Volker Straub

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1938655/publications.pdf

Version: 2024-02-01

197 papers	11,850 citations	23567 58 h-index	33894 99 g-index
206 all docs	206 docs citations	206 times ranked	12807 citing authors

#	Article	IF	Citations
1	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
2	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
3	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. Human Mutation, 2015, 36, 395-402.	2.5	507
4	Animal Models for Muscular Dystrophy Show Different Patterns of Sarcolemmal Disruption. Journal of Cell Biology, 1997, 139, 375-385.	5.2	441
5	Prevalence of genetic muscle disease in Northern England: in-depth analysis of a muscle clinic population. Brain, 2009, 132, 3175-3186.	7.6	414
6	Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. Brain, 2007, 130, 2725-2735.	7.6	385
7	Safety and efficacy of drisapersen for the treatment of Duchenne muscular dystrophy (DEMAND II): an exploratory, randomised, placebo-controlled phase 2 study. Lancet Neurology, The, 2014, 13, 987-996.	10.2	279
8	Managing Duchenne muscular dystrophy â€“ The additive effect of spinal surgery and home nocturnal ventilation in improving survival. Neuromuscular Disorders, 2007, 17, 470-475.	0.6	273
9	A founder mutation in Anoctamin 5 is a major cause of limb girdle muscular dystrophy. Brain, 2011, 134, 171-182.	7.6	254
10	Mutations in the FKRP gene can cause muscle-eye-brain disease and Walker-Warburg syndrome. Journal of Medical Genetics, 2004, 41, e61-e61.	3.2	243
11	Phenotypic spectrum associated with mutations in the fukutin-related protein gene. Annals of Neurology, 2003, 53, 537-542.	5.3	219
12	Long-term benefits and adverse effects of intermittent versus daily glucocorticoids in boys with Duchenne muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 698-705.	1.9	201
13	Secondary calpain3 deficiency in 2q-linked muscular dystrophy. Neurology, 2001, 56, 869-877.	1.1	163
14	Genotypeâ€“phenotype correlation in a large population of muscular dystrophy patients with LAMA2 mutations. Neuromuscular Disorders, 2010, 20, 241-250.	0.6	154
15	The burden of Duchenne muscular dystrophy. Neurology, 2014, 83, 529-536.	1.1	149
16	Quantitative Muscle MRI as an Assessment Tool for Monitoring Disease Progression in LGMD2I: A Multicentre Longitudinal Study. PLoS ONE, 2013, 8, e70993.	2.5	148
17	Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. Human Mutation, 2012, 33, 981-988.	2.5	145
18	Development of Exon Skipping Therapies for Duchenne Muscular Dystrophy: A Critical Review and a Perspective on the Outstanding Issues. Nucleic Acid Therapeutics, 2017, 27, 251-259.	3.6	144

#	Article	IF	Citations
19	Serum matrix metalloproteinase-9 (MMP-9) as a biomarker for monitoring disease progression in Duchenne muscular dystrophy (DMD). Neuromuscular Disorders, 2011, 21, 569-578.	0.6	132
20	Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. Journal of Neuromuscular Diseases, 2017, 4, 293-306.	2.6	125
21	Affinity proteomics within rare diseases: a <scp>BIO</scp>â€<scp>NMD</scp> study for blood biomarkers of muscular dystrophies. EMBO Molecular Medicine, 2014, 6, 918-936.	6.9	105
22	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. Nature Medicine, 2021, 27, 1197-1204.	30.7	96
23	Towards harmonization of protocols for MRI outcome measures in skeletal muscle studies: Consensus recommendations from two TREAT-NMD NMR workshops, 2 May 2010, Stockholm, Sweden, 1â€“2 October 2009, Paris, France. Neuromuscular Disorders, 2012, 22, S54-S67.	0.6	94
24	The TREAT-NMD Duchenne Muscular Dystrophy Registries: Conception, Design, and Utilization by Industry and Academia. Human Mutation, 2013, 34, 1449-1457.	2.5	94
