# Jose T Bras

List of Publications by Year
in descending order



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|---|---|---|---|
| 156 papers | 20,872 citations | 31976 53 h-index | 11939 134 g-index |
| 176 all docs | 176 docs citations | 176 times ranked | 25390 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Diagnosis and management of dementia with Lewy bodies. Neurology, 2017, 89, 88-100. | 1.1 | 2,805 |
| 2 | TREM2 Variants in Alzheimer's Disease. New England Journal of Medicine, 2013, 368, 117-127. | 27.0 | 2,385 |
| 3 | Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430. | 21.4 | 1,962 |
| 4 | Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. Nature Genetics, 2014, 46, 989-993. | 21.4 | 1,685 |
| 5 | 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 |
| 6 | Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, . | 12.6 | 1,085 |
| 7 | Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384. | 21.4 | 783 |
| 8 | Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. Brain, 2009, 132, 1783-1794. | 7.6 | 612 |
| 9 | Rare coding variants in the phospholipase D3 gene confer risk for Alzheimerâ€™s disease. Nature, 2014, 505, 550-554. | 27.8 | 425 |
| 10 | A Multicenter Study of Glucocerebrosidase Mutations in Dementia With Lewy Bodies. JAMA Neurology, 2013, 70, 727. | 9.0 | 374 |
| 11 | The age factor in Alzheimerâ€™s disease. Genome Medicine, 2015, 7, 106. | 8.2 | 271 |
| 12 | SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614. | 5.3 | 257 |
| 13 | Mutation of the parkinsonism gene ATP13A2 causes neuronal ceroid-lipofuscinosis. Human Molecular Genetics, 2012, 21, 2646-2650. | 2.9 | 231 |
| 14 | Mutations in ANO3 Cause Dominant Craniocervical Dystonia: Ion Channel Implicated in Pathogenesis. American Journal of Human Genetics, 2012, 91, 1041-1050. | 6.2 | 224 |
| 15 | DYT16, a novel young-onset dystonia-parkinsonism disorder: identification of a segregating mutation in the stress-response protein PRKRA. Lancet Neurology, The, 2008, 7, 207-215. | 10.2 | 202 |
| 16 | Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. Lancet Neurology, The, 2018, 17, 64-74. | 10.2 | 195 |
| 17 | Genetic analysis implicates APOE, SNCA and suggests lysosomal dysfunction in the etiology of dementia with Lewy bodies. Human Molecular Genetics, 2014, 23, 6139-6146. | 2.9 | 178 |
| 18 | Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009. | 2.9 | 176 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Age at symptom onset and death and disease duration in genetic frontotemporal dementia: an international retrospective cohort study. Lancet Neurology, The, 2020, 19, 145-156. | 10.2 | 175 |
| 20 | Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492. | 7.6 | 169 |
| 21 | The role of TREM2 in Alzheimer's disease and other neurodegenerative disorders. Lancet Neurology, The, 2018, 17, 721-730. | 10.2 | 161 |
| 22 | SQSTM1Mutations in French Patients With Frontotemporal Dementia or Frontotemporal Dementia With Amyotrophic Lateral Sclerosis. JAMA Neurology, 2013, 70, 1403-10. | 9.0 | 153 |
| 23 | Investigating the Causal Relationship of C-Reactive Protein with 32 Complex Somatic and Psychiatric Outcomes: A Large-Scale Cross-Consortium Mendelian Randomization Study. PLoS Medicine, 2016, 13, e1001976. | 8.4 | 150 |
| 24 | Genetic Analysis of Inherited Leukodystrophies. JAMA Neurology, 2013, 70, 875. | 9.0 | 147 |
