# Hugh J Mcmillan

List of Publications by Year
in descending order



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|---|---|---|---|
| 122 papers | 3,057 citations | 159358 30 h-index | 197535 49 g-index |
| 127 all docs | 127 docs citations | 127 times ranked | 4448 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Myostatin inhibitor ACEâ€031 treatment of ambulatory boys with Duchenne muscular dystrophy: Results of a randomized, placeboâ€controlled clinical trial. Muscle and Nerve, 2017, 55, 458-464. | 1.0 | 176 |
| 2 | Hepatotoxicity following administration of onasemnogene abeparvovec (AVXS-101) for the treatment of spinal muscularÂatrophy. Journal of Hepatology, 2021, 74, 560-566. | 1.8 | 143 |
| 3 | Life expectancy at birth in Duchenne muscular dystrophy: a systematic review and meta-analysis. European Journal of Epidemiology, 2020, 35, 643-653. | 2.5 | 132 |
| 4 | The use of intravenous bisphosphonate therapy to treat vertebral fractures due to osteoporosis among boys with Duchenne muscular dystrophy. Osteoporosis International, 2012, 23, 2703-2711. | 1.3 | 115 |
| 5 | Emergence of the Primary Pediatric Stroke Center. Stroke, 2014, 45, 2018-2023. | 1.0 | 108 |
| 6 | Onasemnogene abeparvovec for presymptomatic infants with two copies of SMN2 at risk for spinal muscular atrophy type 1: the Phase III SPR1NT trial. Nature Medicine, 2022, 28, 1381-1389. | 15.2 | 99 |
| 7 | Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. Neuromuscular Disorders, 2021, 31, 574-582. | 0.3 | 94 |
| 8 | Pan-viral serology implicates enteroviruses in acute flaccid myelitis. Nature Medicine, 2019, 25, 1748-1752. | 15.2 | 93 |
| 9 | Onasemnogene abeparvovec for presymptomatic infants with three copies of SMN2 at risk for spinal muscular atrophy: the Phase III SPR1NT trial. Nature Medicine, 2022, 28, 1390-1397. | 15.2 | 93 |
| 10 | Debunking Occam's razor: Diagnosing multiple genetic diseases in families by wholeâ€exome sequencing. Clinical Genetics, 2017, 92, 281-289. | 1.0 | 92 |
| 11 | Medical studentsâ€™ perception of lesbian, gay, bisexual, and transgender (LGBT) discrimination in their learning environment and their self-reported comfort level for caring for LGBT patients: a survey study. Medical Education Online, 2017, 22, 1368850. | 1.1 | 88 |
| 12 | Abnormal fatty acid metabolism is a core component of spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1519-1532. | 1.7 | 72 |
| 13 | Serum Transaminase Levels in Boys With Duchenne and Becker Muscular Dystrophy. Pediatrics, 2011, 127, e132-e136. | 1.0 | 63 |
| 14 | Childhood chronic inflammatory demyelinating polyradiculoneuropathy: Combined analysis of a large cohort and eleven published series. Neuromuscular Disorders, 2013, 23, 103-111. | 0.3 | 62 |
| 15 | Specific combination of compound heterozygous mutations in 17Î²-hydroxysteroid dehydrogenase type 4 (HSD17B4) defines a new subtype of D-bifunctional protein deficiency. Orphanet Journal of Rare Diseases, 2012, 7, 90. | 1.2 | 60 |
| 16 | The time to and determinants of first fractures in boys with Duchenne muscular dystrophy. Osteoporosis International, 2017, 28, 597-608. | 1.3 | 59 |
| 17 | Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. Contemporary Clinical Trials, 2017, 58, 34-39. | 0.8 | 56 |
| 18 | Ophthalmoplegic Migraine: Inflammatory Neuropathy with Secondary Migraine?. Canadian Journal of Neurological Sciences, 2007, 34, 349-355. | 0.3 | 55 |

