Volker Straub

List of Publications by Citations

181 papers	8,310 citations	50 h-index	86 g-index
206 ext. papers	10,206 ext. citations	5.9 avg, IF	5.47 L-index

#	Paper	IF	Citations
181	The Human Phenotype Ontology in 2017. *Nucleic Acids Research*, **2017**, 45, D865-D876	20.1	507
180	Animal models for muscular dystrophy show different patterns of sarcolemmal disruption. *Journal of Cell Biology*, **1997**, 139, 375-85	7.3	411
179	Prevalence of genetic muscle disease in Northern England: in-depth analysis of a muscle clinic population. *Brain*, **2009**, 132, 3175-86	11.2	340
178	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. *Science Translational Medicine*, **2017**, 9,	17.5	338
177	The TREAT-NMD DMD Global Database: analysis of more than 7,000 Duchenne muscular dystrophy mutations. *Human Mutation*, **2015**, 36, 395-402	4.7	338
176	Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. *Brain*, **2007**, 130, 2725-35	11.2	322
175	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, 987-96	24.1	249
174	Managing Duchenne muscular dystrophy--the additive effect of spinal surgery and home nocturnal ventilation in improving survival. *Neuromuscular Disorders*, **2007**, 17, 470-5	2.9	215
173	Mutations in the FKRP gene can cause muscle-eye-brain disease and Walker-Warburg syndrome. *Journal of Medical Genetics*, **2004**, 41, e61	5.8	197
172	Phenotypic spectrum associated with mutations in the fukutin-related protein gene. *Annals of Neurology*, **2003**, 53, 537-42	9.4	193
171	A founder mutation in Anoctamin 5 is a major cause of limb-girdle muscular dystrophy. *Brain*, **2011**, 134, 171-182	11.2	157
170	Secondary calpain3 deficiency in 2q-linked muscular dystrophy: titin is the candidate gene. *Neurology*, **2001**, 56, 869-77	6.5	151
169	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	5.5	146
168	Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. *Human Mutation*, **2012**, 33, 981-8	4.7	119
167	Quantitative muscle MRI as an assessment tool for monitoring disease progression in LGMD2I: a multicentre longitudinal study. *PLoS ONE*, **2013**, 8, e70993	3.7	116
166	Genotype-phenotype correlation in a large population of muscular dystrophy patients with LAMA2 mutations. *Neuromuscular Disorders*, **2010**, 20, 241-50	2.9	114
165	Serum matrix metalloproteinase-9 (MMP-9) as a biomarker for monitoring disease progression in Duchenne muscular dystrophy (DMD). *Neuromuscular Disorders*, **2011**, 21, 569-78	2.9	110

164	The burden of Duchenne muscular dystrophy: an international, cross-sectional study. *Neurology*, **2014**, 83, 529-36	6.5	108
163	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	4.8	100
162	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	5	84
161	Affinity proteomics within rare diseases: a BIO-NMD study for blood biomarkers of muscular dystrophies. *EMBO Molecular Medicine*, **2014**, 6, 918-36	12	81
160	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, 1-2 October 2009, Paris, France. *Neuromuscular Disorders*, **2012**, 22 Suppl 2, S54-67	2.9	81
159	The TREAT-NMD Duchenne muscular dystrophy registries: conception, design, and utilization by industry and academia. *Human Mutation*, **2013**, 34, 1449-57	4.7	78
158	Inheritance patterns and phenotypic features of myofibrillar myopathy associated with a BAG3 mutation. *Neuromuscular Disorders*, **2010**, 20, 438-42	2.9	77
157	TREAT-NMD workshop: pattern recognition in genetic muscle diseases using muscle MRI: 25-26 February 2011, Rome, Italy. *Neuromuscular Disorders*, **2012**, 22 Suppl 2, S42-53	2.9	73
156	Muscle magnetic resonance imaging in congenital myopathies due to ryanodine receptor type 1 gene mutations. *Archives of Neurology*, **2011**, 68, 1171-9		72
155	New aspects on patients affected by dysferlin deficient muscular dystrophy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2010**, 81, 946-53	5.5	63
154	Magnetic resonance imaging in Duchenne muscular dystrophy: longitudinal assessment of natural history over 18 months. *Muscle and Nerve*, **2013**, 48, 586-8	3.4	62
153	Quantitative magnetic resonance imaging in limb-girdle muscular dystrophy 2I: a multinational cross-sectional study. *PLoS ONE*, **2014**, 9, e90377	3.7	62
152	Validation of genetic modifiers for Duchenne muscular dystrophy: a multicentre study assessing SPP1 and LTBP4 variants. *Journal of Neurology, Neurosurgery and Psychiatry*, **2015**, 86, 1060-5	5.5	61
151	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, 152-63	5.5	60
150	Muscle MRI findings in limb girdle muscular dystrophy type 2L. *Neuromuscular Disorders*, **2012**, 22 Suppl 2, S122-9	2.9	60
149	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. *Annals of Neurology*, **2018**, 83, 1105-1124	9.4	59
148	Stakeholder cooperation to overcome challenges in orphan medicine development: the example of Duchenne muscular dystrophy. *Lancet Neurology, The*, **2016**, 15, 882-890	24.1	58
147	Titin founder mutation is a common cause of myofibrillar myopathy with early respiratory failure. *Journal of Neurology, Neurosurgery and Psychiatry*, **2014**, 85, 331-8	5.5	58

