Kathryn N North

List of Publications by Year
in descending order

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412 papers	26,166 citations	5126 86 h-index	11282 141 g-index
439 all docs	439 docs citations	439 times ranked	24320 citing authors

#	Article	IF	Citations
1	Sex- and age-related differences in autistic behaviours in children with neurofibromatosis type 1. Journal of Autism and Developmental Disorders, 2023, 53, 2835-2850.	1.7	2
2	The MMAAS Project: An Observational Human Study Investigating the Effect of Anabolic Androgenic Steroid Use on Gene Expression and the Molecular Mechanism of Muscle Memory. Clinical Journal of Sport Medicine, 2023, 33, e115-e122.	0.9	2
3	The mediating role of ADHD symptoms between executive function and social skills in children with neurofibromatosis type 1. Child Neuropsychology, 2022, 28, 318-336.	0.8	2
4	Loss of Î±-actinin-3 confers protection from eccentric contraction damage in fast-twitch EDL muscles from aged mdx dystrophic mice by reducing pathological fibre branching. Human Molecular Genetics, 2022, 31, 1417-1429.	1.4	2
5	Delineating the autistic phenotype in children with neurofibromatosis type 1. Molecular Autism, 2022, 13, 3.	2.6	8
6	Response to MÃ¶rseburg etÂal.. American Journal of Human Genetics, 2022, 109, 973.	2.6	2
7	Absence of the Z-disc protein Î±-actinin-3 impairs the mechanical stability of Actn3KO mouse fast-twitch muscle fibres without altering their contractile properties or twitch kinetics. Skeletal Muscle, 2022, 12, .	1.9	3
8	Autism in neurofibromatosis type 1: misuse of covariance to dismiss autistic trait burden. Developmental Medicine and Child Neurology, 2021, 63, 233-234.	1.1	4
9	Cognition, ADHD Symptoms, and Functional Impairment in Children and Adolescents With Neurofibromatosis Type 1. Journal of Attention Disorders, 2021, 25, 1177-1186.	1.5	32
10	Loss of Î±-actinin-3 during human evolution provides superior cold resilience and muscle heat generation. American Journal of Human Genetics, 2021, 108, 446-457.	2.6	32
11	ACTN3 genotype influences skeletal muscle mass regulation and response to dexamethasone. Science Advances, 2021, 7, .	4.7	7
12	Generating an iPSC line (with isogenic control) from the PBMCs of an ACTA1 (p.Gly148Asp) nemaline myopathy patient. Stem Cell Research, 2021, 54, 102429.	0.3	3
13	Dystrophin-negative slow-twitch soleus muscles are not susceptible to eccentric contraction induced injury over the lifespan of the mdx mouse. American Journal of Physiology - Cell Physiology, 2021, 321, C704-C720.	2.1	11
14	Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution. American Journal of Human Genetics, 2021, 108, 1551-1557.	2.6	36
15	Profiling the Word Reading Abilities of School-Age Children with Neurofibromatosis Type 1. Journal of the International Neuropsychological Society, 2021, 27, 484-496.	1.2	7
16	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	3.0	94
17	Auditory Dysfunction Among Individuals With Neurofibromatosis Type 1. JAMA Network Open, 2021, 4, e2136842.	2.8	3
18	Lifespan Analysis of Dystrophic mdx Fast-Twitch Muscle Morphology and Its Impact on Contractile Function. Frontiers in Physiology, 2021, 12, 771499.	1.3	9

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19	A brief history of human disease genetics. Nature, 2020, 577, 179-189.	13.7	441
20	TCTEX1D1 is a genetic modifier of disease progression in Duchenne muscular dystrophy. European Journal of Human Genetics, 2020, 28, 815-825.	1.4	36
21	LATE BREAKING NEWS E-POSTER PRESENTATION. Neuromuscular Disorders, 2020, 30, S170.	0.3	0
22	Isolated Extensor Digitorum Longus Muscles from Old mdx Dystrophic Mice Show Little Force Recovery 120 Minutes after Eccentric Damage. Biophysical Journal, 2020, 118, 121a.	0.2	0
