Barbara Garavaglia

List of Publications by Year
in descending order

180 papers	6,891 citations	61984 43 h-index	85541 71 g-index
183 all docs	183 docs citations	183 times ranked	11112 citing authors

#	Article	IF	Citations
1	Exome Sequencing Reveals De Novo WDR45 Mutations Causing a Phenotypically Distinct, X-Linked Dominant Form of NBIA. American Journal of Human Genetics, 2012, 91, 1144-1149.	6.2	309
2	Age-related Iron Deposition in the Basal Ganglia: Quantitative Analysis in Healthy Subjects. Radiology, 2009, 252, 165-172.	7.3	266
3	Beta-propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. Brain, 2013, 136, 1708-1717.	7.6	203
4	Mitochondrial DNA haplogroup K is associated with a lower risk of Parkinson's disease in Italians. European Journal of Human Genetics, 2005, 13, 748-752.	2.8	197
5	Ethylmalonic Encephalopathy Is Caused by Mutations in ETHE1, a Gene Encoding a Mitochondrial Matrix Protein. American Journal of Human Genetics, 2004, 74, 239-252.	6.2	192
6	Genotypic and phenotypic spectrum of PANK2 mutations in patients with neurodegeneration with brain iron accumulation. Annals of Neurology, 2006, 59, 248-256.	5.3	184
7	Exome Sequence Reveals Mutations in CoA Synthase as a Cause of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2014, 94, 11-22.	6.2	176
8	The relevance of gender in Parkinsonâ€™s disease: a review. Journal of Neurology, 2017, 264, 1583-1607.	3.6	171
9	Iron-related MRI images in patients with pantothenate kinase-associated neurodegeneration (PKAN) treated with deferiprone: Results of a phase II pilot trial. Movement Disorders, 2011, 26, 1755-1759.	3.9	125
10	Carnitine stimulation of pyruvate dehydrogenase complex (PDHC) in isolated human skeletal muscle mitochondria. Muscle and Nerve, 1988, 11, 720-724.	2.2	114
11	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	6.2	109
12	Metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations. Molecular Genetics and Metabolism, 2012, 105, 463-471.	1.1	106
13	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	6.2	96
14	Propionylcarnitine excretion in propionic and methylmalonic acidurias: a cause of carnitine deficiency. Clinica Chimica Acta, 1984, 139, 13-21.	1.1	92
15	Isolated limb dystonia as presenting feature of Parkin disease. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 827-828.	1.9	91
16	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. American Journal of Human Genetics, 2016, 99, 1229-1244.	6.2	91
17	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	6.2	88
18	New genes and pathomechanisms in mitochondrial disorders unraveled by NGS technologies. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, 1326-1335.	1.0	87

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19	Mitochondrial dysfunction in Parkinson disease: evidence in mutant PARK2 fibroblasts. Frontiers in Genetics, 2015, 6, 78.	2.3	77
20	Myoclonusâ€“dystonia syndrome: Clinical presentation, disease course, and genetic features in 11 families. Movement Disorders, 2008, 23, 28-34.	3.9	75
21	PINK1heterozygous rare variants: prevalence, significance and phenotypic spectrum. Human Mutation, 2008, 29, 565-565.	2.5	74
22	Coenzyme A corrects pathological defects in human neurons of <scp>PANK</scp> 2â€associated neurodegeneration. EMBO Molecular Medicine, 2016, 8, 1197-1211.	6.9	74
23	SUSCEPTIBILITY TO DYT1 DYSTONIA IN EUROPEAN PATIENTS IS MODIFIED BY THE D216H POLYMORPHISM. Neurology, 2008, 70, 2261-2262.	1.1	73
24	Rhabdomyolysis and acute encephalopathy in late onset medium chain acyl-CoA dehydrogenase deficiency.. Journal of Neurology, Neurosurgery and Psychiatry, 1995, 58, 209-214.	1.9	71
25	Phenotype and genotype variation in primary carnitine deficiency. Genetics in Medicine, 2001, 3, 387-392.	2.4	71
