Barbara Garavaglia

List of Publications by Year
in descending order

180 papers	6,891 citations	61945 43 h-index	85498 71 g-index
183 all docs	183 docs citations	183 times ranked	11112 citing authors

#	Article	IF	Citations
1	Genetics, sex, and gender. Journal of Neuroscience Research, 2023, 101, 553-562.	1.3	12
2	Neurodevelopmental disorder and lateâ€onset degenerative parkinsonism in a patient with a WDR45 defect. Movement Disorders Clinical Practice, 2022, 9, 110-112.	0.8	2
3	AOPEP variants as a novel cause of recessive dystonia: Generalized dystonia and dystonia-parkinsonism. Parkinsonism and Related Disorders, 2022, 97, 52-56.	1.1	7
4	Cerebrospinal fluid neuropathological biomarkers in beta-propeller protein-associated neurodegeneration, with complicated parkinsonian phenotype. Parkinsonism and Related Disorders, 2022, 98, 38-40.	1.1	0
5	The Role of VPS35 in the Pathobiology of Parkinsonâ€™s Disease. Cellular and Molecular Neurobiology, 2021, 41, 199-227.	1.7	35
6	<scp>EIF2AK2</scp> Missense Variants Associated with Early Onset Generalized Dystonia. Annals of Neurology, 2021, 89, 485-497.	2.8	32
7	Expanding the genetic spectrum of primary familial brain calcification due to SLC2OA2 mutations: a case series. Neurogenetics, 2021, 22, 65-70.	0.7	4
8	Impulse control behavior in GBA-mutated parkinsonian patients. Journal of the Neurological Sciences, 2021, 421, 117291.	0.3	5
9	<scp>YY1</scp>â€Related Dystonia: Clinical Aspects and Longâ€Term Response to Deep Brain Stimulation. Movement Disorders, 2021, 36, 1461-1462.	2.2	16
10	<scp>THAP1</scp> Dystonia with Globus Pallidus <scp>T2</scp> Hypointensity: A Report of Two Cases. Movement Disorders, 2021, 36, 1463-1464.	2.2	3
11	Myopathic mitochondrial DNA depletion syndrome associated with biallelic variants in LIG3. Brain, 2021, 144, e74-e74.	3.7	5
12	Pediatric Paroxysmal Exercise-Induced Neurological Symptoms: Clinical Spectrum and Diagnostic Algorithm. Frontiers in Neurology, 2021, 12, 658178.	1.1	4
13	Clinical, molecular and glycophenotype insights in SLC39A8-CDG. Orphanet Journal of Rare Diseases, 2021, 16, 307.	1.2	4
14	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.	0.7	3
15	Multiple Genetic Rare Variants in Autism Spectrum Disorders: A Single-Center Targeted NGS Study. Applied Sciences (Switzerland), 2021, 11, 8096.	1.3	0
16	Clinical and instrumental characterization of GBA-related Parkinson's disease: Focus on cardiovascular and sudomotor autonomic dysfunction and other non-motor features. Does the type of mutation matter?. Journal of the Neurological Sciences, 2021, 429, 117644.	0.3	0
17	Parkinson's disease-dementia in trans LRP10 and GBA variants: Response to deep brain stimulation. Parkinsonism and Related Disorders, 2021, 92, 72-75.	1.1	5
18	Governance of Access in Biobanking: The Case of Telethon Network of Genetic Biobanks. Biopreservation and Biobanking, 2021, 19, 483-492.	0.5	1

