Carlo Minetti

List of Publications by Year
in descending order

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244 papers	12,291 citations	16411 64 h-index	34900 98 g-index
249 all docs	249 docs citations	249 times ranked	13747 citing authors

#	Article	IF	Citations
1	An integrated approach to the evaluation of patients with asymptomatic or minimally symptomatic <scp>hyperCKemia</scp>. Muscle and Nerve, 2022, 65, 96-104.	1.0	10
2	P2X7 Receptor Antagonist Reduces Fibrosis and Inflammation in a Mouse Model of Alpha-Sarcoglycan Muscular Dystrophy. Pharmaceuticals, 2022, 15, 89.	1.7	11
3	Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. Cerebellum, 2022, , 1.	1.4	5
4	Ocular phenotype and electroretinogram abnormalities in Lafora disease and correlation with disease stage. Journal of Neurology, 2022, 269, 3597-3604.	1.8	3
5	Epilepsy Course and Developmental Trajectories in STXBP1 -DEE. Neurology: Genetics, 2022, 8, .	0.9	24
6	Anterior chest wall deformities in children with neurofibromatosis type 1. Acta Paediatrica, International Journal of Paediatrics, 2021, 110, 594-595.	0.7	1
7	Atypical Presentation of Aromatic L-Amino Acid Decarboxylase Deficiency with Developmental Epileptic Encephalopathy. Journal of Pediatric Epilepsy, 2021, 10, 124-127.	0.1	4
8	Temporalâ€parietalâ€occipital epilepsy in GEFS+ associated with SCN1A mutation. Epileptic Disorders, 2021, 23, 397-401.	0.7	2
9	Italian cohort of Lafora disease: Clinical features, disease evolution, and genotype-phenotype correlations. Journal of the Neurological Sciences, 2021, 424, 117409.	0.3	11
10	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.	1.0	8
11	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. Neurology, 2021, 97, e577-e586.	1.5	11
12	Neuromuscular and Neuroendocrinological Features Associated With ZC4H2-Related Arthrogryposis Multiplex Congenita in a Sicilian Family: A Case Report. Frontiers in Neurology, 2021, 12, 704747.	1.1	6
13	Epileptic encephalopathy caused by <scp>ARV1</scp> deficiency: Refinement of the genotypeâ€“phenotype spectrum and functional impact on <scp>GPI</scp>â€anchored proteins. Clinical Genetics, 2021, 100, 607-614.	1.0	6
14	Hyperkinetic stereotyped movements in a boy with biallelic CNTNAP2 variants. Italian Journal of Pediatrics, 2021, 47, 208.	1.0	5
15	The Role of Muscle Biopsy in Diagnostic Process of Infant Hypotonia: From Clinical Classification to the Genetic Outcome. Frontiers in Neurology, 2021, 12, 735488.	1.1	7
16	New Trends and Most Promising Therapeutic Strategies for Epilepsy Treatment. Frontiers in Neurology, 2021, 12, 753753.	1.1	23
17	Cortical tremor: a tantalizing conundrum between cortex and cerebellum. Brain, 2020, 143, e87-e87.	3.7	7
18	Deep learning for neonatal seizure detection: a friend rather than foe. The Lancet Child and Adolescent Health, 2020, 4, 711-712.	2.7	1

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19	eATP/P2X7R Axis: An Orchestrated Pathway Triggering Inflammasome Activation in Muscle Diseases. International Journal of Molecular Sciences, 2020, 21, 5963.	1.8	11
20	Loss of Wwox Perturbs Neuronal Migration and Impairs Early Cortical Development. Frontiers in Neuroscience, 2020, 14, 644.	1.4	22
21	Targeted re-sequencing for early diagnosis of genetic causes of childhood epilepsy: the Italian experience from the â€˜beyond epilepsyâ€™ project. Italian Journal of Pediatrics, 2020, 46, 92.	1.0	17
22	Distal motor neuropathy associated with novel EMILIN1 mutation. Neurobiology of Disease, 2020, 137, 104757.	2.1	6
23	Genotype-phenotype correlations in patients with de novo KCNQ2 pathogenic variants. Neurology: Genetics, 2020, 6, e528.	0.9	24
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	Tele-monitoring in paediatric and young home-ventilated neuromuscular patients: A multicentre case-control trial. Journal of Telemedicine and Telecare, 2019, 25, 414-424.	1.4	21
26	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
27	Novel homozygous TSFM pathogenic variant associated with encephalocardiomyopathy with sensorineural hearing loss and peculiar neuroradiologic findings. Neurogenetics, 2019, 20, 165-172.	0.7	8
28	Microbiota-gut brain axis involvement in neuropsychiatric disorders. Expert Review of Neurotherapeutics, 2019, 19, 1037-1050.	1.4	116
29	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	3.7	43
30	Abnormal circadian rhythm in patients with GRIN1-related developmental epileptic encephalopathy. European Journal of Paediatric Neurology, 2019, 23, 657-661.	0.7	4
31	Pelizaeusâ€“Merzbacher Disease due to PLP1 Frameshift Mutation in a Female with Nonrandom Skewed X-Chromosome Inactivation. Neuropediatrics, 2019, 50, 268-270.	0.3	1
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	No evidence for a BRD 2 promoter hypermethylation inÂblood leukocytes of Europeans with juvenile myoclonicÂepilepsy. Epilepsia, 2019, 60, e31-e36.	2.6	4
34	Primary muscle involvement in a 15â€yearâ€old girl with the recurrent homozygous c.362dupC variant in FKBP14. American Journal of Medical Genetics, Part A, 2019, 179, 317-321.	0.7	3
35	The Danger Signal Extracellular ATP Is Involved in the Immunomediated Damage of Î±-Sarcoglycanâ€“Deficient Muscular Dystrophy. American Journal of Pathology, 2019, 189, 354-369.	1.9	9
36	Muscle Expression of Type I and Type <scp>II</scp> Interferons Is Increased in Juvenile Dermatomyositis and Related to Clinical and Histologic Features. Arthritis and Rheumatology, 2019, 71, 1011-1021.	2.9	55