23	Generation of four iPSC lines from Neurofibromatosis Type 1 patients. Stem Cell Research, 2020, 49, 102013.	0.3	1
24	Eosinophil function in adipose tissue is regulated by KrÃ¼ppel-like factor 3 (KLF3). Nature Communications, 2020, 11, 2922.	5.8	35
25	Social skills and autism spectrum disorder symptoms in children with neurofibromatosis type 1: evidence for clinical trial outcomes. Developmental Medicine and Child Neurology, 2020, 62, 813-819.	1.1	13
26	Visual spatial learning outcomes for clinical trials in neurofibromatosis type 1. Annals of Clinical and Translational Neurology, 2020, 7, 245-249.	1.7	9
27	Attention to faces in social context in children with neurofibromatosis type 1. Developmental Medicine and Child Neurology, 2019, 61, 174-180.	1.1	11
28	Australian Genomics: A Federated Model for Integrating Genomics into Healthcare. American Journal of Human Genetics, 2019, 105, 7-14.	2.6	75
29	A â€œhuman knockoutâ€•model to investigate the influence of the Î±-actinin-3 protein on exercise-induced mitochondrial adaptations. Scientific Reports, 2019, 9, 12688.	1.6	13
30	Building a learning community of Australian clinical genomics: a social network study of the Australian Genomic Health Alliance. BMC Medicine, 2019, 17, 44.	2.3	22
31	A transformative translational change programme to introduce genomics into healthcare: a complexity and implementation science study protocol. BMJ Open, 2019, 9, e024681.	0.8	21
32	Understanding autism spectrum disorder and social functioning in children with neurofibromatosis type 1: protocol for a cross-sectional multimodal study. BMJ Open, 2019, 9, e030601.	0.8	11
33	Reproducibility of cognitive endpoints in clinical trials: lessons from neurofibromatosis type 1. Annals of Clinical and Translational Neurology, 2019, 6, 2555-2565.	1.7	24
34	Integrating Genomics into Healthcare: A Global Responsibility. American Journal of Human Genetics, 2019, 104, 13-20.	2.6	264
35	Preliteracy impairments in children with neurofibromatosis type 1. Developmental Medicine and Child Neurology, 2018, 60, 703-710.	1.1	15
36	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	2.8	93

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37	Monitoring of optic nerve function in Neurofibromatosis 2 children with optic nerve sheath meningiomas using multifocal visual evoked potentials. Journal of Clinical Neuroscience, 2018, 50, 262-267.	0.8	5
38	Nemaline myopathy and distal arthrogryposis associated with an autosomal recessiveÂTNNT3 Âsplice variant. Human Mutation, 2018, 39, 383-388.	1.1	48
39	The Effect of ACTN3 Gene Doping on Skeletal Muscle Performance. American Journal of Human Genetics, 2018, 102, 845-857.	2.6	17
40	Impaired engagement of the ventral attention system in neurofibromatosis type 1. Brain Imaging and Behavior, 2018, 12, 499-508.	1.1	12
41	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	1.5	148
42	Is evolutionary loss our gain? The role of ACTN3 p.Arg577Ter (R577X) genotype in athletic performance, ageing, and disease. Human Mutation, 2018, 39, 1774-1787.	1.1	50
43	Branched fibers from old fast-twitch dystrophic muscles are the sites of terminal damage in muscular dystrophy. American Journal of Physiology - Cell Physiology, 2018, 314, C662-C674.	2.1	23
44	Effects of methylphenidate on cognition and behaviour in children with neurofibromatosis type 1: a study protocol for a randomised placebo-controlled crossover trial. BMJ Open, 2018, 8, e021800.	0.8	12
45	No association between ACTN3 R577X and ACE I/D polymorphisms and endurance running times in 698 Caucasian athletes. BMC Genomics, 2018, 19, 13.	1.2	65
46	Social Function and Autism Spectrum Disorder in Children and Adults with Neurofibromatosis Type 1: a Systematic Review and Meta-Analysis. Neuropsychology Review, 2018, 28, 317-340.	2.5	45