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19	Thickness mapping of individual retinal layers and sectors by Spectralis SpectralÂDomainâ€optical Coherence Tomography in Autosomal Dominant Optic Atrophy. Acta Ophthalmologica, 2020, 98, e390.	0.6	0
20	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	3.7	28
21	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.	2.8	70
22	MYORG -related disease is associated with central pontine calcifications and atypical parkinsonism. Neurology: Genetics, 2020, 6, e399.	0.9	13
23	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.	1.8	11
24	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. American Journal of Human Genetics, 2020, 106, 412-421.	2.6	47
25	Idiopathic brain calcification in a patient with hereditary hemochromatosis. BMC Neurology, 2020, 20, 113.	0.8	1
26	Rewiring of the Human Mitochondrial Interactome during Neuronal Reprogramming Reveals Regulators of the Respirasome and Neurogenesis. IScience, 2019, 19, 1114-1132.	1.9	38
27	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	3.7	43
28	Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A singleâ€center cohort study. Movement Disorders, 2019, 34, 1516-1527.	2.2	55
29	DMT1 Expression and Iron Levels at the Crossroads Between Aging and Neurodegeneration. Frontiers in Neuroscience, 2019, 13, 575.	1.4	29
30	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.	1.0	29
31	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	2.8	54
32	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	2.6	88
33	Adult diagnosis of Cockayne syndrome. Neurology, 2019, 93, 854-855.	1.5	3
34	Pallidal Deep Brain Stimulation in DYT6 Dystonia: Clinical Outcome and Predictive Factors for Motor Improvement. Journal of Clinical Medicine, 2019, 8, 2163.	1.0	25
35	Kufs disease due to mutation ofCLN6: clinical, pathological and molecular genetic features. Brain, 2019, 142, 59-69.	3.7	28
36	Restless Legs Syndrome in NKX2-1-related chorea: An expansion of the disease spectrum. Brain and Development, 2019, 41, 250-256.	0.6	6

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37	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.	0.8	20
38	Benign hereditary chorea and deletions outside NKX2-1: What's the role of MBIP?. European Journal of Medical Genetics, 2018, 61, 581-584.	0.7	9
39	The relevance of gene panels in movement disorders diagnosis: A lab perspective. European Journal of Paediatric Neurology, 2018, 22, 285-291.	0.7	32
40	Diagnosis and treatment of pediatric onset isolated dystonia. European Journal of Paediatric Neurology, 2018, 22, 238-244.	0.7	9
41	SLC19A3 related disorder: Treatment implication and clinical outcome of 2 new patients. European Journal of Paediatric Neurology, 2018, 22, 332-335.	0.7	6
42	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.	1.2	32
43	Clinical and Biochemical Features in a Patient With Mitochondrial Fission Factor Gene Alteration. Frontiers in Genetics, 2018, 9, 625.	1.1	34
44	Mitochondrial dysfunction in fibroblasts of Multiple System Atrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3588-3597.	1.8	32
45	R106C TFG variant causes infantile neuroaxonal dystrophy â€œplusâ€•syndrome. Neurogenetics, 2018, 19, 179-187.	0.7	11
46	Primary brain calcification: an international study reporting novel variants and associated phenotypes. European Journal of Human Genetics, 2018, 26, 1462-1477.	1.4	48
47	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. Orphanet Journal of Rare Diseases, 2018, 13, 135.	1.2	17
48	DYT2 screening in early-onset isolated dystonia. European Journal of Paediatric Neurology, 2017, 21, 269-271.	0.7	13
49	Fast Progression of Cerebellar Atrophy in PLA2G6-Associated Infantile Neuronal Axonal Dystrophy. Cerebellum, 2017, 16, 742-745.	1.4	6
50	Long-term follow-up in spastic paraplegia due to SPG56/CYP2U1: age-dependency rather than genetic variability?. Journal of Neurology, 2017, 264, 586-588.	1.8	9
51	ADCY5-related movement disorders: Frequency, disease course and phenotypic variability in a cohort of paediatric patients. Parkinsonism and Related Disorders, 2017, 41, 37-43.	1.1	67
52	Phenotype and natural history of variant late infantile ceroidâ€lipofuscinosis 5. Developmental Medicine and Child Neurology, 2017, 59, 815-821.	1.1	31
53	The relevance of gender in Parkinsonâ€™s disease: a review. Journal of Neurology, 2017, 264, 1583-1607.	1.8	171
54	A PDE10A de novo mutation causes childhoodâ€onset chorea with diurnal fluctuations. Movement Disorders, 2017, 32, 1646-1647.	2.2	13

