John Vissing

List of Publications by Year in descending order

401 papers	13,895 citations	20817 60 h-index	39675 94 g-index
413 all docs	413 docs citations	413 times ranked	10878 citing authors

#	Article	IF	Citations
1	Paternal Inheritance of Mitochondrial DNA. New England Journal of Medicine, 2002, 347, 576-580.	27.0	595
2	Safety and efficacy of eculizumab in anti-acetylcholine receptor antibody-positive refractory generalised myasthenia gravis (REGAIN): a phase 3, randomised, double-blind, placebo-controlled, multicentre study. Lancet Neurology, The, 2017, 16, 976-986.	10.2	472
3	Recombination of Human Mitochondrial DNA. Science, 2004, 304, 981-981.	12.6	253
4	The spectrum of exercise tolerance in mitochondrial myopathies: a study of 40 patients. Brain, 2003, 126, 413-423.	7.6	226
5	The Effect of Oral Sucrose on Exercise Tolerance in Patients with McArdle's Disease. New England Journal of Medicine, 2003, 349, 2503-2509.	27.0	215
6	High prevalence and phenotypeÃ¢Â€Â“genotype correlations of limb girdle muscular dystrophy type 2I in Denmark. Annals of Neurology, 2006, 59, 808-815.	5.3	201
7	McArdle disease: a clinical review. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 1182-1188.	1.9	197
8	Safety, efficacy, and tolerability of efgartigimod in patients with generalised myasthenia gravis (ADAPT): a multicentre, randomised, placebo-controlled, phase 3 trial. Lancet Neurology, The, 2021, 20, 526-536.	10.2	194
9	Aerobic training is safe and improves exercise capacity in patients with mitochondrial myopathy. Brain, 2006, 129, 3402-3412.	7.6	184
10	A nonischemic forearm exercise test for McArdle disease. Annals of Neurology, 2002, 52, 153-159.	5.3	163
11	Identification and Characterization of a Common Set of Complex I Assembly Intermediates in Mitochondria from Patients with Complex I Deficiency. Journal of Biological Chemistry, 2003, 278, 43081-43088.	3.4	163
12	Mitochondrial encephalomyopathy with elevated methylmalonic acid is caused by SUCLA2 mutations. Brain, 2007, 130, 853-861.	7.6	162
13	Longâ€term safety and efficacy of eculizumab in generalized myasthenia gravis. Muscle and Nerve, 2019, 60, 14-24.	2.2	162
14	Quantitative Muscle MRI as an Assessment Tool for Monitoring Disease Progression in LGMD2I: A Multicentre Longitudinal Study. PLoS ONE, 2013, 8, e70993.	2.5	148
15	Aerobic conditioning: An effective therapy in McArdle's disease. Annals of Neurology, 2006, 59, 922-928.	5.3	146
16	Cardiac manifestations of myotonic dystrophy type 1. International Journal of Cardiology, 2012, 160, 82-88.	1.7	146
17	Novel POLG mutations in progressive external ophthalmoplegia mimicking mitochondrial neurogastrointestinal encephalomyopathy. European Journal of Human Genetics, 2003, 11, 547-549.	2.8	145
18	Exercise-Induced Changes in Local Cerebral Glucose Utilization in the Rat. Journal of Cerebral Blood Flow and Metabolism, 1996, 16, 729-736.	4.3	141

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19	Spontaneous "Second Wind" and Glucose-Induced Second "Second Wind" in McArdle Disease. Archives of Neurology, 2002, 59, 1395-402.	4.5	138
20	Aerobic training improves exercise performance in facioscapulohumeral muscular dystrophy. Neurology, 2005, 64, 1064-1066.	1.1	124
21	European consensus for starting and stopping enzyme replacement therapy in adult patients with Pompe disease: a 10â€year experience. European Journal of Neurology, 2017, 24, 768.	3.3	118
22	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. Human Mutation, 2012, 33, 949-959.	2.5	115
23	Guidance for the management of myasthenia gravis (MG) and Lambert-Eaton myasthenic syndrome (LEMS) during the COVID-19 pandemic. Journal of the Neurological Sciences, 2020, 412, 116803.	0.6	110
