Isabella Moroni

List of Publications by Year
in descending order

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Version: 2024-02-01

164 papers	6,675 citations	50273 46 h-index	76898 74 g-index
171 all docs	171 docs citations	171 times ranked	9391 citing authors

#	Article	IF	Citations
1	Kearns-Sayre syndrome: expanding spectrum of a â€œnovelâ€•mitochondrial leukomyeloencephalopathy. Neurological Sciences, 2022, 43, 2081.	1.9	1
2	Clinical practice guideline for the management of paediatric Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 530-538.	1.9	10
3	Expanding the clinical-pathological and genetic spectrum of RYR1-related congenital myopathies with cores and minicores: an Italian population study. Acta Neuropathologica Communications, 2022, 10, 54.	5.2	3
4	Validation of the Italian version of the pediatric <scp>CMT</scp> quality of life outcome measure. Journal of the Peripheral Nervous System, 2022, 27, 127-130.	3.1	3
5	Milder presentation of TELO2-related syndrome in two sisters homozygous for the p.Arg609His pathogenic variant. European Journal of Medical Genetics, 2021, 64, 104116.	1.3	5
6	Next-generation sequencing application to investigate skeletal muscle channelopathies in a large cohort of Italian patients. Neuromuscular Disorders, 2021, 31, 336-347.	0.6	13
7	<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
8	Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. Journal of Clinical Medicine, 2021, 10, 2063.	2.4	8
9	Association Between Body Mass Index and Disability in Children With Charcot-Marie-Tooth Disease. Neurology, 2021, 97, e1727-e1736.	1.1	2
10	Clinical, imaging, biochemical and molecular features in Leigh syndrome: a study from the Italian network of mitochondrial diseases. Orphanet Journal of Rare Diseases, 2021, 16, 413.	2.7	16
11	Alexander disease evolution over time: data from an Italian cohort of pediatric-onset patients. Molecular Genetics and Metabolism, 2021, 134, 353-358.	1.1	6
12	RARS1â€•related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93.	3.7	18
13	Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. Neurogenetics, 2020, 21, 87-96.	1.4	14
14	More than an â€˜atypicalâ€™ phenotype: dual molecular diagnosis of autoimmune lymphoproliferative syndrome and Becker muscular dystrophy. British Journal of Haematology, 2020, 191, 291-294.	2.5	4
15	A Recurrent Pathogenic Variant of INPP5K Underlies Autosomal Recessive Congenital Muscular Dystrophy With Cataracts and Intellectual Disability: Evidence for a Founder Effect in Southern Italy. Frontiers in Genetics, 2020, 11, 565868.	2.3	8
16	Refining clinical trial inclusion criteria to optimize the standardized response mean of the CMTPedS. Annals of Clinical and Translational Neurology, 2020, 7, 1713-1715.	3.7	5
17	Novel mutation identification and copy number variant detection via exome sequencing in congenital muscular dystrophy. Molecular Genetics & Genomic Medicine, 2020, 8, e1387.	1.2	3
18	Expanding the phenotypic spectrum of TRIM2 â€•associated Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2020, 25, 429-432.	3.1	4

