Michelle A Farrar Mbbs

List of Publications by Year
in descending order

87	2,845	186265 28	206112 48
papers	citations	h-index	g-index

91	91	91	3134
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Multidisciplinary perspectives and practices of wheelchair prescription for children with neuromuscular conditions. Disability and Rehabilitation: Assistive Technology, 2023, 18, 166-174.	2.2	3
2	Integrating newborn screening for spinal muscular atrophy into health care systems: an Australian pilot programme. Developmental Medicine and Child Neurology, 2022, 64, 625-632.	2.1	34
3	Restoration of Nusinersen Levels Following Treatment Interruption in People With Spinal Muscular Atrophy: Simulations Based on a Population Pharmacokinetic Model. CNS Drugs, 2022, 36, 181-190.	5.9	6
4	The changing therapeutic landscape of spinal muscular atrophy. Australian Journal of General Practice, 2022, 51, 38-42.	0.8	5
5	Clinical practice guideline for the management of paediatric Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 530-538.	1.9	10
6	Onasemnogene abeparvovec in spinal muscular atrophy: an Australian experience of safety and efficacy. Annals of Clinical and Translational Neurology, 2022, 9, 339-350.	3.7	32
7	Ethical aspects of the changing landscape for spinal muscular atrophy management in Australia. Australian Journal of General Practice, 2022, 51, 131-135.	0.8	2
8	Axonal excitability changes in children with spinal muscular atrophy treated with nusinersen. Journal of Physiology, 2022, 600, 95-109.	2.9	7
9	Family, healthcare professional, and societal preferences for the treatment of infantile spinal muscular atrophy: A discrete choice experiment. Developmental Medicine and Child Neurology, 2022, 64, 753-761.	2.1	5
10	The involvement of rare disease patient organisations in therapeutic innovation across rare paediatric neurological conditions: a narrative review. Orphanet Journal of Rare Diseases, 2022, 17, 167.	2.7	9
11	Onasemnogene abeparvovec for the treatment of spinal muscular atrophy. Expert Opinion on Biological Therapy, 2022, 22, 1075-1090.	3.1	13
12	Delivering paediatric precision medicine: Genomic and environmental considerations along the causal pathway of childhood neurodevelopmental disorders. Developmental Medicine and Child Neurology, 2022, 64, 1077-1084.	2.1	7
13	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.	30.7	99
14	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.	30.7	93
15	Real-world respiratory and bulbar comorbidities of SMA type 1 children treated with nusinersen: 2-Year single centre Australian experience. Paediatric Respiratory Reviews, 2021, 39, 54-60.	1.8	16
16	Congenital subpendymal giant cell astrocytoma in children with tuberous sclerosis complex: growth patterns and neurological outcome. Pediatric Research, 2021, 89, 1447-1451.	2.3	5
17	Thrombotic Microangiopathy Following Onasemnogene Abeparvovec for Spinal Muscular Atrophy: A Case Series. Journal of Pediatrics, 2021, 231, 265-268.	1.8	107
18	Motor unit changes in children with symptomatic spinal muscular atrophy treated with nusinersen. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 78-85.	1.9	33

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19	â€œWe needed thisâ€¦ perspectives of parents and healthcare professionals involved in a pilot newborn screening program for spinal muscular atrophy. EClinicalMedicine, 2021, 33, 100742.	7.1	22
20	Population pharmacokinetics-based recommendations for a single delayed or missed dose of nusinersen. Neuromuscular Disorders, 2021, 31, 310-318.	0.6	10
21	Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. Neuromuscular Disorders, 2021, 31, 574-582.	0.6	94
22	Growth and nutrition in pediatric neuromuscular disorders. Clinical Nutrition, 2021, 40, 4341-4348.	5.0	12
23	Expert recommendations and clinical considerations in the use of onasemnogene abeparvovec gene therapy for spinal muscular atrophy. Muscle and Nerve, 2021, 64, 413-427.	2.2	30
24	Clinically Responsive Genomic Analysis Pipelines. Journal of Molecular Diagnostics, 2021, 23, 894-905.	2.8	10
25	Newborn screening for spinal muscular atrophy with disease-modifying therapies: a cost-effectiveness analysis. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 1296-1304.	1.9	40
26	â€œItâ€™s not just the wheelchair, itâ€™s everything elseâ€¦ Australian parentsâ€™ perspectives of wheelchair prescription for children with neuromuscular disorders. Disability and Rehabilitation, 2020, 42, 3457-3466.	1.8	8
27	The implementation of newborn screening for spinal muscular atrophy: the Australian experience. Genetics in Medicine, 2020, 22, 557-565.	2.4	90
28	Next generation sequencing of human enterovirus strains from an outbreak of enterovirus A71 shows applicability to outbreak investigations. Journal of Clinical Virology, 2020, 122, 104216.	3.1	4
29	Spinal muscular atrophy â€” the dawning of a new era. Nature Reviews Neurology, 2020, 16, 593-594.	10.1	12
30	Personalized medicine for children with spinal muscular atrophy: Toward the holy grail. Muscle and Nerve, 2020, 62, 425-426.	2.2	2
31	Peripheral nerve maturation and excitability properties from early childhood: Comparison of motor and sensory nerves. Clinical Neurophysiology, 2020, 131, 2452-2459.	1.5	3
32	Treating adults with spinal muscular atrophy with nusinersen. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1139-1139.	1.9	1
33	Great expectations: virus-mediated gene therapy in neurological disorders. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 849-860.	1.9	16
34	Prenusinersen economic and health-related quality of life burden of spinal muscular atrophy. Neurology, 2020, 95, e1-e10.	1.1	30
35	Health, wellbeing and lived experiences of adults with SMA: a scoping systematic review. Orphanet Journal of Rare Diseases, 2020, 15, 70.	2.7	32
36	Nutritional practices in pediatric patients with neuromuscular disorders. Nutrition Reviews, 2020, 78, 857-865.	5.8	5