24	Aerobic training in patients with myotonic dystrophy type 1. Annals of Neurology, 2005, 57, 754-757.	5.3	102
25	Muscle Glycogenosis Due to Phosphoglucomutase 1 Deficiency. New England Journal of Medicine, 2009, 361, 425-427.	27.0	101
26	Endurance training improves fitness and strength in patients with Becker muscular dystrophy. Brain, 2008, 131, 2824-2831.	7.6	100
27	Loss-of-function mutations inSCN4Acause severe foetal hypokinesia or â€˜classicalâ€™ congenital myopathy. Brain, 2016, 139, 674-691.	7.6	100
28	Efficacy and Safety of Rozanolixizumab in Moderate to Severe Generalized Myasthenia Gravis. Neurology, 2021, 96, e853-e865.	1.1	97
29	Bezafibrate in skeletal muscle fatty acid oxidation disorders. Neurology, 2014, 82, 607-613.	1.1	96
30	A diagnostic cycle test for McArdle's disease. Annals of Neurology, 2003, 54, 539-542.	5.3	93
31	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
32	Open-Label Trial of Anti-TNF-Î± in Dermato- and Polymyositis Treated Concomitantly with Methotrexate. European Neurology, 2008, 59, 159-163.	1.4	92
33	Leg muscle involvement in facioscapulohumeral muscular dystrophy assessed by MRI. Journal of Neurology, 2006, 253, 1437-1441.	3.6	91
34	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
35	Tissue specific distribution of the 3243A->G mtDNA mutation. Journal of Medical Genetics, 2006, 43, 671-677.	3.2	87
36	A heterozygous 21-bp deletion inCAPN3causes dominantly inherited limb girdle muscular dystrophy. Brain, 2016, 139, 2154-2163.	7.6	87

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37	Treatment of Mitochondrial Neurogastrointestinal Encephalomyopathy With Dialysis. Archives of Neurology, 2007, 64, 435.	4.5	86
38	COVID-19-associated risks and effects in myasthenia gravis (CARE-MG). Lancet Neurology, The, 2020, 19, 970-971.	10.2	85
39	Fuel utilization in subjects with carnitine palmitoyltransferase 2 gene mutations. Annals of Neurology, 2005, 57, 60-66.	5.3	81
40	Severe paraspinal muscle involvement in facioscapulohumeral muscular dystrophy. Neurology, 2014, 83, 1178-1183.	1.1	81
41	Quantitative Magnetic Resonance Imaging in Limb-Girdle Muscular Dystrophy 2I: A Multinational Cross-Sectional Study. PLoS ONE, 2014, 9, e90377.	2.5	81
42	Endurance training: An effective and safe treatment for patients with LGMD2I. Neurology, 2007, 68, 59-61.	1.1	79
43	Fat Replacement of Paraspinal Muscles with Aging in Healthy Adults. Medicine and Science in Sports and Exercise, 2017, 49, 595-601.	0.4	78
44	Role of 5â€²AMPâ€activated protein kinase in glycogen synthase activity and glucose utilization: insights from patients with McArdle's disease. Journal of Physiology, 2002, 541, 979-989.	2.9	76
45	Effect of Oral Sucrose Shortly Before Exercise on Work Capacity in McArdle Disease. Archives of Neurology, 2008, 65, 786-9.	4.5	74
46	Lactate production and clearance in exercise. Effects of training. A miniâ€review. Scandinavian Journal of Medicine and Science in Sports, 1998, 8, 127-131.	2.9	73
47	Late-onset Pompe disease is prevalent in unclassified limb-girdle muscular dystrophies. Molecular Genetics and Metabolism, 2013, 110, 287-289.	1.1	73
48	MRI as outcome measure in facioscapulohumeral muscular dystrophy: 1-year follow-up of 45 patients. Journal of Neurology, 2017, 264, 438-447.	3.6	72
49	Multisystem disorder associated with a missense mutation in the mitochondrial cytochromeb gene. Annals of Neurology, 2001, 50, 540-543.	5.3	71
50	Oxidative capacity correlates with muscle mutation load in mitochondrial myopathy. Annals of Neurology, 2003, 54, 86-92.	5.3	71
51	Muscle Phenotype and Mutation Load in 51 Persons With the 3243A>G Mitochondrial DNA Mutation. Archives of Neurology, 2006, 63, 1701.	4.5	71
52	Bimagrumab vs Optimized Standard of Care for Treatment of Sarcopenia in Community-Dwelling Older Adults. JAMA Network Open, 2020, 3, e2020836.	5.9	71