25	TREAT-NMD workshop: Pattern recognition in genetic muscle diseases using muscle MRI. Neuromuscular Disorders, 2012, 22, S42-S53.	0.6	93
26	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
27	Inheritance patterns and phenotypic features of myofibrillar myopathy associated with a BAG3 mutation. Neuromuscular Disorders, 2010, 20, 438-442.	0.6	90
28	Muscle Magnetic Resonance Imaging in Congenital Myopathies Due to Ryanodine Receptor Type 1 Gene Mutations. Archives of Neurology, 2011, 68, 1171.	4.5	89
29	A heterozygous 21-bp deletion inCAPN3causes dominantly inherited limb girdle muscular dystrophy. Brain, 2016, 139, 2154-2163.	7.6	87
30	Validation of genetic modifiers for Duchenne muscular dystrophy: a multicentre study assessingSPP1andLTBP4variants. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 1060-1065.	1.9	86
31	Healthâ€related quality of life in patients with Duchenne muscular dystrophy: a multinational, crossâ€sectional study. Developmental Medicine and Child Neurology, 2016, 58, 508-515.	2.1	82
32	Quantifying the burden of caregiving in Duchenne muscular dystrophy. Journal of Neurology, 2016, 263, 906-915.	3.6	82
33	Limb-girdle muscular dystrophies â€” international collaborations for translational research. Nature Reviews Neurology, 2016, 12, 294-309.	10.1	81
34	Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1071-1081.	1.9	81
35	Quantitative Magnetic Resonance Imaging in Limb-Girdle Muscular Dystrophy 2I: A Multinational Cross-Sectional Study. PLoS ONE, 2014, 9, e90377.	2.5	81
36	New aspects on patients affected by dysferlin deficient muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 946-953.	1.9	79

#	Article	IF	Citations
37	Muscle MRI findings in limb girdle muscular dystrophy type 2L. Neuromuscular Disorders, 2012, 22, S122-S129.	0.6	77
38	Stakeholder cooperation to overcome challenges in orphan medicine development: the example of Duchenne muscular dystrophy. Lancet Neurology, The, 2016, 15, 882-890.	10.2	77
39	Mapping the differences in care for 5,000 Spinal Muscular Atrophy patients, a survey of 24 national registries in North America, Australasia and Europe. Journal of Neurology, 2014, 261, 152-163.	3.6	76
40	Improving recognition of Duchenne muscular dystrophy: a retrospective case note review. Archives of Disease in Childhood, 2014, 99, 1074-1077.	1.9	75
41	The Clinical Outcome Study for dysferlinopathy. Neurology: Genetics, 2016, 2, e89.	1.9	75
42	Congenital muscular dystrophies in the UK population: Clinical and molecular spectrum of a large cohort diagnosed over a 12-year period. Neuromuscular Disorders, 2017, 27, 793-803.	0.6	75
43	Dystrophin quantification. Neurology, 2014, 83, 2062-2069.	1.1	73
44	Fibronectin is a serum biomarker for <scp>D</scp>uchenne muscular dystrophy. Proteomics - Clinical Applications, 2014, 8, 269-278.	1.6	73
45	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850.	3.6	72
46	The Classification, Natural History and Treatment of the Limb Girdle Muscular Dystrophies. Journal of Neuromuscular Diseases, 2015, 2, S7-S19.	2.6	72
47	Biochemical Characterization of Patients With In-Frame or Out-of-FrameDMDDeletions Pertinent to Exon 44 or 45 Skipping. JAMA Neurology, 2014, 71, 32.	9.0	71
48	Titin founder mutation is a common cause of myofibrillar myopathy with early respiratory failure. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 331-338.	1.9	71
49	Muscle-Derived Proteins as Serum Biomarkers for Monitoring Disease Progression in Three Forms of Muscular Dystrophy. Journal of Neuromuscular Diseases, 2015, 2, 241-255.	2.6	71
50	Association Study of Exon Variants in the NF-Î²B and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	6.2	71
51	Magnetic resonance imaging in duchenne muscular dystrophy: Longitudinal assessment of natural history over 18 months. Muscle and Nerve, 2013, 48, 586-588.	2.2	70
