

Volker Straub

List of Publications by Year in descending order

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Version: 2024-02-01

197
papers

11,850
citations

23500

58
h-index

33814

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. <i>Nucleic Acids Research</i> , 2017, 45, D865-D876.	6.5	699
2	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. <i>Science Translational Medicine</i> , 2017, 9, .	5.8	516
3	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. <i>Human Mutation</i> , 2015, 36, 395-402.	1.1	507
4	Animal Models for Muscular Dystrophy Show Different Patterns of Sarcolemmal Disruption. <i>Journal of Cell Biology</i> , 1997, 139, 375-385.	2.3	441
5	Prevalence of genetic muscle disease in Northern England: in-depth analysis of a muscle clinic population. <i>Brain</i> , 2009, 132, 3175-3186.	3.7	414
6	Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. <i>Brain</i> , 2007, 130, 2725-2735.	3.7	385
7	Safety and efficacy of drisapersen for the treatment of Duchenne muscular dystrophy (DEMAND II): an exploratory, randomised, placebo-controlled phase 2 study. <i>Lancet Neurology</i> , The, 2014, 13, 987-996.	4.9	279
8	Managing Duchenne muscular dystrophy – The additive effect of spinal surgery and home nocturnal ventilation in improving survival. <i>Neuromuscular Disorders</i> , 2007, 17, 470-475.	0.3	273
9	A founder mutation in Anoctamin 5 is a major cause of limb girdle muscular dystrophy. <i>Brain</i> , 2011, 134, 171-182.	3.7	254
10	Mutations in the FKRP gene can cause muscle-eye-brain disease and Walker-Warburg syndrome. <i>Journal of Medical Genetics</i> , 2004, 41, e61-e61.	1.5	243
11	Phenotypic spectrum associated with mutations in the fukutin-related protein gene. <i>Annals of Neurology</i> , 2003, 53, 537-542.	2.8	219
12	Long-term benefits and adverse effects of intermittent versus daily glucocorticoids in boys with Duchenne muscular dystrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 698-705.	0.9	201
13	Secondary calpain3 deficiency in 2q-linked muscular dystrophy. <i>Neurology</i> , 2001, 56, 869-877.	1.5	163
14	Genotype–phenotype correlation in a large population of muscular dystrophy patients with LAMA2 mutations. <i>Neuromuscular Disorders</i> , 2010, 20, 241-250.	0.3	154
15	The burden of Duchenne muscular dystrophy. <i>Neurology</i> , 2014, 83, 529-536.	1.5	149
16	Quantitative Muscle MRI as an Assessment Tool for Monitoring Disease Progression in LGMD2I: A Multicentre Longitudinal Study. <i>PLoS ONE</i> , 2013, 8, e70993.	1.1	148
17	Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. <i>Human Mutation</i> , 2012, 33, 981-988.	1.1	145
18	Development of Exon Skipping Therapies for Duchenne Muscular Dystrophy: A Critical Review and a Perspective on the Outstanding Issues. <i>Nucleic Acid Therapeutics</i> , 2017, 27, 251-259.	2.0	144