| 25 | Insights into TREM2 biology by network analysis of human brain gene expression data. Neurobiology of Aging, 2013, 34, 2699-2714. | 3.1 | 145 |
| 26 | Serum neurofilament light chain in genetic frontotemporal dementia: a longitudinal, multicentre cohort study. Lancet Neurology, The, 2019, 18, 1103-1111. | 10.2 | 128 |
| 27 | Mutations in PNKP Cause Recessive Ataxia with Oculomotor Apraxia Type 4. American Journal of Human Genetics, 2015, 96, 474-479. | 6.2 | 127 |
| 28 | Emerging pathways in genetic Parkinson's disease: Potential role of ceramide metabolism in Lewy body disease. FEBS Journal, 2008, 275, 5767-5773. | 4.7 | 121 |
| 29 | A novel compound heterozygous mutation in TREM2 found in a Turkish frontotemporal dementia-like family. Neurobiology of Aging, 2013, 34, 2890.e1-2890.e5. | 3.1 | 113 |
| 30 | Use of next-generation sequencing and other whole-genome strategies to dissect neurological disease. Nature Reviews Neuroscience, 2012, 13, 453-464. | 10.2 | 110 |
| 31 | Missense variant in TREML2 protects against Alzheimer's disease. Neurobiology of Aging, 2014, 35, 1510.e19-1510.e26. | 3.1 | 110 |
| 32 | Hyposmia and cognitive impairment in Gaucher disease patients and carriers. Movement Disorders, 2012, 27, 526-532. | 3.9 | 108 |
| 33 | NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. Neurobiology of Aging, 2017, 57, 247.e9-247.e13. | 3.1 | 108 |
| 34 | Complete screening for glucocerebrosidase mutations in Parkinson disease patients from Portugal. Neurobiology of Aging, 2009, 30, 1515-1517. | 3.1 | 97 |
| 35 | NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12. | 3.1 | 96 |
| 36 | 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 |

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|---|---|---|---|
| 37 | The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52. | 2.4 | 94 |
| 38 | Genetic risk factors for the posterior cortical atrophy variant of Alzheimer's disease. Alzheimer's and Dementia, 2016, 12, 862-871. | 0.8 | 93 |
| 39 | Loss-of-function mutations in RAB39B are associated with typical early-onset Parkinson disease. Neurology: Genetics, 2015, 1, e9. | 1.9 | 90 |
| 40 | SnapShot: Genetics of Alzheimerâ€™s Disease. Cell, 2013, 155, 968-968.e1. | 28.9 | 86 |
| 41 | Homozygous TREM2 mutation in a family with atypical frontotemporal dementia. Neurobiology of Aging, 2014, 35, 2419.e23-2419.e25. | 3.1 | 84 |
| 42 | Microdeletion in a FAAH pseudogene identified in a patient with high anandamide concentrations and pain insensitivity. British Journal of Anaesthesia, 2019, 123, e249-e253. | 3.4 | 82 |
| 43 | SnapShot: Genetics of Parkinsonâ€™s Disease. Cell, 2015, 160, 570-570.e1. | 28.9 | 79 |
| 44 | Genome-wide analysis of genetic correlation in dementia with Lewy bodies, Parkinson's and Alzheimer's diseases. Neurobiology of Aging, 2016, 38, 214.e7-214.e10. | 3.1 | 78 |
| 45 | TYROBP genetic variants in early-onset Alzheimer's disease. Neurobiology of Aging, 2016, 48, 222.e9-222.e15. | 3.1 | 69 |
| 46 | The Genetics of Dementia with Lewy Bodies: Current Understanding and Future Directions. Current Neurology and Neuroscience Reports, 2018, 18, 67. | 4.2 | 69 |
| 47 | Alzheimerâ€™s disease polygenic risk score as a predictor of conversion from mild-cognitive impairment. Translational Psychiatry, 2019, 9, 154. | 4.8 | 69 |
