# Enza Maria Valente

List of Publications by Year
in descending order



| 275 | 27,988 | 16451 64 | 6131 159 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 286 | 286 | 286 | 37089 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222. | 9.1 | 4,701 |
| 2 | Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544. | 9.1 | 3,122 |
| 3 | Hereditary Early-Onset Parkinson's Disease Caused by Mutations in PINK1. Science, 2004, 304, 1158-1160. | 12.6 | 3,060 |
| 4 | An SCN9A channelopathy causes congenital inability to experience pain. Nature, 2006, 444, 894-898. | 27.8 | 1,353 |
| 5 | Localization of a Novel Locus for Autosomal Recessive Early-Onset Parkinsonism, PARK6, on Human Chromosome 1p35-p36. American Journal of Human Genetics, 2001, 68, 895-900. | 6.2 | 459 |
| 6 | PINK1 mutations are associated with sporadic earlyâ€onset parkinsonism. Annals of Neurology, 2004, 56, 336-341. | 5.3 | 447 |
| 7 | Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. American Journal of Medical Genetics, Part A, 2015, 167, 296-312. | 1.2 | 447 |
| 8 | Mitochondrial import and enzymatic activity of PINK1 mutants associated to recessive parkinsonism. Human Molecular Genetics, 2005, 14, 3477-3492. | 2.9 | 413 |
| 9 | Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. Nature Genetics, 2009, 41, 1032-1036. | 21.4 | 383 |
| 10 | Parkes Weber syndrome, vein of Galen aneurysmal malformation, and other fast-flow vascular anomalies are caused byRASA1 mutations. Human Mutation, 2008, 29, 959-965. | 2.5 | 382 |
| 11 | Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725. | 6.2 | 375 |
| 12 | Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625. | 21.4 | 368 |
| 13 | Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179. | 6.2 | 352 |
| 14 | Motile and nonâ€motile cilia in human pathology: from function to phenotypes. Journal of Pathology, 2017, 241, 294-309. | 4.5 | 341 |
| 15 | Joubert Syndrome and related disorders. Orphanet Journal of Rare Diseases, 2010, 5, 20. | 2.7 | 325 |
| 16 | Joubert syndrome: congenital cerebellar ataxia with the molar tooth. Lancet Neurology, The, 2013, 12, 894-905. | 10.2 | 307 |
| 17 | Association of LRRK2 exonic variants with susceptibility to Parkinson's disease: a caseâ€“control study. Lancet Neurology, The, 2011, 10, 898-908. | 10.2 | 294 |
| 18 | Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625. | 21.4 | 261 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Translation Initiator EIF4G1 Mutations in Familial Parkinson Disease. American Journal of Human Genetics, 2011, 89, 398-406. | 6.2 | 250 |
| 20 | PINK1 and BECN1 relocalize at mitochondria-associated membranes during mitophagy and promote ER-mitochondria tethering and autophagosome formation. Autophagy, 2017, 13, 654-669. | 9.1 | 249 |
| 21 | The Parkinson-associated protein PINK1 interacts with Beclin1 and promotes autophagy. Cell Death and Differentiation, 2010, 17, 962-974. | 11.2 | 233 |
| 22 | Pallidal stimulation improves pantothenate kinaseâ€“associated neurodegeneration. Annals of Neurology, 2005, 57, 738-741. | 5.3 | 227 |
| 23 | Dystonia. Nature Reviews Disease Primers, 2018, 4, 25. | 30.5 | 223 |
| 24 | Primary cilia in neurodevelopmental disorders. Nature Reviews Neurology, 2014, 10, 27-36. | 10.1 | 215 |
| 25 | Clinical and subclinical dopaminergic dysfunction in PARK6-linked parkinsonism: An18F-dopa PET study. Annals of Neurology, 2002, 52, 849-853. | 5.3 | 192 |
