Ichizo Nishino

List of Publications by Year
in descending order

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553	33,869	8755 75	4991 167
papers	citations	h-index	g-index

619	619	619	40878
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
2	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	9.1	3,122
3	Guidelines for the use and interpretation of assays for monitoring autophagy in higher eukaryotes. Autophagy, 2008, 4, 151-175.	9.1	2,064
4	Primary LAMP-2 deficiency causes X-linked vacuolar cardiomyopathy and myopathy (Danon disease). Nature, 2000, 406, 906-910.	27.8	865
5	Adiponectin and AdipoR1 regulate PGC-1Î± and mitochondria by Ca2+ and AMPK/SIRT1. Nature, 2010, 464, 1313-1319.	27.8	859
6	Thymidine Phosphorylase Gene Mutations in MNGIE, a Human Mitochondrial Disorder. Science, 1999, 283, 689-692.	12.6	827
7	Post-translational disruption of dystroglycanâ€“ligand interactions in congenital muscular dystrophies. Nature, 2002, 418, 417-421.	27.8	747
8	Fatal infantile cardioencephalomyopathy with COX deficiency and mutations in SCO2, a COX assembly gene. Nature Genetics, 1999, 23, 333-337.	21.4	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. Journal of Biological Chemistry, 2004, 279, 41114-41123.	3.4	488
10	Distinctive patterns of microRNA expression in primary muscular disorders. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 17016-17021.	7.1	458
11	Human PTRF mutations cause secondary deficiency of caveolins resulting in muscular dystrophy with generalized lipodystrophy. Journal of Clinical Investigation, 2009, 119, 2623-2633.	8.2	350
12	Mitochondrial neurogastrointestinal encephalomyopathy: An autosomal recessive disorder due to thymidine phosphorylase mutations. Annals of Neurology, 2000, 47, 792-800.	5.3	324
13	Misregulated alternative splicing of BIN1 is associated with T tubule alterations and muscle weakness in myotonic dystrophy. Nature Medicine, 2011, 17, 720-725.	30.7	299
14	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. Nature Genetics, 2019, 51, 1222-1232.	21.4	265
15	LARGE can functionally bypass Î±-dystroglycan glycosylation defects in distinct congenital muscular dystrophies. Nature Medicine, 2004, 10, 696-703.	30.7	253
16	Identification and characterization of PDGFRÎ±+ mesenchymal progenitors in human skeletal muscle. Cell Death and Disease, 2014, 5, e1186-e1186.	6.3	241
17	Central core disease is due to RYR1 mutations in more than 90% of patients. Brain, 2006, 129, 1470-1480.	7.6	233
18	Clinical features and prognosis in anti-SRP and anti-HMGCR necrotising myopathy. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 1038-1044.	1.9	229

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19	The sarcolemmal proteins dysferlin and caveolin-3 interact in skeletal muscle. Human Molecular Genetics, 2001, 10, 1761-1766.	2.9	214
20	Distal myopathy with rimmed vacuoles is allelic to hereditary inclusion body myopathy. Neurology, 2002, 59, 1689-1693.	1.1	209
21	Altered Thymidine Metabolism Due to Defects of Thymidine Phosphorylase. Journal of Biological Chemistry, 2002, 277, 4128-4133.	3.4	209
22	Molecular features of the CAG repeats and clinical manifestation of Machado–Joseph disease. Human Molecular Genetics, 1995, 4, 807-812.	2.9	191
23	Autophagic degradation of nuclear components in mammalian cells. Autophagy, 2009, 5, 795-804.	9.1	189
24	Mutations in DNMT3B Modify Epigenetic Repression of the D4Z4 Repeat and the Penetrance of Facioscapulohumeral Dystrophy. American Journal of Human Genetics, 2016, 98, 1020-1029.	6.2	188
25	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 6-18.	6.2	186