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19	Myopathic changes associated with psychomotor delay and seizures caused by a novel homozygous mutation in TBCK. Muscle and Nerve, 2020, 62, 266-271.	2.2	3
20	Validation of the Italian version of the <scp>Charcotâ€Marieâ€Tooth</scp> disease Pediatric Scale. Journal of the Peripheral Nervous System, 2020, 25, 138-142.	3.1	5
21	A longitudinal study of CMT1A using Rasch analysis based CMT neuropathy and examination scores. Neurology, 2020, 94, e884-e896.	1.1	29
22	The Search for Molecular Markers in a Gene-Orphan Case Study of a Pediatric Spinal Cord Pilocytic Astrocytoma. Cancer Genomics and Proteomics, 2020, 17, 117-130.	2.0	6
23	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	7.6	39
24	Estimating the impact of COVID-19 pandemic on services provided by Italian Neuromuscular Centers: an Italian Association of Myology survey of the acute phase. Acta Myologica, 2020, 39, 57-66.	1.5	24
25	Pathogenic Variants in STXBP1 and in Genes for GABAa Receptor Subunities Cause Atypical Rett/Rett-like Phenotypes. International Journal of Molecular Sciences, 2019, 20, 3621.	4.1	29
26	Variation in SIPA1L2 is correlated with phenotype modification in Charcotâ€“ Marieâ€“ Tooth disease type 1A. Annals of Neurology, 2019, 85, 316-330.	5.3	33
27	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
28	Epileptic phenotypes in children with earlyâ€onset mitochondrial diseases. Acta Neurologica Scandinavica, 2019, 140, 184-193.	2.1	15
29	Balance impairment in pediatric charcotâ€“marieâ€“tooth disease. Muscle and Nerve, 2019, 60, 242-249.	2.2	22
30	Spontaneous MRI improvement and absence of cerebral calcification in Aicardi-GoutiÃ¨res syndrome: Diagnostic and disease-monitoring implications. Molecular Genetics and Metabolism, 2019, 126, 489-494.	1.1	10
31	Exome sequencing detects compound heterozygous nonsense LAMA2 mutations in two siblings with atypical phenotype and nearly normal brain MRI. Neuromuscular Disorders, 2019, 29, 376-380.	0.6	9
32	Clinical-genetic features and peculiar muscle histopathology in infantileDNM1L-related mitochondrial epileptic encephalopathy. Human Mutation, 2019, 40, 601-618.	2.5	31
33	Clinical spectrum of PTEN mutation in pediatric patients. A bicenter experience. European Journal of Medical Genetics, 2019, 62, 103596.	1.3	39
34	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	2.5	22
35	SLC19A3 related disorder: Treatment implication and clinical outcome of 2 new patients. European Journal of Paediatric Neurology, 2018, 22, 332-335.	1.6	6
36	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

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37	Compound heterozygous missense and deep intronic variants in NDUFAF6 unraveled by exome sequencing and mRNA analysis. Journal of Human Genetics, 2018, 63, 563-568.	2.3	15
38	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26
39	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. Brain, 2018, 141, 3319-3330.	7.6	25
40	Electromyographic and biomechanical analysis of step negotiation in Charcot Marie Tooth subjects whose level walk is not impaired. Gait and Posture, 2018, 62, 497-504.	1.4	6
41	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	2.7	61
42	Multi-system disorder and severe recurrent rhabdomyolysis due to TANGO2 mutations in a 3-year-old child. Neuromuscular Disorders, 2018, 28, S41-S42.	0.6	0
43	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. Orphanet Journal of Rare Diseases, 2018, 13, 135.	2.7	17
44	The noncoding RNA AK127244 in 2p16.3 locus: A new susceptibility region for neuropsychiatric disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 557-562.	1.7	4
45	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86
46	Mutations in INPP5K Cause a Form of Congenital Muscular Dystrophy Overlapping Marinesco-SjÃ¶gren Syndrome and Dystroglycanopathy. American Journal of Human Genetics, 2017, 100, 537-545.	6.2	67
47	Dysregulated mitophagy and mitochondrial organization in optic atrophy due to OPA1 mutations. Neurology, 2017, 88, 131-142.	1.1	81
48	221st ENMC International Workshop:. Neuromuscular Disorders, 2017, 27, 1138-1142.	0.6	10
49	Thiamine-responsive disease due to mutation of tpk1 : Importance of avoiding misdiagnosis. Neurology, 2017, 89, 870-871.	1.1	13
50	Responsiveness of gait analysis parameters in a cohort of 71 CMT subjects. Neuromuscular Disorders, 2017, 27, 1029-1037.	0.6	10
51	Natural history of Charcotâ€Marieâ€Tooth disease during childhood. Annals of Neurology, 2017, 82, 353-359.	5.3	50
52	Multi-system disorder and severe recurrent rhabdomyolysis due to TANGO2 mutations in a 3 year-old child. Neuromuscular Disorders, 2017, 27, S207.	0.6	2
53	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	3.6	32
54	Not only dominant, not only optic atrophy: expanding the clinical spectrum associated with OPA1 mutations. Orphanet Journal of Rare Diseases, 2017, 12, 89.	2.7	39