53	Effect of diet on exercise tolerance in carnitine palmitoyltransferase II deficiency. Neurology, 2003, 61, 559-561.	1.1	69
54	Difference in allelic expression of the CLCN1 gene and the possible influence on the myotonia congenita phenotype. European Journal of Human Genetics, 2004, 12, 738-743.	2.8	69

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55	Limb girdle muscular dystrophies: classification, clinical spectrum and emerging therapies. Current Opinion in Neurology, 2016, 29, 635-641.	3.6	69
56	A new mitochondrial tRNA ^{Met} gene mutation in a patient with dystrophic muscle and exercise intolerance. Neurology, 1998, 50, 1875-1878.	1.1	67
57	No spontaneous second wind in muscle phosphofructokinase deficiency. Neurology, 2004, 62, 82-86.	1.1	64
58	Anoctamin 5 muscular dystrophy in Denmark: prevalence, genotypes, phenotypes, cardiac findings, and muscle protein expression. Journal of Neurology, 2013, 260, 2084-2093.	3.6	63
59	Effect of deficient muscular glycogenolysis on extramuscular fuel production in exercise. Journal of Applied Physiology, 1992, 72, 1773-1779.	2.5	62
60	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. Genetics in Medicine, 2020, 22, 1478-1488.	2.4	62
61	Cardiac Involvement in Patients With Limb-Girdle Muscular Dystrophy Type 2 and Becker Muscular Dystrophy. Archives of Neurology, 2008, 65, 1196-201.	4.5	61
62	Reduced levels of skeletal muscle Na ⁺ K ⁺ -ATPase in McArdle disease. Neurology, 1998, 50, 37-40.	1.1	60
63	LGMD2I presenting with a characteristic Duchenne or Becker muscular dystrophy phenotype. Neurology, 2005, 64, 1635-1637.	1.1	60
64	Sympathetic activation in exercise is not dependent on muscle acidosis. Direct evidence from studies in metabolic myopathies.. Journal of Clinical Investigation, 1998, 101, 1654-1660.	8.2	60
65	Carbohydrate- and protein-rich diets in McArdle disease: effects on exercise capacity. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 1359-1363.	1.9	59
66	Safety, tolerability, pharmacokinetics, pharmacodynamics, and exploratory efficacy of the novel enzyme replacement therapy avalglucosidase alfa (neoGAA) in treatment-naÃ¯ve and alglucosidase alfa-treated patients with late-onset Pompe disease: A phase 1, open-label, multicenter, multinational, ascending dose study. Neuromuscular Disorders, 2019, 29, 167-186.	0.6	59
67	Cardiac involvement in myotonic dystrophy: a nationwide cohort study. European Heart Journal, 2014, 35, 2158-2164.	2.2	56
68	Diagnosis of Pompe Disease. JAMA Neurology, 2013, 70, 923.	9.0	55
69	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	1.9	55
70	Fuel utilization in patients with very long-chain acyl-coa dehydrogenase deficiency. Annals of Neurology, 2004, 56, 279-283.	5.3	53
71	A forearm exercise screening test for mitochondrial myopathy. Neurology, 2002, 58, 1533-1538.	1.1	52
72	Clinical and molecular characterization of limbâ€girdle muscular dystrophy due to LAMA2 mutations. Muscle and Nerve, 2011, 44, 703-709.	2.2	52

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73	Exercise fuel mobilization in mitochondrial myopathy: A metabolic dilemma. Annals of Neurology, 1996, 40, 655-662.	5.3	51
74	Splice mutations preserve myophosphorylase activity that ameliorates the phenotype in McArdle disease. Brain, 2009, 132, 1545-1552.	7.6	51
75	Two- and 6-minute walk tests assess walking capability equally in neuromuscular diseases. Neurology, 2016, 86, 442-445.	1.1	51
76	Is muscle glycogenolysis impaired in X-linked phosphorylase b kinase deficiency?. Neurology, 2008, 70, 1876-1882.	1.1	50
77	Endocrine function in 97 patients with myotonic dystrophy type 1. Journal of Neurology, 2012, 259, 912-920.	3.6	50
