# Kathryn N North

List of Publications by Year
in descending order

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| | | 4370 | 9839 |
|---|---|---|---|
| 412 | 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. | 9.4 | 1,124 |
| 2 | ACTN3 Genotype Is Associated with Human Elite Athletic Performance. American Journal of Human Genetics, 2003, 73, 627-631. | 2.6 | 708 |
| 3 | Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, . | 5.8 | 516 |
| 4 | The nature and frequency of cognitive deficits in children with neurofibromatosis type 1. Neurology, 2005, 65, 1037-1044. | 1.5 | 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. | 3.3 | 458 |
| 6 | Neurofibromatosis type 1. Journal of the American Academy of Dermatology, 2009, 61, 1-14. | 0.6 | 443 |
| 7 | A brief history of human disease genetics. Nature, 2020, 577, 179-189. | 13.7 | 441 |
| 8 | Mutations in the skeletal muscle Î±-actin gene in patients with actin myopathy and nemaline myopathy. Nature Genetics, 1999, 23, 208-212. | 9.4 | 389 |
| 9 | Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. Brain, 2007, 130, 2725-2735. | 3.7 | 385 |
| 10 | A common nonsense mutation results in Î±-actinin-3 deficiency in the general population. Nature Genetics, 1999, 21, 353-354. | 9.4 | 378 |
| 11 | Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487. | 1.0 | 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. | 1.4 | 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. | 9.4 | 278 |
| 14 | Diagnostic approach to the congenital muscular dystrophies. Neuromuscular Disorders, 2014, 24, 289-311. | 0.3 | 275 |
| 15 | Cognitive function and academic performance in neurofibrornatosis 1. Neurology, 1997, 48, 1121-1127. | 1.5 | 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. | 1.4 | 266 |
| 17 | Integrating Genomics into Healthcare: A Global Responsibility. American Journal of Human Genetics, 2019, 104, 13-20. | 2.6 | 264 |
| 18 | MURC/Cavin-4 and cavin family members form tissue-specific caveolar complexes. Journal of Cell Biology, 2009, 185, 1259-1273. | 2.3 | 243 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders, 2014, 24, 97-116. | 0.3 | 239 |
| 20 | Nemaline myopathy: A clinical study of 143 cases. Annals of Neurology, 2001, 50, 312-320. | 2.8 | 236 |
| 21 | Mutations in SIL1 cause Marinesco-SjÃ¶gren syndrome, a cerebellar ataxia with cataract and myopathy. Nature Genetics, 2005, 37, 1312-1314. | 9.4 | 232 |
| 22 | Neurofibromatosis type 1 and optic pathway gliomas Follow-up of 54 patients. Ophthalmology, 2004, 111, 568-577. | 2.5 | 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. | 1.1 | 203 |
| 24 | A gene for speed? The evolution and function of ?-actinin-3. BioEssays, 2004, 26, 786-795. | 1.2 | 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. | 2.6 | 197 |
| 26 | Microwave radiation can alter protein conformation without bulk heating. FEBS Letters, 2003, 543, 93-97. | 1.3 | 191 |
| 27 | 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 |
| 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. | 1.5 | 182 |
| 30 | Cerebrovascular complications in Ehlers-Danlos syndrome type IV. Annals of Neurology, 1995, 38, 960-964. | 2.8 | 176 |
| 31 | A federated ecosystem for sharing genomic, clinical data. Science, 2016, 352, 1278-1280. | 6.0 | 175 |
| 32 | Brain involvement in muscular dystrophies with defective dystroglycan glycosylation. Annals of Neurology, 2008, 64, 573-582. | 2.8 | 172 |
| 33 | Genes and human elite athletic performance. Human Genetics, 2005, 116, 331-339. | 1.8 | 171 |
| 34 | SEPN1: Associated with congenital fiber-type disproportion and insulin resistance. Annals of Neurology, 2006, 59, 546-552. | 2.8 | 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. | 1.4 | 165 |