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55	Toward the Standardization of Mitochondrial Proteomics: The Italian Mitochondrial Human Proteome Project Initiative. Journal of Proteome Research, 2017, 16, 4319-4329.	1.8	66
56	Thiamine-responsive disease due to mutation of tpk1 : Importance of avoiding misdiagnosis. Neurology, 2017, 89, 870-871.	1.5	13
57	A Map of Human Mitochondrial Protein Interactions Linked to Neurodegeneration Reveals New Mechanisms of Redox Homeostasis and NF-Î²B Signaling. Cell Systems, 2017, 5, 564-577.e12.	2.9	44
58	Rare causes of early-onset dystonia-parkinsonism with cognitive impairment: a de novo PSEN-1 mutation. Neurogenetics, 2017, 18, 175-178.	0.7	23
59	Ferrous Iron Up-regulation in Fibroblasts of Patients with Beta Propeller Protein-Associated Neurodegeneration (BPAN). Frontiers in Genetics, 2017, 8, 18.	1.1	20
60	Patient Affected by Beta-Propeller Protein-Associated Neurodegeneration: A Therapeutic Attempt with Iron Chelation Therapy. Frontiers in Neurology, 2017, 8, 385.	1.1	18
61	Assessment of the retinal posterior pole in dominant optic atrophy by spectral-domain optical coherence tomography and microperimetry. PLoS ONE, 2017, 12, e0174560.	1.1	17
62	Novel GNAL mutation with intra-familial clinical heterogeneity: Expanding the phenotype. Parkinsonism and Related Disorders, 2016, 22, e135.	1.1	0
63	Novel GNAL mutation with intra-familial clinical heterogeneity: Expanding the phenotype. Parkinsonism and Related Disorders, 2016, 23, 66-71.	1.1	35
64	Neuropsychological assessment in patients with Parkinson's disease associated with PARK2 gene mutations: A case-control study. Parkinsonism and Related Disorders, 2016, 22, e167-e168.	1.1	0
65	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.	0.7	29
66	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	2.6	96
67	Neurological Disorders Associated with Striatal Lesions: Classification and Diagnostic Approach. Current Neurology and Neuroscience Reports, 2016, 16, 54.	2.0	23
68	The Neuronal Ceroid Lipofuscinoses: A Case-Based Overview. Journal of Pediatric Biochemistry, 2016, 06, 060-065.	0.2	3
69	Mutational analysis of COQ2 in patients with MSA in Italy. Neurobiology of Aging, 2016, 45, 213.e1-213.e2.	1.5	25
70	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.	0.7	24
71	Coenzyme A corrects pathological defects in human neurons of <scp>PANK</scp> 2â€associated neurodegeneration. EMBO Molecular Medicine, 2016, 8, 1197-1211.	3.3	74
72	Childhood-onset ATP1A3-related conditions: Report of two new cases of phenotypic spectrum. Parkinsonism and Related Disorders, 2016, 30, 81-82.	1.1	18

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73	Disorders of Glycolysis and the Pentose Phosphate Pathway. , 2016, , 149-160.		4
74	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. American Journal of Human Genetics, 2016, 99, 1229-1244.	2.6	91
75	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.	1.2	40
76	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.	0.3	5
77	Autonomic dysfunction in Parkinson's disease associated with common glucocerebrosidase gene mutations. Parkinsonism and Related Disorders, 2016, 22, e44.	1.1	0
78	Autonomic dysfunction in Parkinson's disease associated with common glucocerebrosidase gene mutations. Parkinsonism and Related Disorders, 2016, 22, e26.	1.1	1
79	Novel GNAL mutation with intra-familial clinical heterogeneity: Expanding the phenotype. Parkinsonism and Related Disorders, 2016, 22, e77.	1.1	0
80	New genes and pathomechanisms in mitochondrial disorders unraveled by NGS technologies. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, 1326-1335.	0.5	87
81	Clinical and genetic features of paroxysmal kinesigenic dyskinesia in Italian patients. European Journal of Paediatric Neurology, 2016, 20, 152-157.	0.7	14
82	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.	0.8	2
83	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	2.6	109
84	Establishing a human neuronal derived-iPSC model to clarify the pathogenetic mechanism for PKAN. Journal of the Neurological Sciences, 2015, 357, e43.	0.3	0
85	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.	1.4	63
86	Mitochondrial dysfunction in Parkinson disease: evidence in mutant PARK2 fibroblasts. Frontiers in Genetics, 2015, 6, 78.	1.1	77
87	Mitochondrial Complex III Deficiency Caused by TTC19 Defects: Report of a Novel Mutation and Review of Literature. JIMD Reports, 2015, 22, 115-120.	0.7	15
88	A slowly progressive mitochondrial encephalomyopathy widens the spectrum of AIFM1 disorders. Neurology, 2015, 84, 2193-2195.	1.5	47
89	Cerebrospinal Fluid Monoamine Metabolite Analysis in Pediatric Movement Disorders. Journal of Child Neurology, 2015, 30, 1800-1805.	0.7	3
90	TheCACNA1BR1389H variant is not associated with myoclonus-dystonia in a large European multicentric cohort. Human Molecular Genetics, 2015, 24, 5326-5329.	1.4	28