| 48 | SnapShot: Genetics of ALS and FTD. Cell, 2015, 160, 798-798.e1. | 28.9 | 68 |
| 49 | Linkage, whole genome sequence, and biological data implicate variants in RAB10 in Alzheimerâ€™s disease resilience. Genome Medicine, 2017, 9, 100. | 8.2 | 67 |
| 50 | Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416. | 9.0 | 66 |
| 51 | 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 |
| 52 | Study protocol: Insight 46 â€“ a neuroscience sub-study of the MRC National Survey of Health and Development. BMC Neurology, 2017, 17, 75. | 1.8 | 64 |
| 53 | A systematic screening to identifyde novomutations causing sporadic early-onset Parkinson's disease. Human Molecular Genetics, 2015, 24, 6711-6720. | 2.9 | 59 |
| 54 | Next generation sequencing techniques in neurological diseases: redefining clinical and molecular associations. Human Molecular Genetics, 2014, 23, R47-R53. | 2.9 | 57 |

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|---|---|---|---|
| 55 | 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 |
| 56 | Influence of Single Nucleotide Polymorphisms in <b><i>COMT</i></b>, <b><i>MAO-A</i></b> and <b><i>BDNF</i></b> Genes on Dyskinesias and Levodopa Use in Parkinson's Disease. Neurodegenerative Diseases, 2014, 13, 24-28. | 1.4 | 56 |
| 57 | Investigating the role of rare coding variability in Mendelian dementia genes ( APP , PSEN1 , PSEN2 , GRN) Tj ETQq1 1 0.784314 rgBT | 3.1 | 53 |
| 58 | Analysis of Parkinson disease patients from Portugal for mutations in SNCA, PRKN, PINK1 and LRRK2. BMC Neurology, 2008, 8, 1. | 1.8 | 52 |
| 59 | Additional rare variant analysis in Parkinsonâ€™s disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. Human Molecular Genetics, 2016, 25, ddw348. | 2.9 | 48 |
| 60 | 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 |
| 61 | Functional network resilience to pathology in presymptomatic genetic frontotemporal dementia. Neurobiology of Aging, 2019, 77, 169-177. | 3.1 | 47 |
| 62 | Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342. | 12.8 | 44 |
| 63 | A rare loss-of-function variant of ADAM17 is associated with late-onset familial Alzheimer disease. Molecular Psychiatry, 2020, 25, 629-639. | 7.9 | 42 |
| 64 | Progression of Behavioral Disturbances and Neuropsychiatric Symptoms in Patients With Genetic Frontotemporal Dementia. JAMA Network Open, 2021, 4, e2030194. | 5.9 | 42 |
| 65 | Identification of sixteen novel candidate genes for late onset Parkinsonâ€™s disease. Molecular Neurodegeneration, 2021, 16, 35. | 10.8 | 41 |
| 66 | ABCA7 p.G215S as potential protective factor for Alzheimer's disease. Neurobiology of Aging, 2016, 46, 235.e1-235.e9. | 3.1 | 37 |
| 67 | Brain functional network integrity sustains cognitive function despite atrophy in presymptomatic genetic frontotemporal dementia. Alzheimer's and Dementia, 2021, 17, 500-514. | 0.8 | 36 |
| 68 | Current concepts and controversies in the pathogenesis of Parkinsonâ€™s disease dementia and Dementia with Lewy Bodies. F1000Research, 2017, 6, 1604. | 1.6 | 35 |
| 69 | Exome sequencing in a consanguineous family clinically diagnosed with early-onset Alzheimer's disease identifies a homozygous CTSF mutation. Neurobiology of Aging, 2016, 46, 236.e1-236.e6. | 3.1 | 34 |
| 70 | Influence of Coding Variability in APP-AÎ² Metabolism Genes in Sporadic Alzheimerâ€™s Disease. PLoS ONE, 2016, 11, e0150079. | 2.5 | 34 |