78	Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. Annals of Clinical and Translational Neurology, 2014, 1, 88-98.	3.7	50
79	Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. Neurology, 2015, 84, 1772-1781.	1.1	50
80	Effect of aerobic training in patients with spinal and bulbar muscular atrophy (Kennedy disease). Neurology, 2009, 72, 317-323.	1.1	49
81	Calpain 3 is important for muscle regeneration: Evidence from patients with limb girdle muscular dystrophies. BMC Musculoskeletal Disorders, 2012, 13, 43.	1.9	49
82	Mutations inCOA3cause isolated complex IV deficiency associated with neuropathy, exercise intolerance, obesity, and short stature. Journal of Medical Genetics, 2015, 52, 203-207.	3.2	49
83	The antimyotonic effect of lamotrigine in non-dystrophic myotonias: a double-blind randomized study. Brain, 2017, 140, 2295-2305.	7.6	49
84	A decline in PABPN1 induces progressive muscle weakness in Oculopharyngeal muscle dystrophy and in muscle aging. Aging, 2013, 5, 412-426.	3.1	49
85	Fat metabolism during exercise in patients with McArdle disease. Neurology, 2009, 72, 718-724.	1.1	48
86	Aerobic training and postexercise protein in facioscapulohumeral muscular dystrophy. Neurology, 2015, 85, 396-403.	1.1	48
87	31P-MRS of skeletal muscle is not a sensitive diagnostic test for mitochondrial myopathy. Journal of Neurology, 2007, 254, 29-37.	3.6	47
88	The cytochrome b p.278Y>C mutation causative of a multisystem disorder enhances superoxide production and alters supramolecular interactions of respiratory chain complexes. Human Molecular Genetics, 2013, 22, 2141-2151.	2.9	46
89	MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscular Disorders, 2019, 29, 827-841.	0.6	46
90	MRI in Neuromuscular Diseases: An Emerging Diagnostic Tool and Biomarker for Prognosis and Efficacy. Annals of Neurology, 2020, 88, 669-681.	5.3	46

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91	New patterns of inheritance in mitochondrial disease. Biochemical and Biophysical Research Communications, 2003, 310, 247-251.	2.1	45
92	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	7.6	45
93	Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. Neurology, 2020, 94, e1094-e1102.	1.1	45
94	Shortâ€•and longâ€•term effects of endurance training in patients with mitochondrial myopathy. European Journal of Neurology, 2009, 16, 1336-1339.	3.3	44
95	High prevalence of cardiac involvement in patients with myotonic dystrophy type 1: A cross-sectional study. International Journal of Cardiology, 2014, 174, 31-36.	1.7	44
96	LAMA2â€•related myopathy: Frequency among congenital and limbâ€girdle muscular dystrophies. Muscle and Nerve, 2015, 52, 547-553.	2.2	44
97	Axial myopathy: an overlooked feature of muscle diseases. Brain, 2016, 139, 13-22.	7.6	44
98	Resistance training in patients with limbâ€girdle and becker muscular dystrophies. Muscle and Nerve, 2013, 47, 163-169.	2.2	43
99	Myocardial fibrosis in patients with myotonic dystrophy type 1: a cardiovascular magnetic resonance study. Journal of Cardiovascular Magnetic Resonance, 2014, 16, 59.	3.3	43
100	Training improves oxidative capacity, but not function, in spinal muscular atrophy type III. Muscle and Nerve, 2015, 52, 240-244.	2.2	43
101	Contractile properties are disrupted in Becker muscular dystrophy, but not in limb girdle type 2I. Annals of Neurology, 2016, 80, 466-471.	5.3	43
102	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	3.6	43
103	Clinical presentation and mutations in Danish patients with Wilson disease. European Journal of Human Genetics, 2011, 19, 935-941.	2.8	42
104	Oral branched-chain amino acids do not improve exercise capacity in McArdle disease. Neurology, 1998, 51, 1456-1459.	1.1	41
105	Muscle Phosphoglycerate Mutase Deficiency Revisited. Archives of Neurology, 2009, 66, 394-8.	4.5	40