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19	Serum matrix metalloproteinase-9 (MMP-9) as a biomarker for monitoring disease progression in Duchenne muscular dystrophy (DMD). <i>Neuromuscular Disorders</i> , 2011, 21, 569-578.	0.3	132
20	Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. <i>Journal of Neuromuscular Diseases</i> , 2017, 4, 293-306.	1.1	125
21	Affinity proteomics within rare diseases: a <sc>BIO</sc>â€<sc>NMD</sc> study for blood biomarkers of muscular dystrophies. <i>EMBO Molecular Medicine</i> , 2014, 6, 918-936.	3.3	105
22	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. <i>Nature Medicine</i> , 2021, 27, 1197-1204.	15.2	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. <i>Neuromuscular Disorders</i> , 2012, 22, S54-S67.	0.3	94
24	The TREAT-NMD Duchenne Muscular Dystrophy Registries: Conception, Design, and Utilization by Industry and Academia. <i>Human Mutation</i> , 2013, 34, 1449-1457.	1.1	94
25	TREAT-NMD workshop: Pattern recognition in genetic muscle diseases using muscle MRI. <i>Neuromuscular Disorders</i> , 2012, 22, S42-S53.	0.3	93
26	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. <i>Annals of Neurology</i> , 2018, 83, 1105-1124.	2.8	93
27	Inheritance patterns and phenotypic features of myofibrillar myopathy associated with a BAG3 mutation. <i>Neuromuscular Disorders</i> , 2010, 20, 438-442.	0.3	90
28	Muscle Magnetic Resonance Imaging in Congenital Myopathies Due to Ryanodine Receptor Type 1 Gene Mutations. <i>Archives of Neurology</i> , 2011, 68, 1171.	4.9	89
29	A heterozygous 21-bp deletion in<i>CAPN3</i> causes dominantly inherited limb girdle muscular dystrophy. <i>Brain</i> , 2016, 139, 2154-2163.	3.7	87
30	Validation of genetic modifiers for Duchenne muscular dystrophy: a multicentre study assessing<i>SPP1</i> and<i>LTBP4</i> variants. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 1060-1065.	0.9	86
31	Healthâ€related quality of life in patients with Duchenne muscular dystrophy: a multinational, crossâ€sectional study. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 508-515.	1.1	82
32	Quantifying the burden of caregiving in Duchenne muscular dystrophy. <i>Journal of Neurology</i> , 2016, 263, 906-915.	1.8	82
33	Limb-girdle muscular dystrophies â€ international collaborations for translational research. <i>Nature Reviews Neurology</i> , 2016, 12, 294-309.	4.9	81
34	Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 1071-1081.	0.9	81
35	Quantitative Magnetic Resonance Imaging in Limb-Girdle Muscular Dystrophy 2I: A Multinational Cross-Sectional Study. <i>PLoS ONE</i> , 2014, 9, e90377.	1.1	81
36	New aspects on patients affected by dysferlin deficient muscular dystrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, 946-953.	0.9	79

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37	Muscle MRI findings in limb girdle muscular dystrophy type 2L. <i>Neuromuscular Disorders</i> , 2012, 22, S122-S129.	0.3	77
38	Stakeholder cooperation to overcome challenges in orphan medicine development: the example of Duchenne muscular dystrophy. <i>Lancet Neurology</i> , The, 2016, 15, 882-890.	4.9	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. <i>Journal of Neurology</i> , 2014, 261, 152-163.	1.8	76
40	Improving recognition of Duchenne muscular dystrophy: a retrospective case note review. <i>Archives of Disease in Childhood</i> , 2014, 99, 1074-1077.	1.0	75
41	The Clinical Outcome Study for dysferlinopathy. <i>Neurology: Genetics</i> , 2016, 2, e89.	0.9	75
42	Congenital muscular dystrophies in the UK population: Clinical and molecular spectrum of a large cohort diagnosed over a 12-year period. <i>Neuromuscular Disorders</i> , 2017, 27, 793-803.	0.3	75
43	Dystrophin quantification. <i>Neurology</i> , 2014, 83, 2062-2069.	1.5	73
44	Fibronectin is a serum biomarker for Duchenne muscular dystrophy. <i>Proteomics - Clinical Applications</i> , 2014, 8, 269-278.	0.8	73
45	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. <i>Journal of Neurology</i> , 2012, 259, 838-850.	1.8	72
46	The Classification, Natural History and Treatment of the Limb Girdle Muscular Dystrophies. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, S7-S19.	1.1	72
47	Biochemical Characterization of Patients With In-Frame or Out-of-Frame DMD Deletions Pertinent to Exon 44 or 45 Skipping. <i>JAMA Neurology</i> , 2014, 71, 32.	4.5	71
48	Titin founder mutation is a common cause of myofibrillar myopathy with early respiratory failure. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 331-338.	0.9	71
49	Muscle-Derived Proteins as Serum Biomarkers for Monitoring Disease Progression in Three Forms of Muscular Dystrophy. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 241-255.	1.1	71
50	Association Study of Exon Variants in the NF- κ B and TGF β Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2016, 99, 1163-1171.	2.6	71
51	Magnetic resonance imaging in duchenne muscular dystrophy: Longitudinal assessment of natural history over 18 months. <i>Muscle and Nerve</i> , 2013, 48, 586-588.	1.0	70
52	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. <i>Journal of Neuromuscular Diseases</i> , 2014, 1, 75-90.	1.1	69
53	Mutations in INPP5K, Encoding a Phosphoinositide 5-Phosphatase, Cause Congenital Muscular Dystrophy with Cataracts and Mild Cognitive Impairment. <i>American Journal of Human Genetics</i> , 2017, 100, 523-536.	2.6	67
54	Why are some patients with Duchenne muscular dystrophy dying young: An analysis of causes of death in North East England. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 904-909.	0.7	66

