

Ichizo Nishino

List of Publications by Year in descending order

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552
papers

33,869
citations

10070

75
h-index

5739

167
g-index

619
all docs

619
docs citations

619
times ranked

44213
citing authors

#	ARTICLE	IF	CITATIONS
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). <i>Autophagy</i> , 2016, 12, 1-222.	4.3	4,701
2	Guidelines for the use and interpretation of assays for monitoring autophagy. <i>Autophagy</i> , 2012, 8, 445-544.	4.3	3,122
3	Guidelines for the use and interpretation of assays for monitoring autophagy in higher eukaryotes. <i>Autophagy</i> , 2008, 4, 151-175.	4.3	2,064
4	Primary LAMP-2 deficiency causes X-linked vacuolar cardiomyopathy and myopathy (Danon disease). <i>Nature</i> , 2000, 406, 906-910.	13.7	865
5	Adiponectin and AdipoR1 regulate PGC-1 β and mitochondria by Ca ²⁺ and AMPK/SIRT1. <i>Nature</i> , 2010, 464, 1313-1319.	13.7	859
6	Thymidine Phosphorylase Gene Mutations in MNGIE, a Human Mitochondrial Disorder. <i>Science</i> , 1999, 283, 689-692.	6.0	827
7	Post-translational disruption of dystroglycan α ligand interactions in congenital muscular dystrophies. <i>Nature</i> , 2002, 418, 417-421.	13.7	747
8	Fatal infantile cardioencephalomyopathy with COX deficiency and mutations in SCO2, a COX assembly gene. <i>Nature Genetics</i> , 1999, 23, 333-337.	9.4	556
9	Skeletal Muscle FOXO1 (FKHR) Transgenic Mice Have Less Skeletal Muscle Mass, Down-regulated Type I (Slow Twitch/Red Muscle) Fiber Genes, and Impaired Glycemic Control. <i>Journal of Biological Chemistry</i> , 2004, 279, 41114-41123.	1.6	488
10	Distinctive patterns of microRNA expression in primary muscular disorders. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 17016-17021.	3.3	458
11	Human PTRF mutations cause secondary deficiency of caveolins resulting in muscular dystrophy with generalized lipodystrophy. <i>Journal of Clinical Investigation</i> , 2009, 119, 2623-2633.	3.9	350
12	Mitochondrial neurogastrointestinal encephalomyopathy: An autosomal recessive disorder due to thymidine phosphorylase mutations. <i>Annals of Neurology</i> , 2000, 47, 792-800.	2.8	324
13	Misregulated alternative splicing of BIN1 is associated with T tubule alterations and muscle weakness in myotonic dystrophy. <i>Nature Medicine</i> , 2011, 17, 720-725.	15.2	299
14	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. <i>Nature Genetics</i> , 2019, 51, 1222-1232.	9.4	265
15	LARGE can functionally bypass α -dystroglycan glycosylation defects in distinct congenital muscular dystrophies. <i>Nature Medicine</i> , 2004, 10, 696-703.	15.2	253
16	Identification and characterization of PDGFR α ⁺ mesenchymal progenitors in human skeletal muscle. <i>Cell Death and Disease</i> , 2014, 5, e1186-e1186.	2.7	241
17	Central core disease is due to RYR1 mutations in more than 90% of patients. <i>Brain</i> , 2006, 129, 1470-1480.	3.7	233
18	Clinical features and prognosis in anti-SRP and anti-HMGCR necrotising myopathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 1038-1044.	0.9	229

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19	The sarcolemmal proteins dysferlin and caveolin-3 interact in skeletal muscle. <i>Human Molecular Genetics</i> , 2001, 10, 1761-1766.	1.4	214
20	Distal myopathy with rimmed vacuoles is allelic to hereditary inclusion body myopathy. <i>Neurology</i> , 2002, 59, 1689-1693.	1.5	209
21	Altered Thymidine Metabolism Due to Defects of Thymidine Phosphorylase. <i>Journal of Biological Chemistry</i> , 2002, 277, 4128-4133.	1.6	209
22	Molecular features of the CAG repeats and clinical manifestation of Machado-Joseph disease. <i>Human Molecular Genetics</i> , 1995, 4, 807-812.	1.4	191
23	Autophagic degradation of nuclear components in mammalian cells. <i>Autophagy</i> , 2009, 5, 795-804.	4.3	189
24	Mutations in DNMT3B Modify Epigenetic Repression of the D4Z4 Repeat and the Penetrance of Facioscapulohumeral Dystrophy. <i>American Journal of Human Genetics</i> , 2016, 98, 1020-1029.	2.6	188
25	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy. <i>American Journal of Human Genetics</i> , 2013, 93, 6-18.	2.6	186
26	Allogeneic stem cell transplantation corrects biochemical derangements in MNGIE. <i>Neurology</i> , 2006, 67, 1458-1460.	1.5	172
27	Structural and Functional Mutations of the Perlecan Gene Cause Schwartz-Jampel Syndrome, with Myotonic Myopathy and Chondrodysplasia. <i>American Journal of Human Genetics</i> , 2002, 70, 1368-1375.	2.6	168
28	Prophylactic treatment with sialic acid metabolites precludes the development of the myopathic phenotype in the DMRV-hIBM mouse model. <i>Nature Medicine</i> , 2009, 15, 690-695.	15.2	167
29	<i>TMEM43</i> mutations in emeryâ€dreifuss muscular dystrophyâ€related myopathy. <i>Annals of Neurology</i> , 2011, 69, 1005-1013.	2.8	164
30	Inflammatory myopathy with anti-signal recognition particle antibodies: case series of 100 patients. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 61.	1.2	156
31	Localization of a gene for myoclonus-dystonia to chromosome 7q21-q31. <i>Annals of Neurology</i> , 1999, 46, 794-798.	2.8	154
32	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. <i>Journal of Clinical Investigation</i> , 2014, 124, 4693-4708.	3.9	153
33	Actin mutations are one cause of congenital fibre type disproportion. <i>Annals of Neurology</i> , 2004, 56, 689-694.	2.8	149
34	239th ENMC International Workshop: Classification of dermatomyositis, Amsterdam, the Netherlands, 14â€16 December 2018. <i>Neuromuscular Disorders</i> , 2020, 30, 70-92.	0.3	148
35	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. <i>American Journal of Human Genetics</i> , 2013, 93, 1108-1117.	2.6	147
36	Fukutin gene mutations cause dilated cardiomyopathy with minimal muscle weakness. <i>Annals of Neurology</i> , 2006, 60, 597-602.	2.8	140

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37	Reduction of UDP-N-acetylglucosamine 2-Epimerase/N-Acetylmannosamine Kinase Activity and Sialylation in Distal Myopathy with Rimmed Vacuoles. <i>Journal of Biological Chemistry</i> , 2004, 279, 11402-11407.	1.6	139
38	CXorf6 is a causative gene for hypospadias. <i>Nature Genetics</i> , 2006, 38, 1369-1371.	9.4	136
39	A Gne knockout mouse expressing human GNE D176V mutation develops features similar to distal myopathy with rimmed vacuoles or hereditary inclusion body myopathy. <i>Human Molecular Genetics</i> , 2007, 16, 2669-2682.	1.4	136
40	Lysosomal myopathies: An excessive build-up in autophagosomes is too much to handle. <i>Neuromuscular Disorders</i> , 2008, 18, 521-529.	0.3	136
41	Worldwide distribution and broader clinical spectrum of muscle-eye-brain disease. <i>Human Molecular Genetics</i> , 2003, 12, 527-534.	1.4	133
42	Malignant Hyperthermia in Japan. <i>Anesthesiology</i> , 2006, 104, 1146-1154.	1.3	132
43	Dominant mutations in ORAI1 cause tubular aggregate myopathy with hypocalcemia via constitutive activation of store-operated Ca ²⁺ channels. <i>Human Molecular Genetics</i> , 2015, 24, 637-648.	1.4	132
44	GNE myopathy: current update and future therapy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 385-392.	0.9	131
45	Unbalanced deoxynucleotide pools cause mitochondrial DNA instability in thymidine phosphorylase-deficient mice. <i>Human Molecular Genetics</i> , 2009, 18, 714-722.	1.4	123
46	VMA21 deficiency prevents vacuolar ATPase assembly and causes autophagic vacuolar myopathy. <i>Acta Neuropathologica</i> , 2013, 125, 439-457.	3.9	119
47	ETFDH mutations, CoQ10 levels, and respiratory chain activities in patients with riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. <i>Neuromuscular Disorders</i> , 2009, 19, 212-216.	0.3	118
48	Sarcoplasmic MxA expression. <i>Neurology</i> , 2017, 88, 493-500.	1.5	118
49	Skeletal Muscle Involvement in Antisynthetase Syndrome. <i>JAMA Neurology</i> , 2017, 74, 992.	4.5	117
50	Deficiency of Î±-Dystroglycan in Muscleâ€“Eyeâ€“Brain Disease. <i>Biochemical and Biophysical Research Communications</i> , 2002, 291, 1283-1286.	1.0	115
51	A Congenital Muscular Dystrophy with Mitochondrial Structural Abnormalities Caused by Defective De Novo Phosphatidylcholine Biosynthesis. <i>American Journal of Human Genetics</i> , 2011, 88, 845-851.	2.6	115
52	Defects in amphiphysin 2 (BIN1) and triads in several forms of centronuclear myopathies. <i>Acta Neuropathologica</i> , 2011, 121, 253-266.	3.9	113
53	Danon disease: a phenotypic expression of LAMP-2 deficiency. <i>Acta Neuropathologica</i> , 2015, 129, 391-398.	3.9	112
54	A Gne knockout mouse expressing human V572L mutation develops features similar to distal myopathy with rimmed vacuoles or hereditary inclusion body myopathy. <i>Human Molecular Genetics</i> , 2007, 16, 115-128.	1.4	111

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55	Autophagic Vacuolar Myopathy. <i>Seminars in Pediatric Neurology</i> , 2006, 13, 90-95.	1.0	110
56	Autophagic vacuolar myopathies. <i>Current Neurology and Neuroscience Reports</i> , 2003, 3, 64-69.	2.0	108
57	Definitive Diagnosis of Mitochondrial Neurogastrointestinal Encephalomyopathy by Biochemical Assays. <i>Clinical Chemistry</i> , 2004, 50, 120-124.	1.5	107
58	Unifying Nomenclature for the Isoforms of the Lysosomal Membrane Protein LAMP-2. <i>Traffic</i> , 2005, 6, 1058-1061.	1.3	107
59	Defects of intergenomic communication: autosomal disorders that cause multiple deletions and depletion of mitochondrial DNA. <i>Seminars in Cell and Developmental Biology</i> , 2001, 12, 417-427.	2.3	105
60	Overexpression of Peroxisome Proliferator-Activated Receptor δ Co-Activator-1 α Leads to Muscle Atrophy with Depletion of ATP. <i>American Journal of Pathology</i> , 2006, 169, 1129-1139.	1.9	96
61	Defects of Vps15 in skeletal muscles lead to autophagic vacuolar myopathy and lysosomal disease. <i>EMBO Molecular Medicine</i> , 2013, 5, 870-890.	3.3	96
62	Cell-Surface Protein Profiling Identifies Distinctive Markers of Progenitor Cells in Human Skeletal Muscle. <i>Stem Cell Reports</i> , 2016, 7, 263-278.	2.3	95
63	Centronuclear myopathy in mice lacking a novel muscle-specific protein kinase transcriptionally regulated by MEF2. <i>Genes and Development</i> , 2005, 19, 2066-2077.	2.7	93
64	Myoclonus epilepsy associated with ragged-red fibers: A G-to-A mutation at nucleotide pair 8363 in mitochondrial tRNA ^{Lys} in two families. <i>Muscle and Nerve</i> , 1997, 20, 271-278.	1.0	92
65	Mitochondrial Neurogastrointestinal Encephalomyopathy Syndrome Maps to Chromosome 22q13.32-qter. <i>American Journal of Human Genetics</i> , 1998, 63, 526-533.	2.6	91
66	Emerin-Lacking Mice Show Minimal Motor and Cardiac Dysfunctions with Nuclear-Associated Vacuoles. <i>American Journal of Pathology</i> , 2006, 168, 907-917.	1.9	91
67	Safety and efficacy of intravenous bimagrumab in inclusion body myositis (RESILIENT): a randomised, double-blind, placebo-controlled phase 2b trial. <i>Lancet Neurology</i> , The, 2019, 18, 834-844.	4.9	91
68	Primary collagen VI deficiency is the second most common congenital muscular dystrophy in Japan. <i>Neurology</i> , 2007, 69, 1035-1042.	1.5	90
69	Filamin C plays an essential role in the maintenance of the structural integrity of cardiac and skeletal muscles, revealed by the medaka mutant zacro. <i>Developmental Biology</i> , 2012, 361, 79-89.	0.9	90
70	Expansion of GGC Repeat in GIPC1 Is Associated with Oculopharyngodistal Myopathy. <i>American Journal of Human Genetics</i> , 2020, 106, 793-804.	2.6	90
71	N ϵ -WASP is required for Amphiphysin α -BIN α -dependent nuclear positioning and triad organization in skeletal muscle and is involved in the pathophysiology of centronuclear myopathy. <i>EMBO Molecular Medicine</i> , 2014, 6, 1455-1475.	3.3	87
72	Mechanisms of Genomic Instabilities Underlying Two Common Fragile-Site-Associated Loci, PARK2 and DMD, in Germ Cell and Cancer Cell Lines. <i>American Journal of Human Genetics</i> , 2010, 87, 75-89.	2.6	85