| 26 | AHI1 is required for photoreceptor outer segment development and is a modifier for retinal degeneration in nephronophthisis. Nature Genetics, 2010, 42, 175-180. | 21.4 | 171 |
| 27 | PRRT2 gene mutations. Neurology, 2012, 79, 2115-2121. | 1.1 | 159 |
| 28 | CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199. | 21.4 | 157 |
| 29 | Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38. | 5.3 | 154 |
| 30 | Dopa-responsive dystonia: A clinical and molecular genetic study. Annals of Neurology, 1998, 44, 649-656. | 5.3 | 153 |
| 31 | Deep brain stimulation in myoclonus-dystonia syndrome. Movement Disorders, 2004, 19, 724-727. | 3.9 | 140 |
| 32 | Mutant Pink1 induces mitochondrial dysfunction in a neuronal cell model of Parkinsonâ€™s disease by disturbing calcium flux. Journal of Neurochemistry, 2009, 108, 1561-1574. | 3.9 | 139 |
| 33 | CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113. | 6.2 | 137 |
| 34 | Transcriptional regulator PRDM12 is essential for human pain perception. Nature Genetics, 2015, 47, 803-808. | 21.4 | 137 |
| 35 | The diverse phenotype and genotype of pantothenate kinase-associated neurodegeneration. Neurology, 2005, 64, 1810-1812. | 1.1 | 136 |
| 36 | PINK1 protects against cell death induced by mitochondrial depolarization, by phosphorylating Bcl-xL and impairing its pro-apoptotic cleavage. Cell Death and Differentiation, 2013, 20, 920-930. | 11.2 | 136 |

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| 37 | Diffusion Tensor Imaging in Joubert Syndrome. American Journal of Neuroradiology, 2007, 28, 1929-1933. | 2.4 | 134 |
| 38 | Clinical and Molecular Genetics of Leber's Congenital Amaurosis: A Multicenter Study of Italian Patients. , 2007, 48, 4284. | | 131 |
| 39 | Genotypes and phenotypes of Joubert syndrome and related disorders. European Journal of Medical Genetics, 2008, 51, 1-23. | 1.3 | 127 |
| 40 | AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534. | 5.3 | 125 |
| 41 | Defective temporal processing of sensory stimuli in DYT1 mutation carriers: a new endophenotype of dystonia?. Brain, 2006, 130, 134-142. | 7.6 | 122 |
| 42 | DYT13, a novel primary torsion dystonia locus, maps to chromosome 1p36.13-36.32 in an Italian family with cranial-cervical or upper limb onset. Annals of Neurology, 2001, 49, 362-366. | 5.3 | 118 |
| 43 | A useful electrophysiologic parameter for diagnosis of carpal tunnel syndrome. , 1996, 19, 48-53. | | 114 |
| 44 | A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146. | 2.5 | 113 |
| 45 | Mitochondrial damage-associated inflammation highlights biomarkers in PRKN/PINK1 parkinsonism. Brain, 2020, 143, 3041-3051. | 7.6 | 105 |
| 46 | Distinguishing the four genetic causes of jouberts syndrome-related disorders. Annals of Neurology, 2005, 57, 513-519. | 5.3 | 104 |
| 47 | Park6â€linked parkinsonism occurs in several european families. Annals of Neurology, 2002, 51, 14-18. | 5.3 | 98 |
| 48 | MKS5 and CEP290 Dependent Assembly Pathway of the Ciliary Transition Zone. PLoS Biology, 2016, 14, e1002416. | 5.6 | 98 |
| 49 | Phenotypic spectrum of alpha-synuclein mutations: New insights from patients and cellular models. Parkinsonism and Related Disorders, 2016, 22, S16-S20. | 2.2 | 98 |
| 50 | MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442. | 2.5 | 96 |
| 51 | Independent and joint effects of the MAPT and SNCA genes in Parkinson disease. Annals of Neurology, 2011, 69, 778-792. | 5.3 | 92 |
| 52 | Description, Nomenclature, and Mapping of a Novel Cerebello-Renal Syndrome with the Molar Tooth Malformation. American Journal of Human Genetics, 2003, 73, 663-670. | 6.2 | 91 |