26	Allogeneic stem cell transplantation corrects biochemical derangements in MNGIE. Neurology, 2006, 67, 1458-1460.	1.1	172
27	Structural and Functional Mutations of the Perlecan Gene Cause Schwartz-Jampel Syndrome, with Myotonic Myopathy and Chondrodysplasia. American Journal of Human Genetics, 2002, 70, 1368-1375.	6.2	168
28	Prophylactic treatment with sialic acid metabolites precludes the development of the myopathic phenotype in the DMRV-hIBM mouse model. Nature Medicine, 2009, 15, 690-695.	30.7	167
29	TMEM43 mutations in emeryâ€dreifuss muscular dystrophyâ€related myopathy. Annals of Neurology, 2011, 69, 1005-1013.	5.3	164
30	Inflammatory myopathy with anti-signal recognition particle antibodies: case series of 100 patients. Orphanet Journal of Rare Diseases, 2015, 10, 61.	2.7	156
31	Localization of a gene for myoclonus-dystonia to chromosome 7q21-q31. Annals of Neurology, 1999, 46, 794-798.	5.3	154
32	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708.	8.2	153
33	Actin mutations are one cause of congenital fibre type disproportion. Annals of Neurology, 2004, 56, 689-694.	5.3	149
34	239th ENMC International Workshop: Classification of dermatomyositis, Amsterdam, the Netherlands, 14â€“16 December 2018. Neuromuscular Disorders, 2020, 30, 70-92.	0.6	148
35	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 1108-1117.	6.2	147
36	Fukutin gene mutations cause dilated cardiomyopathy with minimal muscle weakness. Annals of Neurology, 2006, 60, 597-602.	5.3	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. Journal of Biological Chemistry, 2004, 279, 11402-11407.	3.4	139
38	CXorf6 is a causative gene for hypospadias. Nature Genetics, 2006, 38, 1369-1371.	21.4	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. Human Molecular Genetics, 2007, 16, 2669-2682.	2.9	136
40	Lysosomal myopathies: An excessive build-up in autophagosomes is too much to handle. Neuromuscular Disorders, 2008, 18, 521-529.	0.6	136
41	Worldwide distribution and broader clinical spectrum of muscle-eye-brain disease. Human Molecular Genetics, 2003, 12, 527-534.	2.9	133
42	Malignant Hyperthermia in Japan. Anesthesiology, 2006, 104, 1146-1154.	2.5	132
43	Dominant mutations in ORAI1 cause tubular aggregate myopathy with hypocalcemia via constitutive activation of store-operated Ca2+ channels. Human Molecular Genetics, 2015, 24, 637-648.	2.9	132
44	GNE myopathy: current update and future therapy. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 385-392.	1.9	131
45	Unbalanced deoxynucleotide pools cause mitochondrial DNA instability in thymidine phosphorylase-deficient mice. Human Molecular Genetics, 2009, 18, 714-722.	2.9	123
46	VMA21 deficiency prevents vacuolar ATPase assembly and causes autophagic vacuolar myopathy. Acta Neuropathologica, 2013, 125, 439-457.	7.7	119
47	ETFDH mutations, CoQ10 levels, and respiratory chain activities in patients with riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Neuromuscular Disorders, 2009, 19, 212-216.	0.6	118
48	Sarcoplasmic MxA expression. Neurology, 2017, 88, 493-500.	1.1	118
49	Skeletal Muscle Involvement in Antisynthetase Syndrome. JAMA Neurology, 2017, 74, 992.	9.0	117
50	Deficiency of Î±-Dystroglycan in Muscleâ€“Eyeâ€“Brain Disease. Biochemical and Biophysical Research Communications, 2002, 291, 1283-1286.	2.1	115
51	A Congenital Muscular Dystrophy with Mitochondrial Structural Abnormalities Caused by Defective De Novo Phosphatidylcholine Biosynthesis. American Journal of Human Genetics, 2011, 88, 845-851.	6.2	115
