Claudio Bruno

List of Publications by Year
in descending order

202	8,994	36303 51	60623 81
papers	citations	h-index	g-index

205	205	205	9177
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Spinal muscular atrophy: state of the art and new therapeutic strategies. Neurological Sciences, 2022, 43, 615-624.	1.9	13
2	Clinical and genetic spectrum of a large cohort of patients with Î´-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606.	7.6	11
3	An integrated approach to the evaluation of patients with asymptomatic or minimally symptomatic <scp>hyperCKemia</scp>. Muscle and Nerve, 2022, 65, 96-104.	2.2	10
4	P2X7 Receptor Antagonist Reduces Fibrosis and Inflammation in a Mouse Model of Alpha-Sarcoglycan Muscular Dystrophy. Pharmaceuticals, 2022, 15, 89.	3.8	11
5	Nusinersen efficacy data for 24â€month in type 2 and 3 spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2022, 9, 404-409.	3.7	22
6	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
7	Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894.	3.6	2
8	Respiratory Trajectories in Type 2 and 3 Spinal Muscular Atrophy in the iSMAC Cohort Study. Neurology, 2021, 96, e587-e599.	1.1	36
9	Type I SMA â€œnew natural historyâ€: longâ€term data in nusinersenâ€treated patients. Annals of Clinical and Translational Neurology, 2021, 8, 548-557.	3.7	35
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.	2.4	8
11	Nusinersen in pediatric and adult patients with type III spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2021, 8, 1622-1634.	3.7	27
12	North Star Ambulatory Assessment changes in ambulant Duchenne boys amenable to skip exons 44, 45, 51, and 53: A 3 year follow up. PLoS ONE, 2021, 16, e0253882.	2.5	6
13	The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 479-488.	0.6	0
14	Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. Neuromuscular Disorders, 2021, 31, 596-602.	0.6	29
15	Risdiplam-Treated Infants with Type 1 Spinal Muscular Atrophy versus Historical Controls. New England Journal of Medicine, 2021, 385, 427-435.	27.0	137
16	SMA-miRs (miR-181a-5p, -324-5p, and -451a) are overexpressed in spinal muscular atrophy skeletal muscle and serum samples. ELife, 2021, 10, .	6.0	13
17	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
18	The Role of Muscle Biopsy in Diagnostic Process of Infant Hypotonia: From Clinical Classification to the Genetic Outcome. Frontiers in Neurology, 2021, 12, 735488.	2.4	7

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19	Anterior cingulate and medial prefrontal cortex response to systematically controlled tonal dissonance during passive music listening. Human Brain Mapping, 2020, 41, 46-66.	3.6	10
20	Novel compound heterozygous pathogenic variants in nucleotide-binding protein like protein (NUBPL) cause leukoencephalopathy with multi-systemic involvement. Molecular Genetics and Metabolism, 2020, 129, 26-34.	1.1	9
21	Creation and implementation of a European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC registry). Orphanet Journal of Rare Diseases, 2020, 15, 187.	2.7	3
22	Genotypeâ€“phenotype correlations in recessive titinopathies. Genetics in Medicine, 2020, 22, 2029-2040.	2.4	35
23	eATP/P2X7R Axis: An Orchestrated Pathway Triggering Inflammasome Activation in Muscle Diseases. International Journal of Molecular Sciences, 2020, 21, 5963.	4.1	11
24	The Genetic Landscape of Dystrophin Mutations in Italy: A Nationwide Study. Frontiers in Genetics, 2020, 11, 131.	2.3	49
25	Respiratory Needs in Patients with Type 1 Spinal Muscular Atrophy TreatedÂwith Nusinersen. Journal of Pediatrics, 2020, 219, 223-228.e4.	1.8	51
26	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	3.7	36
27	Tele-monitoring in paediatric and young home-ventilated neuromuscular patients: A multicentre case-control trial. Journal of Telemedicine and Telecare, 2019, 25, 414-424.	2.7	21
28	Novel homozygous TSFM pathogenic variant associated with encephalocardiomyopathy with sensorineural hearing loss and peculiar neuroradiologic findings. Neurogenetics, 2019, 20, 165-172.	1.4	8
29	Nusinersen in type 1 spinal muscular atrophy: Twelveâ€month realâ€world data. Annals of Neurology, 2019, 86, 443-451.	5.3	83
30	Long-term natural history data in Duchenne muscular dystrophy ambulant patients with mutations amenable to skip exons 44, 45, 51 and 53. PLoS ONE, 2019, 14, e0218683.	2.5	47
31	Clinical and neuroimaging features of the m.10197G>A mtDNA mutation: New case reports and expansion of the phenotype variability. Journal of the Neurological Sciences, 2019, 399, 69-75.	0.6	8
32	Italian recommendations for diagnosis and management of congenital myasthenic syndromes. Neurological Sciences, 2019, 40, 457-468.	1.9	24
33	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.	3.8	9
34	Study Design of STR1VE-EU, a Phase 3 Trial of AVXS-101 Gene-Replacement Therapy (GRT) in Patients With Spinal Muscular Atrophy Type 1 (SMA1) in Europe. , 2019, 50, .		0
35	Unusual white matter involvement in EAST syndrome associated with novel KCNJ10 mutations. Journal of Neurology, 2018, 265, 1419-1425.	3.6	8
36	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.6	24