| 53 | The <scp>C</scp>ontursi <scp>F</scp>amily 20 <scp>Y</scp>ears <scp>L</scp>ater: <scp>I</scp>ntrafamilial <scp>P</scp>henotypic <scp>V</scp>ariability of the <scp>SNCA</scp> p.<scp>A</scp>53T <scp>M</scp>utation. Movement Disorders, 2016, 31, 257-258. | 3.9 | 86 |
| 54 | Genetic and clinical heterogeneity in paroxysmal kinesigenic dyskinesia: Evidence for a third EKD gene. Movement Disorders, 2002, 17, 717-725. | 3.9 | 85 |

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|---|---|---|---|
| 55 | Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969. | 12.6 | 84 |
| 56 | GNAO1 encephalopathy. Neurology: Genetics, 2017, 3, e143. | 1.9 | 84 |
| 57 | <scp>GBAâ€Related</scp> Parkinson's Disease: Dissection of Genotypeâ€“Phenotype Correlates in a Large Italian Cohort. Movement Disorders, 2020, 35, 2106-2111. | 3.9 | 83 |
| 58 | Clinical and genetic studies in hereditary spastic paraplegia with thin corpus callosum. Neurology, 2004, 62, 262-268. | 1.1 | 82 |
| 59 | The PINK1 phenotype can be indistinguishable from idiopathic Parkinson disease. Neurology, 2005, 64, 1958-1960. | 1.1 | 81 |
| 60 | The epsilon-sarcoglycan gene in myoclonic syndromes. Neurology, 2005, 64, 737-739. | 1.1 | 80 |
| 61 | CC2D2Amutations in Meckel and Joubert syndromes indicate a genotype-phenotype correlation. Human Mutation, 2009, 30, 1574-1582. | 2.5 | 80 |
| 62 | NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a. | 2.5 | 77 |
| 63 | Mitochondrial dysfunction in Parkinson disease: evidence in mutant PARK2 fibroblasts. Frontiers in Genetics, 2015, 6, 78. | 2.3 | 77 |
| 64 | Joubert syndrome and related disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1879-1888. | 1.8 | 75 |
| 65 | PINK1heterozygous rare variants: prevalence, significance and phenotypic spectrum. Human Mutation, 2008, 29, 565-565. | 2.5 | 74 |
| 66 | A new family with paroxysmal exercise induced dystonia and migraine: a clinical and genetic study. Journal of Neurology, Neurosurgery and Psychiatry, 2000, 68, 609-614. | 1.9 | 72 |
| 67 | Whole gene deletion and splicing mutations expand thePINK1 genotypic spectrum. Human Mutation, 2007, 28, 98-98. | 2.5 | 66 |
| 68 | Biallelic mutations in the 3â€² exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. Nature Genetics, 2017, 49, 457-464. | 21.4 | 66 |
| 69 | Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249. | 1.2 | 66 |
| 70 | Phenotypic characterisation of autosomal recessive PARK6-linked parkinsonism in three unrelated Italian families. Movement Disorders, 2001, 16, 999-1006. | 3.9 | 65 |
| 71 | Infantile ascending hereditary spastic paralysis (IAHSP). Neurology, 2003, 60, 674-682. | 1.1 | 64 |
| 72 | RPGRIP1L mutations are mainly associated with the cerebelloâ€renal phenotype of Joubert syndromeâ€related disorders. Clinical Genetics, 2008, 74, 164-170. | 2.0 | 64 |

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|---|---|---|---|
| 73 | Delineation and Diagnostic Criteria of Oral-Facial-Digital Syndrome Type VI. Orphanet Journal of Rare Diseases, 2012, 7, 4. | 2.7 | 64 |
| 74 | Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078. | 2.8 | 64 |
| 75 | Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602. | 6.0 | 64 |
| 76 | Mutations in B9D1 and MKS1 cause mild Joubert syndrome: expanding the genetic overlap with the lethal ciliopathy Meckel syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 72. | 2.7 | 63 |
