Kathryn N North

List of Publications by Year
in descending order

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		4388	9861
413	26,166	86	141
papers	citations	h-index	g-index

439	439	439	22543
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in ACTN4, encoding Î±-actinin-4, cause familial focal segmental glomerulosclerosis. Nature Genetics, 2000, 24, 251-256.	21.4	1,124
2	ACTN3 Genotype Is Associated with Human Elite Athletic Performance. American Journal of Human Genetics, 2003, 73, 627-631.	6.2	708
3	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
4	The nature and frequency of cognitive deficits in children with neurofibromatosis type 1. Neurology, 2005, 65, 1037-1044.	1.1	510
5	Distinctive patterns of microRNA expression in primary muscular disorders. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 17016-17021.	7.1	458
6	Neurofibromatosis type 1. Journal of the American Academy of Dermatology, 2009, 61, 1-14.	1.2	443
7	A brief history of human disease genetics. Nature, 2020, 577, 179-189.	27.8	441
8	Mutations in the skeletal muscle Î±-actin gene in patients with actin myopathy and nemaline myopathy. Nature Genetics, 1999, 23, 208-212.	21.4	389
9	Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. Brain, 2007, 130, 2725-2735.	7.6	385
10	A common nonsense mutation results in Î±-actinin-3 deficiency in the general population. Nature Genetics, 1999, 21, 353-354.	21.4	378
11	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	2.2	357
12	Differential expression of the actin-binding proteins, alpha-actinin-2 and -3, in different species: implications for the evolution of functional redundancy. Human Molecular Genetics, 2001, 10, 1335-1346.	2.9	299
13	Loss of ACTN3 gene function alters mouse muscle metabolism and shows evidence of positive selection in humans. Nature Genetics, 2007, 39, 1261-1265.	21.4	278
14	Diagnostic approach to the congenital muscular dystrophies. Neuromuscular Disorders, 2014, 24, 289-311.	0.6	275
15	Cognitive function and academic performance in neurofibrornatosis 1. Neurology, 1997, 48, 1121-1127.	1.1	270
16	An Actn3 knockout mouse provides mechanistic insights into the association between Â-actinin-3 deficiency and human athletic performance. Human Molecular Genetics, 2008, 17, 1076-1086.	2.9	266
17	Integrating Genomics into Healthcare: A Global Responsibility. American Journal of Human Genetics, 2019, 104, 13-20.	6.2	264
18	MURC/Cavin-4 and cavin family members form tissue-specific caveolar complexes. Journal of Cell Biology, 2009, 185, 1259-1273.	5.2	243

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19	Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders, 2014, 24, 97-116.	0.6	239
20	Nemaline myopathy: A clinical study of 143 cases. Annals of Neurology, 2001, 50, 312-320.	5.3	236
21	Mutations in SIL1 cause Marinesco-SjÃ¶gren syndrome, a cerebellar ataxia with cataract and myopathy. Nature Genetics, 2005, 37, 1312-1314.	21.4	232
22	Neurofibromatosis type 1 and optic pathway gliomas Follow-up of 54 patients. Ophthalmology, 2004, 111, 568-577.	5.2	205
23	Learning disabilities in children with neurofibromatosis type 1: subtypes, cognitive profile, and attention-deficitâ€“hyperactivity disorder. Developmental Medicine and Child Neurology, 2006, 48, 973.	2.1	203
24	A gene for speed? The evolution and function of ?-actinin-3. BioEssays, 2004, 26, 786-795.	2.5	197
25	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.	6.2	197
26	Microwave radiation can alter protein conformation without bulk heating. FEBS Letters, 2003, 543, 93-97.	2.8	191