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37	A 5-center experience with intrathecal administration of nusinersen in SMA1 in Italy letter to the editor of european journal of pediatric neurology regarding the manuscript â€œsingle-center experience with intrathecal administration of nusinersen in children with spinal muscular atrophy type 1â€• written by pechmann and colleaguesâ€• European Journal of Paediatric Neurology, 2018, 22, 729-731.	1.6	5
38	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	9.0	69
39	Zidovudine ameliorates pathology in the mouse model of Duchenne muscular dystrophy via P2RX7 purinoceptor antagonism. Acta Neuropathologica Communications, 2018, 6, 27.	5.2	30
40	Expanding the histopathological spectrum of CFL2â€related myopathies. Clinical Genetics, 2018, 93, 1234-1239.	2.0	11
41	Clinical and molecular consequences of exon 78 deletion in DMD gene. Journal of Human Genetics, 2018, 63, 761-764.	2.3	7
42	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	1.9	55
43	Detection of early nocturnal hypoventilation in neuromuscular disorders. Journal of International Medical Research, 2018, 46, 1153-1161.	1.0	22
44	Broad phenotypic spectrum and genotype-phenotype correlations in GMPPB-related dystroglycanopathies: an Italian cross-sectional study. Orphanet Journal of Rare Diseases, 2018, 13, 170.	2.7	26
45	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. Genes, 2018, 9, 524.	2.4	7
46	Upper limb function in Duchenne muscular dystrophy: 24 month longitudinal data. PLoS ONE, 2018, 13, e0199223.	2.5	45
47	An observational study of functional abilities in infants, children, and adults with type 1 SMA. Neurology, 2018, 91, e696-e703.	1.1	24
48	Nusinersen in type 1 SMA infants, children and young adults: Preliminary results on motor function. Neuromuscular Disorders, 2018, 28, 582-585.	0.6	67
49	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86
50	Diagnosis of Duchenne Muscular Dystrophy in Italy in the last decade: Critical issues and areas for improvements. Neuromuscular Disorders, 2017, 27, 447-451.	0.6	42
51	Muscle MRI in neutral lipid storage disease (NLSD). Journal of Neurology, 2017, 264, 1334-1342.	3.6	15
52	Mutations in GMPPB Presenting with Pseudometabolic Myopathy. JIMD Reports, 2017, 38, 23-31.	1.5	8
53	Expanded access program with Nusinersen in SMA type I in Italy: Strengths and pitfalls of a successful experience. Neuromuscular Disorders, 2017, 27, 1084-1086.	0.6	38
54	Gait disturbance and lower limb pain in a patient with PIK3CA -related disorder. European Journal of Medical Genetics, 2017, 60, 655-657.	1.3	3

