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	Refining the mutational spectrum and geneâ€“phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. Journal of Medical Genetics, 2022, 59, 399-409.	3.2	13
2	ARF1 haploinsufficiency causes periventricular nodular heterotopia with variable clinical expressivity. Journal of Medical Genetics, 2022, 59, 781-784.	3.2	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.	3.2	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.	2.8	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.	2.5	9
6	Adult onset familiar dystonia-plus syndrome: A novel presentation of IRF2BPL-associated neurodegeneration. Parkinsonism and Related Disorders, 2022, 94, 22-24.	2.2	6
7	Get Your Molar Tooth Right: Joubert Syndrome Misdiagnosis Unmasked by Whole-Exome Sequencing. Cerebellum, 2022, 21, 1144-1150.	2.5	7
8	AMBRA1 regulates mitophagy by interacting with ATAD3A and promoting PINK1 stability. Autophagy, 2022, 18, 1752-1762.	9.1	25
9	Dairy Intake and Parkinson's Disease: A Mendelian Randomization Study. Movement Disorders, 2022, 37, 857-864.	3.9	15
10	PINK1 Protects against Staurosporine-Induced Apoptosis by Interacting with Beclin1 and Impairing Its Pro-Apoptotic Cleavage. Cells, 2022, 11, 678.	4.1	11
11	Patient-derived cellular models of primary ciliopathies. Journal of Medical Genetics, 2022, , jmedgenet-2021-108315.	3.2	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.	1.6	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.	2.5	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.	5.3	5
15	Genome-wide Association and Meta-analysis of Age at Onset in Parkinson Disease. Neurology, 2022, 99, .	1.1	25
16	Superior Cerebellar Atrophy: An Imaging Clue to Diagnose ITPR1-Related Disorders. International Journal of Molecular Sciences, 2022, 23, 6723.	4.1	4
17	<scp>TWNK</scp> in Parkinson's Disease: A Movement Disorder and Mitochondrial Disease Center Perspective Study. Movement Disorders, 2022, 37, 1938-1943.	3.9	10
18	The Interaction between <scp>HLAâ€DRB1</scp> and Smoking in Parkinson's Disease Revisited. Movement Disorders, 2022, 37, 1929-1937.	3.9	4

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19	Heterozygous KIF1A variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. Journal of Medical Genetics, 2021, 58, 475-483.	3.2	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.	2.9	0
21	Parkin Deficiency Appears Not to Be Associated with Cardiac Damage in Parkinson's Disease. Movement Disorders, 2021, 36, 271-273.	3.9	4
22	Posterior Cortical Atrophy phenotype in a GBA N370S mutation carrier: a case report. BMC Neurology, 2021, 21, 17.	1.8	3
23	Phenotypic Definition and Genotype-Phenotype Correlates in PMPCA-Related Disease. Applied Sciences (Switzerland), 2021, 11, 748.	2.5	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.	4.1	16
25	Profiling the Biochemical Signature of GBAâ€Related Parkinson's Disease in Peripheral Blood Mononuclear Cells. Movement Disorders, 2021, 36, 1267-1272.	3.9	22
26	Novel unconventional variants expand the allelic spectrum of OPHN1 gene. American Journal of Medical Genetics, Part A, 2021, 185, 1575-1581.	1.2	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.	4.1	21
28	A Clinical-Based Diagnostic Approach to Cerebellar Atrophy in Children. Applied Sciences (Switzerland), 2021, 11, 2333.	2.5	0
29	CASK related disorder: Epilepsy and developmental outcome. European Journal of Paediatric Neurology, 2021, 31, 61-69.	1.6	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.	2.2	2
31	Histologic heterogeneity and syndromic associations of non-ampullary duodenal polyps and superficial mucosal lesions. Digestive and Liver Disease, 2021, 53, 1647-1654.	0.9	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.	1.8	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.	2.6	6
34	Xâ€Linked Parkinsonism: Phenotypic and Genetic Heterogeneity. Movement Disorders, 2021, 36, 1511-1525.	3.9	10
35	RFC1 expansions are a common cause of idiopathic sensory neuropathy. Brain, 2021, 144, 1542-1550.	7.6	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.	0.7	2

