Hanns Lochmller

List of Publications by Year in Descending Order

456 papers	18,911 citations	73 h-index	111 g-index
485 ext. papers	22,324 ext. citations	5.9 avg, IF	6.17 L-index

#	Paper	IF	Citations
456	Novel insights into PORCN mutations, associated phenotypes and pathophysiological aspects.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 29	4.2	0
455	Multispectral optoacoustic tomography for non-invasive disease phenotyping in pediatric spinal muscular atrophy patients. *Photoacoustics*, **2022**, 25, 100315	9	1
454	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	4.8	1
453	Case Report: Advanced Skeletal Muscle Imaging in S-Adenosylhomocysteine Hydrolase Deficiency and Further Insight Into Muscle Pathology.. *Frontiers in Pediatrics*, **2022**, 10, 847445	3.4	
452	Congenital myasthenic syndrome: Correlation between clinical features and molecular diagnosis. *European Journal of Neurology*, **2021**,	6	2
451	SMArtCARE Real-World Data on Drug Treatment for Spinal Muscular Atrophy **2021**, 52,		
450	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. *Brain*, **2021**,	11.2	1
449	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	11	3
448	Expanding the Phenotypic Spectrum of -Associated Distal Arthrogryposis. *Children*, **2021**, 8,	2.8	1
447	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**,	9.4	1
446	Disease monitoring programs of rare genetic diseases: transparent data sharing between academic and commercial stakeholders. *Orphanet Journal of Rare Diseases*, **2021**, 16, 141	4.2	3
445	Associations Between Variant Repeat Interruptions and Clinical Outcomes in Myotonic Dystrophy Type 1. *Neurology: Genetics*, **2021**, 7, e572	3.8	4
444	Results from a 3-year Non-interventional, Observational Disease Monitoring Program in Adults with GNE Myopathy. *Journal of Neuromuscular Diseases*, **2021**, 8, 225-234	5	1
443	Recessive VAMP1 mutations associated with severe congenital myasthenic syndromes - A recognizable clinical phenotype. *European Journal of Paediatric Neurology*, **2021**, 31, 54-60	3.8	3
442	INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. *Brain*, **2021**, 144, 2427-2442	11.2	2
441	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	5.3	3
440	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	5	2

439	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	5	1
438	Clinical Outcome Evaluations and CBT Response Prediction in Myotonic Dystrophy. *Journal of Neuromuscular Diseases*, **2021**, 8, 1031-1046	5	0
437	Expanding the clinical and molecular spectrum of ATP6V1A related metabolic cutis laxa. *Journal of Inherited Metabolic Disease*, **2021**, 44, 972-986	5.4	2
436	Molecular pathophysiology of human MICU1 deficiency. *Neuropathology and Applied Neurobiology*, **2021**, 47, 840-855	5.2	2
435	Costs of Illness of Spinal Muscular Atrophy: A Systematic Review. *Applied Health Economics and Health Policy*, **2021**, 19, 501-520	3.4	6
434	Dysregulation of GSK3 Target Proteins in Skin Fibroblasts of Myotonic Dystrophy Type 1 (DM1) Patients. *Journal of Neuromuscular Diseases*, **2021**, 8, 603-619	5	2
433	A founder mutation in the GMPPB gene [c.1000G > 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	3	2
432	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	6.4	2
431	A de novo CSDE1 variant causing neurodevelopmental delay, intellectual disability, neurologic and psychiatric symptoms in a child of consanguineous parents. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	0
430	Autosomal recessive variants in alter the tubulin ring complex leading to neurodevelopmental disease. *IScience*, **2021**, 24, 101948	6.1	1
429	Long Term Follow-Up on Pediatric Cases With Congenital Myasthenic Syndromes-A Retrospective Single Centre Cohort Study. *Frontiers in Human Neuroscience*, **2020**, 14, 560860	3.3	4
428	Economic Costs of Myasthenia Gravis: A Systematic Review. *Pharmacoeconomics*, **2020**, 38, 715-728	4.4	7
427	Blood-derived biomarkers correlate with clinical progression in Duchenne muscular dystrophy. *Journal of Neuromuscular Diseases*, **2020**, 7, 231-246	5	8
426	Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. *Lancet Neurology, The*, **2020**, 19, 522-532	24.1	21
425	A National Spinal Muscular Atrophy Registry for Real-World Evidence. *Canadian Journal of Neurological Sciences*, **2020**, 47, 810-815	1	3
424	An improved method for culturing myotubes on laminins for the robust clustering of postsynaptic machinery. *Scientific Reports*, **2020**, 10, 4524	4.9	5
423	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. *Journal of Neuromuscular Diseases*, **2020**, 7, 153-166	5	10
422	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. *Journal of Molecular Diagnostics*, **2020**, 22, 1205-1215	5.1	8

