

Nardo Nardocci

List of Publications by Year in descending order

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Version: 2024-02-01

173
papers

7,773
citations

66315

42
h-index

62565

80
g-index

179
all docs

179
docs citations

179
times ranked

10161
citing authors

#	ARTICLE	IF	CITATIONS
1	Refining the mutational spectrum and geneâ€‘phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. <i>Journal of Medical Genetics</i> , 2022, 59, 399-409.	1.5	13
2	Psychiatric autoimmune conditions in children and adolescents: Is catatonia a severity marker?. <i>Progress in Neuro-Psychopharmacology and Biological Psychiatry</i> , 2021, 104, 110028.	2.5	11
3	Globus pallidus internus activity during simultaneous bilateral microelectrode recordings in status dystonicus. <i>Acta Neurochirurgica</i> , 2021, 163, 211-217.	0.9	3
4	Deep brain stimulation versus pallidotomy for status dystonicus: a single-center case series. <i>Journal of Neurosurgery</i> , 2021, 134, 197-207.	0.9	14
5	<scp><i>YY1</i></scp>â€™Related Dystonia: Clinical Aspects and Longâ€™Term Response to Deep Brain Stimulation. <i>Movement Disorders</i> , 2021, 36, 1461-1462.	2.2	16
6	Automatic imitation in youngsters with Gilles de la Tourette syndrome: A behavioral study. <i>Child Neuropsychology</i> , 2021, 27, 782-798.	0.8	4
7	<scp>THAP1</scp> Dystonia with Globus Pallidus <scp>T2</scp> Hypointensity: A Report of Two Cases. <i>Movement Disorders</i> , 2021, 36, 1463-1464.	2.2	3
8	Pediatric Paroxysmal Exercise-Induced Neurological Symptoms: Clinical Spectrum and Diagnostic Algorithm. <i>Frontiers in Neurology</i> , 2021, 12, 658178.	1.1	4
9	Movement disorders in patients with <scp>R</scp>ett syndrome: A systematic review of evidence and associated clinical considerations. <i>Psychiatry and Clinical Neurosciences</i> , 2021, 75, 369-393.	1.0	11
10	Early-onset bradykinetic rigid syndrome and reflex seizures in a child with PURA syndrome. <i>Epileptic Disorders</i> , 2021, 23, 745-748.	0.7	3
11	In-depth phenotyping of movement disorders in WARS2 encephalopathy. <i>Journal of the Neurological Sciences</i> , 2021, 429, 117675.	0.3	0
12	Status dystonicus induced by deep brain stimulation surgery. <i>Neurological Sciences</i> , 2020, 41, 729-730.	0.9	4
13	Lossâ€™ofâ€™Function Variants in <scp>HOPS</scp> Complex Genes <scp><i>VPS16</i></scp> and <scp><i>VPS41</i></scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. <i>Annals of Neurology</i> , 2020, 88, 867-877.	2.8	70
14	Cardiac phenotype in <i>ATP1A3</i>-related syndromes. <i>Neurology</i> , 2020, 95, e2866-e2879.	1.5	19
15	Vascular Remodeling in Moyamoya Angiopathy: From Peripheral Blood Mononuclear Cells to Endothelial Cells. <i>International Journal of Molecular Sciences</i> , 2020, 21, 5763.	1.8	15
16	SCN8A splicing mutation causing skipping of the exon 15 associated with intellectual disability and cortical myoclonus. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2020, 82, 56-58.	0.9	0
17	Long term perceptions of illness and self after Deep Brain Stimulation in pediatric dystonia: A narrative research. <i>European Journal of Paediatric Neurology</i> , 2020, 26, 61-67.	0.7	7
18	Postural Control in Children with Cerebellar Ataxia. <i>Applied Sciences (Switzerland)</i> , 2020, 10, 1606.	1.3	20