| 71 | The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. NeuroImage, 2019, 189, 645-654. | 4.2 | 33 |
| 72 | A novel human pain insensitivity disorder caused by a point mutation in ZFHX2. Brain, 2018, 141, 365-376. | 7.6 | 32 |

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|---|---|---|---|
| 73 | Mendelian adult-onset leukodystrophy genes in Alzheimer's disease: critical influence of CSF1R and NOTCH3. Neurobiology of Aging, 2018, 66, 179.e17-179.e29. | 3.1 | 32 |
| 74 | Polygenic risk score in postmortem diagnosed sporadic early-onset Alzheimer's disease. Neurobiology of Aging, 2018, 62, 244.e1-244.e8. | 3.1 | 30 |
| 75 | Heritability and genetic variance of dementia with Lewy bodies. Neurobiology of Disease, 2019, 127, 492-501. | 4.4 | 29 |
| 76 | Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42. | 5.3 | 29 |
| 77 | Youngâ€onset parkinsonism due to homozygous duplication of Î±â€synuclein in a consanguineous family. Movement Disorders, 2012, 27, 1829-1830. | 3.9 | 27 |
| 78 | Genetic architecture of common non-Alzheimerâ€™s disease dementias. Neurobiology of Disease, 2020, 142, 104946. | 4.4 | 27 |
| 79 | Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. Acta Neuropathologica Communications, 2020, 8, 5. | 5.2 | 27 |
| 80 | A data-driven disease progression model of fluid biomarkers in genetic frontotemporal dementia. Brain, 2022, 145, 1805-1817. | 7.6 | 27 |
| 81 | Genome-wide association of polygenic risk extremes for Alzheimer's disease in the UK Biobank. Scientific Reports, 2022, 12, 8404. | 3.3 | 27 |
| 82 | A nonsense mutation in PRNP associated with clinical Alzheimer's disease. Neurobiology of Aging, 2014, 35, 2656.e13-2656.e16. | 3.1 | 26 |
| 83 | The Chihuahua dog: A new animal model for neuronal ceroid lipofuscinosis CLN7 disease?. Journal of Neuroscience Research, 2016, 94, 339-347. | 2.9 | 26 |
| 84 | Social cognition impairment in genetic frontotemporal dementia within the GENFI cohort. Cortex, 2020, 133, 384-398. | 2.4 | 26 |
| 85 | Polygenic risk scores for Alzheimer's disease are related to dementia risk in APOE É›4 negatives. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2021, 13, e12142. | 2.4 | 25 |
| 86 | Conceptual framework for the definition of preclinical and prodromal frontotemporal dementia. Alzheimer's and Dementia, 2022, 18, 1408-1423. | 0.8 | 24 |
| 87 | <scp>RARS</scp>2 mutations in a sibship with infantile spasms. Epilepsia, 2016, 57, e97-e102. | 5.1 | 23 |
| 88 | Genetics Underlying Atypical Parkinsonism and Related Neurodegenerative Disorders. International Journal of Molecular Sciences, 2015, 16, 24629-24655. | 4.1 | 21 |
| 89 | Mutation analysis of sporadic early-onset Alzheimer's disease using the NeuroX array. Neurobiology of Aging, 2017, 49, 215.e1-215.e8. | 3.1 | 21 |
| 90 | KCNN2 mutation in autosomalâ€dominant tremulous myoclonusâ€dystonia. European Journal of Neurology, 2020, 27, 1471-1477. | 3.3 | 21 |

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|---|---|---|---|
| 91 | Stratifying the Presymptomatic Phase of Genetic Frontotemporal Dementia by Serum <scp>NfL</scp> and <scp>pNfH</scp>: A Longitudinal Multicentre Study. Annals of Neurology, 2022, 91, 33-47. | 5.3 | 21 |
| 92 | Analysis of brain atrophy and local gene expression in genetic frontotemporal dementia. Brain Communications, 2020, 2, . | 3.3 | 20 |