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55	Intrafamilial phenotypic variability in Andersenâ€“Tawil syndrome: A diagnostic challenge in a potentially treatable condition. Neuromuscular Disorders, 2017, 27, 294-297.	0.6	14
56	Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes. Journal of Neurology, 2017, 264, 102-111.	3.6	38
57	Congenital myasthenic syndrome: phenotypic variability in patients harbouring p.T159P mutation in gene. Acta Myologica, 2017, 36, 28-32.	1.5	4
58	Biallelic Mutations in DNM1L are Associated with a Slowly Progressive Infantile Encephalopathy. Human Mutation, 2016, 37, 898-903.	2.5	64
59	Magnetic resonance imaging spectrum of succinate dehydrogenaseâ€“related infantile leukoencephalopathy. Annals of Neurology, 2016, 79, 379-386.	5.3	34
60	Screening for SH3TC2 gene mutations in a series of demyelinating recessive Charcotâ€Marieâ€Tooth disease (CMT4). Journal of the Peripheral Nervous System, 2016, 21, 142-149.	3.1	32
61	COA7(C1orf163/RESA1) mutations associated with mitochondrial leukoencephalopathy and cytochrome c oxidase deficiency. Journal of Medical Genetics, 2016, 53, 846-849.	3.2	40
62	New Mutations in NEB Gene Discovered by Targeted Next-Generation Sequencing in Nemaline Myopathy Italian Patients. Journal of Molecular Neuroscience, 2016, 59, 351-359.	2.3	17
63	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
64	Phenotypic Variability of Childhood Charcot-Marie-Tooth Disease. JAMA Neurology, 2016, 73, 645.	9.0	71
65	Neurological Disorders Associated with Striatal Lesions: Classification and Diagnostic Approach. Current Neurology and Neuroscience Reports, 2016, 16, 54.	4.2	23
66	Very mild features of dysequilibrium syndrome associated with a novel VLDLR missense mutation. Neurogenetics, 2016, 17, 191-195.	1.4	9
67	Unusual presentations and intrafamilial phenotypic variability in infantile onset Alexander disease. Neurological Sciences, 2016, 37, 973-977.	1.9	7
68	Response to: Mitochondrial neuropathy affects peripheral and cranial nerves and is primary or secondary or both. Neuromuscular Disorders, 2016, 26, 549.	0.6	0
69	SEPN1-related myopathy in three patients: novel mutations and diagnostic clues. European Journal of Pediatrics, 2016, 175, 1113-1118.	2.7	17
70	LYRM7mutations cause a multifocal cavitating leukoencephalopathy with distinct MRI appearance. Brain, 2016, 139, 782-794.	7.6	51
71	â€œMitochondrial neuropathiesâ€: A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.6	37
72	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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73	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	6.0	64
74	CMT subtypes and disease burden in patients enrolled in the Inherited Neuropathies Consortium natural history study: a cross-sectional analysis. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 873-878.	1.9	249
75	Mitochondrial leukoencephalopathy and complex II deficiency associated with a recessive SDHB mutation with reduced penetrance. Molecular Genetics and Metabolism Reports, 2015, 5, 51-54.	1.1	16
76	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.1	75
77	Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. American Journal of Medical Genetics, Part A, 2015, 167, 296-312.	1.2	447
78	Non-coding VMA21 deletions cause X-linked Myopathy with Excessive Autophagy. Neuromuscular Disorders, 2015, 25, 207-211.	0.6	25
79	Bone and Spinal Muscular Atrophy. Bone, 2015, 79, 116-120.	2.9	51
80	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
81	A slowly progressive mitochondrial encephalomyopathy widens the spectrum of AIFM1 disorders. Neurology, 2015, 84, 2193-2195.	1.1	47
82	Centronuclear myopathies: genotypeâ€“phenotype correlation and frequency of defined genetic forms in an Italian cohort. Journal of Neurology, 2015, 262, 1728-1740.	3.6	51
83	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	3.6	68
84	Childhood onset of acquired neuromyotonia: Association with a ganglioneuroma. Muscle and Nerve, 2015, 51, 620-621.	2.2	1
85	Genotypeâ€“phenotype characteristics and baseline natural history of heritable neuropathies caused by mutations in theMPZgene. Brain, 2015, 138, 3180-3192.	7.6	80
86	Pearson Syndrome: A Retrospective Cohort Study from the Marrow Failure Study Group of A.I.E.O.P. (Associazione Italiana Emato-Oncologia Pediatrica). JIMD Reports, 2015, 26, 37-43.	1.5	39
87	Dysregulated mitophagy and mitochondrial transport in sensori-motor neuropathy due to â€œDominant Optic Atrophyâ€•plus with OPA1 (Optic Atrophy 1) mutations. Neuromuscular Disorders, 2015, 25, S185-S186.	0.6	0
88	Truncating and Missense Mutations in IGHMBP2 Cause Charcot-Marie Tooth Disease Type 2. American Journal of Human Genetics, 2014, 95, 590-601.	6.2	75
89	A Novel Homozygous YARS2 Mutation in Two Italian Siblings and a Review of Literature. JIMD Reports, 2014, 20, 95-101.	1.5	19
90	Cavitating leukoencephalopathy with multiple mitochondrial dysfunction syndrome and NFU1 mutations. Frontiers in Genetics, 2014, 5, 412.	2.3	49