52	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Journal of Neuromuscular Diseases, 2014, 1, 75-90.	2.6	69
53	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
54	Why are some patients with Duchenne muscular dystrophy dying young: An analysis of causes of death in North East England. European Journal of Paediatric Neurology, 2016, 20, 904-909.	1.6	66

#	Article	IF	Citations
55	Congenital muscular dystrophy with laminin Î±2 chain deficiency: Identification of a new intermediate phenotype and correlation of clinical findings to muscle immunohistochemistry. European Journal of Pediatrics, 1996, 155, 968-976.	2.7	65
56	Contrast agent-enhanced magnetic resonance imaging of skeletal muscle damage in animal models of muscular dystrophy. Magnetic Resonance in Medicine, 2000, 44, 655-659.	3.0	65
57	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. Genetics in Medicine, 2020, 22, 1478-1488.	2.4	62
58	Prevalence of Pompe disease in 3,076 patients with hyperCKemia and limb-girdle muscular weakness. Neurology, 2016, 87, 295-298.	1.1	60
59	MR imaging in Duchenne muscular dystrophy: Quantification of T₁â€weighted signal, contrast uptake, and the effects of exercise. Journal of Magnetic Resonance Imaging, 2009, 30, 1130-1138.	3.4	59
60	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
61	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
62	Short stature and pubertal delay in Duchenne muscular dystrophy. Archives of Disease in Childhood, 2016, 101, 101-106.	1.9	58
63	Long-Term Safety and Efficacy Data of Golodirsen in Ambulatory Patients with Duchenne Muscular Dystrophy Amenable to Exon 53 Skipping: A First-in-human, Multicenter, Two-Part, Open-Label, Phase 1/2 Trial. Nucleic Acid Therapeutics, 2022, 32, 29-39.	3.6	58
64	Late onset in dysferlinopathy widens the clinical spectrum. Neuromuscular Disorders, 2008, 18, 288-290.	0.6	57
65	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. Contemporary Clinical Trials, 2017, 58, 34-39.	1.8	56
66	Fractures and Linear Growth in a Nationwide Cohort of Boys With Duchenne Muscular Dystrophy With and Without Glucocorticoid Treatment. JAMA Neurology, 2019, 76, 701.	9.0	56
67	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	1.9	55
68	Compliance to Care Guidelines for Duchenne Muscular Dystrophy. Journal of Neuromuscular Diseases, 2015, 2, 63-72.	2.6	51
69	Reduced serum myostatin concentrations associated with genetic muscle disease progression. Journal of Neurology, 2017, 264, 541-553.	3.6	51
70	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
71	Tracking disease progression nonâ€invasively in Duchenne and Becker muscular dystrophies. Journal of Cachexia, Sarcopenia and Muscle, 2018, 9, 715-726.	7.3	47
72	MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscular Disorders, 2019, 29, 827-841.	0.6	46

#	Article	IF	Citations
73	Exploration of New Contrasts, Targets, and MR Imaging and Spectroscopy Techniques for Neuromuscular Disease â€“ A Workshop Report of Working Group 3 of the Biomedicine and Molecular Biosciences COST Action BM1304 MYO-MRI. Journal of Neuromuscular Diseases, 2019, 6, 1-30.	2.6	46
74	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	7.6	45
75	Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. Neurology, 2020, 94, e1094-e1102.	1.1	45
76	Exome sequences versus sequential gene testing in the UK highly specialised Service for Limb Girdle Muscular Dystrophy. Orphanet Journal of Rare Diseases, 2017, 12, 151.	2.7	44
77	Development and Application of an Ultrasensitive Hybridization-Based ELISA Method for the Determination of Peptide-Conjugated Phosphorodiamidate Morpholino Oligonucleotides. Nucleic Acid Therapeutics, 2015, 25, 275-284.	3.6	43
