Enza Maria Valente

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2682221/publications.pdf

Version: 2024-02-01

275 papers	27,988 citations	16411 64 h-index	6113 159 g-index
286 all docs	286 docs citations	286 times ranked	37089 citing authors

#	Article	IF	Citations
1	Refining the mutational spectrum and geneâ€“phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. Journal of Medical Genetics, 2022, 59, 399-409.	1.5	13
2	ARF1 haploinsufficiency causes periventricular nodular heterotopia with variable clinical expressivity. Journal of Medical Genetics, 2022, 59, 781-784.	1.5	7
3	SUFU haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. Journal of Medical Genetics, 2022, 59, 888-894.	1.5	19
4	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
5	Clinical variability at the mild end of BRAT1 â€related spectrum: Evidence from two families with genotypeâ€“phenotype discordance. Human Mutation, 2022, 43, 67-73.	1.1	9
6	Adult onset familiar dystonia-plus syndrome: A novel presentation of IRF2BPL-associated neurodegeneration. Parkinsonism and Related Disorders, 2022, 94, 22-24.	1.1	6
7	Get Your Molar Tooth Right: Joubert Syndrome Misdiagnosis Unmasked by Whole-Exome Sequencing. Cerebellum, 2022, 21, 1144-1150.	1.4	7
8	AMBRA1 regulates mitophagy by interacting with ATAD3A and promoting PINK1 stability. Autophagy, 2022, 18, 1752-1762.	4.3	25
9	Dairy Intake and Parkinson's Disease: A Mendelian Randomization Study. Movement Disorders, 2022, 37, 857-864.	2.2	15
10	PINK1 Protects against Staurosporine-Induced Apoptosis by Interacting with Beclin1 and Impairing Its Pro-Apoptotic Cleavage. Cells, 2022, 11, 678.	1.8	11
11	Patient-derived cellular models of primary ciliopathies. Journal of Medical Genetics, 2022, , jmedgenet-2021-108315.	1.5	5
12	Genotypeâ€“phenotype correlates in Joubert syndrome: A review. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 72-88.	0.7	37
13	Biallelic mutations in PSMC3IP are associated with secondary amenorrhea: expanding the spectrum of prematureÂovarian insufficiency. Journal of Assisted Reproduction and Genetics, 2022, 39, 1177-1181.	1.2	3
14	An Integrated Phenotypic and Genotypic Approach Reveals a Highâ€Risk Subtype Association for <scp>EBF3</scp> Missense Variants Affecting the Zinc Finger Domain. Annals of Neurology, 2022, 92, 138-153.	2.8	5
15	Genome-wide Association and Meta-analysis of Age at Onset in Parkinson Disease. Neurology, 2022, 99, .	1.5	25
16	Superior Cerebellar Atrophy: An Imaging Clue to Diagnose ITPR1-Related Disorders. International Journal of Molecular Sciences, 2022, 23, 6723.	1.8	4
17	<scp>TWNK</scp> in Parkinson's Disease: A Movement Disorder and Mitochondrial Disease Center Perspective Study. Movement Disorders, 2022, 37, 1938-1943.	2.2	10
18	The Interaction between <scp>HLAâ€DRB1</scp> and Smoking in Parkinson's Disease Revisited. Movement Disorders, 2022, 37, 1929-1937.	2.2	4

#	Article	IF	Citations
19	Heterozygous KIF1A variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. Journal of Medical Genetics, 2021, 58, 475-483.	1.5	21
20	Visual Evoked Potentials in Joubert Syndrome: A Suggested Useful Method for Evaluating Future Approaches Targeted to Improve Visual Pathwaysâ€™ Function. Advances in Therapy, 2021, 38, 278-289.	1.3	0
21	Parkin Deficiency Appears Not to Be Associated with Cardiac Damage in Parkinson's Disease. Movement Disorders, 2021, 36, 271-273.	2.2	4
22	Posterior Cortical Atrophy phenotype in a GBA N370S mutation carrier: a case report. BMC Neurology, 2021, 21, 17.	0.8	3
23	Phenotypic Definition and Genotype-Phenotype Correlates in PMPCA-Related Disease. Applied Sciences (Switzerland), 2021, 11, 748.	1.3	1
24	Genome-Wide DNA Methylation Analysis of a Cohort of 41 Patients Affected by Oculo-Auriculo-Vertebral Spectrum (OAVS). International Journal of Molecular Sciences, 2021, 22, 1190.	1.8	16
25	Profiling the Biochemical Signature of GBAâ€Related Parkinson's Disease in Peripheral Blood Mononuclear Cells. Movement Disorders, 2021, 36, 1267-1272.	2.2	22
26	Novel unconventional variants expand the allelic spectrum of OPHN1 gene. American Journal of Medical Genetics, Part A, 2021, 185, 1575-1581.	0.7	3
27	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.	1.8	21
28	A Clinical-Based Diagnostic Approach to Cerebellar Atrophy in Children. Applied Sciences (Switzerland), 2021, 11, 2333.	1.3	0
29	CASK related disorder: Epilepsy and developmental outcome. European Journal of Paediatric Neurology, 2021, 31, 61-69.	0.7	7
30	Genetic characterization of a cohort with familial parkinsonism and cognitive-behavioral syndrome: A Next Generation Sequencing study. Parkinsonism and Related Disorders, 2021, 84, 82-90.	1.1	2
31	Histologic heterogeneity and syndromic associations of non-ampullary duodenal polyps and superficial mucosal lesions. Digestive and Liver Disease, 2021, 53, 1647-1654.	0.4	1
32	Challenges and resources in adult life with Joubert syndrome: issues from an international classification of functioning (ICF) perspective. Disability and Rehabilitation, 2021, , 1-8.	0.9	1
33	Diagnostic Yield and Cost-Effectiveness of â€œDynamicâ€•Exome Analysis in Epilepsy with Neurodevelopmental Disorders: A Tertiary-Center Experience in Northern Italy. Diagnostics, 2021, 11, 948.	1.3	6
34	Xâ€Linked Parkinsonism: Phenotypic and Genetic Heterogeneity. Movement Disorders, 2021, 36, 1511-1525.	2.2	10
35	RFC1 expansions are a common cause of idiopathic sensory neuropathy. Brain, 2021, 144, 1542-1550.	3.7	63
36	Establishment of three Joubert syndrome-derived induced pluripotent stem cell (iPSC) lines harbouring compound heterozygous mutations in CC2D2A gene. Stem Cell Research, 2021, 54, 102430.	0.3	2