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55	Respiratory pattern in a FSDH paediatric population. Respiratory Medicine, 2017, 126, 132.	2.9	0
56	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	3.6	32
57	Neural mechanisms underlying valence inferences to sound: The role of the right angular gyrus. Neuropsychologia, 2017, 102, 144-162.	1.6	6
58	Neutral Lipid Storage Diseases: clinical/genetic features and natural history in a large cohort of Italian patients. Orphanet Journal of Rare Diseases, 2017, 12, 90.	2.7	49
59	Congenital myopathies: clinical phenotypes and new diagnostic tools. Italian Journal of Pediatrics, 2017, 43, 101.	2.6	80
60	Tele-monitoring in paediatric neuromuscular patients requiring home mechanical ventilation, multicentric study. , 2017, , .		0
61	Timed Rise from Floor as a Predictor of Disease Progression in Duchenne Muscular Dystrophy: An Observational Study. PLoS ONE, 2016, 11, e0151445.	2.5	32
62	Clinical expression of facioscapulohumeral muscular dystrophy in carriers of 1â€“3 D4Z4 reduced alleles: experience of the FSHD Italian National Registry. BMJ Open, 2016, 6, e007798.	1.9	60
63	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
64	Registries versus tertiary care centers: How do we measure standards of care in Duchenne muscular dystrophy?. Neuromuscular Disorders, 2016, 26, 261-263.	0.6	3
65	Respiratory pattern in a FSHD pediatric population. Respiratory Medicine, 2016, 119, 78-80.	2.9	7
66	Categorizing natural history trajectories of ambulatory function measured by the 6-minute walk distance in patients with Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, 576-583.	0.6	57
67	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.	3.3	18
68	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.1	92
69	Dolichol-phosphate mannose synthase depletion in zebrafish leads to dystrophic muscle with hypoglycosylated Î±-dystroglycan. Biochemical and Biophysical Research Communications, 2016, 477, 137-143.	2.1	17
70	The ubiquitin ligase tripartite-motif-protein 32 is induced in Duchenne muscular dystrophy. Laboratory Investigation, 2016, 96, 862-871.	3.7	23
71	Genetic and Early Clinical Manifestations of Females Heterozygous for Duchenne/Becker Muscular Dystrophy. Pediatric Neurology, 2016, 55, 58-63.	2.1	37
72	Interstitial 9p24.3 deletion involving only DOCK8 and KANK1 genes in two patients with non-overlapping phenotypic traits. European Journal of Medical Genetics, 2016, 59, 20-25.	1.3	17

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73	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. Neuromuscular Disorders, 2016, 26, 292-299.	0.6	25
74	â€œMitochondrial neuropathiesâ€: A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.6	37
75	Health-related quality of life and functional changes in DMD: A 12-month longitudinal cohort study. Neuromuscular Disorders, 2016, 26, 189-196.	0.6	32
76	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.1	75
77	Benefits of glucocorticoids in non-ambulant boys/men with Duchenne muscular dystrophy: A multicentric longitudinal study using the Performance of Upper Limb test. Neuromuscular Disorders, 2015, 25, 749-753.	0.6	41
78	Reading impairment in Duchenne muscular dystrophy: A pilot study to investigate similarities and differences with developmental dyslexia. Research in Developmental Disabilities, 2015, 45-46, 168-177.	2.2	13
79	Paternal germline mosaicism in collagen VI related myopathies. European Journal of Paediatric Neurology, 2015, 19, 533-536.	1.6	15
80	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
81	Early onset cardiomyopathy associated with the mitochondrial tRNALeu(UUR) 3271T>C MELAS mutation. Biochemical and Biophysical Research Communications, 2015, 458, 601-604.	2.1	14
82	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	3.6	68
83	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.	1.5	15
84	Inferior Olivary Nucleus Involvement in Pediatric Neurodegenerative Disorders: Does It Play a Role in Neuroimaging Pattern-Recognition Approach?. Neuropediatrics, 2015, 46, 104-109.	0.6	4
85	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.	3.8	42
86	Long Term Natural History Data in Ambulant Boys with Duchenne Muscular Dystrophy: 36-Month Changes. PLoS ONE, 2014, 9, e108205.	2.5	98
87	MotorPlex provides accurate variant detection across large muscle genes both in single myopathic patients and in pools of DNA samples. Acta Neuropathologica Communications, 2014, 2, 100.	5.2	76
88	6 Minute Walk Test in Duchenne MD Patients with Different Mutations: 12 Month Changes. PLoS ONE, 2014, 9, e83400.	2.5	65
89	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. Neuromuscular Disorders, 2014, 24, 201-206.	0.6	83
90	Acitretinâ€Responsive Ichthyosis in Chanarinâ€“Dorfman Syndrome with a Novel Mutation in the <scp>ABHD</scp>5/<scp>CGI</scp>â€58 Gene. Pediatric Dermatology, 2014, 31, 612-614.	0.9	13