27	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 6-18.	6.2	186
28	Neurofibromatosis Type 1. American Journal of Medical Genetics Part A, 2000, 97, 119-127.	2.4	182
29	Specific learning disability in children with neurofibromatosis type 1. Neurology, 1994, 44, 878-878.	1.1	182
30	Cerebrovascular complications in Ehlers-Danlos syndrome type IV. Annals of Neurology, 1995, 38, 960-964.	5.3	176
31	A federated ecosystem for sharing genomic, clinical data. Science, 2016, 352, 1278-1280.	12.6	175
32	Brain involvement in muscular dystrophies with defective dystroglycan glycosylation. Annals of Neurology, 2008, 64, 573-582.	5.3	172
33	Genes and human elite athletic performance. Human Genetics, 2005, 116, 331-339.	3.8	171
34	SEPN1: Associated with congenital fiber-type disproportion and insulin resistance. Annals of Neurology, 2006, 59, 546-552.	5.3	165
35	Association analysis of the ACTN3 R577X polymorphism and complex quantitative body composition and performance phenotypes in adolescent Greeks. European Journal of Human Genetics, 2007, 15, 88-93.	2.8	165
36	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. JAMA Neurology, 2015, 72, 1424.	9.0	164

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37	Nemaline myopathy: current concepts. The ENMC International Consortium and Nemaline Myopathy.. Journal of Medical Genetics, 1997, 34, 705-713.	3.2	158
38	Genes for Elite Power and Sprint Performance: ACTN3 Leads the Way. Sports Medicine, 2013, 43, 803-817.	6.5	158
39	Dominant collagen VI mutations are a common cause of Ullrich congenital muscular dystrophy. Human Molecular Genetics, 2004, 14, 279-293.	2.9	156
40	Mutations in BICD2 Cause Dominant Congenital Spinal Muscular Atrophy and Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 92, 965-973.	6.2	156
41	Recessive mutations in RYR1 are a common cause of congenital fiber type disproportion. Human Mutation, 2010, 31, E1544-E1550.	2.5	153
42	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708.	8.2	153
43	Mutations in TPM3 are a common cause of congenital fiber type disproportion. Annals of Neurology, 2008, 63, 329-337.	5.3	152
44	The expanding phenotype of laminin alpha2 chain (merosin) abnormalities: case series and review. Journal of Medical Genetics, 2001, 38, 649-657.	3.2	150
45	Actin mutations are one cause of congenital fibre type disproportion. Annals of Neurology, 2004, 56, 689-694.	5.3	149
46	Large-scale GWAS identifies multiple loci for hand grip strength providing biological insights into muscular fitness. Nature Communications, 2017, 8, 16015.	12.8	149
47	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	3.5	148
48	Consensus Statement on Standard of Care for Congenital Myopathies. Journal of Child Neurology, 2012, 27, 363-382.	1.4	147
49	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.	6.2	147
50	Nemaline Myopathy Caused by Mutations in the Muscle Î±-Skeletal-Actin Gene. American Journal of Human Genetics, 2001, 68, 1333-1343.	6.2	144
51	Dominant Mutations in KBTBD13, a Member of the BTB/Kelch Family, Cause Nemaline Myopathy with Cores. American Journal of Human Genetics, 2010, 87, 842-847.	6.2	143
52	Gliomas presenting after age 10 in individuals with neurofibromatosis type 1 (NF1). Neurology, 2002, 59, 759-761.	1.1	139
53	Social skills of children with neurofibromatosis type 1. Developmental Medicine and Child Neurology, 2004, 46, 553-63.	2.1	137
54	Assessment of executive function and attention in children with neurofibromatosis type 1: Relationships between cognitive measures and real-world behavior. Child Neuropsychology, 2011, 17, 313-329.	1.3	131