| 36 | Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. JAMA Neurology, 2015, 72, 1424. | 4.5 | 164 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Nemaline myopathy: current concepts. The ENMC International Consortium and Nemaline Myopathy.. Journal of Medical Genetics, 1997, 34, 705-713. | 1.5 | 158 |
| 38 | Genes for Elite Power and Sprint Performance: ACTN3 Leads the Way. Sports Medicine, 2013, 43, 803-817. | 3.1 | 158 |
| 39 | Dominant collagen VI mutations are a common cause of Ullrich congenital muscular dystrophy. Human Molecular Genetics, 2004, 14, 279-293. | 1.4 | 156 |
| 40 | 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 |
| 41 | Recessive mutations in RYR1 are a common cause of congenital fiber type disproportion. Human Mutation, 2010, 31, E1544-E1550. | 1.1 | 153 |
| 42 | Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708. | 3.9 | 153 |
| 43 | Mutations in TPM3 are a common cause of congenital fiber type disproportion. Annals of Neurology, 2008, 63, 329-337. | 2.8 | 152 |
| 44 | The expanding phenotype of laminin alpha2 chain (merosin) abnormalities: case series and review. Journal of Medical Genetics, 2001, 38, 649-657. | 1.5 | 150 |
| 45 | Actin mutations are one cause of congenital fibre type disproportion. Annals of Neurology, 2004, 56, 689-694. | 2.8 | 149 |
| 46 | Large-scale GWAS identifies multiple loci for hand grip strength providing biological insights into muscular fitness. Nature Communications, 2017, 8, 16015. | 5.8 | 149 |
| 47 | BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752. | 1.5 | 148 |
| 48 | Consensus Statement on Standard of Care for Congenital Myopathies. Journal of Child Neurology, 2012, 27, 363-382. | 0.7 | 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. | 2.6 | 147 |
| 50 | Nemaline Myopathy Caused by Mutations in the Muscle Î±-Skeletal-Actin Gene. American Journal of Human Genetics, 2001, 68, 1333-1343. | 2.6 | 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. | 2.6 | 143 |
| 52 | Gliomas presenting after age 10 in individuals with neurofibromatosis type 1 (NF1). Neurology, 2002, 59, 759-761. | 1.5 | 139 |
| 53 | Social skills of children with neurofibromatosis type 1. Developmental Medicine and Child Neurology, 2004, 46, 553-63. | 1.1 | 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. | 0.8 | 131 |

| # | Article | IF | Citations |
|---|---|---|---|
| 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. | 0.3 | 127 |
| 57 | Neurofibromatosis Type 1: Review of the First 200 Patients in an Australian Clinic. Journal of Child Neurology, 1993, 8, 395-402. | 0.7 | 122 |
| 58 | Clinical course correlates poorly with muscle pathology in nemaline myopathy. Neurology, 2003, 60, 665-673. | 1.5 | 120 |
| 59 | ACTN3 and ACE Genotypes in Elite Jamaican and US Sprinters. Medicine and Science in Sports and Exercise, 2010, 42, 107-112. | 0.2 | 120 |
| 60 | Ferlins: Regulators of Vesicle Fusion for Auditory Neurotransmission, Receptor Trafficking and Membrane Repair. Traffic, 2012, 13, 185-194. | 1.3 | 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. | 0.3 | 118 |
| 62 | ACTN3. Exercise and Sport Sciences Reviews, 2007, 35, 30-34. | 1.6 | 118 |
| 63 | MRI findings in children with neurofibromatosis type 1: a prospective study. Pediatric Radiology, 1996, 26, 478-487. | 1.1 | 117 |
| 64 | ACTN3 genotype influences muscle performance through the regulation of calcineurin signaling. Journal of Clinical Investigation, 2013, 123, 4255-4263. | 3.9 | 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. | 3.1 | 113 |
| 66 | COGNITIVE FUNCTION AND ACADEMIC PERFORMANCE IN CHILDREN WITH NEUROFIBROMATOSIS TYPE 1. Developmental Medicine and Child Neurology, 1995, 37, 427-436. | 1.1 | 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. | 0.3 | 107 |
