Benedikt Schoser

List of Publications by Year
in descending order

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Version: 2024-02-01

204 papers	9,463 citations	30070 54 h-index	48315 88 g-index

246 all docs	246 docs citations	246 times ranked	10230 citing authors

#	Article	IF	Citations
1	Non-ATGâ€“initiated translation directed by microsatellite expansions. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 260-265.	7.1	826
2	Misregulated alternative splicing of BIN1 is associated with T tubule alterations and muscle weakness in myotonic dystrophy. Nature Medicine, 2011, 17, 720-725.	30.7	299
3	Anti-LRP4 autoantibodies in AChR- and MuSK-antibody-negative myasthenia gravis. Journal of Neurology, 2012, 259, 427-435.	3.6	297
4	Transplantation of Genetically Corrected Human iPSC-Derived Progenitors in Mice with Limb-Girdle Muscular Dystrophy. Science Translational Medicine, 2012, 4, 140ra89.	12.4	269
5	Enzyme replacement therapy with alglucosidase alfa in 44 patients with late-onset glycogen storage disease type 2: 12-month results of an observational clinical trial. Journal of Neurology, 2010, 257, 91-97.	3.6	229
6	Late onset Pompe disease: Clinical and neurophysiological spectrum of 38 patients including long-term follow-up in 18 patients. Neuromuscular Disorders, 2007, 17, 698-706.	0.6	208
7	Survival and long-term outcomes in late-onset Pompe disease following alglucosidase alfa treatment: a systematic review and meta-analysis. Journal of Neurology, 2017, 264, 621-630.	3.6	183
8	Enzyme replacement therapy in late-onset Pompe disease: a systematic literature review. Journal of Neurology, 2013, 260, 951-959.	3.6	168
9	Myofibrillar Myopathies: A Clinical and Myopathological Guide. Brain Pathology, 2009, 19, 483-492.	4.1	164
10	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. American Journal of Human Genetics, 2011, 88, 162-172.	6.2	153
11	Anti-agrin autoantibodies in myasthenia gravis. Neurology, 2014, 82, 1976-1983.	1.1	151
12	An X-Linked Myopathy with Postural Muscle Atrophy and Generalized Hypertrophy, Termed XMPMA, Is Caused by Mutations in FHL1. American Journal of Human Genetics, 2008, 82, 88-99.	6.2	148
13	Dystrophin-deficient pigs provide new insights into the hierarchy of physiological derangements of dystrophic muscle. Human Molecular Genetics, 2013, 22, 4368-4382.	2.9	134
14	Polyglucosan body myopathy caused by defective ubiquitin ligase RBCK1. Annals of Neurology, 2013, 74, 914-919.	5.3	132
15	Safety and Treatment Effects of Nusinersen in Longstanding Adult 5q-SMA Type 3 â€“ A Prospective Observational Study. Journal of Neuromuscular Diseases, 2019, 6, 453-465.	2.6	132
16	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
17	Consensus-based care recommendations for adults with myotonic dystrophy type 1. Neurology: Clinical Practice, 2018, 8, 507-520.	1.6	115
18	MSH3 modifies somatic instability and disease severity in Huntingtonâ€™s and myotonic dystrophy type 1. Brain, 2019, 142, 1876-1886.	7.6	114