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73	Increased Expression of Wild-Type or a Centronuclear Myopathy Mutant of Dynamin 2 in Skeletal Muscle of Adult Mice Leads to Structural Defects and Muscle Weakness. <i>American Journal of Pathology</i> , 2011, 178, 2224-2235.	1.9	84
74	Homozygous nonsense variant in <i>LRIF1</i> associated with facioscapulohumeral muscular dystrophy. <i>Neurology</i> , 2020, 94, e2441-e2447.	1.5	84
75	Myopathy Associated With Antibodies to Signal Recognition Particle. <i>Archives of Neurology</i> , 2012, 69, 728-32.	4.9	82
76	Autophagic Vacuoles with Sarcolemmal Features Delineate Danon Disease and Related Myopathies. <i>Journal of Neuropathology and Experimental Neurology</i> , 2005, 64, 513-522.	0.9	81
77	Disrupted prenatal RNA processing and myogenesis in congenital myotonic dystrophy. <i>Genes and Development</i> , 2017, 31, 1122-1133.	2.7	80
78	Genetic diagnosis of Duchenne/Becker muscular dystrophy using next-generation sequencing: validation analysis of DMD mutations. <i>Journal of Human Genetics</i> , 2016, 61, 483-489.	1.1	79
79	CGG expansion in NOTCH2NLC is associated with oculopharyngodistal myopathy with neurological manifestations. <i>Acta Neuropathologica Communications</i> , 2020, 8, 204.	2.4	76
80	Genotype and phenotype analyses in 136 patients with single large-scale mitochondrial DNA deletions. <i>Journal of Human Genetics</i> , 2008, 53, 598-606.	1.1	75
81	Muscle choline kinase beta defect causes mitochondrial dysfunction and increased mitophagy. <i>Human Molecular Genetics</i> , 2011, 20, 3841-3851.	1.4	75
82	Clinical and genetic analysis of lipid storage myopathies. <i>Muscle and Nerve</i> , 2009, 39, 333-342.	1.0	74
83	Statins and Myotoxic Effects Associated With Anti-3-Hydroxy-3-Methylglutaryl-Coenzyme A Reductase Autoantibodies. <i>Medicine (United States)</i> , 2015, 94, e416.	0.4	74
84	Inflammatory myopathy associated with PD-1 inhibitors. <i>Journal of Autoimmunity</i> , 2019, 100, 105-113.	3.0	73
85	Mutations in the satellite cell gene MEGF10 cause a recessive congenital myopathy with minicores. <i>Neurogenetics</i> , 2012, 13, 115-124.	0.7	68
86	Where are we moving in the classification of idiopathic inflammatory myopathies?. <i>Current Opinion in Neurology</i> , 2020, 33, 590-603.	1.8	68
87	Inflammatory changes in infantile-onset LMNA-associated myopathy. <i>Neuromuscular Disorders</i> , 2011, 21, 563-568.	0.3	67
88	Autophagy in Lysosomal Myopathies. <i>Brain Pathology</i> , 2012, 22, 82-88.	2.1	67
89	Analysis of mouse models of cytochrome c oxidase deficiency owing to mutations in <i>Sco2</i> . <i>Human Molecular Genetics</i> , 2010, 19, 170-180.	1.4	66
90	Biallelic Mutations in MYPN , Encoding Myopalladin, Are Associated with Childhood-Onset, Slowly Progressive Nemaline Myopathy. <i>American Journal of Human Genetics</i> , 2017, 100, 169-178.	2.6	66

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91	Transgenic Monkey Model of the Polyglutamine Diseases Recapitulating Progressive Neurological Symptoms. <i>ENeuro</i> , 2017, 4, ENEURO.0250-16.2017.	0.9	66
92	A new congenital muscular dystrophy with mitochondrial structural abnormalities. , 1998, 21, 40-47.		64
93	MNGIE: from nuclear DNA to mitochondrial DNA. <i>Neuromuscular Disorders</i> , 2001, 11, 7-10.	0.3	64
94	Distal myopathy with rimmed vacuoles and hereditary inclusion body myopathy. <i>Current Neurology and Neuroscience Reports</i> , 2005, 5, 61-65.	2.0	64
95	Congenital neuromuscular disease with uniform type 1 fiber and RYR1 mutation. <i>Neurology</i> , 2008, 70, 114-122.	1.5	64
96	Pediatric necrotizing myopathy associated with anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase antibodies. <i>Rheumatology</i> , 2017, 56, 287-293.	0.9	64
97	GNE myopathy: New name and new mutation nomenclature. <i>Neuromuscular Disorders</i> , 2014, 24, 387-389.	0.3	61
98	Hepatitis C virus infection in inclusion body myositis. <i>Neurology</i> , 2016, 86, 211-217.	1.5	61
99	Classification of idiopathic inflammatory myopathies: pathology perspectives. <i>Current Opinion in Neurology</i> , 2019, 32, 704-714.	1.8	61
100	Lipid Storage Myopathy. <i>Current Neurology and Neuroscience Reports</i> , 2011, 11, 97-103.	2.0	60
101	Perifascicular necrosis in anti-synthetase syndrome beyond anti-Jo-1. <i>Brain</i> , 2016, 139, e50-e50.	3.7	60
102	Protein and gene analyses of dysferlinopathy in a large group of Japanese muscular dystrophy patients. <i>Journal of the Neurological Sciences</i> , 2003, 211, 23-28.	0.3	58
103	Dysferlin Interacts with Affixin (β 2-Parvin) at the Sarcolemma. <i>Journal of Neuropathology and Experimental Neurology</i> , 2005, 64, 334-340.	0.9	57
104	Nuclear changes in skeletal muscle extend to satellite cells in autosomal dominant Emery-Dreifuss muscular dystrophy/limb-girdle muscular dystrophy 1B. <i>Neuromuscular Disorders</i> , 2009, 19, 29-36.	0.3	57
105	Characteristics of Japanese Duchenne and Becker muscular dystrophy patients in a novel Japanese national registry of muscular dystrophy (Remudy). <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 60.	1.2	56
106	LAMP2-deficient human B cells exhibit altered MHC class II presentation of exogenous antigens. <i>Immunology</i> , 2010, 131, 318-330.	2.0	55
107	The cathepsin L gene is a direct target of FOXO1 in skeletal muscle. <i>Biochemical Journal</i> , 2010, 427, 171-178.	1.7	55
108	Characterization of the Asian myopathy patients with <i>VCP</i> mutations. <i>European Journal of Neurology</i> , 2012, 19, 501-509.	1.7	55

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109	Rigid spine syndrome caused by a novel mutation in four-and-a-half LIM domain 1 gene (FHL1). <i>Neuromuscular Disorders</i> , 2008, 18, 959-961.	0.3	54
110	Establishment of an Improved Mouse Model for Infantile Neuroaxonal Dystrophy That Shows Early Disease Onset and Bears a Point Mutation in Pla2g6. <i>American Journal of Pathology</i> , 2009, 175, 2257-2263.	1.9	54
111	The First Molecular Evidence That Autophagy Relates Rimmed Vacuole Formation in Chloroquine Myopathy. <i>Journal of Biochemistry</i> , 2002, 131, 647-651.	0.9	53
112	Clinical and histological findings associated with autoantibodies detected by RNA immunoprecipitation in inflammatory myopathies. <i>Journal of Neuroimmunology</i> , 2014, 274, 202-208.	1.1	53
113	Ullrich congenital muscular dystrophy: clinicopathological features, natural history and pathomechanism(s). <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 280-287.	0.9	53
114	DNAJB6 myopathy in an Asian cohort and cytoplasmic/nuclear inclusions. <i>Neuromuscular Disorders</i> , 2013, 23, 269-276.	0.3	52
115	Sialyllactose ameliorates myopathic phenotypes in symptomatic GNE myopathy model mice. <i>Brain</i> , 2014, 137, 2670-2679.	3.7	52
116	Integrated Diagnosis Project for Inflammatory Myopathies: An association between autoantibodies and muscle pathology. <i>Autoimmunity Reviews</i> , 2017, 16, 693-700.	2.5	52
117	Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy. <i>American Journal of Human Genetics</i> , 2016, 99, 950-961.	2.6	51
118	Targeted massively parallel sequencing and histological assessment of skeletal muscles for the molecular diagnosis of inherited muscle disorders. <i>Journal of Medical Genetics</i> , 2017, 54, 104-110.	1.5	51
119	Distal lipid storage myopathy due to PNPLA2 mutation. <i>Neuromuscular Disorders</i> , 2008, 18, 671-674.	0.3	50
120	Mutation profile of the GNE gene in Japanese patients with distal myopathy with rimmed vacuoles (GNE) Tj ETQq0 0.0 rgBT /Overlock 100	0.9	50
121	Autophagy in a Mouse Model of Distal Myopathy with Rimmed Vacuoles or Hereditary Inclusion Body Myopathy. <i>Autophagy</i> , 2007, 3, 396-398.	4.3	49
122	Increase in number of sporadic inclusion body myositis (sIBM) in Japan. <i>Journal of Neurology</i> , 2012, 259, 554-556.	1.8	49
123	A patient-derived iPSC model revealed oxidative stress increases facioscapulohumeral muscular dystrophy-causative <i>DUX4</i> . <i>Human Molecular Genetics</i> , 2018, 27, 4024-4035.	1.4	49
124	Heterozygous UDP-GlcNAc 2-epimerase and N-acetylmannosamine kinase domain mutations in the GNE gene result in a less severe GNE myopathy phenotype compared to homozygous N-acetylmannosamine kinase domain mutations. <i>Journal of the Neurological Sciences</i> , 2012, 318, 100-105.	0.3	47
125	Congenital muscular dystrophy with fatty liver and infantile-onset cataract caused by TRAPPC11 mutations: broadening of the phenotype. <i>Skeletal Muscle</i> , 2015, 5, 29.	1.9	47
126	HLA-DRB1 alleles in immune-mediated necrotizing myopathy. <i>Neurology</i> , 2016, 87, 1954-1955.	1.5	47

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127	Localization of Calpain 3 in Human Skeletal Muscle and Its Alteration in Limb-Girdle Muscular Dystrophy 2A Muscle. <i>Journal of Biochemistry</i> , 2003, 133, 659-664.	0.9	46
128	NOVEL <i>FHL1</i> MUTATIONS IN FATAL AND BENIGN REDUCING BODY MYOPATHY. <i>Neurology</i> , 2009, 72, 375-376.	1.5	46
129	Defects in autophagosome-lysosome fusion underlie Vici syndrome, a neurodevelopmental disorder with multisystem involvement. <i>Scientific Reports</i> , 2017, 7, 3552.	1.6	46
130	Hypoparathyroidism and insulin-dependent diabetes mellitus in a patient with Kearns-Sayre syndrome harbouring a mitochondrial DNA deletion. <i>Clinical Endocrinology</i> , 1996, 45, 637-641.	1.2	43
131	Comprehensive analysis for genetic diagnosis of Dystrophinopathies in Japan. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 149.	1.2	43
132	Subcellular Localization of Fukutin and Fukutin-Related Protein in Muscle Cells. <i>Journal of Biochemistry</i> , 2004, 135, 709-712.	0.9	42
133	Congenital muscular dystrophy with glycosylation defects of α -dystroglycan in Japan. <i>Neuromuscular Disorders</i> , 2005, 15, 342-348.	0.3	42
134	Novel Lamp-2 gene mutation and successful treatment with heart transplantation in a large family with Danon disease. <i>Muscle and Nerve</i> , 2006, 33, 393-397.	1.0	42
135	Infantile facioscapulohumeral muscular dystrophy revisited: Expansion of clinical phenotypes in patients with a very short EcoRI fragment. <i>Neuromuscular Disorders</i> , 2013, 23, 298-305.	0.3	42
136	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. <i>Journal of Human Genetics</i> , 2014, 59, 581-583.	1.1	42
137	Sialic acid deficiency is associated with oxidative stress leading to muscle atrophy and weakness in GNE myopathy. <i>Human Molecular Genetics</i> , 2017, 26, 3081-3093.	1.4	42
138	The 3260 mutation in mitochondrial DNA can cause mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)., 1996, 19, 1603-1604.		41
139	Characterization of MTM1 mutations in 31 Japanese families with myotubular myopathy, including a patient carrying 240kb deletion in Xq28 without male hypogonadism. <i>Neuromuscular Disorders</i> , 2005, 15, 245-252.	0.3	41
140	Mutation analysis of the GNE gene in distal myopathy with rimmed vacuoles (DMRV) patients in Thailand. <i>Muscle and Nerve</i> , 2006, 34, 775-778.	1.0	41
141	Aberrant neuromuscular junctions and delayed terminal muscle fiber maturation in α -dystroglycanopathies. <i>Human Molecular Genetics</i> , 2006, 15, 1279-1289.	1.4	41
142	A Novel Mutation in the Mitochondrial tRNAThr Gene Associated with a Mitochondrial Encephalomyopathy. <i>Biochemical and Biophysical Research Communications</i> , 1996, 225, 180-185.	1.0	40
143	Proteolysis of α -dystroglycan in muscular diseases. <i>Neuromuscular Disorders</i> , 2005, 15, 336-341.	0.3	40
144	Peracetylated N-Acetylmannosamine, a Synthetic Sugar Molecule, Efficiently Rescues Muscle Phenotype and Biochemical Defects in Mouse Model of Sialic Acid-deficient Myopathy. <i>Journal of Biological Chemistry</i> , 2012, 287, 2689-2705.	1.6	40

