

Michelle A Farrar Mbbs

List of Publications by Year in descending order

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Version: 2024-02-01

87
papers

2,845
citations

201385

27
h-index

205818

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. <i>World Journal of Clinical Pediatrics</i> , 2015, 4, 66.	0.6	199
2	Emerging therapies and challenges in spinal muscular atrophy. <i>Annals of Neurology</i> , 2017, 81, 355-368.	2.8	157
3	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. <i>American Journal of Human Genetics</i> , 2013, 93, 1108-1117.	2.6	147
4	The Genetics of Spinal Muscular Atrophy: Progress and Challenges. <i>Neurotherapeutics</i> , 2015, 12, 290-302.	2.1	110
5	Thrombotic Microangiopathy Following Onasemnogene Abeparvec for Spinal Muscular Atrophy: A Case Series. <i>Journal of Pediatrics</i> , 2021, 231, 265-268.	0.9	107
6	Clinical Characteristics and Functional Motor Outcomes of Enterovirus 71 Neurological Disease in Children. <i>JAMA Neurology</i> , 2016, 73, 300.	4.5	106
7	Pathophysiological Insights Derived by Natural History and Motor Function of Spinal Muscular Atrophy. <i>Journal of Pediatrics</i> , 2013, 162, 155-159.	0.9	104
8	Onasemnogene abeparvec for presymptomatic infants with two copies of SMN2 at risk for spinal muscular atrophy type 1: the Phase III SPR1NT trial. <i>Nature Medicine</i> , 2022, 28, 1381-1389.	15.2	99
9	Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. <i>Neuromuscular Disorders</i> , 2021, 31, 574-582.	0.3	94
10	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. <i>Annals of Neurology</i> , 2018, 83, 1105-1124.	2.8	93
11	Onasemnogene abeparvec for presymptomatic infants with three copies of SMN2 at risk for spinal muscular atrophy: the Phase III SPR1NT trial. <i>Nature Medicine</i> , 2022, 28, 1390-1397.	15.2	93
12	The implementation of newborn screening for spinal muscular atrophy: the Australian experience. <i>Genetics in Medicine</i> , 2020, 22, 557-565.	1.1	90
13	Integrating exome sequencing into a diagnostic pathway for epileptic encephalopathy: Evidence of clinical utility and cost effectiveness. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 186-199.	0.6	83
14	Pediatric chemotherapy induced peripheral neuropathy: A systematic review of current knowledge. <i>Cancer Treatment Reviews</i> , 2016, 50, 118-128.	3.4	69
15	An Integrated Safety Analysis of Infants and Children with Symptomatic Spinal Muscular Atrophy (SMA) Treated with Nusinersen in Seven Clinical Trials. <i>CNS Drugs</i> , 2019, 33, 919-932.	2.7	69
16	Neurophysiological and clinical outcomes in chemotherapy-induced neuropathy in cancer. <i>Clinical Neurophysiology</i> , 2017, 128, 1166-1175.	0.7	50
17	Biomarkers and the Development of a Personalized Medicine Approach in Spinal Muscular Atrophy. <i>Frontiers in Neurology</i> , 2019, 10, 898.	1.1	49
18	Nusinersen for SMA: expanded access programme. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 937-942.	0.9	46