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|---|---|---|---|
| 19 | Risk of Intracranial Hemorrhage Following Intravenous tPA (Tissue-Type Plasminogen Activator) for Acute Stroke Is Low in Children. Stroke, 2020, 51, 542-548. | 1.0 | 52 |
| 20 | Brainstem compression: a predictor of postoperative cerebellar mutism. Child's Nervous System, 2009, 25, 677-681. | 0.6 | 49 |
| 21 | Bone Health and Osteoporosis Management of the Patient With Duchenne Muscular Dystrophy. Pediatrics, 2018, 142, S34-S42. | 1.0 | 48 |
| 22 | Congenital Visual Impairment and Progressive Microcephaly Due to Lysylâ€“Transfer Ribonucleic Acid (RNA) Synthetase (KARS) Mutations. Journal of Child Neurology, 2015, 30, 1037-1043. | 0.7 | 47 |
| 23 | Lumbar puncture simulation in pediatric residency training: improving procedural competence and decreasing anxiety. BMC Medical Education, 2016, 16, 198. | 1.0 | 43 |
| 24 | Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, . | 4.2 | 43 |
| 25 | Effect of Different Corticosteroid Dosing Regimens on Clinical Outcomes in Boys With Duchenne Muscular Dystrophy. JAMA - Journal of the American Medical Association, 2022, 327, 1456. | 3.8 | 43 |
| 26 | Evidence for clinical, genetic and biochemical variability in spinal muscular atrophy with progressive myoclonic epilepsy. Clinical Genetics, 2014, 86, 558-563. | 1.0 | 42 |
| 27 | Compound heterozygous mutations in glycyl-tRNA synthetase are a proposed cause of systemic mitochondrial disease. BMC Medical Genetics, 2014, 15, 36. | 2.1 | 41 |
| 28 | WATERSHED INFARCTION DUE TO ACUTE HYPEREOSINOPHILIA. Neurology, 2008, 70, 80-82. | 1.5 | 34 |
| 29 | Histomorphometry and Bone Matrix Mineralization Before and After Bisphosphonate Treatment in Boys With Duchenne Muscular Dystrophy: A Paired Transiliac Biopsy Study. Journal of Bone and Mineral Research, 2016, 31, 1060-1069. | 3.1 | 34 |
| 30 | Length of delay to birth of a second litter in dwarf hamsters (Phodopus): Evidence for post-implantation embryonic diapause. The Journal of Experimental Zoology, 1997, 278, 106-114. | 1.4 | 30 |
| 31 | Expert recommendations and clinical considerations in the use of onasemnogene abeparvovec gene therapy for spinal muscular atrophy. Muscle and Nerve, 2021, 64, 413-427. | 1.0 | 30 |
| 32 | Recessive mutations in ATP8A2 cause severe hypotonia, cognitive impairment, hyperkinetic movement disorders and progressive optic atrophy. Orphanet Journal of Rare Diseases, 2018, 13, 86. | 1.2 | 29 |
| 33 | Duchenne Muscular Dystrophy: Canadian Paediatric Neuromuscular Physicians Survey. Canadian Journal of Neurological Sciences, 2010, 37, 195-205. | 0.3 | 27 |
| 34 | Hereditary neuropathy with liability to pressure palsies in childhood: Case series and literature update. Neuromuscular Disorders, 2015, 25, 693-698. | 0.3 | 26 |
| 35 | Obesity and Endocrine Management of the Patient With Duchenne Muscular Dystrophy. Pediatrics, 2018, 142, S43-S52. | 1.0 | 26 |
| 36 | A checklist for clinical trials in rare disease: obstacles and anticipatory actionsâ€”lessons learned from the FOR-DMD trial. Trials, 2018, 19, 291. | 0.7 | 26 |

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|---|---|---|---|
| 37 | Bi-allelic variants in the mitochondrial RNase P subunit PRORP cause mitochondrial tRNA processing defects and pleiotropic multisystem presentations. American Journal of Human Genetics, 2021, 108, 2195-2204. | 2.6 | 26 |
