Hanns LochmÃ¼ller

List of Publications by Year
in descending order

470 papers	25,030 citations	6254 80 h-index	15266 126 g-index
485 all docs	485 docs citations	485 times ranked	24955 citing authors

#	Article	IF	Citations
1	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
2	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	14.5	539
3	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. Human Mutation, 2015, 36, 395-402.	2.5	507
4	Phenotypic spectrum associated with mutations of the mitochondrial polymerase Â gene. Brain, 2006, 129, 1674-1684.	7.6	397
5	Prevalence, incidence and carrier frequency of 5qâ€“linked spinal muscular atrophy â€“ a literature review. Orphanet Journal of Rare Diseases, 2017, 12, 124.	2.7	391
6	Mutations in dynamin 2 cause dominant centronuclear myopathy. Nature Genetics, 2005, 37, 1207-1209.	21.4	390
7	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	6.2	305
8	The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene. Brain, 2007, 130, 2037-2044.	7.6	298
9	Mildly affected patients with spinal muscular atrophy are partially protected by an increased SMN2 copy number. Human Genetics, 2006, 119, 422-428.	3.8	292
10	A Mutation in the Dimerization Domain of Filamin C Causes a Novel Type of Autosomal Dominant Myofibrillar Myopathy. American Journal of Human Genetics, 2005, 77, 297-304.	6.2	268
11	Mutation in TACO1, encoding a translational activator of COX I, results in cytochrome c oxidase deficiency and late-onset Leigh syndrome. Nature Genetics, 2009, 41, 833-837.	21.4	260
12	A founder mutation in Anoctamin 5 is a major cause of limb girdle muscular dystrophy. Brain, 2011, 134, 171-182.	7.6	254
13	Dok-7 Mutations Underlie a Neuromuscular Junction Synaptopathy. Science, 2006, 313, 1975-1978.	12.6	247
14	SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. Nature Genetics, 2009, 41, 654-656.	21.4	233
15	Mutations in SIL1 cause Marinesco-SjÃ¶gren syndrome, a cerebellar ataxia with cataract and myopathy. Nature Genetics, 2005, 37, 1312-1314.	21.4	232
16	Alterations in the ankyrin domain of TRPV4 cause congenital distal SMA, scapuloperoneal SMA and HMSN2C. Nature Genetics, 2010, 42, 160-164.	21.4	228
17	An agrin minigene rescues dystrophic symptoms in a mouse model for congenital muscular dystrophy. Nature, 2001, 413, 302-307.	27.8	222
18	Late onset Pompe disease: Clinical and neurophysiological spectrum of 38 patients including long-term follow-up in 18 patients. Neuromuscular Disorders, 2007, 17, 698-706.	0.6	208

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19	Risk of developing a mitochondrial DNA deletion disorder. Lancet, The, 2004, 364, 592-596.	13.7	201
20	Mutations and polymorphisms of the skeletal muscle ÃŽÂ±-actin gene (ACTA1). Human Mutation, 2009, 30, 1267-1277.	2.5	198
21	Autosomal-Dominant Distal Myopathy Associated with a Recurrent Missense Mutation in the Gene Encoding the Nuclear Matrix Protein, Matrin 3. American Journal of Human Genetics, 2009, 84, 511-518.	6.2	161
22	RD-Connect: An Integrated Platform Connecting Databases, Registries, Biobanks and Clinical Bioinformatics for Rare Disease Research. Journal of General Internal Medicine, 2014, 29, 780-787.	2.6	159
23	Dystrophin Expression in Muscles of mdx Mice After Adenovirus-MediatedIn VivoGene Transfer. Human Gene Therapy, 1996, 7, 129-140.	2.7	158
24	Future of Rare Diseases Research 2017â€“2027: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 21-27.	3.1	154
25	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. American Journal of Human Genetics, 2011, 88, 162-172.	6.2	153
26	The burden of Duchenne muscular dystrophy. Neurology, 2014, 83, 529-536.	1.1	149
27	Mutation History of the Roma/Gypsies. American Journal of Human Genetics, 2004, 75, 596-609.	6.2	148
28	Pathological consequences of VCP mutations on human striated muscle. Brain, 2007, 130, 381-393.	7.6	148
29	An X-Linked Myopathy with Postural Muscle Atrophy and Generalized Hypertrophy, Termed XMPMA, Is Caused by Mutations in FHL1. American Journal of Human Genetics, 2008, 82, 88-99.	6.2	148
30	Quantitative Muscle MRI as an Assessment Tool for Monitoring Disease Progression in LGMD2I: A Multicentre Longitudinal Study. PLoS ONE, 2013, 8, e70993.	2.5	148
31	Escobar Syndrome Is a Prenatal Myasthenia Caused by Disruption of the Acetylcholine Receptor Fetal Î³ Subunit. American Journal of Human Genetics, 2006, 79, 303-312.	6.2	146
32	Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. Human Mutation, 2012, 33, 981-988.	2.5	145
33	Phenotypical spectrum of DOK7 mutations in congenital myasthenic syndromes. Brain, 2007, 130, 1497-1506.	7.6	143
34	Mitochondrial Phosphateâ€“Carrier Deficiency: A Novel Disorder of Oxidative Phosphorylation. American Journal of Human Genetics, 2007, 80, 478-484.	6.2	142
35	Infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). Annals of Neurology, 2003, 54, 719-724.	5.3	141
36	Diagnostic value of muscle MRI in differentiating LGMD2I from other LGMDs. Journal of Neurology, 2005, 252, 538-547.	3.6	136