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19	White matter and cerebellar involvement in alternating hemiplegia of childhood. <i>Journal of Neurology</i> , 2020, 267, 1300-1311.	1.8	10
20	The Movement disorder associated with NMDAR antibody-encephalitis is complex and characteristic: an expert video-rating study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 724-726.	0.9	71
21	Frequency and phenotypic spectrum of <i>KMT2B</i> dystonia in childhood: A single-center cohort study. <i>Movement Disorders</i> , 2019, 34, 1516-1527.	2.2	55
22	Molecular Genetics and Interferon Signature in the Italian Aicardi Goutières Syndrome Cohort: Report of 12 New Cases and Literature Review. <i>Journal of Clinical Medicine</i> , 2019, 8, 750.	1.0	29
23	Safety and efficacy of deferiprone for pantothenate kinase-associated neurodegeneration: a randomised, double-blind, controlled trial and an open-label extension study. <i>Lancet Neurology</i> , The, 2019, 18, 631-642.	4.9	102
24	Epileptic phenotypes in children with early-onset mitochondrial diseases. <i>Acta Neurologica Scandinavica</i> , 2019, 140, 184-193.	1.0	15
25	Screening of SLC2A1 in a large cohort of patients suspected for Glut1 deficiency syndrome: identification of novel variants and associated phenotypes. <i>Journal of Neurology</i> , 2019, 266, 1439-1448.	1.8	18
26	Pallidal Deep Brain Stimulation in DYT6 Dystonia: Clinical Outcome and Predictive Factors for Motor Improvement. <i>Journal of Clinical Medicine</i> , 2019, 8, 2163.	1.0	25
27	GEN-O-MA project: an Italian network studying clinical course and pathogenic pathways of moyamoya disease study protocol and preliminary results. <i>Neurological Sciences</i> , 2019, 40, 561-570.	0.9	15
28	Substantia Nigra Swelling and Dentate Nucleus T2 Hyperintensity May Be Early Magnetic Resonance Imaging Signs of Propeller Protein-associated Neurodegeneration. <i>Movement Disorders Clinical Practice</i> , 2019, 6, 51-56.	0.8	20
29	Long-term educational program to limit the burden of neurological disorders in Sub-Saharan Africa: report from an Italy-Mozambique cooperation on epilepsy in children. <i>European Journal of Neurology</i> , 2018, 25, e39.	1.7	4
30	Benign hereditary chorea and deletions outside NKX2-1: What's the role of MBIP?. <i>European Journal of Medical Genetics</i> , 2018, 61, 581-584.	0.7	9
31	Pediatric NMDAR encephalitis: A single center observation study with a closer look at movement disorders. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 301-307.	0.7	34
32	CANS: Childhood acute neuropsychiatric syndromes. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 316-320.	0.7	16
33	Diagnosis and treatment of pediatric onset isolated dystonia. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 238-244.	0.7	9
34	ATP1A3-related disorders: An update. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 257-263.	0.7	54
35	SLC19A3 related disorder: Treatment implication and clinical outcome of 2 new patients. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 332-335.	0.7	6
36	A causality algorithm to guide diagnosis and treatment of catatonia due to autoimmune conditions in children and adolescents. <i>Schizophrenia Research</i> , 2018, 200, 68-76.	1.1	19