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37	Novel mutation in sarcotubular myopathy. Acta Myologica, 2019, 38, 8-12.	1.5	13
38	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.3	24
39	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	4.5	69
40	Spinal motor neuron involvement in a patient with homozygous PRUNE mutation. European Journal of Paediatric Neurology, 2018, 22, 541-543.	0.7	10
41	Erratum to â€œDe novo 12q22.q23.3 duplication associated with temporal lobe epilepsyâ€•[Seizure 57 (2018) 63â€“65]. Seizure: the Journal of the British Epilepsy Association, 2018, 57, R1.	0.9	0
42	Clinical and molecular consequences of exon 78 deletion in DMD gene. Journal of Human Genetics, 2018, 63, 761-764.	1.1	7
43	De novo 12q22.q23.3 duplication associated with temporal lobe epilepsy. Seizure: the Journal of the British Epilepsy Association, 2018, 57, 63-65.	0.9	0
44	Inflammatory myopathy in a patient with collagen VI mutations. Scandinavian Journal of Rheumatology, 2018, 47, 166-167.	0.6	0
45	Detection of early nocturnal hypoventilation in neuromuscular disorders. Journal of International Medical Research, 2018, 46, 1153-1161.	0.4	22
46	Characterization of MDPL Fibroblasts Carrying the Recurrent p.Ser605del Mutation in POLD1 Gene. DNA and Cell Biology, 2018, 37, 1061-1067.	0.9	20
47	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. Genes, 2018, 9, 524.	1.0	7
48	Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. American Journal of Human Genetics, 2018, 103, 431-439.	2.6	62
49	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	1.0	86
50	Widening the Heterogeneity of Leigh Syndrome: Clinical, Biochemical, and Neuroradiologic Features in a Patient Harboring a NDUFA10 Mutation. JIMD Reports, 2017, 37, 37-43.	0.7	13
51	Mutations in GMPPB Presenting with Pseudometabolic Myopathy. JIMD Reports, 2017, 38, 23-31.	0.7	8
52	De novo 12q22.q23.3 duplication associated with temporal lobe epilepsy. Seizure: the Journal of the British Epilepsy Association, 2017, 50, 80-82.	0.9	4
53	Novel AMPD2 mutation in pontocerebellar hypoplasia, dysmorphisms, and teeth abnormalities. Neurology: Genetics, 2017, 3, e179.	0.9	22
54	Respiratory pattern in a FSDH paediatric population. Respiratory Medicine, 2017, 126, 132.	1.3	0

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55	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	1.8	32
56	Congenital myopathies: clinical phenotypes and new diagnostic tools. Italian Journal of Pediatrics, 2017, 43, 101.	1.0	80
57	Tele-monitoring in paediatric neuromuscular patients requiring home mechanical ventilation, multicentric study. , 2017, , .		0
58	<scp>WES</scp> in a family trio suggests involvement of <scp>TECPR2</scp> in a complex form of progressive motor neuron disease. Clinical Genetics, 2016, 90, 182-185.	1.0	17
59	MYH7-related myopathies: clinical, histopathological and imaging findings in a cohort of Italian patients. Orphanet Journal of Rare Diseases, 2016, 11, 91.	1.2	70
60	White matter involvement in a family with a novel PDGFB mutation. Neurology: Genetics, 2016, 2, e77.	0.9	19
61	Respiratory pattern in a FSHD pediatric population. Respiratory Medicine, 2016, 119, 78-80.	1.3	7
62	Antiepileptic Drugs Under Investigation for Treatment of Focal Epilepsy. Clinical Neuropharmacology, 2016, 39, 281-287.	0.2	6
63	Beyond spinal muscular atrophy with lower extremity dominance: cerebellar hypoplasia associated with a novel mutation in <scp>BICD</scp>2. European Journal of Neurology, 2016, 23, e19-21.	1.7	18
64	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.5	92
65	Novel treatment perspectives from advances in understanding of genetic epilepsy syndromes. Expert Opinion on Orphan Drugs, 2016, 4, 485-490.	0.5	0
66	The ubiquitin ligase tripartite-motif-protein 32 is induced in Duchenne muscular dystrophy. Laboratory Investigation, 2016, 96, 862-871.	1.7	23
67	Genetic and Early Clinical Manifestations of Females Heterozygous for Duchenne/Becker Muscular Dystrophy. Pediatric Neurology, 2016, 55, 58-63.	1.0	37
68	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. Neuromuscular Disorders, 2016, 26, 292-299.	0.3	25
69	STXBP1 encephalopathy. Neurology, 2016, 86, 954-962.	1.5	264
70	â€œMitochondrial neuropathiesâ€: A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.3	37
71	Health-related quality of life and functional changes in DMD: A 12-month longitudinal cohort study. Neuromuscular Disorders, 2016, 26, 189-196.	0.3	32
72	Psychiatric comorbidities in patients from seven families with autosomal dominant cortical tremor, myoclonus, and epilepsy. Epilepsy and Behavior, 2016, 56, 38-43.	0.9	21

