Volker Straub

List of Publications by Year
in descending order

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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	Clinical and genetic spectrum of a large cohort of patients with Î´-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606.	7.6	11
2	Patient reported quality of life in limb girdle muscular dystrophy. Neuromuscular Disorders, 2022, 32, 57-64.	0.6	3
3	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
4	Cardiac and pulmonary findings in dysferlinopathy: A 3â€year, longitudinal study. Muscle and Nerve, 2022, 65, 531-540.	2.2	9
5	Clinicoâ€genetic spectrum of limbâ€girdle muscular weakness in Austria: A multicentre cohort study. European Journal of Neurology, 2022, , .	3.3	4
6	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
7	FXR1-related congenital myopathy: expansion of the clinical and genetic spectrum. Journal of Medical Genetics, 2022, 59, 1069-1074.	3.2	1
8	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
9	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
10	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
11	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. Nature Communications, 2022, 13, 2306.	12.8	20
12	Comparison of strength testing modalities in dysferlinopathy. Muscle and Nerve, 2022, 66, 159-166.	2.2	3
13	Long-term Safety and Efficacy of Avalglucosidase Alfa in Patients With Late-Onset Pompe Disease. Neurology, 2022, 99, .	1.1	16
14	Skeletal muscle magnetic resonance imaging in <scp>Pompe</scp> disease. Muscle and Nerve, 2021, 63, 640-650.	2.2	18
15	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
16	Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. Annals of Neurology, 2021, 89, 967-978.	5.3	17
17	Miyoshi myopathy and limb girdle muscular dystrophy R2 are the same disease. Neuromuscular Disorders, 2021, 31, 265-280.	0.6	18
18	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15

#	Article	IF	Citations
19	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. Nature Medicine, 2021, 27, 1197-1204.	30.7	96
20	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
21	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
22	Health related quality of life in young, steroid-naÃ¯ve boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 1161-1168.	0.6	4
23	A cryptic intronic LAMA2 insertion in a boy with mild congenital muscular dystrophy type 1A. Neuromuscular Disorders, 2021, 31, 660-665.	0.6	0
24	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
25	The impact of testosterone therapy on quality of life in adolescents with Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 1259-1265.	0.6	4
26	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
27	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
28	Intellectual disability in paediatric patients with genetic muscle diseases. Neuromuscular Disorders, 2021, 31, 988-997.	0.6	4
29	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
30	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
31	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
32	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	3.6	43
33	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
34	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
35	Multisystem proteinopathy due to a homozygous p.Arg159His VCP mutation. Neurology, 2020, 94, e785-e796.	1.1	15
36	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

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37	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
38	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. European Journal of Neurology, 2020, 27, 2604-2615.	3.3	16
39	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
40	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	7.6	45
41	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
42	<scp>GGPS1</scp> Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. Annals of Neurology, 2020, 88, 332-347.	5.3	22
43	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
44	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. Genetics in Medicine, 2020, 22, 1478-1488.	2.4	62
45	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. Journal of Neuromuscular Diseases, 2020, 7, 153-166.	2.6	18
46	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. Frontiers in Genetics, 2020, 11, 605.	2.3	9
47	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
48	Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. Neurology, 2020, 94, e1094-e1102.	1.1	45
49	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
50	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
51	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
52	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
53	A decade of optimizing drug development for rare neuromuscular disorders through TACT. Nature Reviews Drug Discovery, 2020, 19, 1-2.	46.4	12
54	ANO5 mutations in the Polish limb girdle muscular dystrophy patients: Effects on the protein structure. Scientific Reports, 2019, 9, 11533.	3.3	11

