Benedikt Schoser

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3716041/publications.pdf

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	The effect of methocarbamol and mexiletine on murine muscle spindle function. Muscle and Nerve, 2022, 66, 96-105.	2.2	2
2	Successful treatment with azacitidine in VEXAS syndrome with prominent myofasciitis. Rheumatology, 2022, 61, e117-e119.	1.9	16
3	Small fiber involvement is independent from clinical pain in late-onset Pompe disease. Orphanet Journal of Rare Diseases, 2022, 17, 177.	2.7	1
4	BNIP3 Is Involved in Muscle Fiber Atrophy in Late-Onset Pompe Disease Patients. American Journal of Pathology, 2022, , .	3.8	3
5	Long-term Safety and Efficacy of Avalglucosidase Alfa in Patients With Late-Onset Pompe Disease. Neurology, 2022, 99, .	1.1	16
6	Slowly Progressive Limb-Girdle Weakness and HyperCKemia â€“ Limb Girdle Muscular Dystrophy or Anti-3-Hydroxy-3-Methylglutaryl-CoA-Reductase-Myopathy?. Journal of Neuromuscular Diseases, 2022, , 1-8.	2.6	2
7	Type-2 muscle fiber atrophy is associated with sarcopenia in elderly men with hip fracture. Experimental Gerontology, 2021, 144, 111171.	2.8	20
8	Non-dystrophic myotonias: clinical and mutation spectrum of 70 German patients. Journal of Neurology, 2021, 268, 1708-1720.	3.6	19
9	Dyslexia and cognitive impairment in adult patients with myotonic dystrophy type 1: a clinical prospective analysis. Journal of Neurology, 2021, 268, 484-492.	3.6	0
10	Uptake of mossâ€derived human recombinant GAA in Gaa â^'/â^' mice. JIMD Reports, 2021, 59, 81-89.	1.5	3
11	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
12	The impact of interrupting enzyme replacement therapy in late-onset Pompe disease. Journal of Neurology, 2021, 268, 2943-2950.	3.6	6
13	Associations Between Variant Repeat Interruptions and Clinical Outcomes in Myotonic Dystrophy Type 1. Neurology: Genetics, 2021, 7, e572.	1.9	10
14	The risks of using non-specific outcome measures to capture activities of daily living in myotonic dystrophy type 2 - Response. Neuromuscular Disorders, 2021, 31, 369.	0.6	0
15	New developments in myotonic dystrophies from a multisystemic perspective. Current Opinion in Neurology, 2021, 34, 738-747.	3.6	3
16	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
17	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
18	Modern Gestalt approach to neuromuscular disorders. Current Opinion in Neurology, 2021, Publish Ahead of Print, .	3.6	0

#	Article	IF	Citations
19	Late-onset neuromuscular disorders in the differential diagnosis of sarcopenia. BMC Neurology, 2021, 21, 241.	1.8	6
20	CTG-Repeat Detection in Primary Human Myoblasts of Myotonic Dystrophy Type 1. Frontiers in Neuroscience, 2021, 15, 686735.	2.8	0
21	Clinical Outcome Evaluations and CBT Response Prediction in Myotonic Dystrophy. Journal of Neuromuscular Diseases, 2021, 8, 1031-1046.	2.6	4
22	Differential Diagnosis of Acquired and Hereditary Neuropathies in Children and Adolescentsâ€"Consensus-Based Practice Guidelines. Children, 2021, 8, 687.	1.5	4
23	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
24	Influence of IGF-I serum concentration on muscular regeneration capacity in patients with sarcopenia. BMC Musculoskeletal Disorders, 2021, 22, 807.	1.9	7
25	Congenital myopathy and epidermolysis bullosa due to PLEC variant. Neuromuscular Disorders, 2021, 31, 1212-1217.	0.6	4
26	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
27	Cutaneous T-cell lymphoma mimicking myopathy with lipoatrophy. Neuromuscular Disorders, 2021, , .	0.6	0
28	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
29	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
30	A systematic review on the definition of rhabdomyolysis. Journal of Neurology, 2020, 267, 877-882.	3.6	101
31	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
32	An integrative correlation of myopathology, phenotype and genotype in late onset Pompe disease. Neuropathology and Applied Neurobiology, 2020, 46, 359-374.	3.2	13
33	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
34	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
35	How to capture activities of daily living in myotonic dystrophy type 2?. Neuromuscular Disorders, 2020, 30, 796-806.	0.6	2
36	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