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73	The leukodystrophy protein FAM126A (hyccin) regulates PtdIns(4)P synthesis at the plasmaÂmembrane. Nature Cell Biology, 2016, 18, 132-138.	4.6	91
74	Safety of Overnight Switch from Brand-Name to Generic Levetiracetam. Clinical Drug Investigation, 2016, 36, 87-91.	1.1	21
75	Novel GABRG2 mutations cause familial febrile seizures. Neurology: Genetics, 2015, 1, e35.	0.9	29
76	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.5	75
77	Early onset cardiomyopathy associated with the mitochondrial tRNALeu(UUR) 3271T>C MELAS mutation. Biochemical and Biophysical Research Communications, 2015, 458, 601-604.	1.0	14
78	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	1.8	68
79	Expanding the Clinical and Magnetic Resonance Spectrum of Leukoencephalopathy with Thalamus and Brainstem Involvement and High Lactate (LTBL) in a Patient Harboring a Novel EARS2 Mutation. JIMD Reports, 2015, 23, 85-89.	0.7	15
80	Enhancement of Muscle T Regulatory Cells and Improvement of Muscular Dystrophic Process in mdx Mice by Blockade of Extracellular ATP/P2X Axis. American Journal of Pathology, 2015, 185, 3349-3360.	1.9	42
81	Validation of a score tool for measurement of histological severity in juvenile dermatomyositis and association with clinical severity of disease. Annals of the Rheumatic Diseases, 2015, 74, 204-210.	0.5	56
82	Impairment of ceramide synthesis causes a novel progressive myoclonus epilepsy. Annals of Neurology, 2014, 76, 206-212.	2.8	80
83	Inter-society consensus document on treatment and prevention of bronchiolitis in newborns and infants. Italian Journal of Pediatrics, 2014, 40, 65.	1.0	129
84	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	1.8	119
85	Functional characterization of the c.<scp>462delA</scp> mutation in the <scp>NDUFS4</scp> subunit gene of mitochondrial complex I. Clinical Genetics, 2014, 86, 99-101.	1.0	4
86	Vaccination recommendations for patients with neuromuscular disease. Vaccine, 2014, 32, 5893-5900.	1.7	20
87	Novel DyneinDYNC1H1Neck and Motor Domain Mutations Link Distal Spinal Muscular Atrophy and Abnormal Cortical Development. Human Mutation, 2014, 35, 298-302.	1.1	77
88	Myoclonus in mitochondrial disorders. Movement Disorders, 2014, 29, 722-728.	2.2	33
89	EXOSC3 mutations in isolated cerebellar hypoplasia and spinal anterior horn involvement. Journal of Neurology, 2013, 260, 1866-1870.	1.8	28
90	VMA21 deficiency prevents vacuolar ATPase assembly and causes autophagic vacuolar myopathy. Acta Neuropathologica, 2013, 125, 439-457.	3.9	119

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91	Genetic testing in benign familial epilepsies of the first year of life: Clinical and diagnostic significance. Epilepsia, 2013, 54, 425-436.	2.6	110
92	Novel FAM126A mutations in hypomyelination and congenital cataract disease. Biochemical and Biophysical Research Communications, 2013, 439, 369-372.	1.0	12
93	Subclinical myopathy in a child with neutral lipid storage disease and mutations in the PNPLA2 gene. Biochemical and Biophysical Research Communications, 2013, 430, 241-244.	1.0	28
94	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.5	157
95	Hypomyelination and congenital cataract: Identification of novel mutations in two unrelated families. European Journal of Paediatric Neurology, 2013, 17, 108-111.	0.7	7
96	Autosomal recessive epilepsy associated with contactin 2 mutation is different from familial cortical tremor, myoclonus and epilepsy. Brain, 2013, 136, e253-e253.	3.7	7
97	Magnetic Resonance Imaging â€œTigroid Patternâ€•in Alexander Disease. Neuropediatrics, 2013, 44, 174-176.	0.3	9
98	Different electroclinical picture of generalized epilepsy in two families with 15q13.3 microdeletion. Epilepsia, 2013, 54, e69-73.	2.6	14
99	Earlyâ€•onset absence epilepsy: <scp>SLC</scp>2<scp>A</scp>1 gene analysis and treatment evolution. European Journal of Neurology, 2013, 20, 856-859.	1.7	19
100	Clinical dissection of early onset absence epilepsy in children and prognostic implications. Epilepsia, 2013, 54, 1761-1770.	2.6	14
101	Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322.	4.9	61
102	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.5	81
103	Allelic and phenotypic heterogeneity in 49 Italian patients with the muscle form of CPTâ€‖ deficiency. Clinical Genetics, 2012, 82, 232-239.	1.0	40
104	â€˜Autoimmune epilepsyâ€™ or exasperated search for the etiology of seizures of unknown origin?. Epilepsy and Behavior, 2012, 25, 440-441.	0.9	5
105	Fatigue and exercise intolerance in mitochondrial diseases. Literature revision and experience of the Italian Network of mitochondrial diseases. Neuromuscular Disorders, 2012, 22, S226-S229.	0.3	38
106	Hyccin, the Molecule Mutated in the Leukodystrophy Hypomyelination and Congenital Cataract (HCC), Is a Neuronal Protein. PLoS ONE, 2012, 7, e32180.	1.1	20
107	Quality of Life in Duchenne Muscular Dystrophy: The Subjective Impact on Children and Parents. Journal of Child Neurology, 2011, 26, 707-713.	0.7	69
108	Caveolinopathies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2011, 101, 135-142.	1.0	45

