Hugh J Mcmillan

List of Publications by Citations

110 papers	1,717 citations	24 h-index	37 g-index
127 ext. papers	2,336 ext. citations	4.6 avg, IF	4.76 L-index

#	Paper	IF	Citations
110	Myostatin inhibitor ACE-031 treatment of ambulatory boys with Duchenne muscular dystrophy: Results of a randomized, placebo-controlled clinical trial. *Muscle and Nerve*, **2017**, 55, 458-464	3.4	124
109	The use of intravenous bisphosphonate therapy to treat vertebral fractures due to osteoporosis among boys with Duchenne muscular dystrophy. *Osteoporosis International*, **2012**, 23, 2703-11	5.3	95
108	Emergence of the primary pediatric stroke center: impact of the thrombolysis in pediatric stroke trial. *Stroke*, **2014**, 45, 2018-23	6.7	78
107	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by whole-exome sequencing. *Clinical Genetics*, **2017**, 92, 281-289	4	69
106	Specific combination of compound heterozygous mutations in 17β-hydroxysteroid dehydrogenase type 4 (HSD17B4) defines a new subtype of D-bifunctional protein deficiency. *Orphanet Journal of Rare Diseases*, **2012**, 7, 90	4.2	58
105	Pan-viral serology implicates enteroviruses in acute flaccid myelitis. *Nature Medicine*, **2019**, 25, 1748-1752	30.5	54
104	Medical students' 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	4.4	50
103	Serum transaminase levels in boys with Duchenne and Becker muscular dystrophy. *Pediatrics*, **2011**, 127, e132-6	7.4	50
102	The time to and determinants of first fractures in boys with Duchenne muscular dystrophy. *Osteoporosis International*, **2017**, 28, 597-608	5.3	48
101	Ophthalmoplegic migraine: inflammatory neuropathy with secondary migraine?. *Canadian Journal of Neurological Sciences*, **2007**, 34, 349-55	1	48
100	Childhood chronic inflammatory demyelinating polyradiculoneuropathy: combined analysis of a large cohort and eleven published series. *Neuromuscular Disorders*, **2013**, 23, 103-11	2.9	45
99	Life expectancy at birth in Duchenne muscular dystrophy: a systematic review and meta-analysis. *European Journal of Epidemiology*, **2020**, 35, 643-653	12.1	44
98	Compound heterozygous mutations in glycyl-tRNA synthetase are a proposed cause of systemic mitochondrial disease. *BMC Medical Genetics*, **2014**, 15, 36	2.1	40
97	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. *Contemporary Clinical Trials*, **2017**, 58, 34-39	2.3	39
96	Congenital Visual Impairment and Progressive Microcephaly Due to Lysyl-Transfer Ribonucleic Acid (RNA) Synthetase (KARS) Mutations: The Expanding Phenotype of Aminoacyl-Transfer RNA Synthetase Mutations in Human Disease. *Journal of Child Neurology*, **2015**, 30, 1037-43	2.5	39
95	Abnormal fatty acid metabolism is a core component of spinal muscular atrophy. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1519-1532	5.3	39
94	Brainstem compression: a predictor of postoperative cerebellar mutism. *Childls Nervous System*, **2009**, 25, 677-81	1.7	38

