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	A review of major causative genes in congenital myopathies. Journal of Human Genetics, 2023, 68, 215-225.	1.1	10
2	Anti-nuclear matrix protein 2 antibody-positive inflammatory myopathies represent extensive myositis without dermatomyositis-specific rash. Rheumatology, 2022, 61, 1222-1227.	0.9	32
3	Successful treatment of congenital myasthenic syndrome caused by a novel compound heterozygous variant in RAPSN. Brain and Development, 2022, 44, 50-55.	0.6	4
4	Visualizing Muscle Sialic Acid Expression in the GNED207VTgGne-/- Cmah-/- Model of GNE Myopathy: A Comparison of Dietary and Gene Therapy Approaches. Journal of Neuromuscular Diseases, 2022, 9, 53-71.	1.1	6
5	Deep convolutional neural network-based algorithm for muscle biopsy diagnosis. Laboratory Investigation, 2022, 102, 220-226.	1.7	6
6	DA-Raf and the MEK inhibitor trametinib reverse skeletal myocyte differentiation inhibition or muscle atrophy caused by myostatin and GDF11 through the non-Smad Rasâ€“ERK pathway. Journal of Biochemistry, 2022, 171, 109-122.	0.9	5
7	TRAPPC11â€related muscular dystrophy with hypoglycosylation of alphaâ€dystroglycan in skeletal muscle and brain. Neuropathology and Applied Neurobiology, 2022, 48, .	1.8	13
8	Case of antiâ€nuclear matrix protein 2 antibodyâ€positive juvenile dermatomyositis preceded by linear cutaneous lupus erythematosus on the face. Journal of Dermatology, 2022, 49, e18.	0.6	0
9	Assessment of thrombocytopenia, sleep apnea, and cardiac involvement in <scp>GNE</scp> myopathy patients. Muscle and Nerve, 2022, 65, 284-290.	1.0	8
10	A Japanese Patient with Hereditary Myopathy with Early Respiratory Failure Due to the p.P31732L Mutation of Titin. Internal Medicine, 2022, 61, 1587-1592.	0.3	2
11	Maximal Multistage Shuttle Run Test-induced Myalgia in a Patient with Muscle Phosphorylase B Kinase Deficiency. Internal Medicine, 2022, , .	0.3	1
12	Extra-muscular manifestations in GNE myopathy patients: A nationwide repository questionnaire survey in Japan. Clinical Neurology and Neurosurgery, 2022, 212, 107057.	0.6	7
13	Nemaline Myopathy Initially Diagnosed as Right Heart Failure with Type 2 Respiratory Failure. Internal Medicine, 2022, 61, 1897-1901.	0.3	1
14	Imagingâ€based evaluation of pathogenicity by novel DNM2 variants associated with centronuclear myopathy. Human Mutation, 2022, 43, 169-179.	1.1	4
15	A case of delayed diagnosis of Becker muscular dystrophy due to underlying developmental disorders. Brain and Development, 2022, 44, 259-262.	0.6	1
16	TNNI1 Mutated in Autosomal Dominant Proximal Arthrogryposis. Neurology: Genetics, 2022, 8, e649.	0.9	0
17	Identification of a novel mutation and genotypeâ€“phenotype relationship in MEGF10 myopathy. Neuromuscular Disorders, 2022, 32, 436-440.	0.3	1
18	Mutation spectrum of primary lipid storage myopathies. Annals of Indian Academy of Neurology, 2022, 25, 106.	0.2	7

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19	Resection of Gastric Cancer Remitted Anti-signal Recognition Particle Myopathy. Internal Medicine, 2022, 61, 2509-2515.	0.3	2
20	Advances in understanding of the natural history, mechanism, extraâ€muscular manifestations and treatment of GNE myopathy. Neurology and Clinical Neuroscience, 2022, 10, 289-297.	0.2	0
21	Tulobuterol is a potential therapeutic drug in congenital myasthenic syndrome. Pediatrics International, 2022, 64, e15115.	0.2	2
22	Frontal lobe-dominant cerebral blood flow reduction and atrophy can be progressive in Duchenne muscular dystrophy. Neuromuscular Disorders, 2022, 32, 477-485.	0.3	4
23	Myoglobinopathy affecting facial and oropharyngeal muscles. Neuromuscular Disorders, 2022, , .	0.3	0