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109	Mitochondrial myopathy in a child with a muscle-restricted mutation in the mitochondrial transfer RNAAsn gene. Biochemical and Biophysical Research Communications, 2011, 412, 518-521.	1.0	5
110	Corrigendum to the Letter â€œTemporal lobe epilepsy and hippocampal malrotation: Is there a causal association?â€•[Epilepsy & Behavior 18 (2010) 502â€“504]. Epilepsy and Behavior, 2011, 20, 593.	0.9	0
111	Lumping encephalopathies with inflammation-mediated status epilepticus: Is there enough evidence?. Epilepsy and Behavior, 2011, 20, 592.	0.9	1
112	Natural history and long-term evolution in families with autosomal dominant cortical tremor, myoclonus, and epilepsy. Epilepsia, 2011, 52, 1245-1250.	2.6	45
113	Muscular dystrophy with marked Dysferlin deficiency is consistently caused by primary dysferlin gene mutations. European Journal of Human Genetics, 2011, 19, 974-980.	1.4	67
114	A proofâ€ofâ€concept trial of the whey protein alfaâ€lactalbumin in chronic cortical myoclonus. Movement Disorders, 2011, 26, 2573-2575.	2.2	9
115	Therapeutic approaches in the treatment of juvenile dermatomyositis in patients with recent-onset disease and in those experiencing disease flare: An international multicenter PRINTO study. Arthritis and Rheumatism, 2011, 63, 3142-3152.	6.7	47
116	A clinical and genetic study of 33 new cases with early-onset absence epilepsy. Epilepsy Research, 2011, 95, 221-226.	0.8	15
117	Hyperactivation of oxidative mitochondrial metabolism in epithelial cancer cells in situ. Cell Cycle, 2011, 10, 4047-4064.	1.3	256
118	Hypomyelination and Congenital Cataract. Archives of Neurology, 2011, 68, 1191.	4.9	22
119	West syndrome associated with 14q12 duplications harboring FOXG1. Neurology, 2011, 76, 1600-1602.	1.5	49
120	TBC1D24, an ARF6-Interacting Protein, Is Mutated in Familial Infantile Myoclonic Epilepsy. American Journal of Human Genetics, 2010, 87, 365-370.	2.6	134
121	Old drugs do the trick in childhood absence epilepsy. Nature Reviews Neurology, 2010, 6, 420-421.	4.9	3
122	The spectrum of GNE mutations: allelic heterogeneity for a common phenotype. Neurological Sciences, 2010, 31, 377-380.	0.9	17
123	Familial nonkinesigenic paroxysmal dyskinesia and intracranial calcifications: A new syndrome?. Movement Disorders, 2010, 25, 2468-2470.	2.2	2
124	Caveolinopathies: from the biology of caveolin-3 to human diseases. European Journal of Human Genetics, 2010, 18, 137-145.	1.4	238
125	S100B protein and near infrared spectroscopy in preterm and term newborns. Frontiers in Bioscience - Elite, 2010, E2, 159-164.	0.9	12
126	LESIONAL REFLEX EPILEPSY ASSOCIATED WITH THE THOUGHT OF FOOD. Neurology, 2010, 75, 288-289.	1.5	2