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91	Overlapping phenotypes in complex spastic paraplegias SPG11, SPG15, SPG35 and SPG48. Brain, 2014, 137, 1907-1920.	7.6	133
92	Mutations in APOPT1, Encoding a Mitochondrial Protein, Cause Cavitating Leukoencephalopathy with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 2014, 95, 315-325.	6.2	64
93	A fourth case of POMT2-related limb girdle muscle dystrophy with mild reduction of Î±-dystroglycan glycosylation. European Journal of Paediatric Neurology, 2014, 18, 404-408.	1.6	17
94	Doubleâ€trouble in pediatric neurology: Myotonia congenita combined with charcotâ€“marieâ€“tooth disease type 1a. Muscle and Nerve, 2014, 50, 145-147.	2.2	6
95	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	3.6	119
96	Clinical, histological and genetic characterisation of patients with tubular aggregate myopathy caused by mutations in STIM1. Journal of Medical Genetics, 2014, 51, 824-833.	3.2	72
97	G.P.170. Neuromuscular Disorders, 2014, 24, 856-857.	0.6	0
98	G.P.251. Neuromuscular Disorders, 2014, 24, 892.	0.6	2
99	G.P.136. Neuromuscular Disorders, 2014, 24, 841-842.	0.6	1
100	Myoclonus in mitochondrial disorders. Movement Disorders, 2014, 29, 722-728.	3.9	33
101	Mitochondrial Diseases in Childhood. Current Molecular Medicine, 2014, 14, 1069-1078.	1.3	3
102	Changes of gait pattern in children with Charcot-Marie-Tooth disease type 1A: a 18 months follow-up study. Journal of NeuroEngineering and Rehabilitation, 2013, 10, 65.	4.6	19
103	Dandy-Walker malformation and Wisconsin syndrome: novel cases add further insight into the genotype-phenotype correlations of 3q23q25 deletions. Orphanet Journal of Rare Diseases, 2013, 8, 75.	2.7	19
104	Novel PTRF mutation in a child with mild myopathy and very mild congenital lipodystrophy. BMC Medical Genetics, 2013, 14, 89.	2.1	40
105	SETX mutations are a frequent genetic cause of juvenile and adult onset cerebellar ataxia with neuropathy and elevated serum alpha-fetoprotein. Orphanet Journal of Rare Diseases, 2013, 8, 123.	2.7	31
106	Peripheral neuropathy in mitochondrial disorders. Lancet Neurology, The, 2013, 12, 1011-1024.	10.2	101
107	P.1.11 Development of a registry and a database for a nation-wide Italian collaborative network on congenital muscular dystrophy. Neuromuscular Disorders, 2013, 23, 744-745.	0.6	0
108	Early neurodevelopmental assessment in Duchenne muscular dystrophy. Neuromuscular Disorders, 2013, 23, 451-455.	0.6	63