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37	â€œThe Whole Game is Changing and Youâ€™ve Got Hopeâ€: Australian Perspectives on Treatment Decision Making in Spinal Muscular Atrophy. Patient, 2020, 13, 389-400.	2.7	22
38	Parentsâ€™ perceptions of power wheelchair prescription for children with a neuromuscular disorder: a scoping review. Disability and Rehabilitation, 2019, 41, 2750-2757.	1.8	7
39	Myotonic dystrophy type 1: clinical manifestations in children and adolescents. Archives of Disease in Childhood, 2019, 104, 48-52.	1.9	31
40	An Integrated Safety Analysis of Infants and Children with Symptomatic Spinal Muscular Atrophy (SMA) Treated with Nusinersen in Seven Clinical Trials. CNS Drugs, 2019, 33, 919-932.	5.9	69
41	Biomarkers and the Development of a Personalized Medicine Approach in Spinal Muscular Atrophy. Frontiers in Neurology, 2019, 10, 898.	2.4	49
42	Importance of muscle biopsy to establish pathogenicity of DMD missense and splice variants. Neuromuscular Disorders, 2019, 29, 913-919.	0.6	19
43	â€œGetting ready for the adult worldâ€: how adults with spinal muscular atrophy perceive and experience healthcare, transition and well-being. Orphanet Journal of Rare Diseases, 2019, 14, 74.	2.7	27
44	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.	1.9	1
45	066â€…Avxs-101 gene-replacement therapy (GRT) for spinal muscular atrophy type 1 (SMA1): pivotal phase 3 study (STR1VE) update. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, A22.1-A22.	1.9	3
46	Onasemnogene Abeparvovec Gene-Replacement Therapy (GRT) in Pre-symptomatic Spinal Muscular Atrophy (SMA): SPR1NT Study Update. Neuropediatrics, 2019, 50, .	0.6	0
47	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
48	New and developing therapies in spinal muscular atrophy. Paediatric Respiratory Reviews, 2018, 28, 3-10.	1.8	15
49	Integrating exome sequencing into a diagnostic pathway for epileptic encephalopathy: Evidence of clinical utility and cost effectiveness. Molecular Genetics & Genomic Medicine, 2018, 6, 186-199.	1.2	83
50	Nusinersen for SMA: expanded access programme. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 937-942.	1.9	46
51	Multimodal quantitative examination of nerve function in colorectal cancer patients prior to chemotherapy. Muscle and Nerve, 2018, 57, 615-621.	2.2	2
52	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. Brain, 2018, 141, 3319-3330.	7.6	25
53	Financial, opportunity and psychosocial costs of spinal muscular atrophy: an exploratory qualitative analysis of Australian carer perspectives. BMJ Open, 2018, 8, e020907.	1.9	45
54	Cannabis for paediatric epilepsy: challenges and conundrums. Medical Journal of Australia, 2018, 208, 132-136.	1.7	6