| 38 | Association Between Chronic Aspiration and Chronic Airway Infection with Pseudomonas aeruginosa and Other Gram-Negative Bacteria in Children with Cerebral Palsy. Lung, 2016, 194, 307-314. | 1.4 | 25 |
| 39 | Autoimmune Neuromuscular Disorders in Childhood. Current Treatment Options in Neurology, 2011, 13, 590-607. | 0.7 | 24 |
| 40 | Chemotherapy-induced Peripheral Neuropathy Among Paediatric Oncology Patients. Canadian Journal of Neurological Sciences, 2014, 41, 442-447. | 0.3 | 23 |
| 41 | Divergent reproductive endocrinology of the estrous cycle and pregnancy in dwarf hamsters (Phodopus). Comparative Biochemistry and Physiology Part A, Molecular & Integrative Physiology, 1999, 124, 53-67. | 0.8 | 20 |
| 42 | Association of Early-Onset Spasticity and Risk for Cognitive Impairment With Mutations at Amino Acid 499 in SPAST. Journal of Child Neurology, 2018, 33, 329-332. | 0.7 | 20 |
| 43 | Compound heterozygous CACNA1H mutations associated with severe congenital amyotrophy. Channels, 2019, 13, 153-161. | 1.5 | 20 |
| 44 | Evolutionary Change in the Endocrinology of Behavioral Receptivity: Divergent Roles for Progesterone and Prolactin within the Genus Phodopus1. Biology of Reproduction, 1998, 59, 30-38. | 1.2 | 19 |
| 45 | Detailed Biochemical and Bioenergetic Characterization of FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion. JIMD Reports, 2015, 27, 1-9. | 0.7 | 19 |
| 46 | Disability, Quality of Life, and Pain Coping in Pediatric Migraine: An Observational Study. Journal of Child Neurology, 2017, 32, 717-724. | 0.7 | 19 |
| 47 | Chiari 1 malformation and holocord syringomyelia presenting as abrupt onset foot drop. Child's Nervous System, 2011, 27, 183-186. | 0.6 | 18 |
| 48 | Newborn Screening for Spinal Muscular Atrophy: Ontario Testing and Follow-up Recommendations. Canadian Journal of Neurological Sciences, 2021, 48, 504-511. | 0.3 | 18 |
| 49 | Acute asymmetrical spinal infarct secondary to fibrocartilaginous embolism. Child's Nervous System, 2015, 31, 487-491. | 0.6 | 17 |
| 50 | Homozygous contiguous gene deletion of 13q12 causing LGMD2C and ARSACS in the same patient. Muscle and Nerve, 2009, 39, 396-399. | 1.0 | 16 |
| 51 | The CNDR: Collaborating to Translate New Therapies for Canadians. Canadian Journal of Neurological Sciences, 2013, 40, 698-704. | 0.3 | 16 |
| 52 | Adolescent onset cognitive regression and neuropsychiatric symptoms associated with the A140V <scp>MECP</scp>2 mutation. Developmental Medicine and Child Neurology, 2014, 56, 91-94. | 1.1 | 16 |
| 53 | Autosomal recessive axonal polyneuropathy in a sibling pair due to a novel homozygous mutation in IGHMBP2. Neuromuscular Disorders, 2015, 25, 794-799. | 0.3 | 16 |
| 54 | Costs of Illness of Spinal Muscular Atrophy: A Systematic Review. Applied Health Economics and Health Policy, 2021, 19, 501-520. | 1.0 | 16 |

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|---|---|---|---|
| 55 | Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <scp>PIGQ</scp>: Report of seven new subjects and review of the literature. Journal of Inherited Metabolic Disease, 2020, 43, 1321-1332. | 1.7 | 15 |
| 56 | Novel MPZ mutations and congenital hypomyelinating neuropathy. Neuromuscular Disorders, 2010, 20, 725-729. | 0.3 | 14 |
| 57 | Utility and practice of electrodiagnostic testing in the pediatric population: An AANEM consensus statement. Muscle and Nerve, 2020, 61, 143-155. | 1.0 | 14 |
| 58 | A splice variant in ATAD3A expands the clinical and genetic spectrum of Harel-Yoon syndrome. Neurology: Genetics, 2020, 6, e452. | 0.9 | 14 |