#	Article	IF	Citations
37	Spectrum of Disease Severity in Nonsyndromic Patients With Mutations in the CEP290 Gene: A Multicentric Longitudinal Study. , 2021, 62, 1.		3
38	Preexisting Bipolar Disorder Influences the Subsequent Phenotype of Parkinson's Disease. Movement Disorders, 2021, 36, 2840-2852.	2.2	8
39	LIPAD (LRRK2/Luebeck International Parkinson's Disease) Study Protocol: Deep Phenotyping of an International Genetic Cohort. Frontiers in Neurology, 2021, 12, 710572.	1.1	3
40	Social prediction in pediatric patients with congenital, non-progressive malformations of the cerebellum: From deficits in predicting movements to rehabilitation in virtual reality. Cortex, 2021, 144, 82-98.	1.1	8
41	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. Nephrology Dialysis Transplantation, 2020, 35, 1195-1202.	0.4	15
42	A novel IRF2BPL truncating variant is associated with endolysosomal storage. Molecular Biology Reports, 2020, 47, 711-714.	1.0	16
43	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	0.7	66
44	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi006-A from a patient affected by an autosomal recessive form of long QT syndrome type 1. Stem Cell Research, 2020, 42, 101658.	0.3	4
45	Development of SaraHome: A novel, well-accepted, technology-based assessment tool for patients with ataxia. Computer Methods and Programs in Biomedicine, 2020, 188, 105257.	2.6	21
46	Mitochondrial damage-associated inflammation highlights biomarkers in PRKN/PINK1 parkinsonism. Brain, 2020, 143, 3041-3051.	3.7	105
47	Electroretinographic Assessment in Joubert Syndrome: A Suggested Objective Method to Evaluate the Effectiveness of Future Targeted Treatment. Advances in Therapy, 2020, 37, 3827-3838.	1.3	5
48	<scp>GBAâ€Related</scp> Parkinson's Disease: Dissection of Genotypeâ€“Phenotype Correlates in a Large Italian Cohort. Movement Disorders, 2020, 35, 2106-2111.	2.2	83
49	Mitochondria and Parkinsonâ€™s disease: a complex (III) liaison. Brain, 2020, 143, 3175-3178.	3.7	0
50	KCTD17-related myoclonus-dystonia syndrome: clinical and electrophysiological findings of a patient with atypical late onset. Parkinsonism and Related Disorders, 2020, 78, 129-133.	1.1	5
51	Alazami syndrome: Phenotypic expansion and clinical resemblance to Smithâ€“Lemliâ€“Opitz syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2722-2726.	0.7	3
52	<scp>APP</scp>â€Related Corticobasal Syndrome: Expanding the List of Corticobasal Degeneration Look Alikes. Movement Disorders Clinical Practice, 2020, 7, 849-851.	0.8	6
53	Generation of induced pluripotent stem cell (iPSC) lines from a Joubert syndrome patient with compound heterozygous mutations in C5orf42 gene. Stem Cell Research, 2020, 49, 102007.	0.3	3
54	Clinical and Molecular Characterization of a Novel Progranulin Deletion Associated with Different Phenotypes. Journal of Alzheimer's Disease, 2020, 76, 341-347.	1.2	5

