Michelle A Farrar Mbbs

List of Publications by Year
in descending order

87 papers	2,845 citations	186265 28 h-index	206112 48 g-index
91 all docs	91 docs citations	91 times ranked	3134 citing authors

#	Article	IF	Citations
1	Congenital and childhood myotonic dystrophy: Current aspects of disease and future directions. World Journal of Clinical Pediatrics, 2015, 4, 66.	2.1	199
2	Emerging therapies and challenges in spinal muscular atrophy. Annals of Neurology, 2017, 81, 355-368.	5.3	157
3	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
4	The Genetics of Spinal Muscular Atrophy: Progress and Challenges. Neurotherapeutics, 2015, 12, 290-302.	4.4	110
5	Thrombotic Microangiopathy Following Onasemnogene Abeparvovec for Spinal Muscular Atrophy: A Case Series. Journal of Pediatrics, 2021, 231, 265-268.	1.8	107
6	Clinical Characteristics and Functional Motor Outcomes of Enterovirus 71 Neurological Disease in Children. JAMA Neurology, 2016, 73, 300.	9.0	106
7	Pathophysiological Insights Derived by Natural History and Motor Function of Spinal Muscular Atrophy. Journal of Pediatrics, 2013, 162, 155-159.	1.8	104
8	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
9	Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. Neuromuscular Disorders, 2021, 31, 574-582.	0.6	94
10	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
11	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
12	The implementation of newborn screening for spinal muscular atrophy: the Australian experience. Genetics in Medicine, 2020, 22, 557-565.	2.4	90
13	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
14	Pediatric chemotherapy induced peripheral neuropathy: A systematic review of current knowledge. Cancer Treatment Reviews, 2016, 50, 118-128.	7.7	69
15	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
16	Neurophysiological and clinical outcomes in chemotherapy-induced neuropathy in cancer. Clinical Neurophysiology, 2017, 128, 1166-1175.	1.5	50
17	Biomarkers and the Development of a Personalized Medicine Approach in Spinal Muscular Atrophy. Frontiers in Neurology, 2019, 10, 898.	2.4	49
18	Nusinersen for SMA: expanded access programme. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 937-942.	1.9	46

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19	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
20	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
21	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
22	Novel motor phenotypes in patients with VRK1 mutations without pontocerebellar hypoplasia. Neurology, 2016, 87, 65-70.	1.1	38
23	Dysfunction of axonal membrane conductances in adolescents and young adults with spinal muscular atrophy. Brain, 2011, 134, 3185-3197.	7.6	35
24	Fracture in Duchenne Muscular Dystrophy. Journal of Child Neurology, 2016, 31, 1181-1187.	1.4	34
25	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
26	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
27	Health, wellbeing and lived experiences of adults with SMA: a scoping systematic review. Orphanet Journal of Rare Diseases, 2020, 15, 70.	2.7	32
28	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
29	Myotonic dystrophy type 1: clinical manifestations in children and adolescents. Archives of Disease in Childhood, 2019, 104, 48-52.	1.9	31
30	Prenusinersen economic and health-related quality of life burden of spinal muscular atrophy. Neurology, 2020, 95, e1-e10.	1.1	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.	2.2	30
32	Combined Endovascular and Open Surgery for Four-Vessel Cerebrovascular Occlusive Disease. Journal of Endovascular Therapy, 2001, 8, 62-66.	1.5	29
33	Cannabidiol for treating drugâ€resistant epilepsy in children: the New South Wales experience. Medical Journal of Australia, 2018, 209, 217-221.	1.7	28
34	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
35	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. Brain, 2018, 141, 3319-3330.	7.6	25
36	Evolution of peripheral nerve function in humans: novel insights from motor nerve excitability. Journal of Physiology, 2013, 591, 273-286.	2.9	24