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19	Financial, opportunity and psychosocial costs of spinal muscular atrophy: an exploratory qualitative analysis of Australian carer perspectives. <i>BMJ Open</i> , 2018, 8, e020907.	0.8	45
20	Newborn screening for spinal muscular atrophy with disease-modifying therapies: a cost-effectiveness analysis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 1296-1304.	0.9	40
21	Safety and efficacy of progressive resistance exercise for Charcot-Marie-Tooth disease in children: a randomised, double-blind, sham-controlled trial. <i>The Lancet Child and Adolescent Health</i> , 2017, 1, 106-113.	2.7	39
22	Novel motor phenotypes in patients with <i>VRK1</i> mutations without pontocerebellar hypoplasia. <i>Neurology</i> , 2016, 87, 65-70.	1.5	38
23	Dysfunction of axonal membrane conductances in adolescents and young adults with spinal muscular atrophy. <i>Brain</i> , 2011, 134, 3185-3197.	3.7	35
24	Fracture in Duchenne Muscular Dystrophy. <i>Journal of Child Neurology</i> , 2016, 31, 1181-1187.	0.7	34
25	Integrating newborn screening for spinal muscular atrophy into health care systems: an Australian pilot programme. <i>Developmental Medicine and Child Neurology</i> , 2022, 64, 625-632.	1.1	34
26	Motor unit changes in children with symptomatic spinal muscular atrophy treated with nusinersen. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 78-85.	0.9	33
27	Health, wellbeing and lived experiences of adults with SMA: a scoping systematic review. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 70.	1.2	32
28	Onasemnogene abeparvovec in spinal muscular atrophy: an Australian experience of safety and efficacy. <i>Annals of Clinical and Translational Neurology</i> , 2022, 9, 339-350.	1.7	32
29	Myotonic dystrophy type 1: clinical manifestations in children and adolescents. <i>Archives of Disease in Childhood</i> , 2019, 104, 48-52.	1.0	31
30	Prenusinersen economic and health-related quality of life burden of spinal muscular atrophy. <i>Neurology</i> , 2020, 95, e1-e10.	1.5	30
31	Expert recommendations and clinical considerations in the use of onasemnogene abeparvovec gene therapy for spinal muscular atrophy. <i>Muscle and Nerve</i> , 2021, 64, 413-427.	1.0	30
32	Combined Endovascular and Open Surgery for Four-Vessel Cerebrovascular Occlusive Disease. <i>Journal of Endovascular Therapy</i> , 2001, 8, 62-66.	0.8	29
33	Cannabidiol for treating drug-resistant epilepsy in children: the New South Wales experience. <i>Medical Journal of Australia</i> , 2018, 209, 217-221.	0.8	28
34	“Getting ready for the adult world”: how adults with spinal muscular atrophy perceive and experience healthcare, transition and well-being. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 74.	1.2	27
35	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. <i>Brain</i> , 2018, 141, 3319-3330.	3.7	25
36	Evolution of peripheral nerve function in humans: novel insights from motor nerve excitability. <i>Journal of Physiology</i> , 2013, 591, 273-286.	1.3	24