47	The relationship between deficit in digit span and genotype in nonsense mutation Duchenne muscular dystrophy. Neurology, 2018, 91, e1215-e1219.	1.5	12
48	Evidence for ACTN3 as a genetic modifier of Duchenne muscular dystrophy. Nature Communications, 2017, 8, 14143.	5.8	58
49	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	5.8	516
50	Can in-the-moment diary methods measure health-related quality of life in Duchenne muscular dystrophy?. Quality of Life Research, 2017, 26, 1145-1152.	1.5	8
51	Atypical Local Interference Affects Global Processing in Children with Neurofibromatosis Type 1. Journal of the International Neuropsychological Society, 2017, 23, 446-450.	1.2	3
52	The neural basis of deficient response inhibition in children with neurofibromatosis type 1: Evidence from a functional MRI study. Cortex, 2017, 93, 1-11.	1.1	14
53	Exploring the relationship between Î±-actinin-3 deficiency and obesity in mice and humans. International Journal of Obesity, 2017, 41, 1154-1157.	1.6	9
54	Clinical characterisation of a large international congenital titinopathy cohort. Neuromuscular Disorders, 2017, 27, S37.	0.3	0

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55	Androgen Action via the Androgen Receptor in Neurons Within the Brain Positively Regulates Muscle Mass in Male Mice. Endocrinology, 2017, 158, 3684-3695.	1.4	26
56	Do titin developmental isoforms contribute to the pathogenesis of congenital titinopathy?. Neuromuscular Disorders, 2017, 27, S237-S238.	0.3	1
57	Large-scale GWAS identifies multiple loci for hand grip strength providing biological insights into muscular fitness. Nature Communications, 2017, 8, 16015.	5.8	149
58	Cost-effectiveness of massively parallel sequencing for diagnosis of paediatric muscle diseases. Npj Genomic Medicine, 2017, 2, .	1.7	67
59	Neurofibromatosis Type 2. Journal of Child Neurology, 2017, 32, 9-22.	0.7	75
60	The kids are OK: it is discrimination not sameâ€sex parents that harms children. Medical Journal of Australia, 2017, 207, 374-375.	0.8	13
61	Facial emotion recognition, face scan paths, and face perception in children with neurofibromatosis type 1.. Neuropsychology, 2017, 31, 361-370.	1.0	13
62	Social Competence in Children with Neurofibromatosis Type 1: Relationships with Psychopathology and Cognitive Ability. Journal of Childhood & Developmental Disorders, 2016, 02, .	0.3	7
63	No Evidence of a Common DNA Variant Profile Specific to World Class Endurance Athletes. PLoS ONE, 2016, 11, e0147330.	1.1	96
64	Uptake of health monitoring and disease selfâ€management in Australian adults with neurofibromatosis type 1: strategies to improve care. Clinical Genetics, 2016, 89, 385-391.	1.0	6
65	The Effect of Heterozygosity for the ACTN3 Null Allele on Human Muscle Performance. Medicine and Science in Sports and Exercise, 2016, 48, 509-520.	0.2	14
66	Targeted Re-Sequencing Emulsion PCR Panel for Myopathies: Results in 94 Cases. Journal of Neuromuscular Diseases, 2016, 3, 209-225.	1.1	18
67	TOR1AIP1 as a cause of cardiac failure and recessive limb-girdle muscular dystrophy. Neuromuscular Disorders, 2016, 26, 500-503.	0.3	38
68	ACTN3 R577X and ACE I/D gene variants influence performance in elite sprinters: a multi-cohort study. BMC Genomics, 2016, 17, 285.	1.2	106
69	Patient-reported outcomes of pain and physical functioning in neurofibromatosis clinical trials. Neurology, 2016, 87, S4-S12.	1.5	36
70	Phonics Training Improves Reading in Children with Neurofibromatosis Type 1: A Prospective Intervention Trial. Journal of Pediatrics, 2016, 177, 219-226.e2.	0.9	10
71	Neurocognitive outcomes in neurofibromatosis clinical trials. Neurology, 2016, 87, S21-30.	1.5	16