52	Defects in amphiphysin 2 (BIN1) and triads in several forms of centronuclear myopathies. Acta Neuropathologica, 2011, 121, 253-266.	7.7	113
53	Danon disease: a phenotypic expression of LAMP-2 deficiency. Acta Neuropathologica, 2015, 129, 391-398.	7.7	112
54	A Gne knockout mouse expressing human V572L mutation develops features similar to distal myopathy with rimmed vacuoles or hereditary inclusion body myopathy. Human Molecular Genetics, 2007, 16, 115-128.	2.9	111

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55	Autophagic Vacuolar Myopathy. Seminars in Pediatric Neurology, 2006, 13, 90-95.	2.0	110
56	Autophagic vacuolar myopathies. Current Neurology and Neuroscience Reports, 2003, 3, 64-69.	4.2	108
57	Definitive Diagnosis of Mitochondrial Neurogastrointestinal Encephalomyopathy by Biochemical Assays. Clinical Chemistry, 2004, 50, 120-124.	3.2	107
58	Unifying Nomenclature for the Isoforms of the Lysosomal Membrane Protein LAMP-2. Traffic, 2005, 6, 1058-1061.	2.7	107
59	Defects of intergenomic communication: autosomal disorders that cause multiple deletions and depletion of mitochondrial DNA. Seminars in Cell and Developmental Biology, 2001, 12, 417-427.	5.0	105
60	Overexpression of Peroxisome Proliferator-Activated Receptor Î³ Co-Activator-1Î± Leads to Muscle Atrophy with Depletion of ATP. American Journal of Pathology, 2006, 169, 1129-1139.	3.8	96
61	Defects of Vps15 in skeletal muscles lead to autophagic vacuolar myopathy and lysosomal disease. EMBO Molecular Medicine, 2013, 5, 870-890.	6.9	96
62	Cell-Surface Protein Profiling Identifies Distinctive Markers of Progenitor Cells in Human Skeletal Muscle. Stem Cell Reports, 2016, 7, 263-278.	4.8	95
63	Centronuclear myopathy in mice lacking a novel muscle-specific protein kinase transcriptionally regulated by MEF2. Genes and Development, 2005, 19, 2066-2077.	5.9	93
64	Myoclonus epilepsy associated with ragged-red fibers: A G-to-A mutation at nucleotide pair 8363 in mitochondrial tRNALys in two families. Muscle and Nerve, 1997, 20, 271-278.	2.2	92
65	Mitochondrial Neurogastrointestinal Encephalomyopathy Syndrome Maps to Chromosome 22q13.32-qter. American Journal of Human Genetics, 1998, 63, 526-533.	6.2	91
66	Emerin-Lacking Mice Show Minimal Motor and Cardiac Dysfunctions with Nuclear-Associated Vacuoles. American Journal of Pathology, 2006, 168, 907-917.	3.8	91
67	Safety and efficacy of intravenous bimagrumab in inclusion body myositis (RESILIENT): a randomised, double-blind, placebo-controlled phase 2b trial. Lancet Neurology, The, 2019, 18, 834-844.	10.2	91
68	Primary collagen VI deficiency is the second most common congenital muscular dystrophy in Japan. Neurology, 2007, 69, 1035-1042.	1.1	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. Developmental Biology, 2012, 361, 79-89.	2.0	90
70	Expansion of GGC Repeat in GIPC1 Is Associated with Oculopharyngodistal Myopathy. American Journal of Human Genetics, 2020, 106, 793-804.	6.2	90
71	Nâ€‹<scp>WASP</scp> is required for Amphiphysinâ€‹2/<scp>BIN</scp>1â€‹dependent nuclear positioning and triad organization in skeletal muscle and is involved in the pathophysiology of centronuclear myopathy. EMBO Molecular Medicine, 2014, 6, 1455-1475.	6.9	87
72	Mechanisms of Genomic Instabilities Underlying Two Common Fragile-Site-Associated Loci, PARK2 and DMD, in Germ Cell and Cancer Cell Lines. American Journal of Human Genetics, 2010, 87, 75-89.	6.2	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. American Journal of Pathology, 2011, 178, 2224-2235.	3.8	84
74	Homozygous nonsense variant in LRIF1 associated with facioscapulohumeral muscular dystrophy. Neurology, 2020, 94, e2441-e2447.	1.1	84