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37	Motor cortical dysfunction develops in spinocerebellar ataxia type 3. Clinical Neurophysiology, 2016, 127, 3418-3424.	1.5	22
38	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
39	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
40	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
41	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
42	Importance of muscle biopsy to establish pathogenicity of DMD missense and splice variants. Neuromuscular Disorders, 2019, 29, 913-919.	0.6	19
43	Inherited Paediatric Motor Neuron Disorders: Beyond Spinal Muscular Atrophy. Neural Plasticity, 2017, 2017, 1-22.	2.2	18
44	Acute, Reversible Axonal Energy Failure During Stroke-Like Episodes in MELAS. Pediatrics, 2010, 126, e734-e739.	2.1	17
45	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
46	Great expectations: virus-mediated gene therapy in neurological disorders. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 849-860.	1.9	16
47	Corticomotoneuronal Integrity and Adaptation in Spinal Muscular Atrophy. Archives of Neurology, 2012, 69, 467.	4.5	15
48	Polyarticular Arthritis and Spinal Muscular Atrophy in Acid Ceramidase Deficiency. Pediatrics, 2016, 138, .	2.1	15
49	New and developing therapies in spinal muscular atrophy. Paediatric Respiratory Reviews, 2018, 28, 3-10.	1.8	15
50	Axonal dysfunction, dysmyelination, and conduction failure in hereditary neuropathy with liability to pressure palsies. Muscle and Nerve, 2014, 49, 858-865.	2.2	14
51	Neurophysiological profile of peripheral neuropathy associated with childhood mitochondrial disease. Mitochondrion, 2016, 30, 162-167.	3.4	13
52	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
53	Onasemnogene abeparvovec for the treatment of spinal muscular atrophy. Expert Opinion on Biological Therapy, 2022, 22, 1075-1090.	3.1	13
54	Teaching doctors how to diagnose paroxysmal events: a comparison of two educational methods. Medical Education, 2008, 42, 909-914.	2.1	12

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55	Spinal muscular atrophy â€" the dawning of a new era. Nature Reviews Neurology, 2020, 16, 593-594.	10.1	12
56	Growth and nutrition in pediatric neuromuscular disorders. Clinical Nutrition, 2021, 40, 4341-4348.	5.0	12
57	The relationship of body habitus and respiratory function in Duchenne muscular dystrophy. Respiratory Medicine, 2016, 119, 35-40.	2.9	11
58	Burning pain: axonal dysfunction in erythromelalgia. Pain, 2017, 158, 900-911.	4.2	11
59	Teaching paediatric epilepsy to medical students: A randomised crossover trial. Journal of Paediatrics and Child Health, 2009, 45, 727-730.	0.8	10
60	Population pharmacokinetics-based recommendations for a single delayed or missed dose of nusinersen. Neuromuscular Disorders, 2021, 31, 310-318.	0.6	10
61	Clinically Responsive Genomic Analysis Pipelines. Journal of Molecular Diagnostics, 2021, 23, 894-905.	2.8	10
62	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
63	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
64	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
65	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
66	Axonal excitability changes in children with spinal muscular atrophy treated with nusinersen. Journal of Physiology, 2022, 600, 95-109.	2.9	7
67	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
68	Cannabis for paediatric epilepsy: challenges and conundrums. Medical Journal of Australia, 2018, 208, 132-136.	1.7	6
69	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
70	Nutritional practices in pediatric patients with neuromuscular disorders. Nutrition Reviews, 2020, 78, 857-865.	5.8	5
71	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
72	The changing therapeutic landscape of spinal muscular atrophy. Australian Journal of General Practice, 2022, 51, 38-42.	0.8	5

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73	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
74	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
75	Glial mitochondropathy in infantile neuroaxonal dystrophy: pathophysiological and therapeutic implications. Brain, 2016, 139, e67-e67.	7.6	4
76	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
77	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
78	Multidisciplinary perspectives and practices of wheelchair prescription for children with neuromuscular conditions. Disability and Rehabilitation: Assistive Technology, 2023, 18, 166-174.	2.2	3
79	Peripheral nerve maturation and excitability properties from early childhood: Comparison of motor and sensory nerves. Clinical Neurophysiology, 2020, 131, 2452-2459.	1.5	3
80	Multimodal quantitative examination of nerve function in colorectal cancer patients prior to chemotherapy. Muscle and Nerve, 2018, 57, 615-621.	2.2	2
81	Spinal muscular atrophy: A modifiable disease emerges. Paediatric Respiratory Reviews, 2018, 28, 1-2.	1.8	2
82	Screening for spinal muscular atrophy. Medical Journal of Australia, 2018, 209, 147-148.	1.7	2
83	Personalized medicine for children with spinal muscular atrophy: Toward the holy grail. Muscle and Nerve, 2020, 62, 425-426.	2.2	2
84	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
85	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
86	Treating adults with spinal muscular atrophy with nusinersen. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1139-1139.	1.9	1
87	Onasemnogene Abeparvovec Gene-Replacement Therapy (GRT) in Pre-symptomatic Spinal Muscular Atrophy (SMA): SPR1NT Study Update. Neuropediatrics, 2019, 50, .	0.6	0