26	Lateâ€onset riboflavinâ€responsive myopathy with combined multiple acyl coenzyme A dehydrogenase and respiratory chain deficiency. Neurology, 1994, 44, 2153-2153.	1.1	71
27	Lossâ€ofâ€Function Variants in <scp>HOPS</scp> Complex Genes <scp>VPS16</scp> and <scp>VPS41</scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. Annals of Neurology, 2020, 88, 867-877.	5.3	70
28	Functional analysis of mutations in the OCTN2 transporter causing primary carnitine deficiency: Lack of genotype-phenotype correlation. Human Mutation, 2000, 16, 401-407.	2.5	69
29	ADCY5-related movement disorders: Frequency, disease course and phenotypic variability in a cohort of paediatric patients. Parkinsonism and Related Disorders, 2017, 41, 37-43.	2.2	67
30	Toward the Standardization of Mitochondrial Proteomics: The Italian Mitochondrial Human Proteome Project Initiative. Journal of Proteome Research, 2017, 16, 4319-4329.	3.7	66
31	Molecular and functional analysis ofSLC25A20 mutations causing carnitine-acylcarnitine translocase deficiency. Human Mutation, 2004, 24, 312-320.	2.5	63
32	The EuroBioBank Network: 10 years of hands-on experience of collaborative, transnational biobanking for rare diseases. European Journal of Human Genetics, 2015, 23, 1116-1123.	2.8	63
33	Hypoparathyroidism in mitochondrial trifunctional protein deficiency. Journal of Pediatrics, 1996, 129, 159-162.	1.8	62
34	Mitochondrial iron and energetic dysfunction distinguish fibroblasts and induced neurons from pantothenate kinase-associated neurodegeneration patients. Neurobiology of Disease, 2015, 81, 144-153.	4.4	61
35	Analysis of the ?-sarcoglycan gene in familial and sporadic myoclonus-dystonia: Evidence for genetic heterogeneity. Movement Disorders, 2003, 18, 1047-1051.	3.9	58
36	Peripheral sensory-motor polyneuropathy, pigmentary retinopathy, and fatal cardiomyopathy in long-chain 3-hydroxy-acyl-CoA dehydrogenase deficiency. European Journal of Pediatrics, 1992, 151, 121-126.	2.7	57

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37	Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A singleâ€center cohort study. Movement Disorders, 2019, 34, 1516-1527.	3.9	55
38	Imaging of dopaminergic dysfunction with [¹²³ I]FP-CIT SPECT in early-onset parkin disease. Neurology, 2004, 63, 2097-2103.	1.1	54
39	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	5.3	54
40	Neonatal dopa-responsive extrapyramidal syndrome in twins with recessive GTPCH deficiency. Neurology, 2003, 60, 335-337.	1.1	50
41	Cavitating leukoencephalopathy with multiple mitochondrial dysfunction syndrome and NFU1 mutations. Frontiers in Genetics, 2014, 5, 412.	2.3	49
42	Syndrome of encephalopathy, petechiae, and ethylmalonic aciduria. Pediatric Neurology, 1997, 17, 165-170.	2.1	48
43	Primary brain calcification: an international study reporting novel variants and associated phenotypes. European Journal of Human Genetics, 2018, 26, 1462-1477.	2.8	48
44	A slowly progressive mitochondrial encephalomyopathy widens the spectrum of AIFM1 disorders. Neurology, 2015, 84, 2193-2195.	1.1	47
45	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. American Journal of Human Genetics, 2020, 106, 412-421.	6.2	47
46	Variant late infantile ceroid lipofuscinoses associated with novel mutations in CLN6. Biochemical and Biophysical Research Communications, 2009, 379, 892-897.	2.1	45
47	cDNA cloning and mitochondrial import of the beta-subunit of the human electron-transfer flavoprotein. FEBS Journal, 1993, 213, 1003-1008.	0.2	44
48	Clinical and molecular heterogeneity in veryâ€“long-chain acyl-coenzyme a dehydrogenase deficiency. Pediatric Neurology, 2000, 22, 98-105.	2.1	44
49	Skin fibroblasts from pantothenate kinase-associated neurodegeneration patients show altered cellular oxidative status and have defective iron-handling properties. Human Molecular Genetics, 2012, 21, 4049-4059.	2.9	44