| 77 | RFC1 expansions are a common cause of idiopathic sensory neuropathy. Brain, 2021, 144, 1542-1550. | 7.6 | 63 |
| 78 | Impulsive-compulsive behaviors in parkin -associated Parkinson disease. Neurology, 2016, 87, 1436-1441. | 1.1 | 61 |
| 79 | Surgical prognosis in carpal tunnel syndrome: usefulness of a preoperative neurophysiological assessment. Acta Neurologica Scandinavica, 1996, 94, 343-346. | 2.1 | 59 |
| 80 | Analysis of the ?-sarcoglycan gene in familial and sporadic myoclonus-dystonia: Evidence for genetic heterogeneity. Movement Disorders, 2003, 18, 1047-1051. | 3.9 | 58 |
| 81 | Mutation ofPOC1Bin a Severe Syndromic Retinal Ciliopathy. Human Mutation, 2014, 35, 1153-1162. | 2.5 | 57 |
| 82 | A family study on primary blepharospasm. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 77, 252-254. | 1.9 | 56 |
| 83 | Global investigation and meta-analysis of the C9orf72 (G <sub>4</sub> C <sub>2</sub> ) <sub>n</sub> repeat in Parkinson disease. Neurology, 2014, 83, 1906-1913. | 1.1 | 56 |
| 84 | Mutations inCEP120cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615. | 3.2 | 55 |
| 85 | Assessing the role ofDRD5 andDYT1 in two different caseâ€“control series with primary blepharospasm. Movement Disorders, 2007, 22, 162-166. | 3.9 | 54 |
| 86 | Genomeâ€wide association study in musician's dystonia: A risk variant at the arylsulfatase G locus?. Movement Disorders, 2014, 29, 921-927. | 3.9 | 53 |
| 87 | PINK1 in the limelight: multiple functions of an eclectic protein in human health and disease. Journal of Pathology, 2017, 241, 251-263. | 4.5 | 52 |
| 88 | Ataxia, Intellectual Disability, and Ocular Apraxia with Cerebellar Cysts: A New Disease?. Cerebellum, 2014, 13, 79-88. | 2.5 | 50 |
| 89 | Phenotypic variability of PINK1 expression: 12 Years' clinical follow-up of two Italian families. Movement Disorders, 2014, 29, 1561-1566. | 3.9 | 48 |
| 90 | PINK1 homozygous W437X mutation in a patient with apparent dominant transmission of parkinsonism. Movement Disorders, 2006, 21, 1265-1267. | 3.9 | 46 |

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|---|---|---|---|
| 91 | Primary torsion dystonia: the search for genes is not over. Journal of Neurology, Neurosurgery and Psychiatry, 1999, 67, 395-397. | 1.9 | 45 |
| 92 | Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. American Journal of Human Genetics, 2017, 101, 552-563. | 6.2 | 45 |
| 93 | Novel locus for autosomal dominant pure hereditary spastic paraplegia (SPG19) maps to chromosome 9q33-q34. Annals of Neurology, 2002, 51, 681-685. | 5.3 | 44 |
| 94 | Pontine tegmental cap dysplasia: developmental and cognitive outcome in three adolescent patients. Orphanet Journal of Rare Diseases, 2011, 6, 36. | 2.7 | 44 |
| 95 | Familial blepharospasm is inherited as an autosomal dominant trait and relates to a novel unassigned gene. Movement Disorders, 2003, 18, 207-212. | 3.9 | 43 |
| 96 | Spastic paraplegia with thin corpus callosum: description of 20 new families, refinement of the SPG11 locus, candidate gene analysis and evidence of genetic heterogeneity. Neurogenetics, 2006, 7, 149-156. | 1.4 | 43 |
| 97 | Mutation screening of the DYT6/THAP1 gene in Italy. Movement Disorders, 2009, 24, 2424-2427. | 3.9 | 43 |
| 98 | Clinical, neuroradiological and molecular characterization of cerebellar dysplasia with cysts (Porettiâ€“Boltshauser syndrome). European Journal of Human Genetics, 2016, 24, 1262-1267. | 2.8 | 43 |
| 99 | Phenotypic characterization of DYT13 primary torsion dystonia. Movement Disorders, 2004, 19, 200-206. | 3.9 | 40 |