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37	The ubiquitin-selective chaperone CDC-48/p97 links myosin assembly to human myopathy. Nature Cell Biology, 2007, 9, 379-390.	10.3	135
38	Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. Brain, 2008, 131, 747-759.	7.6	134
39	Dystrophin-deficient pigs provide new insights into the hierarchy of physiological derangements of dystrophic muscle. Human Molecular Genetics, 2013, 22, 4368-4382.	2.9	134
40	Clinical and morphological phenotype of the filamin myopathy: a study of 31 German patients. Brain, 2007, 130, 3250-3264.	7.6	132
41	Serum matrix metalloproteinase-9 (MMP-9) as a biomarker for monitoring disease progression in Duchenne muscular dystrophy (DMD). Neuromuscular Disorders, 2011, 21, 569-578.	0.6	132
42	Safety and Treatment Effects of Nusinersen in Longstanding Adult 5q-SMA Type 3 â€“ A Prospective Observational Study. Journal of Neuromuscular Diseases, 2019, 6, 453-465.	2.6	132
43	Life expectancy at birth in Duchenne muscular dystrophy: a systematic review and meta-analysis. European Journal of Epidemiology, 2020, 35, 643-653.	5.7	132
44	Partial deficiency of the C-terminal-domain phosphatase of RNA polymerase II is associated with congenital cataracts facial dysmorphism neuropathy syndrome. Nature Genetics, 2003, 35, 185-189.	21.4	129
45	Targeted Next-Generation Sequencing of a 12.5 Mb Homozygous Region Reveals ANO10 Mutations in Patients with Autosomal-Recessive Cerebellar Ataxia. American Journal of Human Genetics, 2010, 87, 813-819.	6.2	125
46	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
47	Sympathetic innervation controls homeostasis of neuromuscular junctions in health and disease. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 746-750.	7.1	123
48	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. Nature Communications, 2014, 5, 4287.	12.8	120
49	Congenital myasthenic syndromes due to mutations inALG2andALG14. Brain, 2013, 136, 944-956.	7.6	117
50	Recommendations for Improving the Quality of Rare Disease Registries. International Journal of Environmental Research and Public Health, 2018, 15, 1644.	2.6	116
51	Factors Influencing the Efficacy, Longevity, and Safety of Electroporation-Assisted Plasmid-Based Gene Transfer into Mouse Muscles. Molecular Therapy, 2004, 10, 447-455.	8.2	115
52	Muscle fibres and cultured muscle cells express the B7.1/2-related inducible co-stimulatory molecule, ICOSL: implications for the pathogenesis of inflammatory myopathies. Brain, 2003, 126, 1026-1035.	7.6	112
53	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. Brain, 2009, 132, 3165-3174.	7.6	112
54	International Charter of principles for sharing bio-specimens and data. European Journal of Human Genetics, 2015, 23, 721-728.	2.8	112