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145	Aberrant Myokine Signaling in Congenital Myotonic Dystrophy. <i>Cell Reports</i> , 2017, 21, 1240-1252.	2.9	40
146	Megaconial congenital muscular dystrophy due to loss-of-function mutations in choline kinase β . <i>Current Opinion in Neurology</i> , 2013, 26, 536-543.	1.8	39
147	Association of Dermatomyositis Sine Dermatitis With Anti-“Nuclear Matrix Protein 2 Autoantibodies. <i>JAMA Neurology</i> , 2020, 77, 872.	4.5	39
148	Limb-Girdle Muscular Dystrophy Due to Emerin Gene Mutations. <i>Archives of Neurology</i> , 2007, 64, 1038.	4.9	38
149	Reversible infantile respiratory chain deficiency: A clinical and molecular study. <i>Annals of Neurology</i> , 2010, 68, 845-854.	2.8	38
150	Identification of Variants in the 4q35 Gene <i>FAT1</i> in Patients with a Facioscapulohumeral Dystrophy-Like Phenotype. <i>Human Mutation</i> , 2015, 36, 443-453.	1.1	38
151	Nationwide patient registry for GNE myopathy in Japan. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 150.	1.2	37
152	<i>DAG1</i> mutations associated with asymptomatic hyperCKemia and hypoglycosylation of β -dystroglycan. <i>Neurology</i> , 2015, 84, 273-279.	1.5	37
153	Dysferlin expression in tubular aggregates: their possible relationship to endoplasmic reticulum stress. <i>Acta Neuropathologica</i> , 2003, 105, 603-609.	3.9	36
154	Muscle weakness correlates with muscle atrophy and precedes the development of inclusion body or rimmed vacuoles in the mouse model of DMRV/hIBM. <i>Physiological Genomics</i> , 2008, 35, 106-115.	1.0	36
155	Positive association between <i>STAT4</i> polymorphisms and polymyositis/dermatomyositis in a Japanese population. <i>Annals of the Rheumatic Diseases</i> , 2012, 71, 1646-1650.	0.5	36
156	Prednisolone improves walking in Japanese Duchenne muscular dystrophy patients. <i>Journal of Neurology</i> , 2013, 260, 3023-3029.	1.8	36
157	Severe nemaline myopathy caused by mutations of the stop codon of the skeletal muscle alpha actin gene (<i>ACTA1</i>). <i>Neuromuscular Disorders</i> , 2006, 16, 541-547.	0.3	35
158	Alternative splicing of myomesin 1 gene is aberrantly regulated in myotonic dystrophy type 1. <i>Genes To Cells</i> , 2011, 16, 961-972.	0.5	35
159	Muscle glycogen storage disease 0 presenting recurrent syncope with weakness and myalgia. <i>Neuromuscular Disorders</i> , 2012, 22, 162-165.	0.3	35
160	Phenotypic stratification and genotype-phenotype correlation in a heterogeneous, international cohort of GNE myopathy patients: First report from the GNE myopathy Disease Monitoring Program, registry portion. <i>Neuromuscular Disorders</i> , 2018, 28, 158-168.	0.3	35
161	Molecular pathomechanism of distal myopathy with rimmed vacuoles. <i>Acta Myologica</i> , 2005, 24, 80-3.	1.5	35
162	Mutations of calpain 3 gene in patients with sporadic limb-girdle muscular dystrophy in Japan. <i>Journal of the Neurological Sciences</i> , 1999, 171, 31-37.	0.3	34

#	ARTICLE	IF	CITATIONS
163	Very low penetrance in 85 Japanese families with facioscapulohumeral muscular dystrophy 1A. <i>Journal of Medical Genetics</i> , 2004, 41, 12e-12.	1.5	34
164	Dysferlin mutation analysis in a group of Italian patients with limb-girdle muscular dystrophy and Miyoshi myopathy. <i>European Journal of Neurology</i> , 2004, 11, 657-661.	1.7	34
165	Humanin expression in skeletal muscles of patients with chronic progressive external ophthalmoplegia. <i>Journal of Human Genetics</i> , 2006, 51, 555-558.	1.1	34
166	GNE myopathy: A prospective natural history study of disease progression. <i>Neuromuscular Disorders</i> , 2014, 24, 380-386.	0.3	34
167	Isolated inclusion body myopathy caused by a multisystem proteinopathyâ€linked <i>hnRNPA1</i> mutation. <i>Neurology: Genetics</i> , 2015, 1, e23.	0.9	34
168	Dermatomyositis. <i>Neurology</i> , 2022, 98, .	1.5	34
169	Characterization of Danon disease in a male patient and his affected mother. <i>Neuromuscular Disorders</i> , 2003, 13, 708-711.	0.3	33
170	Dysferlinopathy associated with rigid spine syndrome. <i>Neuropathology</i> , 2004, 24, 341-346.	0.7	33
171	Sarcolemmopathy: Muscular Dystrophies with Cell Membrane Defects. <i>Brain Pathology</i> , 2001, 11, 218-230.	2.1	33
172	Muscular dystrophy with large mitochondria associated with mutations in the <i>CHKB</i> gene in three British patients: Extending the clinical and pathological phenotype. <i>Neuromuscular Disorders</i> , 2013, 23, 549-556.	0.3	33
173	Molecular pathomechanisms and cell-type-specific disease phenotypes of MELAS caused by mutant mitochondrial tRNA ^{Trp} . <i>Acta Neuropathologica Communications</i> , 2015, 3, 52.	2.4	33
174	GNE myopathy in India. <i>Neurology India</i> , 2013, 61, 371.	0.2	32
175	<i>DNM1L</i> -related encephalopathy in infancy with Leigh syndromeâ€like phenotype and suppressionâ€burst. <i>Clinical Genetics</i> , 2016, 90, 472-474.	1.0	32
176	Anti-nuclear matrix protein 2 antibody-positive inflammatory myopathies represent extensive myositis without dermatomyositis-specific rash. <i>Rheumatology</i> , 2022, 61, 1222-1227.	0.9	32
177	A new congenital form of X-linked autophagic vacuolar myopathy. <i>Neurology</i> , 2005, 65, 1132-1134.	1.5	31
178	Allelic heterogeneity of GNE gene mutation in two Tunisian families with autosomal recessive inclusion body myopathy. <i>Neuromuscular Disorders</i> , 2005, 15, 361-363.	0.3	31
179	Asymptomatic hyperCKemia in a case of Danon disease due to a missense mutation in <i>Lamp-2</i> gene. <i>Neuromuscular Disorders</i> , 2005, 15, 409-411.	0.3	31
180	Expression profiling of muscles from Fukuyama-type congenital muscular dystrophy and laminin- α 2 deficient congenital muscular dystrophy; is congenital muscular dystrophy a primary fibrotic disease?. <i>Biochemical and Biophysical Research Communications</i> , 2006, 342, 489-502.	1.0	31

#	ARTICLE	IF	CITATIONS
181	Unfolded protein response and aggresome formation in hereditary reducing-body myopathy. <i>Muscle and Nerve</i> , 2007, 35, 322-326.	1.0	31
182	Homozygous female Becker muscular dystrophy. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1052-1055.	0.7	31
183	Effects of enzyme replacement therapy on five patients with advanced late-onset glycogen storage disease type II: a 2-year follow-up study. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 301-310.	1.7	31
184	RNA sequencing solved the most common but unrecognized NEB pathogenic variant in Japanese nemaline myopathy. <i>Genetics in Medicine</i> , 2019, 21, 1629-1638.	1.1	31
185	Clinical practice guidance for juvenile dermatomyositis (JDM) 2018-Update. <i>Modern Rheumatology</i> , 2020, 30, 411-423.	0.9	31
186	Alternative splicing of clathrin heavy chain contributes to the switch from coated pits to plaques. <i>Journal of Cell Biology</i> , 2020, 219, .	2.3	31
187	Recessive <i>RYR1</i> mutations in a patient with severe congenital nemaline myopathy with ophthalmoplegia identified through massively parallel sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 772-778.	0.7	30
188	Clinicopathologic Features of Oculopharyngodistal Myopathy With <i>LRP12</i> CGG Repeat Expansions Compared With Other Oculopharyngodistal Myopathy Subtypes. <i>JAMA Neurology</i> , 2021, 78, 853.	4.5	30
189	Muscular dystrophies. <i>Current Opinion in Neurology</i> , 2002, 15, 539-544.	1.8	29
190	Sub-cellular localisation of fukutin related protein in different cell lines and in the muscle of patients with MDC1C and LGMD2I. <i>Neuromuscular Disorders</i> , 2005, 15, 836-843.	0.3	29
191	Asymptomatic Sporadic Dysferlinopathy Presenting with Elevation of Serum Creatine Kinase. Typical Distribution of Muscle Involvement Shown by MRI but not by CT. <i>Internal Medicine</i> , 2008, 47, 305-307.	0.3	29
192	Alternative splicing of PDLIM3/ALP, for β -actinin-associated LIM protein 3, is aberrant in persons with myotonic dystrophy. <i>Biochemical and Biophysical Research Communications</i> , 2011, 409, 64-69.	1.0	29
193	Confirmation of the efficacy of vitamin B ₆ supplementation for McArdle disease by follow-up muscle biopsy. <i>Muscle and Nerve</i> , 2012, 45, 436-440.	1.0	29
194	Congenital generalized lipodystrophy type 4 with muscular dystrophy: Clinical and pathological manifestations in early childhood. <i>Neuromuscular Disorders</i> , 2013, 23, 441-444.	0.3	29
195	<i>IBA57</i> mutations abrogate iron-sulfur cluster assembly leading to cavitating leukoencephalopathy. <i>Neurology: Genetics</i> , 2017, 3, e184.	0.9	29
196	Reduced cell anchorage may cause sarcolemma-specific collagen VI deficiency in Ullrich disease. <i>Neurology</i> , 2007, 69, 1043-1049.	1.5	28
197	Specific phosphorylation of Ser458 of A-type lamins in <i>LMNA</i> -associated myopathy patients. <i>Journal of Cell Science</i> , 2010, 123, 3893-3900.	1.2	28
198	Anti-Signal Recognition Particle Myopathy in the First Decade of Life. <i>Pediatric Neurology</i> , 2011, 45, 114-116.	1.0	28

