

Hanns Lochmüller

List of Publications by Year in descending order

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470
papers

25,030
citations

6233

80
h-index

15683

125
g-index

485
all docs

485
docs citations

485
times ranked

24955
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	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. <i>Nucleic Acids Research</i> , 2019, 47, D1018-D1027.	6.5	539
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	Phenotypic spectrum associated with mutations of the mitochondrial polymerase β gene. <i>Brain</i> , 2006, 129, 1674-1684.	3.7	397
5	Prevalence, incidence and carrier frequency of 5q-linked spinal muscular atrophy – a literature review. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 124.	1.2	391
6	Mutations in dynamin 2 cause dominant centronuclear myopathy. <i>Nature Genetics</i> , 2005, 37, 1207-1209.	9.4	390
7	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. <i>American Journal of Human Genetics</i> , 2017, 100, 695-705.	2.6	305
8	The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene. <i>Brain</i> , 2007, 130, 2037-2044.	3.7	298
9	Mildly affected patients with spinal muscular atrophy are partially protected by an increased SMN2 copy number. <i>Human Genetics</i> , 2006, 119, 422-428.	1.8	292
10	A Mutation in the Dimerization Domain of Filamin C Causes a Novel Type of Autosomal Dominant Myofibrillar Myopathy. <i>American Journal of Human Genetics</i> , 2005, 77, 297-304.	2.6	268
11	Mutation in TACO1, encoding a translational activator of COX I, results in cytochrome c oxidase deficiency and late-onset Leigh syndrome. <i>Nature Genetics</i> , 2009, 41, 833-837.	9.4	260
12	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
13	Dok-7 Mutations Underlie a Neuromuscular Junction Synaptopathy. <i>Science</i> , 2006, 313, 1975-1978.	6.0	247
14	SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. <i>Nature Genetics</i> , 2009, 41, 654-656.	9.4	233
15	Mutations in SIL1 cause Marinesco-Sjögren syndrome, a cerebellar ataxia with cataract and myopathy. <i>Nature Genetics</i> , 2005, 37, 1312-1314.	9.4	232
16	Alterations in the ankyrin domain of TRPV4 cause congenital distal SMA, scapuloperoneal SMA and HMSN2C. <i>Nature Genetics</i> , 2010, 42, 160-164.	9.4	228
17	An agrin minigene rescues dystrophic symptoms in a mouse model for congenital muscular dystrophy. <i>Nature</i> , 2001, 413, 302-307.	13.7	222
18	Late onset Pompe disease: Clinical and neurophysiological spectrum of 38 patients including long-term follow-up in 18 patients. <i>Neuromuscular Disorders</i> , 2007, 17, 698-706.	0.3	208

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19	Risk of developing a mitochondrial DNA deletion disorder. <i>Lancet, The</i> , 2004, 364, 592-596.	6.3	201
20	Mutations and polymorphisms of the skeletal muscle β -actin gene (<i>ACTA1</i>). <i>Human Mutation</i> , 2009, 30, 1267-1277.	1.1	198
21	Autosomal-Dominant Distal Myopathy Associated with a Recurrent Missense Mutation in the Gene Encoding the Nuclear Matrix Protein, Matrin 3. <i>American Journal of Human Genetics</i> , 2009, 84, 511-518.	2.6	161
22	RD-Connect: An Integrated Platform Connecting Databases, Registries, Biobanks and Clinical Bioinformatics for Rare Disease Research. <i>Journal of General Internal Medicine</i> , 2014, 29, 780-787.	1.3	159
23	Dystrophin Expression in Muscles of mdx Mice After Adenovirus-Mediated <i>In Vivo</i> Gene Transfer. <i>Human Gene Therapy</i> , 1996, 7, 129-140.	1.4	158
24	Future of Rare Diseases Research 2017–2027: An IRDiRC Perspective. <i>Clinical and Translational Science</i> , 2018, 11, 21-27.	1.5	154
25	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. <i>American Journal of Human Genetics</i> , 2011, 88, 162-172.	2.6	153
26	The burden of Duchenne muscular dystrophy. <i>Neurology</i> , 2014, 83, 529-536.	1.5	149
27	Mutation History of the Roma/Gypsies. <i>American Journal of Human Genetics</i> , 2004, 75, 596-609.	2.6	148
28	Pathological consequences of VCP mutations on human striated muscle. <i>Brain</i> , 2007, 130, 381-393.	3.7	148
29	An X-Linked Myopathy with Postural Muscle Atrophy and Generalized Hypertrophy, Termed XMPMA, Is Caused by Mutations in FHL1. <i>American Journal of Human Genetics</i> , 2008, 82, 88-99.	2.6	148
30	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
31	Escobar Syndrome Is a Prenatal Myasthenia Caused by Disruption of the Acetylcholine Receptor Fetal β 3 Subunit. <i>American Journal of Human Genetics</i> , 2006, 79, 303-312.	2.6	146
32	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
33	Phenotypical spectrum of DOK7 mutations in congenital myasthenic syndromes. <i>Brain</i> , 2007, 130, 1497-1506.	3.7	143
34	Mitochondrial Phosphate Carrier Deficiency: A Novel Disorder of Oxidative Phosphorylation. <i>American Journal of Human Genetics</i> , 2007, 80, 478-484.	2.6	142
35	Infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). <i>Annals of Neurology</i> , 2003, 54, 719-724.	2.8	141
36	Diagnostic value of muscle MRI in differentiating LGMD2I from other LGMDs. <i>Journal of Neurology</i> , 2005, 252, 538-547.	1.8	136