421	Change over time in ability to perform activities of daily living in myotonic dystrophy type 1. *Journal of Neurology*, **2020**, 267, 3235-3242	5.5	2
420	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. *Frontiers in Genetics*, **2020**, 11, 605	4.5	3
419	Life expectancy at birth in Duchenne muscular dystrophy: a systematic review and meta-analysis. *European Journal of Epidemiology*, **2020**, 35, 643-653	12.1	44
418	Severe congenital myasthenic syndrome associated with novel biallelic mutation of the CHRND gene. *Neuromuscular Disorders*, **2020**, 30, 336-339	2.9	1
417	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	10.3	37
416	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	3.2	8
415	Congenital myasthenic syndrome due to DOK7 mutation in a cohort of patients with NunexplainedN limb-girdle muscular weakness. *Journal of Clinical Neuroscience*, **2020**, 75, 195-198	2.2	0
414	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	5.3	4
413	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. *EMBO Journal*, **2020**, 39, e105364	13	15
412	Confirmation of TACO1 as a Leigh Syndrome Disease Gene in Two Additional Families. *Journal of Neuromuscular Diseases*, **2020**, 7, 301-308	5	3
411	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	6.1	1
410	Activities of daily living in myotonic dystrophy type 1. *Acta Neurologica Scandinavica*, **2020**, 141, 380-387	3.8	4
409	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	10.3	14
408	A novel, pathogenic dinucleotide deletion in the mitochondrial MT-TY gene causing myasthenia-like features. *Neuromuscular Disorders*, **2020**, 30, 661-668	2.9	5
407	Biallelic loss of function variants in SYT2 cause a treatable congenital onset presynaptic myasthenic syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2272-2283	2.5	8
406	Behr syndrome and hypertrophic cardiomyopathy in a family with a novel UCHL1 deletion. *Journal of Neurology*, **2020**, 267, 3643-3649	5.5	3
405	-related autosomal recessive encephalopathy in 2 Turkish children. *Neurology: Genetics*, **2020**, 6, e392	3.8	4
404	Loss of supervillin causes myopathy with myofibrillar disorganization and autophagic vacuoles. *Brain*, **2020**, 143, 2406-2420	11.2	7

403	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	4.2	8
402	A Phase 2 Study of AMO-02 (Tideglusib) in Congenital and Childhood-Onset Myotonic Dystrophy Type 1 (DM1). *Pediatric Neurology*, **2020**, 112, 84-93	2.9	16
401	Congenital myasthenic syndrome with mild intellectual disability caused by a recurrent SLC25A1 variant. *European Journal of Human Genetics*, **2020**, 28, 373-377	5.3	6
400	Severe neurodevelopmental disease caused by a homozygous TLK2 variant. *European Journal of Human Genetics*, **2020**, 28, 383-387	5.3	4
399	Clinical presentation and proteomic signature of patients with TANGO2 mutations. *Journal of Inherited Metabolic Disease*, **2020**, 43, 297-308	5.4	17
398	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	4.1	2
397	De novo variant in SCN4A causes neonatal sodium channel myotonia with general muscle stiffness and respiratory failure. *Neuromuscular Disorders*, **2019**, 29, 907-909	2.9	3
396	Safety and Treatment Effects of Nusinersen in Longstanding Adult 5q-SMA Type 3 - A Prospective Observational Study. *Journal of Neuromuscular Diseases*, **2019**, 6, 453-465	5	75
395	SMArtCARE□□A platform to collect real-life outcome data of patients with spinal muscular atrophy. *Orphanet Journal of Rare Diseases*, **2019**, 14, 18	4.2	33
394	A phase 3 randomized study evaluating sialic acid extended-release for GNE myopathy. *Neurology*, **2019**, 92, e2109-e2117	6.5	21
393	genotype explains 20% of phenotypic variability in GNE myopathy. *Neurology: Genetics*, **2019**, 5, e308	3.8	11
392	Correction of pseudoexon splicing caused by a novel intronic dysferlin mutation. *Annals of Clinical and Translational Neurology*, **2019**, 6, 642-654	5.3	12
391	Salbutamol modifies the neuromuscular junction in a mouse model of ColQ myasthenic syndrome. *Human Molecular Genetics*, **2019**, 28, 2339-2351	5.6	18
390	Dihydropyridine Receptor Congenital Myopathy In A Consangineous Turkish Family. *Journal of Neuromuscular Diseases*, **2019**, 6, 377-384	5	4
389	Increasing phenotypic annotation improves the diagnostic rate of exome sequencing in a rare neuromuscular disorder. *Human Mutation*, **2019**, 40, 1797-1812	4.7	9
388	"Be an ambassador for change that you would like to see": 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	4.2	5
387	Phenotype may predict the clinical course of facioscapolohumeral muscular dystrophy. *Muscle and Nerve*, **2019**, 59, 711-713	3.4	10
386	MACF1 links Rapsyn to microtubule- and actin-binding proteins to maintain neuromuscular synapses. *Journal of Cell Biology*, **2019**, 218, 1686-1705	7.3	22