78	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	3.6	43
79	Effect of Different Corticosteroid Dosing Regimens on Clinical Outcomes in Boys With Duchenne Muscular Dystrophy. JAMA - Journal of the American Medical Association, 2022, 327, 1456.	7.4	43
80	The Diagnostic Value of MRI Pattern Recognition in Distal Myopathies. Frontiers in Neurology, 2018, 9, 456.	2.4	42
81	Interventions for muscular dystrophy: molecular medicines entering the clinic. Lancet, The, 2009, 374, 1849-1856.	13.7	41
82	Detection of variants in dystroglycanopathy-associated genes through the application of targeted whole-exome sequencing analysis to a large cohort of patients with unexplained limb-girdle muscle weakness. Skeletal Muscle, 2018, 8, 23.	4.2	40
83	Complex phenotypes associated with STIM1 mutations in both coiled coil and EF-hand domains. Neuromuscular Disorders, 2017, 27, 861-872.	0.6	39
84	Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 576-585.	1.9	38
85	Respiratory and upper limb function as outcome measures in ambulant and non-ambulant subjects with Duchenne muscular dystrophy: A prospective multicentre study. Neuromuscular Disorders, 2019, 29, 261-268.	0.6	36
86	Albumin targeting of damaged muscle fibres in the mdx mouse can be monitored by MRI. Neuromuscular Disorders, 2004, 14, 791-796.	0.6	35
87	Novel mutations in DNAJB6 gene cause a very severe early-onset limb-girdle muscular dystrophy 1D disease. Neuromuscular Disorders, 2015, 25, 835-842.	0.6	35
88	Where do we stand in trial readiness for autosomal recessive limb girdle muscular dystrophies?. Neuromuscular Disorders, 2016, 26, 111-125.	0.6	31
89	MSTO1 mutations cause mtDNA depletion, manifesting as muscular dystrophy with cerebellar involvement. Acta Neuropathologica, 2019, 138, 1013-1031.	7.7	31
90	Two recurrent mutations are associated with GNE myopathy in the North of Britain. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1359-1365.	1.9	30

#	Article	IF	Citations
91	Prophylactic oral bisphosphonate therapy in duchenne muscular dystrophy. Muscle and Nerve, 2016, 54, 79-85.	2.2	30
92	Respiratory involvement in ambulant and non-ambulant patients with facioscapulohumeral muscular dystrophy. Journal of Neurology, 2017, 264, 1271-1280.	3.6	30
93	The clinical-phenotype continuum in DYNC1H1-related disordersâ€"genomic profiling and proposal for a novel classification. Journal of Human Genetics, 2020, 65, 1003-1017.	2.3	30
94	POGLUT1 biallelic mutations cause myopathy with reduced satellite cells, Î±-dystroglycan hypoglycosylation and a distinctive radiological pattern. Acta Neuropathologica, 2020, 139, 565-582.	7.7	29
95	Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. Brain, 2017, 140, 37-48.	7.6	28
96	Natural history of limb girdle muscular dystrophy R9 over 6Âyears: searching for trial endpoints. Annals of Clinical and Translational Neurology, 2019, 6, 1033-1045.	3.7	28
97	Recurrent TTN metatranscriptâ€only c.39974â€"11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. Human Mutation, 2020, 41, 403-411.	2.5	28
98	Phenotypic variability of TRPV4 related neuropathies. Neuromuscular Disorders, 2015, 25, 516-521.	0.6	27
99	Clinical and neuroimaging findings in two brothers with limb girdle muscular dystrophy due to LAMA2 mutations. Neuromuscular Disorders, 2017, 27, 170-174.	0.6	27
100	SMCHD1 mutation spectrum for facioscapulohumeral muscular dystrophy type 2 (FSHD2) and Bosma arhinia microphthalmia syndrome (BAMS) reveals disease-specific localisation of variants in the ATPase domain. Journal of Medical Genetics, 2019, 56, 693-700.	3.2	27
101	POPDC3 Gene Variants Associate with a New Form of Limb Girdle Muscular Dystrophy. Annals of Neurology, 2019, 86, 832-843.	5.3	27