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91	Electroclinical spectrum of the neuronal ceroid lipofuscinoses associated with CLN6 mutations. Neurology, 2015, 85, 316-324.	1.5	40
92	Mitochondrial iron and energetic dysfunction distinguish fibroblasts and induced neurons from pantothenate kinase-associated neurodegeneration patients. Neurobiology of Disease, 2015, 81, 144-153.	2.1	61
93	A family with paroxysmal nonkinesigenic dyskinesias (PNKD): Evidence of mitochondrial dysfunction. European Journal of Paediatric Neurology, 2015, 19, 64-68.	0.7	13
94	Downbeat nystagmus as the presenting symptom of infantile neuroaxonal dystrophy: A case report. Brain and Development, 2015, 37, 270-272.	0.6	6
95	A Case of Infantile Neuroaxonal Dystrophy of Neonatal Onset. Journal of Child Neurology, 2015, 30, 368-370.	0.7	12
96	A Novel Homozygous YARS2 Mutation in Two Italian Siblings and a Review of Literature. JIMD Reports, 2014, 20, 95-101.	0.7	19
97	Cavitating leukoencephalopathy with multiple mitochondrial dysfunction syndrome and NFU1 mutations. Frontiers in Genetics, 2014, 5, 412.	1.1	49
98	Running apraxia as a presenting symptom of neuronal ceroid lipofuscinosis 6. Movement Disorders, 2014, 29, 277-278.	2.2	1
99	Novel DYT11 gene mutation in patients without dopaminergic deficit (SWEDD) screened for dystonia. Neurology, 2014, 83, 1155-1162.	1.5	22
100	Novel phenotype in a family with infantile convulsions and paroxysmal choreoathetosis syndrome and PRRT2 gene mutation. Brain and Development, 2014, 36, 183-184.	0.6	6
101	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.	2.6	176
102	Isolated limb dystonia as presenting feature of Parkin disease. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 827-828.	0.9	91
103	Molecular epidemiology of childhood neuronal ceroid-lipofuscinosis in Italy. Orphanet Journal of Rare Diseases, 2013, 8, 19.	1.2	42
104	Telethon Network of Genetic Biobanks: a key service for diagnosis and research on rare diseases. Orphanet Journal of Rare Diseases, 2013, 8, 129.	1.2	39
105	Beta-propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. Brain, 2013, 136, 1708-1717.	3.7	203
106	Human L-ferritin deficiency is characterized by idiopathic generalized seizures and atypical restless leg syndrome. Journal of Experimental Medicine, 2013, 210, 1779-1791.	4.2	39
107	Expanding the clinical phenotype of DYT5 mutations: Is multiple system atrophy a possible one?. Neurology, 2013, 81, 301-302.	1.5	10
108	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.	1.4	44