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127	TEMPORAL LOBE ABNORMALITIES ON BRAIN MRI IN HEALTHY VOLUNTEERS: A PROSPECTIVE CASE-CONTROL STUDY. Neurology, 2010, 75, 377-378.	1.5	15
128	Suicide-Related Events in Patients Treated with Antiepileptic Drugs. New England Journal of Medicine, 2010, 363, 1873-1874.	13.9	3
129	Gallowayâ€“Mowat syndrome: An early-onset progressive encephalopathy with intractable epilepsy associated to renal impairment. Two novel cases and review of literature. Seizure: the Journal of the British Epilepsy Association, 2010, 19, 132-135.	0.9	26
130	Temporal lobe epilepsy and hippocampal malrotation: Is there a causal association?. Epilepsy and Behavior, 2010, 18, 502-504.	0.9	9
131	Congenital muscular dystrophies with cognitive impairment. Neurology, 2010, 75, 898-903.	1.5	27
132	Therapeutic Potential of Proteasome Inhibition in Duchenne and Becker Muscular Dystrophies. American Journal of Pathology, 2010, 176, 1863-1877.	1.9	71
133	One Hundred Twenty-One Dystrophin Point Mutations Detected from Stored DNA Samples by Combinatorial Denaturing High-Performance Liquid Chromatography. Journal of Molecular Diagnostics, 2010, 12, 65-73.	1.2	17
134	Congenital muscular dystrophies with defective glycosylation of dystroglycan. Neurology, 2009, 72, 1802-1809.	1.5	166
135	Autoantibodies to glutamic acid decarboxylase (GAD) in focal and generalized epilepsy: A study on 233 patients. Journal of Neuroimmunology, 2009, 211, 120-123.	1.1	74
136	Clinical and translational implications of the caveolin gene family: lessons from mouse models and human genetic disorders. Laboratory Investigation, 2009, 89, 614-623.	1.7	76
137	Familial benign nonprogressive myoclonic epilepsies. Epilepsia, 2009, 50, 37-40.	2.6	11
138	White matter lesions in spastic paraplegia with mutations in SPG5/CYP7B1. Neuromuscular Disorders, 2009, 19, 62-65.	0.3	48
139	Caveolin-1 (P132L), a Common Breast Cancer Mutation, Confers Mammary Cell Invasiveness and Defines a Novel Stem Cell/Metastasis-Associated Gene Signature. American Journal of Pathology, 2009, 174, 1650-1662.	1.9	73
140	Loss of Caveolin-3 Induces a Lactogenic Microenvironment that Is Protective Against Mammary Tumor Formation. American Journal of Pathology, 2009, 174, 613-629.	1.9	20
141	Caveolin-1â^'/â^' Null Mammary Stromal Fibroblasts Share Characteristics with Human Breast Cancer-Associated Fibroblasts. American Journal of Pathology, 2009, 174, 746-761.	1.9	123
142	Dramatic response to levetiracetam in post-ischaemic Holmes' tremor. BMJ Case Reports, 2009, 2009, bcr0820080643-bcr0820080643.	0.2	11
143	Chitosan may decrease serum valproate and increase the risk of seizure reappearance. BMJ: British Medical Journal, 2009, 339, b3751-b3751.	2.4	6
144	Benign adult familial myoclonic epilepsy (BAFME): evidence of an extended founder haplotype on chromosome 2p11.1-q12.2 in five Italian families. Neurogenetics, 2008, 9, 139-142.	0.7	43

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145	Caveolin-3 T78M and T78K missense mutations lead to different phenotypes in vivo and in vitro. Laboratory Investigation, 2008, 88, 275-283.	1.7	37
146	Inclusion body myopathy, Pagetâ€™s disease of the bone and frontotemporal dementia: recurrence of the VCP R155H mutation in an Italian family and implications for genetic counselling. Clinical Genetics, 2008, 74, 54-60.	1.0	37
147	Refractory, life-threatening status epilepticus in a 3-year-old girl. Lancet Neurology, The, 2008, 7, 278-284.	4.9	24
148	Clinical and genetic characterization of Chanarinâ€“Dorfman syndrome. Biochemical and Biophysical Research Communications, 2008, 369, 1125-1128.	1.0	72
149	Aquaporin-4 expression is severely reduced in human sarcoglycanopathies and dysferlinopathies. Cell Cycle, 2008, 7, 2199-2207.	1.3	20
150	Hypomyelination and Congenital Cataract: Neuroimaging Features of a Novel Inherited White Matter Disorder. American Journal of Neuroradiology, 2008, 29, 301-305.	1.2	25
151	Localized Treatment with a Novel FDA-Approved Proteasome Inhibitor Blocks the Degradation of Dystrophin and Dystrophin-Associated Proteins in mdx Mice. Cell Cycle, 2007, 6, 1242-1248.	1.3	67
152	Randomized, double-blind, placebo-controlled trial of phenylbutyrate in spinal muscular atrophy. Neurology, 2007, 68, 51-55.	1.5	159
153	Gene expression profiling in the early phases of DMD: a constant molecular signature characterizes DMD muscle from early postnatal life throughout disease progression. FASEB Journal, 2007, 21, 1210-1226.	0.2	209
154	Muscle and the Cerebellum. Neuroradiology Journal, 2007, 20, 427-433.	0.6	0
155	Inherited neuromyotonia: A clinical and genetic study of a family. Neuromuscular Disorders, 2007, 17, 23-27.	0.3	16
156	The Hammersmith functional score correlates with the SMN2 copy number: A multicentric study. Neuromuscular Disorders, 2007, 17, 400-403.	0.3	47
157	Null mutations and lethal congenital form of glycogen storage disease type IV. Biochemical and Biophysical Research Communications, 2007, 361, 445-450.	1.0	29
158	POMT2 gene mutation in limb-girdle muscular dystrophy with inflammatory changes. Biochemical and Biophysical Research Communications, 2007, 363, 1033-1037.	1.0	91
159	Potential drug targets within bone morphogenetic protein signaling pathways. Current Opinion in Pharmacology, 2007, 7, 325-333.	1.7	27
160	Caveolin-1(â^'/â^') and Caveolin-2(â^'/â^')-Deficient Mice Both Display Numerous Skeletal Muscle Abnormalities, with Tubular Aggregate Formation. American Journal of Pathology, 2007, 170, 316-333.	1.9	59
161	Truncation of Caveolin-3 causes autosomal-recessive Rippling Muscle Disease. Journal of Neurology, Neurosurgery and Psychiatry, 2007, 79, 735-737.	0.9	10
162	International consensus on a proposed score system for muscle biopsy evaluation in patients with juvenile dermatomyositis: A tool for potential use in clinical trials. Arthritis and Rheumatism, 2007, 57, 1192-1201.	6.7	132