102	Longitudinal serum biomarker screening identifies malate dehydrogenase 2 as candidate prognostic biomarker for Duchenne muscular dystrophy. Journal of Cachexia, Sarcopenia and Muscle, 2020, 11, 505-517.	7.3	27
103	The TREAT-NMD care and trial site registry: an online registry to facilitate clinical research for neuromuscular diseases. Orphanet Journal of Rare Diseases, 2013, 8, 171.	2.7	26
104	Elusive sources of variability of dystrophin rescue by exon skipping. Skeletal Muscle, 2015, 5, 44.	4.2	26
105	A checklist for clinical trials in rare disease: obstacles and anticipatory actionsâ€"lessons learned from the FOR-DMD trial. Trials, 2018, 19, 291.	1.6	26
106	Muscular dystrophy with arrhythmia caused by loss-of-function mutations in BVES. Neurology: Genetics, 2019, 5, e321.	1.9	26
107	Mutational spectrum and phenotypic variability of VCP-related neurological disease in the UK. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 680-681.	1.9	25
108	Re-evaluation of the phenotype caused by the commonMATR3p.Ser85Cys mutation in a new family. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 448-450.	1.9	24

#	Article	IF	Citations
109	Economic Evaluation in Duchenne Muscular Dystrophy: Model Frameworks for Cost-Effectiveness Analysis. Pharmacoeconomics, 2017, 35, 249-258.	3.3	24
110	FSHD type 2 and Bosma arhinia microphthalmia syndrome. Neurology, 2018, 91, e562-e570.	1.1	24
111	The UK Myotonic Dystrophy Patient Registry: facilitating and accelerating clinical research. Journal of Neurology, 2017, 264, 979-988.	3.6	23
112	<scp>GGPS1</scp> Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. Annals of Neurology, 2020, 88, 332-347.	5.3	22
113	The Latin American experience with a next generation sequencing genetic panel for recessive limb-girdle muscular weakness and Pompe disease. Orphanet Journal of Rare Diseases, 2020, 15, 11.	2.7	22
114	The TREAT-NMD advisory committee for therapeutics (TACT): an innovative de-risking model to foster orphan drug development. Orphanet Journal of Rare Diseases, 2015, 10, 49.	2.7	21
115	Identification of GAA variants through whole exome sequencing targeted to a cohort of 606 patients with unexplained limb-girdle muscle weakness. Orphanet Journal of Rare Diseases, 2017, 12, 173.	2.7	21
116	Limb girdle muscular dystrophy due to mutations in POMT2. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 506-512.	1.9	21
117	Conserved expression of truncated telethonin in a patient with limb-girdle muscular dystrophy 2G. Neuromuscular Disorders, 2015, 25, 349-352.	0.6	20
118	Assessment of disease progression in dysferlinopathy. Neurology, 2019, 92, .	1.1	20
119	Global FKRP Registry: observations in more than 300 patients with Limb Girdle Muscular Dystrophy R9. Annals of Clinical and Translational Neurology, 2020, 7, 757-766.	3.7	20
120	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. Nature Communications, 2022, 13, 2306.	12.8	20
121	Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1224-1226.	1.9	19
122	Subepicardial dysfunction leads to global left ventricular systolic impairment in patients with limb girdle muscular dystrophy 2I. European Journal of Heart Failure, 2013, 15, 986-994.	7.1	18
123	A novel recessive TTN founder variant is a common cause of distal myopathy in the Serbian population. European Journal of Human Genetics, 2017, 25, 572-581.	2.8	18
124	A â€˜second truncationâ€™ in TTN causes early onset recessive muscular dystrophy. Neuromuscular Disorders, 2017, 27, 1009-1017.	0.6	18
125	Improving translatability of preclinical studies for neuromuscular disorders: lessons from the TREAT-NMD Advisory Committee for Therapeutics (TACT). DMM Disease Models and Mechanisms, 2020, 13, .	2.4	18