385	Quality of life of patients with spinal muscular atrophy: A systematic review. *European Journal of Paediatric Neurology*, **2019**, 23, 347-356	3.8	27
384	CHRNG-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	2.5	3
383	Incomplete description of the current body of evidence of the health economics of Duchenne muscular dystrophy. *Orphanet Journal of Rare Diseases*, **2019**, 14, 75	4.2	
382	Health-Related Quality of Life in Patients with Adult-Onset Myotonic Dystrophy Type 1: A Systematic Review. *Patient*, **2019**, 12, 365-373	3.7	6
381	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	5	6
380	Targeted therapies for congenital myasthenic syndromes: systematic review and steps towards a treatabolome. *Emerging Topics in Life Sciences*, **2019**, 3, 19-37	3.5	25
379	Analyzing walking speeds with ankle and wrist worn accelerometers in a cohort with myotonic dystrophy. *Disability and Rehabilitation*, **2019**, 41, 2972-2978	2.4	8
378	Modulation of Agrin and RhoA Pathways Ameliorates Movement Defects and Synapse Morphology in MYO9A-Depleted Zebrafish. *Cells*, **2019**, 8,	7.9	3
377	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	4.2	12
376	Analysis of the functional capacity outcome measures for myotonic dystrophy. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1487-1497	5.3	4
375	Disease burden of myotonic dystrophy type 1. *Journal of Neurology*, **2019**, 266, 998-1006	5.5	16
374	237th ENMC International Workshop: GNE myopathy - current and future research Hoofddorp, The Netherlands, 14-16 September 2018. *Neuromuscular Disorders*, **2019**, 29, 401-410	2.9	2
373	Congenital myasthenic syndrome caused by novel COL13A1 mutations. *Journal of Neurology*, **2019**, 266, 1107-1112	5.5	12
372	Genetic determinants of disease severity in the myotonic dystrophy type 1 OPTIMISTIC cohort. *Neurology*, **2019**, 93, e995-e1009	6.5	43
371	A Review of International Biobanks and Networks: Success Factors and Key Benchmarks-A 10-Year Retrospective Review. *Biopreservation and Biobanking*, **2019**, 17, 512-519	2.1	5
370	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. *Nucleic Acids Research*, **2019**, 47, D1018-D1027	20.1	333
369	SIL1 deficiency causes degenerative changes of peripheral nerves and neuromuscular junctions in fish, mice and human. *Neurobiology of Disease*, **2019**, 124, 218-229	7.5	7
368	Assessment of disease progression in dysferlinopathy: A 1-year cohort study. *Neurology*, **2019**,	6.5	11

367	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	2.9	15
366	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	2.4	7
365	The beta-adrenergic agonist salbutamol modulates neuromuscular junction formation in zebrafish models of human myasthenic syndromes. *Human Molecular Genetics*, **2018**, 27, 1556-1564	5.6	16
364	MYO9A deficiency in motor neurons is associated with reduced neuromuscular agrin secretion. *Human Molecular Genetics*, **2018**, 27, 1434-1446	5.6	8
363	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	3.4	6
362	RD-Connect, NeurOmics and EURenOmics: collaborative European initiative for rare diseases. *European Journal of Human Genetics*, **2018**, 26, 778-785	5.3	39
361	Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophy-like disease. *Genetics in Medicine*, **2018**, 20, 1224-1235	8.1	18
360	Tracking disease progression non-invasively in Duchenne and Becker muscular dystrophies. *Journal of Cachexia, Sarcopenia and Muscle*, **2018**, 9, 715-726	10.3	32
359	Mutations in glycyl-tRNA synthetase impair mitochondrial metabolism in neurons. *Human Molecular Genetics*, **2018**, 27, 2187-2204	5.6	15
358	Mobility shift of beta-dystroglycan as a marker of gene-related muscular dystrophy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 762-768	5.5	13
357	A common CHRNE mutation in Brazilian patients with congenital myasthenic syndrome. *Journal of Neurology*, **2018**, 265, 708-713	5.5	9
356	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	5.3	27
355	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. *Human Molecular Genetics*, **2018**, 27, 1186-1195	5.6	40
354	Phenotypic stratification and genotype-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	2.9	20
353	Clinical and research strategies for limb-girdle congenital myasthenic syndromes. *Annals of the New York Academy of Sciences*, **2018**, 1412, 102-112	6.5	9
352	Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo mutation. *Neurology*, **2018**, 90, e1842-e1848	6.5	3
351	How to Spot Congenital Myasthenic Syndromes Resembling the Lambert-Eaton Myasthenic Syndrome? A Brief Review of Clinical, Electrophysiological, and Genetics Features. *NeuroMolecular Medicine*, **2018**, 20, 205-214	4.6	3
350	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	2.5	17

