Hanns LochmÃ¼ller

List of Publications by Year
in descending order

470 papers	25,030 citations	7251 80 h-index	17891 125 g-index
485 all docs	485 docs citations	485 times ranked	26904 citing authors

#	Article	IF	Citations
1	A de novo <scp>CSDE1</scp> variant causing neurodevelopmental delay, intellectual disability, neurologic and psychiatric symptoms in a child of consanguineous parents. American Journal of Medical Genetics, Part A, 2022, 188, 283-291.	0.7	1
2	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. Brain, 2022, 145, 1507-1518.	3.7	14
3	Distinct and Recognisable Muscle MRI Pattern in a Series of Adults Harbouring an Identical GMPPB Gene Mutation. Journal of Neuromuscular Diseases, 2022, 9, 95-109.	1.1	4
4	Congenital myasthenic syndrome: Correlation between clinical features and molecular diagnosis. European Journal of Neurology, 2022, 29, 833-842.	1.7	14
5	Multispectral optoacoustic tomography for non-invasive disease phenotyping in pediatric spinal muscular atrophy patients. Photoacoustics, 2022, 25, 100315.	4.4	16
6	Novel insights into PORCN mutations, associated phenotypes and pathophysiological aspects. Orphanet Journal of Rare Diseases, 2022, 17, 29.	1.2	3
7	NCAM1 and GDF15 are biomarkers of Charcot-Marie-Tooth disease in patients and mice. Brain, 2022, 145, 3999-4015.	3.7	12
8	The RDâ€Connect Genomeâ€Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. Human Mutation, 2022, , .	1.1	18
9	Serum miRNAs as biomarkers for the rare types of muscular dystrophy. Neuromuscular Disorders, 2022, 32, 332-346.	0.3	5
10	Cathepsin D as biomarker in cerebrospinal fluid of nusinersenâ€treated patients with spinal muscular atrophy. European Journal of Neurology, 2022, 29, 2084-2096.	1.7	13
11	Circulating small RNA signatures differentiate accurately the subtypes of muscular dystrophies: small-RNA next-generation sequencing analytics and functional insights. RNA Biology, 2022, 19, 507-518.	1.5	1
12	Case Report: Advanced Skeletal Muscle Imaging in S-Adenosylhomocysteine Hydrolase Deficiency and Further Insight Into Muscle Pathology. Frontiers in Pediatrics, 2022, 10, 847445.	0.9	1
13	Collagen VI Regulates Motor Circuit Plasticity and Motor Performance by Cannabinoid Modulation. Journal of Neuroscience, 2022, 42, 1557-1573.	1.7	1
14	MYTHO: A novel regulator of autophagy and skeletal muscle health. FASEB Journal, 2022, 36, .	0.2	0
15	Expanding the clinical and molecular spectrum of <scp>ATP6V1A</scp> related metabolic cutis laxa. Journal of Inherited Metabolic Disease, 2021, 44, 972-986.	1.7	7
16	Molecular pathophysiology of human MICU1 deficiency. Neuropathology and Applied Neurobiology, 2021, 47, 840-855.	1.8	15
17	Costs of Illness of Spinal Muscular Atrophy: A Systematic Review. Applied Health Economics and Health Policy, 2021, 19, 501-520.	1.0	16
18	Disease monitoring programs of rare genetic diseases: transparent data sharing between academic and commercial stakeholders. Orphanet Journal of Rare Diseases, 2021, 16, 141.	1.2	6