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55	C2C12 Co-culture on a fibroblast substratum enables sustained survival of contractile, highly differentiated myotubes with peripheral nuclei and adult fast myosin expression. Cytoskeleton, 2004, 58, 200-211.	4.4	129
56	Deficiency of a skeletal muscle isoform of Î±-actinin (Î±-actinin-3) in merosin-positive congenital muscular dystrophy. Neuromuscular Disorders, 1996, 6, 229-235.	0.6	127
57	Neurofibromatosis Type 1: Review of the First 200 Patients in an Australian Clinic. Journal of Child Neurology, 1993, 8, 395-402.	1.4	122
58	Clinical course correlates poorly with muscle pathology in nemaline myopathy. Neurology, 2003, 60, 665-673.	1.1	120
59	ACTN3 and ACE Genotypes in Elite Jamaican and US Sprinters. Medicine and Science in Sports and Exercise, 2010, 42, 107-112.	0.4	120
60	Ferlins: Regulators of Vesicle Fusion for Auditory Neurotransmission, Receptor Trafficking and Membrane Repair. Traffic, 2012, 13, 185-194.	2.7	119
61	Principal mutation hotspot for central core disease and related myopathies in the C-terminal transmembrane region of the RYR1 gene. Neuromuscular Disorders, 2003, 13, 151-157.	0.6	118
62	ACTN3. Exercise and Sport Sciences Reviews, 2007, 35, 30-34.	3.0	118
63	MRI findings in children with neurofibromatosis type 1: a prospective study. Pediatric Radiology, 1996, 26, 478-487.	2.0	117
64	ACTN3 genotype influences muscle performance through the regulation of calcineurin signaling. Journal of Clinical Investigation, 2013, 123, 4255-4263.	8.2	113
65	Direct-to-consumer genetic testing for predicting sports performance and talent identification: Consensus statement. British Journal of Sports Medicine, 2015, 49, 1486-1491.	6.7	113
66	COGNITIVE FUNCTION AND ACADEMIC PERFORMANCE IN CHILDREN WITH NEUROFIBROMATOSIS TYPE 1. Developmental Medicine and Child Neurology, 1995, 37, 427-436.	2.1	110
67	Genotype?phenotype correlations in nemaline myopathy caused by mutations in the genes for nebulin and skeletal muscle ?-actin. Neuromuscular Disorders, 2004, 14, 461-470.	0.6	107
68	Mutation Update: The Spectra of Nebulin Variants and Associated Myopathies. Human Mutation, 2014, 35, 1418-1426.	2.5	107
69	Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. Neurology, 2016, 86, 391-398.	1.1	107
70	ACTN3 R577X and ACE I/D gene variants influence performance in elite sprinters: a multi-cohort study. BMC Genomics, 2016, 17, 285.	2.8	106
71	Homozygosity for a nonsense mutation in the alpha-tropomyosin slow gene TPM3 in a patient with severe infantile nemaline myopathy. Neuromuscular Disorders, 1999, 9, 573-579.	0.6	105
72	Natural history of cognitive deficits and their relationship to MRI T2-hyperintensities in NF1. Neurology, 2003, 60, 1139-1145.	1.1	105

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73	Outcome of noninvasive ventilation in children with neuromuscular disease. Neurology, 2007, 68, 198-201.	1.1	102
74	Review Article : Cognitive Deficits in Neurofibromatosis 1. Journal of Child Neurology, 2002, 17, 605-612.	1.4	101
75	The ACTN3 R577X Polymorphism in East and West African Athletes. Medicine and Science in Sports and Exercise, 2007, 39, 1985-1988.	0.4	100
76	Expanding the clinical, pathological and MRI phenotype of DNM2-related centronuclear myopathy. Neuromuscular Disorders, 2010, 20, 229-237.	0.6	100
77	Limbâ€“girdle muscular dystrophy: Diagnostic evaluation, frequency and clues to pathogenesis. Neuromuscular Disorders, 2008, 18, 34-44.	0.6	99