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37	R106C TFG variant causes infantile neuroaxonal dystrophy ÆplusÆ-syndrome. <i>Neurogenetics</i> , 2018, 19, 179-187.	0.7	11
38	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 135.	1.2	17
39	The noncoding RNA AK127244 in 2p16.3 locus: A new susceptibility region for neuropsychiatric disorders. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018, 177, 557-562.	1.1	4
40	DYT2 screening in early-onset isolated dystonia. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 269-271.	0.7	13
41	Alternating Hemiplegia and Epilepsia Partialis Continua: A new phenotype for a novel compound TBC1D24 mutation. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2017, 47, 71-73.	0.9	16
42	ADCY5-related movement disorders: Frequency, disease course and phenotypic variability in a cohort of paediatric patients. <i>Parkinsonism and Related Disorders</i> , 2017, 41, 37-43.	1.1	67
43	Phenotype and natural history of variant late infantile ceroidÆipofuscinosis 5. <i>Developmental Medicine and Child Neurology</i> , 2017, 59, 815-821.	1.1	31
44	Consensus clinical management guideline for pantothenate kinase-associated neurodegeneration (PKAN). <i>Molecular Genetics and Metabolism</i> , 2017, 120, 278-287.	0.5	64
45	A <i>PDE10A</i> de novo mutation causes childhoodÆonset chorea with diurnal fluctuations. <i>Movement Disorders</i> , 2017, 32, 1646-1647.	2.2	13
46	Clinical rating scale for pantothenate kinaseÆassociated neurodegeneration: A pilot study. <i>Movement Disorders</i> , 2017, 32, 1620-1630.	2.2	18
47	Thiamine-responsive disease due to mutation of <i>tpk1</i> : Importance of avoiding misdiagnosis. <i>Neurology</i> , 2017, 89, 870-871.	1.5	13
48	Recognizing the Common Origins of Dystonia and the Development of Human Movement: A Manifesto of Unmet Needs in Isolated Childhood Dystonias. <i>Frontiers in Neurology</i> , 2016, 7, 226.	1.1	28
49	Elevated aspartate aminotransferase and lactate dehydrogenase levels are a constant finding in <i>PLA2G6</i> Æassociated neurodegeneration. <i>European Journal of Neurology</i> , 2016, 23, e24-5.	1.7	2
50	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. <i>American Journal of Human Genetics</i> , 2016, 98, 763-771.	2.6	96
51	Neurological Disorders Associated with Striatal Lesions: Classification and Diagnostic Approach. <i>Current Neurology and Neuroscience Reports</i> , 2016, 16, 54.	2.0	23
52	Childhood-onset ATP1A3-related conditions: Report of two new cases of phenotypic spectrum. <i>Parkinsonism and Related Disorders</i> , 2016, 30, 81-82.	1.1	18
53	Cognitive and neuropsychological evolution in children with anti-NMDAR encephalitis. <i>Journal of Neurology</i> , 2016, 263, 765-771.	1.8	38
54	Clinical and genetic features of paroxysmal kinesigenic dyskinesia in Italian patients. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 152-157.	0.7	14

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55	Vasculogenic and Angiogenic Pathways in Moyamoya Disease. <i>Current Medicinal Chemistry</i> , 2016, 23, 315-345.	1.2	44
56	Inherited Isolated Dystonia in Children. <i>Journal of Pediatric Neurology</i> , 2015, 13, 174-179.	0.0	3
57	Clinical profile of patients with ATP1A3 mutations in Alternating Hemiplegia of Childhood—a study of 155 patients. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 123.	1.2	117
58	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. <i>American Journal of Human Genetics</i> , 2015, 96, 938-947.	2.6	109
59	Faulty cardiac repolarization reserve in alternating hemiplegia of childhood broadens the phenotype. <i>Brain</i> , 2015, 138, 2859-2874.	3.7	30
60	Mitochondrial dysfunction in Parkinson disease: evidence in mutant PARK2 fibroblasts. <i>Frontiers in Genetics</i> , 2015, 6, 78.	1.1	77
61	Focal seizure, focal dyskinesia, or both? A complex motor phenomenon reveals anti-NMDAR encephalitis. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2015, 27, 16-18.	0.9	13
62	Cerebrospinal Fluid Monoamine Metabolite Analysis in Pediatric Movement Disorders. <i>Journal of Child Neurology</i> , 2015, 30, 1800-1805.	0.7	3
63	The <i>CACNA1B</i> R1389H variant is not associated with myoclonus-dystonia in a large European multicentric cohort. <i>Human Molecular Genetics</i> , 2015, 24, 5326-5329.	1.4	28
64	Posteroventrolateral pallidotomy through implanted DBS electrodes monitored by recording local field potentials. <i>British Journal of Neurosurgery</i> , 2015, 29, 888-890.	0.4	6
65	Electroclinical spectrum of the neuronal ceroid lipofuscinoses associated with <i>CLN6</i> mutations. <i>Neurology</i> , 2015, 85, 316-324.	1.5	40
66	A family with paroxysmal nonkinesigenic dyskinesias (PNKD): Evidence of mitochondrial dysfunction. <i>European Journal of Paediatric Neurology</i> , 2015, 19, 64-68.	0.7	13
67	Sporadic and familial glut1ds Italian patients: A wide clinical variability. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2015, 24, 28-32.	0.9	32
68	Downbeat nystagmus as the presenting symptom of infantile neuroaxonal dystrophy: A case report. <i>Brain and Development</i> , 2015, 37, 270-272.	0.6	6
69	Pallidal stimulation for acquired dystonia due to cerebral palsy: beyond 5 years. <i>European Journal of Neurology</i> , 2015, 22, 426.	1.7	58
70	Running apraxia as a presenting symptom of neuronal ceroid lipofuscinosis 6. <i>Movement Disorders</i> , 2014, 29, 277-278.	2.2	1
71	Exome Sequence Reveals Mutations in CoA Synthase as a Cause of Neurodegeneration with Brain Iron Accumulation. <i>American Journal of Human Genetics</i> , 2014, 94, 11-22.	2.6	176
72	Molecular epidemiology of childhood neuronal ceroid-lipofuscinosis in Italy. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 19.	1.2	42

