John Vissing

List of Publications by Citations

357 papers	9,826 citations	53 h-index	81 g-index
413 ext. papers	11,813 ext. citations	5.2 avg, IF	6.25 L-index

#	Paper	IF	Citations
357	Paternal inheritance of mitochondrial DNA. *New England Journal of Medicine*, **2002**, 347, 576-80	59.2	473
356	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	24.1	278
355	Recombination of human mitochondrial DNA. *Science*, **2004**, 304, 981	33.3	203
354	The spectrum of exercise tolerance in mitochondrial myopathies: a study of 40 patients. *Brain*, **2003**, 126, 413-23	11.2	177
353	The effect of oral sucrose on exercise tolerance in patients with McArdle's disease. *New England Journal of Medicine*, **2003**, 349, 2503-9	59.2	175
352	Aerobic training is safe and improves exercise capacity in patients with mitochondrial myopathy. *Brain*, **2006**, 129, 3402-12	11.2	151
351	Mitochondrial encephalomyopathy with elevated methylmalonic acid is caused by SUCLA2 mutations. *Brain*, **2007**, 130, 853-61	11.2	148
350	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, 43081-8	5.4	147
349	Novel POLG mutations in progressive external ophthalmoplegia mimicking mitochondrial neurogastrointestinal encephalomyopathy. *European Journal of Human Genetics*, **2003**, 11, 547-9	5.3	131
348	Exercise-induced changes in local cerebral glucose utilization in the rat. *Journal of Cerebral Blood Flow and Metabolism*, **1996**, 16, 729-36	7.3	125
347	Aerobic conditioning: an effective therapy in McArdle's disease. *Annals of Neurology*, **2006**, 59, 922-8	9.4	120
346	High prevalence and phenotype-genotype correlations of limb girdle muscular dystrophy type 2I in Denmark. *Annals of Neurology*, **2006**, 59, 808-15	9.4	118
345	Quantitative muscle MRI as an assessment tool for monitoring disease progression in LGMD2I: a multicentre longitudinal study. *PLoS ONE*, **2013**, 8, e70993	3.7	116
344	McArdle disease: a clinical review. *Journal of Neurology, Neurosurgery and Psychiatry*, **2010**, 81, 1182-8	5.5	110
343	Spontaneous "second wind" and glucose-induced second "second wind" in McArdle disease: oxidative mechanisms. *Archives of Neurology*, **2002**, 59, 1395-402		108
342	Aerobic training improves exercise performance in facioscapulohumeral muscular dystrophy. *Neurology*, **2005**, 64, 1064-6	6.5	107
341	Cardiac manifestations of myotonic dystrophy type 1. *International Journal of Cardiology*, **2012**, 160, 82-8	3.2	105

340	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. *Human Mutation*, **2012**, 33, 949-59	4.7	91
339	Muscle glycogenosis due to phosphoglucomutase 1 deficiency. *New England Journal of Medicine*, **2009**, 361, 425-7	59.2	89
338	A nonischemic forearm exercise test for McArdle disease. *Annals of Neurology*, **2002**, 52, 153-9	9.4	86
337	Endurance training improves fitness and strength in patients with Becker muscular dystrophy. *Brain* , **2008**, 131, 2824-31	11.2	82
336	Aerobic training in patients with myotonic dystrophy type 1. *Annals of Neurology*, **2005**, 57, 754-7	9.4	82
335	European consensus for starting and stopping enzyme replacement therapy in adult patients with Pompe disease: a 10-year experience. *European Journal of Neurology*, **2017**, 24, 768-e31	6	81
334	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	3.2	81
333	A diagnostic cycle test for McArdle disease. *Annals of Neurology*, **2003**, 54, 539-42	9.4	79
332	Loss-of-function mutations in SCN4A cause severe foetal hypokinesia or classical congenital myopathy. *Brain*, **2016**, 139, 674-91	11.2	76
331	Open-label trial of anti-TNF-alpha in dermato- and polymyositis treated concomitantly with methotrexate. *European Neurology*, **2008**, 59, 159-63	2.1	76
330	Treatment of mitochondrial neurogastrointestinal encephalomyopathy with dialysis. *Archives of Neurology*, **2007**, 64, 435-8		76
329	Long-term safety and efficacy of eculizumab in generalized myasthenia gravis. *Muscle and Nerve*, **2019**, 60, 14-24	3.4	76
328	Tissue specific distribution of the 3243A->G mtDNA mutation. *Journal of Medical Genetics*, **2006**, 43, 671-5	5.8	73
327	Leg muscle involvement in facioscapulohumeral muscular dystrophy assessed by MRI. *Journal of Neurology*, **2006**, 253, 1437-41	5.5	73
326	Role of 5AMP-activated protein kinase in glycogen synthase activity and glucose utilization: insights from patients with McArdle disease. *Journal of Physiology*, **2002**, 541, 979-89	3.9	68
325	Bezafibrate in skeletal muscle fatty acid oxidation disorders: a randomized clinical trial. *Neurology*, **2014**, 82, 607-13	6.5	67
324	Severe paraspinal muscle involvement in facioscapulohumeral muscular dystrophy. *Neurology*, **2014** , 83, 1178-83	6.5	66
323	Endurance training: an effective and safe treatment for patients with LGMD2I. *Neurology*, **2007**, 68, 59-61	6.5	66

322	Multisystem disorder associated with a missense mutation in the mitochondrial cytochrome b gene. *Annals of Neurology*, **2001**, 50, 540-3	9.4	66
321	A new mitochondrial tRNA(Met) gene mutation in a patient with dystrophic muscle and exercise intolerance. *Neurology*, **1998**, 50, 1875-8	6.5	66
320	Fuel utilization in subjects with carnitine palmitoyltransferase 2 gene mutations. *Annals of Neurology*, **2005**, 57, 60-6	9.4	64
319	Quantitative magnetic resonance imaging in limb-girdle muscular dystrophy 2I: a multinational cross-sectional study. *PLoS ONE*, **2014**, 9, e90377	3.7	62
318	Difference in allelic expression of the CLCN1 gene and the possible influence on the myotonia congenita phenotype. *European Journal of Human Genetics*, **2004**, 12, 738-43	5.3	60
317	Oxidative capacity correlates with muscle mutation load in mitochondrial myopathy. *Annals of Neurology*, **2003**, 54, 86-92	9.4	60
316	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. *Annals of Neurology*, **2018**, 83, 1105-1124	9.4	59
315	Effect of diet on exercise tolerance in carnitine palmitoyltransferase II deficiency. *Neurology*, **2003**, 61, 559-61	6.5	59
314	Effect of deficient muscular glycogenolysis on extramuscular fuel production in exercise. *Journal of Applied Physiology*, **1992**, 72, 1773-9	3.7	59
313	Late-onset Pompe disease is prevalent in unclassified limb-girdle muscular dystrophies. *Molecular Genetics and Metabolism*, **2013**, 110, 287-9	3.7	58
312	Muscle phenotype and mutation load in 51 persons with the 3243A>G mitochondrial DNA mutation. *Archives of Neurology*, **2006**, 63, 1701-6		58
311	Fat Replacement of Paraspinal Muscles with Aging in Healthy Adults. *Medicine and Science in Sports and Exercise*, **2017**, 49, 595-601	1.2	57
310	A heterozygous 21-bp deletion in CAPN3 causes dominantly inherited limb girdle muscular dystrophy. *Brain*, **2016**, 139, 2154-63	11.2	56
309	Effect of oral sucrose shortly before exercise on work capacity in McArdle disease. *Archives of Neurology*, **2008**, 65, 786-9		56
308	Lactate production and clearance in exercise. Effects of training. A mini-review. *Scandinavian Journal of Medicine and Science in Sports*, **1998**, 8, 127-31	4.6	56
307	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	24.1	55
306	No spontaneous second wind in muscle phosphofructokinase deficiency. *Neurology*, **2004**, 62, 82-6	6.5	55
305	MRI as outcome measure in facioscapulohumeral muscular dystrophy: 1-year follow-up of 45 patients. *Journal of Neurology*, **2017**, 264, 438-447	5.5	52