93	Evidence for clinical, genetic and biochemical variability in spinal muscular atrophy with progressive myoclonic epilepsy. *Clinical Genetics*, **2014**, 86, 558-63	4	36
92	Hepatotoxicity following administration of onasemnogene abeparvovec (AVXS-101) for the treatment of spinal muscular´atrophy. *Journal of Hepatology*, **2021**, 74, 560-566	13.4	34
91	Lumbar puncture simulation in pediatric residency training: improving procedural competence and decreasing anxiety. *BMC Medical Education*, **2016**, 16, 198	3.3	32
90	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, 1060-9	6.3	31
89	Bone Health and Osteoporosis Management of the Patient With Duchenne Muscular Dystrophy. *Pediatrics*, **2018**, 142, S34-S42	7.4	27
88	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		26
87	Watershed infarction due to acute hypereosinophilia. *Neurology*, **2008**, 70, 80-2	6.5	24
86	Duchenne muscular dystrophy: Canadian paediatric neuromuscular physicians survey. *Canadian Journal of Neurological Sciences*, **2010**, 37, 195-205	1	21
85	Risk of Intracranial Hemorrhage Following Intravenous tPA (Tissue-Type Plasminogen Activator) for Acute Stroke Is Low in Children. *Stroke*, **2020**, 51, 542-548	6.7	21
84	Hereditary neuropathy with liability to pressure palsies in childhood: Case series and literature update. *Neuromuscular Disorders*, **2015**, 25, 693-8	2.9	19
83	Autoimmune neuromuscular disorders in childhood. *Current Treatment Options in Neurology*, **2011**, 13, 590-607	4.4	18
82	A checklist for clinical trials in rare disease: obstacles and anticipatory actions-lessons learned from the FOR-DMD trial. *Trials*, **2018**, 19, 291	2.8	17
81	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	2.6	17
80	Association Between Chronic Aspiration and Chronic Airway Infection with Pseudomonas aeruginosa and Other Gram-Negative Bacteria in Children with Cerebral Palsy. *Lung*, **2016**, 194, 307-14	2.9	17
79	Acute asymmetrical spinal infarct secondary to fibrocartilaginous embolism. *Childls Nervous System*, **2015**, 31, 487-91	1.7	16
78	Recessive mutations in ATP8A2 cause severe hypotonia, cognitive impairment, hyperkinetic movement disorders and progressive optic atrophy. *Orphanet Journal of Rare Diseases*, **2018**, 13, 86	4.2	16
77	Chemotherapy-induced Peripheral Neuropathy Among Paediatric Oncology Patients. *Canadian Journal of Neurological Sciences*, **2014**, 41, 442-7	1	16
76	Evolutionary change in the endocrinology of behavioral receptivity: divergent roles for progesterone and prolactin within the genus Phodopus. *Biology of Reproduction*, **1998**, 59, 30-8	3.9	16

75	Detailed Biochemical and Bioenergetic Characterization of FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion. *JIMD Reports*, **2016**, 27, 1-9	1.9	15
74	Compound heterozygous CACNA1H mutations associated with severe congenital amyotrophy. *Channels*, **2019**, 13, 153-161	3	14
73	Adolescent onset cognitive regression and neuropsychiatric symptoms associated with the A140V MECP2 mutation. *Developmental Medicine and Child Neurology*, **2014**, 56, 91-4	3.3	14
72	The CNDR: collaborating to translate new therapies for Canadians. *Canadian Journal of Neurological Sciences*, **2013**, 40, 698-704	1	13
71	Disability, Quality of Life, and Pain Coping in Pediatric Migraine: An Observational Study. *Journal of Child Neurology*, **2017**, 32, 717-724	2.5	12
70	Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. *Neuromuscular Disorders*, **2021**, 31, 574-582	2.9	12
69	Obesity and Endocrine Management of the Patient With Duchenne Muscular Dystrophy. *Pediatrics*, **2018**, 142, S43-S52	7.4	12
68	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	2.5	11
67	Homozygous contiguous gene deletion of 13q12 causing LGMD2C and ARSACS in the same patient. *Muscle and Nerve*, **2009**, 39, 396-9	3.4	11
66	Survival Motor Neuron Protein is Released from Cells in Exosomes: A Potential Biomarker for Spinal Muscular Atrophy. *Scientific Reports*, **2017**, 7, 13859	4.9	10
65	Autosomal recessive axonal polyneuropathy in a sibling pair due to a novel homozygous mutation in IGHMBP2. *Neuromuscular Disorders*, **2015**, 25, 794-9	2.9	10
64	Diagnosis and outcome of childhood perineurioma. *Childls Nervous System*, **2016**, 32, 1555-60	1.7	10
63	Chiari 1 malformation and holocord syringomyelia presenting as abrupt onset foot drop. *Childls Nervous System*, **2011**, 27, 183-6	1.7	10
62	Novel MPZ mutations and congenital hypomyelinating neuropathy. *Neuromuscular Disorders*, **2010**, 20, 725-9	2.9	10
61	Electrophysiologic evidence for anterior horn cell disease in amyoplasia. *Pediatric Neurology*, **2010**, 43, 142-7	2.9	8
60	Cranial nerve hypertrophy in pediatric chronic inflammatory demyelinating polyradiculoneuropathy. *Pediatric Radiology*, **2010**, 40 Suppl 1, S176	2.8	8
59	Enhanced cGAS-STING-dependent interferon signaling associated with mutations in ATAD3A. *Journal of Experimental Medicine*, **2021**, 218,	16.6	8
58	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		7