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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.	3.9	8
39	LIPAD (LRRK2/Luebeck International Parkinson's Disease) Study Protocol: Deep Phenotyping of an International Genetic Cohort. Frontiers in Neurology, 2021, 12, 710572.	2.4	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.	2.4	8
41	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. Nephrology Dialysis Transplantation, 2020, 35, 1195-1202.	0.7	15
42	A novel IRF2BPL truncating variant is associated with endolysosomal storage. Molecular Biology Reports, 2020, 47, 711-714.	2.3	16
43	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	1.2	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.	0.7	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.	4.7	21
46	Mitochondrial damage-associated inflammation highlights biomarkers in PRKN/PINK1 parkinsonism. Brain, 2020, 143, 3041-3051.	7.6	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.	2.9	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.	3.9	83
49	Mitochondria and Parkinsonâ€™s disease: a complex (III) liaison. Brain, 2020, 143, 3175-3178.	7.6	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.	2.2	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.	1.2	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.	1.5	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.	0.7	3
54	Clinical and Molecular Characterization of a Novel Progranulin Deletion Associated with Different Phenotypes. Journal of Alzheimer's Disease, 2020, 76, 341-347.	2.6	5

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55	Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801.	1.1	26
56	Mechanisms of neurodegeneration in Parkinsonâ€™s disease: keep neurons in the PINK1. Mechanisms of Ageing and Development, 2020, 189, 111277.	4.6	11
57	Movement Disorders in Genetic Pediatric Ataxias. Movement Disorders Clinical Practice, 2020, 7, 383-393.	1.5	3
58	Awareness of rare and genetic neurological diseases among italian neurologist. A national survey. Neurological Sciences, 2020, 41, 1567-1570.	1.9	2
59	PINK1 and Parkin: The odd couple. Neuroscience Research, 2020, 159, 25-33.	1.9	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.	4.5	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.	0.7	3
62	Agenesis of the putamen and globus pallidus caused by recessive mutations in the homeobox gene GSX2. Brain, 2019, 142, 2965-2978.	7.6	12
63	Twenty years on: Myoclonusâ€dystonia and Îµâ€sarcoglycan â€" neurodevelopment, channel, and signaling dysfunction. Movement Disorders, 2019, 34, 1588-1601.	3.9	31
64	Toward the elimination of bias in Pediatric Research. Pediatric Research, 2019, 86, 680-681.	2.3	0
65	Production and characterization of human induced pluripotent stem cells (iPSC) CSSi007-A (4383) from Joubert Syndrome. Stem Cell Research, 2019, 38, 101480.	0.7	3
66	Prenatal findings in oralâ€facialâ€digital syndrome type VI: Report of three cases and literature review. Prenatal Diagnosis, 2019, 39, 652-655.	2.3	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.	0.7	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.	0.7	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.	0.7	2
70	LRP10: A novel disease gene bridging Parkinson's disease and dementia with Lewy body. Movement Disorders, 2019, 34, 47-47.	3.9	3
71	Atypical childhood-onset neuroaxonal dystrophy in an Indian girl. Journal of Pediatric Neurosciences, 2019, 14, 90.	0.3	2
72	Solving Mendelian Mysteries: The Non-coding Genome May Hold the Key. Cell, 2018, 172, 889-891.	28.9	9