#	Article	IF	Citations
55	Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801.	1.5	26
56	Mechanisms of neurodegeneration in Parkinsonâ€™s disease: keep neurons in the PINK1. Mechanisms of Ageing and Development, 2020, 189, 111277.	2.2	11
57	Movement Disorders in Genetic Pediatric Ataxias. Movement Disorders Clinical Practice, 2020, 7, 383-393.	0.8	3
58	Awareness of rare and genetic neurological diseases among italian neurologist. A national survey. Neurological Sciences, 2020, 41, 1567-1570.	0.9	2
59	PINK1 and Parkin: The odd couple. Neuroscience Research, 2020, 159, 25-33.	1.0	8
60	The spectrum of brainstem malformations associated to mutations of the tubulin genes family: MRI and DTI analysis. European Radiology, 2019, 29, 770-782.	2.3	22
61	Generation of two human induced pluripotent stem cell (hiPSC) lines from a long QT syndrome South African founder population. Stem Cell Research, 2019, 39, 101510.	0.3	3
62	Agenesis of the putamen and globus pallidus caused by recessive mutations in the homeobox gene GSX2. Brain, 2019, 142, 2965-2978.	3.7	12
63	Twenty years on: Myoclonusâ€dystonia and Îµâ€sarcoglycan â€” neurodevelopment, channel, and signaling dysfunction. Movement Disorders, 2019, 34, 1588-1601.	2.2	31
64	Toward the elimination of bias in Pediatric Research. Pediatric Research, 2019, 86, 680-681.	1.1	0
65	Production and characterization of human induced pluripotent stem cells (iPSC) CSSi007-A (4383) from Joubert Syndrome. Stem Cell Research, 2019, 38, 101480.	0.3	3
66	Prenatal findings in oralâ€facialâ€digital syndrome type VI: Report of three cases and literature review. Prenatal Diagnosis, 2019, 39, 652-655.	1.1	1
67	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi005-A from a patient carrying the KCNQ1-R190W mutation. Stem Cell Research, 2019, 37, 101437.	0.3	1
68	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi007-A from a Long QT Syndrome type 1 patient carrier of two common variants in the NOS1AP gene. Stem Cell Research, 2019, 36, 101416.	0.3	2
69	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi004-A from a carrier of the KCNQ1-R594Q mutation. Stem Cell Research, 2019, 37, 101431.	0.3	2
70	LRP10: A novel disease gene bridging Parkinson's disease and dementia with Lewy body. Movement Disorders, 2019, 34, 47-47.	2.2	3
71	Atypical childhood-onset neuroaxonal dystrophy in an Indian girl. Journal of Pediatric Neurosciences, 2019, 14, 90.	0.2	2
72	Solving Mendelian Mysteries: The Non-coding Genome May Hold the Key. Cell, 2018, 172, 889-891.	13.5	9

#	Article	IF	Citations
73	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi002-A from a patient affected by the Jervell and Lange-Nielsen syndrome and carrier of two compound heterozygous mutations on the KCNQ1 gene. Stem Cell Research, 2018, 29, 157-161.	0.3	3
74	Intrafamilial variability in a polish family harbouring a frameshift THAP1 mutation. Journal of the Neurological Sciences, 2018, 388, 158.	0.3	0
75	Production and characterization of human induced pluripotent stem cells (iPSCs) from Joubert Syndrome: CSSi001-A (2850). Stem Cell Research, 2018, 27, 74-77.	0.3	3
76	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi003-A from a patient affected by an autosomal recessive form of Long QT Syndrome type 1. Stem Cell Research, 2018, 29, 170-173.	0.3	6
77	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.	0.7	22
78	Establishment of stable iPS-derived human neural stem cell lines suitable for cell therapies. Cell Death and Disease, 2018, 9, 937.	2.7	36
79	Dystonia. Nature Reviews Disease Primers, 2018, 4, 25.	18.1	223
80	Between SCA5 and SCAR14: delineation of the SPTBN2 p.R480W-associated phenotype. European Journal of Human Genetics, 2018, 26, 928-929.	1.4	17
81	Wholeâ€exome sequencing for variant discovery in blepharospasm. Molecular Genetics & Genomic Medicine, 2018, 6, 601-626.	0.6	20
82	Macular staphyloma in patients affected by Joubert syndrome with retinal dystrophy: a new finding detected by SD-OCT. Documenta Ophthalmologica, 2018, 137, 25-36.	1.0	7
83	Genetics of cerebellar disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 154, 267-286.	1.0	3
84	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.	1.0	24
85	Progressive Supranuclear Palsyâ€“Like Phenotype in a GBA E326K Mutation Carrier. Movement Disorders Clinical Practice, 2017, 4, 444-446.	0.8	14
86	DYT2 screening in early-onset isolated dystonia. European Journal of Paediatric Neurology, 2017, 21, 269-271.	0.7	13
87	Biallelic mutations in the 3â€² exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. Nature Genetics, 2017, 49, 457-464.	9.4	66
88	PINK1 and BECN1 relocalize at mitochondria-associated membranes during mitophagy and promote ER-mitochondria tethering and autophagosome formation. Autophagy, 2017, 13, 654-669.	4.3	249
89	Early-onset head titubation in a child with Poretti-Boltshauser syndrome. Neurology, 2017, 88, 1478-1479.	1.5	5
90	KMT2B: A new twist in dystonia genetics. Movement Disorders, 2017, 32, 529-529.	2.2	2