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163	Phenotypic characterization of hypomyelination and congenital cataract. Annals of Neurology, 2007, 62, 121-127.	2.8	39
164	Brain MRI Findings in Severe Myoclonic Epilepsy in Infancy and Genotype?Phenotype Correlations. Epilepsia, 2007, 48, 1092-1096.	2.6	89
165	Cerebellar atrophy without cerebellar cortex hyperintensity in infantile neuroaxonal dystrophy (INAD) due to PLA2G6 mutation. European Journal of Paediatric Neurology, 2007, 11, 175-177.	0.7	28
166	Caveolin-1 Deficiency (â^'/â^') Conveys Premalignant Alterations in Mammary Epithelia, with Abnormal Lumen Formation, Growth Factor Independence, and Cell Invasiveness. American Journal of Pathology, 2006, 168, 292-309.	1.9	66
167	Multiplex real-time PCR for detection of deletions and duplications in dystrophin gene. Biochemical and Biophysical Research Communications, 2006, 339, 145-150.	1.0	35
168	Somatic and germline mosaicisms in Severe Myoclonic Epilepsy of Infancy. Biochemical and Biophysical Research Communications, 2006, 341, 489-493.	1.0	102
169	A new method for analysis of mitochondrial DNA point mutations and assess levels of heteroplasmy. Biochemical and Biophysical Research Communications, 2006, 342, 387-393.	1.0	33
170	Reliability of the Hammersmith functional motor scale for spinal muscular atrophy in a multicentric study. Neuromuscular Disorders, 2006, 16, 93-98.	0.3	64
171	A Novel SCN2A Mutation in Family with Benign Familial Infantile Seizures. Epilepsia, 2006, 47, 218-220.	2.6	74
172	Linkage Analysis and Disease Models in Benign Familial Infantile Seizures: A Study of 16 Families. Epilepsia, 2006, 47, 1029-1034.	2.6	23
173	Deficiency of hyccin, a newly identified membrane protein, causes hypomyelination and congenital cataract. Nature Genetics, 2006, 38, 1111-1113.	9.4	82
174	GDAP1 mutation in autosomal recessive Charcot-Marie-Tooth with pyramidal features. Journal of Neurology, 2006, 253, 1234-1235.	1.8	9
175	McArdle disease: the mutation spectrum ofPYGMin a large Italian cohort. Human Mutation, 2006, 27, 718-718.	1.1	52
176	POMGnT1 Mutations in Congenital Muscular Dystrophy. Archives of Neurology, 2006, 63, 1491.	4.9	38
177	Epileptic seizures can follow high doses of oral vardenafil. BMJ: British Medical Journal, 2006, 333, 785.	2.4	17
178	Cryptic chromosome deletions involving SCN1A in severe myoclonic epilepsy of infancy. Neurology, 2006, 67, 1230-1235.	1.5	80
179	Pharmacological rescue of the dystrophin-glycoprotein complex in Duchenne and Becker skeletal muscle explants by proteasome inhibitor treatment. American Journal of Physiology - Cell Physiology, 2006, 290, C577-C582.	2.1	57
180	Dramatic response to levetiracetam in post-ischaemic Holmes' tremor. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 78, 438-439.	0.9	36