146	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. *Journal of Neurology*, **2012**, 259, 838-50	5.5	58
145	Contrast agent-enhanced magnetic resonance imaging of skeletal muscle damage in animal models of muscular dystrophy. *Magnetic Resonance in Medicine*, **2000**, 44, 655-9	4.4	58
144	Quantifying the burden of caregiving in Duchenne muscular dystrophy. *Journal of Neurology*, **2016**, 263, 906-915	5.5	56
143	A heterozygous 21-bp deletion in CAPN3 causes dominantly inherited limb girdle muscular dystrophy. *Brain*, **2016**, 139, 2154-63	11.2	56
142	Fibronectin is a serum biomarker for Duchenne muscular dystrophy. *Proteomics - Clinical Applications*, **2014**, 8, 269-78	3.1	55
141	Limb-girdle muscular dystrophies - international collaborations for translational research. *Nature Reviews Neurology*, **2016**, 12, 294-309	15	55
140	MR imaging in Duchenne muscular dystrophy: quantification of T1-weighted signal, contrast uptake, and the effects of exercise. *Journal of Magnetic Resonance Imaging*, **2009**, 30, 1130-8	5.6	54
139	Health-related quality of life in patients with Duchenne muscular dystrophy: a multinational, cross-sectional study. *Developmental Medicine and Child Neurology*, **2016**, 58, 508-15	3.3	54
138	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	2.9	53
137	Biochemical characterization of patients with in-frame or out-of-frame DMD deletions pertinent to exon 44 or 45 skipping. *JAMA Neurology*, **2014**, 71, 32-40	17.2	52
136	Congenital muscular dystrophy with laminin alpha 2 chain deficiency: identification of a new intermediate phenotype and correlation of clinical findings to muscle immunohistochemistry. *European Journal of Pediatrics*, **1996**, 155, 968-76	4.1	52
135	Dystrophin quantification: Biological and translational research implications. *Neurology*, **2014**, 83, 2062-9	8.5	51
134	Muscle-Derived Proteins as Serum Biomarkers for Monitoring Disease Progression in Three Forms of Muscular Dystrophy. *Journal of Neuromuscular Diseases*, **2015**, 2, 241-255	5	50
133	Improving recognition of Duchenne muscular dystrophy: a retrospective case note review. *Archives of Disease in Childhood*, **2014**, 99, 1074-7	2.2	48
132	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. *Journal of Neuromuscular Diseases*, **2014**, 1, 75-90	5	47
131	The Classification, Natural History and Treatment of the Limb Girdle Muscular Dystrophies. *Journal of Neuromuscular Diseases*, **2015**, 2, S7-S19	5	47
130	The Clinical Outcome Study for dysferlinopathy: An international multicenter study. *Neurology: Genetics*, **2016**, 2, e89	3.8	44
129	Late onset in dysferlinopathy widens the clinical spectrum. *Neuromuscular Disorders*, **2008**, 18, 288-90	2.9	44

