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	Habitual Physical Activity in Patients with Myasthenia Gravis Assessed by Accelerometry and Questionnaire. Journal of Neuromuscular Diseases, 2022, 9, 161-169.	2.6	4
2	Diagnostic interest of whole-body MRI in early- and late-onset LAMA2 muscular dystrophies: a large international cohort. Journal of Neurology, 2022, 269, 2414-2429.	3.6	5
3	Causes of symptom dissatisfaction in patients with generalized myasthenia gravis. Journal of Neurology, 2022, 269, 3086-3093.	3.6	6
4	Axial muscle involvement in patients with limb girdle muscular dystrophy type <scp>R9</scp>. Muscle and Nerve, 2022, 65, 405-414.	2.2	3
5	No effect of resveratrol on fatty acid oxidation or exercise capacity in patients with fatty acid oxidation disorders: A randomized clinical crossâ€over trial. Journal of Inherited Metabolic Disease, 2022, 45, 517-528.	3.6	7
6	Growth Factors Do Not Improve Muscle Function in Young or Adult mdx Mice. Biomedicines, 2022, 10, 304.	3.2	0
7	No effect of triheptanoin in patients with phosphofructokinase deficiency. Neuromuscular Disorders, 2022, , .	0.6	1
8	No effect of oral ketone ester supplementation on exercise capacity in patients with <scp>McArdle</scp> disease and healthy controls: A randomized placeboâ€controlled crossâ€over study. Journal of Inherited Metabolic Disease, 2022, 45, 502-516.	3.6	11
9	Botulinum toxin treatment improves dysphagia in patients with oculopharyngeal muscular dystrophy and sporadic inclusion body myositis. Journal of Neurology, 2022, 269, 4154-4160.	3.6	6
10	Novel truncating variants in <scp>FGD1</scp> detected in two Danish families with <scp>Aarskogâ€“Scott</scp> syndrome and myopathic features. American Journal of Medical Genetics, Part A, 2022, 188, 2251-2257.	1.2	3
11	Preclinical Research in McArdle Disease: A Review of Research Models and Therapeutic Strategies. Genes, 2022, 13, 74.	2.4	4
12	Three novel FHL1 variants cause a mild phenotype of Emeryâ€Dreifuss muscular dystrophy. Human Mutation, 2022, 43, 1234-1238.	2.5	2
13	High Resolution Analysis of DMPK Hypermethylation and Repeat Interruptions in Myotonic Dystrophy Type 1. Genes, 2022, 13, 970.	2.4	10
14	Long-term Safety and Efficacy of Avalglucosidase Alfa in Patients With Late-Onset Pompe Disease. Neurology, 2022, 99, .	1.1	16
15	Efficacy and Safety of Rozanolixizumab in Moderate to Severe Generalized Myasthenia Gravis. Neurology, 2021, 96, e853-e865.	1.1	97
16	Episodic hyperCKaemia may be a feature of Î±â€methylacylâ€coenzyme A racemase deficiency. European Journal of Neurology, 2021, 28, 729-731.	3.3	5
17	251st ENMC international workshop: Polyglucosan storage myopathies 13â€“15 December 2019, Hoofddorp, the Netherlands. Neuromuscular Disorders, 2021, 31, 466-477.	0.6	4
18	Antimyostatin Treatment in Health and Disease: The Story of Great Expectations and Limited Success. Cells, 2021, 10, 533.	4.1	19

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19	Combined Muscle Biopsy and Comprehensive Electrophysiology in General Anesthesia is Valuable in Diagnosis of Neuromuscular Disease in Children. Neuropediatrics, 2021, 52, 462-468.	0.6	0
20	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
21	Exercise Testing, Physical Training and Fatigue in Patients with Mitochondrial Myopathy Related to mtDNA Mutations. Journal of Clinical Medicine, 2021, 10, 1796.	2.4	8
22	Function, structure and quality of striated muscles in the lower extremities in patients with late onset Pompe Diseaseâ€”an MRI study. PeerJ, 2021, 9, e10928.	2.0	2
23	Patients With Becker Muscular Dystrophy Have Severe Paraspinal Muscle Involvement. Frontiers in Neurology, 2021, 12, 613483.	2.4	2
24	No effect of resveratrol in patients with mitochondrial myopathy: A crossâ€over randomized controlled trial. Journal of Inherited Metabolic Disease, 2021, 44, 1186-1198.	3.6	4
25	Autophagy is affected in patients with hypokalemic periodic paralysis: an involvement in vacuolar myopathy?. Acta Neuropathologica Communications, 2021, 9, 109.	5.2	2