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55	Congenital muscular dystrophy with laminin Î±2 chain deficiency: Identification of a new intermediate phenotype and correlation of clinical findings to muscle immunohistochemistry. <i>European Journal of Pediatrics</i> , 1996, 155, 968-976.	1.3	65
56	Contrast agent-enhanced magnetic resonance imaging of skeletal muscle damage in animal models of muscular dystrophy. <i>Magnetic Resonance in Medicine</i> , 2000, 44, 655-659.	1.9	65
57	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. <i>Genetics in Medicine</i> , 2020, 22, 1478-1488.	1.1	62
58	Prevalence of Pompe disease in 3,076 patients with hyperCKemia and limb-girdle muscular weakness. <i>Neurology</i> , 2016, 87, 295-298.	1.5	60
59	MR imaging in Duchenne muscular dystrophy: Quantification of T ₁ -weighted signal, contrast uptake, and the effects of exercise. <i>Journal of Magnetic Resonance Imaging</i> , 2009, 30, 1130-1138.	1.9	59
60	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. <i>Neuromuscular Disorders</i> , 2019, 29, 167-186.	0.3	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. <i>Lancet Neurology</i> , The, 2021, 20, 1012-1026.	4.9	59
62	Short stature and pubertal delay in Duchenne muscular dystrophy. <i>Archives of Disease in Childhood</i> , 2016, 101, 101-106.	1.0	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. <i>Nucleic Acid Therapeutics</i> , 2022, 32, 29-39.	2.0	58
64	Late onset in dysferlinopathy widens the clinical spectrum. <i>Neuromuscular Disorders</i> , 2008, 18, 288-290.	0.3	57
65	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. <i>Contemporary Clinical Trials</i> , 2017, 58, 34-39.	0.8	56
66	Fractures and Linear Growth in a Nationwide Cohort of Boys With Duchenne Muscular Dystrophy With and Without Glucocorticoid Treatment. <i>JAMA Neurology</i> , 2019, 76, 701.	4.5	56
67	MRI in sarcoglycanopathies: a large international cohort study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 72-77.	0.9	55
68	Compliance to Care Guidelines for Duchenne Muscular Dystrophy. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 63-72.	1.1	51
69	Reduced serum myostatin concentrations associated with genetic muscle disease progression. <i>Journal of Neurology</i> , 2017, 264, 541-553.	1.8	51
70	Prospective exploratory muscle biopsy, imaging, and functional assessment in patients with late-onset Pompe disease treated with alglucosidase alfa: The EMBASSY Study. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 115-123.	0.5	49
71	Tracking disease progression noninvasively in Duchenne and Becker muscular dystrophies. <i>Journal of Cachexia, Sarcopenia and Muscle</i> , 2018, 9, 715-726.	2.9	47
72	MYO-MRI diagnostic protocols in genetic myopathies. <i>Neuromuscular Disorders</i> , 2019, 29, 827-841.	0.3	46