#	ARTICLE	IF	CITATIONS
199	Acid phosphatase-positive globular inclusions is a good diagnostic marker for two patients with adult-onset Pompe disease lacking disease specific pathology. <i>Neuromuscular Disorders</i> , 2012, 22, 389-393.	0.3	28
200	Clinical utility gene card for: Centronuclear and myotubular myopathies. <i>European Journal of Human Genetics</i> , 2012, 20, 1101-1101.	1.4	28
201	Psychiatric and neurodevelopmental aspects of Becker muscular dystrophy. <i>Neuromuscular Disorders</i> , 2019, 29, 930-939.	0.3	28
202	Biallelic variants in <i>LIG3</i> cause a novel mitochondrial neurogastrointestinal encephalomyopathy. <i>Brain</i> , 2021, 144, 1451-1466.	3.7	28
203	LAMP-2 Positive Vacuolar Myopathy with Dilated Cardiomyopathy. <i>Internal Medicine</i> , 2007, 46, 757-760.	0.3	27
204	Rapidly progressive scoliosis and respiratory deterioration in Ullrich congenital muscular dystrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 982-988.	0.9	27
205	A Nationwide Survey on Danon Disease in Japan. <i>International Journal of Molecular Sciences</i> , 2018, 19, 3507.	1.8	27
206	Novel LAMP-2 Mutation in a Family With Danon Disease Presenting With Hypertrophic Cardiomyopathy. <i>Circulation Journal</i> , 2009, 73, 376-380.	0.7	26
207	A novel mutation in the LMNA gene causes congenital muscular dystrophy with dropped head and brain involvement. <i>Neuromuscular Disorders</i> , 2012, 22, 149-151.	0.3	26
208	Reduced Dnmt3a increases Gdf5 expression with suppressed satellite cell differentiation and impaired skeletal muscle regeneration. <i>FASEB Journal</i> , 2018, 32, 1452-1467.	0.2	26
209	Pembrolizumab-induced Ocular Myasthenia Gravis with Anti-titin Antibody and Necrotizing Myopathy. <i>Internal Medicine</i> , 2019, 58, 1635-1638.	0.3	26
210	A novel RyR1-selective inhibitor prevents and rescues sudden death in mouse models of malignant hyperthermia and heat stroke. <i>Nature Communications</i> , 2021, 12, 4293.	5.8	26
211	Germline mosaicism of a novel mutation in lysosome-associated membrane protein-2 deficiency (Danon) Tj ETQq1 1,0.784314.rgBT / 0.2.8 25	2.8	25
212	Efficacy and Safety of Bimagrumab in Sporadic Inclusion Body Myositis. <i>Neurology</i> , 2021, 96, e1595-e1607.	1.5	25
213	cDNA microarray analysis of individual Duchenne muscular dystrophy patients. <i>Human Molecular Genetics</i> , 2003, 12, 595-600.	1.4	25
214	Thymidine Phosphorylase Deficiency Causes MNGIE: An Autosomal Recessive Mitochondrial Disorder. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2004, 23, 1217-1225.	0.4	24
215	Functional analysis of SERCA1b, a highly expressed SERCA1 variant in myotonic dystrophy type 1 muscle. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2015, 1852, 2042-2047.	1.8	24
216	Early onset of cardiomyopathy and intellectual disability in a girl with Danon disease associated with a de novo novel mutation of the LAMP2 gene. <i>Neuropathology</i> , 2016, 36, 561-565.	0.7	24

#	ARTICLE	IF	CITATIONS
217	Cardiopulmonary dysfunction in patients with limb-girdle muscular dystrophy 2A. <i>Muscle and Nerve</i> , 2017, 55, 465-469.	1.0	24
218	Small-Vessel Vasculopathy Due to Aberrant Autophagy in LAMP-2 Deficiency. <i>Scientific Reports</i> , 2018, 8, 3326.	1.6	24
219	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. <i>American Journal of Human Genetics</i> , 2020, 107, 1078-1095.	2.6	24
220	Pathologic Features of Anti-Mi-2 Dermatomyositis. <i>Neurology</i> , 2021, 96, e448-e459.	1.5	24
221	The C2A domain in dysferlin is important for association with MG53 (TRIM72). <i>PLOS Currents</i> , 2012, 4, e5035add8caff4.	1.4	24
222	Characterization of lobulated fibers in limb girdle muscular dystrophy type 2A by gene expression profiling. <i>Neuroscience Research</i> , 2007, 57, 513-521.	1.0	23
223	Recent advances in distal myopathy with rimmed vacuoles (DMRV) or hIBM: treatment perspectives. <i>Current Opinion in Neurology</i> , 2008, 21, 596-600.	1.8	23
224	Defective Myotilin Homodimerization Caused by a Novel Mutation in MYOT Exon 9 in the First Japanese Limb Girdle Muscular Dystrophy 1A Patient. <i>Journal of Neuropathology and Experimental Neurology</i> , 2009, 68, 701-707.	0.9	23
225	Respiratory dysfunction in patients severely affected by GNE myopathy (distal myopathy with rimmed) <i>Tj ETQq1 1 0.784314 rgBT /Ov</i>	0.3	23
226	Ultrasound-enhanced delivery of Morpholino with Bubble liposomes ameliorates the myotonia of myotonic dystrophy model mice. <i>Scientific Reports</i> , 2013, 3, 2242.	1.6	23
227	<i>ABLIM1</i> splicing is abnormal in skeletal muscle of patients with <i>DM</i> 1 and regulated by <i>MBNL</i> , <i>CELF</i> and <i>PTBP</i> 1. <i>Genes To Cells</i> , 2015, 20, 121-134.	0.5	23
228	Necklace cytoplasmic bodies in hereditary myopathy with early respiratory failure. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 483-489.	0.9	23
229	Cardiac autophagic vacuolation in severe X-linked myopathy with excessive autophagy. <i>Neuromuscular Disorders</i> , 2017, 27, 185-187.	0.3	23
230	Zmynd17 controls muscle mitochondrial quality and whole-body metabolism. <i>FASEB Journal</i> , 2018, 32, 5012-5025.	0.2	23
231	Exon skipping induced by nonsense/frameshift mutations in DMD gene results in Becker muscular dystrophy. <i>Human Genetics</i> , 2020, 139, 247-255.	1.8	23
232	<i>COVID-19</i> -associated myositis may be dermatomyositis. <i>Muscle and Nerve</i> , 2021, 63, E9-E10.	1.0	23
233	Successful treatment of a novel type I interferonopathy due to a de novo <i>PSMB9</i> gene mutation with a Janus kinase inhibitor. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 639-644.	1.5	23
234	Mitochondrial abnormalities in selenium-deficient myopathy. , 1998, 21, 637-639.		22

#	ARTICLE	IF	CITATIONS
235	Rapid and accurate diagnosis of facioscapulohumeral muscular dystrophy. <i>Neuromuscular Disorders</i> , 2006, 16, 256-261.	0.3	22
236	Phospholipid synthetic defect and mitophagy in muscle disease. <i>Autophagy</i> , 2011, 7, 1559-1561.	4.3	22
237	Respiratory and cardiac function in Japanese patients with dysferlinopathy. <i>Muscle and Nerve</i> , 2016, 53, 394-401.	1.0	22
238	<i>GNE</i> genotype explains 20% of phenotypic variability in <i>GNE</i> myopathy. <i>Neurology: Genetics</i> , 2019, 5, e308.	0.9	22
239	Vascular Involvement in a Patient with Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, and Stroke-Like Episodes. <i>American Journal of the Medical Sciences</i> , 2005, 329, 265-266.	0.4	21
240	Chapter 19 Monitoring Autophagy in Muscle Diseases. <i>Methods in Enzymology</i> , 2009, 453, 379-396.	0.4	21
241	Different effects of novel mtDNA G3242A and G3244A base changes adjacent to a common A3243G mutation in patients with mitochondrial disorders. <i>Mitochondrion</i> , 2009, 9, 115-122.	1.6	21
242	Manumycin A corrects aberrant splicing of <i>Clcn1</i> in myotonic dystrophy type 1 (DM1) mice. <i>Scientific Reports</i> , 2013, 3, 2142.	1.6	21
243	COX6A2 variants cause a muscle-specific cytochrome c oxidase deficiency. <i>Annals of Neurology</i> , 2019, 86, 193-202.	2.8	21
244	Mutant BIN1-Dynamin 2 complexes dysregulate membrane remodeling in the pathogenesis of centronuclear myopathy. <i>Journal of Biological Chemistry</i> , 2021, 296, 100077.	1.6	21
245	Comprehensive target capture/next-generation sequencing as a second-tier diagnostic approach for congenital muscular dystrophy in Taiwan. <i>PLoS ONE</i> , 2017, 12, e0170517.	1.1	21
246	MTM1 gene mutations in Japanese patients with the severe infantile form of myotubular myopathy. <i>Neuromuscular Disorders</i> , 1998, 8, 453-458.	0.3	20
247	Newly recognized exons induced by a splicing abnormality from an intronic mutation of the dystrophin gene resulting in Duchenne muscular dystrophy. , 1999, 13, 170-170.		20
248	FSHD-like patients without 4q35 deletion. <i>Journal of the Neurological Sciences</i> , 2004, 219, 89-93.	0.3	20
249	Gene expression analyses in X-linked myotubular myopathy. <i>Neurology</i> , 2005, 65, 732-737.	1.5	20
250	Expression of MBNL and CELF mRNA transcripts in muscles with myotonic dystrophy. <i>Neuromuscular Disorders</i> , 2007, 17, 306-312.	0.3	20
251	Mutational Analysis of Fukutin Gene in Dilated Cardiomyopathy and Hypertrophic Cardiomyopathy. <i>Circulation Journal</i> , 2009, 73, 158-161.	0.7	20
252	A 13-year-old girl with proximal weakness and hypertrophic cardiomyopathy with danon disease. <i>Muscle and Nerve</i> , 2010, 41, 879-882.	1.0	20

#	ARTICLE	IF	CITATIONS
253	A mutation in a rare type of intron in a sodium-channel gene results in aberrant splicing and causes myotonia. <i>Human Mutation</i> , 2011, 32, 773-782.	1.1	20
254	Rimmed Vacuoles in Becker Muscular Dystrophy Have Similar Features with Inclusion Myopathies. <i>PLoS ONE</i> , 2012, 7, e52002.	1.1	20
255	A nationwide survey on Marinesco-Sjögren syndrome in Japan. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 58.	1.2	20
256	Milder forms of muscular dystrophy associated with <i>POMGNT2</i> mutations. <i>Neurology: Genetics</i> , 2015, 1, e33.	0.9	20
257	Mutations in the J domain of DNAJB6 cause dominant distal myopathy. <i>Neuromuscular Disorders</i> , 2020, 30, 38-46.	0.3	20
258	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. <i>Nature Communications</i> , 2022, 13, 2306.	5.8	20
259	Danon disease: A novel Lamp-2 gene mutation in a family with four affected members. <i>Neuromuscular Disorders</i> , 2008, 18, 167-174.	0.3	19
260	Pyruvate Improved Insulin Secretion Status in a Mitochondrial Diabetes Mellitus Patient. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 1924-1926.	1.8	19
261	Muscle Weakness and Fibrosis Due to Cell Autonomous and Non-cell Autonomous Events in Collagen VI Deficient Congenital Muscular Dystrophy. <i>EBioMedicine</i> , 2017, 15, 193-202.	2.7	19
262	ADSSL1 myopathy is the most common nemaline myopathy in Japan with variable clinical features. <i>Neurology</i> , 2020, 95, e1500-e1511.	1.5	19
263	Mitochondrial Encephalomyopathy with Elderly Onset of Stroke-Like Episodes. <i>Internal Medicine</i> , 1996, 35, 991-995.	0.3	18
264	Affixin activates Rac1 via β PIX in C2C12 myoblast. <i>FEBS Letters</i> , 2008, 582, 1189-1196.	1.3	18
265	Central nervous system and muscle involvement in an adolescent patient with riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. <i>Brain and Development</i> , 2010, 32, 669-672.	0.6	18
266	Elevated urinary β 2 microglobulin in the first identified Japanese family afflicted by X-linked myopathy with excessive autophagy. <i>Neuromuscular Disorders</i> , 2013, 23, 911-916.	0.3	18
267	Limb-girdle muscular dystrophy type 2I is not rare in Taiwan. <i>Neuromuscular Disorders</i> , 2013, 23, 675-681.	0.3	18
268	Congenital fiber type disproportion myopathy caused by LMNA mutations. <i>Journal of the Neurological Sciences</i> , 2014, 340, 94-98.	0.3	18
269	Congenital autophagic vacuolar myopathy is allelic to X-linked myopathy with excessive autophagy. <i>Neurology</i> , 2015, 84, 1714-1716.	1.5	18
270	A novel mutation in the proteolytic domain of LONP1 causes atypical CODAS syndrome. <i>Journal of Human Genetics</i> , 2017, 62, 653-655.	1.1	18