#	Article	IF	Citations
91	GNAO1 encephalopathy. Neurology: Genetics, 2017, 3, e143.	0.9	84
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.	2.6	45
93	Anterior Mesencephalic Cap Dysplasia: Novel Brain Stem Malformative Features Associated with Joubert Syndrome. American Journal of Neuroradiology, 2017, 38, 2385-2390.	1.2	15
94	Genetic Paradoxes in an Italian Family with <scp>PARK</scp>2 Multiexon Duplication. Movement Disorders Clinical Practice, 2017, 4, 889-892.	0.8	1
95	A novel PMCA3 mutation in an ataxic patient with hypomorphic phosphomannomutase 2 (PMM2) heterozygote mutations: Biochemical characterization of the pump defect. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 3303-3312.	1.8	17
96	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	2.3	36
97	Motile and nonâ€motile cilia in human pathology: from function to phenotypes. Journal of Pathology, 2017, 241, 294-309.	2.1	341
98	PINK1 in the limelight: multiple functions of an eclectic protein in human health and disease. Journal of Pathology, 2017, 241, 251-263.	2.1	52
99	The multiple faces of TOR1A: different inheritance, different phenotype. Brain, 2017, 140, 2764-2767.	3.7	3
100	MKS5 and CEP290 Dependent Assembly Pathway of the Ciliary Transition Zone. PLoS Biology, 2016, 14, e1002416.	2.6	98
101	â€œFork and bracketâ€•syndrome expands the spectrum of SBF1 -related sensory motor polyneuropathies. Neurology: Genetics, 2016, 2, e61.	0.9	11
102	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.	2.2	86
103	Elevated aspartate aminotransferase and lactate dehydrogenase levels are a constant finding in <scp>PLA</scp>2G6â€associated neurodegeneration. European Journal of Neurology, 2016, 23, e24-5.	1.7	2
104	Clinical features for diagnosis and management of patients with PRDM12 congenital insensitivity to pain. Journal of Medical Genetics, 2016, 53, 533-535.	1.5	34
105	Clinical, neuroradiological and molecular characterization of cerebellar dysplasia with cysts (Porettiâ€“Boltshauser syndrome). European Journal of Human Genetics, 2016, 24, 1262-1267.	1.4	43
106	Impulsive-compulsive behaviors in parkin -associated Parkinson disease. Neurology, 2016, 87, 1436-1441.	1.5	61
107	Cognitive, adaptive, and behavioral features in Joubert syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3115-3124.	0.7	22
108	Mutations inCEP120cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615.	1.5	55

#	Article	IF	Citations
109	Very mild features of dysequilibrium syndrome associated with a novel VLDLR missense mutation. Neurogenetics, 2016, 17, 191-195.	0.7	9
110	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	4.3	4,701
111	Exposure to low-dose rotenone precipitates synaptic plasticity alterations in PINK1 heterozygous knockout mice. Neurobiology of Disease, 2016, 91, 21-36.	2.1	36
112	Phenotypic spectrum of alpha-synuclein mutations: New insights from patients and cellular models. Parkinsonism and Related Disorders, 2016, 22, S16-S20.	1.1	98
113	A rare case of cerebellar agenesis: a probabilistic Constrained Spherical Deconvolution tractographic study. Brain Imaging and Behavior, 2016, 10, 158-167.	1.1	13
114	Brain Connectivity Changes in Autosomal Recessive Parkinson Disease: A Model for the Sporadic Form. PLoS ONE, 2016, 11, e0163980.	1.1	10
115	AlteredPLP1splicing causes hypomyelination of early myelinating structures. Annals of Clinical and Translational Neurology, 2015, 2, 648-661.	1.7	27
116	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	2.8	64
117	Transcriptional regulator PRDM12 is essential for human pain perception. Nature Genetics, 2015, 47, 803-808.	9.4	137
118	Candidate genes for Parkinson disease: Lessons from pathogenesis. Clinica Chimica Acta, 2015, 449, 68-76.	0.5	25
119	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.	0.7	447
120	Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. Human Genetics, 2015, 134, 123-126.	1.8	30
121	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.	0.3	27
122	Mitochondrial dysfunction in Parkinson disease: evidence in mutant PARK2 fibroblasts. Frontiers in Genetics, 2015, 6, 78.	1.1	77
123	Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38.	2.8	154
124	Infantile and childhood onset <scp>PLA</scp>2G6â€associated neurodegeneration in a large North African cohort. European Journal of Neurology, 2015, 22, 178-186.	1.7	25
125	Cognitive rehabilitation in a child with Joubert Syndrome: Developmental trends and adaptive changes in a single case report. Research in Developmental Disabilities, 2015, 47, 375-384.	1.2	15
126	Genetics and Molecular Biology of Parkinson Disease. , 2015, , 243-257.		0