#	Article	IF	Citations
37	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
38	Validation of Motor Outcome Measures in Myotonic Dystrophy Type 2. Frontiers in Neurology, 2020, 11, 306.	2.4	9
39	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. Journal of Neuromuscular Diseases, 2020, 7, 153-166.	2.6	18
40	Progressive external ophthalmoplegia due to a recurrent de novo m.15990C>T MT-TP (mt-tRNAPro) gene variant. Neuromuscular Disorders, 2020, 30, 346-350.	0.6	4
41	Interrelation between Sarcopenia and the Number of Motor Neurons in Patients with Parkinsonian Syndromes. Gerontology, 2020, 66, 409-415.	2.8	19
42	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
43	248th ENMC International Workshop: Myotonic dystrophies: Molecular approaches for clinical purposes, framing a European molecular research network, Hoofddorp, the Netherlands, 11â€“13 October 2019. Neuromuscular Disorders, 2020, 30, 521-531.	0.6	1
44	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
45	Regional Variation of Thigh Muscle Composition in Healthy Controls and Patients with Myotonic Dystrophy Type 2, Limb Girdle Muscular Dystrophy Type 2A, and Pompeâ€™s Disease. , 2020, 24, .		0
46	Title is missing!. , 2020, 15, e0231000.		0
47	Title is missing!. , 2020, 15, e0231000.		0
48	Title is missing!. , 2020, 15, e0231000.		0
49	Title is missing!. , 2020, 15, e0231000.		0
50	Title is missing!. , 2020, 15, e0231000.		0
51	Title is missing!. , 2020, 15, e0231000.		0
52	Mannose 6â€phosphonate labelling: A key for processing the therapeutic enzyme in Pompe disease. Journal of Cellular and Molecular Medicine, 2019, 23, 6499-6503.	3.6	4
53	PRECLINICAL APPROACHES AND EARLY CLINICAL RESULTS. Neuromuscular Disorders, 2019, 29, S122.	0.6	2
54	Consensus-based care recommendations for adults with myotonic dystrophy type 2. Neurology: Clinical Practice, 2019, 9, 343-353.	1.6	41

#	Article	IF	Citations
55	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
56	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
57	MSH3 modifies somatic instability and disease severity in Huntingtonâ€™s and myotonic dystrophy type 1. Brain, 2019, 142, 1876-1886.	7.6	114
58	Decreased water T₂ in fatty infiltrated skeletal muscles of patients with neuromuscular diseases. NMR in Biomedicine, 2019, 32, e4111.	2.8	20
59	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
60	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	2.4	25
61	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
62	First-in-human study of advanced and targeted acid Î±-glucosidase (AT-GAA) (ATB200/AT2221) in patients with Pompe disease: preliminary functional assessment results from the ATB200-02 trial. Molecular Genetics and Metabolism, 2019, 126, S86.	1.1	4
63	A Systematic Review of the Health Economics of Pompe Disease. PharmacoEconomics - Open, 2019, 3, 479-493.	1.8	10
64	A genetic modifier of symptom onset in Pompe disease. EBioMedicine, 2019, 43, 553-561.	6.1	32
65	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
66	Genetic determinants of disease severity in the myotonic dystrophy type 1 OPTIMISTIC cohort. Neurology, 2019, 93, e995-e1009.	1.1	71
67	Sarcopenia â€“ Endocrinological and Neurological Aspects. Experimental and Clinical Endocrinology and Diabetes, 2019, 6, 8-22.	1.2	23
68	Cannabis use in myotonic dystrophy patients in Germany and USA: a pilot survey. Journal of Neurology, 2019, 266, 530-532.	3.6	8
69	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
70	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
71	Assessing metabolic profiles in human myoblasts from patients with late-onset Pompe disease. Annals of Translational Medicine, 2019, 7, 277-277.	1.7	6
72	Pompe disease: what are we missing?. Annals of Translational Medicine, 2019, 7, 292-292.	1.7	17