72	Variants in SLC18A3 , vesicular acetylcholine transporter, cause congenital myasthenic syndrome. Neurology, 2016, 87, 1442-1448.	1.5	46

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73	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. American Journal of Human Genetics, 2016, 99, 1086-1105.	2.6	45
74	Analysis of a large international cohort confirms that recessively inherited loss-of-function TTN mutations cause prenatal or infant-onset muscle disease, often complicated by early cardiorespiratory involvement. Neuromuscular Disorders, 2016, 26, S89.	0.3	0
75	Current status and recommendations for biomarkers and biobanking in neurofibromatosis. Neurology, 2016, 87, S40-8.	1.5	23
76	Sleep and pulmonary outcomes for clinical trials of airway plexiform neurofibromas in NF1. Neurology, 2016, 87, S13-20.	1.5	15
77	Current whole-body MRI applications in the neurofibromatoses. Neurology, 2016, 87, S31-9.	1.5	65
78	Randomized placebo-controlled study of lovastatin in children with neurofibromatosis type 1. Neurology, 2016, 87, 2575-2584.	1.5	76
79	Disease Burden and Symptom Structure of Autism in Neurofibromatosis Type 1. JAMA Psychiatry, 2016, 73, 1276.	6.0	90
80	Theory of mind in children with Neurofibromatosis Type 1.. Neuropsychology, 2016, 30, 439-448.	1.0	27
81	A federated ecosystem for sharing genomic, clinical data. Science, 2016, 352, 1278-1280.	6.0	175
82	The Future of Genomic Research in Athletic Performance and Adaptation to Training. Medicine and Sport Science, 2016, 61, 55-67.	1.4	35
83	Diagnosis and etiology of congenital muscular dystrophy: We are halfway there. Annals of Neurology, 2016, 80, 101-111.	2.8	57
84	Lack of MG53 in human heart precludes utility as a biomarker of myocardial injury or endogenous cardioprotective factor. Cardiovascular Research, 2016, 110, 178-187.	1.8	46
85	How does Î±-actinin-3 deficiency alter muscle function? Mechanistic insights into ACTN3 , the â€˜gene for speedâ€™. Biochimica Et Biophysica Acta - Molecular Cell Research, 2016, 1863, 686-693.	1.9	57
86	Prominent scapulae mimicking an inherited myopathy expands the phenotype of CHD7-related disease. European Journal of Human Genetics, 2016, 24, 1216-1219.	1.4	2
87	Rodent models for resolving extremes of exercise and health. Physiological Genomics, 2016, 48, 82-92.	1.0	20
88	Athlome Project Consortium: a concerted effort to discover genomic and other â€œomicâ€•markers of athletic performance. Physiological Genomics, 2016, 48, 183-190.	1.0	96
89	Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. Neurology, 2016, 86, 391-398.	1.5	107
90	Analysis of theACTN3heterozygous genotype suggests that Î±-actinin-3 controls sarcomeric composition and muscle function in a dose-dependent fashion. Human Molecular Genetics, 2016, 25, 866-877.	1.4	35

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91	TREAT-NMD (translational research in Europe, assessment and treatment for neuromuscular) Tj ETQq1 1 0.784314 rgBT /Overlock 10	0.3	0
92	Associate Professor Nigel Clarke PhD, MBChB, FRACP (1966â€“2015). Neuromuscular Disorders, 2015, 25, 977-978.	0.3	0
93	Muscle weakness in children with neurofibromatosis type 1. Developmental Medicine and Child Neurology, 2015, 57, 733-736.	1.1	21
94	Direct-to-consumer genetic testing for predicting sports performance and talent identification: Consensus statement. British Journal of Sports Medicine, 2015, 49, 1486-1491.	3.1	113
95	Recessive ACTA1 variant causes congenital muscular dystrophy with rigid spine. European Journal of Human Genetics, 2015, 23, 883-886.	1.4	23
96	Altered Ca2+ Kinetics Associated with Î±-Actinin-3 Deficiency May Explain Positive Selection for ACTN3 Null Allele in Human Evolution. PLoS Genetics, 2015, 11, e1004862.	1.5	39