50	A Map of Human Mitochondrial Protein Interactions Linked to Neurodegeneration Reveals New Mechanisms of Redox Homeostasis and NF-Î¹B Signaling. Cell Systems, 2017, 5, 564-577.e12.	6.2	44
51	Primary carnitine deficiency. Neurology, 1991, 41, 1691-1691.	1.1	44
52	A neurophysiological study of myoclonus in patients with DYT11 myoclonusâ€dystonia syndrome. Movement Disorders, 2008, 23, 2041-2048.	3.9	43
53	Mutation screening of the DYT6/THAP1 gene in Italy. Movement Disorders, 2009, 24, 2424-2427.	3.9	43
54	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43

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55	Parkin analysis in early onset Parkinson's disease. Parkinsonism and Related Disorders, 2008, 14, 326-333.	2.2	42
56	Molecular epidemiology of childhood neuronal ceroid-lipofuscinosis in Italy. Orphanet Journal of Rare Diseases, 2013, 8, 19.	2.7	42
57	Mutations and polymorphisms of the gene encoding the Î²-subunit of the electron transfer flavoprotein in three patients with glutaric acidemia type II. Human Molecular Genetics, 1994, 3, 429-435.	2.9	41
58	Purification and properties of carnitine acetyltransferase from human liver. FEBS Journal, 1990, 189, 539-546.	0.2	40
59	Clinical and neuropsychological correlates in two brothers with pantothenate kinaseâ€“associated neurodegeneration. Movement Disorders, 2005, 20, 208-212.	3.9	40
60	High frequency stimulation of the subthalamic nucleus is efficacious in Parkin disease. Journal of Neurology, 2005, 252, 208-211.	3.6	40
61	Electroclinical spectrum of the neuronal ceroid lipofuscinoses associated with CLN6 mutations. Neurology, 2015, 85, 316-324.	1.1	40
62	The alliance between genetic biobanks and patient organisations: the experience of the telethon network of genetic biobanks. Orphanet Journal of Rare Diseases, 2016, 11, 142.	2.7	40
63	Telethon Network of Genetic Biobanks: a key service for diagnosis and research on rare diseases. Orphanet Journal of Rare Diseases, 2013, 8, 129.	2.7	39
64	Human L-ferritin deficiency is characterized by idiopathic generalized seizures and atypical restless leg syndrome. Journal of Experimental Medicine, 2013, 210, 1779-1791.	8.5	39
65	C19orf12 and FA2H Mutations Are Rare in Italian Patients With Neurodegeneration With Brain Iron Accumulation. Seminars in Pediatric Neurology, 2012, 19, 75-81.	2.0	38
66	Rewiring of the Human Mitochondrial Interactome during Neuronal Reprogramming Reveals Regulators of the Respirasome and Neurogenesis. IScience, 2019, 19, 1114-1132.	4.1	38
67	Frequency of DYT1 mutation in early onset primary dystonia in Italian patients. Movement Disorders, 2002, 17, 407-408.	3.9	36
68	Novel GNAL mutation with intra-familial clinical heterogeneity: Expanding the phenotype. Parkinsonism and Related Disorders, 2016, 23, 66-71.	2.2	35
69	The Role of VPS35 in the Pathobiology of Parkinsonâ€™s Disease. Cellular and Molecular Neurobiology, 2021, 41, 199-227.	3.3	35
70	The â€œEye-of-the-Tigerâ€•Sign may be Absent in the Early Stages of Classic Pantothenate Kinase Associated Neurodegeneration. Neuropediatrics, 2011, 42, 159-162.	0.6	34
71	Clinical and Biochemical Features in a Patient With Mitochondrial Fission Factor Gene Alteration. Frontiers in Genetics, 2018, 9, 625.	2.3	34
72	Mild or absent clinical signs in twin sisters with short-chain acyl-CoA dehydrogenase deficiency. European Journal of Pediatrics, 1998, 157, 317-320.	2.7	33

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73	Encephalopathy, petechiae, and acrocyanosis with ethylmalonic aciduria associated with muscle cytochrome c oxidase deficiency. Journal of Pediatrics, 1994, 125, 843.	1.8	32
74	The relevance of gene panels in movement disorders diagnosis: A lab perspective. European Journal of Paediatric Neurology, 2018, 22, 285-291.	1.6	32