| 59 | Survival Motor Neuron Protein is Released from Cells in Exosomes: A Potential Biomarker for Spinal Muscular Atrophy. Scientific Reports, 2017, 7, 13859. | 1.6 | 13 |
| 60 | Periodic breathing in patients with NALCN mutations. Journal of Human Genetics, 2018, 63, 1093-1096. | 1.1 | 13 |
| 61 | Novel variants in TUBA1A cause congenital fibrosis of the extraocular muscles with or without malformations of cortical brain development. European Journal of Human Genetics, 2021, 29, 816-826. | 1.4 | 13 |
| 62 | A Randomized, Double-Blind, Placebo-Controlled, Global Phase 3 Study of Edasalonexent in Pediatric Patients with Duchenne Muscular Dystrophy: Results of the PolarisDMD Trial. Journal of Neuromuscular Diseases, 2021, 8, 769-784. | 1.1 | 13 |
| 63 | Onasemnogene abeparvovec for the treatment of spinal muscular atrophy. Expert Opinion on Biological Therapy, 2022, 22, 1075-1090. | 1.4 | 13 |
| 64 | 2022 HRS expert consensus statement on evaluation and management of arrhythmic risk in neuromuscular disorders. Heart Rhythm, 2022, 19, e61-e120. | 0.3 | 13 |
| 65 | Electrophysiologic Evidence for Anterior Horn Cell Disease in Amyoplasia. Pediatric Neurology, 2010, 43, 142-147. | 1.0 | 12 |
| 66 | Pediatric sciatic neuropathy associated with neoplasms. Muscle and Nerve, 2011, 43, 183-188. | 1.0 | 12 |
| 67 | A Novel Mutation in MARS in a Patient with Charcot-Marie-Tooth Disease, Axonal, Type 2U with Congenital Onset. Journal of Neuromuscular Diseases, 2019, 6, 333-339. | 1.1 | 12 |
| 68 | Diagnosis and outcome of childhood perineurioma. Child's Nervous System, 2016, 32, 1555-1560. | 0.6 | 11 |
| 69 | The Canadian Neuromuscular Disease Registry: Connecting patients to national and international research opportunities. Paediatrics and Child Health, 2018, 23, 20-26. | 0.3 | 11 |
| 70 | Higher-Quality Data Collection Is Critical to Establish the Safety and Efficacy of Pediatric Mechanical Thrombectomy. Stroke, 2021, 52, 1213-1221. | 1.0 | 10 |
| 71 | A Report of Hereditary Neuropathy with Liability to Pressure Palsy (HNPP) Presenting with Brachial Plexopathy: The Value of Complete Electrodiagnostic Testing. American Journal of Electroneurodiagnostic Technology, 2011, 51, 183-190. | 0.3 | 9 |
| 72 | Cranial nerve hypertrophy in pediatric chronic inflammatory demyelinating polyradiculoneuropathy. Pediatric Radiology, 2010, 40, 176-176. | 1.1 | 8 |

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|---|---|---|---|
| 73 | Achalasia, chronic sensory neuropathy, and N-type calcium channel autoantibodies: beneficial response to IVIG. Clinical Journal of Gastroenterology, 2010, 3, 78-82. | 0.4 | 8 |
| 74 | Canadian Paediatric Neurology Workforce Survey and Consensus Statement. Canadian Journal of Neurological Sciences, 2016, 43, 402-409. | 0.3 | 8 |
| 75 | Myotonic Myopathy With Secondary Joint and Skeletal Anomalies From the c.2386C>G, p.L796V Mutation in SCN4A. Frontiers in Neurology, 2020, 11, 77. | 1.1 | 8 |
| 76 | Routine lung volume recruitment in boys with Duchenne muscular dystrophy: a randomised clinical trial. Thorax, 2022, 77, 805-811. | 2.7 | 8 |
| 77 | Congenital Nemaline Myopathy: The Value of Magnetic Resonance Imaging of Muscle. Canadian Journal of Neurological Sciences, 2015, 42, 338-340. | 0.3 | 7 |
| 78 | We need a â€œmade in Canadaâ€•orphan drug framework. Cmaj, 2017, 189, E1274-E1275. | 0.9 | 7 |