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91	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	3.6	119
92	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.	2.0	4
93	Vaccination recommendations for patients with neuromuscular disease. Vaccine, 2014, 32, 5893-5900.	3.8	20
94	Novel DyneinDYNC1H1Neck and Motor Domain Mutations Link Distal Spinal Muscular Atrophy and Abnormal Cortical Development. Human Mutation, 2014, 35, 298-302.	2.5	77
95	Myoclonus in mitochondrial disorders. Movement Disorders, 2014, 29, 722-728.	3.9	33
96	The 6 Minute Walk Test and Performance of Upper Limb in Ambulant Duchenne Muscular Dystrophy Boys. PLOS Currents, 2014, 6, .	1.4	24
97	Neuromuscular Disorders in Zebrafish: State of the Art and Future Perspectives. NeuroMolecular Medicine, 2013, 15, 405-419.	3.4	10
98	Dopamine-agonist responsive Parkinsonism in a patient with the SANDO syndrome caused by POLG mutation. BMC Medical Genetics, 2013, 14, 105.	2.1	25
99	Neuromuscular Disorders of Glycogen Metabolism. Current Neurology and Neuroscience Reports, 2013, 13, 333.	4.2	14
100	Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients. Journal of Inherited Metabolic Disease, 2013, 36, 43-53.	3.6	70
101	Centronuclear myopathy related to dynamin 2 mutations: Clinical, morphological, muscle imaging and genetic features of an Italian cohort. Neuromuscular Disorders, 2013, 23, 229-238.	0.6	53
102	Novel mutations in the fukutin gene in a boy with asymptomatic hyperCKemia. Neuromuscular Disorders, 2013, 23, 1010-1015.	0.6	5
103	Duchenne muscular dystrophy and epilepsy. Neuromuscular Disorders, 2013, 23, 313-315.	0.6	60
104	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.	2.1	28
105	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.1	157
106	Metabolic Consequences of Adipose Triglyceride Lipase Deficiency in Humans: An In Vivo Study in Patients With Neutral Lipid Storage Disease With Myopathy. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1540-E1548.	3.6	23
107	Mitochondrial Encephalomyopathy Due to a Novel Mutation inACAD9. JAMA Neurology, 2013, 70, 1177.	9.0	38
108	Susceptibility-Weighted Angiography of Intracranial Blood Products and Calcifications Compared to Gradient Echo Sequence. Neuroradiology Journal, 2013, 26, 493-500.	1.2	15

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109	24 Month Longitudinal Data in Ambulant Boys with Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e52512.	2.5	99
110	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.1	81
111	Clinical features and new molecular findings in muscle phosphofructokinase deficiency (GSD type VII). Neuromuscular Disorders, 2012, 22, 325-330.	0.6	44
112	Allelic and phenotypic heterogeneity in 49 Italian patients with the muscle form of CPTâ€Ⅱ deficiency. Clinical Genetics, 2012, 82, 232-239.	2.0	40
113	TRPV4 mutations in children with congenital distal spinal muscular atrophy. Neurogenetics, 2012, 13, 195-203.	1.4	31
114	Respiratory and cardiac function in congenital muscular dystrophies with alpha dystroglycan deficiency. Neuromuscular Disorders, 2012, 22, 685-689.	0.6	31
115	Intermittentâ€relapsing pyruvate dehydrogenase complex deficiency: a case with clinical, biochemical, and neuroradiological reversibility. Developmental Medicine and Child Neurology, 2012, 54, 472-476.	2.1	18
116	Involvement of the Globus Pallidus in Giant Axonal Neuropathy. Pediatric Neurology, 2012, 47, 382-384.	2.1	3
117	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.	2.1	5
118	Cerebellar hypoplasia and brainstem thinning associated with severe white matter and basal ganglia abnormalities in a child with an mtDNA deletion. Journal of Inherited Metabolic Disease, 2011, 34, 1225-1227.	3.6	3
119	Genotype-phenotype correlations in a large series of patients with muscle type CPT II deficiency. Neurological Research, 2011, 33, 24-32.	1.3	46
120	The spectrum of GNE mutations: allelic heterogeneity for a common phenotype. Neurological Sciences, 2010, 31, 377-380.	1.9	17
121	Caveolinopathies: from the biology of caveolin-3 to human diseases. European Journal of Human Genetics, 2010, 18, 137-145.	2.8	238
122	SMN transcript levels in leukocytes of SMA patients determined by absolute real-time PCR. European Journal of Human Genetics, 2010, 18, 52-58.	2.8	47
123	Cavitating Leukoencephalopathy in a Child Carrying the MitochondrialA8344GMutation: Fig 1.. American Journal of Neuroradiology, 2010, 31, E78-E79.	2.4	17
124	North Star Ambulatory Assessment, 6-minute walk test and timed items in ambulant boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2010, 20, 712-716.	0.6	171
125	Therapeutic Potential of Proteasome Inhibition in Duchenne and Becker Muscular Dystrophies. American Journal of Pathology, 2010, 176, 1863-1877.	3.8	71
126	Screening of ARHSP-TCC patients expands the spectrum ofSPG11mutations and includes a large scale gene deletion. Human Mutation, 2009, 30, E500-E519.	2.5	53