24	Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes. Journal of Neurology, 2022, 269, 4129-4140.	1.8	2
25	Intranuclear inclusions in skin biopsies are not limited to neuronal intranuclear inclusion disease but can also be seen in oculopharyngodistal myopathy. Neuropathology and Applied Neurobiology, 2022, 48, .	1.8	14
26	Reliability of antinuclear matrix protein 2 antibody assays in idiopathic inflammatory myopathies is dependent on target protein properties. Journal of Dermatology, 2022, 49, 441-447.	0.6	3
27	Dermatomyositis. Neurology, 2022, 98, .	1.5	34
28	Muscle biochemical and pathological diagnosis in Pompe disease. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 1142-1145.	0.9	0
29	Sudden cardiac death prevention in an Emeryâ€“Dreifuss muscular dystrophy patient. Pediatrics International, 2022, 64, e15204.	0.2	0
30	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. Nature Communications, 2022, 13, 2306.	5.8	20
31	A 78-year-old Japanese male with late-onset PHKA1-associated distal myopathy: Case report and literature review. Neuromuscular Disorders, 2022, , .	0.3	1
32	A 7â€yearâ€old female with hypotonia and scoliosis. Brain Pathology, 2022, 32, .	2.1	2
33	Determining neurodevelopmental manifestations in Duchenne muscular dystrophy using a battery of brief tests. Journal of the Neurological Sciences, 2022, 440, 120340.	0.3	2
34	Hyperglycemic Crisis in Patients With Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like Episodes (MELAS). Pediatric Neurology, 2021, 114, 1-4.	1.0	2
35	Cricopharyngeal bar on videofluoroscopy: high specificity for inclusion body myositis. Journal of Neurology, 2021, 268, 1016-1024.	1.8	9
36	A case of eosinophilic fasciitis without skin manifestations: a case report in a patient with lupus and literature review. Clinical Rheumatology, 2021, 40, 2477-2483.	1.0	2

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37	Pathologic Features of Anti-Mi-2 Dermatomyositis. Neurology, 2021, 96, e448-e459.	1.5	24
38	Betaâ€propeller proteinâ€“associated neurodegeneration presenting Rettâ€like features: A case report and literature review. American Journal of Medical Genetics, Part A, 2021, 185, 579-583.	0.7	3
39	The wide-ranging clinical and genetic features in Japanese families with valosin-containing protein proteinopathy. Neurobiology of Aging, 2021, 100, 120.e1-120.e6.	1.5	8
40	Clinicopathological findings of a mitochondrial encephalopathy, lactic acidosis, and strokeâ€like episodes/Leigh syndrome overlap patient with a novel m. 3482A>G mutation in MTâ€ND1. Neuropathology, 2021, 41, 84-90.	0.7	4
41	A case of sporadic lateâ€onset nemaline myopathy without monoclonal gammopathy of unknown significance/human immunodeficiency virus successfully treated with intravenous gamma globulin. Clinical and Experimental Neuroimmunology, 2021, 12, 124-126.	0.5	1
42	<scp>COVID</scp>â€19â€associated myositis may be dermatomyositis. Muscle and Nerve, 2021, 63, E9-E10.	1.0	23
43	Mutant BIN1-Dynamin 2 complexes dysregulate membrane remodeling in the pathogenesis of centronuclear myopathy. Journal of Biological Chemistry, 2021, 296, 100077.	1.6	21
44	Monoclonal gammopathy of renal significance (MGRS)-related AL amyloidosis complicated by amyloid myopathy: a case report. BMC Nephrology, 2021, 22, 74.	0.8	3
45	Efficacy and Safety of Bimagrumab in Sporadic Inclusion Body Myositis. Neurology, 2021, 96, e1595-e1607.	1.5	25
46	A novel deletion in the C-terminal region of HSPB8 in a family with rimmed vacuolar myopathy. Journal of Human Genetics, 2021, 66, 965-972.	1.1	6