349	Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 1224-1226	5.5	14
348	Whole-exome sequencing identifies mutations in in a mild form of Carey-Fineman-Ziter syndrome. *Neurology: Genetics*, **2018**, 4, e226	3.8	4
347	Compliance to care guidelines for Duchenne muscular dystrophy in Italy. *Neuromuscular Disorders*, **2018**, 28, 100	2.9	2
346	Mass spectrometry-based protein analysis to unravel the tissue pathophysiology in Duchenne muscular dystrophy. *Proteomics - Clinical Applications*, **2018**, 12, 1700071	3.1	17
345	Benign and malignant tumors in the UK myotonic dystrophy patient registry. *Muscle and Nerve*, **2018**, 57, 316-320	3.4	11
344	Chronic pain has a strong impact on quality of life in facioscapulohumeral muscular dystrophy. *Muscle and Nerve*, **2018**, 57, 380-387	3.4	17
343	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	2.9	9
342	Intersection of Proteomics and Genomics to "Solve the Unsolved" in Rare Disorders such as Neurodegenerative and Neuromuscular Diseases. *Proteomics - Clinical Applications*, **2018**, 12, 1700073	3.1	19
341	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	5	9
340	A checklist for clinical trials in rare disease: obstacles and anticipatory actions-lessons learned from the FOR-DMD trial. *Trials*, **2018**, 19, 291	2.8	17
339	Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 1071-1081	5.5	43
338	GNE myopathy: from clinics and genetics to pathology and research strategies. *Orphanet Journal of Rare Diseases*, **2018**, 13, 70	4.2	18
337	Recommendations for Improving the Quality of Rare Disease Registries. *International Journal of Environmental Research and Public Health*, **2018**, 15,	4.6	56
336	Duchenne muscular dystrophy and caregiver burden: a systematic review. *Developmental Medicine and Child Neurology*, **2018**, 60, 987-996	3.3	31
335	GFPT1 deficiency in muscle leads to myasthenia and myopathy in mice. *Human Molecular Genetics*, **2018**, 27, 3218-3232	5.6	10
334	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	2.9	9
333	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	24.1	62
332	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	5.5	21

331	Risk of skin cancer among patients with myotonic dystrophy type 1 based on primary care physician data from the U.K. Clinical Practice Research Datalink. *International Journal of Cancer*, **2018**, 142, 1174-1181	7.5	19
330	Progress in Rare Diseases Research 2010-2016: An IRDiRC Perspective. *Clinical and Translational Science*, **2018**, 11, 11-20	4.9	63
329	Future of Rare Diseases Research 2017-2027: An IRDiRC Perspective. *Clinical and Translational Science*, **2018**, 11, 21-27	4.9	86
328	Neuromuscular Junction Changes in a Mouse Model of Charcot-Marie-Tooth Disease Type 4C. *International Journal of Molecular Sciences*, **2018**, 19,	6.3	11
327	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	4.2	10
326	Clinical variability of early-onset congenital myasthenic syndrome due to biallelic RAPSN mutations in Brazil. *Neuromuscular Disorders*, **2018**, 28, 961-964	2.9	6
325	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	10.7	24
324	GNE myopathy in the bedouin population of Kuwait: Genetics, prevalence, and clinical description. *Muscle and Nerve*, **2018**, 58, 700-707	3.4	3
323	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	4.2	12
322	Biochemical and pathological changes result from mutated Caveolin-3 in muscle. *Skeletal Muscle*, **2018**, 8, 28	5.1	12
321	The Human Phenotype Ontology in 2017. *Nucleic Acids Research*, **2017**, 45, D865-D876	20.1	507
320	Reduced serum myostatin concentrations associated with genetic muscle disease progression. *Journal of Neurology*, **2017**, 264, 541-553	5.5	34
319	IRDiRC Recognized Resources: a new mechanism to support scientists to conduct efficient, high-quality research for rare diseases. *European Journal of Human Genetics*, **2017**, 25, 162-165	5.3	26
318	Predictors of Health-Related Quality of Life in boys with Duchenne muscular dystrophy from six European countries. *Journal of Neurology*, **2017**, 264, 709-723	5.5	15
317	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	5.5	10
316	Genetic heterogeneity of motor neuropathies. *Neurology*, **2017**, 88, 1226-1234	6.5	54
315	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	11	40
314	Intragenic deletion detected by whole-genome sequencing in congenital myasthenic syndromes. *Neurology: Genetics*, **2017**, 3, e152	3.8	3