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19	Associations Between Variant Repeat Interruptions and Clinical Outcomes in Myotonic Dystrophy Type 1. Neurology: Genetics, 2021, 7, e572.	0.9	10
20	Results from a 3-year Non-interventional, Observational Disease Monitoring Program in Adults with GNE Myopathy. Journal of Neuromuscular Diseases, 2021, 8, 225-234.	1.1	9
21	Recessive VAMP1 mutations associated with severe congenital myasthenic syndromes â€“ A recognizable clinical phenotype. European Journal of Paediatric Neurology, 2021, 31, 54-60.	0.7	7
22	INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. Brain, 2021, 144, 2427-2442.	3.7	7
23	Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). European Journal of Human Genetics, 2021, 29, 1348-1353.	1.4	10
24	Biomarkers in Duchenne and Becker muscular dystrophies. Muscle and Nerve, 2021, 64, 4-5.	1.0	0
25	A Canadian Adult Spinal Muscular Atrophy Outcome Measures Toolkit: Results of a National Consensus using a Modified Delphi Method. Journal of Neuromuscular Diseases, 2021, 8, 579-588.	1.1	7
26	E-Health & Innovation to Overcome Barriers in Neuromuscular Diseases. Report from the 1st eNMD Congress: Nice, France, March 22-23, 2019. Journal of Neuromuscular Diseases, 2021, 8, 743-754.	1.1	2
27	Clinical Outcome Evaluations and CBT Response Prediction in Myotonic Dystrophy. Journal of Neuromuscular Diseases, 2021, 8, 1031-1046.	1.1	4
28	Dysregulation of GSK3Î²-Target Proteins in Skin Fibroblasts of Myotonic Dystrophy Type 1 (DM1) Patients. Journal of Neuromuscular Diseases, 2021, 8, 603-619.	1.1	2
29	A founder mutation in theÂGMPPBÂgene [c.1000Gâ€‰>â€‰A (p.Asp334Asn)] causes a mild form of limb-girdle muscular dystrophy/congenital myasthenic syndrome (LGMD/CMS) in South Indian patients. Neurogenetics, 2021, 22, 271-285.	0.7	7
30	miR-223-3p and miR-24-3p as novel serum-based biomarkers for myotonic dystrophy type 1. Molecular Therapy - Methods and Clinical Development, 2021, 23, 169-183.	1.8	6
31	Autosomal recessive variants in TUBGCP2 alter the Î³-tubulin ring complex leading to neurodevelopmental disease. IScience, 2021, 24, 101948.	1.9	6
32	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	2.6	11
33	Expanding the Phenotypic Spectrum of ECEL1-Associated Distal Arthrogryposis. Children, 2021, 8, 909.	0.6	4
34	Homozygous WASHC4 variant in two sisters causes a syndromic phenotype defined by dysmorphisms, intellectual disability, profound developmental disorder, and skeletal muscle involvement. Journal of Pathology, 2021, , .	2.1	5
35	SMArtCARE Real-World Data on Drug Treatment for Spinal Muscular Atrophy. , 2021, 52, .		0
36	Noninvasive Imaging in Pediatric Spinal Muscular Atrophy Patients Using Multispectral Optoacoustic Tomography: A Proof-of-Concept Study. Neuropediatrics, 2021, 52, .	0.3	0

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37	Editorial: Molecular Mechanisms Underlying Assembly and Maintenance of the Neuromuscular Junction. Frontiers in Molecular Neuroscience, 2021, 14, 797832.	1.4	0
38	Congenital myasthenic syndrome in a cohort of patients with â€˜doubleâ€™ seronegative myasthenia gravis. Arquivos De Neuro-Psiquiatria, 2021, , .	0.3	3
39	Congenital myasthenic syndrome with mild intellectual disability caused by a recurrent SLC25A1 variant. European Journal of Human Genetics, 2020, 28, 373-377.	1.4	20
40	Severe neurodevelopmental disease caused by a homozygous TLK2 variant. European Journal of Human Genetics, 2020, 28, 383-387.	1.4	6
41	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	1.7	43
42	Activities of daily living in myotonic dystrophy type 1. Acta Neurologica Scandinavica, 2020, 141, 380-387.	1.0	7
43	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.	2.9	27
44	A novel, pathogenic dinucleotide deletion in the mitochondrial MT-TY gene causing myasthenia-like features. Neuromuscular Disorders, 2020, 30, 661-668.	0.3	8
45	Biallelic loss of function variants in <scp>SYT2</scp> cause a treatable congenital onset presynaptic myasthenic syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2272-2283.	0.7	20
46	Behr syndrome and hypertrophic cardiomyopathy in a family with a novel UCHL1 deletion. Journal of Neurology, 2020, 267, 3643-3649.	1.8	8
47	COL4A1 -related autosomal recessive encephalopathy in 2 Turkish children. Neurology: Genetics, 2020, 6, e392.	0.9	9
48	Loss of supervillin causes myopathy with myofibrillar disorganization and autophagic vacuoles. Brain, 2020, 143, 2406-2420.	3.7	15
49	A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome. Orphanet Journal of Rare Diseases, 2020, 15, 206.	1.2	21
50	A Phase 2 Study of AMO-02 (Tideglusib) in Congenital and Childhood-Onset Myotonic Dystrophy Type 1 (DM1). Pediatric Neurology, 2020, 112, 84-93.	1.0	44
51	Long Term Follow-Up on Pediatric Cases With Congenital Myasthenic Syndromesâ€”A Retrospective Single Centre Cohort Study. Frontiers in Human Neuroscience, 2020, 14, 560860.	1.0	14
52	Economic Costs of Myasthenia Gravis: A Systematic Review. Pharmacoeconomics, 2020, 38, 715-728.	1.7	22
53	Blood-derived biomarkers correlate with clinical progression in Duchenne muscular dystrophy. Journal of Neuromuscular Diseases, 2020, 7, 231-246.	1.1	20
54	Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. Lancet Neurology, The, 2020, 19, 522-532.	4.9	36