106	Anti-gravity training improves walking capacity and postural balance in patients with muscular dystrophy. Neuromuscular Disorders, 2014, 24, 492-498.	0.6	40
107	Exercise tolerance in carnitine palmitoyltransferase II deficiency with IV and oral glucose. Neurology, 2002, 59, 1046-1051.	1.1	39
108	Muscle phosphorylase kinase deficiency. Neurology, 2012, 78, 265-268.	1.1	39

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109	Effect of sildenafil on skeletal and cardiac muscle in <scp>B</scp>ecker muscular dystrophy. Annals of Neurology, 2014, 76, 550-557.	5.3	39
110	The exercise metaboreflex is maintained in the absence of muscle acidosis: insights from muscle microdialysis in humans with McArdle's disease. Journal of Physiology, 2001, 537, 641-649.	2.9	38
111	Multiple mtDNA deletions with features of MNGIE. Neurology, 2002, 59, 926-929.	1.1	38
112	Exercise intolerance in Glycogen Storage Disease Type III: Weakness or energy deficiency?. Molecular Genetics and Metabolism, 2013, 109, 14-20.	1.1	38
113	Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 576-585.	1.9	38
114	Growth and differentiation factor 15 as a biomarker for mitochondrial myopathy. Mitochondrion, 2020, 50, 35-41.	3.4	38
115	Safety and efficacy of omaveloxolone in patients with mitochondrial myopathy. Neurology, 2020, 94, e687-e698.	1.1	38
116	Effect of Fuels on Exercise Capacity in Muscle Phosphoglycerate Mutase Deficiency. Archives of Neurology, 2005, 62, 1440.	4.5	37
117	Exercise intolerance in mitochondrial myopathy is not related to lactic acidosis. Annals of Neurology, 2001, 49, 672-676.	5.3	36
118	Muscle structural changes in mitochondrial myopathy relate to genotype. Journal of Neurology, 2003, 250, 1328-1334.	3.6	36
119	Progression of cardiac involvement in patients with limb-girdle type 2 and Becker muscular dystrophies: A 9-year follow-up study. International Journal of Cardiology, 2015, 182, 403-411.	1.7	36
120	Cycle ergometry is not a sensitive diagnostic test for mitochondrial myopathy. Journal of Neurology, 2003, 250, 293-299.	3.6	35
121	Physical training for McArdle disease. The Cochrane Library, 2011, , CD007931.	2.8	34
122	Phenotypes, genotypes, and prevalence of congenital myopathies older than 5 years in Denmark. Neurology: Genetics, 2017, 3, e140.	1.9	34
123	Paternal comeback in mitochondrial DNA inheritance. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 1475-1476.	7.1	34
124	Decreased insulin action in skeletal muscle from patients with McArdle's disease. American Journal of Physiology - Endocrinology and Metabolism, 2002, 282, E1267-E1275.	3.5	33
125	Decrement of compound muscle action potential is related to mutation type in myotonia congenita. Muscle and Nerve, 2003, 27, 449-455.	2.2	33
126	Late onset of stroke-like episode associated with a 3256Câ†'T point mutation of mitochondrial DNA. Journal of the Neurological Sciences, 2003, 214, 17-20.	0.6	33

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127	Relationship between muscle inflammation and fat replacement assessed by MRI in facioscapulohumeral muscular dystrophy. Journal of Neurology, 2019, 266, 1127-1135.	3.6	33
128	Endocrine function over time in patients with myotonic dystrophy type 1. European Journal of Neurology, 2015, 22, 116-122.	3.3	32
129	Zilucoplan: An Investigational Complement C5 Inhibitor for the Treatment of Acetylcholine Receptor Autoantibodyâ€“Positive Generalized Myasthenia Gravis. Expert Opinion on Investigational Drugs, 2021, 30, 483-493.	4.1	32
130	Prevalence of migraine in persons with the 3243A>G mutation in mitochondrial <scp>DNA</scp>. European Journal of Neurology, 2016, 23, 175-181.	3.3	31