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19	36 months observational clinical study of 38 adult Pompe disease patients under alglucosidase alfa enzyme replacement therapy. Journal of Inherited Metabolic Disease, 2012, 35, 837-845.	3.6	109
20	Core Clinical Phenotypes in Myotonic Dystrophies. Frontiers in Neurology, 2018, 9, 303.	2.4	104
21	A systematic review on the definition of rhabdomyolysis. Journal of Neurology, 2020, 267, 877-882.	3.6	101
22	Spinal poly-GA inclusions in a C9orf72 mouse model trigger motor deficits and inflammation without neuron loss. Acta Neuropathologica, 2017, 134, 241-254.	7.7	99
23	Recessive TRAPPC11 Mutations Cause a Disease Spectrum of Limb Girdle Muscular Dystrophy and Myopathy with Movement Disorder and Intellectual Disability. American Journal of Human Genetics, 2013, 93, 181-190.	6.2	98
24	Commonality ofTRIM32mutation in causing sarcotubular myopathy and LGMD2H. Annals of Neurology, 2005, 57, 591-595.	5.3	96
25	Cognitive behavioural therapy with optional graded exercise therapy in patients with severe fatigue with myotonic dystrophy type 1: a multicentre, single-blind, randomised trial. Lancet Neurology, The, 2018, 17, 671-680.	10.2	95
26	Toward deconstructing the phenotype of lateâ€onset Pompe disease. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 80-88.	1.6	92
27	Ectopic expression of cyclin D3 corrects differentiation of DM1 myoblasts through activation of RNA CUG-binding protein, CUGBP1. Experimental Cell Research, 2008, 314, 2266-2278.	2.6	89
28	The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. Journal of Neurology, 2011, 258, 1987-1997.	3.6	87
29	Expansion of CUG RNA repeats causes stress and inhibition of translation in myotonic dystrophy 1 (DM1) cells. FASEB Journal, 2010, 24, 3706-3719.	0.5	86
30	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
31	Diagnostic approach for FSHD revisited: SMCHD1 mutations cause FSHD2 and act as modifiers of disease severity in FSHD1. European Journal of Human Genetics, 2015, 23, 808-816.	2.8	83
32	Myotonic Dystrophies 1 and 2: Complex Diseases with Complex Mechanisms. Current Genomics, 2010, 11, 77-90.	1.6	82
33	Reduction of the Rate of Protein Translation in Patients with Myotonic Dystrophy 2. Journal of Neuroscience, 2009, 29, 9042-9049.	3.6	81
34	The p.G154S mutation of the alpha-B crystallin gene (CRYAB) causes late-onset distal myopathy. Neuromuscular Disorders, 2010, 20, 255-259.	0.6	81
35	Expression of RNA CCUG Repeats Dysregulates Translation and Degradation of Proteins in Myotonic Dystrophy 2 Patients. American Journal of Pathology, 2009, 175, 748-762.	3.8	77
36	Muscle MRI findings in limb girdle muscular dystrophy type 2L. Neuromuscular Disorders, 2012, 22, S122-S129.	0.6	77

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37	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850.	3.6	72
38	Genetic determinants of disease severity in the myotonic dystrophy type 1 OPTIMISTIC cohort. Neurology, 2019, 93, e995-e1009.	1.1	71
39	Therapeutic Approaches in Glycogen Storage Disease Type II/Pompe Disease. Neurotherapeutics, 2008, 5, 569-578.	4.4	69
40	Maximum inspiratory pressure as a clinically meaningful trial endpoint for neuromuscular diseases: a comprehensive review of the literature. Orphanet Journal of Rare Diseases, 2017, 12, 52.	2.7	69
41	Mutations in INPP5K , Encoding a Phosphoinositide 5-Phosphatase, Cause Congenital Muscular Dystrophy with Cataracts and Mild Cognitive Impairment. American Journal of Human Genetics, 2017, 100, 523-536.	6.2	67
42	The impact of antibodies in late-onset Pompe disease: A case series and literature review. Molecular Genetics and Metabolism, 2012, 106, 301-309.	1.1	66
43	Deficiency of the mitochondrial phosphate carrier presenting as myopathy and cardiomyopathy in a family with three affected children. Neuromuscular Disorders, 2011, 21, 803-808.	0.6	65
44	Lambertâ€“Eaton myasthenic syndrome (LEMS): a rare autoimmune presynaptic disorder often associated with cancer. Journal of Neurology, 2017, 264, 1854-1863.	3.6	65
45	Absence of a differentiation defect in muscle satellite cells from DM2 patients. Neurobiology of Disease, 2009, 36, 181-190.	4.4	64
46	The frequency and severity of cardiac involvement in myotonic dystrophy type 2 (DM2): Long-term outcomes. International Journal of Cardiology, 2013, 168, 1147-1153.	1.7	64
47	ANO5Gene Analysis in a Large Cohort of Patients with Anoctaminopathy: Confirmation of Male Prevalence and High Occurrence of the Common Exon 5 Gene Mutation. Human Mutation, 2013, 34, 1111-1118.	2.5	64
48	Strumpellin is a novel valosin-containing protein binding partner linking hereditary spastic paraplegia to protein aggregation diseases. Brain, 2010, 133, 2920-2941.	7.6	62
49	Adult-onset glycogen storage disease type 2: clinico-pathological phenotype revisited. Neuropathology and Applied Neurobiology, 2007, 33, 070615152525006-???.	3.2	61
50	Consequences of mutations within the C terminus of the FHL1 gene. Neurology, 2009, 73, 543-551.	1.1	61
51	rbFOX1/MBNL1 competition for CCUG RNA repeats bindingÂcontributes to myotonic dystrophy typeÂ1/typeÂ2 differences. Nature Communications, 2018, 9, 2009.	12.8	61
52	Prevalence of Pompe disease in 3,076 patients with hyperCKemia and limb-girdle muscular weakness. Neurology, 2016, 87, 295-298.	1.1	60
53	Skeletal Muscle Pathology in X-Linked Myotubular Myopathy: Review With Cross-Species Comparisons. Journal of Neuropathology and Experimental Neurology, 2016, 75, 102-110.	1.7	59
54	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