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55	A National Spinal Muscular Atrophy Registry for Real-World Evidence. Canadian Journal of Neurological Sciences, 2020, 47, 810-815.	0.3	6
56	An improved method for culturing myotubes on laminins for the robust clustering of postsynaptic machinery. Scientific Reports, 2020, 10, 4524.	1.6	13
57	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. Journal of Neuromuscular Diseases, 2020, 7, 153-166.	1.1	18
58	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. Journal of Molecular Diagnostics, 2020, 22, 1205-1215.	1.2	14
59	Change over time in ability to perform activities of daily living in myotonic dystrophy type 1. Journal of Neurology, 2020, 267, 3235-3242.	1.8	3
60	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. Frontiers in Genetics, 2020, 11, 605.	1.1	9
61	Life expectancy at birth in Duchenne muscular dystrophy: a systematic review and meta-analysis. European Journal of Epidemiology, 2020, 35, 643-653.	2.5	132
62	Severe congenital myasthenic syndrome associated with novel biallelic mutation of the CHRND gene. Neuromuscular Disorders, 2020, 30, 336-339.	0.3	2
63	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
64	Multiple acyl-coenzyme A dehydrogenase deficiency shows a possible founder effect and is the most frequent cause of lipid storage myopathy in Iran. Journal of the Neurological Sciences, 2020, 411, 116707.	0.3	14
65	Congenital myasthenic syndrome due to DOK7 mutation in a cohort of patients with â€˜unexplainedâ€™ limb-girdle muscular weakness. Journal of Clinical Neuroscience, 2020, 75, 195-198.	0.8	2
66	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.	1.7	20
67	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364.	3.5	26
68	Confirmation of TACO1 as a Leigh Syndrome Disease Gene in Two Additional Families. Journal of Neuromuscular Diseases, 2020, 7, 301-308.	1.1	8
69	Modulation of the Acetylcholine Receptor Clustering Pathway Improves Neuromuscular Junction Structure and Muscle Strength in a Mouse Model of Congenital Myasthenic Syndrome. Frontiers in Molecular Neuroscience, 2020, 13, 594220.	1.4	5
70	Analyzing walking speeds with ankle and wrist worn accelerometers in a cohort with myotonic dystrophy. Disability and Rehabilitation, 2019, 41, 2972-2978.	0.9	13
71	Modulation of Agrin and RhoA Pathways Ameliorates Movement Defects and Synapse Morphology in MYO9A-Depleted Zebrafish. Cells, 2019, 8, 848.	1.8	10
72	De-duplicating patient records from three independent data sources reveals the incidence of rare neuromuscular disorders in Germany. Orphanet Journal of Rare Diseases, 2019, 14, 152.	1.2	22