78	Magnetic resonance imaging of muscle in nemaline myopathy. Neuromuscular Disorders, 2004, 14, 779-784.	0.6	98
79	No Evidence of a Common DNA Variant Profile Specific to World Class Endurance Athletes. PLoS ONE, 2016, 11, e0147330.	2.5	96
80	Athlome Project Consortium: a concerted effort to discover genomic and other â€œomicâ€•markers of athletic performance. Physiological Genomics, 2016, 48, 183-190.	2.3	96
81	A Gene for Speed: The Emerging Role of Î±-Actinin-3 in Muscle Metabolism. Physiology, 2010, 25, 250-259.	3.1	95
82	Deficiency of Î±-actinin-3 is associated with increased susceptibility to contraction-induced damage and skeletal muscle remodeling. Human Molecular Genetics, 2011, 20, 2914-2927.	2.9	95
83	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	6.5	94
84	Calpains, Cleaved Mini-Dysferlin_{C72}, and L-Type Channels Underpin Calcium-Dependent Muscle Membrane Repair. Journal of Neuroscience, 2013, 33, 5085-5094.	3.6	93
85	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
86	Congenital Fiber Type Disproportionâ€”30 Years On. Journal of Neuropathology and Experimental Neurology, 2003, 62, 977-989.	1.7	92
87	Cognitive and Psychological Profile of Males With Becker Muscular Dystrophy. Journal of Child Neurology, 2008, 23, 155-162.	1.4	92
88	Mutation Update and Genotype-Phenotype Correlations of Novel and Previously Described Mutations inTPM2andTPM3Causing Congenital Myopathies. Human Mutation, 2014, 35, 779-790.	2.5	92
89	Brain structure and function in neurofibromatosis type 1: current concepts and future directions. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 304-309.	1.9	90
90	Disease Burden and Symptom Structure of Autism in Neurofibromatosis Type 1. JAMA Psychiatry, 2016, 73, 1276.	11.0	90

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91	Evidence for a dominant-negative effect in ACTA1 nemaline myopathy caused by abnormal folding, aggregation and altered polymerization of mutant actin isoforms. Human Molecular Genetics, 2004, 13, 1727-1743.	2.9	89
92	Evidence-based guideline summary: Evaluation, diagnosis, and management of congenital muscular dystrophy. Neurology, 2015, 84, 1369-1378.	1.1	88
93	T2 hyperintensities in children with neurofibromatosis type 1 and their relationship to cognitive functioning. Journal of Neurology, Neurosurgery and Psychiatry, 2007, 78, 1088-1091.	1.9	87
94	Lethal neonatal deficiency of carnitine palmitoyltransferase II associated with dysgenesis of the brain and kidneys. Journal of Pediatrics, 1995, 127, 414-420.	1.8	86
95	Single section Western blot. Neurology, 2003, 61, 93-97.	1.1	86
96	Cardiac aquaporin expression in humans, rats, and mice. American Journal of Physiology - Heart and Circulatory Physiology, 2006, 291, H705-H713.	3.2	86
97	Growth in North American white children with neurofibromatosis 1 (NF1). Journal of Medical Genetics, 2000, 37, 933-938.	3.2	85
98	A mutation in alpha-tropomyosinslow affects muscle strength, maturation and hypertrophy in a mouse model for nemaline myopathy. Human Molecular Genetics, 2001, 10, 317-328.	2.9	85
99	Natural history of pulmonary function in collagen VI-related myopathies. Brain, 2013, 136, 3625-3633.	7.6	85
100	Decreased Bone Mineral Density in Neurofibromatosis Type 1. Journal of Pediatric Orthopaedics, 2007, 27, 472-475.	1.2	83
101	Diagnosis and etiology of congenital muscular dystrophy. Neurology, 2008, 71, 312-321.	1.1	83
102	Phenotypic and molecular insights into spinal muscular atrophy due to mutations in BICD2. Brain, 2015, 138, 293-310.	7.6	82
103	Association Analysis of ACE and ACTN3 in Elite Caucasian and East Asian Swimmers. Medicine and Science in Sports and Exercise, 2013, 45, 892-900.	0.4	80
