Hugh J Mcmillan

List of Publications by Year in Descending Order

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	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	
109	Routine lung volume recruitment in boys with Duchenne muscular dystrophy: a randomised clinical trial.. *Thorax*, **2022**,	7.3	1
108	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
107	Neurophysiological Characteristics of Allgrove (Triple A) Syndrome: Case Report and Literature Review. *Child Neurology Open*, **2021**, 8, 2329048X211031059	1.3	0
106	Ontario Newborn Screening for Spinal Muscular Atrophy: The First Year. *Canadian Journal of Neurological Sciences*, **2021**, 1-7	1	
105	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
104	Newborn Screening for Spinal Muscular Atrophy: Ontario Testing and Follow-up Recommendations. *Canadian Journal of Neurological Sciences*, **2021**, 48, 504-511	1	4
103	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
102	Higher-Quality Data Collection Is Critical to Establish the Safety and Efficacy of Pediatric Mechanical Thrombectomy. *Stroke*, **2021**, 52, 1213-1221	6.7	2
101	Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. *Neuromuscular Disorders*, **2021**, 31, 574-582	2.9	12
100	Guidance on Gene Replacement Therapy in Spinal Muscular Atrophy: A Canadian Perspective. *Canadian Journal of Neurological Sciences*, **2021**, 1-4	1	
99	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
98	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
97	Whole genome sequencing reveals biallelic PLA2G6 mutations in siblings with cerebellar atrophy and cap myopathy. *Clinical Genetics*, **2021**, 99, 746-748	4	1
96	Pediatric Hyperacute Arterial Ischemic Stroke Pathways at Canadian Tertiary Care Hospitals. *Canadian Journal of Neurological Sciences*, **2021**, 1-8	1	0
95	Costs of Illness of Spinal Muscular Atrophy: A Systematic Review. *Applied Health Economics and Health Policy*, **2021**, 19, 501-520	3.4	6
94	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

93	Not so Shocking: Electromyography in Pediatrics Remains Feasible and Diagnostically Useful. *Canadian Journal of Neurological Sciences*, **2021**, 1-7	1	0
92	Enhanced cGAS-STING-dependent interferon signaling associated with mutations in ATAD3A. *Journal of Experimental Medicine*, **2021**, 218,	16.6	8
91	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
90	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
89	Intrinsic peripheral nerve and root tumor and pseudotumoral lesions at a tertiary care pediatric hospital. *Childls Nervous System*, **2021**, 37, 1229-1236	1.7	
88	A National Spinal Muscular Atrophy Registry for Real-World Evidence. *Canadian Journal of Neurological Sciences*, **2020**, 47, 810-815	1	3
87	A splice variant in expands the clinical and genetic spectrum of Harel-Yoon syndrome. *Neurology: Genetics*, **2020**, 6, e452	3.8	4
86	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
85	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
84	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
83	Drisapersen associated with elevated serum factor VIII levels in Duchenne muscular dystrophy. *Neurology*, **2020**, 94, 538-540	6.5	1
82	Family Perspectives on Visiting the Pediatric Emergency Department for Migraine: A Qualitative Study. *Pediatric Emergency Care*, **2020**, 36, e310-e317	1.4	1
81	Utility and practice of electrodiagnostic testing in the pediatric population: An AANEM consensus statement. *Muscle and Nerve*, **2020**, 61, 143-155	3.4	6
80	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
79	Inhaled Solvent Abuse Mimicking Chronic Inflammatory Demyelinating Polyradiculoneuropathy. *Child Neurology Open*, **2020**, 7, 2329048X20934914	1.3	1
78	Phosphoserine aminotransferase deficiency: imaging findings in a child with congenital microcephaly. *Journal of Maternal-Fetal and Neonatal Medicine*, **2020**, 33, 1033-1035	2	5
77	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
76	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