#	Article	IF	Citations
127	Primary cilia in neurodevelopmental disorders. Nature Reviews Neurology, 2014, 10, 27-36.	4.9	215
128	Global investigation and meta-analysis of the C9orf72 (G ₄ C ₂) _n repeat in Parkinson disease. Neurology, 2014, 83, 1906-1913.	1.5	56
129	Genomeâ€wide association study in musician's dystonia: A risk variant at the arylsulfatase G locus?. Movement Disorders, 2014, 29, 921-927.	2.2	53
130	Recurrent and fatal akinetic crisis in geneticâ€mitochondrial parkinsonisms. European Journal of Neurology, 2014, 21, 1242-1246.	1.7	14
131	A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	1.1	113
132	Ataxia, Intellectual Disability, and Ocular Apraxia with Cerebellar Cysts: A New Disease?. Cerebellum, 2014, 13, 79-88.	1.4	50
133	The ciliary proteins Meckelin and Jouberin are required for retinoic acid-dependent neural differentiation of mouse embryonic stem cells. Differentiation, 2014, 87, 134-146.	1.0	4
134	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.	1.5	36
135	Mutation ofPOC1Bin a Severe Syndromic Retinal Ciliopathy. Human Mutation, 2014, 35, 1153-1162.	1.1	57
136	PINK1 heterozygous mutations induce subtle alterations in dopamineâ€dependent synaptic plasticity. Movement Disorders, 2014, 29, 41-53.	2.2	40
137	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.	1.2	63
138	Phenotypic variability of PINK1 expression: 12 Years' clinical follow-up of two Italian families. Movement Disorders, 2014, 29, 1561-1566.	2.2	48
139	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.	0.8	40
140	Kohlschutter-Tonz Syndrome: Clinical and Genetic Insights Gained From 16 Cases Deriving From a Close-Knit Village inÂNorthern Israel. Pediatric Neurology, 2014, 50, 421-426.	1.0	17
141	The Molar Tooth Sign Is Pathognomonic for Joubert Syndrome!. Pediatric Neurology, 2014, 50, e15-e16.	1.0	34
142	Parkinson Disease Genetics: A â€œContinuumâ€•from Mendelian to Multifactorial Inheritance. Current Molecular Medicine, 2014, 14, 1079-1088.	0.6	36
143	Dandy-Walker malformation and Wisconsin syndrome: novel cases add further insight into the genotype-phenotype correlations of 3q23q25 deletions. Orphanet Journal of Rare Diseases, 2013, 8, 75.	1.2	19
144	Joubert syndrome and related disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1879-1888.	1.0	75

#	Article	IF	Citations
145	Cohort study of prevalence and phenomenology of tremor in dementia with Lewy bodies. Journal of Neurology, 2013, 260, 1731-1742.	1.8	23
146	Migraine with aura as the predominant phenotype in a family with a PRRT2 mutation. Journal of Neurology, 2013, 260, 656-660.	1.8	17
147	Novel genes and novel pathogenetic mechanisms in adultâ€onset primary dystonia. Movement Disorders, 2013, 28, 440-440.	2.2	1
148	Joubert syndrome: congenital cerebellar ataxia with the molar tooth. Lancet Neurology, The, 2013, 12, 894-905.	4.9	307
149	Populationâ€specific frequencies for LRRK2 susceptibility variants in the genetic epidemiology of Parkinson's disease (GEOâ€PD) consortium. Movement Disorders, 2013, 28, 1740-1744.	2.2	30
150	Alphaâ€synuclein gene duplication: Marked intrafamilial variability in two novel pedigrees. Movement Disorders, 2013, 28, 813-817.	2.2	29
151	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.	0.5	29
152	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.	5.0	136
153	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.	0.7	23
154	Successful subthalamic stimulation, but levodopa-induced dystonia, in a genetic Parkinsonâ€™s disease. Neurological Sciences, 2013, 34, 383-386.	0.9	10
155	A solid quality-control analysis of AB SOLiD short-read sequencing data. Briefings in Bioinformatics, 2013, 14, 684-695.	3.2	8
156	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	1.4	64
157	Clinical utility gene card for: Joubert Syndrome - update 2013. European Journal of Human Genetics, 2013, 21, 1187-1187.	1.4	20
158	Genetic Issues in the Diagnosis of Dystonias. Frontiers in Neurology, 2013, 4, 34.	1.1	27
159	Mutations in Extracellular Matrix GenesNID1andLAMC1Cause Autosomal Dominant Dandy-Walker Malformation and Occipital Cephaloceles. Human Mutation, 2013, 34, 1075-1079.	1.1	38
160	Defining the Epsilonâ€Sarcoglycan (SGCE) Gene Phenotypic Signature in Myoclonusâ€Dystonia: A Reappraisal of Genetic Testing Criteria. Movement Disorders, 2013, 28, 787-794.	2.2	31
161	The syndrome of deafnessâ€dystonia: Clinical and genetic heterogeneity. Movement Disorders, 2013, 28, 795-803.	2.2	25
162	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	6.0	84