| 68 | Mutation Update: The Spectra of Nebulin Variants and Associated Myopathies. Human Mutation, 2014, 35, 1418-1426. | 1.1 | 107 |
| 69 | Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. Neurology, 2016, 86, 391-398. | 1.5 | 107 |
| 70 | 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 |
| 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. | 0.3 | 105 |
| 72 | Natural history of cognitive deficits and their relationship to MRI T2-hyperintensities in NF1. Neurology, 2003, 60, 1139-1145. | 1.5 | 105 |

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|---|---|---|---|
| 73 | Outcome of noninvasive ventilation in children with neuromuscular disease. Neurology, 2007, 68, 198-201. | 1.5 | 102 |
| 74 | Review Article : Cognitive Deficits in Neurofibromatosis 1. Journal of Child Neurology, 2002, 17, 605-612. | 0.7 | 101 |
| 75 | The ACTN3 R577X Polymorphism in East and West African Athletes. Medicine and Science in Sports and Exercise, 2007, 39, 1985-1988. | 0.2 | 100 |
| 76 | Expanding the clinical, pathological and MRI phenotype of DNM2-related centronuclear myopathy. Neuromuscular Disorders, 2010, 20, 229-237. | 0.3 | 100 |
| 77 | Limbâ€“girdle muscular dystrophy: Diagnostic evaluation, frequency and clues to pathogenesis. Neuromuscular Disorders, 2008, 18, 34-44. | 0.3 | 99 |
| 78 | Magnetic resonance imaging of muscle in nemaline myopathy. Neuromuscular Disorders, 2004, 14, 779-784. | 0.3 | 98 |
| 79 | No Evidence of a Common DNA Variant Profile Specific to World Class Endurance Athletes. PLoS ONE, 2016, 11, e0147330. | 1.1 | 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. | 1.0 | 96 |
| 81 | A Gene for Speed: The Emerging Role of Î±-Actinin-3 in Muscle Metabolism. Physiology, 2010, 25, 250-259. | 1.6 | 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. | 1.4 | 95 |
| 83 | GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029. | 3.0 | 94 |
| 84 | Calpains, Cleaved Mini-Dysferlin<sub>C72</sub>, and L-Type Channels Underpin Calcium-Dependent Muscle Membrane Repair. Journal of Neuroscience, 2013, 33, 5085-5094. | 1.7 | 93 |
| 85 | Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124. | 2.8 | 93 |
| 86 | Congenital Fiber Type Disproportionâ€”30 Years On. Journal of Neuropathology and Experimental Neurology, 2003, 62, 977-989. | 0.9 | 92 |
| 87 | Cognitive and Psychological Profile of Males With Becker Muscular Dystrophy. Journal of Child Neurology, 2008, 23, 155-162. | 0.7 | 92 |
| 88 | Mutation Update and Genotype-Phenotype Correlations of Novel and Previously Described Mutations inTPM2andTPM3Causing Congenital Myopathies. Human Mutation, 2014, 35, 779-790. | 1.1 | 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. | 0.9 | 90 |
| 90 | Disease Burden and Symptom Structure of Autism in Neurofibromatosis Type 1. JAMA Psychiatry, 2016, 73, 1276. | 6.0 | 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. | 1.4 | 89 |
| 92 | Evidence-based guideline summary: Evaluation, diagnosis, and management of congenital muscular dystrophy. Neurology, 2015, 84, 1369-1378. | 1.5 | 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. | 0.9 | 87 |
| 94 | Lethal neonatal deficiency of carnitine palmitoyltransferase II associated with dysgenesis of the brain and kidneys. Journal of Pediatrics, 1995, 127, 414-420. | 0.9 | 86 |
| 95 | Single section Western blot. Neurology, 2003, 61, 93-97. | 1.5 | 86 |
| 96 | Cardiac aquaporin expression in humans, rats, and mice. American Journal of Physiology - Heart and Circulatory Physiology, 2006, 291, H705-H713. | 1.5 | 86 |
| 97 | Growth in North American white children with neurofibromatosis 1 (NF1). Journal of Medical Genetics, 2000, 37, 933-938. | 1.5 | 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. | 1.4 | 85 |