75	KARS-related diseases: progressive leukoencephalopathy with brainstem and spinal cord calcifications as new phenotype and a review of literature. Orphanet Journal of Rare Diseases, 2018, 13, 45.	2.7	32
76	Mitochondrial dysfunction in fibroblasts of Multiple System Atrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3588-3597.	3.8	32
77	<scp>EIF2AK2</scp> Missense Variants Associated with Early Onset Generalized Dystonia. Annals of Neurology, 2021, 89, 485-497.	5.3	32
78	Multisystem triglyceride storage disease is due to a specific defect in the degradation of endocellularly synthesized triglycerides. Neurology, 1988, 38, 1107-1107.	1.1	32
79	A Novel Mutation in the SURF1 Gene in a Child With Leigh Disease, Peripheral Neuropathy, and Cytochrome-c Oxidase Deficiency. Journal of Child Neurology, 2002, 17, 233-236.	1.4	31
80	Phenotype and natural history of variant late infantile ceroidâ€lipofuscinosis 5. Developmental Medicine and Child Neurology, 2017, 59, 815-821.	2.1	31
81	The mitochondrial DNA C3303T mutation can cause cardiomyopathy and/or skeletal myopathy. Journal of Pediatrics, 1999, 135, 197-202.	1.8	30
82	Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-GoutiÃ¨res syndrome. European Journal of Paediatric Neurology, 2016, 20, 604-610.	1.6	29
83	DMT1 Expression and Iron Levels at the Crossroads Between Aging and Neurodegeneration. Frontiers in Neuroscience, 2019, 13, 575.	2.8	29
84	Molecular Genetics and Interferon Signature in the Italian Aicardi GoutiÃ¨res Syndrome Cohort: Report of 12 New Cases and Literature Review. Journal of Clinical Medicine, 2019, 8, 750.	2.4	29
85	Riboflavin-responsive glutaric aciduria type II presenting as a leukodystrophy. Pediatric Neurology, 1995, 13, 333-335.	2.1	28
86	Frequency and phenotypes of LRRK2 G2019S mutation in Italian patients with Parkinson's disease. Movement Disorders, 2006, 21, 1232-1235.	3.9	28
87	TheCACNA1BR1389H variant is not associated with myoclonus-dystonia in a large European multicentric cohort. Human Molecular Genetics, 2015, 24, 5326-5329.	2.9	28
88	Kufs disease due to mutation ofCLN6: clinical, pathological and molecular genetic features. Brain, 2019, 142, 59-69.	7.6	28
89	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
90	Very-long-chain acyl-coenzyme A dehydrogenase deficiency in a child with recurrent myoglobinuria. Neuromuscular Disorders, 1998, 8, 3-6.	0.6	27

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91	GTP-cyclohydrolase I gene mutations in patients with autosomal dominant and recessive GTP-CH1 deficiency: Identification and functional characterization of four novel mutations. Journal of Inherited Metabolic Disease, 2004, 27, 455-463.	3.6	27
92	Late onset sporadic Parkinson's disease caused by PINK1 mutations: Clinical and functional study. Movement Disorders, 2008, 23, 881-885.	3.9	25
93	Mutational analysis of COQ2 in patients with MSA in Italy. Neurobiology of Aging, 2016, 45, 213.e1-213.e2.	3.1	25
94	Pallidal Deep Brain Stimulation in DYT6 Dystonia: Clinical Outcome and Predictive Factors for Motor Improvement. Journal of Clinical Medicine, 2019, 8, 2163.	2.4	25
95	Clinical findings in a patient with FARS2 mutations and earlyâ€infantileâ€encephalopathy with epilepsy. American Journal of Medical Genetics, Part A, 2016, 170, 3004-3007.	1.2	24
96	Neurological Disorders Associated with Striatal Lesions: Classification and Diagnostic Approach. Current Neurology and Neuroscience Reports, 2016, 16, 54.	4.2	23
97	Rare causes of early-onset dystonia-parkinsonism with cognitive impairment: a de novo PSEN-1 mutation. Neurogenetics, 2017, 18, 175-178.	1.4	23
98	Acute, severe cardiomyopathy as main symptom of late-onset very long-chain acyl-coenzyme A dehydrogenase deficiency. European Journal of Pediatrics, 1998, 157, 992-995.	2.7	22
