Enza Maria Valente

List of Publications by Year
in descending order

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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	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	4.3	4,701
2	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	4.3	3,122
3	Hereditary Early-Onset Parkinson's Disease Caused by Mutations in PINK1. Science, 2004, 304, 1158-1160.	6.0	3,060
4	An SCN9A channelopathy causes congenital inability to experience pain. Nature, 2006, 444, 894-898.	13.7	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.	2.6	459
6	PINK1 mutations are associated with sporadic early-onset parkinsonism. Annals of Neurology, 2004, 56, 336-341.	2.8	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.	0.7	447
8	Mitochondrial import and enzymatic activity of PINK1 mutants associated to recessive parkinsonism. Human Molecular Genetics, 2005, 14, 3477-3492.	1.4	413
9	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
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.	1.1	382
11	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	2.6	375
12	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	9.4	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.	2.6	352
14	Motile and nonâ€motile cilia in human pathology: from function to phenotypes. Journal of Pathology, 2017, 241, 294-309.	2.1	341
15	Joubert Syndrome and related disorders. Orphanet Journal of Rare Diseases, 2010, 5, 20.	1.2	325
16	Joubert syndrome: congenital cerebellar ataxia with the molar tooth. Lancet Neurology, The, 2013, 12, 894-905.	4.9	307
17	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
18	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	9.4	261

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19	Translation Initiator EIF4G1 Mutations in Familial Parkinson Disease. American Journal of Human Genetics, 2011, 89, 398-406.	2.6	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.	4.3	249
21	The Parkinson-associated protein PINK1 interacts with Beclin1 and promotes autophagy. Cell Death and Differentiation, 2010, 17, 962-974.	5.0	233
22	Pallidal stimulation improves pantothenate kinase-associated neurodegeneration. Annals of Neurology, 2005, 57, 738-741.	2.8	227
23	Dystonia. Nature Reviews Disease Primers, 2018, 4, 25.	18.1	223
24	Primary cilia in neurodevelopmental disorders. Nature Reviews Neurology, 2014, 10, 27-36.	4.9	215
25	Clinical and subclinical dopaminergic dysfunction in PARK6-linked parkinsonism: An18F-dopa PET study. Annals of Neurology, 2002, 52, 849-853.	2.8	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.	9.4	171
27	PRRT2 gene mutations. Neurology, 2012, 79, 2115-2121.	1.5	159
28	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	9.4	157
29	Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38.	2.8	154
30	Dopa-responsive dystonia: A clinical and molecular genetic study. Annals of Neurology, 1998, 44, 649-656.	2.8	153
31	Deep brain stimulation in myoclonus-dystonia syndrome. Movement Disorders, 2004, 19, 724-727.	2.2	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.	2.1	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.	2.6	137
34	Transcriptional regulator PRDM12 is essential for human pain perception. Nature Genetics, 2015, 47, 803-808.	9.4	137
35	The diverse phenotype and genotype of pantothenate kinase-associated neurodegeneration. Neurology, 2005, 64, 1810-1812.	1.5	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.	5.0	136

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37	Diffusion Tensor Imaging in Joubert Syndrome. American Journal of Neuroradiology, 2007, 28, 1929-1933.	1.2	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.	0.7	127
40	AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534.	2.8	125
41	Defective temporal processing of sensory stimuli in DYT1 mutation carriers: a new endophenotype of dystonia?. Brain, 2006, 130, 134-142.	3.7	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.	2.8	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.	1.1	113
45	Mitochondrial damage-associated inflammation highlights biomarkers in PRKN/PINK1 parkinsonism. Brain, 2020, 143, 3041-3051.	3.7	105
46	Distinguishing the four genetic causes of jouberts syndrome-related disorders. Annals of Neurology, 2005, 57, 513-519.	2.8	104
47	Park6-linked parkinsonism occurs in several european families. Annals of Neurology, 2002, 51, 14-18.	2.8	98
48	MKS5 and CEP290 Dependent Assembly Pathway of the Ciliary Transition Zone. PLoS Biology, 2016, 14, e1002416.	2.6	98
49	Phenotypic spectrum of alpha-synuclein mutations: New insights from patients and cellular models. Parkinsonism and Related Disorders, 2016, 22, S16-S20.	1.1	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.	1.1	96
51	Independent and joint effects of the MAPT and SNCA genes in Parkinson disease. Annals of Neurology, 2011, 69, 778-792.	2.8	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.	2.6	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.	2.2	86
54	Genetic and clinical heterogeneity in paroxysmal kinesigenic dyskinesia: Evidence for a third EKD gene. Movement Disorders, 2002, 17, 717-725.	2.2	85