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55	Spinal muscular atrophy: A modifiable disease emerges. Paediatric Respiratory Reviews, 2018, 28, 1-2.	1.8	2
56	Cannabidiol for treating drugâ€resistant epilepsy in children: the New South Wales experience. Medical Journal of Australia, 2018, 209, 217-221.	1.7	28
57	Screening for spinal muscular atrophy. Medical Journal of Australia, 2018, 209, 147-148.	1.7	2
58	Quality of life and excessive daytime sleepiness in children and adolescents with myotonic dystrophy type 1. Sleep Medicine, 2017, 32, 92-96.	1.6	13
59	Neurophysiological and clinical outcomes in chemotherapy-induced neuropathy in cancer. Clinical Neurophysiology, 2017, 128, 1166-1175.	1.5	50
60	Burning pain: axonal dysfunction in erythromelalgia. Pain, 2017, 158, 900-911.	4.2	11
61	Emerging therapies and challenges in spinal muscular atrophy. Annals of Neurology, 2017, 81, 355-368.	5.3	157
62	Safety and efficacy of progressive resistance exercise for Charcot-Marie-Tooth disease in children: a randomised, double-blind, sham-controlled trial. The Lancet Child and Adolescent Health, 2017, 1, 106-113.	5.6	39
63	Inherited Paediatric Motor Neuron Disorders: Beyond Spinal Muscular Atrophy. Neural Plasticity, 2017, 2017, 1-22.	2.2	18
64	Approaches to genetic diagnosis in neuromuscular conditions in the era of next generation sequencing. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 1384-1385.	1.9	4
65	Polyarticular Arthritis and Spinal Muscular Atrophy in Acid Ceramidase Deficiency. Pediatrics, 2016, 138, .	2.1	15
66	Motor cortical dysfunction develops in spinocerebellar ataxia type 3. Clinical Neurophysiology, 2016, 127, 3418-3424.	1.5	22
67	The relationship of body habitus and respiratory function in Duchenne muscular dystrophy. Respiratory Medicine, 2016, 119, 35-40.	2.9	11
68	Novel motor phenotypes in patients with VRK1 mutations without pontocerebellar hypoplasia. Neurology, 2016, 87, 65-70.	1.1	38
69	Glial mitochondropathy in infantile neuroaxonal dystrophy: pathophysiological and therapeutic implications. Brain, 2016, 139, e67-e67.	7.6	4
70	Neurophysiological profile of peripheral neuropathy associated with childhood mitochondrial disease. Mitochondrion, 2016, 30, 162-167.	3.4	13
71	Pediatric chemotherapy induced peripheral neuropathy: A systematic review of current knowledge. Cancer Treatment Reviews, 2016, 50, 118-128.	7.7	69
72	Fracture in Duchenne Muscular Dystrophy. Journal of Child Neurology, 2016, 31, 1181-1187.	1.4	34

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73	Clinical Characteristics and Functional Motor Outcomes of Enterovirus 71 Neurological Disease in Children. JAMA Neurology, 2016, 73, 300.	9.0	106
74	Pathophysiology of motor dysfunction in a childhood motor neuron disease caused by mutations in the riboflavin transporter. Clinical Neurophysiology, 2016, 127, 911-918.	1.5	22
75	Whole Genome Sequencing Identifies a 78 kb Insertion from Chromosome 8 as the Cause of Charcot-Marie-Tooth Neuropathy CMTX3. PLoS Genetics, 2016, 12, e1006177.	3.5	20
76	Congenital and childhood myotonic dystrophy: Current aspects of disease and future directions. World Journal of Clinical Pediatrics, 2015, 4, 66.	2.1	199
77	The Genetics of Spinal Muscular Atrophy: Progress and Challenges. Neurotherapeutics, 2015, 12, 290-302.	4.4	110
78	Axonal dysfunction, dysmyelination, and conduction failure in hereditary neuropathy with liability to pressure palsies. Muscle and Nerve, 2014, 49, 858-865.	2.2	14
79	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
80	Pathophysiological Insights Derived by Natural History and Motor Function of Spinal Muscular Atrophy. Journal of Pediatrics, 2013, 162, 155-159.	1.8	104
81	Evolution of peripheral nerve function in humans: novel insights from motor nerve excitability. Journal of Physiology, 2013, 591, 273-286.	2.9	24
82	Corticomotoneuronal Integrity and Adaptation in Spinal Muscular Atrophy. Archives of Neurology, 2012, 69, 467.	4.5	15
83	Dysfunction of axonal membrane conductances in adolescents and young adults with spinal muscular atrophy. Brain, 2011, 134, 3185-3197.	7.6	35
84	Acute, Reversible Axonal Energy Failure During Stroke-Like Episodes in MELAS. Pediatrics, 2010, 126, e734-e739.	2.1	17
85	Teaching paediatric epilepsy to medical students: A randomised crossover trial. Journal of Paediatrics and Child Health, 2009, 45, 727-730.	0.8	10
86	Teaching doctors how to diagnose paroxysmal events: a comparison of two educational methods. Medical Education, 2008, 42, 909-914.	2.1	12
87	Combined Endovascular and Open Surgery for Four-Vessel Cerebrovascular Occlusive Disease. Journal of Endovascular Therapy, 2001, 8, 62-66.	1.5	29