47	More prominent fibrosis of the cricopharyngeal muscle in inclusion body myositis. Journal of the Neurological Sciences, 2021, 422, 117327.	0.3	3
48	Megaconial congenital muscular dystrophy secondary to novel CHKB mutations resemble atypical Rett syndrome. Journal of Human Genetics, 2021, 66, 813-823.	1.1	9
49	Antimitochondrial Antibody-associated Myopathy with Slowly Progressive Cardiac Dysfunction. Internal Medicine, 2021, 60, 1035-1041.	0.3	5
50	Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2021, 144, 1451-1466.	3.7	28
51	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	2.6	15
52	Inflammatory features in sporadic lateâ€onset nemaline myopathy are independent from monoclonal gammopathy. Brain Pathology, 2021, 31, e12962.	2.1	15
53	Causative variant profile of collagen VI-related dystrophy in Japan. Orphanet Journal of Rare Diseases, 2021, 16, 284.	1.2	6
54	An autopsied case of ADSSL1 myopathy. Neuromuscular Disorders, 2021, 31, 1220-1225.	0.3	3

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55	A Japanese case of oculopharyngeal muscular dystrophy (OPMD) with PABPN1 c.35Gâ€‰>â€‰C; p.Gly12Ala point mutation. BMC Neurology, 2021, 21, 265.	0.8	7
56	A novel RyR1-selective inhibitor prevents and rescues sudden death in mouse models of malignant hyperthermia and heat stroke. Nature Communications, 2021, 12, 4293.	5.8	26
57	Mild form of Danon disease: two case reports. Neuromuscular Disorders, 2021, 31, 1207-1211.	0.3	1
58	A symptomatic male carrier of Duchenne muscular dystrophy with Klinefelter's syndrome mimicking Becker muscular dystrophy. Neuromuscular Disorders, 2021, 31, 666-672.	0.3	0
59	Noteworthy Cardiovascular Involvement with Sporadic Late-onset Nemaline Myopathy. Internal Medicine, 2021, 60, 2327-2332.	0.3	1
60	Clinicopathologic Features of Oculopharyngodistal Myopathy WithLRP12CGG Repeat Expansions Compared With Other Oculopharyngodistal Myopathy Subtypes. JAMA Neurology, 2021, 78, 853.	4.5	30
61	Successful treatment of a novel type I interferonopathy due to a de novo PSMB9 gene mutation with a Janus kinase inhibitor. Journal of Allergy and Clinical Immunology, 2021, 148, 639-644.	1.5	23
62	Marked Respiratory Failure in an Ambulant Patient with Immune-mediated Necrotizing Myopathy and Anti-Kv1.4 and Anti-titin Antibodies. Internal Medicine, 2021, 60, 2671-2675.	0.3	1
63	Underlying diseases in sporadic presentation of high creatine kinase levels in girls. Clinica Chimica Acta, 2021, 519, 198-203.	0.5	2
64	Neuropathy/intranuclear inclusion bodies in oculopharyngodistal myopathy: A case report. ENeurologicalSci, 2021, 24, 100348.	0.5	4
65	A review of core myopathy: central core disease, multiminicore disease, dusty core disease, and core-rod myopathy. Neuromuscular Disorders, 2021, 31, 968-977.	0.3	13
66	A recurrent homozygous ACTN2 variant associated with core myopathy. Acta Neuropathologica, 2021, 142, 785-788.	3.9	10
67	FKRP mutations cause congenital muscular dystrophy 1C and limb-girdle muscular dystrophy 2I in Asian patients. Journal of Clinical Neuroscience, 2021, 92, 215-221.	0.8	2
68	Series: Diagnosis at a Glance. The Journal of the Japanese Society of Internal Medicine, 2021, 110, 315-318.	0.0	0
69	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
70	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
71	Life-threatening muscle complications of COL4A1-related disorder. Brain and Development, 2020, 42, 93-97.	0.6	3
72	239th ENMC International Workshop: Classification of dermatomyositis, Amsterdam, the Netherlands, 14â€“16 December 2018. Neuromuscular Disorders, 2020, 30, 70-92.	0.3	148

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73	Antiâ€HMGCR myopathy following acute Epsteinâ€“Barr virus infection. Muscle and Nerve, 2020, 61, E5-E8.	1.0	7