57	Pediatric sciatic neuropathy associated with neoplasms. *Muscle and Nerve*, **2011**, 43, 183-8	3.4	7
56	Achalasia, chronic sensory neuropathy, and N-type calcium channel autoantibodies: beneficial response to IVIG. *Clinical Journal of Gastroenterology*, **2010**, 3, 78-82	1.1	7
55	Canadian Paediatric Neurology Workforce Survey and Consensus Statement. *Canadian Journal of Neurological Sciences*, **2016**, 43, 402-9	1	7
54	Periodic breathing in patients with NALCN mutations. *Journal of Human Genetics*, **2018**, 63, 1093-1096	4.3	7
53	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in PIGQ: Report of seven new subjects and review of the literature. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1321-1332	5.4	6
52	The Canadian Neuromuscular Disease Registry: Connecting patients to national and international research opportunities. *Paediatrics and Child Health*, **2018**, 23, 20-26	0.7	6
51	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	5	6
50	Utility and practice of electrodiagnostic testing in the pediatric population: An AANEM consensus statement. *Muscle and Nerve*, **2020**, 61, 143-155	3.4	6
49	Expert recommendations and clinical considerations in the use of onasemnogene abeparvovec gene therapy for spinal muscular atrophy. *Muscle and Nerve*, **2021**, 64, 413-427	3.4	6
48	Costs of Illness of Spinal Muscular Atrophy: A Systematic Review. *Applied Health Economics and Health Policy*, **2021**, 19, 501-520	3.4	6
47	Effect of Different Corticosteroid Dosing Regimens on Clinical Outcomes in Boys With Duchenne Muscular Dystrophy: A Randomized Clinical Trial.. *JAMA - Journal of the American Medical Association*, **2022**,	27.4	6
46	Pediatric monomelic amyotrophy: evidence for poliomyelitis in vulnerable populations. *Muscle and Nerve*, **2009**, 40, 860-3	3.4	5
45	Holocord syringomyelia presenting as rapidly progressive foot drop. *Journal of Neurosciences in Rural Practice*, **2011**, 2, 195-6	1.1	5
44	Phosphoserine aminotransferase deficiency: imaging findings in a child with congenital microcephaly. *Journal of Maternal-Fetal and Neonatal Medicine*, **2020**, 33, 1033-1035	2	5
43	We need a "made in Canada" orphan drug framework. *Cmaj*, **2017**, 189, E1274-E1275	3.5	4
42	A splice variant in expands the clinical and genetic spectrum of Harel-Yoon syndrome. *Neurology: Genetics*, **2020**, 6, e452	3.8	4
41	Newborn Screening for Spinal Muscular Atrophy: Ontario Testing and Follow-up Recommendations. *Canadian Journal of Neurological Sciences*, **2021**, 48, 504-511	1	4
40	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	5	4