#	ARTICLE	IF	CITATIONS
271	GNE myopathy in Chinese population: hotspot and novel mutations. <i>Journal of Human Genetics</i> , 2019, 64, 11-16.	1.1	18
272	Two Japanese LGMDR25 patients with a biallelic recurrent nonsense variant of BVES. <i>Neuromuscular Disorders</i> , 2020, 30, 674-679.	0.3	18
273	Sporadic inclusion body myositis in Japanese is associated with the MHC ancestral haplotype 52.1. <i>Neuromuscular Disorders</i> , 2006, 16, 311-315.	0.3	17
274	Csk-homologous kinase interacts with SHPS-1 and enhances neurite outgrowth of PC12 cells. <i>Journal of Neurochemistry</i> , 2008, 105, 101-112.	2.1	17
275	Clinicopathological features of centronuclear myopathy in Japanese populations harboring mutations in dynamin 2. <i>Clinical Neurology and Neurosurgery</i> , 2012, 114, 678-683.	0.6	17
276	Allele-specific Gene Silencing of Mutant mRNA Restores Cellular Function in Ullrich Congenital Muscular Dystrophy Fibroblasts. <i>Molecular Therapy - Nucleic Acids</i> , 2014, 3, e171.	2.3	17
277	Deep morphological analysis of muscle biopsies from type III glycogenesis (GSDIII), debranching enzyme deficiency, revealed stereotyped vacuolar myopathy and autophagy impairment. <i>Acta Neuropathologica Communications</i> , 2019, 7, 167.	2.4	17
278	A novel FKRP gene mutation in a Taiwanese patient with limb-girdle muscular dystrophy 2I. <i>Brain and Development</i> , 2007, 29, 234-238.	0.6	16
279	Diminished binding of mutated collagen VI to the extracellular matrix surrounding myocytes. <i>Muscle and Nerve</i> , 2008, 38, 1192-1195.	1.0	16
280	Eponym. <i>European Journal of Pediatrics</i> , 2011, 170, 1365-1367.	1.3	16
281	Cell stress molecules in the skeletal muscle of GNE myopathy. <i>BMC Neurology</i> , 2013, 13, 24.	0.8	16
282	Lysosomal storage and advanced senescence in the brain of LAMP-2-deficient Danon disease. <i>Acta Neuropathologica</i> , 2013, 125, 459-461.	3.9	16
283	Association between a C8orf13â€“BLK Polymorphism and Polymyositis/Dermatomyositis in the Japanese Population: An Additive Effect with STAT4 on Disease Susceptibility. <i>PLoS ONE</i> , 2014, 9, e90019.	1.1	16
284	Divergent clinical outcomes of alpha-glucosidase enzyme replacement therapy in two siblings with infantile-onset Pompe disease treated in the symptomatic or pre-symptomatic state. <i>Molecular Genetics and Metabolism Reports</i> , 2016, 9, 98-105.	0.4	16
285	Novel <i>TTN</i> mutations and muscle imaging characteristics in congenital titinopathy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1311-1318.	1.7	16
286	Atypical muscle pathology and a survey of cis-mutations in deaf patients harboring a 1555 A-to-G point mutation in the mitochondrial ribosomal RNA gene. <i>Neuromuscular Disorders</i> , 2002, 12, 506-512.	0.3	15
287	Expression of Myoferlin in Skeletal Muscles of Patients with Dysferlinopathy. <i>Tohoku Journal of Experimental Medicine</i> , 2006, 209, 109-116.	0.5	15
288	A preclinical trial of sialic acid metabolites on distal myopathy with rimmed vacuoles/ hereditary inclusion body myopathy, a sugar-deficient myopathy: a review. <i>Therapeutic Advances in Neurological Disorders</i> , 2010, 3, 127-135.	1.5	15

#	ARTICLE	IF	CITATIONS
289	Deep sequencing detects very-low-grade somatic mosaicism in the unaffected mother of siblings with nemaline myopathy. <i>Neuromuscular Disorders</i> , 2014, 24, 642-647.	0.3	15
290	TBCD may be a causal gene in progressive neurodegenerative encephalopathy with atypical infantile spinal muscular atrophy. <i>Journal of Human Genetics</i> , 2017, 62, 473-480.	1.1	15
291	Missing genetic variations in GNE myopathy: rearrangement hotspots encompassing 5'UTR and founder allele. <i>Journal of Human Genetics</i> , 2017, 62, 159-166.	1.1	15
292	A form of muscular dystrophy associated with pathogenic variants in JAG2. <i>American Journal of Human Genetics</i> , 2021, 108, 840-856.	2.6	15
293	Inflammatory features in sporadic late-onset nemaline myopathy are independent from monoclonal gammopathy. <i>Brain Pathology</i> , 2021, 31, e12962.	2.1	15
294	Two novel CAV3 gene mutations in Japanese families. <i>Neuromuscular Disorders</i> , 2004, 14, 810-814.	0.3	14
295	Cys669?Cys713 disulfide bridge formation is a key to dystroglycan cleavage and subunit association. <i>Genes To Cells</i> , 2007, 12, 75-88.	0.5	14
296	A novel POMT2 mutation causes mild congenital muscular dystrophy with normal brain MRI. <i>Brain and Development</i> , 2009, 31, 465-468.	0.6	14
297	Barth syndrome diagnosed in the subclinical stage of heart failure based on the presence of lipid storage myopathy and isolated noncompaction of the ventricular myocardium. <i>European Journal of Pediatrics</i> , 2011, 170, 1481-1484.	1.3	14
298	Rippling is not always electrically silent in rippling muscle disease. <i>Muscle and Nerve</i> , 2011, 43, 601-605.	1.0	14
299	Fatal hepatic hemorrhage by peliosis hepatis in X-linked myotubular myopathy: A case report. <i>Neuromuscular Disorders</i> , 2013, 23, 917-921.	0.3	14
300	Dietary Phosphorus Overload Aggravates the Phenotype of the Dystrophin-Deficient mdx Mouse. <i>American Journal of Pathology</i> , 2014, 184, 3094-3104.	1.9	14
301	Japanese multiple epidermal growth factor 10 (MEGF10) myopathy with novel mutations: A phenotype-genotype correlation. <i>Neuromuscular Disorders</i> , 2016, 26, 604-609.	0.3	14
302	Concurrent positive anti-3-hydroxy-3-methylglutaryl-coenzyme a reductase antibody with reducing body myopathy: Possible double trouble. <i>Neuromuscular Disorders</i> , 2019, 29, 543-548.	0.3	14
303	Anti-RNP antibodies delineate a subgroup of myositis: A systematic retrospective study on 46 patients. <i>Autoimmunity Reviews</i> , 2020, 19, 102465.	2.5	14
304	Obstruction-related dysphagia in inclusion body myositis: Cricopharyngeal bar on videofluoroscopy indicates risk of aspiration. <i>Journal of the Neurological Sciences</i> , 2020, 413, 116764.	0.3	14
305	Intranuclear inclusions in skin biopsies are not limited to neuronal intranuclear inclusion disease but can also be seen in oculopharyngodistal myopathy. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, .	1.8	14
306	Severe lactic acidosis and neonatal death in Pearson syndrome. <i>Journal of Inherited Metabolic Disease</i> , 1997, 20, 43-48.	1.7	13

#	ARTICLE	IF	CITATIONS
307	Functional analysis of ryanodine receptor type 1 p.R2508C mutation in exon 47. <i>Journal of Anesthesia</i> , 2009, 23, 341-346.	0.7	13
308	Novel DYSF mutations in Thai patients with distal myopathy. <i>Clinical Neurology and Neurosurgery</i> , 2009, 111, 613-618.	0.6	13
309	Congenital myotonic dystrophy can show congenital fiber type disproportion pathology. <i>Acta Neuropathologica</i> , 2010, 119, 481-486.	3.9	13
310	Myocerebrohepatopathy spectrum disorder due to POLG mutations: A clinicopathological report. <i>Brain and Development</i> , 2015, 37, 719-724.	0.6	13
311	Limb-girdle Muscular Dystrophy Type 2A with Mutation in CAPN3: The First Report in Taiwan. <i>Pediatrics and Neonatology</i> , 2015, 56, 62-65.	0.3	13
312	Adult-onset Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, and Stroke (MELAS)-like Encephalopathy Diagnosed Based on the Complete Sequencing of Mitochondrial DNA Extracted from Biopsied Muscle without any Myopathic Changes. <i>Internal Medicine</i> , 2017, 56, 95-99.	0.3	13
313	Quantification of lectin fluorescence in GNE myopathy muscle biopsies. <i>Muscle and Nerve</i> , 2018, 58, 286-292.	1.0	13
314	A novel compound heterozygous variant of ECHS1 identified in a Japanese patient with Leigh syndrome. <i>Human Genome Variation</i> , 2019, 6, 19.	0.4	13
315	An autopsy case of peliosis hepatis with X-linked myotubular myopathy. <i>Legal Medicine</i> , 2019, 38, 77-82.	0.6	13
316	Thigh muscle MRI findings in myopathy associated with anti-mitochondrial antibody. <i>Muscle and Nerve</i> , 2020, 61, 81-87.	1.0	13
317	Late-onset MELAS syndrome with mtDNA 14453G>A mutation masquerading as an acute encephalitis: a case report. <i>BMC Neurology</i> , 2020, 20, 247.	0.8	13
318	Clinical, pathological, imaging, and genetic characterization in a Taiwanese cohort with limb-girdle muscular dystrophy. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 160.	1.2	13
319	A review of core myopathy: central core disease, multiminicore disease, dusty core disease, and core-rod myopathy. <i>Neuromuscular Disorders</i> , 2021, 31, 968-977.	0.3	13
320	Dysferlinopathy Fibroblasts Are Defective in Plasma Membrane Repair. <i>PLOS Currents</i> , 2015, 7, .	1.4	13
321	TRAPPC11-related muscular dystrophy with hypoglycosylation of alpha-dystroglycan in skeletal muscle and brain. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, .	1.8	13
322	Thymidine Phosphorylase Gene Mutations Cause Mitochondrial Neurogastrointestinal Encephalomyopathy (MNGIE). <i>Internal Medicine</i> , 2006, 45, 1103-1103.	0.3	12
323	Familial reducing body myopathy. <i>Brain and Development</i> , 2007, 29, 112-116.	0.6	12
324	Myotonic dystrophy type 2 is rare in the Japanese population. <i>Journal of Human Genetics</i> , 2012, 57, 219-220.	1.1	12

#	ARTICLE	IF	CITATIONS
325	In Vivo Characterization of Mutant Myotilins. <i>American Journal of Pathology</i> , 2012, 180, 1570-1580.	1.9	12
326	Chronic Myopathy Associated With Anti- ϵ -Signal Recognition Particle Antibodies Can Be Misdiagnosed As Facioscapulohumeral Muscular Dystrophy. <i>Journal of Clinical Neuromuscular Disease</i> , 2016, 17, 197-206.	0.3	12
327	Clinical, muscle pathological, and genetic features of Japanese facioscapulohumeral muscular dystrophy 2 (FSHD2) patients with SMCHD1 mutations. <i>Neuromuscular Disorders</i> , 2016, 26, 300-308.	0.3	12
328	Impact of muscle biopsy on diagnosis and management of children with neuromuscular diseases: A 10-year retrospective critical review. <i>Journal of Pediatric Surgery</i> , 2018, 53, 489-492.	0.8	12
329	Social involvement issues in patients with Becker muscular dystrophy: A questionnaire survey of subjects from a patient registry. <i>Brain and Development</i> , 2018, 40, 268-277.	0.6	12
330	Sporadic late-onset nemaline myopathy with monoclonal gammopathy of undetermined significance (SLONM-MGUS): An alternative treatment using cyclophosphamide-thalidomide-dexamethasone (CTD) regimen. <i>Neuromuscular Disorders</i> , 2018, 28, 610-613.	0.3	12
331	Characteristics of Japanese Patients with Becker Muscular Dystrophy and Intermediate Muscular Dystrophy in a Japanese National Registry of Muscular Dystrophy (Remudy): Heterogeneity and Clinical Variation. <i>Journal of Neuromuscular Diseases</i> , 2018, 5, 193-203.	1.1	12
332	Anti-signal Recognition Particle Antibody-positive Necrotizing Myopathy with Secondary Cardiomyopathy: The First Myocardial Biopsy- and Multimodal Imaging-proven Case. <i>Internal Medicine</i> , 2019, 58, 3189-3194.	0.3	12
333	The updated retrospective questionnaire study of sporadic inclusion body myositis in Japan. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 155.	1.2	12
334	A primigravida with very-long-chain acyl-CoA dehydrogenase deficiency. <i>Muscle and Nerve</i> , 2014, 49, 295-296.	1.0	11
335	Plasma IP-10 level distinguishes inflammatory myopathy. <i>Neurology</i> , 2015, 85, 293-294.	1.5	11
336	Mitochondrial encephalomyopathy with 15915 mutation: Clinical report. <i>Pediatric Neurology</i> , 1997, 17, 161-164.	1.0	10
337	Mitochondrial neurogastrointestinal encephalomyopathy and thymidine metabolism: results and hypotheses. <i>Mitochondrion</i> , 2002, 2, 143-147.	1.6	10
338	Oculopharyngeal muscular dystrophy with PABPN1 mutation in a Chinese Malaysian woman. <i>Neuromuscular Disorders</i> , 2005, 15, 262-264.	0.3	10
339	A JAPANESE ADULT FORM OF CPT II DEFICIENCY ASSOCIATED WITH A HOMOZYGOUS F383Y MUTATION. <i>Neurology</i> , 2007, 69, 804-806.	1.5	10
340	Oculopharyngeal Muscular Dystrophy Associated with Dementia. <i>Internal Medicine</i> , 2011, 50, 2409-2412.	0.3	10
341	Novel Mutations in the Gene Encoding Acid .ALPHA.-1,4-glucosidase in a Patient with Late-onset Glycogen Storage Disease Type II (Pompe Disease) with Impaired Intelligence. <i>Internal Medicine</i> , 2011, 50, 2987-2991.	0.3	10
342	A pediatric patient with myopathy associated with antibodies to a signal recognition particle. <i>Brain and Development</i> , 2012, 34, 877-880.	0.6	10