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55	Mutations inGMPPBcause congenital myasthenic syndrome and bridge myasthenic disorders with dystroglycanopathies. Brain, 2015, 138, 2493-2504.	7.6	111
56	Missense mutations of ACTA1 cause dominant congenital myopathy with cores. Journal of Medical Genetics, 2004, 41, 842-848.	3.2	110
57	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
58	Limbâ€“girdle muscular dystrophies. Current Opinion in Neurology, 2008, 21, 576-584.	3.6	104
59	Progress in Rare Diseases Research 2010â€“2016: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 11-20.	3.1	104
60	EFNS guideline on diagnosis and management of limb girdle muscular dystrophies. European Journal of Neurology, 2007, 14, 1305-1312.	3.3	103
61	Phenotypes of the N88S Berardinelli-Seip congenital lipodystrophy 2 mutation. Annals of Neurology, 2005, 57, 415-424.	5.3	99
62	Adult-onset cerebellar ataxia due to mutations inCABC1/ADCK3. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 174-178.	1.9	99
63	Congenital myasthenic syndromes: Achievements and limitations of phenotype-guided gene-after-gene sequencing in diagnostic practice: A study of 680 patients. Human Mutation, 2012, 33, 1474-1484.	2.5	99
64	A multi-source approach to determine SMA incidence and research ready population. Journal of Neurology, 2017, 264, 1465-1473.	3.6	98
65	Commonality ofTRIM32mutation in causing sarcotubular myopathy and LGMD2H. Annals of Neurology, 2005, 57, 591-595.	5.3	96
66	Synaptotagmin 2 Mutations Cause an Autosomal-Dominant Form of Lambert-Eaton Myasthenic Syndrome and Nonprogressive Motor Neuropathy. American Journal of Human Genetics, 2014, 95, 332-339.	6.2	96
67	Human muscle cells express a B7â€related molecule, B7â€H1, with strong negative immune regulatory potential: a novel mechanism of counterbalancing the immune attack in idiopathic inflammatory myopathies. FASEB Journal, 2003, 17, 1-16.	0.5	95
68	Cognitive behavioural therapy with optional graded exercise therapy in patients with severe fatigue with myotonic dystrophy type 1: a multicentre, single-blind, randomised trial. Lancet Neurology, The, 2018, 17, 671-680.	10.2	95
69	Attenuated muscle regeneration is a key factor in dysferlin-deficient muscular dystrophy. Human Molecular Genetics, 2009, 18, 1976-1989.	2.9	94
70	The TREAT-NMD Duchenne Muscular Dystrophy Registries: Conception, Design, and Utilization by Industry and Academia. Human Mutation, 2013, 34, 1449-1457.	2.5	94
71	No overall hyposialylation in hereditary inclusion body myopathy myoblasts carrying the homozygous M712T GNE mutation. Biochemical and Biophysical Research Communications, 2005, 328, 221-226.	2.1	93
72	Scapuloperoneal syndrome type Kaeser and a wide phenotypic spectrum of adult-onset, dominant myopathies are associated with the desmin mutation R350P. Brain, 2007, 130, 1485-1496.	7.6	92

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73	Expression of the E6 and E7 Genes of Human Papillomavirus (HPV16) Extends the Life Span of Human Myoblasts. Experimental Cell Research, 1999, 248, 186-193.	2.6	91
74	Inheritance patterns and phenotypic features of myofibrillar myopathy associated with a BAG3 mutation. Neuromuscular Disorders, 2010, 20, 438-442.	0.6	90
75	Reversible molecular pathology of skeletal muscle in spinal muscular atrophy. Human Molecular Genetics, 2011, 20, 4334-4344.	2.9	89
76	Dysferlin associates with the developing Tâ€tubule system in rodent and human skeletal muscle. Muscle and Nerve, 2010, 41, 166-173.	2.2	87
77	The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. Journal of Neurology, 2011, 258, 1987-1997.	3.6	87
78	A heterozygous 21-bp deletion inCAPN3causes dominantly inherited limb girdle muscular dystrophy. Brain, 2016, 139, 2154-2163.	7.6	87
79	Agrin mutations lead to a congenital myasthenic syndrome with distal muscle weakness and atrophy. Brain, 2014, 137, 2429-2443.	7.6	86
80	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
81	Muscle pathology in 57 patients with myotonic dystrophy type 2. Muscle and Nerve, 2004, 29, 275-281.	2.2	82
82	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
83	Quantifying the burden of caregiving in Duchenne muscular dystrophy. Journal of Neurology, 2016, 263, 906-915.	3.6	82
84	The principles of gene therapy for the nervous system. Trends in Neurosciences, 1996, 19, 49-54.	8.6	81
85	Expression of tollâ€like receptors by human muscle cells in vitro and in vivo: TLR3 is highly expressed in inflammatory and HIV myopathies, mediates ILâ€8 release, and upâ€regulation of NKG2Dâ€ligands. FASEB Journal, 2006, 20, 118-120.	0.5	81
86	The p.G154S mutation of the alpha-B crystallin gene (CRYAB) causes late-onset distal myopathy. Neuromuscular Disorders, 2010, 20, 255-259.	0.6	81
87	Genetic heterogeneity of motor neuropathies. Neurology, 2017, 88, 1226-1234.	1.1	81
88	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
89	Quantitative Magnetic Resonance Imaging in Limb-Girdle Muscular Dystrophy 2I: A Multinational Cross-Sectional Study. PLoS ONE, 2014, 9, e90377.	2.5	81
90	The non-classical MHC molecule HLA-G protects human muscle cells from immune-mediated lysis: implications for myoblast transplantation and gene therapy. Brain, 2003, 126, 176-185.	7.6	80