104	Dysferlin, Annexin A1, and Mitsugumin 53 Are Upregulated in Muscular Dystrophy and Localize to Longitudinal Tubules of the T-System With Stretch. Journal of Neuropathology and Experimental Neurology, 2011, 70, 302-313.	1.7	77
105	Randomized placebo-controlled study of lovastatin in children with neurofibromatosis type 1. Neurology, 2016, 87, 2575-2584.	1.1	76
106	A gene for speed: contractile properties of isolated whole EDL muscle from an Î±-actinin-3 knockout mouse. American Journal of Physiology - Cell Physiology, 2008, 295, C897-C904.	4.6	75
107	Neurofibromatosis Type 2. Journal of Child Neurology, 2017, 32, 9-22.	1.4	75
108	Australian Genomics: A Federated Model for Integrating Genomics into Healthcare. American Journal of Human Genetics, 2019, 105, 7-14.	6.2	75

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109	Cerebrovascular dysplasia in neurofibromatosis type 1. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 1165-1170.	1.9	74
110	Defining Î±-skeletal and Î±-cardiac actin expression in human heart and skeletal muscle explains the absence of cardiac involvement in ACTA1 nemaline myopathy. Neuromuscular Disorders, 2005, 15, 829-835.	0.6	73
111	Î±-Actinin-3 deficiency results in reduced glycogen phosphorylase activity and altered calcium handling in skeletal muscle. Human Molecular Genetics, 2010, 19, 1335-1346.	2.9	73
112	Age-related findings on MRI in neurofibromatosis type 1. Pediatric Radiology, 2006, 36, 1048-1056.	2.0	72
113	Mutations in Contactin-1, a Neural Adhesion and Neuromuscular Junction Protein, Cause a Familial Form of Lethal Congenital Myopathy. American Journal of Human Genetics, 2008, 83, 714-724.	6.2	72
114	Loss of IL-15 receptor Î± alters the endurance, fatigability, and metabolic characteristics of mouse fast skeletal muscles. Journal of Clinical Investigation, 2011, 121, 3120-3132.	8.2	72
115	UDP-N-Acetylglucosamine 2-Epimerase/N-Acetylmannosamine Kinase (GNE) Binds to Alpha-Actinin 1: Novel Pathways in Skeletal Muscle?. PLoS ONE, 2008, 3, e2477.	2.5	71
116	Increased connective tissue growth factor associated with cardiac fibrosis in the mdx mouse model of dystrophic cardiomyopathy. International Journal of Experimental Pathology, 2011, 92, 57-65.	1.3	70
117	Corpus Callosum Morphology and Its Relationship to Cognitive Function in Neurofibromatosis Type 1. Journal of Child Neurology, 2010, 25, 834-841.	1.4	69
118	Evidence Based Selection of Commonly Used RT-qPCR Reference Genes for the Analysis of Mouse Skeletal Muscle. PLoS ONE, 2014, 9, e88653.	2.5	69
119	Cost-effectiveness of massively parallel sequencing for diagnosis of paediatric muscle diseases. Npj Genomic Medicine, 2017, 2, .	3.8	67
120	Neonatal-onset propionic acidemia: Neurologic and developmental profiles, and implications for management. Journal of Pediatrics, 1995, 126, 916-922.	1.8	66
121	Aberrant dysferlin trafficking in cells lacking caveolin or expressing dystrophy mutants of caveolin-3. Human Molecular Genetics, 2006, 15, 129-142.	2.9	66
122	Molecular consequences of dominant Bethlem myopathy collagen VI mutations. Annals of Neurology, 2007, 62, 390-405.	5.3	66
123	Congenital myopathies. Current Opinion in Neurology, 2008, 21, 569-575.	3.6	66
124	α-Actinin-3 and Performance. Medicine and Sport Science, 2009, 54, 88-101.	1.4	65
125	Fetal akinesia: review of the genetics of the neuromuscular causes. Journal of Medical Genetics, 2011, 48, 793-801.	3.2	65