#	Article	IF	Citations
181	Mitochondrial DNA Deletion in a Child With Mitochondrial Encephalomyopathy, Growth Hormone Deficiency, and Hypoparathyroidism. Journal of Child Neurology, 2006, 21, 983-985.	0.7	20
182	Loss of Caveolin-1 Causes the Hyper-Proliferation of Intestinal Crypt Stem Cells, with Increased Sensitivity to Whole Body ?-Radiation. Cell Cycle, 2005, 4, 1817-1825.	1.3	73
183	Electroclinical and Genetic Findings in a Family with Cortical Tremor, Myoclonus, and Epilepsy. Epilepsia, 2005, 46, 1993-1995.	2.6	27
184	Different Molecular Mechanisms Leading to White Matter Hypomyelination in Infantile Onset Lysosomal Disorders. Neuropediatrics, 2005, 36, 265-269.	0.3	34
185	Phenotypic Heterogeneity in Two Unrelated Danon Patients Associated with the SameLAMPâ€2Gene Mutation. Neuropediatrics, 2005, 36, 309-313.	0.3	32
186	Caveolin-1-Deficient Mice Have An Increased Mammary Stem Cell Population with Upregulation of Wnt/?-Catenin Signaling. Cell Cycle, 2005, 4, 1808-1816.	1.3	69
187	Chemokine receptor CCR7 is expressed in muscle fibers in juvenile dermatomyositis. Biochemical and Biophysical Research Communications, 2005, 333, 540-543.	1.0	8
188	Caveolinopathies. Neurology, 2004, 62, 538-543.	1.5	177
189	Clinical and genetic heterogeneity of branching enzyme deficiency (glycogenosis type IV). Neurology, 2004, 63, 1053-1058.	1.5	120
190	Middle interhemispheric variant of holoprosencephaly: A very mild clinical case. Neurology, 2004, 63, 2194-2196.	1.5	13
191	Congenital myopathies. Current Neurology and Neuroscience Reports, 2004, 4, 68-73.	2.0	13
192	NovelGNE mutations in Italian families with autosomal recessive hereditary inclusion-body myopathy. Human Mutation, 2004, 23, 632-632.	1.1	52
193	Motor function-muscle strength relationship in spinal muscular atrophy. Muscle and Nerve, 2004, 29, 548-552.	1.0	81
194	Clinical and molecular findings in patients with giant axonal neuropathy (GAN). Neurology, 2004, 62, 13-16.	1.5	62
195	Progressive exercise intolerance associated with a new muscle-restricted nonsense mutation (G142X) in the mitochondrial cytochromeb gene. Muscle and Nerve, 2003, 28, 508-511.	1.0	43
196	Proteasome Inhibitor (MG-132) Treatment of mdx Mice Rescues the Expression and Membrane Localization of Dystrophin and Dystrophin-Associated Proteins. American Journal of Pathology, 2003, 163, 1663-1675.	1.9	111
197	Phosphofructokinase Muscle-Specific Isoform Requires Caveolin-3 Expression for Plasma Membrane Recruitment and Caveolar Targeting. American Journal of Pathology, 2003, 163, 2619-2634.	1.9	32
198	Mitochondrial Myopathy and Respiratory Failure Associated With a New Mutation in the Mitochondrial Transfer Ribonucleic Acid Glutamic Acid Gene. Journal of Child Neurology, 2003, 18, 300-303.	0.7	28

#	Article	IF	Citations
199	Role of Gabapentin in Spinal Muscular Atrophy. Journal of Child Neurology, 2003, 18, 537-541.	0.7	86
200	Dysferlin in a hyperCKaemic patient with caveolin 3 mutation and in C2C12 cells after p38 MAP kinase inhibition. Experimental and Molecular Medicine, 2003, 35, 538-544.	3.2	36
201	Phenotypic behavior of caveolin-3 R26Q, a mutant associated with hyperCKemia, distal myopathy, and rippling muscle disease. American Journal of Physiology - Cell Physiology, 2003, 285, C1150-C1160.	2.1	43
202	Altered aquaporinâ€4 expression in human muscular dystrophies: a common feature?. FASEB Journal, 2002, 16, 1120-1122.	0.2	61
203	Intracellular Retention of Glycosylphosphatidyl Inositol-Linked Proteins in Caveolin-Deficient Cells. Molecular and Cellular Biology, 2002, 22, 3905-3926.	1.1	82
204	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
205	Familial isolated hyperCKaemia associated with a new mutation in the caveolin-3 (CAV-3) gene. Journal of Neurology, Neurosurgery and Psychiatry, 2002, 73, 65-67.	0.9	55
206	Impairment of Caveolae Formation and T-System Disorganization in Human Muscular Dystrophy with Caveolin-3 Deficiency. American Journal of Pathology, 2002, 160, 265-270.	1.9	117
207	Two new mutations in the myophosphorylase gene in Italian patients with McArdle's disease. Neuromuscular Disorders, 2002, 12, 498-500.	0.3	7
208	Caveolin-3 Null Mice Show a Loss of Caveolae, Changes in the Microdomain Distribution of the Dystrophin-Glycoprotein Complex, and T-tubule Abnormalities. Journal of Biological Chemistry, 2001, 276, 21425-21433.	1.6	385
209	Mutation in the CAV3 gene causes partial caveolin-3 deficiency and persistent elevated levels of serum creatine kinase. Neurology, 2000, 54, 1373-1376.	1.5	158
210	Limb-girdle Muscular Dystrophy (LGMD-1C) Mutants of Caveolin-3 Undergo Ubiquitination and Proteasomal Degradation. Journal of Biological Chemistry, 2000, 275, 37702-37711.	1.6	86
211	Transgenic overexpression of caveolin-3 in skeletal muscle fibers induces a Duchenne-like muscular dystrophy phenotype. Proceedings of the National Academy of Sciences of the United States of America, 2000, 97, 9689-9694.	3.3	132
212	Novel Mutation in the CPT II Gene in a Child With Periodic Febrile Myalgia and Myoglobinuria. Journal of Child Neurology, 2000, 15, 390-393.	0.7	15
213	Caveolin-3 Directly Interacts with the C-terminal Tail of Î²-Dystroglycan. Journal of Biological Chemistry, 2000, 275, 38048-38058.	1.6	181
214	Phenotypic Behavior of Caveolin-3 Mutations That Cause Autosomal Dominant Limb Girdle Muscular Dystrophy (LGMD-1C). Journal of Biological Chemistry, 1999, 274, 25632-25641.	1.6	141
215	A novel missense mutation in the glycogen branching enzyme gene in a child with myopathy and hepatopathy. Neuromuscular Disorders, 1999, 9, 403-407.	0.3	25
216	Clinical and genetic heterogeneity in autosomal recessive nemaline myopathy. Neuromuscular Disorders, 1999, 9, 564-572.	0.3	84