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73	Pathophysiology and Treatment of Neurodegeneration With Brain Iron Accumulation in the Pediatric Population. <i>Current Treatment Options in Neurology</i> , 2013, 15, 652-667.	0.7	13
74	Extragenetic factors and clinical penetrance of DYT1 dystonia: an exploratory study. <i>Journal of Neurology</i> , 2013, 260, 1081-1086.	1.8	30
75	Therapeutic Advances in Neurodegeneration with Brain Iron Accumulation. <i>International Review of Neurobiology</i> , 2013, 110, 153-164.	0.9	9
76	EMG-Based Visual-Haptic Biofeedback: A Tool to Improve Motor Control in Children With Primary Dystonia. <i>IEEE Transactions on Neural Systems and Rehabilitation Engineering</i> , 2013, 21, 474-480.	2.7	29
77	Beta-propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. <i>Brain</i> , 2013, 136, 1708-1717.	3.7	203
78	Axonal dystrophies. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2013, 113, 1919-1924.	1.0	7
79	Update on pediatric dystonias: etiology, epidemiology, and management. <i>Degenerative Neurological and Neuromuscular Disease</i> , 2012, 2, 29.	0.7	9
80	Targeting the brain: considerations in 332 consecutive patients treated by deep brain stimulation (DBS) for severe neurological diseases. <i>Neurological Sciences</i> , 2012, 33, 1285-1303.	0.9	28
81	Indicazioni e trattamento chirurgico della distonia dell'etÀ pediatrica. <i>Area Pediatrica</i> , 2012, 13, 93-100.	0.0	0
82	Cortical myoclonus in childhood and juvenile onset Huntington's disease. <i>Parkinsonism and Related Disorders</i> , 2012, 18, 794-797.	1.1	22
83	Metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 463-471.	0.5	106
84	Paroxysmal non-epileptic motor events in childhood: a clinical and video-EEG-polymyographic study. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 334-338.	1.1	17
85	Error-enhancing robot therapy to induce motor control improvement in childhood onset primary dystonia. <i>Journal of NeuroEngineering and Rehabilitation</i> , 2012, 9, 46.	2.4	18
86	Exome Sequencing Reveals De Novo WDR45 Mutations Causing a Phenotypically Distinct, X-Linked Dominant Form of NBIA. <i>American Journal of Human Genetics</i> , 2012, 91, 1144-1149.	2.6	309
87	Global metabolic profiling reveals metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations. <i>Mitochondrion</i> , 2012, 12, 577.	1.6	0
88	De novo mutations in ATP1A3 cause alternating hemiplegia of childhood. <i>Nature Genetics</i> , 2012, 44, 1030-1034.	9.4	345
89	Status dystonicus: Predictors of outcome and progression patterns of underlying disease. <i>Movement Disorders</i> , 2012, 27, 783-788.	2.2	94
90	C19orf12 and FA2H Mutations Are Rare in Italian Patients With Neurodegeneration With Brain Iron Accumulation. <i>Seminars in Pediatric Neurology</i> , 2012, 19, 75-81.	1.0	38