#	ARTICLE	IF	CITATIONS
343	Adult-onset multiple acyl CoA dehydrogenation deficiency associated with an abnormal isoenzyme pattern of serum lactate dehydrogenase. <i>Neuromuscular Disorders</i> , 2012, 22, 159-161.	0.3	10
344	Property of Lysosomal Storage Disease Associated with Midbrain Pathology in the Central Nervous System of Lamp-2-deficient Mice. <i>American Journal of Pathology</i> , 2015, 185, 1713-1723.	1.9	10
345	Nemaline myopathy with KLHL40 mutation presenting as congenital totally locked-in state. <i>Brain and Development</i> , 2015, 37, 887-890.	0.6	10
346	Transient swelling in the globus pallidus and substantia nigra in childhood suggests SENDA/BPAN. <i>Neurology</i> , 2018, 90, 974-976.	1.5	10
347	Congenital myopathy with fiber-type disproportion accompanied by dilated cardiomyopathy in a patient with a novel p.G48A ACTA1 mutation. <i>Journal of the Neurological Sciences</i> , 2018, 393, 142-144.	0.3	10
348	A recurrent homozygous ACTN2 variant associated with core myopathy. <i>Acta Neuropathologica</i> , 2021, 142, 785-788.	3.9	10
349	A review of major causative genes in congenital myopathies. <i>Journal of Human Genetics</i> , 2023, 68, 215-225.	1.1	10
350	Detection of DNA Fragments Encompassing the Deletion Junction of Mitochondrial Genome. <i>Biochemical and Biophysical Research Communications</i> , 1996, 222, 215-219.	1.0	9
351	Nemaline (actin) myopathy with myofibrillar dysgenesis and abnormal ossification. <i>Neuromuscular Disorders</i> , 2009, 19, 485-488.	0.3	9
352	Isolated inflammatory myopathy with rimmed vacuoles presenting with dropped head. <i>Neuromuscular Disorders</i> , 2009, 19, 853-855.	0.3	9
353	Extramuscular manifestations in children with severe congenital myopathy due to ACTA1 gene mutations. <i>Neuromuscular Disorders</i> , 2011, 21, 489-493.	0.3	9
354	MELAS phenotype associated with m.3302A>G mutation in mitochondrial tRNA ^{Leu} (UUR) gene. <i>Brain and Development</i> , 2014, 36, 180-182.	0.6	9
355	Multicenter questionnaire survey for sporadic inclusion body myositis in Japan. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 146.	1.2	9
356	Duchenne muscular dystrophy in a female with compound heterozygous contiguous exon deletions. <i>Neuromuscular Disorders</i> , 2017, 27, 569-573.	0.3	9
357	GNE myopathy caused by a synonymous mutation leading to aberrant mRNA splicing. <i>Neuromuscular Disorders</i> , 2018, 28, 154-157.	0.3	9
358	Efficacy of Prednisolone in Generated Myotubes Derived From Fibroblasts of Duchenne Muscular Dystrophy Patients. <i>Frontiers in Pharmacology</i> , 2018, 9, 1402.	1.6	9
359	Secondary cardiac involvement in anti-SRP-antibody-positive myopathy: an 87-year-old woman with heart failure symptoms as the first clinical presentation. <i>BMC Neurology</i> , 2020, 20, 29.	0.8	9
360	Cricopharyngeal bar on videofluoroscopy: high specificity for inclusion body myositis. <i>Journal of Neurology</i> , 2021, 268, 1016-1024.	1.8	9

#	ARTICLE	IF	CITATIONS
361	Megaconial congenital muscular dystrophy secondary to novel CHKB mutations resemble atypical Rett syndrome. <i>Journal of Human Genetics</i> , 2021, 66, 813-823.	1.1	9
362	Muscle-specific calpain is phosphorylated in its unique insertion region for enrichment in a myofibril fraction. <i>Genes To Cells</i> , 2014, 19, 830-841.	0.5	8
363	A Japanese male with a novel ANO5 mutation with minimal muscle weakness and muscle pain till his late fifties. <i>Neuromuscular Disorders</i> , 2017, 27, 477-480.	0.3	8
364	A patient with slowly progressive adult-onset nemaline myopathy and novel compound heterozygous mutations in the nebulin gene. <i>Journal of the Neurological Sciences</i> , 2017, 373, 254-257.	0.3	8
365	Neurite growth could be impaired by <i>ETFDH</i> mutation but restored by mitochondrial cofactors. <i>Muscle and Nerve</i> , 2017, 56, 479-485.	1.0	8
366	Interpretation of acid phosphatase activity in creatine kinase elevation: A case of Becker muscular dystrophy. <i>Brain and Development</i> , 2018, 40, 837-840.	0.6	8
367	Characteristic findings of skeletal muscle MRI in caveolinopathies. <i>Neuromuscular Disorders</i> , 2018, 28, 857-862.	0.3	8
368	A novel AIFM1 missense mutation in a Japanese patient with ataxic sensory neuropathy and hearing impairment. <i>Journal of the Neurological Sciences</i> , 2020, 409, 116584.	0.3	8
369	The wide-ranging clinical and genetic features in Japanese families with valosin-containing protein proteinopathy. <i>Neurobiology of Aging</i> , 2021, 100, 120.e1-120.e6.	1.5	8
370	Assessment of thrombocytopenia, sleep apnea, and cardiac involvement in <i>GNE</i> myopathy patients. <i>Muscle and Nerve</i> , 2022, 65, 284-290.	1.0	8
371	A Novel Mutation of the <i>GAA</i> Gene in a Patient with Adult-onset Pompe Disease Lacking a Disease-specific Pathology. <i>Internal Medicine</i> , 2013, 52, 2461-2464.	0.3	7
372	Beevor's sign: a potential clinical marker for <i>GNE</i> myopathy. <i>European Journal of Neurology</i> , 2016, 23, e46-8.	1.7	7
373	Diagnosis of dermatomyositis: Autoantibody profile and muscle pathology. <i>Clinical and Experimental Neuroimmunology</i> , 2017, 8, 302-312.	0.5	7
374	Mitophagy in three cases of immune-mediated necrotizing myopathy associated with anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase autoantibodies: ultrastructural and immunohistochemical studies. <i>Neuromuscular Disorders</i> , 2018, 28, 283-288.	0.3	7
375	Genetic and functional analysis of the RYR1 mutation p.Thr84Met revealed a susceptibility to malignant hyperthermia. <i>Journal of Anesthesia</i> , 2018, 32, 174-181.	0.7	7
376	Phenotype of a limb-girdle congenital myasthenic syndrome patient carrying a GFPT1 mutation. <i>Brain and Development</i> , 2019, 41, 470-473.	0.6	7
377	Emery-Dreifuss muscular dystrophy-related myopathy with TMEM43 mutations. <i>Muscle and Nerve</i> , 2019, 59, E5-E7.	1.0	7
378	Anti-HMGCR myopathy following acute Epstein-Barr virus infection. <i>Muscle and Nerve</i> , 2020, 61, E5-E8.	1.0	7

#	ARTICLE	IF	CITATIONS
379	Pregnancy in GNE myopathy patients: a nationwide repository survey in Japan. Orphanet Journal of Rare Diseases, 2020, 15, 245.	1.2	7
380	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. PLoS ONE, 2020, 15, e0237890.	1.1	7
381	Clinical practice with steroid therapy for Duchenne muscular dystrophy: An expert survey in Asia and Oceania. Brain and Development, 2020, 42, 277-288.	0.6	7
382	A Japanese case of oculopharyngeal muscular dystrophy (OPMD) with PABPN1 c.35G>→A; p.Gly12Ala point mutation. BMC Neurology, 2021, 21, 265.	0.8	7
383	Extra-muscular manifestations in GNE myopathy patients: A nationwide repository questionnaire survey in Japan. Clinical Neurology and Neurosurgery, 2022, 212, 107057.	0.6	7
384	Mutation spectrum of primary lipid storage myopathies. Annals of Indian Academy of Neurology, 2022, 25, 106.	0.2	7
385	A case of congenital fiber-type disproportion syndrome presenting dilated cardiomyopathy with <i>ACTA1</i> mutation. Molecular Genetics & Genomic Medicine, 0, , .	0.6	7
386	Malignant hyperthermia susceptibility diagnosed with a family-specific ryanodine receptor gene type 1 mutation. Journal of Anesthesia, 2008, 22, 70-73.	0.7	6
387	Marked left ventricular hypertrophy in a patient with mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes. International Journal of Cardiology, 2008, 129, e77-e80.	0.8	6
388	Mitochondrial Encephalomyopathy Associated with Diabetes Mellitus, Cataract, and Corpus Callosum Atrophy. Internal Medicine, 2008, 47, 441-444.	0.3	6
389	Teaching Neuro <i>Images</i> : Hemiatrophy as a clinical presentation in facioscapulohumeral muscular dystrophy. Neurology, 2009, 73, e24.	1.5	6
390	Distal Myopathy in Multi-minicore Disease. Internal Medicine, 2009, 48, 1759-1762.	0.3	6
391	Novel Mutations of the <i>GNE</i> Gene in Distal Myopathy with Rimmed Vacuoles Presenting with Very Slow Progression. Case Reports in Neurology, 2012, 4, 120-125.	0.3	6
392	Unusual exocrine complication of pancreatitis in mitochondrial disease. Brain and Development, 2013, 35, 654-659.	0.6	6
393	Think worldwide: hereditary myopathy with early respiratory failure (HMERF) may not be rare. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 248-248.	0.9	6
394	A de novo mutation of the MYH7 gene in a large Chinese family with autosomal dominant myopathy. Human Genome Variation, 2015, 2, 15022.	0.4	6
395	Novel <i>TK2</i> mutations as a cause of delayed muscle maturation in mtDNA depletion syndrome. Neurology: Genetics, 2016, 2, e95.	0.9	6
396	Probable high prevalence of limb-girdle muscular dystrophy type 2D in Taiwan. Journal of the Neurological Sciences, 2016, 362, 304-308.	0.3	6

#	ARTICLE	IF	CITATIONS
397	A Homozygous <i>LAMA2</i> Mutation of c.818G>A Caused Partial Merosin Deficiency in a Japanese Patient. <i>Internal Medicine</i> , 2018, 57, 877-882.	0.3	6
398	Three novel recessive DYSF mutations identified in three patients with muscular dystrophy, limb-girdle, type 2B. <i>Journal of the Neurological Sciences</i> , 2018, 395, 169-171.	0.3	6
399	Two novel VCP missense variants identified in Japanese patients with multisystem proteinopathy. <i>Human Genome Variation</i> , 2018, 5, 9.	0.4	6
400	Chronic sarcoid myopathy mimicking sporadic inclusion body myositis. <i>Clinical Neurology and Neurosurgery</i> , 2019, 182, 84-86.	0.6	6
401	A novel deletion in the C-terminal region of HSPB8 in a family with rimmed vacuolar myopathy. <i>Journal of Human Genetics</i> , 2021, 66, 965-972.	1.1	6
402	Causative variant profile of collagen VI-related dystrophy in Japan. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 284.	1.2	6
403	Visualizing Muscle Sialic Acid Expression in the GNE207VTgGne ^{-/-} Cmah ^{-/-} Model of GNE Myopathy: A Comparison of Dietary and Gene Therapy Approaches. <i>Journal of Neuromuscular Diseases</i> , 2022, 9, 53-71.	1.1	6
404	Deep convolutional neural network-based algorithm for muscle biopsy diagnosis. <i>Laboratory Investigation</i> , 2022, 102, 220-226.	1.7	6
405	A case of congenital neuromuscular disease with uniform type 1 fibers. <i>Brain and Development</i> , 2006, 28, 202-205.	0.6	5
406	A unique case of limb-girdle muscular dystrophy type 2A carrying novel compound heterozygous mutations in the human CAPN3 gene. <i>European Journal of Neurology</i> , 2007, 14, 819-822.	1.7	5
407	Selective muscle involvement in a family affected by a second LIM domain mutation of fhl1: An imaging study using computed tomography. <i>Journal of the Neurological Sciences</i> , 2012, 318, 163-167.	0.3	5
408	Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency: A frequent condition in the southern Chinese population. <i>Neurology and Clinical Neuroscience</i> , 2013, 1, 163-167.	0.2	5
409	Late-onset Pompe disease after 4 years of enzyme replacement therapy: An autopsy case. <i>Neurology and Clinical Neuroscience</i> , 2014, 2, 7-9.	0.2	5
410	Muscle from a 20-week-old myotubular myopathy fetus is not myotubular. <i>Neuromuscular Disorders</i> , 2016, 26, 234-235.	0.3	5
411	Phase I clinical trial results of aceneuramic acid for GNE myopathy in Japan. <i>Translational Medicine Communications</i> , 2018, 3, .	0.5	5
412	A novel LMNA mutation identified in a Japanese patient with LMNA-associated congenital muscular dystrophy. <i>Human Genome Variation</i> , 2018, 5, 19.	0.4	5
413	Dropped Head in Sporadic Late-onset Nemaline Myopathy. <i>Internal Medicine</i> , 2019, 58, 1967-1968.	0.3	5
414	237th ENMC International Workshop: GNE myopathy – current and future research Hoofddorp, The Netherlands, 14–16 September 2018. <i>Neuromuscular Disorders</i> , 2019, 29, 401-410.	0.3	5