74	Thigh muscle MRI findings in myopathy associated with antiâ€mitochondrial antibody. Muscle and Nerve, 2020, 61, 81-87.	1.0	13
75	Anti-RNP antibodies delineate a subgroup of myositis: A systematic retrospective study on 46 patients. Autoimmunity Reviews, 2020, 19, 102465.	2.5	14
76	Exon skipping induced by nonsense/frameshift mutations in DMD gene results in Becker muscular dystrophy. Human Genetics, 2020, 139, 247-255.	1.8	23
77	â€œBoule du bicepsâ€in dysferlinopathy. Neurology, 2020, 94, 83-84.	1.5	1
78	Mutations in the J domain of DNAJB6 cause dominant distal myopathy. Neuromuscular Disorders, 2020, 30, 38-46.	0.3	20
79	Needle electromyography, muscle MRI, and muscle pathology: Correlations in idiopathic inflammatory myopathies. Neurology and Clinical Neuroscience, 2020, 8, 28-35.	0.2	1
80	A novel AIFM1 missense mutation in a Japanese patient with ataxic sensory neuronopathy and hearing impairment. Journal of the Neurological Sciences, 2020, 409, 116584.	0.3	8
81	Pregnancy in GNE myopathy patients: a nationwide repository survey in Japan. Orphanet Journal of Rare Diseases, 2020, 15, 245.	1.2	7
82	Respiratory Dysfunction in Becker Muscular Dystrophy Patients: A Case Series and Autopsy Report. Journal of Neuromuscular Diseases, 2020, 7, 425-431.	1.1	1
83	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. American Journal of Human Genetics, 2020, 107, 1078-1095.	2.6	24
84	Tonsillectomy Improved Therapeutic Response in Anti-SRP Myopathy With Chronic Tonsillitis. Frontiers in Immunology, 2020, 11, 595480.	2.2	0
85	An adult nemaline myopathy patient with respiratory and heart failure harboring a novel NEB variant. ENeurologicalSci, 2020, 21, 100268.	0.5	2
86	CGG expansion in NOTCH2NLC is associated with oculopharyngodistal myopathy with neurological manifestations. Acta Neuropathologica Communications, 2020, 8, 204.	2.4	76
87	Selective involvement of semitendinosus in hereditary myopathy with early respiratory failure. Neurology and Clinical Neuroscience, 2020, 8, 428-429.	0.2	0
88	Limb-girdle muscular dystrophy. , 2020, , 437-449.		0
89	Where are we moving in the classification of idiopathic inflammatory myopathies?. Current Opinion in Neurology, 2020, 33, 590-603.	1.8	68
90	Treatment experience of Taiwanese patients with antiâ€3â€hydroxyâ€3â€methylglutarylâ€coenzyme A reductase myopathy. Kaohsiung Journal of Medical Sciences, 2020, 36, 649-655.	0.8	5

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91	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. PLoS ONE, 2020, 15, e0237890.	1.1	7
92	Lightâ€chain amyloid myopathy isolated to skeletal muscles: A case report. Clinical Case Reports (discontinued), 2020, 8, 2869-2873.	0.2	1
93	A case of type 1 facioscapulohumeral muscular dystrophy (FSHD) with restrictive ventilatory defect and congestive heart failure. ENeurologicalSci, 2020, 21, 100284.	0.5	0
94	Evaluation of the Core Formation Process in Congenital Neuromuscular Disease With Uniform Type 1 Fiber and Central Core Disease. Journal of Neuropathology and Experimental Neurology, 2020, 79, 1370-1375.	0.9	4
95	Paramyotonia Congenita with Persistent Distal and Facial Muscle Weakness: A Case Report with Literature Review. Journal of Neuromuscular Diseases, 2020, 7, 193-201.	1.1	5
96	TTN missense variants in two siblings with asymmetric facial and limb weakness. Journal of the Neurological Sciences, 2020, 415, 116885.	0.3	3
97	Expansion of GGC Repeat in GIPC1 Is Associated with Oculopharyngodistal Myopathy. American Journal of Human Genetics, 2020, 106, 793-804.	2.6	90