26	Energy metabolism during exercise in patients with Î²â€enolase deficiency (GSDXIII). JIMD Reports, 2021, 61, 60-66.	1.5	1
27	Myopathy can be a key phenotype of membrin (GOSR2) deficiency. Human Mutation, 2021, 42, 1101-1106.	2.5	3
28	Cardiac Involvement in Women With Pathogenic Dystrophin Gene Variants. Frontiers in Neurology, 2021, 12, 707838.	2.4	8
29	E-Health & Innovation to Overcome Barriers in Neuromuscular Diseases. Report from the 1st eNMD Congress: Nice, France, March 22-23, 2019. Journal of Neuromuscular Diseases, 2021, 8, 743-754.	2.6	2
30	Improving Care and Empowering Adults Living with SMA: A Call to Action in the New Treatment Era. Journal of Neuromuscular Diseases, 2021, 8, 543-551.	2.6	9
31	1st FSHD European Trial Network workshop:Working towards trial readiness across Europe. Neuromuscular Disorders, 2021, 31, 907-918.	0.6	9
32	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
33	Muscle involvement assessed by quantitative magnetic resonance imaging in patients with anoctamin 5 deficiency. European Journal of Neurology, 2021, 28, 3121-3132.	3.3	13
34	Progression or Not â€“ A Small Natural History Study of Genetical Confirmed Congenital Myopathies. Journal of Neuromuscular Diseases, 2021, 8, 647-655.	2.6	1
35	Prolonged fastingâ€induced hyperketosis, hypoglycaemia and impaired fat oxidation in child and adult patients with spinal muscular atrophy type II. Acta Paediatrica, International Journal of Paediatrics, 2021, 110, 3367-3375.	1.5	4
36	Fatigue, physical activity and associated factors in 779 patients with myasthenia gravis. Neuromuscular Disorders, 2021, 31, 716-725.	0.6	13

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37	Plasma lactate responses during and after submaximal handgrip exercise are not diagnostically helpful in mitochondrial myopathy. Mitochondrion, 2021, 60, 21-26.	3.4	0
38	Quantitative Muscle MRI and Clinical Findings in Women With Pathogenic Dystrophin Gene Variants. Frontiers in Neurology, 2021, 12, 707837.	2.4	9
39	Eculizumab in refractory generalized myasthenia gravis previously treated with rituximab: subgroup analysis of <scp>REGAIN</scp> and its extension study. Muscle and Nerve, 2021, 64, 662-669.	2.2	11
40	METABOLIC MYOPATHIES. Neuromuscular Disorders, 2021, 31, S111-S112.	0.6	0
41	LGMD. Neuromuscular Disorders, 2021, 31, S103.	0.6	0
42	LGMD. Neuromuscular Disorders, 2021, 31, S107-S108.	0.6	0
43	Nampt controls skeletal muscle development by maintaining Ca2+ homeostasis and mitochondrial integrity. Molecular Metabolism, 2021, 53, 101271.	6.5	27
44	Muscle biopsy and <scp>MRI</scp> findings in <scp>ANO5</scp>â€related myopathy. Muscle and Nerve, 2021, 64, 743-748.	2.2	6
45	Extreme Hypoxia Causing Brady-Arrythmias During Apnea in Elite Breath-Hold Divers. Frontiers in Physiology, 2021, 12, 712573.	2.8	2
46	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	3.6	43
47	Growth and differentiation factor 15 as a biomarker for mitochondrial myopathy. Mitochondrion, 2020, 50, 35-41.	3.4	38
48	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
49	Vacuoles, Often Containing Glycogen, Are a Consistent Finding in Hypokalemic Periodic Paralysis. Journal of Neuropathology and Experimental Neurology, 2020, 79, 1127-1129.	1.7	2
50	MUSCLE IMAGING â€“ MRI. Neuromuscular Disorders, 2020, 30, S95-S96.	0.6	0
51	249th ENMC International Workshop: The role of brain dystrophin in muscular dystrophy: Implications for clinical care and translational research, Hoofddorp, The Netherlands, November 29thâ€“December 1st 2019. Neuromuscular Disorders, 2020, 30, 782-794.	0.6	14
52	Creation and implementation of a European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC registry). Orphanet Journal of Rare Diseases, 2020, 15, 187.	2.7	3
53	Preserved Capacity for Adaptations in Strength and Muscle Regulatory Factors in Elderly in Response to Resistance Exercise Training and Deconditioning. Journal of Clinical Medicine, 2020, 9, 2188.	2.4	10