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55	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
56	POPDC3 Gene Variants Associate with a New Form of Limb Girdle Muscular Dystrophy. Annals of Neurology, 2019, 86, 832-843.	5.3	27
57	P.183Functional progression in dysferlinopathy: results of a 3-year natural history study. Neuromuscular Disorders, 2019, 29, S102.	0.6	1
58	MSTO1 mutations cause mtDNA depletion, manifesting as muscular dystrophy with cerebellar involvement. Acta Neuropathologica, 2019, 138, 1013-1031.	7.7	31
59	MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscular Disorders, 2019, 29, 827-841.	0.6	46
60	Muscular dystrophy with arrhythmia caused by loss-of-function mutations in BVES. Neurology: Genetics, 2019, 5, e321.	1.9	26
61	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
62	Observational study of clinical outcomes for testosterone treatment of pubertal delay in Duchenne muscular dystrophy. BMC Pediatrics, 2019, 19, 131.	1.7	4
63	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
64	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
65	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
66	Fat oxidation is impaired during exercise in lipin-1 deficiency. Neurology, 2019, 93, e1433-e1438.	1.1	6
67	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
68	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
69	Assessment of disease progression in dysferlinopathy. Neurology, 2019, 92, .	1.1	20
70	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
71	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
72	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

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73	Muskel-MRT bei Myopathien des Kindes- und Jugendalters. Nervenheilkunde, 2019, 38, .	0.0	0
74	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
75	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
76	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
77	Tracking disease progression nonâ€invasively in Duchenne and Becker muscular dystrophies. Journal of Cachexia, Sarcopenia and Muscle, 2018, 9, 715-726.	7.3	47
78	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
79	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
80	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
81	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
82	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	1.9	55
83	Limb girdle muscular dystrophy due to mutations in POMT2. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 506-512.	1.9	21
84	227 th ENMC International Workshop:. Neuromuscular Disorders, 2018, 28, 185-192.	0.6	5
85	Bones and muscular dystrophies: what do we know?. Current Opinion in Neurology, 2018, 31, 583-591.	3.6	6
86	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
87	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
88	The Diagnostic Value of MRI Pattern Recognition in Distal Myopathies. Frontiers in Neurology, 2018, 9, 456.	2.4	42
89	BAG3 myopathy is not always associated with cardiomyopathy. Neuromuscular Disorders, 2018, 28, 798-801.	0.6	11
90	FSHD type 2 and Bosma arhinia microphthalmia syndrome. Neurology, 2018, 91, e562-e570.	1.1	24

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91	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
92	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
93	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
94	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
95	Reduced serum myostatin concentrations associated with genetic muscle disease progression. Journal of Neurology, 2017, 264, 541-553.	3.6	51
96	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
97	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
98	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. Contemporary Clinical Trials, 2017, 58, 34-39.	1.8	56
99	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
100	The UK Myotonic Dystrophy Patient Registry: facilitating and accelerating clinical research. Journal of Neurology, 2017, 264, 979-988.	3.6	23
101	Complex phenotypes associated with STIM1 mutations in both coiled coil and EF-hand domains. Neuromuscular Disorders, 2017, 27, 861-872.	0.6	39
102	Respiratory involvement in ambulant and non-ambulant patients with facioscapulohumeral muscular dystrophy. Journal of Neurology, 2017, 264, 1271-1280.	3.6	30
103	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
104	A â€˜second truncationâ€™ in TTN causes early onset recessive muscular dystrophy. Neuromuscular Disorders, 2017, 27, 1009-1017.	0.6	18
105	A homozygous DPM3 mutation in a patient with alpha-dystroglycan-related limb girdle muscular dystrophy. Neuromuscular Disorders, 2017, 27, 1043-1046.	0.6	10
106	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
107	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
108	North Star Assessment for dysferlinopathy: Longitudinal performance in the clinical outcome study of dysferlinopathy. Neuromuscular Disorders, 2017, 27, S145.	0.6	1