126	The Impact of ADHD on the Cognitive and Academic Functioning of Children With NF1. Developmental Neuropsychology, 2012, 37, 590-600.	1.4	65

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127	Current whole-body MRI applications in the neurofibromatoses. Neurology, 2016, 87, S31-9.	1.1	65
128	No association between ACTN3 R577X and ACE I/D polymorphisms and endurance running times in 698 Caucasian athletes. BMC Genomics, 2018, 19, 13.	2.8	65
129	Mutations inPIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. Human Molecular Genetics, 2015, 24, 6146-6159.	2.9	64
130	Expression of aquaporin 1Bin human cardiac and skeletal muscle. Journal of Molecular and Cellular Cardiology, 2004, 36, 655-662.	1.9	63
131	Mechanisms underlying intranuclear rod formation. Brain, 2007, 130, 3275-3284.	7.6	63
132	The pathogenesis ofACTA1-related congenital fiber type disproportion. Annals of Neurology, 2007, 61, 552-561.	5.3	63
133	An ?tropomyosin mutation alters dimer preference in nemaline myopathy. Annals of Neurology, 2005, 57, 42-49.	5.3	62
134	Calpain cleavage within dysferlin exon 40a releases a synaptotagmin-like module for membrane repair. Molecular Biology of the Cell, 2014, 25, 3037-3048.	2.1	62
135	Collagen VI glycine mutations: Perturbed assembly and a spectrum of clinical severity. Annals of Neurology, 2008, 64, 294-303.	5.3	61
136	Hypertrophy and dietary tyrosine ameliorate the phenotypes of a mouse model of severe nemaline myopathy. Brain, 2011, 134, 3516-3529.	7.6	59
137	Optic gliomas in neurofibromatosis type 1: Role of visual evoked potentials. Pediatric Neurology, 1994, 10, 117-123.	2.1	58
138	Dietary L-Tyrosine Supplementation in Nemaline Myopathy. Journal of Child Neurology, 2008, 23, 609-613.	1.4	58
139	Mental, Motor, and Language Development of Toddlers with Neurofibromatosis Type 1. Journal of Pediatrics, 2011, 158, 660-665.	1.8	58
140	Evidence for ACTN3 as a genetic modifier of Duchenne muscular dystrophy. Nature Communications, 2017, 8, 14143.	12.8	58
141	Diagnosis and etiology of congenital muscular dystrophy: We are halfway there. Annals of Neurology, 2016, 80, 101-111.	5.3	57
142	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.	4.1	57
143	Review Article : Neurofibromatosis 1: Clinical Review and Exceptions to the Rules. Journal of Child Neurology, 2002, 17, 613-621.	1.4	55
144	Whatâ€™s new in congenital myopathies?. Neuromuscular Disorders, 2008, 18, 433-442.	0.6	55

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145	Health-related Quality of Life in Boys With Duchenne Muscular Dystrophy: Agreement Between Parents and Their Sons. Journal of Child Neurology, 2010, 25, 1188-1194.	1.4	55
146	Expanding the phenotype of GMPPB mutations. Brain, 2015, 138, 836-844.	7.6	54
147	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.	2.5	50
148	Autosomal dominant nemaline myopathy with intranuclear rods due to mutation of the skeletal muscle ACTA1 gene: Clinical and pathological variability within a kindred. Neuromuscular Disorders, 2006, 16, 113-121.	0.6	49
149	Phylogenetic analysis of ferlin genes reveals ancient eukaryotic origins. BMC Evolutionary Biology, 2010, 10, 231.	3.2	49
150	Nemaline myopathy and distal arthrogryposis associated with an autosomal recessiveÂTNNT3Âsplice variant. Human Mutation, 2018, 39, 383-388.	2.5	48
151	Neurofibromatosis 1 in childhood. Seminars in Pediatric Neurology, 1998, 5, 231-242.	2.0	47
152	Clinical Approach to the Diagnosis of Congenital Myopathies. Seminars in Pediatric Neurology, 2011, 18, 216-220.	2.0	47
153	The effect of Î±-actinin-3 deficiency on muscle aging. Experimental Gerontology, 2011, 46, 292-302.	2.8	47