97	Evidence-based guideline summary: Evaluation, diagnosis, and management of congenital muscular dystrophy. Neurology, 2015, 84, 1369-1378.	1.5	88
98	Congenital and Other Structural Myopathies. , 2015, , 499-537.		3
99	The Impact of Neurofibromatosis Type 1 on the Health and Wellbeing of Australian Adults. Journal of Genetic Counseling, 2015, 24, 931-944.	0.9	33
100	Expanding the phenotype of GMPPB mutations. Brain, 2015, 138, 836-844.	3.7	54
101	Developmental Trajectories of Young Children with Neurofibromatosis Type 1: A Longitudinal Study from 21 to 40ÂMonths of Age. Journal of Pediatrics, 2015, 166, 1006-1012.e1.	0.9	18
102	All the World's a Stage: Facilitating Discovery Science and Improved Cancer Care through the Global Alliance for Genomics and Health. Cancer Discovery, 2015, 5, 1133-1136.	7.7	45
103	Reply: The p.Ser107Leu inBICD2is a mutation â€˜hot spotâ€™ causing distal spinal muscular atrophy. Brain, 2015, 138, e392-e392.	3.7	1
104	Evidence-based guideline summary: Evaluation, diagnosis, and management of congenital muscular dystrophy: Report of the Guideline Development Subcommittee of the American Academy of Neurology and the Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine. Neurology, 2015, 85, 1432-1433.	1.5	3
105	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. JAMA Neurology, 2015, 72, 1424.	4.5	164
106	Mutations inPIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. Human Molecular Genetics, 2015, 24, 6146-6159.	1.4	64
107	Muscle weakness inTPM3-myopathy is due to reduced Ca²⁺-sensitivity and impaired acto-myosin cross-bridge cycling in slow fibres. Human Molecular Genetics, 2015, 24, 6278-6292.	1.4	38
108	A gene for speed: The influence of ACTN3 on muscle performance in health and disease. Neuromuscular Disorders, 2015, 25, S185.	0.3	0

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109	Mutated HSPB8 causes both neurogenic and myopathic disease with muscle proteinopathy. Neuromuscular Disorders, 2015, 25, S256.	0.3	0
110	Phenotypic and molecular insights into spinal muscular atrophy due to mutations in BICD2. Brain, 2015, 138, 293-310.	3.7	82
111	Results of a two-year pilot study of clinical outcome measures in collagen VI- and laminin alpha2-related congenital muscular dystrophies. Neuromuscular Disorders, 2015, 25, 43-54.	0.3	30
112	LMOD3: the â€œmissing linkâ€•in nemaline myopathy?. Oncotarget, 2015, 6, 26548-26549.	0.8	11
113	Î±â€•actininâ€3 deficiency is associated with increased exercise performance and trainingâ€•induced changes in mitochondrial respiration in humans. FASEB Journal, 2015, 29, 677.27.	0.2	0
114	NF1 is a critical regulator of muscle development and metabolism. Human Molecular Genetics, 2014, 23, 1250-1259.	1.4	40
115	Activating internal ribosome entry to treat Duchenne muscular dystrophy. Nature Medicine, 2014, 20, 987-988.	15.2	3
116	Relationship between cognitive dysfunction, gait, and motor impairment in children and adolescents with neurofibromatosis type 1. Developmental Medicine and Child Neurology, 2014, 56, 468-474.	1.1	39
117	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	1.0	357
118	Mutation Update: The Spectra of Nebulin Variants and Associated Myopathies. Human Mutation, 2014, 35, 1418-1426.	1.1	107
119	The genetic and neuroanatomical basis of social dysfunction: Lessons from neurofibromatosis type 1. Human Brain Mapping, 2014, 35, 2372-2382.	1.9	30
120	Longitudinal assessment of cognition and T2â€•hyperintensities in NF1: An 18â€•year study. American Journal of Medical Genetics, Part A, 2014, 164, 661-665.	0.7	41
121	Î±-Actinin-3 deficiency alters muscle adaptation in response to denervation and immobilization. Human Molecular Genetics, 2014, 23, 1879-1893.	1.4	26