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73	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. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 1-30.	1.1	46
74	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. <i>Brain</i> , 2020, 143, 2696-2708.	3.7	45
75	Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. <i>Neurology</i> , 2020, 94, e1094-e1102.	1.5	45
76	Exome sequences versus sequential gene testing in the UK highly specialised Service for Limb Girdle Muscular Dystrophy. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 151.	1.2	44
77	Development and Application of an Ultrasensitive Hybridization-Based ELISA Method for the Determination of Peptide-Conjugated Phosphorodiamidate Morpholino Oligonucleotides. <i>Nucleic Acid Therapeutics</i> , 2015, 25, 275-284.	2.0	43
78	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). <i>Journal of Neurology</i> , 2020, 267, 45-56.	1.8	43
79	Effect of Different Corticosteroid Dosing Regimens on Clinical Outcomes in Boys With Duchenne Muscular Dystrophy. <i>JAMA - Journal of the American Medical Association</i> , 2022, 327, 1456.	3.8	43
80	The Diagnostic Value of MRI Pattern Recognition in Distal Myopathies. <i>Frontiers in Neurology</i> , 2018, 9, 456.	1.1	42
81	Interventions for muscular dystrophy: molecular medicines entering the clinic. <i>Lancet</i> , The, 2009, 374, 1849-1856.	6.3	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. <i>Skeletal Muscle</i> , 2018, 8, 23.	1.9	40
83	Complex phenotypes associated with STIM1 mutations in both coiled coil and EF-hand domains. <i>Neuromuscular Disorders</i> , 2017, 27, 861-872.	0.3	39
84	Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 576-585.	0.9	38
85	Respiratory and upper limb function as outcome measures in ambulant and non-ambulant subjects with Duchenne muscular dystrophy: A prospective multicentre study. <i>Neuromuscular Disorders</i> , 2019, 29, 261-268.	0.3	36
86	Albumin targeting of damaged muscle fibres in the mdx mouse can be monitored by MRI. <i>Neuromuscular Disorders</i> , 2004, 14, 791-796.	0.3	35
87	Novel mutations in DNAJB6 gene cause a very severe early-onset limb-girdle muscular dystrophy 1D disease. <i>Neuromuscular Disorders</i> , 2015, 25, 835-842.	0.3	35
88	Where do we stand in trial readiness for autosomal recessive limb girdle muscular dystrophies?. <i>Neuromuscular Disorders</i> , 2016, 26, 111-125.	0.3	31
89	MSTO1 mutations cause mtDNA depletion, manifesting as muscular dystrophy with cerebellar involvement. <i>Acta Neuropathologica</i> , 2019, 138, 1013-1031.	3.9	31
90	Two recurrent mutations are associated with GNE myopathy in the North of Britain. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 1359-1365.	0.9	30