#	Article	IF	Citations
217	Muscular dystrophies, alterations in a limited number of cellular pathways?. Current Opinion in Genetics and Development, 1999, 9, 275-282.	1.5	21
218	Localization of the human caveolin-3 gene to the D3S18/D3S4163/D3S4539 locus (3p25), in close proximity to the human oxytocin receptor gene. FEBS Letters, 1999, 452, 177-180.	1.3	21
219	Early Decrease of IIx Myosin Heavy Chain Transcripts in Duchenne Muscular Dystrophy. Biochemical and Biophysical Research Communications, 1999, 255, 466-469.	1.0	35
220	Increased Number of Caveolae and Caveolin-3 Overexpression in Duchenne Muscular Dystrophy. Biochemical and Biophysical Research Communications, 1999, 261, 547-550.	1.0	93
221	Mutations in the caveolin-3 gene cause autosomal dominant limb-girdle muscular dystrophy. Nature Genetics, 1998, 18, 365-368.	9.4	555
222	Disorganization of dystrophin costameric lattice in Becker muscular dystrophy. , 1998, 21, 211-216.		11
223	Very-long-chain acyl-coenzyme A dehydrogenase deficiency in a child with recurrent myoglobinuria. Neuromuscular Disorders, 1998, 8, 3-6.	0.3	27
224	Molecular Genetics of the Caveolin Gene Family: Implications for Human Cancers, Diabetes, Alzheimer Disease, and Muscular Dystrophy. American Journal of Human Genetics, 1998, 63, 1578-1587.	2.6	171
225	Combined defects of muscle phosphofructokinase and AMP deaminase in a child with myoglobinuria. Neurology, 1998, 50, 296-298.	1.5	25
226	Forearm Semi-ischemic Exercise Test in Pediatric Patients. Journal of Child Neurology, 1998, 13, 288-290.	0.7	15
227	Ubiquitin expression in acute steroid myopathy with loss of myosin thick filaments. , 1996, 19, 94-96.		18
228	Severe dystrophinopathy in a patient with congenital hypotonia. Child's Nervous System, 1996, 12, 466-9.	0.6	3
229	Disruption of muscle basal lamina in congenital muscular dystrophy with merosin deficiency. Neurology, 1996, 46, 1354-1354.	1.5	41
230	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). Neurology, 1994, 44, 721-721.	1.5	402
231	Abnormalities in the expression of Î²â€spectrin in Duchenne muscular dystrophy. Neurology, 1994, 44, 1149-1149.	1.5	16
232	Dystrophinopathy in two young boys with exercise-induced cramps and myoglobinuria. European Journal of Pediatrics, 1993, 152, 848-851.	1.3	23
233	Mosaic Expression of Dystrophin in Carriers of Becker's Muscular Dystrophy and the X-Linked Syndrome of Myalgia and Cramps. New England Journal of Medicine, 1992, 327, 1100-1100.	13.9	3
234	Acute quadriplegic myopathy. Neurology, 1992, 42, 2082-2082.	1.5	247

#	Article	IF	Citations
235	Dystrophin at the plasma membrane of human muscle fibers shows a costameric localization. Neuromuscular Disorders, 1992, 2, 99-109.	0.3	69
236	Carrier detection of duchenne and becker muscular dystrophy using muscle dystrophin immunohistochemistry. Arquivos De Neuro-Psiquiatria, 1992, 50, 478-485.	0.3	4
237	Dystrophinopathy in isolated cases of myopathy in females. Neurology, 1992, 42, 967-967.	1.5	149
238	Immunologic study of vinculin in Duchenne muscular dystrophy. Neurology, 1992, 42, 1751-1751.	1.5	24
239	Becker muscular dystrophy or spinal muscular atrophy?â€"Dystrophin studies resolve conflicting results of electromyography and muscle biopsy. Neuromuscular Disorders, 1991, 1, 195-200.	0.3	11
240	Progressive depletion of fast alphaâ€actininâ€positive muscle fibers in Duchenne muscular dystrophy. Neurology, 1991, 41, 1977-1977.	1.5	26
241	Dystrophin deficiency in young girls with sporadic myopathy and normal karyotype. Neurology, 1991, 41, 1288-1288.	1.5	25
242	Mitochondrial Encephalomyopathies. Neurologic Clinics, 1990, 8, 483-506.	0.8	51
243	A Case of Congenital Neuromuscular Disease with Uniform Type I Fibers, Abnormal Mitochondrial Network and Jagged Z-Line. Neuropediatrics, 1985, 16, 162-166.	0.3	13
244	Familial Nemaline Myopathy. Neuropediatrics, 1982, 13, 211-215.	0.3	16