| 99 | Natural history of pulmonary function in collagen VI-related myopathies. Brain, 2013, 136, 3625-3633. | 3.7 | 85 |
| 100 | Decreased Bone Mineral Density in Neurofibromatosis Type 1. Journal of Pediatric Orthopaedics, 2007, 27, 472-475. | 0.6 | 83 |
| 101 | Diagnosis and etiology of congenital muscular dystrophy. Neurology, 2008, 71, 312-321. | 1.5 | 83 |
| 102 | Phenotypic and molecular insights into spinal muscular atrophy due to mutations in BICD2. Brain, 2015, 138, 293-310. | 3.7 | 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. | 0.2 | 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. | 0.9 | 77 |
| 105 | Randomized placebo-controlled study of lovastatin in children with neurofibromatosis type 1. Neurology, 2016, 87, 2575-2584. | 1.5 | 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. | 2.1 | 75 |
| 107 | Neurofibromatosis Type 2. Journal of Child Neurology, 2017, 32, 9-22. | 0.7 | 75 |
| 108 | Australian Genomics: A Federated Model for Integrating Genomics into Healthcare. American Journal of Human Genetics, 2019, 105, 7-14. | 2.6 | 75 |

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|---|---|---|---|
| 109 | Cerebrovascular dysplasia in neurofibromatosis type 1. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 1165-1170. | 0.9 | 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. | 0.3 | 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. | 1.4 | 73 |
| 112 | Age-related findings on MRI in neurofibromatosis type 1. Pediatric Radiology, 2006, 36, 1048-1056. | 1.1 | 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. | 2.6 | 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. | 3.9 | 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. | 1.1 | 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. | 0.6 | 70 |
| 117 | Corpus Callosum Morphology and Its Relationship to Cognitive Function in Neurofibromatosis Type 1. Journal of Child Neurology, 2010, 25, 834-841. | 0.7 | 69 |
| 118 | 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 |
| 119 | Cost-effectiveness of massively parallel sequencing for diagnosis of paediatric muscle diseases. Npj Genomic Medicine, 2017, 2, . | 1.7 | 67 |
| 120 | Neonatal-onset propionic acidemia: Neurologic and developmental profiles, and implications for management. Journal of Pediatrics, 1995, 126, 916-922. | 0.9 | 66 |
| 121 | Aberrant dysferlin trafficking in cells lacking caveolin or expressing dystrophy mutants of caveolin-3. Human Molecular Genetics, 2006, 15, 129-142. | 1.4 | 66 |
| 122 | Molecular consequences of dominant Bethlem myopathy collagen VI mutations. Annals of Neurology, 2007, 62, 390-405. | 2.8 | 66 |
| 123 | Congenital myopathies. Current Opinion in Neurology, 2008, 21, 569-575. | 1.8 | 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. | 1.5 | 65 |
| 126 | The Impact of ADHD on the Cognitive and Academic Functioning of Children With NF1. Developmental Neuropsychology, 2012, 37, 590-600. | 1.0 | 65 |

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|---|---|---|---|
| 127 | Current whole-body MRI applications in the neurofibromatoses. Neurology, 2016, 87, S31-9. | 1.5 | 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. | 1.2 | 65 |
| 129 | Mutations inPIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. Human Molecular Genetics, 2015, 24, 6146-6159. | 1.4 | 64 |
| 130 | Expression of aquaporin 1Bin human cardiac and skeletal muscle. Journal of Molecular and Cellular Cardiology, 2004, 36, 655-662. | 0.9 | 63 |
| 131 | Mechanisms underlying intranuclear rod formation. Brain, 2007, 130, 3275-3284. | 3.7 | 63 |
| 132 | The pathogenesis ofACTA1-related congenital fiber type disproportion. Annals of Neurology, 2007, 61, 552-561. | 2.8 | 63 |