75	Myopathy Associated With Antibodies to Signal Recognition Particle. Archives of Neurology, 2012, 69, 728-32.	4.5	82
76	Autophagic Vacuoles with Sarcolemmal Features Delineate Danon Disease and Related Myopathies. Journal of Neuropathology and Experimental Neurology, 2005, 64, 513-522.	1.7	81
77	Disrupted prenatal RNA processing and myogenesis in congenital myotonic dystrophy. Genes and Development, 2017, 31, 1122-1133.	5.9	80
78	Genetic diagnosis of Duchenne/Becker muscular dystrophy using next-generation sequencing: validation analysis of DMD mutations. Journal of Human Genetics, 2016, 61, 483-489.	2.3	79
79	CGG expansion in NOTCH2NLC is associated with oculopharyngodistal myopathy with neurological manifestations. Acta Neuropathologica Communications, 2020, 8, 204.	5.2	76
80	Genotype and phenotype analyses in 136 patients with single large-scale mitochondrial DNA deletions. Journal of Human Genetics, 2008, 53, 598-606.	2.3	75
81	Muscle choline kinase beta defect causes mitochondrial dysfunction and increased mitophagy. Human Molecular Genetics, 2011, 20, 3841-3851.	2.9	75
82	Clinical and genetic analysis of lipid storage myopathies. Muscle and Nerve, 2009, 39, 333-342.	2.2	74
83	Statins and Myotoxic Effects Associated With Anti-3-Hydroxy-3-Methylglutaryl-Coenzyme A Reductase Autoantibodies. Medicine (United States), 2015, 94, e416.	1.0	74
84	Inflammatory myopathy associated with PD-1 inhibitors. Journal of Autoimmunity, 2019, 100, 105-113.	6.5	73
85	Mutations in the satellite cell gene MEGF10 cause a recessive congenital myopathy with minicores. Neurogenetics, 2012, 13, 115-124.	1.4	68
86	Where are we moving in the classification of idiopathic inflammatory myopathies?. Current Opinion in Neurology, 2020, 33, 590-603.	3.6	68
87	Inflammatory changes in infantile-onset LMNA-associated myopathy. Neuromuscular Disorders, 2011, 21, 563-568.	0.6	67
88	Autophagy in Lysosomal Myopathies. Brain Pathology, 2012, 22, 82-88.	4.1	67
89	Analysis of mouse models of cytochrome c oxidase deficiency owing to mutations in Sco2. Human Molecular Genetics, 2010, 19, 170-180.	2.9	66
90	Biallelic Mutations in MYPN , Encoding Myopalladin, Are Associated with Childhood-Onset, Slowly Progressive Nemaline Myopathy. American Journal of Human Genetics, 2017, 100, 169-178.	6.2	66

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91	Transgenic Monkey Model of the Polyglutamine Diseases Recapitulating Progressive Neurological Symptoms. ENeuro, 2017, 4, ENEURO.0250-16.2017.	1.9	66
92	A new congenital muscular dystrophy with mitochondrial structural abnormalities. , 1998, 21, 40-47.		64
93	MNGIE: from nuclear DNA to mitochondrial DNA. Neuromuscular Disorders, 2001, 11, 7-10.	0.6	64
94	Distal myopathy with rimmed vacuoles and hereditary inclusion body myopathy. Current Neurology and Neuroscience Reports, 2005, 5, 61-65.	4.2	64
95	Congenital neuromuscular disease with uniform type 1 fiber and RYR1 mutation. Neurology, 2008, 70, 114-122.	1.1	64
96	Pediatric necrotizing myopathy associated with anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase antibodies. Rheumatology, 2017, 56, 287-293.	1.9	64
97	GNE myopathy: New name and new mutation nomenclature. Neuromuscular Disorders, 2014, 24, 387-389.	0.6	61
98	Hepatitis C virus infection in inclusion body myositis. Neurology, 2016, 86, 211-217.	1.1	61
99	Classification of idiopathic inflammatory myopathies: pathology perspectives. Current Opinion in Neurology, 2019, 32, 704-714.	3.6	61
100	Lipid Storage Myopathy. Current Neurology and Neuroscience Reports, 2011, 11, 97-103.	4.2	60