| 100 | Atypical phenotypes and clinical variability in a large Italian family with DYT1-primary torsion dystonia. Movement Disorders, 2006, 21, 1782-1784. | 3.9 | 40 |
| 101 | PINK1 heterozygous mutations induce subtle alterations in dopamineâ€dependent synaptic plasticity. Movement Disorders, 2014, 29, 41-53. | 3.9 | 40 |
| 102 | A novel mutation in the endosomal Na+/H+ exchanger NHE6 (SLC9A6) causes Christianson syndrome with electrical status epilepticus during slow-wave sleep (ESES). Epilepsy Research, 2014, 108, 811-815. | 1.6 | 40 |
| 103 | Nephronophthisis type 1 deletion syndrome with neurological symptoms: Prevalence and significance of the association. Kidney International, 2006, 70, 1342-1347. | 5.2 | 39 |
| 104 | Olfactory dysfunction in Parkinsonism caused by PINK1 mutations. Movement Disorders, 2009, 24, 2350-2357. | 3.9 | 39 |
| 105 | The use of muscle biopsy in the diagnosis of undefined ataxia with cerebellar atrophy in children. European Journal of Paediatric Neurology, 2012, 16, 248-256. | 1.6 | 39 |
| 106 | Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180. | 1.2 | 38 |
| 107 | Mutations in Extracellular Matrix GenesNID1andLAMC1Cause Autosomal Dominant Dandy-Walker Malformation and Occipital Cephaloceles. Human Mutation, 2013, 34, 1075-1079. | 2.5 | 38 |
| 108 | Genotypeâ€“phenotype correlates in Joubert syndrome: A review. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 72-88. | 1.6 | 37 |

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| 109 | Ophthalmological findings in Joubert syndrome. Eye, 2010, 24, 222-225. | 2.1 | 36 |
| 110 | The protective effect of LRRK2 p.R1398H on risk of Parkinson's disease is independent of MAPT and SNCA variants. Neurobiology of Aging, 2014, 35, 266.e5-266.e14. | 3.1 | 36 |
| 111 | Exposure to low-dose rotenone precipitates synaptic plasticity alterations in PINK1 heterozygous knockout mice. Neurobiology of Disease, 2016, 91, 21-36. | 4.4 | 36 |
| 112 | Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092. | 4.5 | 36 |
| 113 | Establishment of stable iPS-derived human neural stem cell lines suitable for cell therapies. Cell Death and Disease, 2018, 9, 937. | 6.3 | 36 |
| 114 | Parkinson Disease Genetics: A â€œContinuumâ€•from Mendelian to Multifactorial Inheritance. Current Molecular Medicine, 2014, 14, 1079-1088. | 1.3 | 36 |
| 115 | Severe infantile hyperkalaemic periodic paralysis and paramyotonia congenita: broadening the clinical spectrum associated with the T704M mutation in SCN4A. Journal of Neurology, Neurosurgery and Psychiatry, 2003, 74, 1339-1341. | 1.9 | 35 |
| 116 | Normal Cognitive Functions in Joubert Syndrome. Neuropediatrics, 2009, 40, 287-290. | 0.6 | 35 |
| 117 | The TOR1A polymorphism rs1182 and the risk of spread in primary blepharospasm. Movement Disorders, 2009, 24, 613-616. | 3.9 | 35 |
| 118 | Molecular pathways in sporadic PD. Parkinsonism and Related Disorders, 2012, 18, S71-S73. | 2.2 | 35 |
| 119 | Phenotypic variability of DYT1-PTD: Does the clinical spectrum include psychogenic dystonia?. Movement Disorders, 2002, 17, 1058-1063. | 3.9 | 34 |
| 120 | PARK6 is a common cause of familial parkinsonism. Neurological Sciences, 2002, 23, s117-s118. | 1.9 | 34 |
| 121 | The Molar Tooth Sign Is Pathognomonic for Joubert Syndrome!. Pediatric Neurology, 2014, 50, e15-e16. | 2.1 | 34 |
| 122 | Clinical features for diagnosis and management of patients with PRDM12 congenital insensitivity to pain. Journal of Medical Genetics, 2016, 53, 533-535. | 3.2 | 34 |