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55	Safety and efficacy of avalglucosidase alfa versus alglucosidase alfa in patients with late-onset Pompe disease (COMET): a phase 3, randomised, multicentre trial. Lancet Neurology, The, 2021, 20, 1012-1026.	10.2	59
56	Utility of a next-generation sequencing-based gene panel investigation in German patients with genetically unclassified limb-girdle muscular dystrophy. Journal of Neurology, 2016, 263, 743-750.	3.6	57
57	Nemaline myopathy caused by mutations in the nebulin gene may present as a distal myopathy. Neuromuscular Disorders, 2011, 21, 556-562.	0.6	56
58	High frequency of co-segregating CLCN1 mutations among myotonic dystrophy type 2 patients from Finland and Germany. Journal of Neurology, 2008, 255, 1731-1736.	3.6	55
59	The value of muscle biopsies in Pompe disease: identifying lipofuscin inclusions in juvenile- and adult-onset patients. Acta Neuropathologica Communications, 2014, 2, 2.	5.2	55
60	The clinical relevance of outcomes used in late-onset Pompe disease: can we do better?. Orphanet Journal of Rare Diseases, 2013, 8, 160.	2.7	54
61	Pharmacological and nutritional treatment for McArdle disease (Glycogen Storage Disease type V). The Cochrane Library, 2014, 2014, CD003458.	2.8	54
62	Differential roles of hypoxia and innate immunity in juvenile and adult dermatomyositis. Acta Neuropathologica Communications, 2016, 4, 45.	5.2	52
63	Amifampridine phosphate (Firdapse^{Â®}) is effective and safe in a phase 3 clinical trial in LEMS. Muscle and Nerve, 2016, 53, 717-725.	2.2	51
64	New insights into the protein aggregation pathology in myotilinopathy by combined proteomic and immunolocalization analyses. Acta Neuropathologica Communications, 2016, 4, 8.	5.2	50
65	Cognitive behaviour therapy plus aerobic exercise training to increase activity in patients with myotonic dystrophy type 1 (DM1) compared to usual care (OPTIMISTIC): study protocol for randomised controlled trial. Trials, 2015, 16, 224.	1.6	49
66	Prospective exploratory muscle biopsy, imaging, and functional assessment in patients with late-onset Pompe disease treated with alglucosidase alfa: The EMBASSY Study. Molecular Genetics and Metabolism, 2016, 119, 115-123.	1.1	49
67	Sleepâ€related symptoms and sleepâ€disordered breathing in adult <scp>P</scp>ompe disease. European Journal of Neurology, 2015, 22, 369.	3.3	48
68	50 years to diagnosis: Autosomal dominant tubular aggregate myopathy caused by a novel STIM1 mutation. Neuromuscular Disorders, 2015, 25, 577-584.	0.6	47
69	Reducing Body Myopathy and Other FHL1-Related Muscular Disorders. Seminars in Pediatric Neurology, 2011, 18, 257-263.	2.0	44
70	â€œOrbiting aroundâ€•the orbital myositis: clinical features, differential diagnosis and therapy. Journal of Neurology, 2016, 263, 631-640.	3.6	43
71	Safety and efficacy of cipaglucosidase alfa plus miglustat versus alglucosidase alfa plus placebo in late-onset Pompe disease (PROPEL): an international, randomised, double-blind, parallel-group, phase 3 trial. Lancet Neurology, The, 2021, 20, 1027-1037.	10.2	42
72	A new phenotype of brain iron accumulation with dystonia, optic atrophy, and peripheral neuropathy. Movement Disorders, 2012, 27, 789-793.	3.9	41