75	Pediatric Neurology Workforce in Canada: A 5-Year Update. *Canadian Journal of Neurological Sciences*, **2019**, 46, 566-574	1	1
74	Compound heterozygous CACNA1H mutations associated with severe congenital amyotrophy. *Channels*, **2019**, 13, 153-161	3	14
73	"Owl's Eye" Sign in Acute Flaccid Paralysis. *Canadian Journal of Neurological Sciences*, **2019**, 46, 756-757	1	1
72	Muscle problems in juvenile-onset acid maltase deficiency (Pompe disease). *Paediatrics and Child Health*, **2019**, 24, 270-271	0.7	1
71	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
70	Pan-viral serology implicates enteroviruses in acute flaccid myelitis. *Nature Medicine*, **2019**, 25, 1748-1752	30.5	54
69	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	
68	The Canadian Neuromuscular Disease Registry: Connecting patients to national and international research opportunities. *Paediatrics and Child Health*, **2018**, 23, 20-26	0.7	6
67	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
66	Response to the Canadian Agency for Drugs and Technologies in Health and Institut national d'excellence en santé et en services sociaux decision regarding nusinersen for Spinal Muscular Atrophy. *Canadian Journal of Neurological Sciences*, **2018**, 45, 516-517	1	1
65	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
64	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
63	Bone Health and Osteoporosis Management of the Patient With Duchenne Muscular Dystrophy. *Pediatrics*, **2018**, 142, S34-S42	7.4	27
62	Obesity and Endocrine Management of the Patient With Duchenne Muscular Dystrophy. *Pediatrics*, **2018**, 142, S43-S52	7.4	12
61	Worster-Drought Syndrome Associated With Mutations. *Child Neurology Open*, **2018**, 5, 2329048X18791083	1.3	2
60	Periodic breathing in patients with NALCN mutations. *Journal of Human Genetics*, **2018**, 63, 1093-1096	4.3	7
59	Case 2: Gross Motor Regression in an 18-month-old Girl. *Pediatrics in Review*, **2017**, 38, 95	1.1	2
58	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by whole-exome sequencing. *Clinical Genetics*, **2017**, 92, 281-289	4	69

57	Disability, Quality of Life, and Pain Coping in Pediatric Migraine: An Observational Study. *Journal of Child Neurology*, **2017**, 32, 717-724	2.5	12
56	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. *Contemporary Clinical Trials*, **2017**, 58, 34-39	2.3	39
55	Mononeuritis multiplex associated with minocycline in an adolescent. *Muscle and Nerve*, **2017**, 56, E33-E35	3.4	0
54	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
53	We need a "made in Canada" orphan drug framework. *Cmaj*, **2017**, 189, E1274-E1275	3.5	4
52	Medical studentsRperception 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
51	The time to and determinants of first fractures in boys with Duchenne muscular dystrophy. *Osteoporosis International*, **2017**, 28, 597-608	5.3	48
50	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
49	Acquired and Hereditary Neuropathies **2017**, 245-264		
48	Neuromuscular Complications in the Critically Ill Child **2017**, 355-369		
47	Radiculopathies and Plexopathies **2017**, 221-243		
46	Detailed Biochemical and Bioenergetic Characterization of FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion. *JIMD Reports*, **2016**, 27, 1-9	1.9	15
45	Lumbar puncture simulation in pediatric residency training: improving procedural competence and decreasing anxiety. *BMC Medical Education*, **2016**, 16, 198	3.3	32
44	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
43	Congenital Trismus From Brainstem Dysgenesis: Case Report and Review of Literature. *Pediatrics*, **2016**, 138,	7.4	1
42	Diagnosis and outcome of childhood perineurioma. *Childls Nervous System*, **2016**, 32, 1555-60	1.7	10
41	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
40	Canadian Paediatric Neurology Workforce Survey and Consensus Statement. *Canadian Journal of Neurological Sciences*, **2016**, 43, 402-9	1	7