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109	Metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations. Molecular Genetics and Metabolism, 2012, 105, 463-471.	0.5	106
110	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.	2.6	309
111	Global metabolic profiling reveals metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations. Mitochondrion, 2012, 12, 577.	1.6	0
112	Early-onset neurodegeneration with brain iron accumulation due to PANK2 mutation. Brain and Development, 2012, 34, 536-538.	0.6	6
113	C19orf12 and FA2H Mutations Are Rare in Italian Patients With Neurodegeneration With Brain Iron Accumulation. Seminars in Pediatric Neurology, 2012, 19, 75-81.	1.0	38
114	Neurophysiological evaluation of motor corticospinal pathways by TMS in idiopathic early-onset Parkinsonâ€™s disease. Clinical Neurophysiology, 2011, 122, 546-549.	0.7	17
115	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.	2.2	125
116	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.	0.3	34
117	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.	0.6	6
118	Sequence Variations in Mitochondrial Ferritin: Distribution in Healthy Controls and Different Types of Patients. Genetic Testing and Molecular Biomarkers, 2010, 14, 793-796.	0.3	9
119	Age-related Iron Deposition in the Basal Ganglia: Quantitative Analysis in Healthy Subjects. Radiology, 2009, 252, 165-172.	3.6	266
120	Early onset primary dystonia. European Journal of Paediatric Neurology, 2009, 13, 488-492.	0.7	11
121	Cerebellar and pyramidal dysfunctions, palpebral ptosis and weakness as presenting symptoms of PARKâ€2. Movement Disorders, 2009, 24, 303-305.	2.2	14
122	Mutation screening of the DYT6/THAP1 gene in Italy. Movement Disorders, 2009, 24, 2424-2427.	2.2	43
123	Variant late infantile ceroid lipofuscinoses associated with novel mutations in CLN6. Biochemical and Biophysical Research Communications, 2009, 379, 892-897.	1.0	45
124	Myoclonusâ€“dystonia syndrome: Clinical presentation, disease course, and genetic features in 11 families. Movement Disorders, 2008, 23, 28-34.	2.2	75
125	Late onset sporadic Parkinson's disease caused by PINK1 mutations: Clinical and functional study. Movement Disorders, 2008, 23, 881-885.	2.2	25
126	A neurophysiological study of myoclonus in patients with DYT11 myoclonusâ€dystonia syndrome. Movement Disorders, 2008, 23, 2041-2048.	2.2	43

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127	PINK1heterozygous rare variants: prevalence, significance and phenotypic spectrum. Human Mutation, 2008, 29, 565-565.	1.1	74
128	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.	0.3	0
129	Parkin analysis in early onset Parkinson's disease. Parkinsonism and Related Disorders, 2008, 14, 326-333.	1.1	42
130	SUSCEPTIBILITY TO DYT1 DYSTONIA IN EUROPEAN PATIENTS IS MODIFIED BY THE D216H POLYMORPHISM. Neurology, 2008, 70, 2261-2262.	1.5	73
131	Normal cardiovascular reflex testing in patients withparkin disease. Movement Disorders, 2007, 22, 528-532.	2.2	8
132	Multiplex ligationâ€dependent probe amplification assay for simultaneous detection of Parkinson's disease gene rearrangements. Movement Disorders, 2007, 22, 2274-2278.	2.2	20
133	Barth syndrome presenting with acute metabolic decompensation in the neonatal period. Journal of Inherited Metabolic Disease, 2006, 29, 684-684.	1.7	22
134	Frequency and phenotypes of LRRK2 G2019S mutation in Italian patients with Parkinson's disease. Movement Disorders, 2006, 21, 1232-1235.	2.2	28
135	Genotypic and phenotypic spectrum of PANK2 mutations in patients with neurodegeneration with brain iron accumulation. Annals of Neurology, 2006, 59, 248-256.	2.8	184
136	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.	1.4	197
137	Clinical and neuropsychological correlates in two brothers with pantothenate kinaseâ€“associated neurodegeneration. Movement Disorders, 2005, 20, 208-212.	2.2	40
138	High frequency stimulation of the subthalamic nucleus is efficacious in Parkin disease. Journal of Neurology, 2005, 252, 208-211.	1.8	40
139	Infantile neuroaxonal dystrophy and pantothenate kinase-associated neurodegeneration. Neurology, 2004, 63, 922-924.	1.5	14
140	Imaging of dopaminergic dysfunction with [¹²³ I]FP-CIT SPECT in early-onset parkin disease. Neurology, 2004, 63, 2097-2103.	1.5	54
141	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.	1.7	27
142	Molecular and functional analysis ofSLC25A20 mutations causing carnitine-acylcarnitine translocase deficiency. Human Mutation, 2004, 24, 312-320.	1.1	63
143	Ethylmalonic Encephalopathy Is Caused by Mutations in ETHE1, a Gene Encoding a Mitochondrial Matrix Protein. American Journal of Human Genetics, 2004, 74, 239-252.	2.6	192
144	Analysis of the ?-sarcoglycan gene in familial and sporadic myoclonus-dystonia: Evidence for genetic heterogeneity. Movement Disorders, 2003, 18, 1047-1051.	2.2	58