98	Homozygous nonsense variant in LRIF1 associated with facioscapulohumeral muscular dystrophy. Neurology, 2020, 94, e2441-e2447.	1.5	84
99	Neutral Lipid Storage Disease Associated with the <i>PNPLA2</i> Gene: Case Report and Literature Review. European Neurology, 2020, 83, 317-322.	0.6	4
100	Clinical, imaging, morphologic, and molecular features of X-linked VMA21-related myopathy in two unrelated Brazilian families. Journal of the Neurological Sciences, 2020, 415, 116977.	0.3	2
101	Severe cardiac involvement with preserved truncated dystrophin expression in Becker muscular dystrophy by +1G>A DMD splice-site mutation: a case report. Journal of Human Genetics, 2020, 65, 903-909.	1.1	3
102	Late-onset MELAS syndrome with mtDNA 14453Gâ†'A mutation masquerading as an acute encephalitis: a case report. BMC Neurology, 2020, 20, 247.	0.8	13
103	Two Japanese LGMDR25 patients with a biallelic recurrent nonsense variant of BVES. Neuromuscular Disorders, 2020, 30, 674-679.	0.3	18
104	Obstruction-related dysphagia in inclusion body myositis: Cricopharyngeal bar on videofluoroscopy indicates risk of aspiration. Journal of the Neurological Sciences, 2020, 413, 116764.	0.3	14
105	Clinical, pathological, imaging, and genetic characterization in a Taiwanese cohort with limb-girdle muscular dystrophy. Orphanet Journal of Rare Diseases, 2020, 15, 160.	1.2	13
106	Collagen â...¥â€related myopathy with subacute presentation of hypercapnic respiratory failure following pneumonia. Neurology and Clinical Neuroscience, 2020, 8, 320-322.	0.2	0
107	ADSSL1 myopathy is the most common nemaline myopathy in Japan with variable clinical features. Neurology, 2020, 95, e1500-e1511.	1.5	19
108	Clinical practice guidance for juvenile dermatomyositis (JDM) 2018-Update. Modern Rheumatology, 2020, 30, 411-423.	0.9	31

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109	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
110	Secondary cardiac involvement in anti-SRP-antibody-positive myopathy: an 87-year-old woman with heart failure symptoms as the first clinical presentation. BMC Neurology, 2020, 20, 29.	0.8	9
111	Duchenne muscular dystrophyâ€“like phenotype in an LGMD2I patient with novel FKRP gene variants. Human Genome Variation, 2020, 7, 12.	0.4	4
112	Association of Dermatomyositis Sine Dermatitis With Antiâ€“Nuclear Matrix Protein 2 Autoantibodies. JAMA Neurology, 2020, 77, 872.	4.5	39
113	Alternative splicing of clathrin heavy chain contributes to the switch from coated pits to plaques. Journal of Cell Biology, 2020, 219, .	2.3	31
114	Lysosomal membrane disorders: lysosome-associated membrane protein-2 deficiency (Danon disease). , 2020, , 567-574.		1
115	Adult-onset Repeat Rhabdomyolysis with a Very Long-chain Acyl-CoA Dehydrogenase Deficiency Due to Compound Heterozygous <i>ACADVL</i> Mutations. Internal Medicine, 2020, 59, 2729-2732.	0.3	4
116	Series: Diagnosis at a Glance. The Journal of the Japanese Society of Internal Medicine, 2020, 109, 2411-2413.	0.0	0
117	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
118	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
119	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
120	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
121	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
122	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis. , 2020, 15, e0237890.		0
123	Anti-signal Recognition Particle Antibody-positive Necrotizing Myopathy with Secondary Cardiomyopathy: The First Myocardial Biopsy- and Multimodal Imaging-proven Case. Internal Medicine, 2019, 58, 3189-3194.	0.3	12
124	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.	4.9	91