39	Hereditary neuropathy with liability to pressure palsies in childhood: Case series and literature update. *Neuromuscular Disorders*, **2015**, 25, 693-8	2.9	19
38	Acute asymmetrical spinal infarct secondary to fibrocartilaginous embolism. *Childls Nervous System*, **2015**, 31, 487-91	1.7	16
37	Chronic Inflammatory Demyelinating Polyradiculoneuropathy **2015**, 398-417		1
36	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
35	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
34	Overview of Pediatric Peripheral Neuropathies **2015**, 274-288		1
33	Congenital Nemaline Myopathy: The Value of Magnetic Resonance Imaging of Muscle. *Canadian Journal of Neurological Sciences*, **2015**, 42, 338-40	1	3
32	Compound heterozygous mutations in glycyl-tRNA synthetase are a proposed cause of systemic mitochondrial disease. *BMC Medical Genetics*, **2014**, 15, 36	2.1	40
31	Emergence of the primary pediatric stroke center: impact of the thrombolysis in pediatric stroke trial. *Stroke*, **2014**, 45, 2018-23	6.7	78
30	Chemotherapy-induced Peripheral Neuropathy Among Paediatric Oncology Patients. *Canadian Journal of Neurological Sciences*, **2014**, 41, 442-7	1	16
29	Subdural haemorrhage and severe coagulopathy resulting in transtentorial uncal herniation in a neonate undergoing therapeutic hypothermia. *BMJ Case Reports*, **2014**, 2014,	0.9	3
28	Evidence for clinical, genetic and biochemical variability in spinal muscular atrophy with progressive myoclonic epilepsy. *Clinical Genetics*, **2014**, 86, 558-63	4	36
27	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
26	Childhood chronic inflammatory demyelinating polyradiculoneuropathy: combined analysis of a large cohort and eleven published series. *Neuromuscular Disorders*, **2013**, 23, 103-11	2.9	45
25	The CNDR: collaborating to translate new therapies for Canadians. *Canadian Journal of Neurological Sciences*, **2013**, 40, 698-704	1	13
24	Inflammatory Changes in Limb Girdle Muscular Dystrophy Type 2I. *Canadian Journal of Neurological Sciences*, **2013**, 40, 875-7	1	3
23	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
22	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

21	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
20	Chiari 1 malformation and holocord syringomyelia presenting as abrupt onset foot drop. *Childls Nervous System*, **2011**, 27, 183-6	1.7	10
19	Autoimmune neuromuscular disorders in childhood. *Current Treatment Options in Neurology*, **2011**, 13, 590-607	4.4	18
18	Pediatric sciatic neuropathy associated with neoplasms. *Muscle and Nerve*, **2011**, 43, 183-8	3.4	7
17	Serum transaminase levels in boys with Duchenne and Becker muscular dystrophy. *Pediatrics*, **2011**, 127, e132-6	7.4	50
16	Holocord syringomyelia presenting as rapidly progressive foot drop. *Journal of Neurosciences in Rural Practice*, **2011**, 2, 195-6	1.1	5
15	Commentary. *Journal of Neurosciences in Rural Practice*, **2011**, 2, 196-7	1.1	
14	Duchenne muscular dystrophy: Canadian paediatric neuromuscular physicians survey. *Canadian Journal of Neurological Sciences*, **2010**, 37, 195-205	1	21
13	Brachial plexopathy and nonaccidental injury: role of the neurologist. *Journal of Child Neurology*, **2010**, 25, 620-2	2.5	3
12	Novel MPZ mutations and congenital hypomyelinating neuropathy. *Neuromuscular Disorders*, **2010**, 20, 725-9	2.9	10
11	Electrophysiologic evidence for anterior horn cell disease in amyoplasia. *Pediatric Neurology*, **2010**, 43, 142-7	2.9	8
10	Cranial nerve hypertrophy in pediatric chronic inflammatory demyelinating polyradiculoneuropathy. *Pediatric Radiology*, **2010**, 40 Suppl 1, S176	2.8	8
9	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
8	Homozygous contiguous gene deletion of 13q12 causing LGMD2C and ARSACS in the same patient. *Muscle and Nerve*, **2009**, 39, 396-9	3.4	11
7	Pediatric monomelic amyotrophy: evidence for poliomyelitis in vulnerable populations. *Muscle and Nerve*, **2009**, 40, 860-3	3.4	5
6	Brainstem compression: a predictor of postoperative cerebellar mutism. *Childls Nervous System*, **2009**, 25, 677-81	1.7	38
5	Watershed infarction due to acute hypereosinophilia. *Neurology*, **2008**, 70, 80-2	6.5	24
4	Ophthalmoplegic migraine: inflammatory neuropathy with secondary migraine?. *Canadian Journal of Neurological Sciences*, **2007**, 34, 349-55	1	48

3	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
2	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
1	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