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145	Neonatal dopa-responsive extrapyramidal syndrome in twins with recessive GTPCH deficiency. Neurology, 2003, 60, 335-337.	1.5	50
146	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.	0.7	31
147	Frequency of DYT1 mutation in early onset primary dystonia in Italian patients. Movement Disorders, 2002, 17, 407-408.	2.2	36
148	Phenotype and genotype variation in primary carnitine deficiency. Genetics in Medicine, 2001, 3, 387-392.	1.1	71
149	Lethal neonatal presentation of carnitine palmitoyltransferase I deficiency. Journal of Inherited Metabolic Disease, 2001, 24, 601-602.	1.7	9
150	Functional analysis of mutations in the OCTN2 transporter causing primary carnitine deficiency: Lack of genotype-phenotype correlation. Human Mutation, 2000, 16, 401-407.	1.1	69
151	Fatal neonatal outcome in a case of muscular mitochondrial DNA depletion. Journal of Inherited Metabolic Disease, 2000, 23, 755-757.	1.7	5
152	Mutation Analysis of the GCDH Gene in Italian and Portuguese Patients with Glutaric Aciduria Type I. Molecular Genetics and Metabolism, 2000, 71, 535-537.	0.5	20
153	Clinical and molecular heterogeneity in veryâ€“long-chain acyl-coenzyme a dehydrogenase deficiency. Pediatric Neurology, 2000, 22, 98-105.	1.0	44
154	Medium-chain triglyceride loading test in carnitine-acylcarnitine translocase deficiency: Insights on treatment. Journal of Inherited Metabolic Disease, 1999, 22, 733-739.	1.7	20
155	The mitochondrial DNA C3303T mutation can cause cardiomyopathy and/or skeletal myopathy. Journal of Pediatrics, 1999, 135, 197-202.	0.9	30
156	Mild or absent clinical signs in twin sisters with short-chain acyl-CoA dehydrogenase deficiency. European Journal of Pediatrics, 1998, 157, 317-320.	1.3	33
157	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.	1.3	22
158	Very-long-chain acyl-coenzyme A dehydrogenase deficiency in a child with recurrent myoglobinuria. Neuromuscular Disorders, 1998, 8, 3-6.	0.3	27
159	Syndrome of encephalopathy, petechiae, and ethylmalonic aciduria. Pediatric Neurology, 1997, 17, 165-170.	1.0	48
160	Hypoparathyroidism in mitochondrial trifunctional protein deficiency. Journal of Pediatrics, 1996, 129, 159-162.	0.9	62
161	Sudden infant death and multiple acyl-CoA dehydrogenation disorders. European Journal of Pediatrics, 1995, 154, 421-422.	1.3	2
162	Clinical and biochemical findings in a Spanish boy with primary carnitine deficiency. Journal of Inherited Metabolic Disease, 1995, 18, 237-240.	1.7	10

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163	Rhabdomyolysis and acute encephalopathy in late onset medium chain acyl-CoA dehydrogenase deficiency.. Journal of Neurology, Neurosurgery and Psychiatry, 1995, 58, 209-214.	0.9	71
164	Riboflavin-responsive glutaric aciduria type II presenting as a leukodystrophy. Pediatric Neurology, 1995, 13, 333-335.	1.0	28
165	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.	1.4	41
166	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.	1.7	1
167	Muscle cytochromec oxidase deficiency in two Italian patients with ethylmalonic aciduria and peculiar clinical phenotype. Journal of Inherited Metabolic Disease, 1994, 17, 301-303.	1.7	20
168	Encephalopathy, petechiae, and acrocyanosis with ethylmalonic aciduria associated with muscle cytochrome c oxidase deficiency. Journal of Pediatrics, 1994, 125, 843.	0.9	32
169	Lateâ€onset riboflavinâ€responsive myopathy with combined multiple acyl coenzyme A dehydrogenase and respiratory chain deficiency. Neurology, 1994, 44, 2153-2153.	1.5	71
170	cDNA cloning and mitochondrial import of the beta-subunit of the human electron-transfer flavoprotein. FEBS Journal, 1993, 213, 1003-1008.	0.2	44
171	Beneficial effect of sodium dichloroacetate in muscle cytochrome C oxidase deficiency. European Journal of Pediatrics, 1993, 152, 537-537.	1.3	10
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