304	Anoctamin 5 muscular dystrophy in Denmark: prevalence, genotypes, phenotypes, cardiac findings, and muscle protein expression. *Journal of Neurology*, **2013**, 260, 2084-93	5.5	52
303	Sympathetic activation in exercise is not dependent on muscle acidosis. Direct evidence from studies in metabolic myopathies. *Journal of Clinical Investigation*, **1998**, 101, 1654-60	15.9	50
302	Diagnosis of Pompe disease: muscle biopsy vs blood-based assays. *JAMA Neurology*, **2013**, 70, 923-7	17.2	49
301	LGMD2I presenting with a characteristic Duchenne or Becker muscular dystrophy phenotype. *Neurology*, **2005**, 64, 1635-7	6.5	49
300	Reduced levels of skeletal muscle Na+K+ -ATPase in McArdle disease. *Neurology*, **1998**, 50, 37-40	6.5	48
299	Exercise fuel mobilization in mitochondrial myopathy: a metabolic dilemma. *Annals of Neurology*, **1996**, 40, 655-62	9.4	46
298	Cardiac involvement in myotonic dystrophy: a nationwide cohort study. *European Heart Journal*, **2014**, 35, 2158-64	9.5	45
297	Is muscle glycogenolysis impaired in X-linked phosphorylase b kinase deficiency?. *Neurology*, **2008**, 70, 1876-82	6.5	45
296	A forearm exercise screening test for mitochondrial myopathy. *Neurology*, **2002**, 58, 1533-8	6.5	45
295	COVID-19-associated risks and effects in myasthenia gravis (CARE-MG). *Lancet Neurology, The*, **2020**, 19, 970-971	24.1	45
294	Limb girdle muscular dystrophies: classification, clinical spectrum and emerging therapies. *Current Opinion in Neurology*, **2016**, 29, 635-41	7.1	45
293	Cardiac involvement in patients with limb-girdle muscular dystrophy type 2 and Becker muscular dystrophy. *Archives of Neurology*, **2008**, 65, 1196-201		44
292	Splice mutations preserve myophosphorylase activity that ameliorates the phenotype in McArdle disease. *Brain*, **2009**, 132, 1545-52	11.2	43
291	Effect of aerobic training in patients with spinal and bulbar muscular atrophy (Kennedy disease). *Neurology*, **2009**, 72, 317-23	6.5	43
290	Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. *Annals of Clinical and Translational Neurology*, **2014**, 1, 88-98	5.3	42
289	Carbohydrate- and protein-rich diets in McArdle disease: effects on exercise capacity. *Journal of Neurology, Neurosurgery and Psychiatry*, **2008**, 79, 1359-63	5.5	42
288	Fuel utilization in patients with very long-chain acyl-coa dehydrogenase deficiency. *Annals of Neurology*, **2004**, 56, 279-83	9.4	42
287	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, 2141-51	5.6	41

286	Mutations in COA3 cause isolated complex IV deficiency associated with neuropathy, exercise intolerance, obesity, and short stature. *Journal of Medical Genetics*, **2015**, 52, 203-7	5.8	41
285	Clinical and molecular characterization of limb-girdle muscular dystrophy due to LAMA2 mutations. *Muscle and Nerve*, **2011**, 44, 703-9	3.4	40
284	31P-MRS of skeletal muscle is not a sensitive diagnostic test for mitochondrial myopathy. *Journal of Neurology*, **2007**, 254, 29-37	5.5	40
283	Calpain 3 is important for muscle regeneration: evidence from patients with limb girdle muscular dystrophies. *BMC Musculoskeletal Disorders*, **2012**, 13, 43	2.8	39
282	New patterns of inheritance in mitochondrial disease. *Biochemical and Biophysical Research Communications*, **2003**, 310, 247-51	3.4	39
281	Endocrine function in 97 patients with myotonic dystrophy type 1. *Journal of Neurology*, **2012**, 259, 912-20	5.5	38
280	Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. *Neurology*, **2015**, 84, 1772-81	6.5	37
279	Fat metabolism during exercise in patients with McArdle disease. *Neurology*, **2009**, 72, 718-24	6.5	37
278	Effect of sildenafil on skeletal and cardiac muscle in Becker muscular dystrophy. *Annals of Neurology*, **2014**, 76, 550-7	9.4	36
277	Short- and long-term effects of endurance training in patients with mitochondrial myopathy. *European Journal of Neurology*, **2009**, 16, 1336-9	6	36
276	Exercise tolerance in carnitine palmitoyltransferase II deficiency with IV and oral glucose. *Neurology* , **2002**, 59, 1046-51	6.5	36
275	Multiple mtDNA deletions with features of MNGIE. *Neurology*, **2002**, 59, 926-9	6.5	36
274	The antimyotonic effect of lamotrigine in non-dystrophic myotonias: a double-blind randomized study. *Brain*, **2017**, 140, 2295-2305	11.2	35
273	Effect of fuels on exercise capacity in muscle phosphoglycerate mutase deficiency. *Archives of Neurology*, **2005**, 62, 1440-3		35
272	Aerobic training and postexercise protein in facioscapulohumeral muscular dystrophy: RCT study. *Neurology*, **2015**, 85, 396-403	6.5	34
271	MRI in sarcoglycanopathies: a large international cohort study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 72-77	5.5	34
270	Exercise intolerance in Glycogen Storage Disease Type III: weakness or energy deficiency?. *Molecular Genetics and Metabolism*, **2013**, 109, 14-20	3.7	34
269	Oral branched-chain amino acids do not improve exercise capacity in McArdle disease. *Neurology*, **1998**, 51, 1456-9	6.5	34