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37	Motor cortical dysfunction develops in spinocerebellar ataxia type 3. <i>Clinical Neurophysiology</i> , 2016, 127, 3418-3424.	0.7	22
38	Pathophysiology of motor dysfunction in a childhood motor neuron disease caused by mutations in the riboflavin transporter. <i>Clinical Neurophysiology</i> , 2016, 127, 911-918.	0.7	22
39	“The Whole Game is Changing and You’ve Got Hope” Australian Perspectives on Treatment Decision Making in Spinal Muscular Atrophy. <i>Patient</i> , 2020, 13, 389-400.	1.1	22
40	“We needed this” perspectives of parents and healthcare professionals involved in a pilot newborn screening program for spinal muscular atrophy. <i>EClinicalMedicine</i> , 2021, 33, 100742.	3.2	22
41	Whole Genome Sequencing Identifies a 78 kb Insertion from Chromosome 8 as the Cause of Charcot-Marie-Tooth Neuropathy CMTX3. <i>PLoS Genetics</i> , 2016, 12, e1006177.	1.5	20
42	Importance of muscle biopsy to establish pathogenicity of DMD missense and splice variants. <i>Neuromuscular Disorders</i> , 2019, 29, 913-919.	0.3	19
43	Inherited Paediatric Motor Neuron Disorders: Beyond Spinal Muscular Atrophy. <i>Neural Plasticity</i> , 2017, 2017, 1-22.	1.0	18
44	Acute, Reversible Axonal Energy Failure During Stroke-Like Episodes in MELAS. <i>Pediatrics</i> , 2010, 126, e734-e739.	1.0	17
45	Real-world respiratory and bulbar comorbidities of SMA type 1 children treated with nusinersen: 2-Year single centre Australian experience. <i>Paediatric Respiratory Reviews</i> , 2021, 39, 54-60.	1.2	16
46	Great expectations: virus-mediated gene therapy in neurological disorders. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 849-860.	0.9	16
47	Corticomotoneuronal Integrity and Adaptation in Spinal Muscular Atrophy. <i>Archives of Neurology</i> , 2012, 69, 467.	4.9	15
48	Polyarticular Arthritis and Spinal Muscular Atrophy in Acid Ceramidase Deficiency. <i>Pediatrics</i> , 2016, 138, .	1.0	15
49	New and developing therapies in spinal muscular atrophy. <i>Paediatric Respiratory Reviews</i> , 2018, 28, 3-10.	1.2	15
50	Axonal dysfunction, dysmyelination, and conduction failure in hereditary neuropathy with liability to pressure palsies. <i>Muscle and Nerve</i> , 2014, 49, 858-865.	1.0	14
51	Neurophysiological profile of peripheral neuropathy associated with childhood mitochondrial disease. <i>Mitochondrion</i> , 2016, 30, 162-167.	1.6	13
52	Quality of life and excessive daytime sleepiness in children and adolescents with myotonic dystrophy type 1. <i>Sleep Medicine</i> , 2017, 32, 92-96.	0.8	13
53	Onasemnogene abeparvovec for the treatment of spinal muscular atrophy. <i>Expert Opinion on Biological Therapy</i> , 2022, 22, 1075-1090.	1.4	13
54	Teaching doctors how to diagnose paroxysmal events: a comparison of two educational methods. <i>Medical Education</i> , 2008, 42, 909-914.	1.1	12

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55	Spinal muscular atrophy – the dawning of a new era. <i>Nature Reviews Neurology</i> , 2020, 16, 593-594.	4.9	12
56	Growth and nutrition in pediatric neuromuscular disorders. <i>Clinical Nutrition</i> , 2021, 40, 4341-4348.	2.3	12
57	The relationship of body habitus and respiratory function in Duchenne muscular dystrophy. <i>Respiratory Medicine</i> , 2016, 119, 35-40.	1.3	11
58	Burning pain: axonal dysfunction in erythromelalgia. <i>Pain</i> , 2017, 158, 900-911.	2.0	11
59	Teaching paediatric epilepsy to medical students: A randomised crossover trial. <i>Journal of Paediatrics and Child Health</i> , 2009, 45, 727-730.	0.4	10
60	Population pharmacokinetics-based recommendations for a single delayed or missed dose of nusinersen. <i>Neuromuscular Disorders</i> , 2021, 31, 310-318.	0.3	10
61	Clinically Responsive Genomic Analysis Pipelines. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 894-905.	1.2	10
62	Clinical practice guideline for the management of paediatric Charcot-Marie-Tooth disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2022, 93, 530-538.	0.9	10
63	The involvement of rare disease patient organisations in therapeutic innovation across rare paediatric neurological conditions: a narrative review. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 167.	1.2	9
64	“It’s not just the wheelchair, it’s everything else” – Australian parents’ perspectives of wheelchair prescription for children with neuromuscular disorders. <i>Disability and Rehabilitation</i> , 2020, 42, 3457-3466.	0.9	8
65	Parents’ perceptions of power wheelchair prescription for children with a neuromuscular disorder: a scoping review. <i>Disability and Rehabilitation</i> , 2019, 41, 2750-2757.	0.9	7
66	Axonal excitability changes in children with spinal muscular atrophy treated with nusinersen. <i>Journal of Physiology</i> , 2022, 600, 95-109.	1.3	7
67	Delivering paediatric precision medicine: Genomic and environmental considerations along the causal pathway of childhood neurodevelopmental disorders. <i>Developmental Medicine and Child Neurology</i> , 2022, 64, 1077-1084.	1.1	7
68	Cannabis for paediatric epilepsy: challenges and conundrums. <i>Medical Journal of Australia</i> , 2018, 208, 132-136.	0.8	6
69	Restoration of Nusinersen Levels Following Treatment Interruption in People With Spinal Muscular Atrophy: Simulations Based on a Population Pharmacokinetic Model. <i>CNS Drugs</i> , 2022, 36, 181-190.	2.7	6
70	Nutritional practices in pediatric patients with neuromuscular disorders. <i>Nutrition Reviews</i> , 2020, 78, 857-865.	2.6	5
71	Congenital subependymal giant cell astrocytoma in children with tuberous sclerosis complex: growth patterns and neurological outcome. <i>Pediatric Research</i> , 2021, 89, 1447-1451.	1.1	5
72	The changing therapeutic landscape of spinal muscular atrophy. <i>Australian Journal of General Practice</i> , 2022, 51, 38-42.	0.3	5