128	Short stature and pubertal delay in Duchenne muscular dystrophy. *Archives of Disease in Childhood*, **2016**, 101, 101-6	2.2	43
127	Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 1071-1081	5.5	43
126	Association Study of Exon Variants in the NF-B and TGF Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. *American Journal of Human Genetics*, **2016**, 99, 1163-1171	11	42
125	Prevalence of Pompe disease in 3,076 patients with hyperCKemia and limb-girdle muscular weakness. *Neurology*, **2016**, 87, 295-8	6.5	42
124	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	3.8	41
123	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	11	40
122	Compliance to Care Guidelines for Duchenne Muscular Dystrophy. *Journal of Neuromuscular Diseases*, **2015**, 2, 63-72	5	40
121	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. *Contemporary Clinical Trials*, **2017**, 58, 34-39	2.3	39
120	Interventions for muscular dystrophy: molecular medicines entering the clinic. *Lancet, The*, **2009**, 374, 1849-56	40	36
119	Reduced serum myostatin concentrations associated with genetic muscle disease progression. *Journal of Neurology*, **2017**, 264, 541-553	5.5	34
118	MRI in sarcoglycanopathies: a large international cohort study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 72-77	5.5	34
117	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, 115-23	3.7	34
116	Albumin targeting of damaged muscle fibres in the mdx mouse can be monitored by MRI. *Neuromuscular Disorders*, **2004**, 14, 791-6	2.9	33
115	Exploration of New Contrasts, Targets, and MR Imaging and Spectroscopy Techniques for Neuromuscular Disease - 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	5	32
114	Tracking disease progression non-invasively in Duchenne and Becker muscular dystrophies. *Journal of Cachexia, Sarcopenia and Muscle*, **2018**, 9, 715-726	10.3	32
113	Fractures and Linear Growth in a Nationwide Cohort of Boys With Duchenne Muscular Dystrophy With and Without Glucocorticoid Treatment: Results From the UK NorthStar Database. *JAMA Neurology*, **2019**, 76, 701-709	17.2	29
112	Development and Application of an Ultrasensitive Hybridization-Based ELISA Method for the Determination of Peptide-Conjugated Phosphorodiamidate Morpholino Oligonucleotides. *Nucleic Acid Therapeutics*, **2015**, 25, 275-84	4.8	29
111	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	4.2	27

110	Safety, tolerability, pharmacokinetics, pharmacodynamics, and exploratory efficacy of the novel enzyme replacement therapy avalglucosidase alfa (neoGAA) in treatment-naïve and alglucosidase alfa-treated patients with late-onset Pompe disease: A phase 1, open-label, multicenter, multinational, ascending dose study. *Neuromuscular Disorders*, **2019**, 29, 167-186	2.9	27
109	Novel mutations in DNAJB6 gene cause a very severe early-onset limb-girdle muscular dystrophy 1D disease. *Neuromuscular Disorders*, **2015**, 25, 835-42	2.9	26
108	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	5.1	26
107	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. *Genetics in Medicine*, **2020**, 22, 1478-1488	8.1	25
106	Respiratory involvement in ambulant and non-ambulant patients with facioscapulohumeral muscular dystrophy. *Journal of Neurology*, **2017**, 264, 1271-1280	5.5	24
105	Prophylactic oral bisphosphonate therapy in duchenne muscular dystrophy. *Muscle and Nerve*, **2016**, 54, 79-85	3.4	24
104	Where do we stand in trial readiness for autosomal recessive limb girdle muscular dystrophies?. *Neuromuscular Disorders*, **2016**, 26, 111-25	2.9	24
103	Complex phenotypes associated with STIM1 mutations in both coiled coil and EF-hand domains. *Neuromuscular Disorders*, **2017**, 27, 861-872	2.9	23
102	MYO-MRI diagnostic protocols in genetic myopathies. *Neuromuscular Disorders*, **2019**, 29, 827-841	2.9	23
101	Two recurrent mutations are associated with GNE myopathy in the North of Britain. *Journal of Neurology, Neurosurgery and Psychiatry*, **2014**, 85, 1359-65	5.5	23
100	Phenotypic variability of TRPV4 related neuropathies. *Neuromuscular Disorders*, **2015**, 25, 516-21	2.9	22
99	The Diagnostic Value of MRI Pattern Recognition in Distal Myopathies. *Frontiers in Neurology*, **2018**, 9, 456	4.1	22
98	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	4.2	22
97	Elusive sources of variability of dystrophin rescue by exon skipping. *Skeletal Muscle*, **2015**, 5, 44	5.1	22
96	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. *Nature Medicine*, **2021**, 27, 1197-1204	50.5	22
95	MSTO1 mutations cause mtDNA depletion, manifesting as muscular dystrophy with cerebellar involvement. *Acta Neuropathologica*, **2019**, 138, 1013-1031	14.3	20
94	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	2.9	19
93	Re-evaluation of the phenotype caused by the common MATR3 p.Ser85Cys mutation in a new family. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 448-50	5.5	19