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91	Prophylactic oral bisphosphonate therapy in duchenne muscular dystrophy. <i>Muscle and Nerve</i> , 2016, 54, 79-85.	1.0	30
92	Respiratory involvement in ambulant and non-ambulant patients with facioscapulohumeral muscular dystrophy. <i>Journal of Neurology</i> , 2017, 264, 1271-1280.	1.8	30
93	The clinical-phenotype continuum in DYNC1H1-related disordersâ€™ genomic profiling and proposal for a novel classification. <i>Journal of Human Genetics</i> , 2020, 65, 1003-1017.	1.1	30
94	POGLUT1 biallelic mutations cause myopathy with reduced satellite cells, ð±-dystroglycan hypoglycosylation and a distinctive radiological pattern. <i>Acta Neuropathologica</i> , 2020, 139, 565-582.	3.9	29
95	Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. <i>Brain</i> , 2017, 140, 37-48.	3.7	28
96	Natural history of limb girdle muscular dystrophy R9 over 6Âyears: searching for trial endpoints. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1033-1045.	1.7	28
97	Recurrent <i>TTN</i> metatranscriptâ€™only c.39974â€™11T>G splice variant associated with autosomal recessive arthrogyposis multiplex congenita and myopathy. <i>Human Mutation</i> , 2020, 41, 403-411.	1.1	28
98	Phenotypic variability of TRPV4 related neuropathies. <i>Neuromuscular Disorders</i> , 2015, 25, 516-521.	0.3	27
99	Clinical and neuroimaging findings in two brothers with limb girdle muscular dystrophy due to LAMA2 mutations. <i>Neuromuscular Disorders</i> , 2017, 27, 170-174.	0.3	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. <i>Journal of Medical Genetics</i> , 2019, 56, 693-700.	1.5	27
101	<i>POPDC3</i> Gene Variants Associate with a New Form of Limb Girdle Muscular Dystrophy. <i>Annals of Neurology</i> , 2019, 86, 832-843.	2.8	27
102	Longitudinal serum biomarker screening identifies malate dehydrogenase 2 as candidate prognostic biomarker for Duchenne muscular dystrophy. <i>Journal of Cachexia, Sarcopenia and Muscle</i> , 2020, 11, 505-517.	2.9	27
103	The TREAT-NMD care and trial site registry: an online registry to facilitate clinical research for neuromuscular diseases. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 171.	1.2	26
104	Elusive sources of variability of dystrophin rescue by exon skipping. <i>Skeletal Muscle</i> , 2015, 5, 44.	1.9	26
105	A checklist for clinical trials in rare disease: obstacles and anticipatory actionsâ€™lessons learned from the FOR-DMD trial. <i>Trials</i> , 2018, 19, 291.	0.7	26
106	Muscular dystrophy with arrhythmia caused by loss-of-function mutations in <i>BVES</i> . <i>Neurology: Genetics</i> , 2019, 5, e321.	0.9	26
107	Mutational spectrum and phenotypic variability of VCP-related neurological disease in the UK. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 680-681.	0.9	25
108	Re-evaluation of the phenotype caused by the common <i>MATR3</i> p.Ser85Cys mutation in a new family. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 448-450.	0.9	24

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109	Economic Evaluation in Duchenne Muscular Dystrophy: Model Frameworks for Cost-Effectiveness Analysis. <i>Pharmacoeconomics</i> , 2017, 35, 249-258.	1.7	24
110	FSHD type 2 and Bosma arhinia microphthalmia syndrome. <i>Neurology</i> , 2018, 91, e562-e570.	1.5	24
111	The UK Myotonic Dystrophy Patient Registry: facilitating and accelerating clinical research. <i>Journal of Neurology</i> , 2017, 264, 979-988.	1.8	23
112	<sc><i>GGPS1</i></sc> Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. <i>Annals of Neurology</i> , 2020, 88, 332-347.	2.8	22
113	The Latin American experience with a next generation sequencing genetic panel for recessive limb-girdle muscular weakness and Pompe disease. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 11.	1.2	22
114	The TREAT-NMD advisory committee for therapeutics (TACT): an innovative de-risking model to foster orphan drug development. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 49.	1.2	21
115	Identification of GAA variants through whole exome sequencing targeted to a cohort of 606 patients with unexplained limb-girdle muscle weakness. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 173.	1.2	21
116	Limb girdle muscular dystrophy due to mutations in <i>POMT2</i>. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 506-512.	0.9	21
117	Conserved expression of truncated telethonin in a patient with limb-girdle muscular dystrophy 2G. <i>Neuromuscular Disorders</i> , 2015, 25, 349-352.	0.3	20
118	Assessment of disease progression in dysferlinopathy. <i>Neurology</i> , 2019, 92, .	1.5	20
119	Global FKRP Registry: observations in more than 300 patients with Limb Girdle Muscular Dystrophy R9. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 757-766.	1.7	20
120	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. <i>Nature Communications</i> , 2022, 13, 2306.	5.8	20
121	Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 1224-1226.	0.9	19
122	Subepicardial dysfunction leads to global left ventricular systolic impairment in patients with limb girdle muscular dystrophy 2I. <i>European Journal of Heart Failure</i> , 2013, 15, 986-994.	2.9	18
123	A novel recessive TTN founder variant is a common cause of distal myopathy in the Serbian population. <i>European Journal of Human Genetics</i> , 2017, 25, 572-581.	1.4	18
124	A "second truncation"™ in TTN causes early onset recessive muscular dystrophy. <i>Neuromuscular Disorders</i> , 2017, 27, 1009-1017.	0.3	18
125	Improving translatability of preclinical studies for neuromuscular disorders: lessons from the TREAT-NMD Advisory Committee for Therapeutics (TACT). <i>DMM Disease Models and Mechanisms</i> , 2020, 13, .	1.2	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. <i>Journal of Clinical Lipidology</i> , 2020, 14, 459-469.e0.	0.6	18