101	Perifascicular necrosis in anti-synthetase syndrome beyond anti-Jo-1. Brain, 2016, 139, e50-e50.	7.6	60
102	Protein and gene analyses of dysferlinopathy in a large group of Japanese muscular dystrophy patients. Journal of the Neurological Sciences, 2003, 211, 23-28.	0.6	58
103	Dysferlin Interacts with Affixin (Î²-Parvin) at the Sarcolemma. Journal of Neuropathology and Experimental Neurology, 2005, 64, 334-340.	1.7	57
104	Nuclear changes in skeletal muscle extend to satellite cells in autosomal dominant Emery-Dreifuss muscular dystrophy/limb-girdle muscular dystrophy 1B. Neuromuscular Disorders, 2009, 19, 29-36.	0.6	57
105	Characteristics of Japanese Duchenne and Becker muscular dystrophy patients in a novel Japanese national registry of muscular dystrophy (Remudy). Orphanet Journal of Rare Diseases, 2013, 8, 60.	2.7	56
106	LAMPâ€2â€deficient human B cells exhibit altered MHC class II presentation of exogenous antigens. Immunology, 2010, 131, 318-330.	4.4	55
107	The cathepsin L gene is a direct target of FOXO1 in skeletal muscle. Biochemical Journal, 2010, 427, 171-178.	3.7	55
108	Characterization of the Asian myopathy patients with VCP mutations. European Journal of Neurology, 2012, 19, 501-509.	3.3	55

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109	Rigid spine syndrome caused by a novel mutation in four-and-a-half LIM domain 1 gene (FHL1). Neuromuscular Disorders, 2008, 18, 959-961.	0.6	54
110	Establishment of an Improved Mouse Model for Infantile Neuroaxonal Dystrophy That Shows Early Disease Onset and Bears a Point Mutation in Pla2g6. American Journal of Pathology, 2009, 175, 2257-2263.	3.8	54
111	The First Molecular Evidence That Autophagy Relates Rimmed Vacuole Formation in Chloroquine Myopathy. Journal of Biochemistry, 2002, 131, 647-651.	1.7	53
112	Clinical and histological findings associated with autoantibodies detected by RNA immunoprecipitation in inflammatory myopathies. Journal of Neuroimmunology, 2014, 274, 202-208.	2.3	53
113	Ullrich congenital muscular dystrophy: clinicopathological features, natural history and pathomechanism(s). Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 280-287.	1.9	53
114	DNAJB6 myopathy in an Asian cohort and cytoplasmic/nuclear inclusions. Neuromuscular Disorders, 2013, 23, 269-276.	0.6	52
115	Sialyllactose ameliorates myopathic phenotypes in symptomatic GNE myopathy model mice. Brain, 2014, 137, 2670-2679.	7.6	52
116	Integrated Diagnosis Project for Inflammatory Myopathies: An association between autoantibodies and muscle pathology. Autoimmunity Reviews, 2017, 16, 693-700.	5.8	52
117	Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy. American Journal of Human Genetics, 2016, 99, 950-961.	6.2	51
118	Targeted massively parallel sequencing and histological assessment of skeletal muscles for the molecular diagnosis of inherited muscle disorders. Journal of Medical Genetics, 2017, 54, 104-110.	3.2	51
119	Distal lipid storage myopathy due to PNPLA2 mutation. Neuromuscular Disorders, 2008, 18, 671-674.	0.6	50
120	Mutation profile of the GNE gene in Japanese patients with distal myopathy with rimmed vacuoles (GNE) Tj ETQq0 0 0 rgBT /Overlock 10	1.9	50
121	Autophagy in a Mouse Model of Distal Myopathy with Rimmed Vacuoles or Hereditary Inclusion Body Myopathy. Autophagy, 2007, 3, 396-398.	9.1	49
122	Increase in number of sporadic inclusion body myositis (sIBM) in Japan. Journal of Neurology, 2012, 259, 554-556.	3.6	49