125	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. Neuromuscular Disorders, 2019, 29, 644-650.	0.3	2
126	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. Nature Genetics, 2019, 51, 1222-1232.	9.4	265

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127	Novel TTN mutations and muscle imaging characteristics in congenital titinopathy. Annals of Clinical and Translational Neurology, 2019, 6, 1311-1318.	1.7	16
128	The updated retrospective questionnaire study of sporadic inclusion body myositis in Japan. Orphanet Journal of Rare Diseases, 2019, 14, 155.	1.2	12
129	Psychiatric and neurodevelopmental aspects of Becker muscular dystrophy. Neuromuscular Disorders, 2019, 29, 930-939.	0.3	28
130	Deep morphological analysis of muscle biopsies from type III glycogenesis (GSDIII), debranching enzyme deficiency, revealed stereotyped vacuolar myopathy and autophagy impairment. Acta Neuropathologica Communications, 2019, 7, 167.	2.4	17
131	Dropped Head Syndrome Caused by Immune-mediated Necrotizing Myopathy. Internal Medicine, 2019, 58, 3343-3344.	0.3	1
132	Physician-Level Aggregated Classifier for Genetic Muscle Disorders. , 2019, , .		2
133	GNE genotype explains 20% of phenotypic variability in GNE myopathy. Neurology: Genetics, 2019, 5, e308.	0.9	22
134	Pembrolizumab-induced Ocular Myasthenia Gravis with Anti-titin Antibody and Necrotizing Myopathy. Internal Medicine, 2019, 58, 1635-1638.	0.3	26
135	Concurrent positive anti-3-hydroxy-3-methylglutaryl-coenzyme a reductase antibody with reducing body myopathy: Possible double trouble. Neuromuscular Disorders, 2019, 29, 543-548.	0.3	14
136	Two closely spaced mutations in cis result in Ullrich congenital muscular dystrophy. Human Genome Variation, 2019, 6, 21.	0.4	3
137	A novel compound heterozygous variant of ECHS1 identified in a Japanese patient with Leigh syndrome. Human Genome Variation, 2019, 6, 19.	0.4	13
138	COX6A2 variants cause a muscleâ€specific cytochrome c oxidase deficiency. Annals of Neurology, 2019, 86, 193-202.	2.8	21
139	An autopsy case of peliosis hepatis with X-linked myotubular myopathy. Legal Medicine, 2019, 38, 77-82.	0.6	13
140	Chronic sarcoid myopathy mimicking sporadic inclusion body myositis. Clinical Neurology and Neurosurgery, 2019, 182, 84-86.	0.6	6
141	Phenotype of a limb-girdle congenital myasthenic syndrome patient carrying a GFPT1 mutation. Brain and Development, 2019, 41, 470-473.	0.6	7
142	Dropped Head in Sporadic Late-onset Nemaline Myopathy. Internal Medicine, 2019, 58, 1967-1968.	0.3	5
143	Three novel MTM1 pathogenic variants identified in Japanese patients with Xâ€linked myotubular myopathy. Molecular Genetics & Genomic Medicine, 2019, 7, e621.	0.6	4
144	Inflammatory myopathy associated with PD-1 inhibitors. Journal of Autoimmunity, 2019, 100, 105-113.	3.0	73

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145	237th ENMC International Workshop: GNE myopathy â€“ current and future research Hoofddorp, The Netherlands, 14â€“16 September 2018. Neuromuscular Disorders, 2019, 29, 401-410.	0.3	5
146	Paraspinal amyotrophy in DNM-2-related centronuclear myopathy. Journal of the Neurological Sciences, 2019, 407, 116537.	0.3	0
147	Classification of idiopathic inflammatory myopathies: pathology perspectives. Current Opinion in Neurology, 2019, 32, 704-714.	1.8	61
148	RNA sequencing solved the most common but unrecognized NEB pathogenic variant in Japanese nemaline myopathy. Genetics in Medicine, 2019, 21, 1629-1638.	1.1	31
149	GNE myopathy in Chinese population: hotspot and novel mutations. Journal of Human Genetics, 2019, 64, 11-16.	1.1	18
150	Emeryâ€Dreifuss muscular dystrophyâ€related myopathy with TMEM43 mutations. Muscle and Nerve, 2019, 59, E5-E7.	1.0	7