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127	Reliability of the North Star Ambulatory Assessment in a multicentric setting. Neuromuscular Disorders, 2009, 19, 458-461.	0.6	171
128	Unusual presentation of phosphoglycerate mutase deficiency due to two different mutations in PGAM-M gene. Neuromuscular Disorders, 2009, 19, 776-778.	0.6	19
129	Adult polyglucosan body disease: Proton magnetic resonance spectroscopy of the brain and novel mutation in the GBE1 gene. Muscle and Nerve, 2008, 37, 530-536.	2.2	27
130	Peroxisomal acylâ€CoAâ€oxidase deficiency: Two new cases. American Journal of Medical Genetics, Part A, 2008, 146A, 1676-1681.	1.2	31
131	Caveolin-3 T78M and T78K missense mutations lead to different phenotypes in vivo and in vitro. Laboratory Investigation, 2008, 88, 275-283.	3.7	37
132	Facioscapulohumeral muscular dystrophy: hearing loss and other atypical features of patients with large 4q35 deletions. European Journal of Neurology, 2008, 15, 1353-1358.	3.3	45
133	POMT1 and POMT2 mutations in CMD patients: A multicentric Italian study. Neuromuscular Disorders, 2008, 18, 565-571.	0.6	38
134	Unclassified polysaccharidosis of the heart and skeletal muscle in siblings. Molecular Genetics and Metabolism, 2008, 95, 52-58.	1.1	12
135	Clinical and genetic characterization of Chanarinâ€“Dorfman syndrome. Biochemical and Biophysical Research Communications, 2008, 369, 1125-1128.	2.1	72
136	Novel mutations in the adipose triglyceride lipase gene causing neutral lipid storage disease with myopathy. Biochemical and Biophysical Research Communications, 2008, 377, 843-846.	2.1	58
137	Aquaporin-4 expression is severely reduced in human sarcoglycanopathies and dysferlinopathies. Cell Cycle, 2008, 7, 2199-2207.	2.6	20
138	Hypomyelination and Congenital Cataract: Neuroimaging Features of a Novel Inherited White Matter Disorder. American Journal of Neuroradiology, 2008, 29, 301-305.	2.4	25
139	Lipid storage myopathies. Current Opinion in Neurology, 2008, 21, 601-606.	3.6	67
140	Fatal Infantile Cardiac Glycogenosis with Phosphorylase Kinase Deficiency and a Mutation in the Î³2-Subunit of AMP-Activated Protein Kinase. Pediatric Research, 2007, 62, 499-504.	2.3	57
141	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.	0.5	209
142	Clinical and electrophysiological features of epilepsy in Italian patients with CLN8 mutations. Epilepsy and Behavior, 2007, 10, 187-191.	1.7	26
143	The Hammersmith functional score correlates with the SMN2 copy number: A multicentric study. Neuromuscular Disorders, 2007, 17, 400-403.	0.6	47
144	The NDUFB11 gene is not a modifier in Leber hereditary optic neuropathy. Biochemical and Biophysical Research Communications, 2007, 355, 181-187.	2.1	13