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91	New aspects on patients affected by dysferlin deficient muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 946-953.	1.9	79
92	Mutations in the collagen XII gene define a new form of extracellular matrix-related myopathy. Human Molecular Genetics, 2014, 23, 2353-2363.	2.9	79
93	Recessive Mutations in the Î±3 (VI) Collagen Gene COL6A3 Cause Early-Onset Isolated Dystonia. American Journal of Human Genetics, 2015, 96, 883-893.	6.2	79
94	Muscle MRI findings in limb girdle muscular dystrophy type 2L. Neuromuscular Disorders, 2012, 22, S122-S129.	0.6	77
95	Treatment of dysferlinopathy with deflazacort: a double-blind, placebo-controlled clinical trial. Orphanet Journal of Rare Diseases, 2013, 8, 26.	2.7	77
96	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
97	Homozygous mutations incaveolin-3cause a severe form of rippling muscle disease. Annals of Neurology, 2003, 53, 512-520.	5.3	75
98	The Clinical Outcome Study for dysferlinopathy. Neurology: Genetics, 2016, 2, e89.	1.9	75
99	ANO10 mutations cause ataxia and coenzyme Q10 deficiency. Journal of Neurology, 2014, 261, 2192-2198.	3.6	74
100	Mutation in dystrophin-encoding gene affects energy metabolism in mouse myoblasts. Biochemical and Biophysical Research Communications, 2009, 386, 463-466.	2.1	73
101	Fibronectin is a serum biomarker for <scp>D</scp>uchenne muscular dystrophy. Proteomics - Clinical Applications, 2014, 8, 269-278.	1.6	73
102	Differential Short-Term Transduction Efficiency of Adult versus Newborn Mouse Tissues by Adenoviral Recombinants. Experimental and Molecular Pathology, 1995, 62, 131-143.	2.1	72
103	High-Level Dystrophin Expression after Adenovirus-Mediated Dystrophin Minigene Transfer to Skeletal Muscle of Dystrophic Dogs: Prolongation of Expression with Immunosuppression. Human Gene Therapy, 1998, 9, 629-634.	2.7	72
104	Localization of UDP-GlcNAc 2-epimerase/ManAc kinase (GNE) in the Golgi complex and the nucleus of mammalian cells. Experimental Cell Research, 2005, 304, 365-379.	2.6	72
105	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850.	3.6	72
106	Comparative proteomic analyses of Duchenne muscular dystrophy and Becker muscular dystrophy muscles: changes contributing to preserve muscle function in Becker muscular dystrophy patients. Journal of Cachexia, Sarcopenia and Muscle, 2020, 11, 547-563.	7.3	72
107	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
108	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