39	A National Spinal Muscular Atrophy Registry for Real-World Evidence. *Canadian Journal of Neurological Sciences*, **2020**, 47, 810-815	1	3
38	Myotonic Myopathy With Secondary Joint and Skeletal Anomalies From the c.2386C>G, p.L769V Mutation in. *Frontiers in Neurology*, **2020**, 11, 77	4.1	3
37	Congenital Nemaline Myopathy: The Value of Magnetic Resonance Imaging of Muscle. *Canadian Journal of Neurological Sciences*, **2015**, 42, 338-40	1	3
36	Subdural haemorrhage and severe coagulopathy resulting in transtentorial uncal herniation in a neonate undergoing therapeutic hypothermia. *BMJ Case Reports*, **2014**, 2014,	0.9	3
35	Inflammatory Changes in Limb Girdle Muscular Dystrophy Type 2I. *Canadian Journal of Neurological Sciences*, **2013**, 40, 875-7	1	3
34	Brachial plexopathy and nonaccidental injury: role of the neurologist. *Journal of Child Neurology*, **2010**, 25, 620-2	2.5	3
33	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	11	3
32	Case 2: Gross Motor Regression in an 18-month-old Girl. *Pediatrics in Review*, **2017**, 38, 95	1.1	2
31	Higher-Quality Data Collection Is Critical to Establish the Safety and Efficacy of Pediatric Mechanical Thrombectomy. *Stroke*, **2021**, 52, 1213-1221	6.7	2
30	Worster-Drought Syndrome Associated With Mutations. *Child Neurology Open*, **2018**, 5, 2329048X18791083	1.3	2
29	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	3	2
28	Chronic Inflammatory Demyelinating Polyradiculoneuropathy **2015**, 398-417		1
27	Overview of Pediatric Peripheral Neuropathies **2015**, 274-288		1
26	Drisapersen associated with elevated serum factor VIII levels in Duchenne muscular dystrophy. *Neurology*, **2020**, 94, 538-540	6.5	1
25	Congenital Trismus From Brainstem Dysgenesis: Case Report and Review of Literature. *Pediatrics*, **2016**, 138,	7.4	1
24	Response to the Canadian Agency for Drugs and Technologies in Health and Institut national dR excellence en sant et en services sociaux decision regarding nusinersen for Spinal Muscular Atrophy. *Canadian Journal of Neurological Sciences*, **2018**, 45, 516-517	1	1
23	Pediatric Neurology Workforce in Canada: A 5-Year Update. *Canadian Journal of Neurological Sciences*, **2019**, 46, 566-574	1	1
22	"Owl's Eye" Sign in Acute Flaccid Paralysis. *Canadian Journal of Neurological Sciences*, **2019**, 46, 756-757	1	1

21	Muscle problems in juvenile-onset acid maltase deficiency (Pompe disease). *Paediatrics and Child Health*, **2019**, 24, 270-271	0.7	1
20	Family Perspectives on Visiting the Pediatric Emergency Department for Migraine: A Qualitative Study. *Pediatric Emergency Care*, **2020**, 36, e310-e317	1.4	1
19	Inhaled Solvent Abuse Mimicking Chronic Inflammatory Demyelinating Polyradiculoneuropathy. *Child Neurology Open*, **2020**, 7, 2329048X20934914	1.3	1
18	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	5.3	1
17	Whole genome sequencing reveals biallelic PLA2G6 mutations in siblings with cerebellar atrophy and cap myopathy. *Clinical Genetics*, **2021**, 99, 746-748	4	1
16	Routine lung volume recruitment in boys with Duchenne muscular dystrophy: a randomised clinical trial.. *Thorax*, **2022**,	7.3	1
15	Mononeuritis multiplex associated with minocycline in an adolescent. *Muscle and Nerve*, **2017**, 56, E33-E35	3.4	0
14	Neurophysiological Characteristics of Allgrove (Triple A) Syndrome: Case Report and Literature Review. *Child Neurology Open*, **2021**, 8, 2329048X211031059	1.3	0
13	014 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	5.5	0
12	Pediatric Hyperacute Arterial Ischemic Stroke Pathways at Canadian Tertiary Care Hospitals. *Canadian Journal of Neurological Sciences*, **2021**, 1-8	1	0
11	Whole genome sequencing identifies pathogenic RNU4ATAC variants in a child with recurrent encephalitis, microcephaly, and normal stature. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3502-3506	2.5	0
10	Not so Shocking: Electromyography in Pediatrics Remains Feasible and Diagnostically Useful. *Canadian Journal of Neurological Sciences*, **2021**, 1-7	1	0
9	Commentary. *Journal of Neurosciences in Rural Practice*, **2011**, 2, 196-7	1.1	
8	Ontario Newborn Screening for Spinal Muscular Atrophy: The First Year. *Canadian Journal of Neurological Sciences*, **2021**, 1-7	1	
7	Acquired and Hereditary Neuropathies **2017**, 245-264		
6	Neuromuscular Complications in the Critically Ill Child **2017**, 355-369		
5	Radiculopathies and Plexopathies **2017**, 221-243		
4	Guidance on Gene Replacement Therapy in Spinal Muscular Atrophy: A Canadian Perspective. *Canadian Journal of Neurological Sciences*, **2021**, 1-4	1	

3	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	3
2	Intrinsic peripheral nerve and root tumor and pseudotumoral lesions at a tertiary care pediatric hospital. *Childls Nervous System*, **2021**, 37, 1229-1236	1.7
1	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	3.7