Volker Straub

List of Publications by Year in Descending Order

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	Assessing the Relationship of Patient Reported Outcome Measures With Functional Status in Dysferlinopathy: A Rasch Analysis Approach.. *Frontiers in Neurology*, **2022**, 13, 828525	4.1	
180	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
179	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy.. *Nature Communications*, **2022**, 13, 2306	17.4	1
178	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
177	Intellectual disability in paediatric patients with genetic muscle diseases. *Neuromuscular Disorders*, **2021**, 31, 988-997	2.9	0
176	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
175	Miyoshi myopathy and limb girdle muscular dystrophy R2 are the same disease. *Neuromuscular Disorders*, **2021**, 31, 265-280	2.9	3
174	A form of muscular dystrophy associated with pathogenic variants in JAG2. *American Journal of Human Genetics*, **2021**, 108, 840-856	11	3
173	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. *Nature Medicine*, **2021**, 27, 1197-1204	50.5	22
172	Use of EP3533-Enhanced Magnetic Resonance Imaging as a Measure of Disease Progression in Skeletal Muscle of Mice. *Frontiers in Neurology*, **2021**, 12, 636719	4.1	1
171	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	3.7	1
170	A cryptic intronic LAMA2 insertion in a boy with mild congenital muscular dystrophy type 1A. *Neuromuscular Disorders*, **2021**, 31, 660-665	2.9	
169	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	2.9	
168	Skeletal muscle magnetic resonance imaging in Pompe disease. *Muscle and Nerve*, **2021**, 63, 640-650	3.4	6
167	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	6.5	1
166	Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. *Annals of Neurology*, **2021**, 89, 967-978	9.4	5
165	Clinical and genetic spectrum of a large cohort of patients with □sarcoglycan muscular dystrophy. *Brain*, **2021**,	11.2	1

164	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	5	
163	Resting-state functional MRI shows altered default-mode network functional connectivity in Duchenne muscular dystrophy patients. *Brain Imaging and Behavior*, **2021**, 15, 2297-2307	4.1	1
162	GGPS1 Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. *Annals of Neurology*, **2020**, 88, 332-347	9.4	9
161	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
160	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. *Genetics in Medicine*, **2020**, 22, 1478-1488	8.1	25
159	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. *Journal of Neuromuscular Diseases*, **2020**, 7, 153-166	5	10
158	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. *Frontiers in Genetics*, **2020**, 11, 605	4.5	3
157	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
156	Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. *Neurology*, **2020**, 94, e1094-e1102	6.5	16
155	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
154	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
153	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
152	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
151	Recurrent TTN metatranscript-only c.39974-11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. *Human Mutation*, **2020**, 41, 403-411	4.7	10
150	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
149	Multisystem proteinopathy due to a homozygous p.Arg159His mutation: A tale of the unexpected. *Neurology*, **2020**, 94, e785-e796	6.5	9
148	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
147	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	2.9	0

146	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. *European Journal of Neurology*, **2020**, 27, 2604-2615	6	4
145	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
144	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. *Brain*, **2020**, 143, 2696-2708	11.2	15
143	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
142	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
141	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). *Journal of Neurology*, **2020**, 267, 45-56	5.5	19
140	MSTO1 mutations cause mtDNA depletion, manifesting as muscular dystrophy with cerebellar involvement. *Acta Neuropathologica*, **2019**, 138, 1013-1031	14.3	20
139	MYO-MRI diagnostic protocols in genetic myopathies. *Neuromuscular Disorders*, **2019**, 29, 827-841	2.9	23
138	Muscular dystrophy with arrhythmia caused by loss-of-function mutations in. *Neurology: Genetics*, **2019**, 5, e321	3.8	12
137	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
136	Observational study of clinical outcomes for testosterone treatment of pubertal delay in Duchenne muscular dystrophy. *BMC Pediatrics*, **2019**, 19, 131	2.6	2
135	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
134	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
133	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
132	ANO5 mutations in the Polish limb girdle muscular dystrophy patients: Effects on the protein structure. *Scientific Reports*, **2019**, 9, 11533	4.9	6
131	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
130	POPDC3 Gene Variants Associate with a New Form of Limb Girdle Muscular Dystrophy. *Annals of Neurology*, **2019**, 86, 832-843	9.4	12
129	Fat oxidation is impaired during exercise in lipin-1 deficiency. *Neurology*, **2019**, 93, e1433-e1438	6.5	4