126	High prevalence of plasma lipid abnormalities in human and canine Duchenne and Becker muscular dystrophies depicts a new type of primary genetic dyslipidemia. Journal of Clinical Lipidology, 2020, 14, 459-469.e0.	1.5	18

#	Article	IF	Citations
127	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. Journal of Neuromuscular Diseases, 2020, 7, 153-166.	2.6	18
128	Skeletal muscle magnetic resonance imaging in <scp>Pompe</scp> disease. Muscle and Nerve, 2021, 63, 640-650.	2.2	18
129	Miyoshi myopathy and limb girdle muscular dystrophy R2 are the same disease. Neuromuscular Disorders, 2021, 31, 265-280.	0.6	18
130	Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. Annals of Neurology, 2021, 89, 967-978.	5.3	17
131	Deep RNA profiling identified clock and molecular clock genes as pathophysiological signatures in collagen VI myopathy. Journal of Cell Science, 2016, 129, 1671-84.	2.0	16
132	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. European Journal of Neurology, 2020, 27, 2604-2615.	3.3	16
133	Long-term Safety and Efficacy of Avalglucosidase Alfa in Patients With Late-Onset Pompe Disease. Neurology, 2022, 99, .	1.1	16
134	Mobility shift of beta-dystroglycan as a marker ofGMPPBgene-related muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 762-768.	1.9	15
135	A new mutation of the SCGA gene is the cause of a late onset mild phenotype limb girdle muscular dystrophy type 2D with axial involvement. Neuromuscular Disorders, 2018, 28, 633-638.	0.6	15
136	Multisystem proteinopathy due to a homozygous p.Arg159His VCP mutation. Neurology, 2020, 94, e785-e796.	1.1	15
137	Facioscapulohumeral muscular dystrophy 1 patients participating in the UK FSHD registry can be subdivided into 4 patterns of self-reported symptoms. Neuromuscular Disorders, 2020, 30, 315-328.	0.6	15
138	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
139	Guidance in Social and Ethical Issues Related to Clinical, Diagnostic Care and Novel Therapies for Hereditary Neuromuscular Rare Diseases: â€œTranslatingâ€œ the Translational. PLOS Currents, 2013, 5, .	1.4	15
140	MEGF10 related myopathies: A new case with adult onset disease with prominent respiratory failure and review of reported phenotypes. Neuromuscular Disorders, 2018, 28, 48-53.	0.6	13
141	Normalized grip strength is a sensitive outcome measure through all stages of Duchenne muscular dystrophy. Journal of Neurology, 2020, 267, 2022-2028.	3.6	13
142	Cardiac involvement in hereditary myopathy with early respiratory failure. Neurology, 2016, 87, 1031-1035.	1.1	12
143	The effects of ageing on mouse muscle microstructure: a comparative study of timeâ€dependent diffusion MRI and histological assessment. NMR in Biomedicine, 2018, 31, e3881.	2.8	12
144	Noninvasive quantification of fibrosis in skeletal and cardiac muscle in mdx mice using EP3533 enhanced magnetic resonance imaging. Magnetic Resonance in Medicine, 2019, 81, 2728-2735.	3.0	12

#	Article	IF	Citations
145	A decade of optimizing drug development for rare neuromuscular disorders through TACT. Nature Reviews Drug Discovery, 2020, 19, 1-2.	46.4	12
146	Threeâ€year quantitative magnetic resonance imaging and phosphorus magnetic resonance spectroscopy study in lower limb muscle in dysferlinopathy. Journal of Cachexia, Sarcopenia and Muscle, 2022, 13, 1850-1863.	7.3	12
147	S151A Î´-sarcoglycan mutation causes a mild phenotype of cardiomyopathy in mice. European Journal of Human Genetics, 2014, 22, 119-125.	2.8	11
148	The Role of Muscle Imaging in the Diagnosis and Assessment of Children with Genetic Muscle Disease. Neuropediatrics, 2017, 48, 233-241.	0.6	11
149	BAG3 myopathy is not always associated with cardiomyopathy. Neuromuscular Disorders, 2018, 28, 798-801.	0.6	11
150	A novel compound heterozygous mutation in the POMK gene causing limb-girdle muscular dystrophy-dystroglycanopathy in a sib pair. Neuromuscular Disorders, 2018, 28, 614-618.	0.6	11