54	Responsiveness of outcome measures in myotonic dystrophy type 1. Annals of Clinical and Translational Neurology, 2020, 7, 1382-1391.	3.7	2

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55	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
56	COVID-19-associated risks and effects in myasthenia gravis (CARE-MG). Lancet Neurology, The, 2020, 19, 970-971.	10.2	85
57	No effect of oral sucrose or IV glucose during exercise in phosphorylase b kinase deficiency. Neuromuscular Disorders, 2020, 30, 340-345.	0.6	4
58	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. European Journal of Neurology, 2020, 27, 2604-2615.	3.3	16
59	Consistent improvement with eculizumab across muscle groups in myasthenia gravis. Annals of Clinical and Translational Neurology, 2020, 7, 1327-1339.	3.7	16
60	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	7.6	45
61	Editorial: Remaining diagnostic issues and start of a treatment era for muscle diseases. Current Opinion in Neurology, 2020, 33, 587-589.	3.6	0
62	Stable Longitudinal Methylation Levels at the CpG Sites Flanking the CTG Repeat of DMPK in Patients with Myotonic Dystrophy Type 1. Genes, 2020, 11, 936.	2.4	10
63	Preclinical Research in Glycogen Storage Diseases: A Comprehensive Review of Current Animal Models. International Journal of Molecular Sciences, 2020, 21, 9621.	4.1	13
64	Mutation Load of Single, Large-Scale Deletions of mtDNA in Mitotic and Postmitotic Tissues. Frontiers in Genetics, 2020, 11, 547638.	2.3	7
65	Bimagrumab vs Optimized Standard of Care for Treatment of Sarcopenia in Community-Dwelling Older Adults. JAMA Network Open, 2020, 3, e2020836.	5.9	71
66	A quantitative method to assess muscle edema using short TI inversion recovery MRI. Scientific Reports, 2020, 10, 7246.	3.3	8
67	Characteristic muscle signatures assessed by quantitative MRI in patients with Bethlem myopathy. Journal of Neurology, 2020, 267, 2432-2442.	3.6	10
68	Impaired lipolysis in propionic acidemia: A new metabolic myopathy?. JIMD Reports, 2020, 53, 16-21.	1.5	10
69	A New Glycogen Storage Disease Caused by a DominantPYGMMutation. Annals of Neurology, 2020, 88, 274-282.	5.3	5
70	Intrarater reliability and validity of outcome measures in myotonic dystrophy type 1. Neurology, 2020, 94, e2508-e2520.	1.1	7
71	Results of an open label feasibility study of sodium valproate in people with McArdle disease. Neuromuscular Disorders, 2020, 30, 734-741.	0.6	3
72	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. Genetics in Medicine, 2020, 22, 1478-1488.	2.4	62

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73	MRI in Neuromuscular Diseases: An Emerging Diagnostic Tool and Biomarker for Prognosis and Efficacy. Annals of Neurology, 2020, 88, 669-681.	5.3	46
74	A single c.1715G>C calpain 3 gene variant causes dominant calpainopathy with loss of calpain 3 expression and activity. Human Mutation, 2020, 41, 1507-1513.	2.5	15
75	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
76	Permanent muscle weakness in hypokalemic periodic paralysis. Neurology, 2020, 95, e342-e352.	1.1	17
77	Depletion of ATP Limits Membrane Excitability of Skeletal Muscle by Increasing Both ClC1-Open Probability and Membrane Conductance. Frontiers in Neurology, 2020, 11, 541.	2.4	9
78	Contractile properties are impaired in congenital myopathies. Neuromuscular Disorders, 2020, 30, 649-655.	0.6	2
79	Evaluation of inflammatory lesions over 2 years in facioscapulohumeral muscular dystrophy. Neurology, 2020, 95, e1211-e1221.	1.1	27
80	Titrating a modified ketogenic diet for patients with McArdle disease: A pilot study. Journal of Inherited Metabolic Disease, 2020, 43, 778-786.	3.6	20
81	Physical activity in myotonic dystrophy type 1. Journal of Neurology, 2020, 267, 1679-1686.	3.6	7
82	Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. Neurology, 2020, 94, e1094-e1102.	1.1	45
83	Safety and efficacy of omaveloxolone in patients with mitochondrial myopathy. Neurology, 2020, 94, e687-e698.	1.1	38