#	Article	IF	Citations
73	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
74	Novel Pompe disease phenotype: a treatment-related modified phenotype neglecting the brain. Developmental Medicine and Child Neurology, 2018, 60, 536-536.	2.1	3
75	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
76	Falls and resulting fractures in Myotonic Dystrophy: Results from a multinational retrospective survey. Neuromuscular Disorders, 2018, 28, 229-235.	0.6	19
77	A zebrafish model for FHL1-opathy reveals loss-of-function effects of human FHL1 mutations. Neuromuscular Disorders, 2018, 28, 521-531.	0.6	12
78	Nuclear Envelope Transmembrane Proteins in Myotonic Dystrophy Type 1. Frontiers in Physiology, 2018, 9, 1532.	2.8	7
79	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
80	Towards clinical outcome measures in myotonic dystrophy type 2: a systematic review. Current Opinion in Neurology, 2018, 31, 599-609.	3.6	8
81	Pattern of myogenesis and vascular repair in early and advanced lesions of juvenile dermatomyositis. Neuromuscular Disorders, 2018, 28, 973-985.	0.6	12
82	How to Interpret Abnormal Findings of Spirometry and Manometry in Myotonic Dystrophies?. Journal of Neuromuscular Diseases, 2018, 5, 451-459.	2.6	4
83	Editorial: Beyond Borders: Myotonic Dystrophiesâ€“A European Perception. Frontiers in Neurology, 2018, 9, 787.	2.4	2
84	Consensus-based care recommendations for adults with myotonic dystrophy type 1. Neurology: Clinical Practice, 2018, 8, 507-520.	1.6	115
85	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
86	rbFOX1/MBNL1 competition for CCUG RNA repeats bindingÂcontributes to myotonic dystrophy typeÂ1/typeÂ2 differences. Nature Communications, 2018, 9, 2009.	12.8	61
87	Core Clinical Phenotypes in Myotonic Dystrophies. Frontiers in Neurology, 2018, 9, 303.	2.4	104
88	Myotonic Dystrophyâ€"A Progeroid Disease?. Frontiers in Neurology, 2018, 9, 601.	2.4	34
89	Self-diagnosis of a triple trouble. Neuromuscular Disorders, 2018, 28, 825-827.	0.6	2
90	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

#	Article	IF	Citations
91	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
92	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
93	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
94	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
95	Inherited and Acquired Muscle Weakness: A Moving Target for Diagnostic Muscle Biopsy. Neuropediatrics, 2017, 48, 226-232.	0.6	5
96	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
97	Lambertâ€“Eaton myasthenic syndrome (LEMS): a rare autoimmune presynaptic disorder often associated with cancer. Journal of Neurology, 2017, 264, 1854-1863.	3.6	65
98	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
99	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
100	211th ENMC International Workshop:. Neuromuscular Disorders, 2017, 27, 1143-1151.	0.6	6
101	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
102	Rare diagnosis of telethoninopathy (LGMD2G) in a Turkish patient. Neuromuscular Disorders, 2017, 27, 856-860.	0.6	15
103	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
104	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
105	The humanistic burden of Pompe disease: are there still unmet needs? A systematic review. BMC Neurology, 2017, 17, 202.	1.8	31
106	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
107	Differential expression and localization of Ankrd2 isoforms in human skeletal and cardiac muscles. Histochemistry and Cell Biology, 2016, 146, 569-584.	1.7	6
108	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