99	Barth syndrome presenting with acute metabolic decompensation in the neonatal period. Journal of Inherited Metabolic Disease, 2006, 29, 684-684.	3.6	22
100	Novel DYT11 gene mutation in patients without dopaminergic deficit (SWEDD) screened for dystonia. Neurology, 2014, 83, 1155-1162.	1.1	22
101	Bilateral striatal necrosis, dystonia and optic atrophy in two siblings.. Journal of Neurology, Neurosurgery and Psychiatry, 1992, 55, 16-19.	1.9	21
102	Muscle cytochromec oxidase deficiency in two Italian patients with ethylmalonic aciduria and peculiar clinical phenotype. Journal of Inherited Metabolic Disease, 1994, 17, 301-303.	3.6	20
103	Medium-chain triglyceride loading test in carnitine-acylcarnitine translocase deficiency: Insights on treatment. Journal of Inherited Metabolic Disease, 1999, 22, 733-739.	3.6	20
104	Mutation Analysis of the GCDH Gene in Italian and Portuguese Patients with Glutaric Aciduria Type I. Molecular Genetics and Metabolism, 2000, 71, 535-537.	1.1	20
105	Multiplex ligationâ€dependent probe amplification assay for simultaneous detection of Parkinson's disease gene rearrangements. Movement Disorders, 2007, 22, 2274-2278.	3.9	20
106	Ferrous Iron Up-regulation in Fibroblasts of Patients with Beta Propeller Protein-Associated Neurodegeneration (BPAN). Frontiers in Genetics, 2017, 8, 18.	2.3	20
107	Substantia Nigra Swelling and Dentate Nucleus T2 Hyperintensity May Be Early Magnetic Resonance Imaging Signs of Î²â€Propeller Proteinâ€Associated Neurodegeneration. Movement Disorders Clinical Practice, 2019, 6, 51-56.	1.5	20
108	A Novel Homozygous YARS2 Mutation in Two Italian Siblings and a Review of Literature. JIMD Reports, 2014, 20, 95-101.	1.5	19

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109	Childhood-onset ATP1A3-related conditions: Report of two new cases of phenotypic spectrum. Parkinsonism and Related Disorders, 2016, 30, 81-82.	2.2	18
110	Patient Affected by Beta-Propeller Protein-Associated Neurodegeneration: A Therapeutic Attempt with Iron Chelation Therapy. Frontiers in Neurology, 2017, 8, 385.	2.4	18
111	Neurophysiological evaluation of motor corticospinal pathways by TMS in idiopathic early-onset Parkinsonâ€™s disease. Clinical Neurophysiology, 2011, 122, 546-549.	1.5	17
112	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. Orphanet Journal of Rare Diseases, 2018, 13, 135.	2.7	17
113	Assessment of the retinal posterior pole in dominant optic atrophy by spectral-domain optical coherence tomography and microperimetry. PLoS ONE, 2017, 12, e0174560.	2.5	17
114	Clinical diagnosis of long-chain acyl-coenzyme A-dehydrogenase deficiency: Use of stress and fat-loading tests. Journal of Pediatrics, 1991, 119, 77-80.	1.8	16
115	<scp>YY1</scp>â€Related Dystonia: Clinical Aspects and Longâ€Term Response to Deep Brain Stimulation. Movement Disorders, 2021, 36, 1461-1462.	3.9	16
116	Mitochondrial Complex III Deficiency Caused by TTC19 Defects: Report of a Novel Mutation and Review of Literature. JIMD Reports, 2015, 22, 115-120.	1.5	15
117	Infantile neuroaxonal dystrophy and pantothenate kinase-associated neurodegeneration. Neurology, 2004, 63, 922-924.	1.1	14
118	Cerebellar and pyramidal dysfunctions, palpebral ptosis and weakness as presenting symptoms of PARKâ€2. Movement Disorders, 2009, 24, 303-305.	3.9	14
119	Clinical and genetic features of paroxysmal kinesigenic dyskinesia in Italian patients. European Journal of Paediatric Neurology, 2016, 20, 152-157.	1.6	14
120	A family with paroxysmal nonkinesigenic dyskinesias (PNKD): Evidence of mitochondrial dysfunction. European Journal of Paediatric Neurology, 2015, 19, 64-68.	1.6	13
121	DYT2 screening in early-onset isolated dystonia. European Journal of Paediatric Neurology, 2017, 21, 269-271.	1.6	13
122	A PDE10A de novo mutation causes childhoodâ€onset chorea with diurnal fluctuations. Movement Disorders, 2017, 32, 1646-1647.	3.9	13