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73	Analysis of the functional capacity outcome measures for myotonic dystrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1487-1497.	1.7	11
74	Reproductive Cancer Risk Factors in Women With Myotonic Dystrophy (DM): Survey Data From the US and UK DM Registries. Frontiers in Neurology, 2019, 10, 1071.	1.1	5
75	De novo variant in SCN4A causes neonatal sodium channel myotonia with general muscle stiffness and respiratory failure. Neuromuscular Disorders, 2019, 29, 907-909.	0.3	5
76	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.	1.1	132
77	SMArtCAREÂ-ÂA platform to collect real-life outcome data of patients with spinal muscular atrophy. Orphanet Journal of Rare Diseases, 2019, 14, 18.	1.2	67
78	A phase 3 randomized study evaluating sialic acid extended-release for GNE myopathy. Neurology, 2019, 92, e2109-e2117.	1.5	40
79	GNE genotype explains 20% of phenotypic variability in GNE myopathy. Neurology: Genetics, 2019, 5, e308.	0.9	22
80	Correction of pseudoexon splicing caused by a novel intronic dysferlin mutation. Annals of Clinical and Translational Neurology, 2019, 6, 642-654.	1.7	20
81	Salbutamol modifies the neuromuscular junction in a mouse model of ColQ myasthenic syndrome. Human Molecular Genetics, 2019, 28, 2339-2351.	1.4	29
82	Dihydropyridine Receptor Congenital Myopathy In A Consangineous Turkish Family. Journal of Neuromuscular Diseases, 2019, 6, 377-384.	1.1	12
83	Increasing phenotypic annotation improves the diagnostic rate of exome sequencing in a rare neuromuscular disorder. Human Mutation, 2019, 40, 1797-1812.	1.1	22
84	â€œBe an ambassador for change that you would like to seeâ€• a call to action to all stakeholders for co-creation in healthcare and medical research to improve quality of life of people with a neuromuscular disease. Orphanet Journal of Rare Diseases, 2019, 14, 126.	1.2	10
85	Phenotype may predict the clinical course of facioscapolohumeral muscular dystrophy. Muscle and Nerve, 2019, 59, 711-713.	1.0	12
86	MACF1 links Rapsyn to microtubule- and actin-binding proteins to maintain neuromuscular synapses. Journal of Cell Biology, 2019, 218, 1686-1705.	2.3	34
87	Quality of life of patients with spinal muscular atrophy: A systematic review. European Journal of Paediatric Neurology, 2019, 23, 347-356.	0.7	48
88	CHRNGâ€•related nonlethal multiple pterygium syndrome: Muscle imaging pattern and clinical, histopathological, and molecular genetic findings. American Journal of Medical Genetics, Part A, 2019, 179, 915-926.	0.7	11
89	Incomplete description of the current body of evidence of the health economics of Duchenne muscular dystrophy. Orphanet Journal of Rare Diseases, 2019, 14, 75.	1.2	0
90	Health-Related Quality of Life in Patients with Adult-Onset Myotonic Dystrophy Type 1: A Systematic Review. Patient, 2019, 12, 365-373.	1.1	11

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91	The Position of Neuromuscular Patients in Shared Decision Making. Report from the 235th ENMC Workshop: Milan, Italy, January 19-20, 2018. Journal of Neuromuscular Diseases, 2019, 6, 161-172.	1.1	7
92	Targeted therapies for congenital myasthenic syndromes: systematic review and steps towards a treatabolome. Emerging Topics in Life Sciences, 2019, 3, 19-37.	1.1	47
93	Disease burden of myotonic dystrophy type 1. Journal of Neurology, 2019, 266, 998-1006.	1.8	21
94	237th ENMC International Workshop: GNE myopathy â€“ current and future research Hoofddorp, The Netherlands, 14â€“16 September 2018. Neuromuscular Disorders, 2019, 29, 401-410.	0.3	5
95	Congenital myasthenic syndrome caused by novel COL13A1 mutations. Journal of Neurology, 2019, 266, 1107-1112.	1.8	14
96	Genetic determinants of disease severity in the myotonic dystrophy type 1 OPTIMISTIC cohort. Neurology, 2019, 93, e995-e1009.	1.5	71
97	A Review of International Biobanks and Networks: Success Factors and Key Benchmarksâ€”A 10-Year Retrospective Review. Biopreservation and Biobanking, 2019, 17, 512-519.	0.5	10
98	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	6.5	539
99	SIL1 deficiency causes degenerative changes of peripheral nerves and neuromuscular junctions in fish, mice and human. Neurobiology of Disease, 2019, 124, 218-229.	2.1	7
100	Assessment of disease progression in dysferlinopathy. Neurology, 2019, 92, .	1.5	20
101	The oral splicing modifier RG7800 increases full length survival of motor neuron 2 mRNA and survival of motor neuron protein: Results from trials in healthy adults and patients with spinal muscular atrophy. Neuromuscular Disorders, 2019, 29, 21-29.	0.3	30
102	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.	0.9	10
103	The beta-adrenergic agonist salbutamol modulates neuromuscular junction formation in zebrafish models of human myasthenic syndromes. Human Molecular Genetics, 2018, 27, 1556-1564.	1.4	28
104	MYO9A deficiency in motor neurons is associated with reduced neuromuscular agrin secretion. Human Molecular Genetics, 2018, 27, 1434-1446.	1.4	14
105	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.	1.0	10
106	RD-Connect, NeurOmics and EURenOmics: collaborative European initiative for rare diseases. European Journal of Human Genetics, 2018, 26, 778-785.	1.4	55
107	Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophyâ€“like disease. Genetics in Medicine, 2018, 20, 1224-1235.	1.1	31
108	Tracking disease progression nonâ€invasively in Duchenne and Becker muscular dystrophies. Journal of Cachexia, Sarcopenia and Muscle, 2018, 9, 715-726.	2.9	47