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109	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.1	157
110	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64
111	Transitioning outcome measures: relationship between the CMTPedS and CMTNSv2 in children, adolescents, and young adults with Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2013, 18, 177-180.	3.1	15
112	A novel homozygous mutation in SUCLA2 gene identified by exome sequencing. Molecular Genetics and Metabolism, 2012, 107, 403-408.	1.1	38
113	Respiratory and cardiac function in congenital muscular dystrophies with alpha dystroglycan deficiency. Neuromuscular Disorders, 2012, 22, 685-689.	0.6	31
114	Cortical myoclonus in childhood and juvenile onset Huntington's disease. Parkinsonism and Related Disorders, 2012, 18, 794-797.	2.2	22
115	Intracranial calcification in early infantile Krabbe disease: nothing new under the sun. Developmental Medicine and Child Neurology, 2012, 54, 376-379.	2.1	14
116	Gait pattern classification in children with Charcotâ€“Marieâ€“Tooth disease type 1A. Gait and Posture, 2012, 35, 131-137.	1.4	72
117	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. American Journal of Human Genetics, 2012, 91, 541-547.	6.2	167
118	Brownâ€“Vialettoâ€“van Laere and Fazioâ€“Londe overlap syndromes: A clinical, biochemical and genetic study. Neuromuscular Disorders, 2012, 22, 1075-1082.	0.6	36
119	Novel POMGNT1 point mutations and intragenic rearrangements associated with muscle-eye-brain disease. Journal of the Neurological Sciences, 2012, 318, 45-50.	0.6	20
120	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	6.2	192
121	Sequence and Copy Number Analyses of HEXB Gene in Patients Affected by Sandhoff Disease: Functional Characterization of 9 Novel Sequence Variants. PLoS ONE, 2012, 7, e41516.	2.5	22
122	Reliability of instrumented movement analysis as outcome measure in Charcotâ€“Marieâ€“Tooth disease: Results from a multitask locomotor protocol. Gait and Posture, 2011, 34, 36-43.	1.4	25
123	P3.1 Brownâ€“Vialettoâ€“Van Laere and Fazio Londe overlap sindromes: A clinical, biochemical and genetic study in 6 patients. Neuromuscular Disorders, 2011, 21, 682.	0.6	1
124	Outcome measures for Charcotâ€Marieâ€Tooth disease: clinical and neurofunctional assessment in children. Journal of the Peripheral Nervous System, 2011, 16, 237-242.	3.1	16
125	Ataxia with oculomotor apraxia type1 (AOA1): novel and recurrent aprataxin mutations, coenzyme Q10 analyses, and clinical findings in Italian patients. Neurogenetics, 2011, 12, 193-201.	1.4	46
126	Development, reliability and validity of the Charcotâ€Marieâ€Tooth disease Pediatric Scale (CMTPedS). Journal of Foot and Ankle Research, 2011, 4, .	1.9	1

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127	Identification of previously unreported mutations in CHRNA1, CHRNE and RAPSN genes in three unrelated Italian patients with congenital myasthenic syndromes. Journal of Neurology, 2010, 257, 1119-1123.	3.6	11
128	Congenital muscular dystrophies with cognitive impairment. Neurology, 2010, 75, 898-903.	1.1	27
129	Congenital muscular dystrophies with defective glycosylation of dystroglycan. Neurology, 2009, 72, 1802-1809.	1.1	166
130	Fukutin gene mutations in an Italian patient with early onset muscular dystrophy but no central nervous system involvement. Muscle and Nerve, 2009, 39, 845-848.	2.2	9
131	Novel mutations in the GDAP1 gene in patients affected with early-onset axonal Charcot-Marie-Tooth type 4A. Neuromuscular Disorders, 2009, 19, 476-480.	0.6	23
132	EM.P.2.08 Fukutin gene mutations in an Italian patient with early onset muscular dystrophy but no central nervous system involvement. Neuromuscular Disorders, 2009, 19, 554.	0.6	0
133	Clinical, molecular, and protein correlations in a large sample of genetically diagnosed Italian limb girdle muscular dystrophy patients. Human Mutation, 2008, 29, 258-266.	2.5	162
134	POMT1 and POMT2 mutations in CMD patients: A multicentric Italian study. Neuromuscular Disorders, 2008, 18, 565-571.	0.6	38
135	G.P.2.03 Clinical and molecular characterization of 12 patients with defective Î±-dystroglycan glycosylation. Neuromuscular Disorders, 2008, 18, 735-736.	0.6	0
136	G.P.2.07 Alpha-dystroglycanopathy in an Italian patient due to large intragenic and single nucleotide deletions in the POMGnT1 gene. Neuromuscular Disorders, 2008, 18, 737.	0.6	0
137	Spectrum of MMACHC mutations in Italian and Portuguese patients with combined methylmalonic aciduria and homocystinuria, cblC type. Molecular Genetics and Metabolism, 2008, 93, 475-480.	1.1	80
138	Classification of Childhood White Matter Disorders Using Proton MR Spectroscopic Imaging. American Journal of Neuroradiology, 2008, 29, 1270-1275.	2.4	46
139	Phenotypic clustering of lamin A/C mutations in neuromuscular patients. Neurology, 2007, 69, 1285-1292.	1.1	120
140	C.P.3.02 Expanding the clinical spectrum of POMT1 and POMT2 phenotype: A multicentric study in the Italian population. Neuromuscular Disorders, 2007, 17, 869-870.	0.6	0
141	Congenital muscular dystrophy with muscle inflammation alpha dystroglycan glycosylation defect and no mutation in FKRP gene. Journal of the Neurological Sciences, 2006, 243, 47-51.	0.6	6
142	Effects of riboflavin in children with complex II deficiency. Brain and Development, 2006, 28, 576-581.	1.1	74
143	Childhood-onset multifocal motor neuropathy with conduction blocks. Neurology, 2006, 66, 922-924.	1.1	13
144	GJA12 mutations in children with recessive hypomyelinating leukoencephalopathy. Neurology, 2006, 67, 273-279.	1.1	95