#	Article	IF	Citations
163	PRRT2 gene mutations. Neurology, 2012, 79, 2115-2121.	1.5	159
164	Molecular pathways in sporadic PD. Parkinsonism and Related Disorders, 2012, 18, S71-S73.	1.1	35
165	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	9.4	157
166	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	4.3	3,122
167	Lack of sequence variations in THAP1 gene and THAP1-binding sites in TOR1A promoter of DYT1 patients. Movement Disorders, 2012, 27, 917-917.	2.2	3
168	A Nonsense Mutation in the Human Homolog of Drosophila rogdi Causes Kohlschutterâ€“Tonz Syndrome. American Journal of Human Genetics, 2012, 90, 708-714.	2.6	26
169	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.	0.7	39
170	Delineation and Diagnostic Criteria of Oral-Facial-Digital Syndrome Type VI. Orphanet Journal of Rare Diseases, 2012, 7, 4.	1.2	64
171	Mosaic caveolin-3 expression in acquired rippling muscle disease without evidence of myasthenia gravis or acetylcholine receptor autoantibodies. Neuromuscular Disorders, 2011, 21, 194-203.	0.3	16
172	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.	0.9	21
173	Clinical utility gene card for: Joubert syndrome. European Journal of Human Genetics, 2011, 19, 1017-1017.	1.4	0
174	Association of LRRK2 exonic variants with susceptibility to Parkinson's disease: a caseâ€“control study. Lancet Neurology, The, 2011, 10, 898-908.	4.9	294
175	Translation Initiator EIF4G1 Mutations in Familial Parkinson Disease. American Journal of Human Genetics, 2011, 89, 398-406.	2.6	250
176	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.	0.7	22
177	Pontine tegmental cap dysplasia: developmental and cognitive outcome in three adolescent patients. Orphanet Journal of Rare Diseases, 2011, 6, 36.	1.2	44
178	Independent and joint effects of the MAPT and SNCA genes in Parkinson disease. Annals of Neurology, 2011, 69, 778-792.	2.8	92
179	"Gluing" phenotypes together: The case of GLUT1. Neurology, 2011, 77, 934-935.	1.5	3
180	Primary Focal Hyperhidrosis in a New Family Not Linked to Known Loci. Dermatology, 2011, 223, 335-342.	0.9	12

#	Article	IF	Citations
181	Expression profiling in peripheral blood reveals signature for penetrance in DYT1 dystonia. Neurobiology of Disease, 2010, 38, 192-200.	2.1	24
182	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	1.1	77
183	Ophthalmological findings in Joubert syndrome. Eye, 2010, 24, 222-225.	1.1	36
184	The Parkinson-associated protein PINK1 interacts with Beclin1 and promotes autophagy. Cell Death and Differentiation, 2010, 17, 962-974.	5.0	233
185	AHI1 is required for photoreceptor outer segment development and is a modifier for retinal degeneration in nephronophthisis. Nature Genetics, 2010, 42, 175-180.	9.4	171
186	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	9.4	261
187	PINK1 (PARK6) and Parkinson's Disease. Blue Books of Neurology, 2010, 34, 66-82.	0.1	1
188	Joubert Syndrome and related disorders. Orphanet Journal of Rare Diseases, 2010, 5, 20.	1.2	325
189	Advances in the genetics of primary torsion dystonia. F1000 Biology Reports, 2010, 2, .	4.0	6
190	Normal Cognitive Functions in Joubert Syndrome. Neuropediatrics, 2009, 40, 287-290.	0.3	35
191	Joubert syndrome with bilateral polymicrogyria: Clinical and neuropathological findings in two brothers. American Journal of Medical Genetics, Part A, 2009, 149A, 1511-1515.	0.7	22
192	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	0.7	38
193	MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442.	1.1	96
194	CC2D2Amutations in Meckel and Joubert syndromes indicate a genotype-phenotype correlation. Human Mutation, 2009, 30, 1574-1582.	1.1	80
195	The TOR1A polymorphism rs1182 and the risk of spread in primary blepharospasm. Movement Disorders, 2009, 24, 613-616.	2.2	35
196	GIGYF2 variants are not associated with Parkinson's disease in Italy. Movement Disorders, 2009, 24, 1867-1868.	2.2	19
197	Olfactory dysfunction in Parkinsonism caused by PINK1 mutations. Movement Disorders, 2009, 24, 2350-2357.	2.2	39
198	Mutation screening of the DYT6/THAP1 gene in Italy. Movement Disorders, 2009, 24, 2424-2427.	2.2	43