123	A patient-derived iPSC model revealed oxidative stress increases facioscapulohumeral muscular dystrophy-causative DUX4. Human Molecular Genetics, 2018, 27, 4024-4035.	2.9	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. Journal of the Neurological Sciences, 2012, 318, 100-105.	0.6	47
125	Congenital muscular dystrophy with fatty liver and infantile-onset cataract caused by TRAPPC11 mutations: broadening of the phenotype. Skeletal Muscle, 2015, 5, 29.	4.2	47
126	HLA-DRB1 alleles in immune-mediated necrotizing myopathy. Neurology, 2016, 87, 1954-1955.	1.1	47

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127	Localization of Calpain 3 in Human Skeletal Muscle and Its Alteration in Limb-Girdle Muscular Dystrophy 2A Muscle. Journal of Biochemistry, 2003, 133, 659-664.	1.7	46
128	NOVEL FHL1 MUTATIONS IN FATAL AND BENIGN REDUCING BODY MYOPATHY. Neurology, 2009, 72, 375-376.	1.1	46
129	Defects in autophagosome-lysosome fusion underlie Vici syndrome, a neurodevelopmental disorder with multisystem involvement. Scientific Reports, 2017, 7, 3552.	3.3	46
130	Hypoparathyroidism and insulin-dependent diabetes mellitus in a patient with Kearns-Sayre syndrome harbouring a mitochondrial DNA deletion. Clinical Endocrinology, 1996, 45, 637-641.	2.4	43
131	Comprehensive analysis for genetic diagnosis of Dystrophinopathies in Japan. Orphanet Journal of Rare Diseases, 2017, 12, 149.	2.7	43
132	Subcellular Localization of Fukutin and Fukutin-Related Protein in Muscle Cells. Journal of Biochemistry, 2004, 135, 709-712.	1.7	42
133	Congenital muscular dystrophy with glycosylation defects of Î±-dystroglycan in Japan. Neuromuscular Disorders, 2005, 15, 342-348.	0.6	42
134	NovelLamp-2 gene mutation and successful treatment with heart transplantation in a large family with Danon disease. Muscle and Nerve, 2006, 33, 393-397.	2.2	42
135	Infantile facioscapulohumeral muscular dystrophy revisited: Expansion of clinical phenotypes in patients with a very short EcoRI fragment. Neuromuscular Disorders, 2013, 23, 298-305.	0.6	42
136	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. Journal of Human Genetics, 2014, 59, 581-583.	2.3	42
137	Sialic acid deficiency is associated with oxidative stress leading to muscle atrophy and weakness in GNE myopathy. Human Molecular Genetics, 2017, 26, 3081-3093.	2.9	42
138	The 3260 mutation in mitochondrial DNA can cause mitochondrial myopathy, encephalopathy, lactic acidosis, and strokelike 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 hypogenitalism. Neuromuscular Disorders, 2005, 15, 245-252.	0.6	41
140	Mutation analysis of theGNE gene in distal myopathy with rimmed vacuoles (DMRV) patients in Thailand. Muscle and Nerve, 2006, 34, 775-778.	2.2	41
141	Aberrant neuromuscular junctions and delayed terminal muscle fiber maturation in Î±-dystroglycanopathies. Human Molecular Genetics, 2006, 15, 1279-1289.	2.9	41
142	A Novel Mutation in the Mitochondrial tRNAThrGene Associated with a Mitochondrial Encephalomyopathy. Biochemical and Biophysical Research Communications, 1996, 225, 180-185.	2.1	40
143	Proteolysis of Î²-dystroglycan in muscular diseases. Neuromuscular Disorders, 2005, 15, 336-341.	0.6	40
144	Peracetylated N-Acetylmannosamine, a Synthetic Sugar Molecule, Efficiently Rescues Muscle Phenotype and Biochemical Defects in Mouse Model of Sialic Acid-deficient Myopathy. Journal of Biological Chemistry, 2012, 287, 2689-2705.	3.4	40

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145	Aberrant Myokine Signaling in Congenital Myotonic Dystrophy. Cell Reports, 2017, 21, 1240-1252.	6.4	40
146	Megaconial congenital muscular dystrophy due to loss-of-function mutations in choline kinase Î². Current Opinion in Neurology, 2013, 26, 536-543.	3.6	39