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37	The ubiquitin-selective chaperone CDC-48/p97 links myosin assembly to human myopathy. <i>Nature Cell Biology</i> , 2007, 9, 379-390.	4.6	135
38	Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. <i>Brain</i> , 2008, 131, 747-759.	3.7	134
39	Dystrophin-deficient pigs provide new insights into the hierarchy of physiological derangements of dystrophic muscle. <i>Human Molecular Genetics</i> , 2013, 22, 4368-4382.	1.4	134
40	Clinical and morphological phenotype of the filamin myopathy: a study of 31 German patients. <i>Brain</i> , 2007, 130, 3250-3264.	3.7	132
41	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
42	Safety and Treatment Effects of Nusinersen in Longstanding Adult 5q-SMA Type 3 – A Prospective Observational Study. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 453-465.	1.1	132
43	Life expectancy at birth in Duchenne muscular dystrophy: a systematic review and meta-analysis. <i>European Journal of Epidemiology</i> , 2020, 35, 643-653.	2.5	132
44	Partial deficiency of the C-terminal-domain phosphatase of RNA polymerase II is associated with congenital cataracts facial dysmorphism neuropathy syndrome. <i>Nature Genetics</i> , 2003, 35, 185-189.	9.4	129
45	Targeted Next-Generation Sequencing of a 12.5 Mb Homozygous Region Reveals ANO10 Mutations in Patients with Autosomal-Recessive Cerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2010, 87, 813-819.	2.6	125
46	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
47	Sympathetic innervation controls homeostasis of neuromuscular junctions in health and disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 746-750.	3.3	123
48	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. <i>Nature Communications</i> , 2014, 5, 4287.	5.8	120
49	Congenital myasthenic syndromes due to mutations in <i>ALG2</i> and <i>ALG14</i> . <i>Brain</i> , 2013, 136, 944-956.	3.7	117
50	Recommendations for Improving the Quality of Rare Disease Registries. <i>International Journal of Environmental Research and Public Health</i> , 2018, 15, 1644.	1.2	116
51	Factors Influencing the Efficacy, Longevity, and Safety of Electroporation-Assisted Plasmid-Based Gene Transfer into Mouse Muscles. <i>Molecular Therapy</i> , 2004, 10, 447-455.	3.7	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. <i>Brain</i> , 2003, 126, 1026-1035.	3.7	112
53	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. <i>Brain</i> , 2009, 132, 3165-3174.	3.7	112
54	International Charter of principles for sharing bio-specimens and data. <i>European Journal of Human Genetics</i> , 2015, 23, 721-728.	1.4	112

