Smoking is associated with age at disease onset in Parkinson's disease. Parkinsonism and Related Disorders, 2022, 97, 79-83. | 2.2 | 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. | 2.1 | 1 |
| 104 | Identifying the genetic components underlying the pathophysiology of movement disorders. The Application of Clinical Genetics, 2011, 4, 81. | 3.0 | 1 |
| 105 | Reply. Annals of Neurology, 2015, 78, 153-154. | 5.3 | 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. | 3.1 | 1 |
| 107 | Transcriptome analysis in LRRK2 and idiopathic Parkinsonâ€™s disease at different glucose levels. Npj Parkinson's Disease, 2021, 7, 109. | 5.3 | 1 |
| 108 | Reply: Rapidly progressing diffuse Lewy body disease. Movement Disorders, 2011, 26, 2585-2585. | 3.9 | 0 |

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|---|---|---|---|
| 109 | Reply. Annals of Neurology, 2016, 79, 868-868. | 5.3 | 0 |