| 133 | An ?tropomyosin mutation alters dimer preference in nemaline myopathy. Annals of Neurology, 2005, 57, 42-49. | 2.8 | 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. | 0.9 | 62 |
| 135 | Collagen VI glycine mutations: Perturbed assembly and a spectrum of clinical severity. Annals of Neurology, 2008, 64, 294-303. | 2.8 | 61 |
| 136 | Hypertrophy and dietary tyrosine ameliorate the phenotypes of a mouse model of severe nemaline myopathy. Brain, 2011, 134, 3516-3529. | 3.7 | 59 |
| 137 | Optic gliomas in neurofibromatosis type 1: Role of visual evoked potentials. Pediatric Neurology, 1994, 10, 117-123. | 1.0 | 58 |
| 138 | Dietary L-Tyrosine Supplementation in Nemaline Myopathy. Journal of Child Neurology, 2008, 23, 609-613. | 0.7 | 58 |
| 139 | Mental, Motor, and Language Development of Toddlers with Neurofibromatosis Type 1. Journal of Pediatrics, 2011, 158, 660-665. | 0.9 | 58 |
| 140 | Evidence for ACTN3 as a genetic modifier of Duchenne muscular dystrophy. Nature Communications, 2017, 8, 14143. | 5.8 | 58 |
| 141 | Diagnosis and etiology of congenital muscular dystrophy: We are halfway there. Annals of Neurology, 2016, 80, 101-111. | 2.8 | 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. | 1.9 | 57 |
| 143 | Review Article : Neurofibromatosis 1: Clinical Review and Exceptions to the Rules. Journal of Child Neurology, 2002, 17, 613-621. | 0.7 | 55 |
| 144 | Whatâ€™s new in congenital myopathies?. Neuromuscular Disorders, 2008, 18, 433-442. | 0.3 | 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. | 0.7 | 55 |
| 146 | Expanding the phenotype of GMPPB mutations. Brain, 2015, 138, 836-844. | 3.7 | 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. | 1.1 | 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. | 0.3 | 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. | 1.1 | 48 |
| 151 | Neurofibromatosis 1 in childhood. Seminars in Pediatric Neurology, 1998, 5, 231-242. | 1.0 | 47 |
| 152 | Clinical Approach to the Diagnosis of Congenital Myopathies. Seminars in Pediatric Neurology, 2011, 18, 216-220. | 1.0 | 47 |
| 153 | The effect of Î±-actinin-3 deficiency on muscle aging. Experimental Gerontology, 2011, 46, 292-302. | 1.2 | 47 |
| 154 | Outcome of children with neuromuscular disease admitted to paediatric intensive care. Archives of Disease in Childhood, 2004, 89, 170-175. | 1.0 | 46 |
| 155 | Variants in SLC18A3 , vesicular acetylcholine transporter, cause congenital myasthenic syndrome. Neurology, 2016, 87, 1442-1448. | 1.5 | 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. | 1.8 | 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. | 1.4 | 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. | 3.3 | 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. | 7.7 | 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. | 2.6 | 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. | 2.5 | 45 |

| # | Article | IF | Citations |
|---|---|---|---|
| 163 | Hand involvement in children with Charcotâ€“Marie-Tooth disease type 1A. Neuromuscular Disorders, 2008, 18, 970-973. | 0.3 | 44 |
| 164 | Cognitive dysfunction as the major presenting feature of Becker's muscular dystrophy. Neurology, 1996, 46, 461-464. | 1.5 | 43 |
| 165 | Cap disease due to mutation of the beta-tropomyosin gene (TPM2). Neuromuscular Disorders, 2009, 19, 348-351. | 0.3 | 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. | 1.1 | 43 |
| 167 | K7del is a common TPM2 gene mutation associated with nemaline myopathy and raised myofibre calcium sensitivity. Brain, 2013, 136, 494-507. | 3.7 | 42 |
| 168 | Importance and challenge of making an early diagnosis in LMNA -related muscular dystrophy. Neurology, 2012, 78, 1258-1263. | 1.5 | 41 |
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