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73	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	9.0	41
74	Consensus-based care recommendations for adults with myotonic dystrophy type 2. Neurology: Clinical Practice, 2019, 9, 343-353.	1.6	41
75	Novel ANO5 mutations causing hyperâ€CKâ€emia, limb girdle muscular weakness and miyoshi type of muscular dystrophy. Muscle and Nerve, 2012, 45, 740-742.	2.2	40
76	A multistage sequencing strategy pinpoints novel candidate alleles for Emery-Dreifuss muscular dystrophy and supports gene misregulation as its pathomechanism. EBioMedicine, 2020, 51, 102587.	6.1	40
77	Assessing the influence of age and gender on the phenotype of myotonic dystrophy type 2. Journal of Neurology, 2017, 264, 2472-2480.	3.6	38
78	Intragenic deletion ofTRIM32in compound heterozygotes with sarcotubular myopathy/LGMD2H. Human Mutation, 2009, 30, E831-E844.	2.5	37
79	Long-Term Endurance Exercise in Humans Stimulates Cell Fusion of Myoblasts along with Fusogenic Endogenous Retroviral Genes In Vivo. PLoS ONE, 2015, 10, e0132099.	2.5	37
80	Immune-mediated rippling muscle disease with myasthenia gravis: A report of seven patients with long-term follow-up in two. Neuromuscular Disorders, 2009, 19, 223-228.	0.6	36
81	Myotonic Dystrophyâ€"A Progeroid Disease?. Frontiers in Neurology, 2018, 9, 601.	2.4	34
82	RNA Foci, CUGBP1, and ZNF9 Are the Primary Targets of the Mutant CUG and CCUG Repeats Expanded in Myotonic Dystrophies Type 1 and Type 2. American Journal of Pathology, 2011, 179, 2475-2489.	3.8	33
83	A genetic modifier of symptom onset in Pompe disease. EBioMedicine, 2019, 43, 553-561.	6.1	32
84	Pain in adult patients with Pompe disease. Molecular Genetics and Metabolism, 2013, 109, 371-376.	1.1	31
85	The humanistic burden of Pompe disease: are there still unmet needs? A systematic review. BMC Neurology, 2017, 17, 202.	1.8	31
86	Reduction of toxic RNAs in myotonic dystrophies type 1 and type 2 by the RNA helicase p68/DDX5. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 8041-8045.	7.1	30
87	Homozygosity for the common GAA gene splice site mutation c.-32-13T>G in Pompe disease is associated with the classical adult phenotypical spectrum. Neuromuscular Disorders, 2015, 25, 719-724.	0.6	29
88	A novel mutation in the myotilin gene (MYOT) causes a severe form of limb girdle muscular dystrophy 1A (LGMD1A). Journal of Neurology, 2011, 258, 1437-1444.	3.6	27
89	InÂvivo characterization of human myofibrillar myopathy genes in zebrafish. Biochemical and Biophysical Research Communications, 2015, 461, 217-223.	2.1	27
90	Facioscapulohumeral muscular dystrophy presenting with unusual phenotypes and atypical morphological features of vacuolar myopathy. Journal of Neurology, 2010, 257, 1108-1118.	3.6	26