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109	Association Study of Exon Variants in the NF-Î²B and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	6.2	71
110	Genetic determinants of disease severity in the myotonic dystrophy type 1 OPTIMISTIC cohort. Neurology, 2019, 93, e995-e1009.	1.1	71
111	Developmental defects in a zebrafish model for muscular dystrophies associated with the loss of fukutin-related protein (FKRP). Brain, 2008, 131, 1551-1561.	7.6	70
112	â€˜You should at least askâ€™. The expectations, hopes and fears of rare disease patients on large-scale data and biomaterial sharing for genomics research. European Journal of Human Genetics, 2016, 24, 1403-1408.	2.8	70
113	Gentamicin fails to increase dystrophin expression in dystrophin-deficient muscle. Muscle and Nerve, 2003, 27, 624-627.	2.2	69
114	Parkinson syndrome, neuropathy, and myopathy caused by the mutation A8344G (MERRF) in tRNALys. Neurology, 2007, 68, 56-58.	1.1	69
115	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Journal of Neuromuscular Diseases, 2014, 1, 75-90.	2.6	69
116	Antibody-Mediated Targeting of an Adenovirus Vector Modified To Contain a Synthetic Immunoglobulin G-Binding Domain in the Capsid. Journal of Virology, 2003, 77, 2093-2104.	3.4	68
117	Impaired Presynaptic High-Affinity Choline Transporter Causes a Congenital Myasthenic Syndrome with Episodic Apnea. American Journal of Human Genetics, 2016, 99, 753-761.	6.2	68
118	Linker molecules between laminins and dystroglycan ameliorate laminin-Î±2â€“deficient muscular dystrophy at all disease stages. Journal of Cell Biology, 2007, 176, 979-993.	5.2	67
119	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
120	SMArtCAREÂ-ÂA platform to collect real-life outcome data of patients with spinal muscular atrophy. Orphanet Journal of Rare Diseases, 2019, 14, 18.	2.7	67
121	Screening for Carnitine Palmitoyltransferase II Deficiency by Tandem Mass Spectrometry. Journal of Inherited Metabolic Disease, 2002, 25, 17-27.	3.6	66
122	Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. Brain, 2011, 134, 183-195.	7.6	66
123	Human Skeletal Muscleâ€“derived CD133+ Cells Form Functional Satellite Cells After Intramuscular Transplantation in Immunodeficient Host Mice. Molecular Therapy, 2014, 22, 1008-1017.	8.2	66
124	A novel homozygous missense mutation in the GNE gene of a patient with quadriceps-sparing hereditary inclusion body myopathy associated with muscle inflammation. Neuromuscular Disorders, 2003, 13, 830-834.	0.6	65
125	A retrospective clinical study of the treatment of slow-channel congenital myasthenic syndrome. Journal of Neurology, 2012, 259, 474-481.	3.6	65
126	Further evidence for genetic heterogeneity of distal HMN type V, CMT2 with predominant hand involvement and Silver syndrome. Journal of the Neurological Sciences, 2007, 263, 100-106.	0.6	64

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127	5â€² Trans-Splicing Repair of the PLEC1 Gene. Journal of Investigative Dermatology, 2008, 128, 568-574.	0.7	64
128	Acute liver failure with subsequent cirrhosis as the primary manifestation of TRMU mutations. Journal of Inherited Metabolic Disease, 2011, 34, 197-201.	3.6	64
129	ANO5Gene Analysis in a Large Cohort of Patients with Anoctaminopathy: Confirmation of Male Prevalence and High Occurrence of the Common Exon 5 Gene Mutation. Human Mutation, 2013, 34, 1111-1118.	2.5	64
130	The EuroBioBank Network: 10 years of hands-on experience of collaborative, transnational biobanking for rare diseases. European Journal of Human Genetics, 2015, 23, 1116-1123.	2.8	63
131	The International Rare Diseases Research Consortium: Policies and Guidelines to maximize impact. European Journal of Human Genetics, 2017, 25, 1293-1302.	2.8	62
132	Mutation screening of the N-myc downstream-regulated gene 1 (NDRG1) in patients with Charcot-Marie-Tooth Disease. Human Mutation, 2003, 22, 129-135.	2.5	61
133	Adult-onset glycogen storage disease type 2: clinico-pathological phenotype revisited. Neuropathology and Applied Neurobiology, 2007, 33, 070615152525006-???.	3.2	61
134	Long-term follow-up in patients with congenital myasthenic syndrome due to CHAT mutations. European Journal of Paediatric Neurology, 2010, 14, 326-333.	1.6	61
135	Congenital myasthenic syndromes: spotlight on genetic defects of neuromuscular transmission. Expert Reviews in Molecular Medicine, 2007, 9, 1-20.	3.9	60
136	Novel POMGnT1 mutations define broader phenotypic spectrum of muscleâ€“eyeâ€“brain disease. Neurogenetics, 2007, 8, 279-288.	1.4	60
137	Duchenne muscular dystrophy and caregiver burden: a systematic review. Developmental Medicine and Child Neurology, 2018, 60, 987-996.	2.1	59
138	Characterization of human muscle type cofilin (CFL2) in normal and regenerating muscle. FEBS Journal, 2001, 268, 3473-3482.	0.2	58
139	Creatine monohydrate in myotonic dystrophy. Journal of Neurology, 2002, 249, 1717-1722.	3.6	58
140	Homozygosity for CCTG mutation in myotonic dystrophy type 2. Brain, 2004, 127, 1868-1877.	7.6	58
141	Late onset in dysferlinopathy widens the clinical spectrum. Neuromuscular Disorders, 2008, 18, 288-290.	0.6	57
142	Increased susceptibility to ATP via alteration of P2X receptor function in dystrophic mdx mouse muscle cells. FASEB Journal, 2006, 20, 610-620.	0.5	56
143	Nemaline myopathy caused by mutations in the nebulin gene may present as a distal myopathy. Neuromuscular Disorders, 2011, 21, 556-562.	0.6	56
144	Exon Skipping and Gene Transfer Restore Dystrophin Expression in Human Induced Pluripotent Stem Cells-Cardiomyocytes Harboring DMD Mutations. Stem Cells and Development, 2013, 22, 2714-2724.	2.1	56