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145	LAMA2 Gene Analysis in Congenital Muscular Dystrophy. Archives of Neurology, 2005, 62, 1582-6.	4.5	32
146	L-2-hydroxyglutaric aciduria and brain malignant tumors. Neurology, 2004, 62, 1882-1884.	1.1	100
147	Clinical and molecular findings in children with complex I deficiency. Biochimica Et Biophysica Acta - Bioenergetics, 2004, 1659, 136-147.	1.0	231
148	A missense mutation in the mitochondrial ND5 gene associated with a Leigh-MELAS overlap syndrome. Neurology, 2003, 60, 1857-1861.	1.1	68
149	Consciousness Disturbances in Megalencephalic Leukoencephalopathy with Subcortical Cysts. Neuropediatrics, 2003, 34, 211-214.	0.6	19
150	Cerebral White Matter Involvement in Children with Mitochondrial Encephalopathies. Neuropediatrics, 2002, 33, 79-85.	0.6	74
151	X-linked creatine deficiency syndrome: A novel mutation in creatine transporter geneSLC6A8. Annals of Neurology, 2002, 52, 227-231.	5.3	92
152	Encefalomiopatie mitocondriali in etÃ pediatrica: Incidenza dell'accumulo di acido lattico documentato con immagini di spettroscopia RM del protone. The Neuroradiology Journal, 2001, 14, 149-152.	0.1	1
153	Costello Syndrome: a cancer predisposing syndrome?. Clinical Dysmorphology, 2000, 9, 265-268.	0.3	21
154	Clinical, biochemical and neuroradiological findings in L -2-hydroxyglutaric aciduria. Neurological Sciences, 2000, 21, 103-108.	1.9	54
155	3-Methylglutaconic aciduria and hypermethioninaemia in a child with clinical and neuroradiological findings of Leigh disease. Journal of Inherited Metabolic Disease, 1999, 22, 593-598.	3.6	10
156	Le encefalomiopatie mitocondriali in etÃ pediatrica. The Neuroradiology Journal, 1999, 12, 113-117.	0.1	2
157	L-2-Hydroxyglutaric aciduria: MRI in seven cases. Neuroradiology, 1998, 40, 727-733.	2.2	64
158	Congenital muscular dystrophy with merosin deficiency: MRI findings in five patients. Neuroradiology, 1998, 40, 807-811.	2.2	40
159	X-linked adrenoleukodystrophy: first report of the Italian Study Group. Italian Journal of Neurological Sciences, 1998, 19, 315-319.	0.1	7
160	Mitochondrial disease associated with the T8993G mutation of the mitochondrial ATPase 6Âgene: a clinical, biochemical, and molecular study in six families. Journal of Neurology, Neurosurgery and Psychiatry, 1997, 63, 16-22.	1.9	133
161	Mild clinical phenotype in a 12-year-old boy with partial merosin deficiency and central and peripheral nervous system abnormalities. Neuromuscular Disorders, 1996, 6, 377-381.	0.6	49
162	Ryanodine receptor gene point mutation and malignant hyperthermia susceptibility. Journal of Neurology, 1995, 242, 127-133.	3.6	4

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163	Riboflavin-responsive glutaric aciduria type II presenting as a leukodystrophy. Pediatric Neurology, 1995, 13, 333-335.	2.1	28
164	Ubidecarenone in the treatment of mitochondrial myopathies: a multi-center double-blind trial. Journal of the Neurological Sciences, 1990, 100, 70-78.	0.6	109