268	Contractile properties are disrupted in Becker muscular dystrophy, but not in limb girdle type 2I. *Annals of Neurology*, **2016**, 80, 466-71	9.4	34
267	Two- and 6-minute walk tests assess walking capability equally in neuromuscular diseases. *Neurology*, **2016**, 86, 442-5	6.5	33
266	High prevalence of cardiac involvement in patients with myotonic dystrophy type 1: a cross-sectional study. *International Journal of Cardiology*, **2014**, 174, 31-6	3.2	33
265	Resistance training in patients with limb-girdle and becker muscular dystrophies. *Muscle and Nerve*, **2013**, 47, 163-9	3.4	33
264	Muscle phosphoglycerate mutase deficiency revisited. *Archives of Neurology*, **2009**, 66, 394-8		33
263	A decline in PABPN1 induces progressive muscle weakness in oculopharyngeal muscle dystrophy and in muscle aging. *Aging*, **2013**, 5, 412-26	5.6	33
262	Training improves oxidative capacity, but not function, in spinal muscular atrophy type III. *Muscle and Nerve*, **2015**, 52, 240-4	3.4	32
261	Myocardial fibrosis in patients with myotonic dystrophy type 1: a cardiovascular magnetic resonance study. *Journal of Cardiovascular Magnetic Resonance*, **2014**, 16, 59	6.9	32
260	Anti-gravity training improves walking capacity and postural balance in patients with muscular dystrophy. *Neuromuscular Disorders*, **2014**, 24, 492-8	2.9	32
259	Muscle structural changes in mitochondrial myopathy relate to genotype. *Journal of Neurology*, **2003**, 250, 1328-34	5.5	32
258	Exercise intolerance in mitochondrial myopathy is not related to lactic acidosis. *Annals of Neurology* , **2001**, 49, 672-676	9.4	32
257	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, 641-9	3.9	32
256	Late onset of stroke-like episode associated with a 3256C-->T point mutation of mitochondrial DNA. *Journal of the Neurological Sciences*, **2003**, 214, 17-20	3.2	31
255	Muscle phosphorylase kinase deficiency: a neutral metabolic variant or a disease?. *Neurology*, **2012**, 78, 265-8	6.5	30
254	Decrement of compound muscle action potential is related to mutation type in myotonia congenita. *Muscle and Nerve*, **2003**, 27, 449-55	3.4	30
253	Decreased insulin action in skeletal muscle from patients with McArdle's disease. *American Journal of Physiology - Endocrinology and Metabolism*, **2002**, 282, E1267-75	6	29
252	Cycle ergometry is not a sensitive diagnostic test for mitochondrial myopathy. *Journal of Neurology* , **2003**, 250, 293-9	5.5	29
251	Effect of liver glycogen content on glucose production in running rats. *Journal of Applied Physiology* , **1989**, 66, 318-22	3.7	29

250	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	24.1	29
249	Bimagrumab vs Optimized Standard of Care for Treatment of Sarcopenia in Community-Dwelling Older Adults: A Randomized Clinical Trial. *JAMA Network Open*, **2020**, 3, e2020836	10.4	28
248	Axial myopathy: an overlooked feature of muscle diseases. *Brain*, **2016**, 139, 13-22	11.2	28
247	LAMA2-related myopathy: Frequency among congenital and limb-girdle muscular dystrophies. *Muscle and Nerve*, **2015**, 52, 547-53	3.4	28
246	Clinical presentation and mutations in Danish patients with Wilson disease. *European Journal of Human Genetics*, **2011**, 19, 935-41	5.3	28
245	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, 403-11	3.2	27
244	Blocked muscle fat oxidation during exercise in neutral lipid storage disease. *Archives of Neurology*, **2012**, 69, 530-3		27
243	No evidence for paternal inheritance of mtDNA in patients with sporadic mtDNA mutations. *Journal of the Neurological Sciences*, **2004**, 218, 99-101	3.2	27
242	Safety, tolerability, pharmacokinetics, pharmacodynamics, and exploratory efficacy of the novel enzyme replacement therapy avalglucosidase alfa (neoGAA) in treatment-naïve 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	2.9	27
241	Muscle phosphoglycerate mutase deficiency with tubular aggregates: Effect of dantrolene. *Annals of Neurology*, **1999**, 46, 274-277	9.4	26
240	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. *Genetics in Medicine*, **2020**, 22, 1478-1488	8.1	25
239	PGM1 deficiency: Substrate use during exercise and effect of treatment with galactose. *Neuromuscular Disorders*, **2017**, 27, 370-376	2.9	24
238	Safety and efficacy of omaveloxolone in patients with mitochondrial myopathy: MOTOR trial. *Neurology*, **2020**, 94, e687-e698	6.5	24
237	Level of muscle regeneration in limb-girdle muscular dystrophy type 2I relates to genotype and clinical severity. *Skeletal Muscle*, **2011**, 1, 31	5.1	24
236	cDNA analyses of CAPN3 enhance mutation detection and reveal a low prevalence of LGMD2A patients in Denmark. *European Journal of Human Genetics*, **2008**, 16, 935-40	5.3	24
235	Phenotypes, genotypes, and prevalence of congenital myopathies older than 5 years in Denmark. *Neurology: Genetics*, **2017**, 3, e140	3.8	23
234	MYO-MRI diagnostic protocols in genetic myopathies. *Neuromuscular Disorders*, **2019**, 29, 827-841	2.9	23
233	Fat and carbohydrate metabolism during exercise in phosphoglucomutase type 1 deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, E1235-40	5.6	23

232	Efficacy and Safety of Rozanolixizumab in Moderate to Severe Generalized Myasthenia Gravis: A Phase 2 Randomized Control Trial. *Neurology*, **2021**, 96, e853-e865	6.5	23
231	Aerobic training in persons who have recovered from juvenile dermatomyositis. *Neuromuscular Disorders*, **2013**, 23, 962-8	2.9	22
230	Fat and carbohydrate metabolism during exercise in late-onset Pompe disease. *Molecular Genetics and Metabolism*, **2012**, 107, 462-8	3.7	22
229	Change in muscle strength over time in spinal muscular atrophy types II and III. A long-term follow-up study. *Neuromuscular Disorders*, **2012**, 22, 1069-74	2.9	22
228	EFNS review on the role of muscle biopsy in the investigation of myalgia. *European Journal of Neurology*, **2013**, 20, 997-1005	6	22
227	Prevalence of migraine in persons with the 3243A>G mutation in mitochondrial DNA. *European Journal of Neurology*, **2016**, 23, 175-81	6	22
226	Update on new muscle glycogenosis. *Current Opinion in Neurology*, **2017**, 30, 449-456	7.1	21
225	Pharmacologic treatment of downstream of tyrosine kinase 7 congenital myasthenic syndrome. *JAMA Neurology*, **2014**, 71, 350-4	17.2	21
224	Physical training for McArdle disease. *The Cochrane Library*, **2011**, CD007931	5.2	21
223	Paradoxically enhanced glucose production during exercise in humans with blocked glycolysis caused by muscle phosphofructokinase deficiency. *Neurology*, **1996**, 47, 766-71	6.5	21
222	Do carriers of PYGM mutations have symptoms of McArdle disease?. *Neurology*, **2006**, 67, 716-8	6.5	21
221	Muscle involvement in limb-girdle muscular dystrophy with GMPPB deficiency (LGMD2T). *Neurology: Genetics*, **2016**, 2, e112	3.8	21
220	MSTO1 mutations cause mtDNA depletion, manifesting as muscular dystrophy with cerebellar involvement. *Acta Neuropathologica*, **2019**, 138, 1013-1031	14.3	20
219	Exercise in muscle glycogen storage diseases. *Journal of Inherited Metabolic Disease*, **2015**, 38, 551-63	5.4	20
218	Skeletal muscle metabolism is impaired during exercise in glycogen storage disease type III. *Neurology*, **2015**, 84, 1767-71	6.5	20
217	Endocrine function over time in patients with myotonic dystrophy type 1. *European Journal of Neurology*, **2015**, 22, 116-22	6	20
216	Fatigue in patients with spinal muscular atrophy type II and congenital myopathies: evaluation of the fatigue severity scale. *Quality of Life Research*, **2014**, 23, 1479-88	3.7	20
215	Ocular, bulbar, limb, and cardiopulmonary involvement in oculopharyngeal muscular dystrophy. *Acta Neurologica Scandinavica*, **2014**, 130, 125-30	3.8	20