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109	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
110	The Role of Muscle Imaging in the Diagnosis and Assessment of Children with Genetic Muscle Disease. Neuropediatrics, 2017, 48, 233-241.	0.6	11
111	Economic Evaluation in Duchenne Muscular Dystrophy: Model Frameworks for Cost-Effectiveness Analysis. Pharmacoeconomics, 2017, 35, 249-258.	3.3	24
112	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
113	Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. Brain, 2017, 140, 37-48.	7.6	28
114	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
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 dystrophies â€" international collaborations for translational research. Nature Reviews Neurology, 2016, 12, 294-309.	10.1	81
117	Prevalence of Pompe disease in 3,076 patients with hyperCKemia and limb-girdle muscular weakness. Neurology, 2016, 87, 295-298.	1.1	60
118	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
119	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
120	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
121	Cardiac involvement in hereditary myopathy with early respiratory failure. Neurology, 2016, 87, 1031-1035.	1.1	12
122	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
123	The Clinical Outcome Study for dysferlinopathy. Neurology: Genetics, 2016, 2, e89.	1.9	75
124	Association Study of Exon Variants in the NF-Î°B and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	6.2	71
125	Prophylactic oral bisphosphonate therapy in duchenne muscular dystrophy. Muscle and Nerve, 2016, 54, 79-85.	2.2	30
126	Quantifying the burden of caregiving in Duchenne muscular dystrophy. Journal of Neurology, 2016, 263, 906-915.	3.6	82

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127	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
128	A heterozygous 21-bp deletion inCAPN3causes dominantly inherited limb girdle muscular dystrophy. Brain, 2016, 139, 2154-2163.	7.6	87
129	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
130	Where do we stand in trial readiness for autosomal recessive limb girdle muscular dystrophies?. Neuromuscular Disorders, 2016, 26, 111-125.	0.6	31
131	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
132	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
133	Short stature and pubertal delay in Duchenne muscular dystrophy. Archives of Disease in Childhood, 2016, 101, 101-106.	1.9	58
134	Compliance to Care Guidelines for Duchenne Muscular Dystrophy. Journal of Neuromuscular Diseases, 2015, 2, 63-72.	2.6	51
135	Elusive sources of variability of dystrophin rescue by exon skipping. Skeletal Muscle, 2015, 5, 44.	4.2	26
136	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
137	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
138	The Classification, Natural History and Treatment of the Limb Girdle Muscular Dystrophies. Journal of Neuromuscular Diseases, 2015, 2, S7-S19.	2.6	72
139	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
140	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
141	Conserved expression of truncated telethonin in a patient with limb-girdle muscular dystrophy 2G. Neuromuscular Disorders, 2015, 25, 349-352.	0.6	20
142	Phenotypic variability of TRPV4 related neuropathies. Neuromuscular Disorders, 2015, 25, 516-521.	0.6	27
143	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
144	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

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145	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
146	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
147	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
148	The burden of Duchenne muscular dystrophy. Neurology, 2014, 83, 529-536.	1.1	149
149	S151A Î´-sarcoglycan mutation causes a mild phenotype of cardiomyopathy in mice. European Journal of Human Genetics, 2014, 22, 119-125.	2.8	11
150	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
151	The impact of integrated omics technologies for patients with rare diseases. Expert Opinion on Orphan Drugs, 2014, 2, 1211-1219.	0.8	5
152	Dystrophin quantification. Neurology, 2014, 83, 2062-2069.	1.1	73
153	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
154	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
155	Fibronectin is a serum biomarker for <scp>D</scp>uchenne muscular dystrophy. Proteomics - Clinical Applications, 2014, 8, 269-278.	1.6	73
156	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
157	Improving recognition of Duchenne muscular dystrophy: a retrospective case note review. Archives of Disease in Childhood, 2014, 99, 1074-1077.	1.9	75
158	Psycho-organic symptoms as early manifestation of adult onset POMT1-related limb girdle muscular dystrophy. Neuromuscular Disorders, 2014, 24, 990-992.	0.6	8
159	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
160	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Journal of Neuromuscular Diseases, 2014, 1, 75-90.	2.6	69
161	Quantitative Magnetic Resonance Imaging in Limb-Girdle Muscular Dystrophy 2I: A Multinational Cross-Sectional Study. PLoS ONE, 2014, 9, e90377.	2.5	81
162	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

#	Article	IF	Citations
163	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
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