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109	Mutations in glycyl-tRNA synthetase impair mitochondrial metabolism in neurons. Human Molecular Genetics, 2018, 27, 2187-2204.	1.4	26
110	Mobility shift of beta-dystroglycan as a marker ofGMPPBgene-related muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 762-768.	0.9	15
111	A common CHRNE mutation in Brazilian patients with congenital myasthenic syndrome. Journal of Neurology, 2018, 265, 708-713.	1.8	18
112	The RD-Connect Registry & Biobank Finder: a tool for sharing aggregated data and metadata among rare disease researchers. European Journal of Human Genetics, 2018, 26, 631-643.	1.4	33
113	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. Human Molecular Genetics, 2018, 27, 1186-1195.	1.4	52
114	Phenotypic stratification and genotypeâ€“phenotype correlation in a heterogeneous, international cohort of GNE myopathy patients: First report from the GNE myopathy Disease Monitoring Program, registry portion. Neuromuscular Disorders, 2018, 28, 158-168.	0.3	35
115	Clinical and research strategies for limbâ€girdle congenital myasthenic syndromes. Annals of the New York Academy of Sciences, 2018, 1412, 102-112.	1.8	17
116	Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo PTEN mutation. Neurology, 2018, 90, e1842-e1848.	1.5	4
117	How to Spot Congenital Myasthenic Syndromes Resembling the Lambertâ€“Eaton Myasthenic Syndrome? A Brief Review of Clinical, Electrophysiological, and Genetics Features. NeuroMolecular Medicine, 2018, 20, 205-214.	1.8	4
118	Recessive variants of MuSK are associated with late onset CMS and predominant limb girdle weakness. American Journal of Medical Genetics, Part A, 2018, 176, 1594-1601.	0.7	25
119	Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1224-1226.	0.9	19
120	Whole-exome sequencing identifies mutations in MYMK in a mild form of Carey-Fineman-Ziter syndrome. Neurology: Genetics, 2018, 4, e226.	0.9	6
121	Compliance to care guidelines for Duchenne muscular dystrophy in Italy. Neuromuscular Disorders, 2018, 28, 100.	0.3	2
122	Mass spectrometryâ€based protein analysis to unravel the tissue pathophysiology in Duchenne muscular dystrophy. Proteomics - Clinical Applications, 2018, 12, 1700071.	0.8	26
123	Benign and malignant tumors in the UK myotonic dystrophy patient registry. Muscle and Nerve, 2018, 57, 316-320.	1.0	15
124	Chronic pain has a strong impact on quality of life in facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2018, 57, 380-387.	1.0	33
125	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.	0.3	13
126	Intersection of Proteomics and Genomics to â€œSolve the Unsolvedâ€• in Rare Disorders such as Neurodegenerative and Neuromuscular Diseases. Proteomics - Clinical Applications, 2018, 12, 1700073.	0.8	33