#	ARTICLE	IF	CITATIONS
415	Treatment experience of Taiwanese patients with anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase myopathy. <i>Kaohsiung Journal of Medical Sciences</i> , 2020, 36, 649-655.	0.8	5
416	Paramyotonia Congenita with Persistent Distal and Facial Muscle Weakness: A Case Report with Literature Review. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 193-201.	1.1	5
417	Antimitochondrial Antibody-associated Myopathy with Slowly Progressive Cardiac Dysfunction. <i>Internal Medicine</i> , 2021, 60, 1035-1041.	0.3	5
418	Myotubular/centronuclear myopathy and central core disease. <i>Neurology India</i> , 2008, 56, 325.	0.2	5
419	DA-Raf and the MEK inhibitor trametinib reverse skeletal myocyte differentiation inhibition or muscle atrophy caused by myostatin and GDF11 through the non-Smad Ras-ERK pathway. <i>Journal of Biochemistry</i> , 2022, 171, 109-122.	0.9	5
420	Brain perfusion abnormalities in a thinner and amphetamine abuser detected by I-123 IMP scintigraphy. <i>Annals of Nuclear Medicine</i> , 1992, 6, 273-275.	1.2	4
421	Muscle fiber immaturity and inactivity reduce myonecrosis in duchenne muscular dystrophy. <i>Annals of Neurology</i> , 1998, 44, 967-971.	2.8	4
422	Early onset distal muscular dystrophy with normal dysferlin expression. <i>Brain and Development</i> , 2005, 27, 589-591.	0.6	4
423	Left ventriculoplasty for dilated cardiomyopathy in Fukuyama-type muscular dystrophy. <i>European Journal of Cardio-thoracic Surgery</i> , 2011, 40, 514-6.	0.6	4
424	Heteroplasmic m.1624C>T mutation of the mitochondrial tRNA ^{Val} gene in a proband and his mother with repeated consciousness disturbances. <i>Mitochondrion</i> , 2012, 12, 617-622.	1.6	4
425	Juvenile autophagic vacuolar myopathy – a new entity or variant?. <i>Neuropathology and Applied Neurobiology</i> , 2013, 39, 449-453.	1.8	4
426	Clinical and genetic analysis of the first known Asian family with myotonic dystrophy type 2. <i>Journal of Human Genetics</i> , 2014, 59, 129-133.	1.1	4
427	Absence of beta-amyloid deposition in the central nervous system of a transgenic mouse model of distal myopathy with rimmed vacuoles. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2014, 21, 138-139.	1.4	4
428	First Japanese case of muscular dystrophy caused by a mutation in the anoctamin5 gene. <i>Neurology and Clinical Neuroscience</i> , 2015, 3, 150-152.	0.2	4
429	Surface electromyogram and muscle ultrasonography for detection of muscle fasciculations in pediatric peripheral neuropathy. <i>Brain and Development</i> , 2017, 39, 617-620.	0.6	4
430	Author update: Sarcoplasmic MxA expression: A valuable marker of dermatomyositis. <i>Neurology</i> , 2017, 89, 215-215.	1.5	4
431	Syndrome of Inappropriate Antidiuretic Hormone Secretion Associated with Amyotrophic Lateral Sclerosis in a Patient Developing Carbon Dioxide Narcosis. <i>Internal Medicine</i> , 2017, 56, 797-803.	0.3	4
432	Three novel MTM1 pathogenic variants identified in Japanese patients with X-linked myotubular myopathy. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e621.	0.6	4

#	ARTICLE	IF	CITATIONS
433	Evaluation of the Core Formation Process in Congenital Neuromuscular Disease With Uniform Type 1 Fiber and Central Core Disease. <i>Journal of Neuropathology and Experimental Neurology</i> , 2020, 79, 1370-1375.	0.9	4
434	Neutral Lipid Storage Disease Associated with the <i>PNPLA2</i> Gene: Case Report and Literature Review. <i>European Neurology</i> , 2020, 83, 317-322.	0.6	4
435	Duchenne muscular dystrophy-like phenotype in an LGMD2I patient with novel FKRPs gene variants. <i>Human Genome Variation</i> , 2020, 7, 12.	0.4	4
436	Clinicopathological findings of a mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes/Leigh syndrome overlap patient with a novel m. 3482A>G mutation in <i>MTA1</i> . <i>Neuropathology</i> , 2021, 41, 84-90.	0.7	4
437	Successful treatment of congenital myasthenic syndrome caused by a novel compound heterozygous variant in <i>RAPSN</i> . <i>Brain and Development</i> , 2022, 44, 50-55.	0.6	4
438	Neuropathy/intranuclear inclusion bodies in oculopharyngodistal myopathy: A case report. <i>ENeurologicalSci</i> , 2021, 24, 100348.	0.5	4
439	Adult-onset Repeat Rhabdomyolysis with a Very Long-chain Acyl-CoA Dehydrogenase Deficiency Due to Compound Heterozygous <i>ACADVL</i> Mutations. <i>Internal Medicine</i> , 2020, 59, 2729-2732.	0.3	4
440	Imaging-based evaluation of pathogenicity by novel <i>DNM2</i> variants associated with centronuclear myopathy. <i>Human Mutation</i> , 2022, 43, 169-179.	1.1	4
441	Frontal lobe-dominant cerebral blood flow reduction and atrophy can be progressive in Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , 2022, 32, 477-485.	0.3	4
442	A case of Fukuyama-type congenital muscular dystrophy with a very mild mental deficit. <i>Neuromuscular Disorders</i> , 2006, 16, 274-276.	0.3	3
443	Lysosomal myopathies. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2007, 86, 205-214.	1.0	3
444	Lobulated fibers in a patient with 46-year history of limb-girdle muscle weakness. <i>Neuropathology</i> , 2011, 31, 455-457.	0.7	3
445	A case of adult-onset reducing body myopathy presenting a novel clinical feature, asymmetrical involvement of the sternocleidomastoid and trapezius muscles. <i>Journal of the Neurological Sciences</i> , 2014, 343, 206-210.	0.3	3
446	Enzyme-linked immunosorbent assays for diagnosis of immune-mediated necrotizing myopathy. <i>Clinical and Experimental Neuroimmunology</i> , 2016, 7, 291-293.	0.5	3
447	Anti-mitochondrial antibody-associated myositis with eosinophilia and dropped head. <i>ENeurologicalSci</i> , 2018, 11, 15-16.	0.5	3
448	Two closely spaced mutations in cis result in Ullrich congenital muscular dystrophy. <i>Human Genome Variation</i> , 2019, 6, 21.	0.4	3
449	Life-threatening muscle complications of COL4A1-related disorder. <i>Brain and Development</i> , 2020, 42, 93-97.	0.6	3
450	TTN missense variants in two siblings with asymmetric facial and limb weakness. <i>Journal of the Neurological Sciences</i> , 2020, 415, 116885.	0.3	3

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451	Severe cardiac involvement with preserved truncated dystrophin expression in Becker muscular dystrophy by +1G>A DMD splice-site mutation: a case report. <i>Journal of Human Genetics</i> , 2020, 65, 903-909.	1.1	3
452	Beta-actin-associated neurodegeneration presenting Rett-like features: A case report and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 579-583.	0.7	3
453	Monoclonal gammopathy of renal significance (MGRS)-related AL amyloidosis complicated by amyloid myopathy: a case report. <i>BMC Nephrology</i> , 2021, 22, 74.	0.8	3
454	More prominent fibrosis of the cricopharyngeal muscle in inclusion body myositis. <i>Journal of the Neurological Sciences</i> , 2021, 422, 117327.	0.3	3
455	An autopsied case of ADSSL1 myopathy. <i>Neuromuscular Disorders</i> , 2021, 31, 1220-1225.	0.3	3
456	Reliability of antinuclear matrix protein 2 antibody assays in idiopathic inflammatory myopathies is dependent on target protein properties. <i>Journal of Dermatology</i> , 2022, 49, 441-447.	0.6	3
457	Negative result in search for human alpha-dystrobrevin deficiency. <i>Muscle and Nerve</i> , 2003, 28, 387-388.	1.0	2
458	Expression of ARPP-16/19 in Rat Denervated Skeletal Muscle. <i>Journal of Biochemistry</i> , 2003, 134, 57-61.	0.9	2
459	Distal myopathy with rimmed vacuoles in a case of opercular syndrome. <i>Brain and Development</i> , 2006, 28, 458-461.	0.6	2
460	Bilateral occipital cortical dysplasia and white matter T2 hyperintensity with mild non-specific myopathy: Two sibling cases. <i>Brain and Development</i> , 2010, 32, 342-346.	0.6	2
461	Myopathy and neurogenic muscular atrophy in unexpected cardiopulmonary arrest. <i>Pediatrics International</i> , 2011, 53, 159-161.	0.2	2
462	High concentration of middle chain fatty acid in a case of Duchenne muscular dystrophy with severe mental retardation. <i>Pediatrics International</i> , 2012, 54, 137-140.	0.2	2
463	Intranuclear rods myopathy with autonomic dysfunction. <i>Brain and Development</i> , 2013, 35, 686-689.	0.6	2
464	Exome sequencing as a diagnostic tool to identify a causal mutation in genetically highly heterogeneous limb-girdle muscular dystrophy. <i>Journal of Human Genetics</i> , 2013, 58, 564-565.	1.1	2
465	Hypoxic ischemic encephalopathy in a case of intranuclear rod myopathy without any prenatal sentinel event. <i>Brain and Development</i> , 2015, 37, 265-269.	0.6	2
466	Case of McLeod syndrome with a novel genetic mutation. <i>Neurology and Clinical Neuroscience</i> , 2016, 4, 115-117.	0.2	2
467	Severe Glomerular Endothelial Injury Associated with a Short D4Z4 Repeat on Chromosome 4q35. <i>Internal Medicine</i> , 2017, 56, 1849-1853.	0.3	2
468	Case of immune-mediated necrotizing myopathy associated with anti-signal recognition particle antibodies: Dramatic improvement after rituximab, cyclophosphamide, doxorubicin, vincristine and prednisolone therapy for intravascular large B-cell lymphoma. <i>Clinical and Experimental Neuroimmunology</i> , 2018, 9, 177-181.	0.5	2