#	Article	IF	Citations
109	Prevalence of Pompe disease in 3,076 patients with hyperCKemia and limb-girdle muscular weakness. Neurology, 2016, 87, 295-298.	1.1	60
110	Meta-opinion: from screening to diagnosis of Pompe disease â€“ a European perspective. Expert Opinion on Orphan Drugs, 2016, 4, 1075-1078.	0.8	1
111	Diagnostic muscle biopsy: is it still needed on the way to a liquid muscle pathology?. Current Opinion in Neurology, 2016, 29, 602-605.	3.6	4
112	Long-term whole-body vibration training in two late-onset Pompe disease patients. Neurological Sciences, 2016, 37, 1357-1360.	1.9	6
113	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
114	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
115	New insights into the protein aggregation pathology in myotilinopathy by combined proteomic and immunolocalization analyses. Acta Neuropathologica Communications, 2016, 4, 8.	5.2	50
116	Differential roles of hypoxia and innate immunity in juvenile and adult dermatomyositis. Acta Neuropathologica Communications, 2016, 4, 45.	5.2	52
117	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
118	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
119	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
120	â€œOrbiting aroundâ€•the orbital myositis: clinical features, differential diagnosis and therapy. Journal of Neurology, 2016, 263, 631-640.	3.6	43
121	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
122	A Troublesome Debate: When to Start Treatment in Adult Pompe Patients?. Journal of Neuromuscular Diseases, 2015, 2, S8-S8.	2.6	0
123	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
124	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	9.0	41
125	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
126	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

#	Article	IF	Citations
127	50 years to diagnosis: Autosomal dominant tubular aggregate myopathy caused by a novel STIM1 mutation. Neuromuscular Disorders, 2015, 25, 577-584.	0.6	47
128	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
129	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
130	A phase 4 prospective study in patients with adult Pompe disease treated with alglucosidase alfa. Molecular Genetics and Metabolism, 2015, 114, S113-S114.	1.1	3
131	InÂvivo characterization of human myofibrillar myopathy genes in zebrafish. Biochemical and Biophysical Research Communications, 2015, 461, 217-223.	2.1	27
132	Glycogen storage diseases of all types. Journal of Inherited Metabolic Disease, 2015, 38, 389-390.	3.6	4
133	Sleepâ€related symptoms and sleepâ€disordered breathing in adult <scp>P</scp>ompe disease. European Journal of Neurology, 2015, 22, 369.	3.3	48
134	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
135	Muscle histology changes after short term vibration training in healthy controls. Acta Myologica, 2015, 34, 133-8.	1.5	2
136	Minutes of the European POmpe Consortium (EPOC) Meeting March 27 to 28, 2015, Munich, Germany. Acta Myologica, 2015, 34, 141-3.	1.5	5
137	Pharmacological and nutritional treatment for McArdle disease (Glycogen Storage Disease type V). The Cochrane Library, 2014, 2014, CD003458.	2.8	54
138	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
139	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
140	Anti-agrin autoantibodies in myasthenia gravis. Neurology, 2014, 82, 1976-1983.	1.1	151
141	Urge Incontinence and Gastrointestinal Symptoms in Adult Patients with Pompe Disease: A Cross-Sectional Survey. JIMD Reports, 2014, 17, 53-61.	1.5	18
142	Novel recessive myotilin mutation causes severe myofibrillar myopathy. Neurogenetics, 2014, 15, 151-156.	1.4	19
143	Pregnancy and delivery in women with Pompe disease. Molecular Genetics and Metabolism, 2014, 112, 148-153.	1.1	22
144	Alglucosidase alfa: 5 years of experience in late-onset Pompe disease. BMC Musculoskeletal Disorders, 2013, 14, .	1.9	0