| 123 | Subclinical sensory abnormalities in unaffected PINK1 heterozygotes. Journal of Neurology, 2008, 255, 1372-1377. | 3.6 | 31 |
| 124 | Defining the Epsilonâ€Sarcoglycan (SGCE) Gene Phenotypic Signature in Myoclonusâ€Dystonia: A Reappraisal of Genetic Testing Criteria. Movement Disorders, 2013, 28, 787-794. | 3.9 | 31 |
| 125 | Twenty years on: Myoclonusâ€dystonia and Îµâ€sarcoglycan â€” neurodevelopment, channel, and signaling dysfunction. Movement Disorders, 2019, 34, 1588-1601. | 3.9 | 31 |
| 126 | Role of the dopamine D5 receptor (DRD5) as a susceptibility gene for cervical dystonia. Journal of Neurology, Neurosurgery and Psychiatry, 2003, 74, 665-666. | 1.9 | 30 |

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|---|---|---|---|
| 127 | Populationâ€specific frequencies for LRRK2 susceptibility variants in the genetic epidemiology of Parkinson's disease (GEOâ€PD) consortium. Movement Disorders, 2013, 28, 1740-1744. | 3.9 | 30 |
| 128 | Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. Human Genetics, 2015, 134, 123-126. | 3.8 | 30 |
| 129 | Alphaâ€synuclein gene duplication: Marked intrafamilial variability in two novel pedigrees. Movement Disorders, 2013, 28, 813-817. | 3.9 | 29 |
| 130 | SERCA1 protein expression in muscle of patients with Brody disease and Brody syndrome and in cultured human muscle fibers. Molecular Genetics and Metabolism, 2013, 110, 162-169. | 1.1 | 29 |
| 131 | Italian family with cranial cervical dystonia: Clinical and genetic study. Movement Disorders, 1999, 14, 820-825. | 3.9 | 28 |
| 132 | Frequency and phenotypes of LRRK2 G2019S mutation in Italian patients with Parkinson's disease. Movement Disorders, 2006, 21, 1232-1235. | 3.9 | 28 |
| 133 | Unusual familial presentation of epsilonâ€sarcoglycan gene mutation with falls and writer's cramp. Movement Disorders, 2008, 23, 1913-1915. | 3.9 | 27 |
| 134 | Genetic Issues in the Diagnosis of Dystonias. Frontiers in Neurology, 2013, 4, 34. | 2.4 | 27 |
| 135 | AlteredPLP1splicing causes hypomyelination of early myelinating structures. Annals of Clinical and Translational Neurology, 2015, 2, 648-661. | 3.7 | 27 |
| 136 | Protracted late infantile ceroid lipofuscinosis due to TPP1 mutations: Clinical, molecular and biochemical characterization in three sibs. Journal of the Neurological Sciences, 2015, 356, 65-71. | 0.6 | 27 |
| 137 | A Nonsense Mutation in the Human Homolog of Drosophila rogdi Causes Kohlschutterâ€“Tonz Syndrome. American Journal of Human Genetics, 2012, 90, 708-714. | 6.2 | 26 |
| 138 | Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801. | 1.1 | 26 |
| 139 | Late onset sporadic Parkinson's disease caused by PINK1 mutations: Clinical and functional study. Movement Disorders, 2008, 23, 881-885. | 3.9 | 25 |
| 140 | The syndrome of deafnessâ€dystonia: Clinical and genetic heterogeneity. Movement Disorders, 2013, 28, 795-803. | 3.9 | 25 |
| 141 | Candidate genes for Parkinson disease: Lessons from pathogenesis. Clinica Chimica Acta, 2015, 449, 68-76. | 1.1 | 25 |
| 142 | Infantile and childhood onset <scp>PLA</scp>2G6â€associated neurodegeneration in a large North African cohort. European Journal of Neurology, 2015, 22, 178-186. | 3.3 | 25 |
| 143 | AMBRA1 regulates mitophagy by interacting with ATAD3A and promoting PINK1 stability. Autophagy, 2022, 18, 1752-1762. | 9.1 | 25 |
| 144 | Genome-wide Association and Meta-analysis of Age at Onset in Parkinson Disease. Neurology, 2022, 99, . | 1.1 | 25 |