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145	High frequency of co-segregating CLCN1 mutations among myotonic dystrophy type 2 patients from Finland and Germany. Journal of Neurology, 2008, 255, 1731-1736.	3.6	55
146	Therapeutic Strategies in Congenital Myasthenic Syndromes. Neurotherapeutics, 2008, 5, 542-547.	4.4	55
147	NFâ€NF<scp>â€Î²</scp>BBâ€dependent expression of the antiapoptotic factor câ€FLIP is regulated by calpain 3, the protein involved in limbâ€girdle muscular dystrophy type 2A. FASEB Journal, 2008, 22, 1521-1529.	0.5	55
148	European Cross-Sectional Survey ofÂCurrent Care Practices for Duchenne Muscular Dystrophy Reveals Regional andÂAge-Dependent Differences. Journal of Neuromuscular Diseases, 2016, 3, 517-527.	2.6	55
149	Clinical features of the myasthenic syndrome arising from mutations in GMPPB. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 802-809.	1.9	55
150	RD-Connect, NeurOmics and EURenOmics: collaborative European initiative for rare diseases. European Journal of Human Genetics, 2018, 26, 778-785.	2.8	55
151	P2X7 purinoceptor alterations in dystrophic mdx mouse muscles: relationship to pathology and potential target for treatment. Journal of Cellular and Molecular Medicine, 2012, 16, 1026-1037.	3.6	53
152	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. Human Molecular Genetics, 2018, 27, 1186-1195.	2.9	52
153	Ephedrine therapy in eight patients with congenital myasthenic syndrome due to DOK7 mutations. Neuromuscular Disorders, 2009, 19, 828-832.	0.6	51
154	Compliance to Care Guidelines for Duchenne Muscular Dystrophy. Journal of Neuromuscular Diseases, 2015, 2, 63-72.	2.6	51
155	Reduced serum myostatin concentrations associated with genetic muscle disease progression. Journal of Neurology, 2017, 264, 541-553.	3.6	51
156	Identification of novel, therapy-responsive protein biomarkers in a mouse model of Duchenne muscular dystrophy by aptamer-based serum proteomics. Scientific Reports, 2015, 5, 17014.	3.3	50
157	Salbutamol-responsive limb-girdle congenital myasthenic syndrome due to a novel missense mutation and heteroallelic deletion in MUSK. Neuromuscular Disorders, 2014, 24, 31-35.	0.6	49
158	Mutations in GFPT1 that underlie limb-girdle congenital myasthenic syndrome result in reduced cell-surface expression of muscle AChR. Human Molecular Genetics, 2013, 22, 2905-2913.	2.9	48
159	Quality of life of patients with spinal muscular atrophy: A systematic review. European Journal of Paediatric Neurology, 2019, 23, 347-356.	1.6	48
160	Electrophysiologic features of SYT2 mutations causing a treatable neuromuscular syndrome. Neurology, 2015, 85, 1964-1971.	1.1	47
161	Improving the informed consent process in international collaborative rare disease research: effective consent for effective research. European Journal of Human Genetics, 2016, 24, 1248-1254.	2.8	47
162	Tracking disease progression nonâ€invasively in Duchenne and Becker muscular dystrophies. Journal of Cachexia, Sarcopenia and Muscle, 2018, 9, 715-726.	7.3	47

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163	Targeted therapies for congenital myasthenic syndromes: systematic review and steps towards a treatabolome. Emerging Topics in Life Sciences, 2019, 3, 19-37.	2.6	47
164	A third of LGMD2A biopsies have normal calpain 3 proteolytic activity as determined by an in vitro assay. Neuromuscular Disorders, 2007, 17, 148-156.	0.6	46
165	Sarcoglycanopathies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2011, 101, 41-46.	1.8	46
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