92	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). *Journal of Neurology*, **2020**, 267, 45-56	5.5	19
91	The UK Myotonic Dystrophy Patient Registry: facilitating and accelerating clinical research. *Journal of Neurology*, **2017**, 264, 979-988	5.5	18
90	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	4.2	18
89	Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 576-585	5.5	18
88	Natural history of limb girdle muscular dystrophy R9 over 6 years: searching for trial endpoints. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1033-1045	5.3	17
87	A checklist for clinical trials in rare disease: obstacles and anticipatory actions-lessons learned from the FOR-DMD trial. *Trials*, **2018**, 19, 291	2.8	17
86	Clinical and neuroimaging findings in two brothers with limb girdle muscular dystrophy due to LAMA2 mutations. *Neuromuscular Disorders*, **2017**, 27, 170-174	2.9	17
85	Conserved expression of truncated telethonin in a patient with limb-girdle muscular dystrophy 2G. *Neuromuscular Disorders*, **2015**, 25, 349-52	2.9	16
84	Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. *Neurology*, **2020**, 94, e1094-e1102	6.5	16
83	Mutational spectrum and phenotypic variability of VCP-related neurological disease in the UK. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 680-1	5.5	16
82	FSHD type 2 and Bosma arhinia microphthalmia syndrome: Two faces of the same mutation. *Neurology*, **2018**, 91, e562-e570	6.5	16
81	POGLUT1 biallelic mutations cause myopathy with reduced satellite cells, α-dystroglycan hypoglycosylation and a distinctive radiological pattern. *Acta Neuropathologica*, **2020**, 139, 565-582	14.3	16
80	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. *Brain*, **2020**, 143, 2696-2708	11.2	15
79	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	5.3	14
78	Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 1224-1226	5.5	14
77	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	5.8	14
76	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	4.2	14
75	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	10.3	14

74	Mobility shift of beta-dystroglycan as a marker of gene-related muscular dystrophy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 762-768	5.5	13
73	Economic Evaluation in Duchenne Muscular Dystrophy: Model Frameworks for Cost-Effectiveness Analysis. *Pharmacoeconomics*, **2017**, 35, 249-258	4.4	13
72	Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. *Brain*, **2017**, 140, 37-48	11.2	13
71	Subepicardial dysfunction leads to global left ventricular systolic impairment in patients with limb girdle muscular dystrophy 2I. *European Journal of Heart Failure*, **2013**, 15, 986-94	12.3	13
70	Muscular dystrophy with arrhythmia caused by loss-of-function mutations in. *Neurology: Genetics*, **2019**, 5, e321	3.8	12
69	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	4.2	12
68	POPDC3 Gene Variants Associate with a New Form of Limb Girdle Muscular Dystrophy. *Annals of Neurology*, **2019**, 86, 832-843	9.4	12
67	Guidance in social and ethical issues related to clinical, diagnostic care and novel therapies for hereditary neuromuscular rare diseases: "translating" the translational. *PLOS Currents*, **2013**, 5,		12
66	Deep RNA profiling identified CLOCK and molecular clock genes as pathophysiological signatures in collagen VI myopathy. *Journal of Cell Science*, **2016**, 129, 1671-84	5.3	12
65	Limb girdle muscular dystrophy due to mutations in. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 506-512	5.5	12
64	Cardiac involvement in hereditary myopathy with early respiratory failure: A cohort study. *Neurology*, **2016**, 87, 1031-5	6.5	11
63	The clinical-phenotype continuum in DYNC1H1-related disorders-genomic profiling and proposal for a novel classification. *Journal of Human Genetics*, **2020**, 65, 1003-1017	4.3	11
62	Assessment of disease progression in dysferlinopathy: A 1-year cohort study. *Neurology*, **2019**,	6.5	11
61	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. *Journal of Neuromuscular Diseases*, **2020**, 7, 153-166	5	10
60	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	2.9	10
59	Recurrent TTN metatranscript-only c.39974-11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. *Human Mutation*, **2020**, 41, 403-411	4.7	10
58	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,	4.1	10
57	GGPS1 Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. *Annals of Neurology*, **2020**, 88, 332-347	9.4	9