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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.	0.7	3
74	Intrafamilial variability in a polish family harbouring a frameshift THAP1 mutation. Journal of the Neurological Sciences, 2018, 388, 158.	0.6	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.	0.7	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.	0.7	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.	1.3	22
78	Establishment of stable iPS-derived human neural stem cell lines suitable for cell therapies. Cell Death and Disease, 2018, 9, 937.	6.3	36
79	Dystonia. Nature Reviews Disease Primers, 2018, 4, 25.	30.5	223
80	Between SCA5 and SCAR14: delineation of the SPTBN2 p.R480W-associated phenotype. European Journal of Human Genetics, 2018, 26, 928-929.	2.8	17
81	Wholeâ€exome sequencing for variant discovery in blepharospasm. Molecular Genetics & Genomic Medicine, 2018, 6, 601-626.	1.2	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.	2.2	7
83	Genetics of cerebellar disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 154, 267-286.	1.8	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.	2.0	24
85	Progressive Supranuclear Palsyâ€“Like Phenotype in a GBA E326K Mutation Carrier. Movement Disorders Clinical Practice, 2017, 4, 444-446.	1.5	14
86	DYT2 screening in early-onset isolated dystonia. European Journal of Paediatric Neurology, 2017, 21, 269-271.	1.6	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.	21.4	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.	9.1	249
89	Early-onset head titubation in a child with Poretti-Boltshauser syndrome. Neurology, 2017, 88, 1478-1479.	1.1	5
90	KMT2B: A new twist in dystonia genetics. Movement Disorders, 2017, 32, 529-529.	3.9	2

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91	GNAO1 encephalopathy. Neurology: Genetics, 2017, 3, e143.	1.9	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.	6.2	45
93	Anterior Mesencephalic Cap Dysplasia: Novel Brain Stem Malformative Features Associated with Joubert Syndrome. American Journal of Neuroradiology, 2017, 38, 2385-2390.	2.4	15
94	Genetic Paradoxes in an Italian Family with <scp>PARK</scp>2 Multiexon Duplication. Movement Disorders Clinical Practice, 2017, 4, 889-892.	1.5	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.	3.8	17
96	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	4.5	36
97	Motile and nonâ€motile cilia in human pathology: from function to phenotypes. Journal of Pathology, 2017, 241, 294-309.	4.5	341
98	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
99	The multiple faces of TOR1A: different inheritance, different phenotype. Brain, 2017, 140, 2764-2767.	7.6	3
100	MKS5 and CEP290 Dependent Assembly Pathway of the Ciliary Transition Zone. PLoS Biology, 2016, 14, e1002416.	5.6	98
101	â€œFork and bracketâ€•syndrome expands the spectrum of SBF1 -related sensory motor polyneuropathies. Neurology: Genetics, 2016, 2, e61.	1.9	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.	3.9	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.	3.3	2
104	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
105	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
106	Impulsive-compulsive behaviors in parkin -associated Parkinson disease. Neurology, 2016, 87, 1436-1441.	1.1	61
107	Cognitive, adaptive, and behavioral features in Joubert syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3115-3124.	1.2	22
108	Mutations inCEP120cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615.	3.2	55

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109	Very mild features of dysequilibrium syndrome associated with a novel VLDLR missense mutation. Neurogenetics, 2016, 17, 191-195.	1.4	9
110	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
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	Phenotypic spectrum of alpha-synuclein mutations: New insights from patients and cellular models. Parkinsonism and Related Disorders, 2016, 22, S16-S20.	2.2	98
113	A rare case of cerebellar agenesis: a probabilistic Constrained Spherical Deconvolution tractographic study. Brain Imaging and Behavior, 2016, 10, 158-167.	2.1	13
114	Brain Connectivity Changes in Autosomal Recessive Parkinson Disease: A Model for the Sporadic Form. PLoS ONE, 2016, 11, e0163980.	2.5	10
115	AlteredPLP1splicing causes hypomyelination of early myelinating structures. Annals of Clinical and Translational Neurology, 2015, 2, 648-661.	3.7	27
116	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	6.0	64
117	Transcriptional regulator PRDM12 is essential for human pain perception. Nature Genetics, 2015, 47, 803-808.	21.4	137
118	Candidate genes for Parkinson disease: Lessons from pathogenesis. Clinica Chimica Acta, 2015, 449, 68-76.	1.1	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.	1.2	447
120	Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. Human Genetics, 2015, 134, 123-126.	3.8	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.	0.6	27
122	Mitochondrial dysfunction in Parkinson disease: evidence in mutant PARK2 fibroblasts. Frontiers in Genetics, 2015, 6, 78.	2.3	77
123	Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38.	5.3	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.	3.3	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.	2.2	15
126	Genetics and Molecular Biology of Parkinson Disease. , 2015, , 243-257.		0