128	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
127	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
126	Assessment of disease progression in dysferlinopathy: A 1-year cohort study. *Neurology*, **2019**,	6.5	11
125	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
124	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
123	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
122	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
121	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. *Annals of Neurology*, **2018**, 83, 1105-1124	9.4	59
120	Tracking disease progression non-invasively in Duchenne and Becker muscular dystrophies. *Journal of Cachexia, Sarcopenia and Muscle*, **2018**, 9, 715-726	10.3	32
119	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
118	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
117	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
116	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
115	MRI in sarcoglycanopathies: a large international cohort study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 72-77	5.5	34
114	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
113	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
112	The Diagnostic Value of MRI Pattern Recognition in Distal Myopathies. *Frontiers in Neurology*, **2018**, 9, 456	4.1	22
111	BAG3 myopathy is not always associated with cardiomyopathy. *Neuromuscular Disorders*, **2018**, 28, 798-801	2.9	8

110	FSHD type 2 and Bosma arhinia microphthalmia syndrome: Two faces of the same mutation. *Neurology*, **2018**, 91, e562-e570	6.5	16
109	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
108	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
107	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
106	Limb girdle muscular dystrophy due to mutations in. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 506-512	5.5	12
105	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
104	Bones and muscular dystrophies: what do we know?. *Current Opinion in Neurology*, **2018**, 31, 583-591	7.1	3
103	The Human Phenotype Ontology in 2017. *Nucleic Acids Research*, **2017**, 45, D865-D876	20.1	507
102	Reduced serum myostatin concentrations associated with genetic muscle disease progression. *Journal of Neurology*, **2017**, 264, 541-553	5.5	34
101	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
100	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. *Science Translational Medicine*, **2017**, 9,	17.5	338
99	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. *Contemporary Clinical Trials*, **2017**, 58, 34-39	2.3	39
98	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
97	The UK Myotonic Dystrophy Patient Registry: facilitating and accelerating clinical research. *Journal of Neurology*, **2017**, 264, 979-988	5.5	18
96	Complex phenotypes associated with STIM1 mutations in both coiled coil and EF-hand domains. *Neuromuscular Disorders*, **2017**, 27, 861-872	2.9	23
95	Respiratory involvement in ambulant and non-ambulant patients with facioscapulohumeral muscular dystrophy. *Journal of Neurology*, **2017**, 264, 1271-1280	5.5	24
94	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
93	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

92	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
91	A Second truncation In TTN causes early onset recessive muscular dystrophy. *Neuromuscular Disorders*, **2017**, 27, 1009-1017	2.9	9
90	A homozygous DPM3 mutation in a patient with alpha-dystroglycan-related limb girdle muscular dystrophy. *Neuromuscular Disorders*, **2017**, 27, 1043-1046	2.9	8
89	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
88	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
87	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
86	The Role of Muscle Imaging in the Diagnosis and Assessment of Children with Genetic Muscle Disease. *Neuropediatrics*, **2017**, 48, 233-241	1.6	8
85	Economic Evaluation in Duchenne Muscular Dystrophy: Model Frameworks for Cost-Effectiveness Analysis. *Pharmacoeconomics*, **2017**, 35, 249-258	4.4	13
84	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
83	Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. *Brain*, **2017**, 140, 37-48	11.2	13
82	Short stature and pubertal delay in Duchenne muscular dystrophy. *Archives of Disease in Childhood*, **2016**, 101, 101-6	2.2	43
81	Cardiac involvement in hereditary myopathy with early respiratory failure: A cohort study. *Neurology*, **2016**, 87, 1031-5	6.5	11
80	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
79	The Clinical Outcome Study for dysferlinopathy: An international multicenter study. *Neurology: Genetics*, **2016**, 2, e89	3.8	44
78	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
77	Prophylactic oral bisphosphonate therapy in duchenne muscular dystrophy. *Muscle and Nerve*, **2016** , 54, 79-85	3.4	24
76	Quantifying the burden of caregiving in Duchenne muscular dystrophy. *Journal of Neurology*, **2016**, 263, 906-915	5.5	56
75	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