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55	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	6.0	84
56	GNAO1 encephalopathy. Neurology: Genetics, 2017, 3, e143.	0.9	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.	2.2	83
58	Clinical and genetic studies in hereditary spastic paraplegia with thin corpus callosum. Neurology, 2004, 62, 262-268.	1.5	82
59	The PINK1 phenotype can be indistinguishable from idiopathic Parkinson disease. Neurology, 2005, 64, 1958-1960.	1.5	81
60	The epsilon-sarcoglycan gene in myoclonic syndromes. Neurology, 2005, 64, 737-739.	1.5	80
61	CC2D2Amutations in Meckel and Joubert syndromes indicate a genotype-phenotype correlation. Human Mutation, 2009, 30, 1574-1582.	1.1	80
62	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	1.1	77
63	Mitochondrial dysfunction in Parkinson disease: evidence in mutant PARK2 fibroblasts. Frontiers in Genetics, 2015, 6, 78.	1.1	77
64	Joubert syndrome and related disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1879-1888.	1.0	75
65	PINK1heterozygous rare variants: prevalence, significance and phenotypic spectrum. Human Mutation, 2008, 29, 565-565.	1.1	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.	0.9	72
67	Whole gene deletion and splicing mutations expand thePINK1 genotypic spectrum. Human Mutation, 2007, 28, 98-98.	1.1	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.	9.4	66
69	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	0.7	66
70	Phenotypic characterisation of autosomal recessive PARK6-linked parkinsonism in three unrelated Italian families. Movement Disorders, 2001, 16, 999-1006.	2.2	65
71	Infantile ascending hereditary spastic paralysis (IAHSP). Neurology, 2003, 60, 674-682.	1.5	64
72	RPGRIP1L mutations are mainly associated with the cerebelloâ€renal phenotype of Joubert syndromeâ€related disorders. Clinical Genetics, 2008, 74, 164-170.	1.0	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.	1.2	64
74	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
75	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	2.8	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.	1.2	63
77	RFC1 expansions are a common cause of idiopathic sensory neuropathy. Brain, 2021, 144, 1542-1550.	3.7	63
78	Impulsive-compulsive behaviors in parkin -associated Parkinson disease. Neurology, 2016, 87, 1436-1441.	1.5	61
79	Surgical prognosis in carpal tunnel syndrome: usefulness of a preoperative neurophysiological assessment. Acta Neurologica Scandinavica, 1996, 94, 343-346.	1.0	59
80	Analysis of the ?-sarcoglycan gene in familial and sporadic myoclonus-dystonia: Evidence for genetic heterogeneity. Movement Disorders, 2003, 18, 1047-1051.	2.2	58
81	Mutation ofPOC1Bin a Severe Syndromic Retinal Ciliopathy. Human Mutation, 2014, 35, 1153-1162.	1.1	57
82	A family study on primary blepharospasm. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 77, 252-254.	0.9	56
83	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
84	Mutations inCEP120cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615.	1.5	55
85	Assessing the role ofDRD5 andDYT1 in two different caseâ€“control series with primary blepharospasm. Movement Disorders, 2007, 22, 162-166.	2.2	54
86	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
87	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
88	Ataxia, Intellectual Disability, and Ocular Apraxia with Cerebellar Cysts: A New Disease?. Cerebellum, 2014, 13, 79-88.	1.4	50
89	Phenotypic variability of PINK1 expression: 12 Years' clinical follow-up of two Italian families. Movement Disorders, 2014, 29, 1561-1566.	2.2	48
90	PINK1 homozygous W437X mutation in a patient with apparent dominant transmission of parkinsonism. Movement Disorders, 2006, 21, 1265-1267.	2.2	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.	0.9	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.	2.6	45
93	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
94	Pontine tegmental cap dysplasia: developmental and cognitive outcome in three adolescent patients. Orphanet Journal of Rare Diseases, 2011, 6, 36.	1.2	44
95	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
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.	0.7	43
97	Mutation screening of the DYT6/THAP1 gene in Italy. Movement Disorders, 2009, 24, 2424-2427.	2.2	43
98	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
99	Phenotypic characterization of DYT13 primary torsion dystonia. Movement Disorders, 2004, 19, 200-206.	2.2	40
100	Atypical phenotypes and clinical variability in a large Italian family with DYT1-primary torsion dystonia. Movement Disorders, 2006, 21, 1782-1784.	2.2	40
101	PINK1 heterozygous mutations induce subtle alterations in dopamineâ€dependent synaptic plasticity. Movement Disorders, 2014, 29, 41-53.	2.2	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.	0.8	40
103	Nephronophthisis type 1 deletion syndrome with neurological symptoms: Prevalence and significance of the association. Kidney International, 2006, 70, 1342-1347.	2.6	39
104	Olfactory dysfunction in Parkinsonism caused by PINK1 mutations. Movement Disorders, 2009, 24, 2350-2357.	2.2	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.	0.7	39
106	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	0.7	38
107	Mutations in Extracellular Matrix GenesNID1andLAMC1Cause Autosomal Dominant Dandy-Walker Malformation and Occipital Cephaloceles. Human Mutation, 2013, 34, 1075-1079.	1.1	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.	0.7	37