214	Recurrent myoglobinuria and deranged acylcarnitines due to a mutation in the mtDNA MT-CO2 gene. *Neurology*, **2013**, 80, 1908-10	6.5	20
213	Creatine kinase response to high-intensity aerobic exercise in adult-onset muscular dystrophy. *Muscle and Nerve*, **2013**, 48, 897-901	3.4	20
212	Resistance training and aerobic training improve muscle strength and aerobic capacity in chronic inflammatory demyelinating polyneuropathy. *Muscle and Nerve*, **2018**, 57, 70-76	3.4	19
211	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). *Journal of Neurology*, **2020**, 267, 45-56	5.5	19
210	Muscle atrophy reversed by growth factor activation of satellite cells in a mouse muscle atrophy model. *PLoS ONE*, **2014**, 9, e100594	3.7	18
209	Decreased variability of the 6-minute walk test by heart rate correction in patients with neuromuscular disease. *PLoS ONE*, **2014**, 9, e114273	3.7	18
208	Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 576-585	5.5	18
207	Natural history of limb girdle muscular dystrophy R9 over 6 years: searching for trial endpoints. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1033-1045	5.3	17
206	No effect of bezafibrate in patients with CPTII and VLCAD deficiencies. *Journal of Inherited Metabolic Disease*, **2015**, 38, 373-4	5.4	17
205	Patients with medium-chain acyl-coenzyme a dehydrogenase deficiency have impaired oxidation of fat during exercise but no effect of L-carnitine supplementation. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, 1667-75	5.6	17
204	High prevalence of impaired glucose homeostasis and myopathy in asymptomatic and oligosymptomatic 3243A>G mitochondrial DNA mutation-positive subjects. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 2872-9	5.6	17
203	Effect of changes in fat availability on exercise capacity in McArdle disease. *Archives of Neurology*, **2009**, 66, 762-6		17
202	Limited diagnostic value of enzyme analysis in patients with mitochondrial tRNA mutations. *Muscle and Nerve*, **2010**, 41, 607-13	3.4	17
201	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	24.1	17
200	Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. *Neurology*, **2020**, 94, e1094-e1102	6.5	16
199	Lactate metabolism during exercise in patients with mitochondrial myopathy. *Neuromuscular Disorders*, **2013**, 23, 629-36	2.9	16
198	Muscle strength in myasthenia gravis. *Acta Neurologica Scandinavica*, **2014**, 129, 367-73	3.8	16
197	Effects of IV glucose and oral medium-chain triglyceride in patients with VLCAD deficiency. *Neurology*, **2007**, 69, 313-5	6.5	16

196	Characterization of two new dominant ClC-1 channel mutations associated with myotonia. *Muscle and Nerve*, **2003**, 28, 722-32	3.4	16
195	Regulation of hepatic glucose production in running rats studied by glucose infusion. *Journal of Applied Physiology*, **1988**, 65, 2552-7	3.7	16
194	Prevalence and phenotypes of congenital myopathy due to actin 1 gene mutations. *Muscle and Nerve*, **2016**, 53, 388-93	3.4	16
193	Differential Muscle Involvement in Mice and Humans Affected by McArdle Disease. *Journal of Neuropathology and Experimental Neurology*, **2016**, 75, 441-54	3.1	16
192	Glycogen Synthesis in Glycogenin 1-Deficient Patients: A Role for Glycogenin 2 in Muscle. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 2690-2700	5.6	15
191	Mechanisms of exertional fatigue in muscle glycogenoses. *Neuromuscular Disorders*, **2012**, 22 Suppl 3, S168-71	2.9	15
190	No muscle involvement in myoclonus-dystonia caused by epsilon-sarcoglycan gene mutations. *European Journal of Neurology*, **2008**, 15, 525-9	6	15
189	Growth and differentiation factor 15 as a biomarker for mitochondrial myopathy. *Mitochondrion*, **2020**, 50, 35-41	4.9	15
188	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. *Brain*, **2020**, 143, 2696-2708	11.2	15
187	Relationship between muscle inflammation and fat replacement assessed by MRI in facioscapulohumeral muscular dystrophy. *Journal of Neurology*, **2019**, 266, 1127-1135	5.5	15
186	Refining the spinobulbar muscular atrophy phenotype by quantitative MRI and clinical assessments. *Neurology*, **2019**, 92, e548-e559	6.5	15
185	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	4.3	14
184	High-intensity interval training in facioscapulohumeral muscular dystrophy type 1: a randomized clinical trial. *Journal of Neurology*, **2017**, 264, 1099-1106	5.5	14
183	MRI in Neuromuscular Diseases: An Emerging Diagnostic Tool and Biomarker for Prognosis and Efficacy. *Annals of Neurology*, **2020**, 88, 669-681	9.4	14
182	Exercise Therapy in Spinobulbar Muscular Atrophy and Other Neuromuscular Disorders. *Journal of Molecular Neuroscience*, **2016**, 58, 388-93	3.3	14
181	Impaired glycogen breakdown and synthesis in phosphoglucomutase 1 deficiency. *Molecular Genetics and Metabolism*, **2017**, 122, 117-121	3.7	14
180	A New Mouse Model of Limb-Girdle Muscular Dystrophy Type 2I Homozygous for the Common L276I Mutation Mimicking the Mild Phenotype in Humans. *Journal of Neuropathology and Experimental Neurology*, **2015**, 74, 1137-46	3.1	14
179	Aerobic training in patients with anoctamin 5 myopathy and hyperckemia. *Muscle and Nerve*, **2014**, 50, 119-23	3.4	14