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91	Muscle ultrasound in classic infantile and adult Pompe disease: A useful screening tool in adults but not in infants. Neuromuscular Disorders, 2015, 25, 120-126.	0.6	26
92	Proteomic characterization of aggregate components in an intrafamilial variable FHL1-associated myopathy. Neuromuscular Disorders, 2013, 23, 418-426.	0.6	25
93	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	2.4	25
94	CRISPR-cas gene-editing as plausible treatment of neuromuscular and nucleotide-repeat-expansion diseases: A systematic review. PLoS ONE, 2019, 14, e0212198.	2.5	25
95	A role for PLCÎ²1 in myotonic dystrophies type 1 and 2. FASEB Journal, 2012, 26, 3042-3048.	0.5	24
96	208th ENMC International Workshop: Formation of a European Network to develop a European data sharing model and treatment guidelines for Pompe disease Naarden, The Netherlands, 26â€“28 September 2014. Neuromuscular Disorders, 2015, 25, 674-678.	0.6	24
97	Patient-specific protein aggregates in myofibrillar myopathies: Laser microdissection and differential proteomics for identification of plaque components. Proteomics, 2012, 12, 3598-3609.	2.2	23
98	Sarcopenia â€“ Endocrinological and Neurological Aspects. Experimental and Clinical Endocrinology and Diabetes, 2019, 6, 8-22.	1.2	23
99	Side effects of anesthesia in DM2 as compared to DM1: a comparative retrospective study. European Journal of Neurology, 2010, 17, 842-845.	3.3	22
100	Pregnancy and delivery in women with Pompe disease. Molecular Genetics and Metabolism, 2014, 112, 148-153.	1.1	22
101	Two patients with GMPPB mutation: The overlapping phenotypes of limb-girdle myasthenic syndrome and limb-girdle muscular dystrophy dystroglycanopathy. Muscle and Nerve, 2017, 56, 334-340.	2.2	22
102	Facioscapulohumeral muscular dystrophy and Charcot-Marie-Tooth neuropathy 1A - evidence for â€œdouble troubleâ€•overlapping syndromes. BMC Medical Genetics, 2013, 14, 92.	2.1	21
103	Moss-Derived Human Recombinant GAA Provides an Optimized Enzyme Uptake in Differentiated Human Muscle Cells of Pompe Disease. International Journal of Molecular Sciences, 2020, 21, 2642.	4.1	21
104	STIG study: real-world data of long-term outcomes of adults with Pompe disease under enzyme replacement therapy with alglucosidase alfa. Journal of Neurology, 2021, 268, 2482-2492.	3.6	21
105	Spongious Hypertrophic Cardiomyopathy in Patients With Mutations in the Four-and-a-Half LIM Domain 1 Gene. Circulation: Cardiovascular Genetics, 2012, 5, 490-502.	5.1	20
106	Decreased water T₂ in fatty infiltrated skeletal muscles of patients with neuromuscular diseases. NMR in Biomedicine, 2019, 32, e4111.	2.8	20
107	Type-2 muscle fiber atrophy is associated with sarcopenia in elderly men with hip fracture. Experimental Gerontology, 2021, 144, 111171.	2.8	20
108	Novel recessive myotilin mutation causes severe myofibrillar myopathy. Neurogenetics, 2014, 15, 151-156.	1.4	19

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109	Falls and resulting fractures in Myotonic Dystrophy: Results from a multinational retrospective survey. Neuromuscular Disorders, 2018, 28, 229-235.	0.6	19
110	Eight years after an international workshop on myotonic dystrophy patient registries: case study of a global collaboration for a rare disease. Orphanet Journal of Rare Diseases, 2018, 13, 155.	2.7	19
111	Interrelation between Sarcopenia and the Number of Motor Neurons in Patients with Parkinsonian Syndromes. Gerontology, 2020, 66, 409-415.	2.8	19
112	Non-dystrophic myotonias: clinical and mutation spectrum of 70 German patients. Journal of Neurology, 2021, 268, 1708-1720.	3.6	19
113	Urge Incontinence and Gastrointestinal Symptoms in Adult Patients with Pompe Disease: A Cross-Sectional Survey. JIMD Reports, 2014, 17, 53-61.	1.5	18
114	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. Journal of Neuromuscular Diseases, 2020, 7, 153-166.	2.6	18
115	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
116	Safety and efficacy of short- and long-term inspiratory muscle training in late-onset Pompe disease (LOPD): a pilot study. Journal of Neurology, 2019, 266, 133-147.	3.6	17
117	Pompe disease: what are we missing?. Annals of Translational Medicine, 2019, 7, 292-292.	1.7	17
118	Early-Onset Myopathies: Clinical Findings, Prevalence of Subgroups and Diagnostic Approach in a Single Neuromuscular Referral Center in Germany. Journal of Neuromuscular Diseases, 2017, 4, 315-325.	2.6	17
119	Physiology, pathophysiology and diagnostic significance of autophagic changes in skeletal muscle tissue â€“ towards the enigma of rimmed and round vacuoles. , 2009, 28, 59-70.		16
120	Successful treatment with azacitidine in VEXAS syndrome with prominent myofasciitis. Rheumatology, 2022, 61, e117-e119.	1.9	16
121	Long-term Safety and Efficacy of Avalglucosidase Alfa in Patients With Late-Onset Pompe Disease. Neurology, 2022, 99, .	1.1	16
122	Long-term efficiency of intravenously administered immunoglobulin in anti-Yo syndrome with paraneoplastic cerebellar degeneration. Journal of Neurology, 2011, 258, 946-947.	3.6	15
123	Immunohistochemistry on a panel of Emeryâ€“Dreifuss muscular dystrophy samples reveals nuclear envelope proteins as inconsistent markers for pathology. Neuromuscular Disorders, 2017, 27, 338-351.	0.6	15
124	Rare diagnosis of telethoninopathy (LGMD2G) in a Turkish patient. Neuromuscular Disorders, 2017, 27, 856-860.	0.6	15
125	An integrative correlation of myopathology, phenotype and genotype in late onset Pompe disease. Neuropathology and Applied Neurobiology, 2020, 46, 359-374.	3.2	13
126	Water T 2 Mapping in Fatty Infiltrated Thigh Muscles of Patients With Neuromuscular Diseases Using a T 2 â€Prepared 3D Turbo Spin Echo With SPAIR. Journal of Magnetic Resonance Imaging, 2020, 51, 1727-1736.	3.4	13