131	PGM1 deficiency: Substrate use during exercise and effect of treatment with galactose. Neuromuscular Disorders, 2017, 27, 370-376.	0.6	31
132	Phenotype and genotype of muscle ryanodine receptor rhabdomyolysis-myalgia syndrome. Acta Neurologica Scandinavica, 2018, 137, 452-461.	2.1	31
133	MSTO1 mutations cause mtDNA depletion, manifesting as muscular dystrophy with cerebellar involvement. Acta Neuropathologica, 2019, 138, 1013-1031.	7.7	31
134	No evidence for paternal inheritance of mtDNA in patients with sporadic mtDNA mutations. Journal of the Neurological Sciences, 2004, 218, 99-101.	0.6	30
135	â€˜Minimal symptom expressionâ€™ in patients with acetylcholine receptor antibody-positive refractory generalized myasthenia gravis treated with eculizumab. Journal of Neurology, 2020, 267, 1991-2001.	3.6	30
136	Effect of liver glycogen content on glucose production in running rats. Journal of Applied Physiology, 1989, 66, 318-322.	2.5	29
137	Muscle phosphoglycerate mutase deficiency with tubular aggregates: Effect of dantrolene. Annals of Neurology, 1999, 46, 274-277.	5.3	29
138	Blocked Muscle Fat Oxidation During Exercise in Neutral Lipid Storage Disease. Archives of Neurology, 2012, 69, 530.	4.5	29
139	Aerobic training in persons who have recovered from juvenile dermatomyositis. Neuromuscular Disorders, 2013, 23, 962-968.	0.6	29
140	Muscle involvement in limb-girdle muscular dystrophy with GMPPB deficiency (LGMD2T). Neurology: Genetics, 2016, 2, e112.	1.9	29
141	cDNA analyses of CAPN3 enhance mutation detection and reveal a low prevalence of LGMD2A patients in Denmark. European Journal of Human Genetics, 2008, 16, 935-940.	2.8	28
142	Change in muscle strength over time in spinal muscular atrophy types II and III. A long-term follow-up study. Neuromuscular Disorders, 2012, 22, 1069-1074.	0.6	28
143	Safety, tolerability, and preliminary efficacy of an IGF-1 mimetic in patients with spinal and bulbar muscular atrophy: a randomised, placebo-controlled trial. Lancet Neurology, The, 2018, 17, 1043-1052.	10.2	28
144	Natural history of limb girdle muscular dystrophy R9 over 6Âyears: searching for trial endpoints. Annals of Clinical and Translational Neurology, 2019, 6, 1033-1045.	3.7	28

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145	Recurrent TTN metatranscriptâ€only c.39974â€“11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. Human Mutation, 2020, 41, 403-411.	2.5	28
146	Fat and carbohydrate metabolism during exercise in late-onset Pompe disease. Molecular Genetics and Metabolism, 2012, 107, 462-468.	1.1	27
147	<scp>EFNS</scp> review on the role of muscle biopsy in the investigation of myalgia. European Journal of Neurology, 2013, 20, 997-1005.	3.3	27
148	Pharmacologic Treatment of Downstream of Tyrosine Kinase 7 Congenital Myasthenic Syndrome. JAMA Neurology, 2014, 71, 350.	9.0	27
149	Fatigue in patients with spinal muscular atrophy type II and congenital myopathies: evaluation of the fatigue severity scale. Quality of Life Research, 2014, 23, 1479-1488.	3.1	27
150	Exercise in muscle glycogen storage diseases. Journal of Inherited Metabolic Disease, 2015, 38, 551-563.	3.6	27
151	Resistance training and aerobic training improve muscle strength and aerobic capacity in chronic inflammatory demyelinating polyneuropathy. Muscle and Nerve, 2018, 57, 70-76.	2.2	27
152	POPDC3 Gene Variants Associate with a New Form of Limb Girdle Muscular Dystrophy. Annals of Neurology, 2019, 86, 832-843.	5.3	27
153	Evaluation of inflammatory lesions over 2 years in facioscapulohumeral muscular dystrophy. Neurology, 2020, 95, e1211-e1221.	1.1	27
154	Nampt controls skeletal muscle development by maintaining Ca2+ homeostasis and mitochondrial integrity. Molecular Metabolism, 2021, 53, 101271.	6.5	27