178	Aerobic Training in Patients with Congenital Myopathy. *PLoS ONE*, **2016**, 11, e0146036	3.7	14
177	Mitochondrial dysfunction and risk of cancer. *British Journal of Cancer*, **2015**, 112, 1134-40	8.7	13
176	Phenotype and genotype of muscle ryanodine receptor rhabdomyolysis-myalgia syndrome. *Acta Neurologica Scandinavica*, **2018**, 137, 452-461	3.8	13
175	Effect of enzyme replacement therapy on isokinetic strength for all major muscle groups in four patients with Pompe disease-a long-term follow-up. *Molecular Genetics and Metabolism*, **2014**, 112, 40-3	3.7	13
174	Severe axial myopathy in McArdle disease. *JAMA Neurology*, **2014**, 71, 88-90	17.2	13
173	Muscle phenotype in patients with myotonic dystrophy type 1. *Muscle and Nerve*, **2013**, 47, 409-15	3.4	13
172	Sequence variants in SPAST, SPG3A and HSPD1 in hereditary spastic paraplegia. *Journal of the Neurological Sciences*, **2009**, 284, 90-5	3.2	13
171	Impaired energy metabolism and abnormal muscle histology in mut- methylmalonic aciduria. *Neurology*, **2005**, 65, 931-3	6.5	13
170	Ventromedial hypothalamic regulation of hormonal and metabolic responses to exercise. *American Journal of Physiology - Regulatory Integrative and Comparative Physiology*, **1989**, 256, R1019-26	3.2	13
169	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	2.6	13
168	Intact transferrin and total plasma glycoprofiling for diagnosis and therapy monitoring in phosphoglucomutase-I deficiency. *Translational Research*, **2018**, 199, 62-76	11	13
167	POPDC3 Gene Variants Associate with a New Form of Limb Girdle Muscular Dystrophy. *Annals of Neurology*, **2019**, 86, 832-843	9.4	12
166	Becker muscular dystrophy with widespread muscle hypertrophy and a non-sense mutation of exon 2. *Neuromuscular Disorders*, **2013**, 23, 25-8	2.9	12
165	Muscle regeneration and inflammation in patients with facioscapulohumeral muscular dystrophy. *Acta Neurologica Scandinavica*, **2013**, 128, 194-201	3.8	12
164	Phenotype and clinical course in a family with a new de novo Twinkle gene mutation. *Neuromuscular Disorders*, **2008**, 18, 306-9	2.9	12
163	Paternal comeback in mitochondrial DNA inheritance. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 1475-1476	11.5	12
162	Treatment Opportunities in Patients With Metabolic Myopathies. *Current Treatment Options in Neurology*, **2017**, 19, 37	4.4	11
161	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,	7.9	11

160	Protein-carbohydrate supplements improve muscle protein balance in muscular dystrophy patients after endurance exercise: a placebo-controlled crossover study. *American Journal of Physiology - Regulatory Integrative and Comparative Physiology*, **2015**, 308, R123-30	3.2	11
159	MMinimal symptom expressionMn patients with acetylcholine receptor antibody-positive refractory generalized myasthenia gravis treated with eculizumab. *Journal of Neurology*, **2020**, 267, 1991-2001	5.5	11
158	Reliability of the 2- and 6-minute walk tests in neuromuscular diseases. *Journal of Rehabilitation Medicine*, **2017**, 49, 362-366	3.4	11
157	A novel de novo mutation of the mitochondrial tRNAlys gene mt.8340G>a associated with pure myopathy. *Neuromuscular Disorders*, **2014**, 24, 162-6	2.9	11
156	Muscle glycogen synthesis and breakdown are both impaired in glycogenin-1 deficiency. *Neurology*, **2017**, 89, 2491-2494	6.5	11
155	A pilot study of muscle plasma protein changes after exercise. *Muscle and Nerve*, **2014**, 49, 261-6	3.4	11
154	Fat metabolism during exercise in patients with mitochondrial disease. *Archives of Neurology*, **2009**, 66, 365-70		11
153	Differential glucose metabolism in mice and humans affected by McArdle disease. *American Journal of Physiology - Regulatory Integrative and Comparative Physiology*, **2016**, 311, R307-14	3.2	10
152	Lack of IL-6 production during exercise in patients with mitochondrial myopathy. *European Journal of Applied Physiology*, **2001**, 84, 155-7	3.4	10
151	Recurrent TTN metatranscript-only c.39974-11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. *Human Mutation*, **2020**, 41, 403-411	4.7	10
150	Zilucoplan: An Investigational Complement C5 Inhibitor for the Treatment of Acetylcholine Receptor Autoantibody-Positive Generalized Myasthenia Gravis. *Expert Opinion on Investigational Drugs*, **2021**, 30, 483-493	5.9	10
149	Body weight-supported training in Becker and limb girdle 2I muscular dystrophy. *Muscle and Nerve*, **2016**, 54, 239-43	3.4	10
148	L-Carnitine Improves Skeletal Muscle Fat Oxidation in Primary Carnitine Deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 4580-4588	5.6	10
147	Disease progression and outcome measures in spinobulbar muscular atrophy. *Annals of Neurology*, **2018**, 84, 754-765	9.4	10
146	Muscle contractility in spinobulbar muscular atrophy. *Scientific Reports*, **2019**, 9, 4680	4.9	9
145	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	7.3	9
144	Muscle imaging in patients with tubular aggregate myopathy caused by mutations in STIM1. *Neuromuscular Disorders*, **2015**, 25, 898-903	2.9	9
143	Risk of cancer in relatives of patients with myotonic dystrophy: a population-based cohort study. *European Journal of Neurology*, **2014**, 21, 1192-7	6	9

142	Exercise intolerance in mitochondrial myopathy is not related to lactic acidosis. *Annals of Neurology*, **2001**, 49, 672-6	9.4	9
141	Lactate and Energy Metabolism During Exercise in Patients With Blocked Glycogenolysis (McArdle Disease). *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E1096-104	5.6	8
140	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	4.7	8
139	Permanent muscle weakness in hypokalemic periodic paralysis. *Neurology*, **2020**, 95, e342-e352	6.5	8
138	BAG3 myopathy is not always associated with cardiomyopathy. *Neuromuscular Disorders*, **2018**, 28, 798-801	2.9	8
137	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	3.2	8
136	No effect of triheptanoin on exercise performance in McArdle disease. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1949-1960	5.3	8
135	Frequency and phenotype of patients carrying TPM2 and TPM3 gene mutations in a cohort of 94 patients with congenital myopathy. *Neuromuscular Disorders*, **2014**, 24, 325-30	2.9	8
134	Effects of Sildenafil on Cerebrovascular Reactivity in Patients with Becker Muscular Dystrophy. *Neurotherapeutics*, **2017**, 14, 182-190	6.4	8
133	A mitochondrial tRNA(Met) mutation causing developmental delay, exercise intolerance and limb girdle phenotype with onset in early childhood. *European Journal of Paediatric Neurology*, **2015**, 19, 69-71	3.8	7
132	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	2.9	7
131	Towards a European Registry and Biorepository for Patients with Spinal and Bulbar Muscular Atrophy. *Journal of Molecular Neuroscience*, **2016**, 58, 394-400	3.3	7
130	Level of residual enzyme activity modulates the phenotype in phosphoglycerate kinase deficiency. *Neurology*, **2018**, 91, e1077-e1082	6.5	7
129	Hydroxylated Long-Chain Acylcarnitines are Biomarkers of Mitochondrial Myopathy. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 5968-5976	5.6	7
128	Myopathic EMG findings and type II muscle fiber atrophy in patients with Lambert-Eaton myasthenic syndrome. *Clinical Neurophysiology*, **2013**, 124, 1889-92	4.3	7
127	Insulin resistance and increased muscle cytokine levels in patients with mitochondrial myopathy. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, 3757-65	5.6	7
126	Protein turnover and cellular stress in mildly and severely affected muscles from patients with limb girdle muscular dystrophy type 2I. *PLoS ONE*, **2013**, 8, e66929	3.7	7
125	Data from the European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC). *Orphanet Journal of Rare Diseases*, **2020**, 15, 330	4.2	7