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73	Family, healthcare professional, and societal preferences for the treatment of infantile spinal muscular atrophy: A discrete choice experiment. <i>Developmental Medicine and Child Neurology</i> , 2022, 64, 753-761.	1.1	5
74	Approaches to genetic diagnosis in neuromuscular conditions in the era of next generation sequencing. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 1384-1385.	0.9	4
75	Glial mitochondriopathy in infantile neuroaxonal dystrophy: pathophysiological and therapeutic implications. <i>Brain</i> , 2016, 139, e67-e67.	3.7	4
76	Next generation sequencing of human enterovirus strains from an outbreak of enterovirus A71 shows applicability to outbreak investigations. <i>Journal of Clinical Virology</i> , 2020, 122, 104216.	1.6	4
77	SMN2 copy number-dependent SMN2 gene-replacement therapy (GRT) for spinal muscular atrophy type 1 (SMA1): pivotal phase 3 study (STRIVE) update. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, A22.1-A22.	0.9	3
78	Multidisciplinary perspectives and practices of wheelchair prescription for children with neuromuscular conditions. <i>Disability and Rehabilitation: Assistive Technology</i> , 2023, 18, 166-174.	1.3	3
79	Peripheral nerve maturation and excitability properties from early childhood: Comparison of motor and sensory nerves. <i>Clinical Neurophysiology</i> , 2020, 131, 2452-2459.	0.7	3
80	Multimodal quantitative examination of nerve function in colorectal cancer patients prior to chemotherapy. <i>Muscle and Nerve</i> , 2018, 57, 615-621.	1.0	2
81	Spinal muscular atrophy: A modifiable disease emerges. <i>Paediatric Respiratory Reviews</i> , 2018, 28, 1-2.	1.2	2
82	Screening for spinal muscular atrophy. <i>Medical Journal of Australia</i> , 2018, 209, 147-148.	0.8	2
83	Personalized medicine for children with spinal muscular atrophy: Toward the holy grail. <i>Muscle and Nerve</i> , 2020, 62, 425-426.	1.0	2
84	Ethical aspects of the changing landscape for spinal muscular atrophy management in Australia. <i>Australian Journal of General Practice</i> , 2022, 51, 131-135.	0.3	2
85	SMN2 copy number-dependent SMN2 gene-replacement therapy (GRT) in presymptomatic spinal muscular atrophy (SMA): study update. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, A5.3-A6.	0.9	1
86	Treating adults with spinal muscular atrophy with nusinersen. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 1139-1139.	0.9	1
87	Onasemnogene Aporavidine Gene-Replacement Therapy (GRT) in Pre-symptomatic Spinal Muscular Atrophy (SMA): SPRINT Study Update. <i>Neuropediatrics</i> , 2019, 50, .	0.3	0