| 79 | Phosphoserine aminotransferase deficiency: imaging findings in a child with congenital microcephaly. Journal of Maternal-Fetal and Neonatal Medicine, 2020, 33, 1033-1035. | 0.7 | 7 |
| 80 | Characterization of physical literacy in children with chronic medical conditions compared with healthy controls: a cross-sectional study. Applied Physiology, Nutrition and Metabolism, 2021, 46, 1073-1082. | 0.9 | 7 |
| 81 | Ontario Newborn Screening for Spinal Muscular Atrophy: The First Year. Canadian Journal of Neurological Sciences, 2022, 49, 821-823. | 0.3 | 7 |
| 82 | Holocord syringomyelia presenting as rapidly progressive foot drop. Journal of Neurosciences in Rural Practice, 2011, 02, 195-196. | 0.3 | 6 |
| 83 | A National Spinal Muscular Atrophy Registry for Real-World Evidence. Canadian Journal of Neurological Sciences, 2020, 47, 810-815. | 0.3 | 6 |
| 84 | Pediatric monomelic amyotrophy: Evidence for poliomyelitis in vulnerable populations. Muscle and Nerve, 2009, 40, 860-863. | 1.0 | 5 |
| 85 | Intermittent glucocorticoid regimes for younger boys with duchenne muscular dystrophy: Balancing efficacy with side effects. Muscle and Nerve, 2019, 59, 638-639. | 1.0 | 5 |
| 86 | Inflammatory Changes in Limb Girdle Muscular Dystrophy Type 2I. Canadian Journal of Neurological Sciences, 2013, 40, 875-877. | 0.3 | 4 |
| 87 | Nusinersen: Evidence of sustained clinical improvement and lessened fatigue in older ambulatory patients with spinal muscular atrophy. Muscle and Nerve, 2020, 61, 1-2. | 1.0 | 4 |
| 88 | Health related quality of life in young, steroid-naÃ¯ve boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 1161-1168. | 0.3 | 4 |
| 89 | Brachial Plexopathy and Nonaccidental Injury: Role of the Neurologist. Journal of Child Neurology, 2010, 25, 620-622. | 0.7 | 3 |
| 90 | Subdural haemorrhage and severe coagulopathy resulting in transtentorial uncal herniation in a neonate undergoing therapeutic hypothermia. BMJ Case Reports, 2014, 2014, bcr2013203080-bcr2013203080. | 0.2 | 3 |

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|---|---|---|---|
| 91 | Mononeuritis multiplex associated with minocycline in an adolescent. Muscle and Nerve, 2017, 56, E33-E35. | 1.0 | 3 |
| 92 | Worster-Drought Syndrome Associated WithLINSMutations. Child Neurology Open, 2018, 5, 2329048X1879108. | 0.5 | 3 |
| 93 | Whole genome sequencing reveals biallelic <scp>PLA2G6</scp> mutations in siblings with cerebellar atrophy and cap myopathy. Clinical Genetics, 2021, 99, 746-748. | 1.0 | 3 |
| 94 | Pediatric Hyperacute Arterial Ischemic Stroke Pathways at Canadian Tertiary Care Hospitals. Canadian Journal of Neurological Sciences, 2021, , 1-8. | 0.3 | 3 |
| 95 | Whole genome sequencing identifies pathogenic <scp>RNU4ATAC</scp> variants in a child with recurrent encephalitis, microcephaly, and normal stature. American Journal of Medical Genetics, Part A, 2021, 185, 3502-3506. | 0.7 | 3 |
| 96 | Not so Shocking: Electromyography in Pediatrics Remains Feasible and Diagnostically Useful. Canadian Journal of Neurological Sciences, 2022, 49, 696-702. | 0.3 | 3 |
| 97 | Overview of Pediatric Peripheral Neuropathies. , 2015, , 274-288. | | 2 |
| 98 | Case 2: Gross Motor Regression in an 18-month-old Girl. Pediatrics in Review, 2017, 38, 95-95. | 0.2 | 2 |
| 99 | Family Perspectives on Visiting the Pediatric Emergency Department for Migraine. Pediatric Emergency Care, 2017, Publish Ahead of Print, e310-e317. | 0.5 | 2 |