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145	Null mutations and lethal congenital form of glycogen storage disease type IV. Biochemical and Biophysical Research Communications, 2007, 361, 445-450.	2.1	29
146	POMT2 gene mutation in limb-girdle muscular dystrophy with inflammatory changes. Biochemical and Biophysical Research Communications, 2007, 363, 1033-1037.	2.1	91
147	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.	3.8	59
148	Phenotypic characterization of hypomyelination and congenital cataract. Annals of Neurology, 2007, 62, 121-127.	5.3	39
149	Allogeneic bone marrow transplantation for Pearson's syndrome. Bone Marrow Transplantation, 2007, 39, 563-565.	2.4	21
150	Multiplex real-time PCR for detection of deletions and duplications in dystrophin gene. Biochemical and Biophysical Research Communications, 2006, 339, 145-150.	2.1	35
151	A new method for analysis of mitochondrial DNA point mutations and assess levels of heteroplasmy. Biochemical and Biophysical Research Communications, 2006, 342, 387-393.	2.1	33
152	Reliability of the Hammersmith functional motor scale for spinal muscular atrophy in a multicentric study. Neuromuscular Disorders, 2006, 16, 93-98.	0.6	64
153	Deficiency of hyccin, a newly identified membrane protein, causes hypomyelination and congenital cataract. Nature Genetics, 2006, 38, 1111-1113.	21.4	82
154	Novel mutations in CLN8 in Italian variant late infantile neuronal ceroid lipofuscinosis: another genetic hit in the Mediterranean. Neurogenetics, 2006, 7, 111-117.	1.4	43
155	High plasma creatine kinase: review of the literature and proposal for a diagnostic algorithm. Neurological Sciences, 2006, 27, 303-311.	1.9	60
156	McArdle disease: the mutation spectrum ofPYGMin a large Italian cohort. Human Mutation, 2006, 27, 718-718.	2.5	52
157	POMGnT1 Mutations in Congenital Muscular Dystrophy. Archives of Neurology, 2006, 63, 1491.	4.5	38
158	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.	4.6	57
159	Mitochondrial DNA Deletion in a Child With Mitochondrial Encephalomyopathy, Growth Hormone Deficiency, and Hypoparathyroidism. Journal of Child Neurology, 2006, 21, 983-985.	1.4	20
160	Phenotypic Heterogeneity in Two Unrelated Danon Patients Associated with the SameLAMPâ€2Gene Mutation. Neuropediatrics, 2005, 36, 309-313.	0.6	32
161	Chemokine receptor CCR7 is expressed in muscle fibers in juvenile dermatomyositis. Biochemical and Biophysical Research Communications, 2005, 333, 540-543.	2.1	8
162	Severe neonatal onset of glycogenosis type IV: Clinical and laboratory findings leading to diagnosis in two siblings. Journal of Inherited Metabolic Disease, 2004, 27, 609-620.	3.6	21

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163	Congenital myopathies. Current Neurology and Neuroscience Reports, 2004, 4, 68-73.	4.2	13
164	NovelGNE mutations in Italian families with autosomal recessive hereditary inclusion-body myopathy. Human Mutation, 2004, 23, 632-632.	2.5	52
165	Respiratory Complex III Is Required to Maintain Complex I in Mammalian Mitochondria. Molecular Cell, 2004, 13, 805-815.	9.7	402
166	Progressive exercise intolerance associated with a new muscle-restricted nonsense mutation (G142X) in the mitochondrial cytochromeb gene. Muscle and Nerve, 2003, 28, 508-511.	2.2	43
167	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.	1.4	28
168	Leigh Syndrome with COX deficiency and SURF1 gene mutations: MR imaging findings. American Journal of Neuroradiology, 2003, 24, 1188-91.	2.4	69
169	A Novel Mutation in the SURF1 Gene in a Child With Leigh Disease, Peripheral Neuropathy, and Cytochrome-c Oxidase Deficiency. Journal of Child Neurology, 2002, 17, 233-236.	1.4	31
170	Myophosphorylase Deficiency (Glycogenosis Type V McArdle Disease). Current Molecular Medicine, 2002, 2, 189-196.	1.3	63
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