56	The effects of ageing on mouse muscle microstructure: a comparative study of time-dependent diffusion MRI and histological assessment. *NMR in Biomedicine*, **2018**, 31, e3881	4.4	9
55	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	2.9	9
54	A second truncation in TTN causes early onset recessive muscular dystrophy. *Neuromuscular Disorders*, **2017**, 27, 1009-1017	2.9	9
53	Multisystem proteinopathy due to a homozygous p.Arg159His mutation: A tale of the unexpected. *Neurology*, **2020**, 94, e785-e796	6.5	9
52	BAG3 myopathy is not always associated with cardiomyopathy. *Neuromuscular Disorders*, **2018**, 28, 798-801	2.9	8
51	A homozygous DPM3 mutation in a patient with alpha-dystroglycan-related limb girdle muscular dystrophy. *Neuromuscular Disorders*, **2017**, 27, 1043-1046	2.9	8
50	The Role of Muscle Imaging in the Diagnosis and Assessment of Children with Genetic Muscle Disease. *Neuropediatrics*, **2017**, 48, 233-241	1.6	8
49	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*, **2021**,	4.8	8
48	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	4.4	8
47	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	4.9	7
46	Normalized grip strength is a sensitive outcome measure through all stages of Duchenne muscular dystrophy. *Journal of Neurology*, **2020**, 267, 2022-2028	5.5	7
45	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	2.9	7
44	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	2.4	7
43	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	3.4	6
42	ANO5 mutations in the Polish limb girdle muscular dystrophy patients: Effects on the protein structure. *Scientific Reports*, **2019**, 9, 11533	4.9	6
41	S151A Sarcoglycan mutation causes a mild phenotype of cardiomyopathy in mice. *European Journal of Human Genetics*, **2014**, 22, 119-25	5.3	6
40	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	24.1	6
39	Extending the clinical and mutational spectrum of -related myopathies in a non-Hutterite population. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 490-493	5.5	6

38	Skeletal muscle magnetic resonance imaging in Pompe disease. *Muscle and Nerve*, **2021**, 63, 640-650	3.4	6
37	Effect of Different Corticosteroid Dosing Regimens on Clinical Outcomes in Boys With Duchenne Muscular Dystrophy: A Randomized Clinical Trial.. *JAMA - Journal of the American Medical Association*, **2022**,	27.4	6
36	Bisphosphonate use in Duchenne Muscular Dystrophy: why, when to start and when to stop?. *Expert Opinion on Orphan Drugs*, **2016**, 4, 407-416	1.1	5
35	Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. *Annals of Neurology*, **2021**, 89, 967-978	9.4	5
34	Time-dependent diffusion MRI as a probe of microstructural changes in a mouse model of Duchenne muscular dystrophy. *NMR in Biomedicine*, **2020**, 33, e4276	4.4	4
33	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	2.9	4
32	Global FKRP Registry: observations in more than 300 patients with Limb Girdle Muscular Dystrophy R9. *Annals of Clinical and Translational Neurology*, **2020**, 7, 757-766	5.3	4
31	The impact of integrated omics technologies for patients with rare diseases. *Expert Opinion on Orphan Drugs*, **2014**, 2, 1211-1219	1.1	4
30	Undiagnosed genetic muscle disease in the north of England: an in depth phenotype analysis. *PLOS Currents*, **2013**, 5,		4
29	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. *European Journal of Neurology*, **2020**, 27, 2604-2615	6	4
28	Fat oxidation is impaired during exercise in lipin-1 deficiency. *Neurology*, **2019**, 93, e1433-e1438	6.5	4
27	Ultrasensitive Hybridization-Based ELISA Method for the Determination of Phosphorodiamidate Morpholino Oligonucleotides in Biological samples. *Methods in Molecular Biology*, **2017**, 1565, 265-277	1.4	3
26	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. *Frontiers in Genetics*, **2020**, 11, 605	4.5	3
25	Psycho-organic symptoms as early manifestation of adult onset POMT1-related limb girdle muscular dystrophy. *Neuromuscular Disorders*, **2014**, 24, 990-2	2.9	3
24	Miyoshi myopathy and limb girdle muscular dystrophy R2 are the same disease. *Neuromuscular Disorders*, **2021**, 31, 265-280	2.9	3
23	A form of muscular dystrophy associated with pathogenic variants in JAG2. *American Journal of Human Genetics*, **2021**, 108, 840-856	11	3
22	A comparison of the bone and growth phenotype of , and murine models with the C57BL/10 wild-type mouse. *DMM Disease Models and Mechanisms*, **2020**, 13,	4.1	3
21	227 ENMC International Workshop:: Finalizing a plan to guarantee quality in translational research for neuromuscular diseases Heemskerk, Netherlands, 10-11 February 2017. *Neuromuscular Disorders*, **2018**, 28, 185-192	2.9	3

20	Bones and muscular dystrophies: what do we know?. *Current Opinion in Neurology*, **2018**, 31, 583-591	7.1	3
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