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| 145 | Expression profiling in peripheral blood reveals signature for penetrance in DYT1 dystonia. Neurobiology of Disease, 2010, 38, 192-200. | 4.4 | 24 |
| 146 | A novel mutation in <scp>NDUFB11</scp> unveils a new clinical phenotype associated with lactic acidosis and sideroblastic anemia. Clinical Genetics, 2017, 91, 441-447. | 2.0 | 24 |
| 147 | Identification of a novel primary torsion dystonia locus (DYT13) on chromosome 1p36 in an Italian family with cranial-cervical or upper limb onset. Neurological Sciences, 2001, 22, 95-96. | 1.9 | 23 |
| 148 | Novel Italian family supports clinical and genetic heterogeneity of primary adult-onset torsion dystonia. Movement Disorders, 2002, 17, 392-397. | 3.9 | 23 |
| 149 | Cohort study of prevalence and phenomenology of tremor in dementia with Lewy bodies. Journal of Neurology, 2013, 260, 1731-1742. | 3.6 | 23 |
| 150 | Conventional magnetic resonance imaging and diffusion tensor imaging studies in children with novel GPR56 mutations: further delineation of a cobblestone-like phenotype. Neurogenetics, 2013, 14, 77-83. | 1.4 | 23 |
| 151 | Joubert syndrome with bilateral polymicrogyria: Clinical and neuropathological findings in two brothers. American Journal of Medical Genetics, Part A, 2009, 149A, 1511-1515. | 1.2 | 22 |
| 152 | FGF17, a gene involved in cerebellar development, is downregulated in a patient with Dandyâ€“Walker malformation carrying a de novo 8p deletion. Neurogenetics, 2011, 12, 241-245. | 1.4 | 22 |
| 153 | Cognitive, adaptive, and behavioral features in Joubert syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3115-3124. | 1.2 | 22 |
| 154 | Novel mutations in the ciliopathy-associated gene CPLANE1 (C5orf42) cause OFD syndrome type VI rather than Joubert syndrome. European Journal of Medical Genetics, 2018, 61, 585-595. | 1.3 | 22 |
| 155 | The spectrum of brainstem malformations associated to mutations of the tubulin genes family: MRI and DTI analysis. European Radiology, 2019, 29, 770-782. | 4.5 | 22 |
| 156 | Profiling the Biochemical Signature of GBAâ€Related Parkinson's Disease in Peripheral Blood Mononuclear Cells. Movement Disorders, 2021, 36, 1267-1272. | 3.9 | 22 |
| 157 | Suprascapular nerve entrapment: Neurophysiological localization in 6 cases. Acta Orthopaedica, 1996, 67, 482-484. | 1.4 | 21 |
| 158 | Low-Rate Repetitive Nerve Stimulation Protocol in an Italian Cohort of Patients Affected by Recessive Myotonia Congenita. Journal of Clinical Neurophysiology, 2011, 28, 39-44. | 1.7 | 21 |
| 159 | Development of SaraHome: A novel, well-accepted, technology-based assessment tool for patients with ataxia. Computer Methods and Programs in Biomedicine, 2020, 188, 105257. | 4.7 | 21 |
| 160 | Heterozygous KIF1A variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. Journal of Medical Genetics, 2021, 58, 475-483. | 3.2 | 21 |
| 161 | GBA Mutations Influence the Release and Pathological Effects of Small Extracellular Vesicles from Fibroblasts of Patients with Parkinsonâ€™s Disease. International Journal of Molecular Sciences, 2021, 22, 2215. | 4.1 | 21 |
| 162 | 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 |

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|---|---|---|---|
| 163 | Multiplex ligationâ€dependent probe amplification assay for simultaneous detection of Parkinson's disease gene rearrangements. Movement Disorders, 2007, 22, 2274-2278. | 3.9 | 20 |
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