84	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
85	Global FKRP Registry: observations in more than 300 patients with Limb Girdle Muscular Dystrophy R9. Annals of Clinical and Translational Neurology, 2020, 7, 757-766.	3.7	20
86	Quantitative Muscle MRI as Outcome Measure in Patients With Becker Muscular Dystrophyâ€”A 1-Year Follow-Up Study. Frontiers in Neurology, 2020, 11, 613489.	2.4	9
87	Effect of Aerobic Exercise Training and Deconditioning on Oxidative Capacity and Muscle Mitochondrial Enzyme Machinery in Young and Elderly Individuals. Journal of Clinical Medicine, 2020, 9, 3113.	2.4	16
88	Late-onset MADD: a rare cause of cirrhosis and acute liver failure?. Acta Myologica, 2020, 39, 19-23.	1.5	3
89	Phenotypic Spectrum of Î±-Dystroglycanopathies Associated With the c.919T>a Variant in the FKRP Gene in Humans and Mice. Journal of Neuropathology and Experimental Neurology, 2020, 79, 1257-1264.	1.7	5
90	Muscle contractility of leg muscles in patients with mitochondrial myopathies. Mitochondrion, 2019, 46, 221-227.	3.4	5

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91	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
92	Long-term efficacy and safety of eculizumab in Japanese patients with generalized myasthenia gravis: A subgroup analysis of the REGAIN open-label extension study. Journal of the Neurological Sciences, 2019, 407, 116419.	0.6	18
93	Impaired Fat Oxidation During Exercise in Long-Chain Acyl-CoA Dehydrogenase Deficiency Patients and Effect of IV-Glucose. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3610-3613.	3.6	1
94	POPDC3 Gene Variants Associate with a New Form of Limb Girdle Muscular Dystrophy. Annals of Neurology, 2019, 86, 832-843.	5.3	27
95	P.69NEO1 and NEO-EXT studies: exploratory efficacy of repeat avalglucosidase alfa dosing for up to 5 years in participants with late-onset Pompe disease (LOPD). Neuromuscular Disorders, 2019, 29, S60-S61.	0.6	0
96	EP.54Assessment of trunk muscle strength in patients with muscular dystrophies using stationary and hand-held dynamometry: a test-retest reliability study. Neuromuscular Disorders, 2019, 29, S116-S117.	0.6	1
97	P.375Does rhythmic auditory stimulation influence walking speed in the 6-minute walk test in patients with myasthenia gravis?. Neuromuscular Disorders, 2019, 29, S190-S191.	0.6	0
98	No effect of triheptanoin on exercise performance in McArdle disease. Annals of Clinical and Translational Neurology, 2019, 6, 1949-1960.	3.7	17
99	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.	5.2	17
100	Hydroxylated long-chain acylcarnitines are biomarkers of mitochondrial myopathy. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 5968-5976.	3.6	12
101	MSTO1 mutations cause mtDNA depletion, manifesting as muscular dystrophy with cerebellar involvement. Acta Neuropathologica, 2019, 138, 1013-1031.	7.7	31
102	MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscular Disorders, 2019, 29, 827-841.	0.6	46
103	Impairments in contractility and cytoskeletal organisation cause nuclear defects in nemaline myopathy. Acta Neuropathologica, 2019, 138, 477-495.	7.7	25
104	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
105	High-intensity training in patients with spinal and bulbar muscular atrophy. Journal of Neurology, 2019, 266, 1693-1697.	3.6	14
106	NEO1 and NEO-EXT studies: Long-term safety of repeat avalglucosidase alfa dosing for 4.5 years in late-onset Pompe disease patients. Molecular Genetics and Metabolism, 2019, 126, S115-S116.	1.1	0
107	The Pathophysiology of Exercise and Effect of Training in Mitochondrial Myopathies. , 2019, , 331-348.		1
108	Moderateâ€intensity aerobic exercise improves physical fitness in bethlem myopathy. Muscle and Nerve, 2019, 60, 183-188.	2.2	4

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109	Low survival rate and muscle fiber-dependent aging effects in the McArdle disease mouse model. Scientific Reports, 2019, 9, 5116.	3.3	11