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127	IGF-I/IGFBP3/ALS Deficiency in Sarcopenia: Low GHBP Suggests GH Resistance in a Subgroup of Geriatric Patients. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 1698-1707.	3.6	13
128	Unclassified polysaccharidosis of the heart and skeletal muscle in siblings. Molecular Genetics and Metabolism, 2008, 95, 52-58.	1.1	12
129	T2-Weighted Dixon Turbo Spin Echo for Accelerated Simultaneous Grading of Whole-Body Skeletal Muscle Fat Infiltration and Edema in Patients With Neuromuscular Diseases. Journal of Computer Assisted Tomography, 2018, 42, 574-579.	0.9	12
130	A zebrafish model for FHL1-opathy reveals loss-of-function effects of human FHL1 mutations. Neuromuscular Disorders, 2018, 28, 521-531.	0.6	12
131	Pattern of myogenesis and vascular repair in early and advanced lesions of juvenile dermatomyositis. Neuromuscular Disorders, 2018, 28, 973-985.	0.6	12
132	Limb girdle muscular dystrophy 2G in a religious minority of Bulgarian Muslims homozygous for the c.75G>A, p.Trp25X mutation. Neuromuscular Disorders, 2018, 28, 625-632.	0.6	12
133	Pharmacological and nutritional treatment for McArdle disease (Glycogen Storage Disease type V). , 2010, , CD003458.		11
134	Clinicopathological analysis of the homozygous p.W1327X AGL mutation in glycogen storage disease type 3. American Journal of Medical Genetics, Part A, 2008, 146A, 2911-2915.	1.2	10
135	Lifetime exercise intolerance with lactic acidosis as key manifestation of novel compound heterozygous ACAD9 mutations causing complex I deficiency. Neuromuscular Disorders, 2017, 27, 473-476.	0.6	10
136	Charcot-Marie-Tooth disease type 2CC due to a frameshift mutation of the neurofilament heavy polypeptide gene in an Austrian family. Neuromuscular Disorders, 2019, 29, 392-397.	0.6	10
137	A Systematic Review of the Health Economics of Pompe Disease. PharmacoEconomics - Open, 2019, 3, 479-493.	1.8	10
138	A role for cannabinoids in the treatment of myotonia? Report of compassionate use in a small cohort of patients. Journal of Neurology, 2020, 267, 415-421.	3.6	10
139	Associations Between Variant Repeat Interruptions and Clinical Outcomes in Myotonic Dystrophy Type 1. Neurology: Genetics, 2021, 7, e572.	1.9	10
140	Four and a Half LIM Protein 1C (FHL1C): A Binding Partner for Voltage-Gated Potassium Channel Kv1.5. PLoS ONE, 2011, 6, e26524.	2.5	10
141	Comparison of recent pivotal recommendations for the diagnosis and treatment of late-onset Pompe disease using diagnostic nodesâ€"the Pompe disease burden scale. Journal of Neurology, 2019, 266, 2010-2017.	3.6	9
142	Utility of maximum inspiratory and expiratory pressures as a screening method for respiratory insufficiency in slowly progressive neuromuscular disorders. Neuromuscular Disorders, 2020, 30, 640-648.	0.6	9
143	Utility and Results from a Patient-Reported Online Survey in Myotonic Dystrophies Types 1 and 2. European Neurology, 2020, 83, 523-533.	1.4	9
144	Validation of Motor Outcome Measures in Myotonic Dystrophy Type 2. Frontiers in Neurology, 2020, 11, 306.	2.4	9