122	Sequence analysis of the equine ACTN3 gene in Australian horse breeds. Gene, 2014, 538, 88-93.	1.0	12
123	Mutation Update and Genotype-Phenotype Correlations of Novel and Previously Described Mutations inTPM2andTPM3Causing Congenital Myopathies. Human Mutation, 2014, 35, 779-790.	1.1	92
124	G.O.2. Neuromuscular Disorders, 2014, 24, 792-793.	0.3	1
125	G.P.219. Neuromuscular Disorders, 2014, 24, 882-883.	0.3	0
126	G.P.35. Neuromuscular Disorders, 2014, 24, 805.	0.3	1

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127	G.P.271. Neuromuscular Disorders, 2014, 24, 898.	0.3	0
128	Calpain cleavage within dysferlin exon 40a releases a synaptotagmin-like module for membrane repair. Molecular Biology of the Cell, 2014, 25, 3037-3048.	0.9	62
129	Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders, 2014, 24, 97-116.	0.3	239
130	Diagnostic approach to the congenital muscular dystrophies. Neuromuscular Disorders, 2014, 24, 289-311.	0.3	275
131	Genetics and sport performance: current challenges and directions to the future. Revista Brasileira De EducaÃ§Ã£o FÃsica E Esporte: RBEFE, 2014, 28, 177-193.	0.1	28
132	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708.	3.9	153
133	Evidence Based Selection of Commonly Used RT-qPCR Reference Genes for the Analysis of Mouse Skeletal Muscle. PLoS ONE, 2014, 9, e88653.	1.1	69
134	A novel mutation expands the genetic and clinical spectrum of MYH7-related myopathies. Neuromuscular Disorders, 2013, 23, 432-436.	0.3	35
135	Calpains, Cleaved Mini-Dysferlin_{C72}, and L-Type Channels Underpin Calcium-Dependent Muscle Membrane Repair. Journal of Neuroscience, 2013, 33, 5085-5094.	1.7	93
136	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 1108-1117.	2.6	147
137	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 6-18.	2.6	186
138	Social functioning in adults with neurofibromatosis type 1. Research in Developmental Disabilities, 2013, 34, 3393-3399.	1.2	29
139	Paired associate learning in children with neurofibromatosis type 1: implications for clinical trials. Journal of Neurology, 2013, 260, 214-220.	1.8	26
140	Corrigendum to â€œThe Learning Disabilities Network (LeaDNet): Using Neurofibromatosis Type 1 [NF1] as a Paradigm for Translational Researchâ€• , 2013, 161, 236-236.		0
141	P.1.2 Natural history of pulmonary function in collagen VI-related myopathies: An international study. Neuromuscular Disorders, 2013, 23, 741-742.	0.3	0
142	Mutations in GDP-Mannose Pyrophosphorylase B Cause Congenital and Limb-Girdle Muscular Dystrophies Associated with Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 93, 29-41.	2.6	197
143	Mutations in BICD2 Cause Dominant Congenital Spinal Muscular Atrophy and Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 92, 965-973.	2.6	156
144	P.10.21 Next-generation sequencing meets genetic diagnostics: Development of a comprehensive workflow for neuromuscular disorders. Neuromuscular Disorders, 2013, 23, 795.	0.3	0

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145	O.10 Mutations in a new dynein/dynactin adaptor gene cause Dominant Congenital Spinal Muscular Atrophy (DCSMA) and Hereditary Spastic Paraplegia (HSP). Neuromuscular Disorders, 2013, 23, 798.	0.3	0
146	O.18 Systematic identification of causal mutations in Mendelian disorders using exome sequence data. Neuromuscular Disorders, 2013, 23, 850.	0.3	0
147	Cognitive Features that Distinguish Preschool-Age Children with Neurofibromatosis Type 1 from Their Peers: A Matched Case-Control Study. Journal of Pediatrics, 2013, 163, 1479-1483.e1.	0.9	26
148	P.9.4. Neuromuscular Disorders, 2013, 23, 784.	0.3	0
149	Genes for Elite Power and Sprint Performance: ACTN3 Leads the Way. Sports Medicine, 2013, 43, 803-817.	3.1	158
