Claudio Bruno

List of Publications by Year
in descending order

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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	Mutations in the caveolin-3 gene cause autosomal dominant limb-girdle muscular dystrophy. Nature Genetics, 1998, 18, 365-368.	21.4	555
2	Exercise Intolerance Due to Mutations in the CytochromebGene of Mitochondrial DNA. New England Journal of Medicine, 1999, 341, 1037-1044.	27.0	408
3	Respiratory Complex III Is Required to Maintain Complex I in Mammalian Mitochondria. Molecular Cell, 2004, 13, 805-815.	9.7	402
4	Caveolinopathies: from the biology of caveolin-3 to human diseases. European Journal of Human Genetics, 2010, 18, 137-145.	2.8	238
5	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
6	Reliability of the North Star Ambulatory Assessment in a multicentric setting. Neuromuscular Disorders, 2009, 19, 458-461.	0.6	171
7	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
8	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.1	157
9	A Stop-Codon Mutation in the Human mtDNA Cytochrome c Oxidase I Gene Disrupts the Functional Structure of Complex IV. American Journal of Human Genetics, 1999, 65, 611-620.	6.2	148
10	Risdiplam-Treated Infants with Type 1 Spinal Muscular Atrophy versus Historical Controls. New England Journal of Medicine, 2021, 385, 427-435.	27.0	137
11	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	3.6	119
12	Impairment of Caveolae Formation and T-System Disorganization in Human Muscular Dystrophy with Caveolin-3 Deficiency. American Journal of Pathology, 2002, 160, 265-270.	3.8	117
13	Glycogen branching enzyme deficiency in adult polyglucosan body disease. Annals of Neurology, 1993, 33, 88-93.	5.3	112
14	24 Month Longitudinal Data in Ambulant Boys with Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e52512.	2.5	99
15	Long Term Natural History Data in Ambulant Boys with Duchenne Muscular Dystrophy: 36-Month Changes. PLoS ONE, 2014, 9, e108205.	2.5	98
16	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.1	92
17	POMT2 gene mutation in limb-girdle muscular dystrophy with inflammatory changes. Biochemical and Biophysical Research Communications, 2007, 363, 1033-1037.	2.1	91
18	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86

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19	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. Neuromuscular Disorders, 2014, 24, 201-206.	0.6	83
20	Nusinersen in type 1 spinal muscular atrophy: Twelveâ€month realâ€world data. Annals of Neurology, 2019, 86, 443-451.	5.3	83
21	Deficiency of hyccin, a newly identified membrane protein, causes hypomyelination and congenital cataract. Nature Genetics, 2006, 38, 1111-1113.	21.4	82
22	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.1	81
23	Congenital myopathies: clinical phenotypes and new diagnostic tools. Italian Journal of Pediatrics, 2017, 43, 101.	2.6	80
24	Novel DyneinDYNC1H1Neck and Motor Domain Mutations Link Distal Spinal Muscular Atrophy and Abnormal Cortical Development. Human Mutation, 2014, 35, 298-302.	2.5	77
25	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
26	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.1	75
27	Clinical and genetic characterization of Chanarinâ€“Dorfman syndrome. Biochemical and Biophysical Research Communications, 2008, 369, 1125-1128.	2.1	72
28	Therapeutic Potential of Proteasome Inhibition in Duchenne and Becker Muscular Dystrophies. American Journal of Pathology, 2010, 176, 1863-1877.	3.8	71
29	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
30	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	9.0	69
31	Leigh Syndrome with COX deficiency and SURF1 gene mutations: MR imaging findings. American Journal of Neuroradiology, 2003, 24, 1188-91.	2.4	69
32	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	3.6	68
33	Lipid storage myopathies. Current Opinion in Neurology, 2008, 21, 601-606.	3.6	67
34	Nusinersen in type 1 SMA infants, children and young adults: Preliminary results on motor function. Neuromuscular Disorders, 2018, 28, 582-585.	0.6	67
35	In Vivo Regulation of Oxidative Phosphorylation in Cells Harboring a Stop-codon Mutation in Mitochondrial DNA-encoded Cytochrome c Oxidase Subunit I. Journal of Biological Chemistry, 2001, 276, 46925-46932.	3.4	66
36	6 Minute Walk Test in Duchenne MD Patients with Different Mutations: 12 Month Changes. PLoS ONE, 2014, 9, e83400.	2.5	65