#	Article	IF	Citations
199	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. Nature Genetics, 2009, 41, 1032-1036.	9.4	383
200	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.	2.1	139
201	Brainâ€derived neurotrophic factor and risk for primary adultâ€onset cranialâ€cervical dystonia. European Journal of Neurology, 2009, 16, 949-952.	1.7	17
202	PINK1: one protein, multiple neuroprotective functions. Future Neurology, 2009, 4, 575-590.	0.9	4
203	Prevalence and Phenotypic Spectrum of PINK1 Mutations in Parkinsonâ€™s Disease. European Neurological Review, 2009, 4, 40.	0.5	2
204	Subclinical sensory abnormalities in unaffected PINK1 heterozygotes. Journal of Neurology, 2008, 255, 1372-1377.	1.8	31
205	Unusual familial presentation of epsilonâ€sarcoglycan gene mutation with falls and writer's cramp. Movement Disorders, 2008, 23, 1913-1915.	2.2	27
206	Late onset sporadic Parkinson's disease caused by PINK1 mutations: Clinical and functional study. Movement Disorders, 2008, 23, 881-885.	2.2	25
207	PINK1heterozygous rare variants: prevalence, significance and phenotypic spectrum. Human Mutation, 2008, 29, 565-565.	1.1	74
208	Parkes Weber syndrome, vein of Galen aneurysmal malformation, and other fast-flow vascular anomalies are caused byRASA1 mutations. Human Mutation, 2008, 29, 959-965.	1.1	382
209	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179.	2.6	352
210	RPGRIP1L mutations are mainly associated with the cerebelloâ€renal phenotype of Joubert syndromeâ€related disorders. Clinical Genetics, 2008, 74, 164-170.	1.0	64
211	Genetic testing for paediatric neurological disorders. Lancet Neurology, The, 2008, 7, 1113-1126.	4.9	10
212	Impaired body movement representation in DYT1 mutation carriers. Clinical Neurophysiology, 2008, 119, 1864-1869.	0.7	20
213	Genotypes and phenotypes of Joubert syndrome and related disorders. European Journal of Medical Genetics, 2008, 51, 1-23.	0.7	127
214	Computerized gait analysis of Botulinum Toxin treatment in children with cerebral palsy. Disability and Rehabilitation, 2007, 29, 659-664.	0.9	19
215	Diffusion Tensor Imaging in Joubert Syndrome. American Journal of Neuroradiology, 2007, 28, 1929-1933.	1.2	134
216	Clinical and Molecular Genetics of Leber's Congenital Amaurosis: A Multicenter Study of Italian Patients. , 2007, 48, 4284.		131

#	Article	IF	Citations
217	The Expanding Joubert Spectrum. Neuroradiology Journal, 2007, 20, 421-425.	0.6	1
218	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	2.6	137
219	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	2.6	375
220	Whole gene deletion and splicing mutations expand thePINK1 genotypic spectrum. Human Mutation, 2007, 28, 98-98.	1.1	66
221	Assessing the role ofDRD5 andDYT1 in two different caseâ€“control series with primary blepharospasm. Movement Disorders, 2007, 22, 162-166.	2.2	54
222	Multiplex ligationâ€dependent probe amplification assay for simultaneous detection of Parkinson's disease gene rearrangements. Movement Disorders, 2007, 22, 2274-2278.	2.2	20
223	Heterozygous mutations in genes causing parkinsonism: monogenic disorders go complex. Lancet Neurology, The, 2007, 6, 576-578.	4.9	7
224	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	9.4	368
225	An SCN9A channelopathy causes congenital inability to experience pain. Nature, 2006, 444, 894-898.	13.7	1,353
226	Nephronophthisis type 1 deletion syndrome with neurological symptoms: Prevalence and significance of the association. Kidney International, 2006, 70, 1342-1347.	2.6	39
227	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.	0.7	43
228	Frequency and phenotypes of LRRK2 G2019S mutation in Italian patients with Parkinson's disease. Movement Disorders, 2006, 21, 1232-1235.	2.2	28
229	PINK1 homozygous W437X mutation in a patient with apparent dominant transmission of parkinsonism. Movement Disorders, 2006, 21, 1265-1267.	2.2	46
230	Atypical phenotypes and clinical variability in a large Italian family with DYT1-primary torsion dystonia. Movement Disorders, 2006, 21, 1782-1784.	2.2	40
231	AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534.	2.8	125
232	Defective temporal processing of sensory stimuli in DYT1 mutation carriers: a new endophenotype of dystonia?. Brain, 2006, 130, 134-142.	3.7	122
233	A family study on primary blepharospasm. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 77, 252-254.	0.9	56
234	Mitochondrial import and enzymatic activity of PINK1 mutants associated to recessive parkinsonism. Human Molecular Genetics, 2005, 14, 3477-3492.	1.4	413