| 93 | Alzheimerâ€™s Disease Genetics: Review of Novel Loci Associated with Disease. Current Genetic Medicine Reports, 2020, 8, 1-16. | 1.9 | 20 |
| 94 | Mutation Frequency of the Major Frontotemporal Dementia Genes, MAPT, GRN and C9ORF72 in a Turkish Cohort of Dementia Patients. PLoS ONE, 2016, 11, e0162592. | 2.5 | 19 |
| 95 | Mutation of TBCK causes a rare recessive developmental disorder. Neurology: Genetics, 2016, 2, e76. | 1.9 | 19 |
| 96 | Multi-infarct dementia of Swedish type is caused by a 3â€™UTR mutation of COL4A1. Brain, 2017, 140, e29-e29. | 7.6 | 19 |
| 97 | An AARS variant as the likely cause of Swedish type hereditary diffuse leukoencephalopathy with spheroids. Acta Neuropathologica Communications, 2019, 7, 188. | 5.2 | 19 |
| 98 | Faster Cortical Thinning and Surface Area Loss in Presymptomatic and Symptomatic C9orf72 Repeat Expansion Adult Carriers. Annals of Neurology, 2020, 88, 113-122. | 5.3 | 19 |
| 99 | Challenge accepted: uncovering the role of rare genetic variants in Alzheimerâ€™s disease. Molecular Neurodegeneration, 2022, 17, 3. | 10.8 | 19 |
| 100 | Assessment of Parkinson's disease risk loci in Greece. Neurobiology of Aging, 2014, 35, 442.e9-442.e16. | 3.1 | 18 |
| 101 | Clinical, ocular motor, and imaging profile of Niemann-Pick type C heterozygosity. Neurology, 2020, 94, e1702-e1715. | 1.1 | 18 |
| 102 | A Non-APOE Polygenic Risk Score for Alzheimerâ€™s Disease Is Associated With Cerebrospinal Fluid Neurofilament Light in a Representative Sample of Cognitively Unimpaired 70-Year Olds. Journals of Gerontology - Series A Biological Sciences and Medical Sciences, 2021, 76, 983-990. | 3.6 | 18 |
| 103 | CLN6 disease caused by the same mutation originating in Pakistan has varying pathology. European Journal of Paediatric Neurology, 2013, 17, 657-660. | 1.6 | 17 |
| 104 | CYLD variants in frontotemporal dementia associated with severe memory impairment in a Portuguese cohort. Brain, 2020, 143, e67-e67. | 7.6 | 16 |
| 105 | Genetics of synucleins in neurodegenerative diseases. Acta Neuropathologica, 2021, 141, 471-490. | 7.7 | 16 |
| 106 | LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032. | 10.2 | 15 |
| 107 | Adenosine Deaminase Two and Immunoglobulin M Accurately Differentiate Adult Sneddonâ€™s Syndrome of Unknown Cause. Cerebrovascular Diseases, 2018, 46, 257-264. | 1.7 | 15 |
| 108 | A comprehensive screening of copy number variability in dementia with Lewy bodies. Neurobiology of Aging, 2019, 75, 223.e1-223.e10. | 3.1 | 13 |

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|---|---|---|---|
| 109 | Action Myoclonus and Seizure in Kuforâ€Rakeb Syndrome. Movement Disorders Clinical Practice, 2018, 5, 195-199. | 1.5 | 13 |
| 110 | A Phenotype of Atypical Apraxia of Speech in a Family Carrying SQSTM1 Mutation. Journal of Alzheimer's Disease, 2014, 43, 625-630. | 2.6 | 12 |
| 111 | Screening exons 16 and 17 of the amyloid precursor protein gene in sporadic early-onset Alzheimer's disease. Neurobiology of Aging, 2016, 39, 220.e1-220.e7. | 3.1 | 12 |
| 112 | Analysis of C9orf72 repeat expansions in a large international cohort of dementia with Lewy bodies. Neurobiology of Aging, 2017, 49, 214.e13-214.e15. | 3.1 | 12 |
| 113 | An Aged Canid with Behavioral Deficits Exhibits Blood and Cerebrospinal Fluid Amyloid Beta Oligomers. Frontiers in Aging Neuroscience, 2018, 10, 7. | 3.4 | 12 |