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37	Reliability of the Hammersmith functional motor scale for spinal muscular atrophy in a multicentric study. Neuromuscular Disorders, 2006, 16, 93-98.	0.6	64
38	Myophosphorylase Deficiency (Glycogenosis Type V McArdle Disease). Current Molecular Medicine, 2002, 2, 189-196.	1.3	63
39	Infantile encephalopathy associated with the MELAS A3243G mutation. Journal of Pediatrics, 1999, 134, 696-700.	1.8	62
40	High plasma creatine kinase: review of the literature and proposal for a diagnostic algorithm. Neurological Sciences, 2006, 27, 303-311.	1.9	60
41	Duchenne muscular dystrophy and epilepsy. Neuromuscular Disorders, 2013, 23, 313-315.	0.6	60
42	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
43	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
44	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
45	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
46	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
47	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
48	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	1.9	55
49	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
50	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
51	NovelGNE mutations in Italian families with autosomal recessive hereditary inclusion-body myopathy. Human Mutation, 2004, 23, 632-632.	2.5	52
52	McArdle disease: the mutation spectrum ofPYGMin a large Italian cohort. Human Mutation, 2006, 27, 718-718.	2.5	52
53	Identification of novel WFS1 mutations in Italian children with Wolfram syndrome. Human Mutation, 2001, 17, 348-349.	2.5	51
54	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

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55	Respiratory Needs in Patients with Type 1 Spinal Muscular Atrophy TreatedÂwith Nusinersen. Journal of Pediatrics, 2020, 219, 223-228.e4.	1.8	51
56	G8363A Mutation in the Mitochondrial DNA Transfer Ribonucleic Acid Lys Gene: Another Cause of Leigh Syndrome. Journal of Child Neurology, 2000, 15, 759-761.	1.4	50
57	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
58	The Genetic Landscape of Dystrophin Mutations in Italy: A Nationwide Study. Frontiers in Genetics, 2020, 11, 131.	2.3	49
59	Genetic and physiologic analysis of the role of uncoupling protein 3 in human energy homeostasis. Diabetes, 1999, 48, 1890-1895.	0.6	48
60	The Hammersmith functional score correlates with the SMN2 copy number: A multicentric study. Neuromuscular Disorders, 2007, 17, 400-403.	0.6	47
61	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
62	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
63	Genotype-phenotype correlations in a large series of patients with muscle type CPT II deficiency. Neurological Research, 2011, 33, 24-32.	1.3	46
64	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
65	Upper limb function in Duchenne muscular dystrophy: 24 month longitudinal data. PLoS ONE, 2018, 13, e0199223.	2.5	45
66	Clinical features and new molecular findings in muscle phosphofructokinase deficiency (GSD type VII). Neuromuscular Disorders, 2012, 22, 325-330.	0.6	44
67	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
68	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
69	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
70	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
71	A Splice Junction Mutation in the Î±MGene of Phosphorylase Kinase in a Patient with Myopathy. Biochemical and Biophysical Research Communications, 1998, 249, 648-651.	2.1	41
72	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

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73	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
74	Phenotypic characterization of hypomyelination and congenital cataract. Annals of Neurology, 2007, 62, 121-127.	5.3	39
75	POMGnT1 Mutations in Congenital Muscular Dystrophy. Archives of Neurology, 2006, 63, 1491.	4.5	38
76	POMT1 and POMT2 mutations in CMD patients: A multicentric Italian study. Neuromuscular Disorders, 2008, 18, 565-571.	0.6	38
77	Mitochondrial Encephalomyopathy Due to a Novel Mutation inACAD9. JAMA Neurology, 2013, 70, 1177.	9.0	38
78	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
79	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
80	Genetic and Early Clinical Manifestations of Females Heterozygous for Duchenne/Becker Muscular Dystrophy. Pediatric Neurology, 2016, 55, 58-63.	2.1	37
81	â€œMitochondrial neuropathiesâ€: A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.6	37
82	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	3.7	36
83	Respiratory Trajectories in Type 2 and 3 Spinal Muscular Atrophy in the iSMAC Cohort Study. Neurology, 2021, 96, e587-e599.	1.1	36
84	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
85	Genotypeâ€“phenotype correlations in recessive titinopathies. Genetics in Medicine, 2020, 22, 2029-2040.	2.4	35
86	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
87	Glycogen storage diseases of muscle. Current Opinion in Neurology, 1998, 11, 477-484.	3.6	35
88	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
89	Myoclonus in mitochondrial disorders. Movement Disorders, 2014, 29, 722-728.	3.9	33
90	Phenotypic Heterogeneity in Two Unrelated Danon Patients Associated with the SameLAMPâ€2Gene Mutation. Neuropediatrics, 2005, 36, 309-313.	0.6	32