151	ANO5 mutations in the Polish limb girdle muscular dystrophy patients: Effects on the protein structure. Scientific Reports, 2019, 9, 11533.	3.3	11
152	Extending the clinical and mutational spectrum ofTRIM32-related myopathies in a non-Hutterite population. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 490-493.	1.9	11
153	247th ENMC International Workshop: Muscle magnetic resonance imaging - Implementing muscle MRI as a diagnostic tool for rare genetic myopathy cohorts. Hoofddorp, The Netherlands, September 2019. Neuromuscular Disorders, 2020, 30, 938-947.	0.6	11
154	Clinical and genetic spectrum of a large cohort of patients with Î´-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606.	7.6	11
155	A homozygous DPM3 mutation in a patient with alpha-dystroglycan-related limb girdle muscular dystrophy. Neuromuscular Disorders, 2017, 27, 1043-1046.	0.6	10
156	Psychometric analysis of the pediatric quality of life inventory 3.0 neuromuscular module administered to patients with duchenne muscular dystrophy: A rasch analysis. Muscle and Nerve, 2018, 58, 367-373.	2.2	10
157	Psychometric properties of the Zarit Caregiver Burden Interview administered to caregivers to patients with Duchenne muscular dystrophy: a Rasch analysis. Disability and Rehabilitation, 2019, 41, 966-973.	1.8	10
158	One gene, one or many diseases?: Simplifying dysferlinopathy. Neurology, 2010, 75, 298-299.	1.1	9
159	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. Frontiers in Genetics, 2020, 11, 605.	2.3	9
160	Cardiac and pulmonary findings in dysferlinopathy: A 3â€year, longitudinal study. Muscle and Nerve, 2022, 65, 531-540.	2.2	9
161	Psycho-organic symptoms as early manifestation of adult onset POMT1-related limb girdle muscular dystrophy. Neuromuscular Disorders, 2014, 24, 990-992.	0.6	8
162	Resting-state functional MRI shows altered default-mode network functional connectivity in Duchenne muscular dystrophy patients. Brain Imaging and Behavior, 2021, 15, 2297-2307.	2.1	8

#	Article	IF	Citations
163	A comparison of the bone and growth phenotype of mdx, mdx:cmahâ^'/â^' and mdx:utrn+/â^' murine models with the C57BL10 wildtype mouse. DMM Disease Models and Mechanisms, 2020, 13, .	2.4	7
164	Timeâ€dependent diffusion MRI as a probe of microstructural changes in a mouse model of Duchenne muscular dystrophy. NMR in Biomedicine, 2020, 33, e4276.	2.8	7
165	Pubertal induction in adolescents with DMD is associated with high satisfaction, gonadotropin release and increased muscle contractile surface area. European Journal of Endocrinology, 2021, 184, 67-79.	3.7	7
166	Bones and muscular dystrophies: what do we know?. Current Opinion in Neurology, 2018, 31, 583-591.	3.6	6
167	Fat oxidation is impaired during exercise in lipin-1 deficiency. Neurology, 2019, 93, e1433-e1438.	1.1	6
168	North Star Ambulatory Assessment changes in ambulant Duchenne boys amenable to skip exons 44, 45, 51, and 53: A 3 year follow up. PLoS ONE, 2021, 16, e0253882.	2.5	6
169	The impact of integrated omics technologies for patients with rare diseases. Expert Opinion on Orphan Drugs, 2014, 2, 1211-1219.	0.8	5
170	Bisphosphonate use in Duchenne Muscular Dystrophy â€“ why, when to start and when to stop?. Expert Opinion on Orphan Drugs, 2016, 4, 407-416.	0.8	5
171	227 th ENMC International Workshop:. Neuromuscular Disorders, 2018, 28, 185-192.	0.6	5
172	Undiagnosed Genetic Muscle Disease in the North of England: an in Depth Phenotype Analysis. PLOS Currents, 2013, 5, .	1.4	5
173	Combined growth hormone and insulin-like growth factor-1 rescues growth retardation in glucocorticoid-treated mdxmice but does not prevent osteopenia. Journal of Endocrinology, 2022, 253, 63-74.	2.6	5