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55	Mutations in <i>GMPPB</i> cause congenital myasthenic syndrome and bridge myasthenic disorders with dystroglycanopathies. <i>Brain</i> , 2015, 138, 2493-2504.	3.7	111
56	Missense mutations of <i>ACTA1</i> cause dominant congenital myopathy with cores. <i>Journal of Medical Genetics</i> , 2004, 41, 842-848.	1.5	110
57	Affinity proteomics within rare diseases: a <i>BIO</i> NMD study for blood biomarkers of muscular dystrophies. <i>EMBO Molecular Medicine</i> , 2014, 6, 918-936.	3.3	105
58	Limb-girdle muscular dystrophies. <i>Current Opinion in Neurology</i> , 2008, 21, 576-584.	1.8	104
59	Progress in Rare Diseases Research 2010-2016: An IRDiRC Perspective. <i>Clinical and Translational Science</i> , 2018, 11, 11-20.	1.5	104
60	EFNS guideline on diagnosis and management of limb girdle muscular dystrophies. <i>European Journal of Neurology</i> , 2007, 14, 1305-1312.	1.7	103
61	Phenotypes of the N88S Berardinelli-Seip congenital lipodystrophy 2 mutation. <i>Annals of Neurology</i> , 2005, 57, 415-424.	2.8	99
62	Adult-onset cerebellar ataxia due to mutations in <i>CABC1/ADCK3</i> . <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 174-178.	0.9	99
63	Congenital myasthenic syndromes: Achievements and limitations of phenotype-guided gene-after-gene sequencing in diagnostic practice: A study of 680 patients. <i>Human Mutation</i> , 2012, 33, 1474-1484.	1.1	99
64	A multi-source approach to determine SMA incidence and research ready population. <i>Journal of Neurology</i> , 2017, 264, 1465-1473.	1.8	98
65	Commonality of <i>TRIM32</i> mutation in causing sarcotubular myopathy and LGMD2H. <i>Annals of Neurology</i> , 2005, 57, 591-595.	2.8	96
66	Synaptotagmin 2 Mutations Cause an Autosomal-Dominant Form of Lambert-Eaton Myasthenic Syndrome and Nonprogressive Motor Neuropathy. <i>American Journal of Human Genetics</i> , 2014, 95, 332-339.	2.6	96
67	Human muscle cells express a B7-related molecule, B7H1, with strong negative immune regulatory potential: a novel mechanism of counterbalancing the immune attack in idiopathic inflammatory myopathies. <i>FASEB Journal</i> , 2003, 17, 1-16.	0.2	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. <i>Lancet Neurology</i> , The, 2018, 17, 671-680.	4.9	95
69	Attenuated muscle regeneration is a key factor in dysferlin-deficient muscular dystrophy. <i>Human Molecular Genetics</i> , 2009, 18, 1976-1989.	1.4	94
70	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
71	No overall hyposialylation in hereditary inclusion body myopathy myoblasts carrying the homozygous M712T <i>GNE</i> mutation. <i>Biochemical and Biophysical Research Communications</i> , 2005, 328, 221-226.	1.0	93
72	Scapulo-peroneal syndrome type Kaeser and a wide phenotypic spectrum of adult-onset, dominant myopathies are associated with the desmin mutation R350P. <i>Brain</i> , 2007, 130, 1485-1496.	3.7	92

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73	Expression of the E6 and E7 Genes of Human Papillomavirus (HPV16) Extends the Life Span of Human Myoblasts. <i>Experimental Cell Research</i> , 1999, 248, 186-193.	1.2	91
74	Inheritance patterns and phenotypic features of myofibrillar myopathy associated with a BAG3 mutation. <i>Neuromuscular Disorders</i> , 2010, 20, 438-442.	0.3	90
75	Reversible molecular pathology of skeletal muscle in spinal muscular atrophy. <i>Human Molecular Genetics</i> , 2011, 20, 4334-4344.	1.4	89
76	Dysferlin associates with the developing Tâ€tubule system in rodent and human skeletal muscle. <i>Muscle and Nerve</i> , 2010, 41, 166-173.	1.0	87
77	The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. <i>Journal of Neurology</i> , 2011, 258, 1987-1997.	1.8	87
78	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
79	Agrin mutations lead to a congenital myasthenic syndrome with distal muscle weakness and atrophy. <i>Brain</i> , 2014, 137, 2429-2443.	3.7	86
80	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
81	Muscle pathology in 57 patients with myotonic dystrophy type 2. <i>Muscle and Nerve</i> , 2004, 29, 275-281.	1.0	82
82	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
83	Quantifying the burden of caregiving in Duchenne muscular dystrophy. <i>Journal of Neurology</i> , 2016, 263, 906-915.	1.8	82
84	The principles of gene therapy for the nervous system. <i>Trends in Neurosciences</i> , 1996, 19, 49-54.	4.2	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. <i>FASEB Journal</i> , 2006, 20, 118-120.	0.2	81
86	The p.G154S mutation of the alpha-B crystallin gene (<i>CRYAB</i>) causes late-onset distal myopathy. <i>Neuromuscular Disorders</i> , 2010, 20, 255-259.	0.3	81
87	Genetic heterogeneity of motor neuropathies. <i>Neurology</i> , 2017, 88, 1226-1234.	1.5	81
88	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
89	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
90	The non-classical MHC molecule HLA-G protects human muscle cells from immune-mediated lysis: implications for myoblast transplantation and gene therapy. <i>Brain</i> , 2003, 126, 176-185.	3.7	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.	0.9	79
92	Mutations in the collagen XII gene define a new form of extracellular matrix-related myopathy. Human Molecular Genetics, 2014, 23, 2353-2363.	1.4	79
93	Recessive Mutations in the ± 3 (VI) Collagen Gene COL6A3 Cause Early-Onset Isolated Dystonia. American Journal of Human Genetics, 2015, 96, 883-893.	2.6	79
94	Muscle MRI findings in limb girdle muscular dystrophy type 2L. Neuromuscular Disorders, 2012, 22, S122-S129.	0.3	77
95	Treatment of dysferlinopathy with deflazacort: a double-blind, placebo-controlled clinical trial. Orphanet Journal of Rare Diseases, 2013, 8, 26.	1.2	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.	1.8	76
97	Homozygous mutations in caveolin-3 cause a severe form of rippling muscle disease. Annals of Neurology, 2003, 53, 512-520.	2.8	75
98	The Clinical Outcome Study for dysferlinopathy. Neurology: Genetics, 2016, 2, e89.	0.9	75
99	ANO10 mutations cause ataxia and coenzyme Q10 deficiency. Journal of Neurology, 2014, 261, 2192-2198.	1.8	74
100	Mutation in dystrophin-encoding gene affects energy metabolism in mouse myoblasts. Biochemical and Biophysical Research Communications, 2009, 386, 463-466.	1.0	73
101	Fibronectin is a serum biomarker for Duchenne muscular dystrophy. Proteomics - Clinical Applications, 2014, 8, 269-278.	0.8	73
102	Differential Short-Term Transduction Efficiency of Adult versus Newborn Mouse Tissues by Adenoviral Recombinants. Experimental and Molecular Pathology, 1995, 62, 131-143.	0.9	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.	1.4	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.	1.2	72
105	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850.	1.8	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.	2.9	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.	0.9	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.	1.1	71