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91	Therapeutic Advances in Neurodegeneration With Brain Iron Accumulation. <i>Seminars in Pediatric Neurology</i> , 2012, 19, 82-86.	1.0	9
92	Phenomenology of psychogenic movement disorders in children. <i>Movement Disorders</i> , 2012, 27, 1153-1157.	2.2	39
93	Myoclonus-dystonia syndrome. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2011, 100, 563-575.	1.0	110
94	Diagnostic issues in childhood and adult dystonia. <i>Expert Opinion on Medical Diagnostics</i> , 2011, 5, 483-500.	1.6	1
95	Role of UBE3A and ATP10A genes in autism susceptibility region 15q11-q13 in an Italian population: A positive replication for UBE3A. <i>Psychiatry Research</i> , 2011, 185, 33-38.	1.7	29
96	EFNS guidelines on diagnosis and treatment of primary dystonias. <i>European Journal of Neurology</i> , 2011, 18, 5-18.	1.7	350
97	Response to letter by Dr Neil Murray. <i>European Journal of Neurology</i> , 2011, 18, e62-e62.	1.7	0
98	The relationship between group A streptococcal infections and Tourette syndrome: a study on a large service-based cohort. <i>Developmental Medicine and Child Neurology</i> , 2011, 53, 951-957.	1.1	57
99	Ataxia with oculomotor apraxia type1 (AOA1): novel and recurrent aprataxin mutations, coenzyme Q10 analyses, and clinical findings in Italian patients. <i>Neurogenetics</i> , 2011, 12, 193-201.	0.7	46
100	Hemophagocytic lymphohistiocytosis with neurological presentation: MRI findings and a nearly miss diagnosis. <i>Neurological Sciences</i> , 2011, 32, 473-477.	0.9	35
101	Deep brain stimulation for movement disorders. Considerations on 276 consecutive patients. <i>Journal of Neural Transmission</i> , 2011, 118, 1497-1510.	1.4	39
102	Iron-related MRI images in patients with pantothenate kinase-associated neurodegeneration (PKAN) treated with deferiprone: Results of a phase II pilot trial. <i>Movement Disorders</i> , 2011, 26, 1755-1759.	2.2	125
103	The "Eye-of-the-Tiger" Sign may be Absent in the Early Stages of Classic Pantothenate Kinase Associated Neurodegeneration. <i>Neuropediatrics</i> , 2011, 42, 159-162.	0.3	34
104	Reaching and Writing Movements: Sensitive and Reliable Tools to Measure Genetic Dystonia in Children. <i>Journal of Child Neurology</i> , 2011, 26, 822-829.	0.7	23
105	Childhood-onset HAM/TSP with progressive cognitive impairment. <i>Neurological Sciences</i> , 2010, 31, 209-212.	0.9	9
106	Glucose transporter type 1 deficiency: Ketogenic diet in three patients with atypical phenotype. <i>Brain and Development</i> , 2010, 32, 404-408.	0.6	33
107	Sequence Variations in Mitochondrial Ferritin: Distribution in Healthy Controls and Different Types of Patients. <i>Genetic Testing and Molecular Biomarkers</i> , 2010, 14, 793-796.	0.3	9
108	Deep Brain Stimulation Electrode Used for Radiofrequency Lesion of the Globus Pallidus Internus in Dystonia. <i>Stereotactic and Functional Neurosurgery</i> , 2009, 87, 348-352.	0.8	21