#	Article	IF	Citations
145	Enzyme replacement therapy in late-onset Pompe disease: a systematic literature review. Journal of Neurology, 2013, 260, 951-959.	3.6	168
146	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
147	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
148	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
149	Pain in adult patients with Pompe disease. Molecular Genetics and Metabolism, 2013, 109, 371-376.	1.1	31
150	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
151	Proteomic characterization of aggregate components in an intrafamilial variable FHL1-associated myopathy. Neuromuscular Disorders, 2013, 23, 418-426.	0.6	25
152	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
153	Polyglucosan body myopathy caused by defective ubiquitin ligase RBCK1. Annals of Neurology, 2013, 74, 914-919.	5.3	132
154	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
155	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
156	A role for PLCÎ²1 in myotonic dystrophies type 1 and 2. FASEB Journal, 2012, 26, 3042-3048.	0.5	24
157	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
158	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
159	Muscle MRI findings in limb girdle muscular dystrophy type 2L. Neuromuscular Disorders, 2012, 22, S122-S129.	0.6	77
160	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
161	G.P.123 Identification of molecular effects of FHL1 mutations on protein assembly in reducing body myopathy. Neuromuscular Disorders, 2012, 22, 903.	0.6	0
162	A new phenotype of brain iron accumulation with dystonia, optic atrophy, and peripheral neuropathy. Movement Disorders, 2012, 27, 789-793.	3.9	41

#	Article	IF	Citations
163	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
164	Transplantation of Genetically Corrected Human iPSC-Derived Progenitors in Mice with Limb-Girdle Muscular Dystrophy. Science Translational Medicine, 2012, 4, 140ra89.	12.4	269
165	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850.	3.6	72
166	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
167	Anti-LRP4 autoantibodies in AChR- and MuSK-antibody-negative myasthenia gravis. Journal of Neurology, 2012, 259, 427-435.	3.6	297
168	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
169	Nemaline myopathy caused by mutations in the nebulin gene may present as a distal myopathy. Neuromuscular Disorders, 2011, 21, 556-562.	0.6	56
170	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
171	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
172	Reducing Body Myopathy and Other FHL1-Related Muscular Disorders. Seminars in Pediatric Neurology, 2011, 18, 257-263.	2.0	44
173	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. American Journal of Human Genetics, 2011, 88, 162-172.	6.2	153
174	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
175	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
176	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
177	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
178	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
179	Myotonic Dystrophies 1 and 2: Complex Diseases with Complex Mechanisms. Current Genomics, 2010, 11, 77-90.	1.6	82
180	Pharmacological and nutritional treatment for McArdle disease (Glycogen Storage Disease type V). , 2010, , CD003458.		11

#	Article	IF	Citations
181	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
182	Facioscapulohumeral muscular dystrophy presenting with unusual phenotypes and atypical morphological features of vacuolar myopathy. Journal of Neurology, 2010, 257, 1108-1118.	3.6	26
183	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
184	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
185	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
186	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
187	Reduction of the Rate of Protein Translation in Patients with Myotonic Dystrophy 2. Journal of Neuroscience, 2009, 29, 9042-9049.	3.6	81
188	Absence of a differentiation defect in muscle satellite cells from DM2 patients. Neurobiology of Disease, 2009, 36, 181-190.	4.4	64
189	Intragenic deletion ofTRIM32in compound heterozygotes with sarcotubular myopathy/LGMD2H. Human Mutation, 2009, 30, E831-E844.	2.5	37
190	Myotone Dystrophien â€“ und ihre Differenzialdiagnosen. Medizinische Genetik, 2009, 21, 381-392.	0.2	2
191	Myofibrillar Myopathies: A Clinical and Myopathological Guide. Brain Pathology, 2009, 19, 483-492.	4.1	164
192	Consequences of mutations within the C terminus of the FHL1 gene. Neurology, 2009, 73, 543-551.	1.1	61
193	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
194	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
195	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
196	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
197	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
198	Therapeutic Approaches in Glycogen Storage Disease Type II/Pompe Disease. Neurotherapeutics, 2008, 5, 569-578.	4.4	69

#	Article	IF	Citations
199	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
200	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
201	Unclassified polysaccharidosis of the heart and skeletal muscle in siblings. Molecular Genetics and Metabolism, 2008, 95, 52-58.	1.1	12
202	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
203	Adult-onset glycogen storage disease type 2: clinico-pathological phenotype revisited. Neuropathology and Applied Neurobiology, 2007, 33, 070615152525006-???.	3.2	61
204	Commonality ofTRIM32mutation in causing sarcotubular myopathy and LGMD2H. Annals of Neurology, 2005, 57, 591-595.	5.3	96