123	Thiamine-responsive disease due to mutation of tpk1 : Importance of avoiding misdiagnosis. Neurology, 2017, 89, 870-871.	1.1	13
124	MYORG -related disease is associated with central pontine calcifications and atypical parkinsonism. Neurology: Genetics, 2020, 6, e399.	1.9	13
125	A Case of Infantile Neuroaxonal Dystrophy of Neonatal Onset. Journal of Child Neurology, 2015, 30, 368-370.	1.4	12
126	Genetics, sex, and gender. Journal of Neuroscience Research, 2023, 101, 553-562.	2.9	12

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127	Early onset primary dystonia. European Journal of Paediatric Neurology, 2009, 13, 488-492.	1.6	11
128	R106C TFG variant causes infantile neuroaxonal dystrophy â€œplusâ€•syndrome. Neurogenetics, 2018, 19, 179-187.	1.4	11
129	Exploring the Impact of PARK2 Mutations on the Total and Mitochondrial Proteome of Human Skin Fibroblasts. Frontiers in Cell and Developmental Biology, 2020, 8, 423.	3.7	11
130	Beneficial effect of sodium dichloroacetate in muscle cytochrome C oxidase deficiency. European Journal of Pediatrics, 1993, 152, 537-537.	2.7	10
131	Clinical and biochemical findings in a Spanish boy with primary carnitine deficiency. Journal of Inherited Metabolic Disease, 1995, 18, 237-240.	3.6	10
132	Expanding the clinical phenotype of DYT5 mutations: Is multiple system atrophy a possible one?. Neurology, 2013, 81, 301-302.	1.1	10
133	Lethal neonatal presentation of carnitine palmitoyltransferase I deficiency. Journal of Inherited Metabolic Disease, 2001, 24, 601-602.	3.6	9
134	Sequence Variations in Mitochondrial Ferritin: Distribution in Healthy Controls and Different Types of Patients. Genetic Testing and Molecular Biomarkers, 2010, 14, 793-796.	0.7	9
135	Long-term follow-up in spastic paraplegia due to SPG56/CYP2U1: age-dependency rather than genetic variability?. Journal of Neurology, 2017, 264, 586-588.	3.6	9
136	Benign hereditary chorea and deletions outside NKX2-1: What's the role of MBIP?. European Journal of Medical Genetics, 2018, 61, 581-584.	1.3	9
137	Diagnosis and treatment of pediatric onset isolated dystonia. European Journal of Paediatric Neurology, 2018, 22, 238-244.	1.6	9
138	Normal cardiovascular reflex testing in patients with parkin disease. Movement Disorders, 2007, 22, 528-532.	3.9	8
139	AOPEP variants as a novel cause of recessive dystonia: Generalized dystonia and dystonia-parkinsonism. Parkinsonism and Related Disorders, 2022, 97, 52-56.	2.2	7
140	MRI Findings in Patients with Clinical Onset Consistent with Infantile Neuroaxonal Dystrophy (INAD), Literature Review, Clinical and MRI Follow-up. Neuroradiology Journal, 2011, 24, 202-214.	1.2	6
141	Early-onset neurodegeneration with brain iron accumulation due to PANK2 mutation. Brain and Development, 2012, 34, 536-538.	1.1	6
142	Novel phenotype in a family with infantile convulsions and paroxysmal choreoathetosis syndrome and PRRT2 gene mutation. Brain and Development, 2014, 36, 183-184.	1.1	6
143	Downbeat nystagmus as the presenting symptom of infantile neuroaxonal dystrophy: A case report. Brain and Development, 2015, 37, 270-272.	1.1	6
144	Fast Progression of Cerebellar Atrophy in PLA2G6-Associated Infantile Neuronal Axonal Dystrophy. Cerebellum, 2017, 16, 742-745.	2.5	6

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145	SLC19A3 related disorder: Treatment implication and clinical outcome of 2 new patients. European Journal of Paediatric Neurology, 2018, 22, 332-335.	1.6	6
146	Restless Legs Syndrome in NKX2-1-related chorea: An expansion of the disease spectrum. Brain and Development, 2019, 41, 250-256.	1.1	6
147	Fatal neonatal outcome in a case of muscular mitochondrial DNA depletion. Journal of Inherited Metabolic Disease, 2000, 23, 755-757.	3.6	5