124	Exercise training in metabolic myopathies. *Revue Neurologique*, **2016**, 172, 559-565	3	7
123	Homozygosity for Arg1142Gln causes congenital myopathy with variable disease expression. *Neurology: Genetics*, **2018**, 4, e267	3.8	7
122	Nampt controls skeletal muscle development by maintaining Ca homeostasis and mitochondrial integrity. *Molecular Metabolism*, **2021**, 53, 101271	8.8	7
121	1st International Workshop on Clinical trial readiness for sarcoglycanopathies 15-16 November 2016, Evry, France. *Neuromuscular Disorders*, **2017**, 27, 683-692	2.9	6
120	Dysphagia is prevalent in patients with CPEO and single, large-scale deletions in mtDNA. *Mitochondrion*, **2017**, 32, 27-30	4.9	6
119	Impairments in contractility and cytoskeletal organisation cause nuclear defects in nemaline myopathy. *Acta Neuropathologica*, **2019**, 138, 477-495	14.3	6
118	High-intensity training in patients with spinal and bulbar muscular atrophy. *Journal of Neurology*, **2019**, 266, 1693-1697	5.5	6
117	Low survival rate and muscle fiber-dependent aging effects in the McArdle disease mouse model. *Scientific Reports*, **2019**, 9, 5116	4.9	6
116	Congenital myopathies are mainly associated with a mild cardiac phenotype. *Journal of Neurology*, **2019**, 266, 1367-1375	5.5	6
115	Aberrant regulation of epigenetic modifiers contributes to the pathogenesis in patients with selenoprotein N-related myopathies. *Human Mutation*, **2019**, 40, 962-974	4.7	6
114	Evaluation of inflammatory lesions over 2 years in facioscapulohumeral muscular dystrophy. *Neurology*, **2020**, 95, e1211-e1221	6.5	6
113	Titrating a modified ketogenic diet for patients with McArdle disease: A pilot study. *Journal of Inherited Metabolic Disease*, **2020**, 43, 778-786	5.4	6
112	Lecocytes mutation load declines with age in carriers of the m.3243A>G mutation: A 10-year Prospective Cohort. *Clinical Genetics*, **2018**, 93, 925-928	4	6
111	Frequency and Phenotype of Myotubular Myopathy Amongst Danish Patients with Congenital Myopathy Older than 5 Years. *Journal of Neuromuscular Diseases*, **2015**, 2, 167-174	5	6
110	Influence of erythrocyte oxygenation and intravascular ATP on resting and exercising skeletal muscle blood flow in humans with mitochondrial myopathy. *Mitochondrion*, **2012**, 12, 414-22	4.9	6
109	High-resolution melting facilitates mutation screening of PYGM in patients with McArdle disease. *Annals of Human Genetics*, **2009**, 73, 292-7	2.2	6
108	Effect of anaesthetizing the region of the paraventricular hypothalamic nuclei on energy metabolism during exercise in the rat. *Acta Physiologica Scandinavica*, **1994**, 151, 165-72		6
107	Exercise therapy for muscle and lower motor neuron diseases. *Acta Myologica*, **2019**, 38, 215-232	1.6	6

106	Consistent improvement with eculizumab across muscle groups in myasthenia gravis. *Annals of Clinical and Translational Neurology*, **2020**, 7, 1327-1339	5.3	6
105	Stable Longitudinal Methylation Levels at the CpG Sites Flanking the CTG Repeat of in Patients with Myotonic Dystrophy Type 1. *Genes*, **2020**, 11,	4.2	6
104	Muscle phosphoglycerate mutase deficiency with tubular aggregates: effect of dantrolene. *Annals of Neurology*, **1999**, 46, 274-7	9.4	6
103	Muscle reflex and central motor control of neuroendocrine activity, glucose homeostasis and circulation during exercise. *Acta Physiologica Scandinavica Supplementum*, **2000**, 647, 1-26		6
102	Visual impairment in anti-GQ1b positive Miller Fisher syndrome. *Acta Neurologica Scandinavica*, **2001**, 103, 259-60	3.8	6
101	Human growth hormone stabilizes walking and improves strength in a patient with dominantly inherited calpainopathy. *Neuromuscular Disorders*, **2017**, 27, 358-362	2.9	5
100	Skeletal muscle metabolism during prolonged exercise in Pompe disease. *Endocrine Connections*, **2017**, 6, 384-394	3.5	5
99	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	6.3	5
98	Impaired lipolysis in propionic acidemia: A new metabolic myopathy?. *JIMD Reports*, **2020**, 53, 16-21	1.9	5
97	Correlation between myasthenia gravis-activities of daily living (MG-ADL) and quantitative myasthenia gravis (QMG) assessments of anti-acetylcholine receptor antibody-positive refractory generalized myasthenia gravis in the phase 3 regain study. *Muscle and Nerve*, **2018**, 58, E21-E22	3.4	5
96	Collagen XII myopathy with rectus femoris atrophy and collagen XII retention in fibroblasts. *Muscle and Nerve*, **2018**, 57, 1026-1030	3.4	5
95	A new mutation of the fukutin gene causing late-onset limb girdle muscular dystrophy. *Neuromuscular Disorders*, **2013**, 23, 562-7	2.9	5
94	Muscle regeneration in mitochondrial myopathies. *Mitochondrion*, **2013**, 13, 63-70	4.9	5
93	LGMD2L with bone affection: overlapping phenotype of dominant and recessive ANO5-induced disease. *Muscle and Nerve*, **2012**, 46, 829-30	3.4	5
92	Diagnostic challenges in combined multiple sclerosis and centronuclear myopathy. *European Journal of Neurology*, **2000**, 7, 567-71	6	5
91	Deletion of exon 26 of the dystrophin gene is associated with a mild Becker muscular dystrophy phenotype. *Acta Myologica*, **2011**, 30, 182-4	1.6	5
90	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,	5.1	5
89	Antimyostatin Treatment in Health and Disease: The Story of Great Expectations and Limited Success. *Cells*, **2021**, 10,	7.9	5