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109	Association Study of Exon Variants in the NF- κ B and TGF β 2 Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2016, 99, 1163-1171.	2.6	71
110	Genetic determinants of disease severity in the myotonic dystrophy type 1 OPTIMISTIC cohort. <i>Neurology</i> , 2019, 93, e995-e1009.	1.5	71
111	Developmental defects in a zebrafish model for muscular dystrophies associated with the loss of fukutin-related protein (FKRP). <i>Brain</i> , 2008, 131, 1551-1561.	3.7	70
112	“You should at least ask”™. The expectations, hopes and fears of rare disease patients on large-scale data and biomaterial sharing for genomics research. <i>European Journal of Human Genetics</i> , 2016, 24, 1403-1408.	1.4	70
113	Gentamicin fails to increase dystrophin expression in dystrophin-deficient muscle. <i>Muscle and Nerve</i> , 2003, 27, 624-627.	1.0	69
114	Parkinson syndrome, neuropathy, and myopathy caused by the mutation A8344G (MERRF) in tRNALys. <i>Neurology</i> , 2007, 68, 56-58.	1.5	69
115	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
116	Antibody-Mediated Targeting of an Adenovirus Vector Modified To Contain a Synthetic Immunoglobulin G-Binding Domain in the Capsid. <i>Journal of Virology</i> , 2003, 77, 2093-2104.	1.5	68
117	Impaired Presynaptic High-Affinity Choline Transporter Causes a Congenital Myasthenic Syndrome with Episodic Apnea. <i>American Journal of Human Genetics</i> , 2016, 99, 753-761.	2.6	68
118	Linker molecules between laminins and dystroglycan ameliorate laminin- α 2-deficient muscular dystrophy at all disease stages. <i>Journal of Cell Biology</i> , 2007, 176, 979-993.	2.3	67
119	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
120	SMartCARE™ platform to collect real-life outcome data of patients with spinal muscular atrophy. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 18.	1.2	67
121	Screening for Carnitine Palmitoyltransferase II Deficiency by Tandem Mass Spectrometry. <i>Journal of Inherited Metabolic Disease</i> , 2002, 25, 17-27.	1.7	66
122	Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. <i>Brain</i> , 2011, 134, 183-195.	3.7	66
123	Human Skeletal Muscle-derived CD133+ Cells Form Functional Satellite Cells After Intramuscular Transplantation in Immunodeficient Host Mice. <i>Molecular Therapy</i> , 2014, 22, 1008-1017.	3.7	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. <i>Neuromuscular Disorders</i> , 2003, 13, 830-834.	0.3	65
125	A retrospective clinical study of the treatment of slow-channel congenital myasthenic syndrome. <i>Journal of Neurology</i> , 2012, 259, 474-481.	1.8	65
126	Further evidence for genetic heterogeneity of distal HMN type V, CMT2 with predominant hand involvement and Silver syndrome. <i>Journal of the Neurological Sciences</i> , 2007, 263, 100-106.	0.3	64