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127	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 153-166.	1.1	18
128	Skeletal muscle magnetic resonance imaging in <scp>Pompe</scp> disease. <i>Muscle and Nerve</i> , 2021, 63, 640-650.	1.0	18
129	Miyoshi myopathy and limb girdle muscular dystrophy R2 are the same disease. <i>Neuromuscular Disorders</i> , 2021, 31, 265-280.	0.3	18
130	Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. <i>Annals of Neurology</i> , 2021, 89, 967-978.	2.8	17
131	Deep RNA profiling identified clock and molecular clock genes as pathophysiological signatures in collagen VI myopathy. <i>Journal of Cell Science</i> , 2016, 129, 1671-84.	1.2	16
132	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. <i>European Journal of Neurology</i> , 2020, 27, 2604-2615.	1.7	16
133	Long-term Safety and Efficacy of Avalglucosidase Alfa in Patients With Late-Onset Pompe Disease. <i>Neurology</i> , 2022, 99, .	1.5	16
134	Mobility shift of beta-dystroglycan as a marker of <i>GMPPB</i> gene-related muscular dystrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 762-768.	0.9	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. <i>Neuromuscular Disorders</i> , 2018, 28, 633-638.	0.3	15
136	Multisystem proteinopathy due to a homozygous p.Arg159His <i>VCP</i> mutation. <i>Neurology</i> , 2020, 94, e785-e796.	1.5	15
137	Facioscapulohumeral muscular dystrophy 1 patients participating in the UK FSHD registry can be subdivided into 4 patterns of self-reported symptoms. <i>Neuromuscular Disorders</i> , 2020, 30, 315-328.	0.3	15
138	A form of muscular dystrophy associated with pathogenic variants in JAG2. <i>American Journal of Human Genetics</i> , 2021, 108, 840-856.	2.6	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. <i>PLOS Currents</i> , 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. <i>Neuromuscular Disorders</i> , 2018, 28, 48-53.	0.3	13
141	Normalized grip strength is a sensitive outcome measure through all stages of Duchenne muscular dystrophy. <i>Journal of Neurology</i> , 2020, 267, 2022-2028.	1.8	13
142	Cardiac involvement in hereditary myopathy with early respiratory failure. <i>Neurology</i> , 2016, 87, 1031-1035.	1.5	12
143	The effects of ageing on mouse muscle microstructure: a comparative study of timeâ€dependent diffusion MRI and histological assessment. <i>NMR in Biomedicine</i> , 2018, 31, e3881.	1.6	12
144	Noninvasive quantification of fibrosis in skeletal and cardiac muscle in mdx mice using EP3533 enhanced magnetic resonance imaging. <i>Magnetic Resonance in Medicine</i> , 2019, 81, 2728-2735.	1.9	12

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