| 114 | LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032-1033. | 10.2 | 11 |
| 115 | Peripheral GRN mRNA and Serum Progranulin Levels as a Potential Indicator for Both the Presence of Splice Site Mutations and Individuals at Risk for Frontotemporal Dementia. Journal of Alzheimer's Disease, 2019, 67, 159-167. | 2.6 | 11 |
| 116 | The clinical syndrome of dystonia with anarthria/aphonia. Parkinsonism and Related Disorders, 2016, 24, 20-27. | 2.2 | 10 |
| 117 | Late-onset and acute presentation of Brown-Vialetto-Van Laere syndrome in a Brazilian family. Neurology: Genetics, 2018, 4, e215. | 1.9 | 10 |
| 118 | How understudied populations have contributed to our understanding of Alzheimerâ€™s disease genetics. Brain, 2021, 144, 1067-1081. | 7.6 | 10 |
| 119 | <scp>CLN</scp>8 disease caused by large genomic deletions. Molecular Genetics & Genomic Medicine, 2017, 5, 85-91. | 1.2 | 9 |
| 120 | A panel of CSF proteins separates genetic frontotemporal dementia from presymptomatic mutation carriers: a GENFI study. Molecular Neurodegeneration, 2021, 16, 79. | 10.8 | 9 |
| 121 | Prion-like Î±-synuclein pathology in the brain of infants with Krabbe disease. Brain, 2022, 145, 1257-1263. | 7.6 | 9 |
| 122 | Atypical Parkinsonism-Dystonia Syndrome Caused by a Novel DJ1 Mutation. Movement Disorders Clinical Practice, 2014, 1, 45-49. | 1.5 | 8 |
| 123 | Disease-related cortical thinning in presymptomatic granulin mutation carriers. NeuroImage: Clinical, 2021, 29, 102540. | 2.7 | 8 |
| 124 | Genetic variants in glutamate-, AÎ²â^', and tau-related pathways determine polygenic risk for Alzheimer's disease. Neurobiology of Aging, 2021, 101, 299.e13-299.e21. | 3.1 | 7 |
| 125 | Mutations in TYROBP are not a common cause of dementia in a Turkish cohort. Neurobiology of Aging, 2017, 58, 240.e1-240.e3. | 3.1 | 6 |
| 126 | Genetics of dementia in a Finnish cohort. European Journal of Human Genetics, 2018, 26, 827-837. | 2.8 | 6 |

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|---|---|---|---|
| 127 | AP4S1 splice-site mutation in a case of spastic paraplegia type 52 with polymicrogyria. Neurology: Genetics, 2018, 4, e273. | 1.9 | 6 |
| 128 | Genotyping of the Alzheimerâ€™s Disease Genome-Wide Association Study Index Single Nucleotide Polymorphisms in the Brains for Dementia Research Cohort. Journal of Alzheimer's Disease, 2018, 64, 355-362. | 2.6 | 6 |
| 129 | Psychiatric Manifestations of <scp>ATP13A2</scp> Mutations. Movement Disorders Clinical Practice, 2020, 7, 838-841. | 1.5 | 6 |
| 130 | Whole-exome sequencing of Finnish patients with vascular cognitive impairment. European Journal of Human Genetics, 2021, 29, 663-671. | 2.8 | 6 |
| 131 | Genetic analysis reveals novel variants for vascular cognitive impairment. Acta Neurologica Scandinavica, 2022, 146, 42-50. | 2.1 | 6 |
| 132 | Molecular Characterization of Portuguese Patients with Hereditary Cerebellar Ataxia. Cells, 2022, 11, 981. | 4.1 | 6 |
| 133 | A deletion of IDUA exon 10 in a family of Golden Retriever dogs with an attenuated form of mucopolysaccharidosis type I. Journal of Veterinary Internal Medicine, 2020, 34, 1813-1824. | 1.6 | 4 |
| 134 | Mutations in a Sibship with Multifocal Polymyoclonus. Tremor and Other Hyperkinetic Movements, 2017, 7, 452. | 2.0 | 4 |
| 135 | Cognitive composites for genetic frontotemporal dementia: GENFI-Cog. Alzheimer's Research and Therapy, 2022, 14, 10. | 6.2 | 4 |