147	Association of Dermatomyositis Sine Dermatitis With Antiâ€“Nuclear Matrix Protein 2 Autoantibodies. JAMA Neurology, 2020, 77, 872.	9.0	39
148	Limb-Girdle Muscular Dystrophy Due to Emerin Gene Mutations. Archives of Neurology, 2007, 64, 1038.	4.5	38
149	Reversible infantile respiratory chain deficiency: A clinical and molecular study. Annals of Neurology, 2010, 68, 845-854.	5.3	38
150	Identification of Variants in the 4q35 GeneFAT1in Patients with a Facioscapulohumeral Dystrophy-Like Phenotype. Human Mutation, 2015, 36, 443-453.	2.5	38
151	Nationwide patient registry for GNE myopathy in Japan. Orphanet Journal of Rare Diseases, 2014, 9, 150.	2.7	37
152	DAG1 mutations associated with asymptomatic hyperCKemia and hypoglycosylation of Î±-dystroglycan. Neurology, 2015, 84, 273-279.	1.1	37
153	Dysferlin expression in tubular aggregates: their possible relationship to endoplasmic reticulum stress. Acta Neuropathologica, 2003, 105, 603-609.	7.7	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. Physiological Genomics, 2008, 35, 106-115.	2.3	36
155	Positive association betweenSTAT4polymorphisms and polymyositis/dermatomyositis in a Japanese population. Annals of the Rheumatic Diseases, 2012, 71, 1646-1650.	0.9	36
156	Prednisolone improves walking in Japanese Duchenne muscular dystrophy patients. Journal of Neurology, 2013, 260, 3023-3029.	3.6	36
157	Severe nemaline myopathy caused by mutations of the stop codon of the skeletal muscle alpha actin gene (ACTA1). Neuromuscular Disorders, 2006, 16, 541-547.	0.6	35
158	Alternative splicing of myomesin 1 gene is aberrantly regulated in myotonic dystrophy type 1. Genes To Cells, 2011, 16, 961-972.	1.2	35
159	Muscle glycogen storage disease 0 presenting recurrent syncope with weakness and myalgia. Neuromuscular Disorders, 2012, 22, 162-165.	0.6	35
160	Phenotypic stratification and genotypeâ€“phenotype correlation in a heterogeneous, international cohort of GNE myopathy patients: First report from the GNE myopathy Disease Monitoring Program, registry portion. Neuromuscular Disorders, 2018, 28, 158-168.	0.6	35
161	Molecular pathomechanism of distal myopathy with rimmed vacuoles. Acta Myologica, 2005, 24, 80-3.	1.5	35
162	Mutations of calpain 3 gene in patients with sporadic limb-girdle muscular dystrophy in Japan. Journal of the Neurological Sciences, 1999, 171, 31-37.	0.6	34

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163	Very low penetrance in 85 Japanese families with facioscapulohumeral muscular dystrophy 1A. Journal of Medical Genetics, 2004, 41, 12e-12.	3.2	34
164	Dysferlin mutation analysis in a group of Italian patients with limb-girdle muscular dystrophy and Miyoshi myopathy. European Journal of Neurology, 2004, 11, 657-661.	3.3	34
165	Humanin expression in skeletal muscles of patients with chronic progressive external ophthalmoplegia. Journal of Human Genetics, 2006, 51, 555-558.	2.3	34
166	GNE myopathy: A prospective natural history study of disease progression. Neuromuscular Disorders, 2014, 24, 380-386.	0.6	34
167	Isolated inclusion body myopathy caused by a multisystem proteinopathyâ€“linked hnRNPA1 mutation. Neurology: Genetics, 2015, 1, e23.	1.9	34
168	Dermatomyositis. Neurology, 2022, 98, .	1.1	34
169	Characterization of Danon disease in a male patient and his affected mother. Neuromuscular Disorders, 2003, 13, 708-711.	0.6	33
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171	Sarcolemmopathy: Muscular Dystrophies with Cell Membrane Defects. Brain Pathology, 2001, 11, 218-230.	4.1	33
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