154	Outcome of children with neuromuscular disease admitted to paediatric intensive care. Archives of Disease in Childhood, 2004, 89, 170-175.	1.9	46
155	Variants in SLC18A3 , vesicular acetylcholine transporter, cause congenital myasthenic syndrome. Neurology, 2016, 87, 1442-1448.	1.1	46
156	Lack of MG53 in human heart precludes utility as a biomarker of myocardial injury or endogenous cardioprotective factor. Cardiovascular Research, 2016, 110, 178-187.	3.8	46
157	CHARGE association in a child with de novo inverted duplication (14)(q22 â†' q24.3). American Journal of Medical Genetics Part A, 1995, 57, 610-614.	2.4	45
158	Myocardial water handling and the role of aquaporins. Biochimica Et Biophysica Acta - Biomembranes, 2006, 1758, 1043-1052.	2.6	45
159	Utility of positron emission tomography for tumour surveillance in children with neurofibromatosis type 1. European Journal of Nuclear Medicine and Molecular Imaging, 2010, 37, 1309-1317.	6.4	45
160	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.	9.4	45
161	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. American Journal of Human Genetics, 2016, 99, 1086-1105.	6.2	45
162	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.	4.9	45

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163	Hand involvement in children with Charcotâ€“Marie-Tooth disease type 1A. Neuromuscular Disorders, 2008, 18, 970-973.	0.6	44
164	Cognitive dysfunction as the major presenting feature of Becker's muscular dystrophy. Neurology, 1996, 46, 461-464.	1.1	43
165	Cap disease due to mutation of the beta-tropomyosin gene (TPM2). Neuromuscular Disorders, 2009, 19, 348-351.	0.6	43
166	Does attentionâ€deficitâ€“hyperactivity disorder exacerbate executive dysfunction in children with neurofibromatosis type 1?. Developmental Medicine and Child Neurology, 2012, 54, 898-904.	2.1	43
167	K7del is a common TPM2 gene mutation associated with nemaline myopathy and raised myofibre calcium sensitivity. Brain, 2013, 136, 494-507.	7.6	42
168	Importance and challenge of making an early diagnosis in LMNA -related muscular dystrophy. Neurology, 2012, 78, 1258-1263.	1.1	41
169	Longitudinal assessment of cognition and T2â€hyperintensities in NF1: An 18â€year study. American Journal of Medical Genetics, Part A, 2014, 164, 661-665.	1.2	41
170	Idiopathic Hypothalamic Dysfunction With Dilated Unresponsive Pupils: Report of Two Cases. Journal of Child Neurology, 1994, 9, 320-325.	1.4	40
171	Mild Functional Differences of Dynamin 2 Mutations Associated to Centronuclear Myopathy and Charcot-Marie-Tooth Peripheral Neuropathy. PLoS ONE, 2011, 6, e27498.	2.5	40
172	NF1 is a critical regulator of muscle development and metabolism. Human Molecular Genetics, 2014, 23, 1250-1259.	2.9	40
173	National Neurofibromatosis Foundation International Database. American Journal of Medical Genetics Part A, 1993, 45, 88-91.	2.4	39
174	Idiopathic hypothalamic dysfunction: a paraneoplastic syndrome?. Lancet, The, 1995, 346, 1298.	13.7	39
175	Intranuclear rod myopathy: molecular pathogenesis and mechanisms of weakness. Annals of Neurology, 2007, 62, 597-608.	5.3	39
176	The self-concept of children and adolescents with neurofibromatosis type 1. Child: Care, Health and Development, 2007, 33, 401-408.	1.7	39
177	Tropomyosin 4 defines novel filaments in skeletal muscle associated with muscle remodelling/regeneration in normal and diseased muscle. Cytoskeleton, 2008, 65, 73-85.	4.4	39
178	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.	2.1	39
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