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91	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
92	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
93	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	3.6	32
94	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
95	Peroxisomal acylâ€CoAâ€oxidase deficiency: Two new cases. American Journal of Medical Genetics, Part A, 2008, 146A, 1676-1681.	1.2	31
96	TRPV4 mutations in children with congenital distal spinal muscular atrophy. Neurogenetics, 2012, 13, 195-203.	1.4	31
97	Respiratory and cardiac function in congenital muscular dystrophies with alpha dystroglycan deficiency. Neuromuscular Disorders, 2012, 22, 685-689.	0.6	31
98	The mitochondrial DNA C3303T mutation can cause cardiomyopathy and/or skeletal myopathy. Journal of Pediatrics, 1999, 135, 197-202.	1.8	30
99	Zidovudine ameliorates pathology in the mouse model of Duchenne muscular dystrophy via P2RX7 purinoceptor antagonism. Acta Neuropathologica Communications, 2018, 6, 27.	5.2	30
100	Null mutations and lethal congenital form of glycogen storage disease type IV. Biochemical and Biophysical Research Communications, 2007, 361, 445-450.	2.1	29
101	Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. Neuromuscular Disorders, 2021, 31, 596-602.	0.6	29
102	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
103	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
104	Very-long-chain acyl-coenzyme A dehydrogenase deficiency in a child with recurrent myoglobinuria. Neuromuscular Disorders, 1998, 8, 3-6.	0.6	27
105	Manifesting heterozygotes in a Japanese family with a novel mutation in the muscle-specific phosphoglycerate mutase (PGAM-M) gene. Neuromuscular Disorders, 1999, 9, 399-402.	0.6	27
106	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
107	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
108	Clinical and electrophysiological features of epilepsy in Italian patients with CLN8 mutations. Epilepsy and Behavior, 2007, 10, 187-191.	1.7	26

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109	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
110	A novel missense mutation in the glycogen branching enzyme gene in a child with myopathy and hepatopathy. Neuromuscular Disorders, 1999, 9, 403-407.	0.6	25
111	Molecular characterization of McArdleâ€™s disease in two large Finnish families. Journal of the Neurological Sciences, 1999, 165, 121-125.	0.6	25
112	HyperCKemia as the Only Sign of McArdle's Disease in a Child. Journal of Child Neurology, 2000, 15, 137-138.	1.4	25
113	Hypomyelination and Congenital Cataract: Neuroimaging Features of a Novel Inherited White Matter Disorder. American Journal of Neuroradiology, 2008, 29, 301-305.	2.4	25
114	Dopamine-agonist responsive Parkinsonism in a patient with the SANDO syndrome caused by POLG mutation. BMC Medical Genetics, 2013, 14, 105.	2.1	25
115	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. Neuromuscular Disorders, 2016, 26, 292-299.	0.6	25
116	Sudden infant death syndrome (SIDS) in a family with myosphosphorylase deficiency. Neuromuscular Disorders, 1997, 7, 81-83.	0.6	24
117	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.6	24
118	An observational study of functional abilities in infants, children, and adults with type 1 SMA. Neurology, 2018, 91, e696-e703.	1.1	24
119	Italian recommendations for diagnosis and management of congenital myasthenic syndromes. Neurological Sciences, 2019, 40, 457-468.	1.9	24
120	The 6 Minute Walk Test and Performance of Upper Limb in Ambulant Duchenne Muscular Dystrophy Boys. PLOS Currents, 2014, 6, .	1.4	24
121	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
122	The ubiquitin ligase tripartite-motif-protein 32 is induced in Duchenne muscular dystrophy. Laboratory Investigation, 2016, 96, 862-871.	3.7	23
123	A nonsense mutation in the myophosphorylase gene in a Japanese family with McArdle's disease. Neuromuscular Disorders, 1999, 9, 34-37.	0.6	22
124	Detection of early nocturnal hypoventilation in neuromuscular disorders. Journal of International Medical Research, 2018, 46, 1153-1161.	1.0	22
125	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
126	Primary adrenal insufficiency in a child with a mitochondrial DNA deletion. Journal of Inherited Metabolic Disease, 1998, 21, 155-161.	3.6	21