155	Effect of Changes in Fat Availability on Exercise Capacity in McArdle Disease. Archives of Neurology, 2009, 66, 762-6.	4.5	26
156	Level of muscle regeneration in limb-girdle muscular dystrophy type 2I relates to genotype and clinical severity. Skeletal Muscle, 2011, 1, 31.	4.2	26
157	Fat and Carbohydrate Metabolism During Exercise in Phosphoglucomutase Type 1 Deficiency. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1235-E1240.	3.6	26
158	Skeletal muscle metabolism is impaired during exercise in glycogen storage disease type III. Neurology, 2015, 84, 1767-1771.	1.1	26
159	No effect of bezafibrate in patients with CPTII and VLCAD deficiencies. Journal of Inherited Metabolic Disease, 2015, 38, 373-374.	3.6	26
160	High-intensity interval training in facioscapulohumeral muscular dystrophy type 1: a randomized clinical trial. Journal of Neurology, 2017, 264, 1099-1106.	3.6	26
161	Disease progression and outcome measures in spinobulbar muscular atrophy. Annals of Neurology, 2018, 84, 754-765.	5.3	25
162	Impairments in contractility and cytoskeletal organisation cause nuclear defects in nemaline myopathy. Acta Neuropathologica, 2019, 138, 477-495.	7.7	25

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163	Patients With Medium-Chain Acylâ€“Coenzyme A Dehydrogenase Deficiency Have Impaired Oxidation of Fat During Exercise but No Effect of<scp>l</scp>-Carnitine Supplementation. Journal of Clinical Endocrinology and Metabolism, 2013, 98, 1667-1675.	3.6	24
164	Differential Muscle Involvement in Mice and Humans Affected by McArdle Disease. Journal of Neuropathology and Experimental Neurology, 2016, 75, 441-454.	1.7	24
165	Decreased Variability of the 6-Minute Walk Test by Heart Rate Correction in Patients with Neuromuscular Disease. PLoS ONE, 2014, 9, e114273.	2.5	24
166	Do carriers of PYGM mutations have symptoms of McArdle disease?. Neurology, 2006, 67, 716-718.	1.1	23
167	Lactate metabolism during exercise in patients with mitochondrial myopathy. Neuromuscular Disorders, 2013, 23, 629-636.	0.6	23
168	Creatine kinase response to highâ€intensity aerobic exercise in adultâ€onset muscular dystrophy. Muscle and Nerve, 2013, 48, 897-901.	2.2	23
169	Ocular, bulbar, limb, and cardiopulmonary involvement in oculopharyngeal muscular dystrophy. Acta Neurologica Scandinavica, 2014, 130, 125-130.	2.1	23
170	Update on new muscle glycogenosis. Current Opinion in Neurology, 2017, 30, 449-456.	3.6	23
171	Refining the spinobulbar muscular atrophy phenotype by quantitative MRI and clinical assessments. Neurology, 2019, 92, e548-e559.	1.1	23
172	Data from the European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC). Orphanet Journal of Rare Diseases, 2020, 15, 330.	2.7	23
173	Paradoxically enhanced glucose production during exercise in humans with blocked glycolysis caused by muscle phosphofructokinase deficiency. Neurology, 1996, 47, 766-771.	1.1	22
174	Recurrent myoglobinuria and deranged acylcarnitines due to a mutation in the mtDNA MT-CO2 gene. Neurology, 2013, 80, 1908-1910.	1.1	22
175	Muscle Atrophy Reversed by Growth Factor Activation of Satellite Cells in a Mouse Muscle Atrophy Model. PLoS ONE, 2014, 9, e100594.	2.5	22
176	Intact transferrin and total plasma glycoprofiling for diagnosis and therapy monitoring in phosphoglucomutase-I deficiency. Translational Research, 2018, 199, 62-76.	5.0	22
177	Aerobic training in patients with anoctamin 5 myopathy and hyperckemia. Muscle and Nerve, 2014, 50, 119-123.	2.2	21
178	Reliability of the 2- and 6-minute walk tests in neuromuscular diseases. Journal of Rehabilitation Medicine, 2017, 49, 362-366.	1.1	21
179	Characterization of two new dominant ClC-1 channel mutations associated with myotonia. Muscle and Nerve, 2003, 28, 722-732.	2.2	20
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