313	Critical points for an accurate human genome analysis. *Human Mutation*, **2017**, 38, 912-921	4.7	5
312	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. *American Journal of Human Genetics*, **2017**, 100, 695-705	11	200
311	The UK Myotonic Dystrophy Patient Registry: facilitating and accelerating clinical research. *Journal of Neurology*, **2017**, 264, 979-988	5.5	18
310	SPARC Interacts with Actin in Skeletal Muscle in Vitro and in Vivo. *American Journal of Pathology*, **2017**, 187, 457-474	5.8	18
309	Complex phenotypes associated with STIM1 mutations in both coiled coil and EF-hand domains. *Neuromuscular Disorders*, **2017**, 27, 861-872	2.9	23
308	The Increasing Genetic and Phenotypical Diversity of Congenital Myasthenic Syndromes. *Neuropediatrics*, **2017**, 48, 294-308	1.6	32
307	Respiratory involvement in ambulant and non-ambulant patients with facioscapulohumeral muscular dystrophy. *Journal of Neurology*, **2017**, 264, 1271-1280	5.5	24
306	Facilitating orphan drug development: Proceedings of the TREAT-NMD International Conference, December 2015, Washington, DC, USA. *Neuromuscular Disorders*, **2017**, 27, 693-701	2.9	1
305	A novel recessive TTN founder variant is a common cause of distal myopathy in the Serbian population. *European Journal of Human Genetics*, **2017**, 25, 572-581	5.3	14
304	Linked Registries: Connecting Rare Diseases Patient Registries through a Semantic Web Layer. *BioMed Research International*, **2017**, 2017, 8327980	3	13
303	Drosophila studies support a role for a presynaptic synaptotagmin mutation in a human congenital myasthenic syndrome. *PLoS ONE*, **2017**, 12, e0184817	3.7	10
302	Prevalence, incidence and carrier frequency of 5q-linked spinal muscular atrophy - a literature review. *Orphanet Journal of Rare Diseases*, **2017**, 12, 124	4.2	201
301	Exome sequences versus sequential gene testing in the UK highly specialised Service for Limb Girdle Muscular Dystrophy. *Orphanet Journal of Rare Diseases*, **2017**, 12, 151	4.2	27
300	Translocation of molecular chaperones to the titin springs is common in skeletal myopathy patients and affects sarcomere function. *Acta Neuropathologica Communications*, **2017**, 5, 72	7.3	18
299	Mortality Cost of Duchenne Muscular Dystrophy. *Global & Regional Health Technology Assessment*, **2017**, 4, grhta.5000260	0.2	0
298	MRC Centre Neuromuscular Biobank (Newcastle and London): Supporting and facilitating rare and neuromuscular disease research worldwide. *Neuromuscular Disorders*, **2017**, 27, 1054-1064	2.9	9
297	Co-presentation of adult-onset systemic lupus erythematosus and nemaline myopathy. *Rheumatology*, **2017**, 56, 2034-2035	3.9	3
296	A second truncation in TTN causes early onset recessive muscular dystrophy. *Neuromuscular Disorders*, **2017**, 27, 1009-1017	2.9	9

295	A comparative study of care practices for young boys with Duchenne muscular dystrophy between Japan and European countries: Implications of early diagnosis. *Neuromuscular Disorders*, **2017**, 27, 894-904	2.9	4
294	The International Rare Diseases Research Consortium: Policies and Guidelines to maximize impact. *European Journal of Human Genetics*, **2017**, 25, 1293-1302	5.3	45
293	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. *Advances in Experimental Medicine and Biology*, **2017**, 1031, 55-94	3.6	13
292	Natural History, Trial Readiness and Gene Discovery: Advances in Patient Registries for Neuromuscular Disease. *Advances in Experimental Medicine and Biology*, **2017**, 1031, 97-124	3.6	13
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290	Patient Preferences for Treatments of Neuromuscular Diseases: A Systematic Literature Review. *Journal of Neuromuscular Diseases*, **2017**, 4, 285-292	5	3
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