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127	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
128	Allogeneic bone marrow transplantation for Pearson's syndrome. Bone Marrow Transplantation, 2007, 39, 563-565.	2.4	21
129	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
130	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
131	Aquaporin-4 expression is severely reduced in human sarcoglycanopathies and dysferlinopathies. Cell Cycle, 2008, 7, 2199-2207.	2.6	20
132	Vaccination recommendations for patients with neuromuscular disease. Vaccine, 2014, 32, 5893-5900.	3.8	20
133	A Novel Missense Mutation (W797R) in the Myophosphorylase Gene in Spanish Patients With McArdle Disease. Archives of Neurology, 2000, 57, 217.	4.5	19
134	Unusual presentation of phosphoglycerate mutase deficiency due to two different mutations in PGAM-M gene. Neuromuscular Disorders, 2009, 19, 776-778.	0.6	19
135	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
136	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
137	Maternally Inherited Mitochondrial Cardiomyopathy Associated with a C-to-T Transition at Nucleotide 3303 of Mitochondrial DNA in the tRNALeu(UUR) Gene. Pediatric and Developmental Pathology, 1999, 2, 78-85.	1.0	17
138	The spectrum of GNE mutations: allelic heterogeneity for a common phenotype. Neurological Sciences, 2010, 31, 377-380.	1.9	17
139	Cavitating Leukoencephalopathy in a Child Carrying the MitochondrialA8344GMutation: Fig 1.. American Journal of Neuroradiology, 2010, 31, E78-E79.	2.4	17
140	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
141	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
142	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
143	Polymorphic Variants in the Human Mitochondrial CytochromebGene. Molecular Genetics and Metabolism, 1999, 67, 49-52.	1.1	16
144	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

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145	Forearm Semi-ischemic Exercise Test in Pediatric Patients. Journal of Child Neurology, 1998, 13, 288-290.	1.4	15
146	Novel Mutation in the CPT II Gene in a Child With Periodic Febrile Myalgia and Myoglobinuria. Journal of Child Neurology, 2000, 15, 390-393.	1.4	15
147	Susceptibility-Weighted Angiography of Intracranial Blood Products and Calcifications Compared to Gradient Echo Sequence. Neuroradiology Journal, 2013, 26, 493-500.	1.2	15
148	Paternal germline mosaicism in collagen VI related myopathies. European Journal of Paediatric Neurology, 2015, 19, 533-536.	1.6	15
149	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
150	Muscle MRI in neutral lipid storage disease (NLSD). Journal of Neurology, 2017, 264, 1334-1342.	3.6	15
151	A new mutation in the myophosphorylase gene (Asn684Tyr) in a Spanish patient with McArdle's disease. Neuromuscular Disorders, 1999, 9, 171-173.	0.6	14
152	Neuromuscular Disorders of Glycogen Metabolism. Current Neurology and Neuroscience Reports, 2013, 13, 333.	4.2	14
153	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
154	Congenital myopathies. Current Neurology and Neuroscience Reports, 2004, 4, 68-73.	4.2	13
155	The NDUFB11 gene is not a modifier in Leber hereditary optic neuropathy. Biochemical and Biophysical Research Communications, 2007, 355, 181-187.	2.1	13
156	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
157	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
158	Spinal muscular atrophy: state of the art and new therapeutic strategies. Neurological Sciences, 2022, 43, 615-624.	1.9	13
159	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
160	Unclassified polysaccharidosis of the heart and skeletal muscle in siblings. Molecular Genetics and Metabolism, 2008, 95, 52-58.	1.1	12
161	Expanding the histopathological spectrum of CFL2â€related myopathies. Clinical Genetics, 2018, 93, 1234-1239.	2.0	11
162	eATP/P2X7R Axis: An Orchestrated Pathway Triggering Inflammasome Activation in Muscle Diseases. International Journal of Molecular Sciences, 2020, 21, 5963.	4.1	11

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163	Clinical and genetic spectrum of a large cohort of patients with Î´-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606.	7.6	11
164	P2X7 Receptor Antagonist Reduces Fibrosis and Inflammation in a Mouse Model of Alpha-Sarcoglycan Muscular Dystrophy. Pharmaceuticals, 2022, 15, 89.	3.8	11
165	Neuromuscular Disorders in Zebrafish: State of the Art and Future Perspectives. NeuroMolecular Medicine, 2013, 15, 405-419.	3.4	10
166	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
167	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
168	A new mutation in the regulatory domain of the myophosphorylase gene affecting protein dimer contact. , 1999, 22, 1136-1138.		9
169	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
170	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
171	Chemokine receptor CCR7 is expressed in muscle fibers in juvenile dermatomyositis. Biochemical and Biophysical Research Communications, 2005, 333, 540-543.	2.1	8
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