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109	Age-related Iron Deposition in the Basal Ganglia: Quantitative Analysis in Healthy Subjects. <i>Radiology</i> , 2009, 252, 165-172.	3.6	266
110	Measuring participation in children with Gilles de la Tourette syndrome: A pilot study with ICF-CY. <i>Disability and Rehabilitation</i> , 2009, 31, S116-S120.	0.9	6
111	Early onset primary dystonia. <i>European Journal of Paediatric Neurology</i> , 2009, 13, 488-492.	0.7	11
112	Mutation screening of the DYT6/THAP1 gene in Italy. <i>Movement Disorders</i> , 2009, 24, 2424-2427.	2.2	43
113	Variant late infantile ceroid lipofuscinoses associated with novel mutations in CLN6. <i>Biochemical and Biophysical Research Communications</i> , 2009, 379, 892-897.	1.0	45
114	P2.062 MLPA analysis in EOP patients. <i>Parkinsonism and Related Disorders</i> , 2009, 15, S105.	1.1	0
115	Distonie. , 2009, , 393-398.		0
116	Myoclonusâ€dystonia syndrome: Clinical presentation, disease course, and genetic features in 11 families. <i>Movement Disorders</i> , 2008, 23, 28-34.	2.2	75
117	A neurophysiological study of myoclonus in patients with DYT11 myoclonusâ€dystonia syndrome. <i>Movement Disorders</i> , 2008, 23, 2041-2048.	2.2	43
118	Polymyography in the diagnosis of childhood onset movement disorders. <i>European Journal of Paediatric Neurology</i> , 2008, 12, 480-483.	0.7	11
119	Impaired body movement representation in DYT1 mutation carriers. <i>Clinical Neurophysiology</i> , 2008, 119, 1864-1869.	0.7	20
120	SUSCEPTIBILITY TO DYT1 DYSTONIA IN EUROPEAN PATIENTS IS MODIFIED BY THE D216H POLYMORPHISM. <i>Neurology</i> , 2008, 70, 2261-2262.	1.5	73
121	Neurodegeneration associated with genetic defects in phospholipase A ₂ . <i>Neurology</i> , 2008, 71, 1402-1409.	1.5	236
122	Paroxysmal movement disorders in GLUT1 deficiency syndrome. <i>Neurology</i> , 2008, 71, 146-148.	1.5	73
123	Deep Brain Stimulation in Dystonia. , 2008, , 305-319.		1
124	Revelation of a Novel CLN5 Mutation in Early Juvenile Neuronal Ceroid Lipofuscinosis. <i>Neuropediatrics</i> , 2007, 38, 46-49.	0.3	41
125	PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. <i>Nature Genetics</i> , 2006, 38, 752-754.	9.4	497
126	Respiratory failure in infants due to spinal muscular atrophy with respiratory distress type 1. <i>Intensive Care Medicine</i> , 2006, 32, 1851-1855.	3.9	33