150	Natural history of pulmonary function in collagen VI-related myopathies. Brain, 2013, 136, 3625-3633.	3.7	85
151	K7del is a common TPM2 gene mutation associated with nemaline myopathy and raised myofibre calcium sensitivity. Brain, 2013, 136, 494-507.	3.7	42
152	ACTN3 genotype influences muscle performance through the regulation of calcineurin signaling. Journal of Clinical Investigation, 2013, 123, 4255-4263.	3.9	113
153	Young Australian adults with NF1 have poor access to health care, high complication rates, and limited disease knowledge. American Journal of Medical Genetics, Part A, 2013, 161, 659-666.	0.7	26
154	Association Analysis of ACE and ACTN3 in Elite Caucasian and East Asian Swimmers. Medicine and Science in Sports and Exercise, 2013, 45, 892-900.	0.2	80
155	ACTN3 Allele Frequency in Humans Covaries with Global Latitudinal Gradient. PLoS ONE, 2013, 8, e52282.	1.1	29
156	A procedure for the computerized analysis of cleft palate speech transcription. Clinical Linguistics and Phonetics, 2012, 26, 18-38.	0.5	0
157	Importance and challenge of making an early diagnosis in LMNA -related muscular dystrophy. Neurology, 2012, 78, 1258-1263.	1.5	41
158	Autosomal dominant congenital spinal muscular atrophy: a true form of spinal muscular atrophy caused by early loss of anterior horn cells. Brain, 2012, 135, 1714-1723.	3.7	21
159	Does attentionâ€deficitâ€“hyperactivity disorder exacerbate executive dysfunction in children with neurofibromatosis type 1?. Developmental Medicine and Child Neurology, 2012, 54, 898-904.	1.1	43
160	Consensus Statement on Standard of Care for Congenital Myopathies. Journal of Child Neurology, 2012, 27, 363-382.	0.7	147
161	Parent-of-origin in individuals with familial neurofibromatosis type 1 and optic pathway gliomas. Familial Cancer, 2012, 11, 653-656.	0.9	7
162	Sniff nasal inspiratory pressure and sleep disordered breathing in childhood neuromuscular disorders. Neuromuscular Disorders, 2012, 22, 528-533.	0.3	19

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163	The Impact of ADHD on the Cognitive and Academic Functioning of Children With NF1. Developmental Neuropsychology, 2012, 37, 590-600.	1.0	65
164	Treatment for plexiform neurofibromas in patients with NF1. Lancet Oncology, The, 2012, 13, 1175-1176.	5.1	4
165	Mutations in TPM2 and congenital fibre type disproportion. Neuromuscular Disorders, 2012, 22, 955-958.	0.3	21
166	G.P.41 The identification of LGMD2G (TCAP) in Australia. Neuromuscular Disorders, 2012, 22, 831-832.	0.3	12
167	G.P.46 Screening for deletion and duplication mutations in genes implicated in LGMD. Neuromuscular Disorders, 2012, 22, 833.	0.3	0
168	C.I.1 Gene Wars: Episode V. The clinician strikes back. Neuromuscular Disorders, 2012, 22, 838.	0.3	0
169	C.P.15 K7del is a recurrent TPM2 nemaline myopathy mutation associated with joint contractures and increased calcium sensitivity. Neuromuscular Disorders, 2012, 22, 845.	0.3	0
170	S.P.53 Interventions for increasing ankle flexibility in patients with neuromuscular disease: A Cochrane Systematic Review. Neuromuscular Disorders, 2012, 22, 885.	0.3	0
171	S.P.55 Transition and outcomes for young men with Duchenne muscular dystrophy in New South Wales. Neuromuscular Disorders, 2012, 22, 885-886.	0.3	1
172	S.P.21 Clinical outcome measures in Collagen 6 (COL6) and Laminin Î±2(LAMA2) related congenital muscular dystrophy. Neuromuscular Disorders, 2012, 22, 893.	0.3	0
173	RARE MYOPATHIES AND EXPERIMENTAL APPROACHES - POSTER PRESENTATIONS G.P.125 ACTN3 genotype influences skeletal muscle performance through alterations in calcineurin signaling. Neuromuscular Disorders, 2012, 22, 904.	0.3	0
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