174	Ultrasensitive Hybridization-Based ELISA Method for the Determination of Phosphorodiamidate Morpholino Oligonucleotides in Biological samples. Methods in Molecular Biology, 2017, 1565, 265-277.	0.9	4
175	Observational study of clinical outcomes for testosterone treatment of pubertal delay in Duchenne muscular dystrophy. BMC Pediatrics, 2019, 19, 131.	1.7	4
176	Health related quality of life in young, steroid-naÃ¯ve boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 1161-1168.	0.6	4
177	The impact of testosterone therapy on quality of life in adolescents with Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 1259-1265.	0.6	4
178	Intellectual disability in paediatric patients with genetic muscle diseases. Neuromuscular Disorders, 2021, 31, 988-997.	0.6	4
179	Clinicoâ€genetic spectrum of limbâ€girdle muscular weakness in Austria: A multicentre cohort study. European Journal of Neurology, 2022, , .	3.3	4
180	Assessing the Relationship of Patient Reported Outcome Measures With Functional Status in Dysferlinopathy: A Rasch Analysis Approach. Frontiers in Neurology, 2022, 13, 828525.	2.4	4

#	Article	IF	Citations
181	P2.23 Exploratory Rasch analysis of adapted North Star ambulatory assessment in LGMD 2I. Neuromuscular Disorders, 2011, 21, 667.	0.6	3
182	Use of EP3533-Enhanced Magnetic Resonance Imaging as a Measure of Disease Progression in Skeletal Muscle of mdx Mice. Frontiers in Neurology, 2021, 12, 636719.	2.4	3
183	Patient reported quality of life in limb girdle muscular dystrophy. Neuromuscular Disorders, 2022, 32, 57-64.	0.6	3
184	Comparison of strength testing modalities in dysferlinopathy. Muscle and Nerve, 2022, 66, 159-166.	2.2	3
185	Prevalence of Pain within Limb Girdle Muscular Dystrophy R9 and Implications for Other Degenerative Diseases. Journal of Clinical Medicine, 2021, 10, 5517.	2.4	2
186	A Phase 4 Prospective Study in Patients with Adult Pompe Disease Treated with Alglucosidase Alfa. Journal of Neuromuscular Diseases, 2015, 2, S72-S73.	2.6	2
187	A Phase 4 Prospective Study in Patients with Adult Pompe Disease Treated with Alglucosidase Alfa. Journal of Neuromuscular Diseases, 2015, 2, S72-S73.	2.6	1
188	North Star Assessment for dysferlinopathy: Longitudinal performance in the clinical outcome study of dysferlinopathy. Neuromuscular Disorders, 2017, 27, S145.	0.6	1
189	P.183Functional progression in dysferlinopathy: results of a 3-year natural history study. Neuromuscular Disorders, 2019, 29, S102.	0.6	1
190	Effects of Chronic, Maximal Phosphorodiamidate Morpholino Oligomer (PMO) Dosing on Muscle Function and Dystrophin Restoration in a Mouse Model of Duchenne Muscular Dystrophy. Journal of Neuromuscular Diseases, 2021, 8, S369-S381.	2.6	1
191	FXR1-related congenital myopathy: expansion of the clinical and genetic spectrum. Journal of Medical Genetics, 2022, 59, 1069-1074.	3.2	1
192	Neuromuscular Diseases Causing Floppy Infant Syndrome. Pediatric Research, 2011, 70, 27-27.	2.3	0
193	1106â€...The clinical phenotypic spectrum of GFPT1 associated congenital myasthenic syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, e1.79-e1.	1.9	0
194	A cryptic intronic LAMA2 insertion in a boy with mild congenital muscular dystrophy type 1A. Neuromuscular Disorders, 2021, 31, 660-665.	0.6	0
195	TREAT-NMD stakeholder meeting for natural history studies in limb girdle muscular dystrophy 18th June 2019, Amsterdam, The Netherlands. Neuromuscular Disorders, 2021, 31, 899-906.	0.6	0
196	Muskel-MRT bei Myopathien des Kindes- und Jugendalters. Nervenheilkunde, 2019, 38, .	0.0	0
197	Study Design of STR1VE-EU, a Phase 3 Trial of AVXS-101 Gene-Replacement Therapy (GRT) in Patients With Spinal Muscular Atrophy Type 1 (SMA1) in Europe. , 2019, 50, .		0