148	Functional characterization of two novel mutations in TTF-1/NKX2.1 homeodomain in patients with benign hereditary chorea. Journal of the Neurological Sciences, 2016, 360, 78-83.	0.6	5
149	Impulse control behavior in GBA-mutated parkinsonian patients. Journal of the Neurological Sciences, 2021, 421, 117291.	0.6	5
150	Myopathic mitochondrial DNA depletion syndrome associated with biallelic variants in LIG3. Brain, 2021, 144, e74-e74.	7.6	5
151	Parkinson's disease-dementia in trans LRP10 and GBA variants: Response to deep brain stimulation. Parkinsonism and Related Disorders, 2021, 92, 72-75.	2.2	5
152	Disorders of Glycolysis and the Pentose Phosphate Pathway. , 2016, , 149-160.		4
153	Expanding the genetic spectrum of primary familial brain calcification due to SLC20A2 mutations: a case series. Neurogenetics, 2021, 22, 65-70.	1.4	4
154	Pediatric Paroxysmal Exercise-Induced Neurological Symptoms: Clinical Spectrum and Diagnostic Algorithm. Frontiers in Neurology, 2021, 12, 658178.	2.4	4
155	Clinical, molecular and glycophenotype insights in SLC39A8-CDG. Orphanet Journal of Rare Diseases, 2021, 16, 307.	2.7	4
156	A case of Refsum disease with atypical clinical picture in family members. Italian Journal of Neurological Sciences, 1989, 10, 451-454.	0.1	3
157	Cerebrospinal Fluid Monoamine Metabolite Analysis in Pediatric Movement Disorders. Journal of Child Neurology, 2015, 30, 1800-1805.	1.4	3
158	The Neuronal Ceroid Lipofuscinoses: A Case-Based Overview. Journal of Pediatric Biochemistry, 2016, 06, 060-065.	0.2	3
159	Adult diagnosis of Cockayne syndrome. Neurology, 2019, 93, 854-855.	1.1	3
160	<scp>THAP1</scp> Dystonia with Globus Pallidus <scp>T2</scp> Hypointensity: A Report of Two Cases. Movement Disorders, 2021, 36, 1463-1464.	3.9	3
161	Novel deep intronic mutation in PLA2G6 causing early-onset Parkinsonâ€™s disease with brain iron accumulation through pseudo-exon activation. Neurogenetics, 2021, 22, 347-351.	1.4	3
162	Sudden infant death and multiple acyl-CoA dehydrogenation disorders. European Journal of Pediatrics, 1995, 154, 421-422.	2.7	2

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163	Adultâ€Onset Focal Chorea in Fahr's Disease Resulting From <scp>SLC</scp>20A2 Mutation: A Novel Phenotype. Movement Disorders Clinical Practice, 2015, 2, 79-80.	1.5	2
164	Neurodevelopmental disorder and lateâ€onset degenerative parkinsonism in a patient with a WDR45 defect. Movement Disorders Clinical Practice, 2022, 9, 110-112.	1.5	2
165	Impaired degradation of phytanic acid in cells from patients with mitochondriopathies: Evidence for the involvement of ETF and the respiratory chain in phytanic acid ?-oxidation. Journal of Inherited Metabolic Disease, 1994, 17, 527-532.	3.6	1
166	Running apraxia as a presenting symptom of neuronal ceroid lipofuscinosis 6. Movement Disorders, 2014, 29, 277-278.	3.9	1
167	Autonomic dysfunction in Parkinson's disease associated with common glucocerebrosidase gene mutations. Parkinsonism and Related Disorders, 2016, 22, e26.	2.2	1
168	Idiopathic brain calcification in a patient with hereditary hemochromatosis. BMC Neurology, 2020, 20, 113.	1.8	1
169	Governance of Access in Biobanking: The Case of Telethon Network of Genetic Biobanks. Biopreservation and Biobanking, 2021, 19, 483-492.	1.0	1
170	G.P.3.12 Riboflavin-Responsive multiple Acyl-CoA dehydrogenation deficiency (MADD-RR): Clinical, biochemical, molecular genetic and 31 P-MRS studies. Neuromuscular Disorders, 2008, 18, 755.	0.6	0
171	Global metabolic profiling reveals metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations. Mitochondrion, 2012, 12, 577.	3.4	0
172	Establishing a human neuronal derived-iPSC model to clarify the pathogenetic mechanism for PKAN. Journal of the Neurological Sciences, 2015, 357, e43.	0.6	0
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