110	Congenital myopathies are mainly associated with a mild cardiac phenotype. Journal of Neurology, 2019, 266, 1367-1375.	3.6	10
111	Adaptations in Mitochondrial Enzymatic Activity Occurs Independent of Genomic Dosage in Response to Aerobic Exercise Training and Deconditioning in Human Skeletal Muscle. Cells, 2019, 8, 237.	4.1	20
112	Muscle contractility in spinobulbar muscular atrophy. Scientific Reports, 2019, 9, 4680.	3.3	17
113	Aberrant regulation of epigenetic modifiers contributes to the pathogenesis in patients with selenoprotein N â€ related myopathies. Human Mutation, 2019, 40, 962-974.	2.5	13
114	Relationship between muscle inflammation and fat replacement assessed by MRI in facioscapulohumeral muscular dystrophy. Journal of Neurology, 2019, 266, 1127-1135.	3.6	33
115	Longâ€term safety and efficacy of eculizumab in generalized myasthenia gravis. Muscle and Nerve, 2019, 60, 14-24.	2.2	162
116	Absence of p.R50X Pygm read-through in McArdle disease cellular models. DMM Disease Models and Mechanisms, 2019, 13, .	2.4	4
117	Fat oxidation is impaired during exercise in lipin-1 deficiency. Neurology, 2019, 93, e1433-e1438.	1.1	6
118	Expanding the phenotype of filamin-C-related myofibrillar myopathy. Clinical Neurology and Neurosurgery, 2019, 176, 30-33.	1.4	8
119	Refining the spinobulbar muscular atrophy phenotype by quantitative MRI and clinical assessments. Neurology, 2019, 92, e548-e559.	1.1	23
120	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
121	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
122	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
123	Muscle Strength and Aerobic Capacity in Patients with CIDP One Year after Participation in an Exercise Trial. Journal of Neuromuscular Diseases, 2019, 6, 93-97.	2.6	7
124	Genetic analysis of Charcot-Marie-Tooth disease in Denmark and the implementation of a next generation sequencing platform. European Journal of Medical Genetics, 2019, 62, 1-8.	1.3	20
125	Mitochondrial mutation m.3243A>G associates with insulin resistance in non-diabetic carriers. Endocrine Connections, 2019, 8, 829-837.	1.9	4
126	European consensus for starting and stopping enzyme replacement therapy in adult patients with Pompe disease: a 10-year experience. Nervno-Myshechnye Bolezni, 2019, 8, 19-34.	0.4	0

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127	Impaired fat oxidation during exercise in multiple acyl-CoA dehydrogenase deficiency. JIMD Reports, 2019, 46, 79-84.	1.5	2
128	Exercise therapy for muscle and lower motor neuron diseases. Acta Myologica, 2019, 38, 215-232.	1.5	8
129	Progressive fat replacement of muscle contributes to the disease mechanism of patients with single, large-scale deletions of mitochondrial DNA. Neuromuscular Disorders, 2018, 28, 408-413.	0.6	8
130	Remodel mitochondria and get energized. Neurology, 2018, 90, 633-634.	1.1	1
131	Correlation between myasthenia gravisâ˜activities of daily living (MGâ€ADL) and quantitative myasthenia gravis (QMG) assessments of antiâ˜acetylcholine receptor antibodyâ˜positive refractory generalized myasthenia gravis in the phase 3 regain study. Muscle and Nerve, 2018, 58, E21-E22.	2.2	5
132	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
133	233rd ENMC International Workshop:. Neuromuscular Disorders, 2018, 28, 540-549.	0.6	5
134	Phenotype and genotype of muscle ryanodine receptor rhabdomyolysis-myalgia syndrome. Acta Neurologica Scandinavica, 2018, 137, 452-461.	2.1	31
135	Collagen XII myopathy with rectus femoris atrophy and collagen XII retention in fibroblasts. Muscle and Nerve, 2018, 57, 1026-1030.	2.2	11
136	Lecocytes mutation load declines with age in carriers of the m.3243A>G mutation: A 10â€year Prospective Cohort. Clinical Genetics, 2018, 93, 925-928.	2.0	11
137	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
138	Screening for late-onset Pompe disease in western Denmark. Acta Neurologica Scandinavica, 2018, 137, 85-90.	2.1	9
139	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	1.9	55