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469	214th ENMC International Workshop: Establishing an international consortium for gene discovery and clinical research for Congenital Muscle Disease, Heemskerk, the Netherlands, 6â€“18 October 2015. <i>Neuromuscular Disorders</i> , 2019, 29, 644-650.	0.3	2
470	Physician-Level Aggregated Classifier for Genetic Muscle Disorders. , 2019, , .		2
471	An adult nemaline myopathy patient with respiratory and heart failure harboring a novel NEB variant. <i>ENeurologicalSci</i> , 2020, 21, 100268.	0.5	2
472	Clinical, imaging, morphologic, and molecular features of X-linked VMA21-related myopathy in two unrelated Brazilian families. <i>Journal of the Neurological Sciences</i> , 2020, 415, 116977.	0.3	2
473	Hyperglycemic Crisis in Patients With Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like Episodes (MELAS). <i>Pediatric Neurology</i> , 2021, 114, 1-4.	1.0	2
474	A case of eosinophilic fasciitis without skin manifestations: a case report in a patient with lupus and literature review. <i>Clinical Rheumatology</i> , 2021, 40, 2477-2483.	1.0	2
475	Underlying diseases in sporadic presentation of high creatine kinase levels in girls. <i>Clinica Chimica Acta</i> , 2021, 519, 198-203.	0.5	2
476	FGRP mutations cause congenital muscular dystrophy 1C and limb-girdle muscular dystrophy 2I in Asian patients. <i>Journal of Clinical Neuroscience</i> , 2021, 92, 215-221.	0.8	2
477	A Japanese Patient with Hereditary Myopathy with Early Respiratory Failure Due to the p.P31732L Mutation of Titin. <i>Internal Medicine</i> , 2022, 61, 1587-1592.	0.3	2
478	Resection of Gastric Cancer Remitted Anti-signal Recognition Particle Myopathy. <i>Internal Medicine</i> , 2022, 61, 2509-2515.	0.3	2
479	Tulobuterol is a potential therapeutic drug in congenital myasthenic syndrome. <i>Pediatrics International</i> , 2022, 64, e151115.	0.2	2
480	Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes. <i>Journal of Neurology</i> , 2022, 269, 4129-4140.	1.8	2
481	A 7â€“year-old female with hypotonia and scoliosis. <i>Brain Pathology</i> , 2022, 32, .	2.1	2
482	Determining neurodevelopmental manifestations in Duchenne muscular dystrophy using a battery of brief tests. <i>Journal of the Neurological Sciences</i> , 2022, 440, 120340.	0.3	2
483	Reply to Lemmers et al.. <i>Neuromuscular Disorders</i> , 2006, 16, 617-618.	0.3	1
484	Daily or alternative, that is the question: steroid therapy for Duchenne muscular dystrophy patients. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 591-591.	0.9	1
485	The muscle findings in a pediatric patient with live attenuated oral polio vaccine-related flaccid monoplegia. <i>Vaccine</i> , 2014, 32, 5379-5381.	1.7	1
486	Limb-Girdle Muscular Dystrophy. , 2015, , 1113-1120.		1

#	ARTICLE	IF	CITATIONS
487	Ophthalmoplegia in congenital neuromuscular disease with uniform type 1 fiber. <i>Brain and Development</i> , 2015, 37, 459-462.	0.6	1
488	An elderly-onset limb girdle muscular dystrophy type 1B (LGMD1B) with pseudo-hypertrophy of paraspinal muscles. <i>Neuromuscular Disorders</i> , 2016, 26, 593-597.	0.3	1
489	Japanese case of Emery-Dreifuss muscular dystrophy with a novel LMNA missense mutation. <i>Neurology and Clinical Neuroscience</i> , 2016, 4, 124-125.	0.2	1
490	Screening for late-onset Pompe disease in undiagnosed myopathies. <i>Neurology and Clinical Neuroscience</i> , 2017, 5, 60-64.	0.2	1
491	Granuloma formation in a patient with GNE myopathy: A case report. <i>Neuromuscular Disorders</i> , 2017, 27, 183-184.	0.3	1
492	<i>Muscular Dystrophies</i> . , 2017, , 183-192.		1
493	A 31-Year-Old Man with Slowly Progressive Limb Muscle Weakness and Respiratory Insufficiency. <i>Brain Pathology</i> , 2018, 28, 123-124.	2.1	1
494	New Criteria Needed for Antisynthetase Syndrome—Reply. <i>JAMA Neurology</i> , 2018, 75, 259.	4.5	1
495	A mild myopathy with anti-SRP plus anti-PL-12 antibodies successfully treated by oral steroid monotherapy. <i>Journal of the Neurological Sciences</i> , 2018, 388, 7-9.	0.3	1
496	Different clinicopathological features between Japanese siblings with facioscapulohumeral muscular dystrophy 2 with a novel nonsense SMCHD1 mutation (Arg552—). <i>Journal of Clinical Neuroscience</i> , 2018, 58, 215-217.	0.8	1
497	Dropped Head Syndrome Caused by Immune-mediated Necrotizing Myopathy. <i>Internal Medicine</i> , 2019, 58, 3343-3344.	0.3	1
498	“Boule du biceps” in dysferlinopathy. <i>Neurology</i> , 2020, 94, 83-84.	1.5	1
499	Needle electromyography, muscle MRI, and muscle pathology: Correlations in idiopathic inflammatory myopathies. <i>Neurology and Clinical Neuroscience</i> , 2020, 8, 28-35.	0.2	1
500	Respiratory Dysfunction in Becker Muscular Dystrophy Patients: A Case Series and Autopsy Report. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 425-431.	1.1	1
501	Light-chain amyloid myopathy isolated to skeletal muscles: A case report. <i>Clinical Case Reports (discontinued)</i> , 2020, 8, 2869-2873.	0.2	1
502	A case of sporadic late-onset nemaline myopathy without monoclonal gammopathy of unknown significance/human immunodeficiency virus successfully treated with intravenous gamma globulin. <i>Clinical and Experimental Neuroimmunology</i> , 2021, 12, 124-126.	0.5	1
503	Mild form of Danon disease: two case reports. <i>Neuromuscular Disorders</i> , 2021, 31, 1207-1211.	0.3	1
504	Noteworthy Cardiovascular Involvement with Sporadic Late-onset Nemaline Myopathy. <i>Internal Medicine</i> , 2021, 60, 2327-2332.	0.3	1

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505	Marked Respiratory Failure in an Ambulant Patient with Immune-mediated Necrotizing Myopathy and Anti-Kv1.4 and Anti-titin Antibodies. <i>Internal Medicine</i> , 2021, 60, 2671-2675.	0.3	1
506	Myoclonus epilepsy associated with ragged-red fibers: A G-to-A mutation at nucleotide pair 8363 in mitochondrial tRNA ^{Lys} in two families. , 1997, 20, 271.		1
507	Mitochondrial Myopathies. , 2014, , 1335-1353.		1
508	Maximal Multistage Shuttle Run Test-induced Myalgia in a Patient with Muscle Phosphorylase B Kinase Deficiency. <i>Internal Medicine</i> , 2022, , .	0.3	1
509	Lysosomal membrane disorders: lysosome-associated membrane protein-2 deficiency (Danon disease). , 2020, , 567-574.		1
510	Nemaline Myopathy Initially Diagnosed as Right Heart Failure with Type 2 Respiratory Failure. <i>Internal Medicine</i> , 2022, 61, 1897-1901.	0.3	1
511	A case of delayed diagnosis of Becker muscular dystrophy due to underlying developmental disorders. <i>Brain and Development</i> , 2022, 44, 259-262.	0.6	1
512	Identification of a novel mutation and genotype-phenotype relationship in MEGF10 myopathy. <i>Neuromuscular Disorders</i> , 2022, 32, 436-440.	0.3	1
513	A 78-year-old Japanese male with late-onset PHKA1-associated distal myopathy: Case report and literature review. <i>Neuromuscular Disorders</i> , 2022, , .	0.3	1
514	A new form of muscular dystrophy with mitochondrial structural abnormalities. <i>Muscle and Nerve</i> , 2001, 24, 1710-1711.	1.0	0
515	Teaching Neuro <i>Images</i> : Unilateral arm and contralateral leg amyotrophy in FSHD. <i>Neurology</i> , 2012, 79, e46.	1.5	0
516	Additional evidence that the ryanodine receptor gene (RYR1) causes malignant hyperthermia and severe skeletal malformations. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 234-235.	0.7	0
517	Family with centronuclear myopathy as a result of a novel p.R369G <i>DNM2</i> mutation. <i>Neurology and Clinical Neuroscience</i> , 2015, 3, 161-162.	0.2	0
518	Case of <i>dynamitin</i> mutation-related sporadic centronuclear myopathy with peripheral neuropathy. <i>Neurology and Clinical Neuroscience</i> , 2015, 3, 131-133.	0.2	0
519	2 Month-Old Male with Hypotonia. <i>Brain Pathology</i> , 2015, 25, 651-652.	2.1	0
520	Lysosomal Membrane Disorders. , 2015, , 411-417.		0
521	Kyphoscoliosis and easy fatigability in a 14-year-old boy. <i>Neuropathology</i> , 2015, 35, 91-93.	0.7	0
522	Immune-mediated Necrotizing Myopathy (IMNM) and HLA Polymorphisms. <i>Major Histocompatibility Complex</i> , 2017, 24, 46-53.	0.2	0

#	ARTICLE	IF	CITATIONS
523	A 62-year-old Woman with A History of Muscle Pain and Skin Rash for 1 Month. <i>Brain Pathology</i> , 2018, 28, 121-122.	2.1	0
524	Case of elderly-onset multiple acyl-CoA dehydrogenase deficiency with a novel ETFDH mutation shows progressive muscle weakness and rhabdomyolysis. <i>Neurology and Clinical Neuroscience</i> , 2018, 6, 39-41.	0.2	0
525	A new familial distal myopathy in Japan with predominant upper extremities. <i>Journal of the Neurological Sciences</i> , 2018, 390, 205-207.	0.3	0
526	Paraspinal amyotrophy in DNM-2-related centronuclear myopathy. <i>Journal of the Neurological Sciences</i> , 2019, 407, 116537.	0.3	0
527	Tonsillectomy Improved Therapeutic Response in Anti-SRP Myopathy With Chronic Tonsillitis. <i>Frontiers in Immunology</i> , 2020, 11, 595480.	2.2	0
528	Selective involvement of semitendinosus in hereditary myopathy with early respiratory failure. <i>Neurology and Clinical Neuroscience</i> , 2020, 8, 428-429.	0.2	0
529	Limb-girdle muscular dystrophy. , 2020, , 437-449.		0
530	A case of type 1 facioscapulohumeral muscular dystrophy (FSHD) with restrictive ventilatory defect and congestive heart failure. <i>ENeurologicalSci</i> , 2020, 21, 100284.	0.5	0
531	Collagen α 1(I) related myopathy with subacute presentation of hypercapnic respiratory failure following pneumonia. <i>Neurology and Clinical Neuroscience</i> , 2020, 8, 320-322.	0.2	0
532	A symptomatic male carrier of Duchenne muscular dystrophy with Klinefelter's syndrome mimicking Becker muscular dystrophy. <i>Neuromuscular Disorders</i> , 2021, 31, 666-672.	0.3	0
533	Case of anti-nuclear matrix protein 2 antibody-positive juvenile dermatomyositis preceded by linear cutaneous lupus erythematosus on the face. <i>Journal of Dermatology</i> , 2022, 49, e18.	0.6	0
534	Metabolic changes in sialic acid synthesis pathway in GNE myopathy model mice with long-term sialic acid treatment. <i>FASEB Journal</i> , 2012, 26, 551.5.	0.2	0
535	Identification of biomarkers for GNE myopathy. <i>FASEB Journal</i> , 2012, 26, 1122.3.	0.2	0
536	VII. Supplementation therapy for metabolic myopathies. <i>The Journal of the Japanese Society of Internal Medicine</i> , 2018, 107, 1501-1506.	0.0	0
537	Series: Diagnosis at a Glance. <i>The Journal of the Japanese Society of Internal Medicine</i> , 2020, 109, 2411-2413.	0.0	0
538	TNNI1 Mutated in Autosomal Dominant Proximal Arthrogryposis. <i>Neurology: Genetics</i> , 2022, 8, e649.	0.9	0
539	Advances in understanding of the natural history, mechanism, extra-muscular manifestations and treatment of GNE myopathy. <i>Neurology and Clinical Neuroscience</i> , 2022, 10, 289-297.	0.2	0
540	Series: Diagnosis at a Glance. <i>The Journal of the Japanese Society of Internal Medicine</i> , 2021, 110, 315-318.	0.0	0

#	ARTICLE	IF	CITATIONS
541	5. Classification of Myositis and Disease Concept-recent Advances-. The Journal of the Japanese Society of Internal Medicine, 2021, 110, 481-485.	0.0	0
542	Myoglobinopathy affecting facial and oropharyngeal muscles. Neuromuscular Disorders, 2022, , .	0.3	0
543	Sick sinus syndrome concomitant with myopathy associated with anti-mitochondrial antibodies: a case report. European Heart Journal - Case Reports, 0, , .	0.3	0
544	A 10-year-old girl with low-grade B cell lymphoma complicated by anti-nuclear matrix protein 2 autoantibody-positive juvenile dermatomyositis. Rheumatology, 2021, , .	0.9	0
545	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
546	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
547	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
548	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
549	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
550	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
551	Muscle biochemical and pathological diagnosis in Pompe disease. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 1142-1145.	0.9	0
552	Sudden cardiac death prevention in an Emeryâ€Dreifuss muscular dystrophy patient. Pediatrics International, 2022, 64, e15204.	0.2	0