| 136 | Genetic Variants and Related Biomarkers in Sporadic Alzheimerâ€™s Disease. Current Genetic Medicine Reports, 2015, 3, 19-25. | 1.9 | 3 |
| 137 | Two pathologically confirmed cases of novel mutations in the MAPT gene causing frontotemporal dementia. Neurobiology of Aging, 2020, 87, 141.e15-141.e20. | 3.1 | 3 |
| 138 | Patients with progranulin mutations overlap with the progressive dysexecutive syndrome: towards the definition of a frontoparietal dementia phenotype. Brain Communications, 2020, 2, fcaa126. | 3.3 | 3 |
| 139 | Novel MAG Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. Journal of Clinical Medicine, 2020, 9, 1212. | 2.4 | 3 |
| 140 | Vasculitic peripheral neuropathy in deficiency of adenosine deaminase 2. Neuromuscular Disorders, 2021, 31, 891-895. | 0.6 | 2 |
| 141 | Examining empathy deficits across familial forms of frontotemporal dementia within the GENFI cohort. Cortex, 2022, 150, 12-28. | 2.4 | 2 |
| 142 | Exome Sequencing of a Portuguese Cohort of Frontotemporal Dementia Patients: Looking Into the ALS-FTD Continuum. Frontiers in Neurology, 0, 13, . | 2.4 | 2 |
| 143 | Ataxia with oculomotor apraxia is associated with the DNA damage repair pathway. Movement Disorders, 2017, 32, 720-720. | 3.9 | 1 |
| 144 | Rare variants in TP73 in a frontotemporal dementia cohort link this gene with primary progressive aphasia phenotypes. European Journal of Neurology, 2022, , . | 3.3 | 1 |

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|---|---|---|---|
| 145 | Structural brain splitting is a hallmark of Granulin-related frontotemporal dementia. Neurobiology of Aging, 2022, , . | 3.1 | 1 |
| 146 | The <scp>CBIâ€R</scp> detects early behavioural impairment in genetic frontotemporal dementia. Annals of Clinical and Translational Neurology, 2022, 9, 644-658. | 3.7 | 1 |
| 147 | P2â€158: LINKAGE AND WHOLE GENOME SEQUENCE ANALYSIS OF ALZHEIMER'S DISEASE RESILIENCE AND RISK. Alzheimer's and Dementia, 2016, 12, P675. | 0.8 | 0 |
| 148 | P3-091: Investigating SARM1 Variants in Alzheimerâ€™s Disease Cohorts. , 2016, 12, P855-P855. | | 0 |
| 149 | Mouse models of kufor-rakeb disease link Parkinson's disease closer to neuronal ceroid lipofuscinosis, suggesting lysosomal dysfunction as shared mechanism. Movement Disorders, 2017, 32, 209-209. | 3.9 | 0 |
| 150 | [P3â€“101]: MULTIâ€INFARCT DEMENTIA OF SWEDISH TYPE IS CAUSED BY 3â€™UTR COL4A1 MUTATION. Alzheimer's and Dementia, 2017, 13, P973. | 0.8 | 0 |
| 151 | [P3â€“110]: CALCULATING POLYGENIC RISK FOR INDIVIDUALS WITH SPORADIC EARLY ONSET ALZHEIMER's DISEASE. Alzheimer's and Dementia, 2017, 13, P976. | 0.8 | 0 |
| 152 | [P3â€“111]: NOVEL CANDIDATE GENES FOR DEMENTIA WITH LEWY BODIES. Alzheimer's and Dementia, 2017, 13, P977. | 0.8 | 0 |
| 153 | [P3â€“112]: INVESTIGATING GENETIC VARIATION IN ALZHEIMER's DISEASE USING WHOLEâ€EXOME SEQUENCING. Alzheimer's and Dementia, 2017, 13, P977. | 0.8 | 0 |
| 154 | [F5â€“01â€“02]: GENETICS OF DLB AND RELEVANCE FOR MECHANISMS. Alzheimer's and Dementia, 2017, 13, P1444. | 0.8 | 0 |
| 155 | [P4â€“416]: GENETIC CHARACTERIZATION OF A TURKISH DEMENTIA COHORT: FOCUS ON TYROBP. Alzheimer's and Dementia, 2017, 13, P1490. | 0.8 | 0 |
| 156 | A comprehensive analysis of copy number variation in a Turkish dementia cohort. Human Genomics, 2021, 15, 48. | 2.9 | 0 |