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145	Quantitative Muscle MRI in Patients with Neuromuscular Diseasesâ€"Association of Muscle Proton Density Fat Fraction with Semi-Quantitative Grading of Fatty Infiltration and Muscle Strength at the Thigh Region. Diagnostics, 2021, 11, 1056.	2.6	9
146	Transcriptome Analysis in a Primary Human Muscle Cell Differentiation Model for Myotonic Dystrophy Type 1. International Journal of Molecular Sciences, 2021, 22, 8607.	4.1	9
147	Exome Sequencing Identified a Splice Site Mutation in FHL1 that Causes Uruguay Syndrome, an X-Linked Disorder With Skeletal Muscle Hypertrophy and Premature Cardiac Death. Circulation: Cardiovascular Genetics, 2016, 9, 130-135.	5.1	8
148	Towards clinical outcome measures in myotonic dystrophy type 2: a systematic review. Current Opinion in Neurology, 2018, 31, 599-609.	3.6	8
149	Cannabis use in myotonic dystrophy patients in Germany and USA: a pilot survey. Journal of Neurology, 2019, 266, 530-532.	3.6	8
150	Nuclear Envelope Transmembrane Proteins in Myotonic Dystrophy Type 1. Frontiers in Physiology, 2018, 9, 1532.	2.8	7
151	Regional variation of thigh muscle fat infiltration in patients with neuromuscular diseases compared to healthy controls. Quantitative Imaging in Medicine and Surgery, 2021, 11, 2610-2621.	2.0	7
152	Influence of IGF-I serum concentration on muscular regeneration capacity in patients with sarcopenia. BMC Musculoskeletal Disorders, 2021, 22, 807.	1.9	7
153	Differential expression and localization of Ankrd2 isoforms in human skeletal and cardiac muscles. Histochemistry and Cell Biology, 2016, 146, 569-584.	1.7	6
154	Long-term whole-body vibration training in two late-onset Pompe disease patients. Neurological Sciences, 2016, 37, 1357-1360.	1.9	6
155	211th ENMC International Workshop:. Neuromuscular Disorders, 2017, 27, 1143-1151.	0.6	6
156	Towards development of a statistical framework to evaluate myotonic dystrophy type 1 mRNA biomarkers in the context of a clinical trial. PLoS ONE, 2020, 15, e0231000.	2.5	6
157	The impact of interrupting enzyme replacement therapy in late-onset Pompe disease. Journal of Neurology, 2021, 268, 2943-2950.	3.6	6
158	Late-onset neuromuscular disorders in the differential diagnosis of sarcopenia. BMC Neurology, 2021, 21, 241.	1.8	6
159	Assessing metabolic profiles in human myoblasts from patients with late-onset Pompe disease. Annals of Translational Medicine, 2019, 7, 277-277.	1.7	6
160	Identification of variants in MBNL1 in patients with a myotonic dystrophy-like phenotype. European Journal of Human Genetics, 2016, 24, 1467-1472.	2.8	5
161	Inherited and Acquired Muscle Weakness: A Moving Target for Diagnostic Muscle Biopsy. Neuropediatrics, 2017, 48, 226-232.	0.6	5
162	Minutes of the European POmpe Consortium (EPOC) Meeting March 27 to 28, 2015, Munich, Germany. Acta Myologica, 2015, 34, 141-3.	1.5	5

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163	Are Evoked Potentials in Patients With Adult-Onset Pompe Disease Indicative of Clinically Relevant Central Nervous System Involvement?. Journal of Clinical Neurophysiology, 2014, 31, 362-366.	1.7	4
164	Glycogen storage diseases of all types. Journal of Inherited Metabolic Disease, 2015, 38, 389-390.	3.6	4
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