#	Article	IF	Citations
235	Distinguishing the four genetic causes of jouberts syndrome-related disorders. Annals of Neurology, 2005, 57, 513-519.	2.8	104
236	Pallidal stimulation improves pantothenate kinase-associated neurodegeneration. Annals of Neurology, 2005, 57, 738-741.	2.8	227
237	A novel family with an unusual early-onset generalized dystonia. Movement Disorders, 2005, 20, 81-86.	2.2	14
238	The PINK1 phenotype can be indistinguishable from idiopathic Parkinson disease. Neurology, 2005, 64, 1958-1960.	1.5	81
239	A Novel Locus for Autosomal Dominant Cone and Coneâ€"Rod Dystrophies Maps to the 6p Gene Cluster of Retinal Dystrophies. , 2005, 46, 3539.		2
240	The epsilon-sarcoglycan gene in myoclonic syndromes. Neurology, 2005, 64, 737-739.	1.5	80
241	The diverse phenotype and genotype of pantothenate kinase-associated neurodegeneration. Neurology, 2005, 64, 1810-1812.	1.5	136
242	Clinical and genetic studies in hereditary spastic paraplegia with thin corpus callosum. Neurology, 2004, 62, 262-268.	1.5	82
243	Autosomal dominant Brody disease cosegregates with a chromosomal (2;7)(p11.2;p12.1) translocation in an Italian family. European Journal of Human Genetics, 2004, 12, 579-583.	1.4	17
244	PINK1 mutations are associated with sporadic early-onset parkinsonism. Annals of Neurology, 2004, 56, 336-341.	2.8	447
245	Phenotypic characterization of DYT13 primary torsion dystonia. Movement Disorders, 2004, 19, 200-206.	2.2	40
246	Deep brain stimulation in myoclonus-dystonia syndrome. Movement Disorders, 2004, 19, 724-727.	2.2	140
247	Opioid binding in DYT1 primary torsion dystonia: An11C-diprenorphine PET study. Movement Disorders, 2004, 19, 1498-1503.	2.2	17
248	Hereditary Early-Onset Parkinson's Disease Caused by Mutations in PINK1. Science, 2004, 304, 1158-1160.	6.0	3,060
249	Familial blepharospasm is inherited as an autosomal dominant trait and relates to a novel unassigned gene. Movement Disorders, 2003, 18, 207-212.	2.2	43
250	Analysis of the ?-sarcoglycan gene in familial and sporadic myoclonus-dystonia: Evidence for genetic heterogeneity. Movement Disorders, 2003, 18, 1047-1051.	2.2	58
251	Description, Nomenclature, and Mapping of a Novel Cerebello-Renal Syndrome with the Molar Tooth Malformation. American Journal of Human Genetics, 2003, 73, 663-670.	2.6	91
252	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.	0.9	35

#	Article	IF	Citations
253	Role of the dopamine D5 receptor (DRD5) as a susceptibility gene for cervical dystonia. Journal of Neurology, Neurosurgery and Psychiatry, 2003, 74, 665-666.	0.9	30
254	Infantile ascending hereditary spastic paralysis (IAHSP). Neurology, 2003, 60, 674-682.	1.5	64
255	Park6-linked parkinsonism occurs in several european families. Annals of Neurology, 2002, 51, 14-18.	2.8	98
256	Novel locus for autosomal dominant pure hereditary spastic paraplegia (SPG19) maps to chromosome 9q33-q34. Annals of Neurology, 2002, 51, 681-685.	2.8	44
257	Clinical and subclinical dopaminergic dysfunction in PARK6-linked parkinsonism: An18F-dopa PET study. Annals of Neurology, 2002, 52, 849-853.	2.8	192
258	Novel Italian family supports clinical and genetic heterogeneity of primary adult-onset torsion dystonia. Movement Disorders, 2002, 17, 392-397.	2.2	23
259	Genetic and clinical heterogeneity in paroxysmal kinesigenic dyskinesia: Evidence for a third EKD gene. Movement Disorders, 2002, 17, 717-725.	2.2	85
260	Phenotypic variability of DYT1-PTD: Does the clinical spectrum include psychogenic dystonia?. Movement Disorders, 2002, 17, 1058-1063.	2.2	34
261	PARK6 is a common cause of familial parkinsonism. Neurological Sciences, 2002, 23, s117-s118.	0.9	34
262	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.	2.6	459
263	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.	0.9	23
264	Phenotypic characterisation of autosomal recessive PARK6-linked parkinsonism in three unrelated Italian families. Movement Disorders, 2001, 16, 999-1006.	2.2	65
265	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.	2.8	118
266	DYT13, a novel primary torsion dystonia locus, maps to chromosome 1p36.13â€“36.32 in an Italian family with cranial-cervical or upper limb onset. , 2001, 49, 362.		3
267	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.	0.9	72
268	Primary torsion dystonia: the search for genes is not over. Journal of Neurology, Neurosurgery and Psychiatry, 1999, 67, 395-397.	0.9	45
269	Italian family with cranial cervical dystonia: Clinical and genetic study. Movement Disorders, 1999, 14, 820-825.	2.2	28
270	Dopa-responsive dystonia: A clinical and molecular genetic study. Annals of Neurology, 1998, 44, 649-656.	2.8	153

#	Article	IF	Citations
271	Suprascapular nerve entrapment: Neurophysiological localization in 6 cases. Acta Orthopaedica, 1996, 67, 482-484.	1.4	21
272	Surgical prognosis in carpal tunnel syndrome: usefulness of a preoperative neurophysiological assessment. Acta Neurologica Scandinavica, 1996, 94, 343-346.	1.0	59
273	A useful electrophysiologic parameter for diagnosis of carpal tunnel syndrome. , 1996, 19, 48-53.		114
274	Letters to the editor. Muscle and Nerve, 1996, 19, 675-681.	1.0	1
275	Low-rate nerve stimulation during regional ischemia in the diagnosis of muscle glycogenosis. Muscle and Nerve, 1996, 19, 1523-1529.	1.0	2