140	Exercising with blocked muscle glycogenolysis: Adaptation in the McArdle mouse. Molecular Genetics and Metabolism, 2018, 123, 21-27.	1.1	5
141	L-Carnitine Improves Skeletal Muscle Fat Oxidation in Primary Carnitine Deficiency. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4580-4588.	3.6	15
142	Disease progression and outcome measures in spinobulbar muscular atrophy. Annals of Neurology, 2018, 84, 754-765.	5.3	25
143	Homozygosity for SCN4A Arg1142Gln causes congenital myopathy with variable disease expression. Neurology: Genetics, 2018, 4, e267.	1.9	9
144	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

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145	Intact transferrin and total plasma glycoprofiling for diagnosis and therapy monitoring in phosphoglucomutase-I deficiency. Translational Research, 2018, 199, 62-76.	5.0	22
146	BAG3 myopathy is not always associated with cardiomyopathy. Neuromuscular Disorders, 2018, 28, 798-801.	0.6	11
147	Level of residual enzyme activity modulates the phenotype in phosphoglycerate kinase deficiency. Neurology, 2018, 91, e1077-e1082.	1.1	15
148	Altered somatosensory neurovascular response in patients with Becker muscular dystrophy. Brain and Behavior, 2018, 8, e00985.	2.2	1
149	SCA28: Novel Mutation in the AFG3L2 Proteolytic Domain Causes a Mild Cerebellar Syndrome with Selective Type-1 Muscle Fiber Atrophy. Cerebellum, 2017, 16, 62-67.	2.5	16
150	Human growth hormone stabilizes walking and improves strength in a patient with dominantly inherited calpainopathy. Neuromuscular Disorders, 2017, 27, 358-362.	0.6	6
151	Leber hereditary optic neuropathy due to a new ND1 mutation. Ophthalmic Genetics, 2017, 38, 480-485.	1.2	6
152	PGM1 deficiency: Substrate use during exercise and effect of treatment with galactose. Neuromuscular Disorders, 2017, 27, 370-376.	0.6	31
153	1st International Workshop on Clinical trial readiness for sarcoglycanopathies 15â€“16 November 2016, Evry, France. Neuromuscular Disorders, 2017, 27, 683-692.	0.6	9
154	Skeletal muscle metabolism during prolonged exercise in Pompe disease. Endocrine Connections, 2017, 6, 384-394.	1.9	8
155	High-intensity interval training in facioscapulohumeral muscular dystrophy type 1: a randomized clinical trial. Journal of Neurology, 2017, 264, 1099-1106.	3.6	26
156	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
157	Glycogen Synthesis in Glycogenin 1â€“Deficient Patients: A Role for Glycogenin 2 in Muscle. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 2690-2700.	3.6	16
158	DOK7 congenital myasthenia may be associated with severe mitral valve insufficiency. Journal of the Neurological Sciences, 2017, 379, 217-218.	0.6	3
159	Fat Replacement of Paraspinal Muscles with Aging in Healthy Adults. Medicine and Science in Sports and Exercise, 2017, 49, 595-601.	0.4	78
160	Mitochondrial Point Mutation m.3243A>G Associates With Lower Bone Mineral Density, Thinner Cortices, and Reduced Bone Strength: A Case-Control Study. Journal of Bone and Mineral Research, 2017, 32, 2041-2048.	2.8	9
161	Phenotypes, genotypes, and prevalence of congenital myopathies older than 5 years in Denmark. Neurology: Genetics, 2017, 3, e140.	1.9	34
162	Dysphagia is prevalent in patients with CPEO and single, large-scale deletions in mtDNA. Mitochondrion, 2017, 32, 27-30.	3.4	9

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163	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
164	Aerobic training in myotonia congenita: Effect on myotonia and fitness. Muscle and Nerve, 2017, 56, 696-699.	2.2	3
165	Treatment Opportunities in Patients With Metabolic Myopathies. Current Treatment Options in Neurology, 2017, 19, 37.	1.8	14
166	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
167	The antimyotonic effect of lamotrigine in non-dystrophic myotonias: a double-blind randomized study. Brain, 2017, 140, 2295-2305.	7.6	49
168	211th ENMC International Workshop:. Neuromuscular Disorders, 2017, 27, 1143-1151.	0.6	6
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