151	Mitophagy in three cases of immune-mediated necrotizing myopathy associated with anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase autoantibodies: ultrastructural and immunohistochemical studies. Neuromuscular Disorders, 2018, 28, 283-288.	0.3	7
152	Small-Vessel Vasculopathy Due to Aberrant Autophagy in LAMP-2 Deficiency. Scientific Reports, 2018, 8, 3326.	1.6	24
153	Quantification of lectin fluorescence in GNE myopathy muscle biopsies. Muscle and Nerve, 2018, 58, 286-292.	1.0	13
154	Genetic and functional analysis of the RYR1 mutation p.Thr84Met revealed a susceptibility to malignant hyperthermia. Journal of Anesthesia, 2018, 32, 174-181.	0.7	7
155	A 62â€Yearâ€Old Woman with A History of Muscle Pain and Skin Rash for 1 Month. Brain Pathology, 2018, 28, 121-122.	2.1	0
156	A 31â€Yearâ€Old Man with Slowly Progressive Limb Muscle Weakness and Respiratory Insufficiency. Brain Pathology, 2018, 28, 123-124.	2.1	1
157	GNE myopathy caused by a synonymous mutation leading to aberrant mRNA splicing. Neuromuscular Disorders, 2018, 28, 154-157.	0.3	9
158	New Criteria Needed for Antisynthetase Syndromeâ€”Reply. JAMA Neurology, 2018, 75, 259.	4.5	1
159	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.	0.3	35
160	Case of elderly-onset multiple acyl-CoA dehydrogenase deficiency with a novel ETFDH mutation shows progressive muscle weakness and rhabdomyolysis. Neurology and Clinical Neuroscience, 2018, 6, 39-41.	0.2	0
161	Transient swelling in the globus pallidus and substantia nigra in childhood suggests SENDA/BPAN. Neurology, 2018, 90, 974-976.	1.5	10
162	A Homozygous LAMA2 Mutation of c.818G>A Caused Partial Merosin Deficiency in a Japanese Patient. Internal Medicine, 2018, 57, 877-882.	0.3	6

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163	A mild myopathy with anti-SRP plus anti-PL-12 antibodies successfully treated by oral steroid monotherapy. Journal of the Neurological Sciences, 2018, 388, 7-9.	0.3	1
164	Impact of muscle biopsy on diagnosis and management of children with neuromuscular diseases: A 10-year retrospective critical review. Journal of Pediatric Surgery, 2018, 53, 489-492.	0.8	12
165	Social involvement issues in patients with Becker muscular dystrophy: A questionnaire survey of subjects from a patient registry. Brain and Development, 2018, 40, 268-277.	0.6	12
166	Reduced Dnmt3a increases Gdf5 expression with suppressed satellite cell differentiation and impaired skeletal muscle regeneration. FASEB Journal, 2018, 32, 1452-1467.	0.2	26
167	Efficacy of Prednisolone in Generated Myotubes Derived From Fibroblasts of Duchenne Muscular Dystrophy Patients. Frontiers in Pharmacology, 2018, 9, 1402.	1.6	9
168	A Nationwide Survey on Danon Disease in Japan. International Journal of Molecular Sciences, 2018, 19, 3507.	1.8	27
169	Three novel recessive DYSF mutations identified in three patients with muscular dystrophy, limb-girdle, type 2B. Journal of the Neurological Sciences, 2018, 395, 169-171.	0.3	6
170	Different clinicopathological features between Japanese siblings with facioscapulohumeral muscular dystrophy 2 with a novel nonsense SMCHD1 mutation (Arg552â^—). Journal of Clinical Neuroscience, 2018, 58, 215-217.	0.8	1
171	Phase I clinical trial results of aceneuramic acid for GNE myopathy in Japan. Translational Medicine Communications, 2018, 3, .	0.5	5
172	Congenital myopathy with fiber-type disproportion accompanied by dilated cardiomyopathy in a patient with a novel p.G48A ACTA1 mutation. Journal of the Neurological Sciences, 2018, 393, 142-144.	0.3	10
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