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127	5â€² Trans-Splicing Repair of the PLEC1 Gene. <i>Journal of Investigative Dermatology</i> , 2008, 128, 568-574.	0.3	64
128	Acute liver failure with subsequent cirrhosis as the primary manifestation of <i>TRMU</i> mutations. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 197-201.	1.7	64
129	<i>ANO5</i> Gene Analysis in a Large Cohort of Patients with Anoctaminopathy: Confirmation of Male Prevalence and High Occurrence of the Common Exon 5 Gene Mutation. <i>Human Mutation</i> , 2013, 34, 1111-1118.	1.1	64
130	The EuroBioBank Network: 10 years of hands-on experience of collaborative, transnational biobanking for rare diseases. <i>European Journal of Human Genetics</i> , 2015, 23, 1116-1123.	1.4	63
131	The International Rare Diseases Research Consortium: Policies and Guidelines to maximize impact. <i>European Journal of Human Genetics</i> , 2017, 25, 1293-1302.	1.4	62
132	Mutation screening of the N-myc downstream-regulated gene 1 (NDRG1) in patients with Charcot-Marie-Tooth Disease. <i>Human Mutation</i> , 2003, 22, 129-135.	1.1	61
133	Adult-onset glycogen storage disease type 2: clinico-pathological phenotype revisited. <i>Neuropathology and Applied Neurobiology</i> , 2007, 33, 070615152525006-???	1.8	61
134	Long-term follow-up in patients with congenital myasthenic syndrome due to CHAT mutations. <i>European Journal of Paediatric Neurology</i> , 2010, 14, 326-333.	0.7	61
135	Congenital myasthenic syndromes: spotlight on genetic defects of neuromuscular transmission. <i>Expert Reviews in Molecular Medicine</i> , 2007, 9, 1-20.	1.6	60
136	Novel POMGnT1 mutations define broader phenotypic spectrum of muscleâ€“eyeâ€“brain disease. <i>Neurogenetics</i> , 2007, 8, 279-288.	0.7	60
137	Duchenne muscular dystrophy and caregiver burden: a systematic review. <i>Developmental Medicine and Child Neurology</i> , 2018, 60, 987-996.	1.1	59
138	Characterization of human muscle type cofilin (CFL2) in normal and regenerating muscle. <i>FEBS Journal</i> , 2001, 268, 3473-3482.	0.2	58
139	Creatine monohydrate in myotonic dystrophy. <i>Journal of Neurology</i> , 2002, 249, 1717-1722.	1.8	58
140	Homozygosity for CCTG mutation in myotonic dystrophy type 2. <i>Brain</i> , 2004, 127, 1868-1877.	3.7	58
141	Late onset in dysferlinopathy widens the clinical spectrum. <i>Neuromuscular Disorders</i> , 2008, 18, 288-290.	0.3	57
142	Increased susceptibility to ATP via alteration of P2X receptor function in dystrophic mdx mouse muscle cells. <i>FASEB Journal</i> , 2006, 20, 610-620.	0.2	56
143	Nemaline myopathy caused by mutations in the nebulin gene may present as a distal myopathy. <i>Neuromuscular Disorders</i> , 2011, 21, 556-562.	0.3	56
144	Exon Skipping and Gene Transfer Restore Dystrophin Expression in Human Induced Pluripotent Stem Cells-Cardiomyocytes Harboring <i>DMD</i> Mutations. <i>Stem Cells and Development</i> , 2013, 22, 2714-2724.	1.1	56

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145	High frequency of co-segregating CLCN1 mutations among myotonic dystrophy type 2 patients from Finland and Germany. <i>Journal of Neurology</i> , 2008, 255, 1731-1736.	1.8	55
146	Therapeutic Strategies in Congenital Myasthenic Syndromes. <i>Neurotherapeutics</i> , 2008, 5, 542-547.	2.1	55
147	NF- κ B-dependent expression of the antiapoptotic factor cFLIP is regulated by calpain 3, the protein involved in limb-girdle muscular dystrophy type 2A. <i>FASEB Journal</i> , 2008, 22, 1521-1529.	0.2	55
148	European Cross-Sectional Survey of Current Care Practices for Duchenne Muscular Dystrophy Reveals Regional and Age-Dependent Differences. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 517-527.	1.1	55
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