88	Clinical and neurophysiological response to pharmacological treatment of DOK7 congenital myasthenia in an older patient. *Clinical Neurology and Neurosurgery*, **2015**, 130, 168-70	2	4
87	Mutation Load of Single, Large-Scale Deletions of mtDNA in Mitotic and Postmitotic Tissues. *Frontiers in Genetics*, **2020**, 11, 547638	4.5	4
86	A quantitative method to assess muscle edema using short TI inversion recovery MRI. *Scientific Reports*, **2020**, 10, 7246	4.9	4
85	Characteristic muscle signatures assessed by quantitative MRI in patients with Bethlem myopathy. *Journal of Neurology*, **2020**, 267, 2432-2442	5.5	4
84	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	5.3	4
83	Pure exercise intolerance and ophthalmoplegia associated with the m.12,294G□□A mutation in the MT-TL2 gene: a case report. *BMC Musculoskeletal Disorders*, **2017**, 18, 419	2.8	4
82	Screening for late-onset Pompe disease in western Denmark. *Acta Neurologica Scandinavica*, **2018**, 137, 85-90	3.8	4
81	Reply: Dominant LGMD2A: alternative diagnosis or hidden digenism?. *Brain*, **2017**, 140, e8	11.2	4
80	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. *European Journal of Neurology*, **2020**, 27, 2604-2615	6	4
79	Preclinical Research in Glycogen Storage Diseases: A Comprehensive Review of Current Animal Models. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	4
78	Effect of Gender, Disease Duration and Treatment on Muscle Strength in Myasthenia Gravis. *PLoS ONE*, **2016**, 11, e0164092	3.7	4
77	Fat oxidation is impaired during exercise in lipin-1 deficiency. *Neurology*, **2019**, 93, e1433-e1438	6.5	4
76	Expanding the phenotype of filamin-C-related myofibrillar myopathy. *Clinical Neurology and Neurosurgery*, **2019**, 176, 30-33	2	4
75	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	5	4
74	Exercising with blocked muscle glycogenolysis: Adaptation in the McArdle mouse. *Molecular Genetics and Metabolism*, **2018**, 123, 21-27	3.7	4
73	Leber hereditary optic neuropathy due to a new ND1 mutation. *Ophthalmic Genetics*, **2017**, 38, 480-485	1.2	3
72	Aerobic training in myotonia congenita: Effect on myotonia and fitness. *Muscle and Nerve*, **2017**, 56, 696-699	3.4	3
71	Moderate-intensity aerobic exercise improves physical fitness in bethlem myopathy. *Muscle and Nerve*, **2019**, 60, 183-188	3.4	3

70	Exercise Intolerance and Myoglobinuria Associated with a Novel Maternally Inherited MT-ND1 Mutation. *JIMD Reports*, **2016**, 25, 65-70	1.9	3
69	Physical activity in myotonic dystrophy type 1. *Journal of Neurology*, **2020**, 267, 1679-1686	5.5	3
68	233rd ENMC International Workshop:: Clinical Trial Readiness for Calpainopathies, Naarden, The Netherlands, 15-17 September 2017. *Neuromuscular Disorders*, **2018**, 28, 540-549	2.9	3
67	211th ENMC International Workshop:: Development of diagnostic criteria and management strategies for McArdle Disease and related rare glycogenolytic disorders to improve standards of care. 17-19 April 2015, Naarden, The Netherlands. *Neuromuscular Disorders*, **2017**, 27, 1143-1151	2.9	3
66	Aerobic anti-gravity exercise in patients with Charcot-Marie-Tooth disease types 1A and X: A pilot study. *Brain and Behavior*, **2017**, 7, e00794	3.4	3
65	O.22 Dominant inheritance of limb girdle muscular dystrophy type 2A. *Neuromuscular Disorders*, **2011**, 21, 750	2.9	3
64	Myositis in Griscelli syndrome type 2 treated with hematopoietic cell transplantation. *Neuromuscular Disorders*, **2010**, 20, 136-8	2.9	3
63	Late-onset MADD: a rare cause of cirrhosis and acute liver failure?. *Acta Myologica*, **2020**, 39, 19-23	1.6	3
62	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,	5.1	3
61	Quantitative Muscle MRI as Outcome Measure in Patients With Becker Muscular Dystrophy-A 1-Year Follow-Up Study. *Frontiers in Neurology*, **2020**, 11, 613489	4.1	3
60	DOK7 congenital myasthenia may be associated with severe mitral valve insufficiency. *Journal of the Neurological Sciences*, **2017**, 379, 217-218	3.2	2
59	Contractile properties are impaired in congenital myopathies. *Neuromuscular Disorders*, **2020**, 30, 649-655	2.9	2
58	Cytokine genes as potential biomarkers for muscle weakness in OPMD. *Human Molecular Genetics*, **2016**, 25, 4282-4287	5.6	2
57	Muscle contractility of leg muscles in patients with mitochondrial myopathies. *Mitochondrion*, **2019**, 46, 221-227	4.9	2
56	Mitochondrial DNA mutation load in a family with the m.8344A>G point mutation and lipomas: a case study. *Clinical Case Reports (discontinued)*, **2017**, 5, 2034-2039	0.7	2
55	Effect of prior immobilization on muscular glucose clearance in resting and running rats. *American Journal of Physiology - Endocrinology and Metabolism*, **1988**, 255, E456-62	6	2
54	Mitochondrial mutation m.3243A>G associates with insulin resistance in non-diabetic carriers. *Endocrine Connections*, **2019**, 8, 829-837	3.5	2
53	Phenotypic Spectrum of 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	3.1	2