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109	Ophthalmological findings in Joubert syndrome. Eye, 2010, 24, 222-225.	1.1	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.	1.5	36
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	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	2.3	36
113	Establishment of stable iPS-derived human neural stem cell lines suitable for cell therapies. Cell Death and Disease, 2018, 9, 937.	2.7	36
114	Parkinson Disease Genetics: A â€œContinuumâ€•from Mendelian to Multifactorial Inheritance. Current Molecular Medicine, 2014, 14, 1079-1088.	0.6	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.	0.9	35
116	Normal Cognitive Functions in Joubert Syndrome. Neuropediatrics, 2009, 40, 287-290.	0.3	35
117	The TOR1A polymorphism rs1182 and the risk of spread in primary blepharospasm. Movement Disorders, 2009, 24, 613-616.	2.2	35
118	Molecular pathways in sporadic PD. Parkinsonism and Related Disorders, 2012, 18, S71-S73.	1.1	35
119	Phenotypic variability of DYT1-PTD: Does the clinical spectrum include psychogenic dystonia?. Movement Disorders, 2002, 17, 1058-1063.	2.2	34
120	PARK6 is a common cause of familial parkinsonism. Neurological Sciences, 2002, 23, s117-s118.	0.9	34
121	The Molar Tooth Sign Is Pathognomonic for Joubert Syndrome!. Pediatric Neurology, 2014, 50, e15-e16.	1.0	34
122	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
123	Subclinical sensory abnormalities in unaffected PINK1 heterozygotes. Journal of Neurology, 2008, 255, 1372-1377.	1.8	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.	2.2	31
125	Twenty years on: Myoclonusâ€dystonia and Îµâ€sarcoglycan â€" neurodevelopment, channel, and signaling dysfunction. Movement Disorders, 2019, 34, 1588-1601.	2.2	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.	0.9	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.	2.2	30
128	Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. Human Genetics, 2015, 134, 123-126.	1.8	30
129	Alphaâ€synuclein gene duplication: Marked intrafamilial variability in two novel pedigrees. Movement Disorders, 2013, 28, 813-817.	2.2	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.	0.5	29
131	Italian family with cranial cervical dystonia: Clinical and genetic study. Movement Disorders, 1999, 14, 820-825.	2.2	28
132	Frequency and phenotypes of LRRK2 G2019S mutation in Italian patients with Parkinson's disease. Movement Disorders, 2006, 21, 1232-1235.	2.2	28
133	Unusual familial presentation of epsilonâ€sarcoglycan gene mutation with falls and writer's cramp. Movement Disorders, 2008, 23, 1913-1915.	2.2	27
134	Genetic Issues in the Diagnosis of Dystonias. Frontiers in Neurology, 2013, 4, 34.	1.1	27
135	AlteredPLP1splicing causes hypomyelination of early myelinating structures. Annals of Clinical and Translational Neurology, 2015, 2, 648-661.	1.7	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.	0.3	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.	2.6	26
138	Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801.	1.5	26
139	Late onset sporadic Parkinson's disease caused by PINK1 mutations: Clinical and functional study. Movement Disorders, 2008, 23, 881-885.	2.2	25
140	The syndrome of deafnessâ€dystonia: Clinical and genetic heterogeneity. Movement Disorders, 2013, 28, 795-803.	2.2	25
141	Candidate genes for Parkinson disease: Lessons from pathogenesis. Clinica Chimica Acta, 2015, 449, 68-76.	0.5	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.	1.7	25
143	AMBRA1 regulates mitophagy by interacting with ATAD3A and promoting PINK1 stability. Autophagy, 2022, 18, 1752-1762.	4.3	25
144	Genome-wide Association and Meta-analysis of Age at Onset in Parkinson Disease. Neurology, 2022, 99, .	1.5	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.	2.1	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.	1.0	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.	0.9	23
148	Novel Italian family supports clinical and genetic heterogeneity of primary adult-onset torsion dystonia. Movement Disorders, 2002, 17, 392-397.	2.2	23
149	Cohort study of prevalence and phenomenology of tremor in dementia with Lewy bodies. Journal of Neurology, 2013, 260, 1731-1742.	1.8	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.	0.7	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.	0.7	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.	0.7	22
153	Cognitive, adaptive, and behavioral features in Joubert syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3115-3124.	0.7	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.	0.7	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.	2.3	22
156	Profiling the Biochemical Signature of GBAâ€Related Parkinson's Disease in Peripheral Blood Mononuclear Cells. Movement Disorders, 2021, 36, 1267-1272.	2.2	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.	0.9	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.	2.6	21
160	Heterozygous KIF1A variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. Journal of Medical Genetics, 2021, 58, 475-483.	1.5	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.	1.8	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.	1.5	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.	2.2	20
164	Impaired body movement representation in DYT1 mutation carriers. Clinical Neurophysiology, 2008, 119, 1864-1869.	0.7	20
165	Clinical utility gene card for: Joubert Syndrome - update 2013. European Journal of Human Genetics, 2013, 21, 1187-1187.	1.4	20
166	Wholeâ€exome sequencing for variant discovery in blepharospasm. Molecular Genetics & Genomic Medicine, 2018, 6, 601-626.	0.6	20
167	Computerized gait analysis of Botulinum Toxin treatment in children with cerebral palsy. Disability and Rehabilitation, 2007, 29, 659-664.	0.9	19
168	GIGYF2 variants are not associated with Parkinson's disease in Italy. Movement Disorders, 2009, 24, 1867-1868.	2.2	19
169	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
170	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
171	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
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