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127	Congenital myasthenic syndrome with episodic apnoea: clinical, neurophysiological and genetic features in the long-term follow-up of 19 patients. Journal of Neurology, 2018, 265, 194-203.	1.8	36
128	Risk of skin cancer among patients with myotonic dystrophy type 1 based on primary care physician data from the <scp>U</scp>.<scp>K</scp>. <scp>C</scp>linical <scp>P</scp>ractice <scp>R</scp>esearch <scp>D</scp>atalink. International Journal of Cancer, 2018, 142, 1174-1181.	2.3	25
129	Progress in Rare Diseases Research 2010â€“2016: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 11-20.	1.5	104
130	Future of Rare Diseases Research 2017â€“2027: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 21-27.	1.5	154
131	Neuromuscular Junction Changes in a Mouse Model of Charcot-Marie-Tooth Disease Type 4C. International Journal of Molecular Sciences, 2018, 19, 4072.	1.8	24
132	A nomenclature and classification for the congenital myasthenic syndromes: preparing for FAIR data in the genomic era. Orphanet Journal of Rare Diseases, 2018, 13, 211.	1.2	17
133	Clinical variability of early-onset congenital myasthenic syndrome due to biallelic RAPSN mutations in Brazil. Neuromuscular Disorders, 2018, 28, 961-964.	0.3	13
134	Comprehensive RNA-Sequencing Analysis in Serum and Muscle Reveals Novel Small RNA Signatures with Biomarker Potential for DMD. Molecular Therapy - Nucleic Acids, 2018, 13, 1-15.	2.3	41
135	GNE myopathy in the bedouin population of Kuwait: Genetics, prevalence, and clinical description. Muscle and Nerve, 2018, 58, 700-707.	1.0	8
136	Eight years after an international workshop on myotonic dystrophy patient registries: case study of a global collaboration for a rare disease. Orphanet Journal of Rare Diseases, 2018, 13, 155.	1.2	19
137	Biochemical and pathological changes result from mutated Caveolin-3 in muscle. Skeletal Muscle, 2018, 8, 28.	1.9	19
138	Position Statement: Sharing of Clinical Research Data in Spinal Muscular Atrophy to Accelerate Research and Improve Outcomes for Patients. Journal of Neuromuscular Diseases, 2018, 5, 131-133.	1.1	10
139	A checklist for clinical trials in rare disease: obstacles and anticipatory actionsâ€”lessons learned from the FOR-DMD trial. Trials, 2018, 19, 291.	0.7	26
140	Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1071-1081.	0.9	81
141	GNE myopathy: from clinics and genetics to pathology and research strategies. Orphanet Journal of Rare Diseases, 2018, 13, 70.	1.2	36
142	Recommendations for Improving the Quality of Rare Disease Registries. International Journal of Environmental Research and Public Health, 2018, 15, 1644.	1.2	116
143	Duchenne muscular dystrophy and caregiver burden: a systematic review. Developmental Medicine and Child Neurology, 2018, 60, 987-996.	1.1	59
144	GFPT1 deficiency in muscle leads to myasthenia and myopathy in mice. Human Molecular Genetics, 2018, 27, 3218-3232.	1.4	18

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145	Limb girdle muscular dystrophy 2G in a religious minority of Bulgarian Muslims homozygous for the c.75G>A, p.Trp25X mutation. Neuromuscular Disorders, 2018, 28, 625-632.	0.3	12
146	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.	4.9	95
147	P 898. SMArtCARE: Longitudinal Data Collection of Patients with Spinal Muscular Atrophy in German-Speaking Countries. Neuropediatrics, 2018, 49, .	0.3	0
148	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
149	Reduced serum myostatin concentrations associated with genetic muscle disease progression. Journal of Neurology, 2017, 264, 541-553.	1.8	51
150	â€˜IRDiRC Recognized Resourcesâ€™: a new mechanism to support scientists to conduct efficient, high-quality research for rare diseases. European Journal of Human Genetics, 2017, 25, 162-165.	1.4	30
151	Predictors of Health-Related Quality of Life in boys with Duchenne muscular dystrophy from six European countries. Journal of Neurology, 2017, 264, 709-723.	1.8	25
152	Functional impairment in patients with myotonic dystrophy type 1 can be assessed by an ataxia rating scale (SARA). Journal of Neurology, 2017, 264, 701-708.	1.8	12
153	Genetic heterogeneity of motor neuropathies. Neurology, 2017, 88, 1226-1234.	1.5	81
154	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.	2.6	67
155	Intragenic DOK7 deletion detected by whole-genome sequencing in congenital myasthenic syndromes. Neurology: Genetics, 2017, 3, e152.	0.9	4
156	Critical points for an accurate human genome analysis. Human Mutation, 2017, 38, 912-921.	1.1	5
157	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
158	The UK Myotonic Dystrophy Patient Registry: facilitating and accelerating clinical research. Journal of Neurology, 2017, 264, 979-988.	1.8	23
159	SPARC Interacts with Actin in Skeletal Muscle inÂVitro and inÂVivo. American Journal of Pathology, 2017, 187, 457-474.	1.9	29
160	Complex phenotypes associated with STIM1 mutations in both coiled coil and EF-hand domains. Neuromuscular Disorders, 2017, 27, 861-872.	0.3	39
161	The Increasing Genetic and Phenotypical Diversity of Congenital Myasthenic Syndromes. Neuropediatrics, 2017, 48, 294-308.	0.3	43
162	Respiratory involvement in ambulant and non-ambulant patients with facioscapulohumeral muscular dystrophy. Journal of Neurology, 2017, 264, 1271-1280.	1.8	30

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163	Facilitating orphan drug development: Proceedings of the TREAT-NMD International Conference, December 2015, Washington, DC, USA. Neuromuscular Disorders, 2017, 27, 693-701.	0.3	1
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