52	Impaired fat oxidation during exercise in multiple acyl-CoA dehydrogenase deficiency. *JIMD Reports* , **2019**, 46, 79-84	1.9	2
51	Vacuoles, Often Containing Glycogen, Are a Consistent Finding in Hypokalemic Periodic Paralysis. *Journal of Neuropathology and Experimental Neurology*, **2020**, 79, 1127-1129	3.1	2
50	249th ENMC International Workshop: The role of brain dystrophin in muscular dystrophy: Implications for clinical care and translational research, Hoofddorp, The Netherlands, November 29th-December 1st 2019. *Neuromuscular Disorders*, **2020**, 30, 782-794	2.9	2
49	No effect of oral sucrose or IV glucose during exercise in phosphorylase b kinase deficiency. *Neuromuscular Disorders*, **2020**, 30, 340-345	2.9	2
48	Myopathy can be a key phenotype of membrin (GOSR2) deficiency. *Human Mutation*, **2021**, 42, 1101-1106	4.7	2
47	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	5	2
46	The Pathophysiology of Exercise and Effect of Training in Mitochondrial Myopathies **2019**, 331-348		1
45	A New Glycogen Storage Disease Caused by a Dominant PYGM Mutation. *Annals of Neurology*, **2020** , 88, 274-282	9.4	1
44	Intrarater reliability and validity of outcome measures in myotonic dystrophy type 1. *Neurology*, **2020**, 94, e2508-e2520	6.5	1
43	Results of an open label feasibility study of sodium valproate in people with McArdle disease. *Neuromuscular Disorders*, **2020**, 30, 734-741	2.9	1
42	Depletion of ATP Limits Membrane Excitability of Skeletal Muscle by Increasing Both ClC1-Open Probability and Membrane Conductance. *Frontiers in Neurology*, **2020**, 11, 541	4.1	1
41	Polymyositis following autologous haematopoietic stem cell transplantation. *Scandinavian Journal of Rheumatology*, **2016**, 45, 429-31	1.9	1
40	Altered somatosensory neurovascular response in patients with Becker muscular dystrophy. *Brain and Behavior*, **2018**, 8, e00985	3.4	1
39	Muscle biopsies off-set normal cellular signaling in surrounding musculature. *Neuromuscular Disorders*, **2013**, 23, 981-5	2.9	1
38	Metabolic myopathies390-408		1
37	Extreme Hypoxia Causing Brady-Arrythmias During Apnea in Elite Breath-Hold Divers.. *Frontiers in Physiology*, **2021**, 12, 712573	4.6	1
36	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	4.2	1
35	Function, structure and quality of striated muscles in the lower extremities in patients with late onset Pompe Disease-an MRI study. *PeerJ*, **2021**, 9, e10928	3.1	1

34	No effect of resveratrol in patients with mitochondrial myopathy: A cross-over randomized controlled trial. *Journal of Inherited Metabolic Disease*, **2021**, 44, 1186-1198	5.4	1
33	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	5	1
32	1st FSHD European Trial Network workshop:Working towards trial readiness across Europe. *Neuromuscular Disorders*, **2021**, 31, 907-918	2.9	1
31	Absence of p.R50X read-through in McArdle disease cellular models. *DMM Disease Models and Mechanisms*, **2020**, 13,	4.1	1
30	251st ENMC international workshop: Polyglucosan storage myopathies 13-15 December 2019, Hoofddorp, the Netherlands. *Neuromuscular Disorders*, **2021**, 31, 466-477	2.9	1
29	Progression or Not - A Small Natural History Study of Genetical Confirmed Congenital Myopathies. *Journal of Neuromuscular Diseases*, **2021**, 8, 647-655	5	1
28	Fatigue, physical activity and associated factors in 779 patients with myasthenia gravis. *Neuromuscular Disorders*, **2021**, 31, 716-725	2.9	1
27	Diagnostic interest of whole-body MRI in early- and late-onset LAMA2 muscular dystrophies: a large international cohort. *Journal of Neurology*, **2021**, 1	5.5	1
26	Quantitative Muscle MRI and Clinical Findings in Women With Pathogenic Dystrophin Gene Variants. *Frontiers in Neurology*, **2021**, 12, 707837	4.1	1
25	Eculizumab in refractory generalized myasthenia gravis previously treated with rituximab: subgroup analysis of REGAIN and its extension study. *Muscle and Nerve*, **2021**, 64, 662-669	3.4	1
24	Disorders of Muscle Glycogen Metabolism254-264		1
23	High Resolution Analysis of DMPK Hypermethylation and Repeat Interruptions in Myotonic Dystrophy Type 1. *Genes*, **2022**, 13, 970	4.2	1
22	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	5.6	0
21	Causes of symptom dissatisfaction in patients with generalized myasthenia gravis. *Journal of Neurology*, **2021**, 1	5.5	0
20	Muscle biopsy and MRI findings in ANO5-related myopathy. *Muscle and Nerve*, **2021**, 64, 743-748	3.4	0
19	Responsiveness of outcome measures in myotonic dystrophy type 1. *Annals of Clinical and Translational Neurology*, **2020**, 7, 1382-1391	5.3	0
18	Patients With Becker Muscular Dystrophy Have Severe Paraspinal Muscle Involvement. *Frontiers in Neurology*, **2021**, 12, 613483	4.1	0
17	Autophagy is affected in patients with hypokalemic periodic paralysis: an involvement in vacuolar myopathy?. *Acta Neuropathologica Communications*, **2021**, 9, 109	7.3	0

16	Cardiac Involvement in Women With Pathogenic Dystrophin Gene Variants. *Frontiers in Neurology*, **2021**, 12, 707838	4.1	0
15	Episodic hyperCKaemia may be a feature of □methylacyl-coenzyme A racemase deficiency. *European Journal of Neurology*, **2021**, 28, 729-731	6	0
14	Muscle involvement assessed by quantitative magnetic resonance imaging in patients with anoctamin 5 deficiency. *European Journal of Neurology*, **2021**, 28, 3121-3132	6	0
13	Prolonged fasting-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	3.1	0
12	Botulinum toxin treatment improves dysphagia in patients with oculopharyngeal muscular dystrophy and sporadic inclusion body myositis.. *Journal of Neurology*, **2022**, 1	5.5	0
11	Response letter to "Cardiac involvement in myotonic dystrophy type 1--do not forget the loop recorder!". *International Journal of Cardiology*, **2013**, 168, 1541	3.2	
10	Response. *Neuromuscular Disorders*, **2013**, 23, 193	2.9	
9	Differences in genetic defects and morphology of eye- and limb muscles in mitochondrial myopathy. *Acta Ophthalmologica*, **2015**, 93, e306-8	3.7	
8	Cardiac fibrosis in myotonic dystrophy type 1; an early marker of cardiac involvement. *European Heart Journal*, **2013**, 34, P2987-P2987	9.5	
7	Visual impairment in anti-GQ1b positive Miller Fisher syndrome. *Acta Neurologica Scandinavica*, **2001**, 103, 259-260	3.8	
6	Role of metabolic feedback regulation in glucose production of running rats. *American Journal of Physiology - Regulatory Integrative and Comparative Physiology*, **1988**, 255, R400-6	3.2	
5	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	0.2	
4	Combined Muscle Biopsy and Comprehensive Electrophysiology in General Anesthesia is Valuable in Diagnosis of Neuromuscular Disease in Children. *Neuropediatrics*, **2021**, 52, 462-468	1.6	
3	Energy metabolism during exercise in patients with □enolase deficiency (GSDXIII). *JIMD Reports*, **2021**, 61, 60-66	1.9	
2	Effects of rhythmic auditory stimulation on walking during the 6-minute walk test in patients with generalised Myasthenia Gravis. *European Journal of Physiotherapy*,1-000	0.5	
1	Plasma lactate responses during and after submaximal handgrip exercise are not diagnostically helpful in mitochondrial myopathy. *Mitochondrion*, **2021**, 60, 21-26	4.9	