74	A heterozygous 21-bp deletion in CAPN3 causes dominantly inherited limb girdle muscular dystrophy. *Brain*, **2016**, 139, 2154-63	11.2	56
73	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
72	Where do we stand in trial readiness for autosomal recessive limb girdle muscular dystrophies?. *Neuromuscular Disorders*, **2016**, 26, 111-25	2.9	24
71	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
70	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
69	Limb-girdle muscular dystrophies - international collaborations for translational research. *Nature Reviews Neurology*, **2016**, 12, 294-309	15	55
68	Prevalence of Pompe disease in 3,076 patients with hyperCKemia and limb-girdle muscular weakness. *Neurology*, **2016**, 87, 295-8	6.5	42
67	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
66	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
65	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
64	Conserved expression of truncated telethonin in a patient with limb-girdle muscular dystrophy 2G. *Neuromuscular Disorders*, **2015**, 25, 349-52	2.9	16
63	Phenotypic variability of TRPV4 related neuropathies. *Neuromuscular Disorders*, **2015**, 25, 516-21	2.9	22
62	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
61	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
60	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
59	Compliance to Care Guidelines for Duchenne Muscular Dystrophy. *Journal of Neuromuscular Diseases*, **2015**, 2, 63-72	5	40
58	Elusive sources of variability of dystrophin rescue by exon skipping. *Skeletal Muscle*, **2015**, 5, 44	5.1	22
57	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

56	A Phase 4 Prospective Study in Patients with Adult Pompe Disease Treated with Alglucosidase Alfa. *Journal of Neuromuscular Diseases*, **2015**, 2, S72-S73	5	
55	The Classification, Natural History and Treatment of the Limb Girdle Muscular Dystrophies. *Journal of Neuromuscular Diseases*, **2015**, 2, S7-S19	5	47
54	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
53	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
52	A Phase 4 Prospective Study in Patients with Adult Pompe Disease Treated with Alglucosidase Alfa. *Journal of Neuromuscular Diseases*, **2015**, 2, S72-S73	5	1
51	Fibronectin is a serum biomarker for Duchenne muscular dystrophy. *Proteomics - Clinical Applications*, **2014**, 8, 269-78	3.1	55
50	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
49	Improving recognition of Duchenne muscular dystrophy: a retrospective case note review. *Archives of Disease in Childhood*, **2014**, 99, 1074-7	2.2	48
48	Psycho-organic symptoms as early manifestation of adult onset POMT1-related limb girdle muscular dystrophy. *Neuromuscular Disorders*, **2014**, 24, 990-2	2.9	3
47	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
46	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. *Journal of Neuromuscular Diseases*, **2014**, 1, 75-90	5	47
45	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
44	The burden of Duchenne muscular dystrophy: an international, cross-sectional study. *Neurology*, **2014**, 83, 529-36	6.5	108
43	S151A δ-sarcoglycan mutation causes a mild phenotype of cardiomyopathy in mice. *European Journal of Human Genetics*, **2014**, 22, 119-25	5.3	6
42	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
41	The impact of integrated omics technologies for patients with rare diseases. *Expert Opinion on Orphan Drugs*, **2014**, 2, 1211-1219	1.1	4
40	Dystrophin quantification: Biological and translational research implications. *Neurology*, **2014**, 83, 2062-9	6.5	51
39	Affinity proteomics within rare diseases: a BIO-NMD study for blood biomarkers of muscular dystrophies. *EMBO Molecular Medicine*, **2014**, 6, 918-36	12	81

38	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
37	Quantitative magnetic resonance imaging in limb-girdle muscular dystrophy 2I: a multinational cross-sectional study. *PLoS ONE*, **2014**, 9, e90377	3.7	62
36	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
35	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
34	The TREAT-NMD Duchenne muscular dystrophy registries: conception, design, and utilization by industry and academia. *Human Mutation*, **2013**, 34, 1449-57	4.7	78
33	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
32	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
31	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
30	Undiagnosed genetic muscle disease in the north of England: an in depth phenotype analysis. *PLOS Currents*, **2013**, 5,		4
29	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
28	Muscle MRI findings in limb girdle muscular dystrophy type 2L. *Neuromuscular Disorders*, **2012**, 22 Suppl 2, S122-9	2.9	60
27	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
26	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
25	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
24	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. *Journal of Neurology*, **2012**, 259, 838-50	5.5	58
23	1106 The clinical phenotypic spectrum of GFPT1 associated congenital myasthenic syndrome. *Journal of Neurology, Neurosurgery and Psychiatry*, **2012**, 83, e1.79-e1	5.5	
22	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
21	Neuromuscular Diseases Causing Floppy Infant Syndrome. *Pediatric Research*, **2011**, 70, 27-27	3.2	

20	Muscle magnetic resonance imaging in congenital myopathies due to ryanodine receptor type 1 gene mutations. *Archives of Neurology*, **2011**, 68, 1171-9		72
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