| 100 | Response to the Canadian Agency for Drugs and Technologies in Health and Institut national dâ€™excellence en santÃ© et en services sociaux decision regarding nusinersen for Spinal Muscular Atrophy. Canadian Journal of Neurological Sciences, 2018, 45, 516-517. | 0.3 | 2 |
| 101 | â€œOwlâ€™s Eyeâ€•Sign in Acute Flaccid Paralysis. Canadian Journal of Neurological Sciences, 2019, 46, 756-757. | 0.3 | 2 |
| 102 | Muscle problems in juvenile-onset acid maltase deficiency (Pompe disease). Paediatrics and Child Health, 2019, 24, 270-271. | 0.3 | 2 |
| 103 | Developmental delay and neuropsychiatric comorbidities associated with Duchenne and Becker muscular dystrophy. Muscle and Nerve, 2020, 61, 127-128. | 1.0 | 2 |
| 104 | Drisapersen associated with elevated serum factor VIII levels in Duchenne muscular dystrophy. Neurology, 2020, 94, 538-540. | 1.5 | 2 |
| 105 | Congenital muscular dystrophies: New evidence-based guidelines for the diagnosis and management of this evolving group of muscle disorders. Muscle and Nerve, 2015, 51, 791-792. | 1.0 | 1 |
| 106 | Chronic Inflammatory Demyelinating Polyradiculoneuropathy. , 2015, , 398-417. | | 1 |
| 107 | Congenital Trismus From Brainstem Dysgenesis: Case Report and Review of Literature. Pediatrics, 2016, 138, . | 1.0 | 1 |
| 108 | Pediatric Neurology Workforce in Canada: A 5-Year Update. Canadian Journal of Neurological Sciences, 2019, 46, 566-574. | 0.3 | 1 |

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|---|---|---|---|
| 109 | 014â€...AVXS-101 gene-replacement therapy (GRT) in presymptomatic spinal muscular atrophy (SMA): study update. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, A5.3-A6. | 0.9 | 1 |
| 110 | STACKING EXERCISES ATTENUATE THE DECLINE IN FVC AND SICK TIME (STEADFAST). Chest, 2019, 156, A245-A246. | 0.4 | 1 |
| 111 | Sensitivity, specificity, and reliability of the Get Active Questionnaire for identifying children with medically necessary special considerations for physical activity. Applied Physiology, Nutrition and Metabolism, 2019, 44, 736-743. | 0.9 | 1 |
| 112 | Inhaled Solvent Abuse Mimicking Chronic Inflammatory Demyelinating Polyradiculoneuropathy. Child Neurology Open, 2020, 7, 2329048X2093491. | 0.5 | 1 |
| 113 | Neurophysiological Characteristics of Allgrove (Triple A) Syndrome: Case Report and Literature Review. Child Neurology Open, 2021, 8, 2329048X2110310. | 0.5 | 1 |
| 114 | Enhancing human aspects of care with young people with muscular dystrophy: An evaluation of a participatory qualitative study with clinicians. PLoS ONE, 2022, 17, e0263956. | 1.1 | 1 |
| 115 | Guidance on Gene Replacement Therapy in Spinal Muscular Atrophy: A Canadian Perspective. Canadian Journal of Neurological Sciences, 2021, , 1-4. | 0.3 | 0 |
| 116 | Biomarkers in Duchenne and Becker muscular dystrophies. Muscle and Nerve, 2021, 64, 4-5. | 1.0 | 0 |
| 117 | Intrinsic peripheral nerve and root tumor and pseudotumoral lesions at a tertiary care pediatric hospital. Child's Nervous System, 2021, 37, 1229-1236. | 0.6 | 0 |
| 118 | Acquired and Hereditary Neuropathies. , 2017, , 245-264. | | 0 |
| 119 | Neuromuscular Complications in the Critically Ill Child. , 2017, , 355-369. | | 0 |
| 120 | Radiculopathies and Plexopathies. , 2017, , 221-243. | | 0 |
| 121 | Commentary. Journal of Neurosciences in Rural Practice, 2011, 2, 196-7. | 0.3 | 0 |
| 122 | NTRK1-related Hereditary Sensory and Autonomic Neuropathy Type 4: The Role of the Histamine Challenge Test. Child Neurology Open, 2022, 9, 2329048X2211088. | 0.5 | 0 |