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127	Non-DYT1 early-onset primary torsion dystonia: Comparison with DYT1 phenotype and review of the literature. <i>Movement Disorders</i> , 2006, 21, 1411-1418.	2.2	37
128	Genotypic and phenotypic spectrum of PANK2 mutations in patients with neurodegeneration with brain iron accumulation. <i>Annals of Neurology</i> , 2006, 59, 248-256.	2.8	184
129	Stimulation of the globus pallidus internus for childhood-onset dystonia. <i>Movement Disorders</i> , 2005, 20, 1194-1200.	2.2	162
130	Persistent Fixed Torticollis due to Atlanto-Axial Rotatory Fixation: Report of 4 Pediatric Cases. <i>Neuropediatrics</i> , 2005, 36, 45-49.	0.3	8
131	Infantile neuroaxonal dystrophy and pantothenate kinase-associated neurodegeneration. <i>Neurology</i> , 2004, 63, 922-924.	1.5	14
132	GTP-cyclohydrolase I gene mutations in patients with autosomal dominant and recessive GTP-CH1 deficiency: Identification and functional characterization of four novel mutations. <i>Journal of Inherited Metabolic Disease</i> , 2004, 27, 455-463.	1.7	27
133	Analysis of the ?-sarcoglycan gene in familial and sporadic myoclonus-dystonia: Evidence for genetic heterogeneity. <i>Movement Disorders</i> , 2003, 18, 1047-1051.	2.2	58
134	Neonatal dopa-responsive extrapyramidal syndrome in twins with recessive GTPCH deficiency. <i>Neurology</i> , 2003, 60, 335-337.	1.5	50
135	Paroxysmal dyskinesias in childhood. <i>Pediatric Neurology</i> , 2003, 28, 168-172.	1.0	27
136	Consciousness Disturbances in Megalencephalic Leukoencephalopathy with Subcortical Cysts. <i>Neuropediatrics</i> , 2003, 34, 211-214.	0.3	19
137	Myoclonic dystonia as unique presentation of isolated vitamin E deficiency in a young patient. <i>Movement Disorders</i> , 2002, 17, 612-614.	2.2	36
138	Frequency of DYT1 mutation in early onset primary dystonia in Italian patients. <i>Movement Disorders</i> , 2002, 17, 407-408.	2.2	36
139	Life-threatening dystonia-dyskinesias in a child: Successful treatment with bilateral pallidal stimulation. <i>Movement Disorders</i> , 2000, 15, 1010-1012.	2.2	89
140	Neuropsychologic assessment of patients for movement disorder surgery. <i>Movement Disorders</i> , 2000, 15, 771-783.	2.2	91
141	Neuronal ceroid lipofuscinoses: detection of atypical forms. <i>Neurological Sciences</i> , 2000, 21, S57-S61.	0.9	7
142	A pathophysiological study of neuronal ceroid lipofuscinoses in 17 patients: critical review and methodological proposal. <i>Neurological Sciences</i> , 2000, 21, S89-S92.	0.9	4
143	Infantile neuroaxonal dystrophy: neuroradiological studies in 11 patients. <i>Neuroradiology</i> , 1999, 41, 376-380.	1.1	70
144	Autosomal recessive Rolandic epilepsy with paroxysmal exercise-induced dystonia and writer's cramp: Delineation of the syndrome and gene mapping to chromosome 16p12-11.2. <i>Annals of Neurology</i> , 1999, 45, 344-352.	2.8	153

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145	Infantile neuroaxonal dystrophy. <i>Neurology</i> , 1999, 52, 1472-1472.	1.5	108
146	Tourettism as clinical presentation of Huntington's disease with onset in childhood. <i>Italian Journal of Neurological Sciences</i> , 1998, 19, 383-385.	0.1	11
147	Neuronal ceroid lipofuscinoses: a review. <i>Italian Journal of Neurological Sciences</i> , 1998, 19, 271-276.	0.1	3
148	Concomitant deficiency of Î²- and Î³-sarcoglycans in 20 Î±-sarcoglycan (adhelin)-deficient patients: immunohistochemical analysis and clinical aspects. <i>Acta Neuropathologica</i> , 1997, 94, 28-35.	3.9	42
149	Acquired hemidystonia in childhood: A clinical and neuroradiological study of thirteen patients. <i>Pediatric Neurology</i> , 1996, 15, 108-113.	1.0	28
150	Transient paroxysmal dystonia in an infant possibly induced by cisapride. <i>Italian Journal of Neurological Sciences</i> , 1996, 17, 157-159.	0.1	4
151	Homozygosity mapping of Hallervorden-Spatz syndrome to chromosome 20p12.3-p13. <i>Nature Genetics</i> , 1996, 14, 479-481.	9.4	158
152	Neurological Disorders, other than Stroke, Associated with Antiphospholipid Antibodies in Childhood. <i>Neuropediatrics</i> , 1996, 27, 149-153.	0.3	63
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