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127	Primary cilia in neurodevelopmental disorders. Nature Reviews Neurology, 2014, 10, 27-36.	10.1	215
128	Global investigation and meta-analysis of the C9orf72 (G ₄ C ₂) _n repeat in Parkinson disease. Neurology, 2014, 83, 1906-1913.	1.1	56
129	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
130	Recurrent and fatal akinetic crisis in geneticâ€mitochondrial parkinsonisms. European Journal of Neurology, 2014, 21, 1242-1246.	3.3	14
131	A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	2.5	113
132	Ataxia, Intellectual Disability, and Ocular Apraxia with Cerebellar Cysts: A New Disease?. Cerebellum, 2014, 13, 79-88.	2.5	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.	1.9	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.	3.1	36
135	Mutation ofPOC1Bin a Severe Syndromic Retinal Ciliopathy. Human Mutation, 2014, 35, 1153-1162.	2.5	57
136	PINK1 heterozygous mutations induce subtle alterations in dopamineâ€dependent synaptic plasticity. Movement Disorders, 2014, 29, 41-53.	3.9	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.	2.7	63
138	Phenotypic variability of PINK1 expression: 12 Years' clinical follow-up of two Italian families. Movement Disorders, 2014, 29, 1561-1566.	3.9	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.	1.6	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.	2.1	17
141	The Molar Tooth Sign Is Pathognomonic for Joubert Syndrome!. Pediatric Neurology, 2014, 50, e15-e16.	2.1	34
142	Parkinson Disease Genetics: A â€œContinuumâ€•from Mendelian to Multifactorial Inheritance. Current Molecular Medicine, 2014, 14, 1079-1088.	1.3	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.	2.7	19
144	Joubert syndrome and related disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1879-1888.	1.8	75

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145	Cohort study of prevalence and phenomenology of tremor in dementia with Lewy bodies. Journal of Neurology, 2013, 260, 1731-1742.	3.6	23
146	Migraine with aura as the predominant phenotype in a family with a PRRT2 mutation. Journal of Neurology, 2013, 260, 656-660.	3.6	17
147	Novel genes and novel pathogenetic mechanisms in adultâ€onset primary dystonia. Movement Disorders, 2013, 28, 440-440.	3.9	1
148	Joubert syndrome: congenital cerebellar ataxia with the molar tooth. Lancet Neurology, The, 2013, 12, 894-905.	10.2	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.	3.9	30
150	Alphaâ€synuclein gene duplication: Marked intrafamilial variability in two novel pedigrees. Movement Disorders, 2013, 28, 813-817.	3.9	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.	1.1	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.	11.2	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.	1.4	23
154	Successful subthalamic stimulation, but levodopa-induced dystonia, in a genetic Parkinsonâ€™s disease. Neurological Sciences, 2013, 34, 383-386.	1.9	10
155	A solid quality-control analysis of AB SOLiD short-read sequencing data. Briefings in Bioinformatics, 2013, 14, 684-695.	6.5	8
156	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
157	Clinical utility gene card for: Joubert Syndrome - update 2013. European Journal of Human Genetics, 2013, 21, 1187-1187.	2.8	20
158	Genetic Issues in the Diagnosis of Dystonias. Frontiers in Neurology, 2013, 4, 34.	2.4	27
159	Mutations in Extracellular Matrix GenesNID1andLAMC1Cause Autosomal Dominant Dandy-Walker Malformation and Occipital Cephaloceles. Human Mutation, 2013, 34, 1075-1079.	2.5	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.	3.9	31
161	The syndrome of deafnessâ€dystonia: Clinical and genetic heterogeneity. Movement Disorders, 2013, 28, 795-803.	3.9	25
162	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	12.6	84

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163	PRRT2 gene mutations. Neurology, 2012, 79, 2115-2121.	1.1	159
164	Molecular pathways in sporadic PD. Parkinsonism and Related Disorders, 2012, 18, S71-S73.	2.2	35
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