Manoj P Menezes

List of Publications by Year
in descending order

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56	2,381	318942	242451
papers	citations	23 h-index	47 g-index

71	71	71	4770
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	1.1	45
2	Clinical practice guideline for the management of paediatric Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 530-538.	0.9	10
3	Replicating and redesigning ankle-foot orthoses with 3D printing for children with Charcot-Marie-Tooth disease. Gait and Posture, 2022, 96, 73-80.	0.6	0
4	Paediatric neurocysticercosis in high income countries. European Journal of Paediatric Neurology, 2022, 39, 88-95.	0.7	0
5	Advances in the management of <scp>Charcotâ€“Marieâ€“Tooth</scp> disease in childhood. Developmental Medicine and Child Neurology, 2022, 64, 931-932.	1.1	0
6	Clinical, Genetic, and Disability Profile of Pediatric Distal Hereditary Motor Neuropathy. Neurology, 2021, 96, e423-e432.	1.5	5
7	Psychiatric comorbidity is common in dystonia and other movement disorders. Archives of Disease in Childhood, 2021, 106, 62-67.	1.0	4
8	Reliability and sensitivity of radiographic measures of hip dysplasia in childhood Charcot-Marie-Tooth disease. HIP International, 2021, , 112070002110275.	0.9	0
9	Association Between Body Mass Index and Disability in Children With Charcot-Marie-Tooth Disease. Neurology, 2021, 97, e1727-e1736.	1.5	2
10	Refining clinical trial inclusion criteria to optimize the standardized response mean of the CMTPedS. Annals of Clinical and Translational Neurology, 2020, 7, 1713-1715.	1.7	5
11	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. Brain Communications, 2020, 2, fcaa178.	1.5	17
12	Prenusinersen economic and health-related quality of life burden of spinal muscular atrophy. Neurology, 2020, 95, e1-e10.	1.5	30
13	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	3.7	39
14	Surgical outcomes of cavovarus foot deformity in children with Charcot-Marie-Tooth disease. Neuromuscular Disorders, 2019, 29, 427-436.	0.3	18
15	Balance impairment in pediatric charcotâ€“marieâ€“tooth disease. Muscle and Nerve, 2019, 60, 242-249.	1.0	22
16	Feasibility of designing, manufacturing and delivering 3D printed ankleâ€foot orthoses: a systematic review. Journal of Foot and Ankle Research, 2019, 12, 11.	0.7	56
17	Yield of comparative genomic hybridization microarray in pediatric neurology practice. Neurology: Genetics, 2019, 5, e367.	0.9	4
18	Magnetic resonance imaging in enterovirusâ€71, myelin oligodendrocyte glycoprotein antibody, aquaporinâ€4 antibody, and multiple sclerosisâ€associated myelitis in children. Developmental Medicine and Child Neurology, 2019, 61, 1108-1116.	1.1	22

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19	Magnetic resonance imaging of the anterior compartment of the lower leg is a biomarker for weakness, disability, and impaired gait in childhood Charcotâ€“Marieâ€“Tooth disease. Muscle and Nerve, 2019, 59, 213-217.	1.0	7
20	Functional outcome measures for infantile Charcotâ€Marieâ€Tooth disease: a systematic review. Journal of the Peripheral Nervous System, 2018, 23, 99-107.	1.4	1
21	Unique clinical and neurophysiologic profile of a cohort of children with CMTX3. Neurology, 2018, 90, e1706-e1710.	1.5	3
22	Nusinersen for SMA: expanded access programme. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 937-942.	0.9	46
23	Genetic, Radiologic, and Clinical Variability in Brown-Vialetto-van Laere Syndrome. Seminars in Pediatric Neurology, 2018, 26, 2-9.	1.0	24
24	The First Case of Riboflavin Transporter Deficiency in sub-Saharan Africa. Seminars in Pediatric Neurology, 2018, 26, 10-14.	1.0	19
25	Established and novel measures of upper limb impairment in children with Charcotâ€Marieâ€tooth disease type 1A and riboflavin transporter deficiency type 2. Journal of the Peripheral Nervous System, 2018, 23, 29-35.	1.4	3
26	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. Brain, 2018, 141, 3319-3330.	3.7	25
27	Diagnostic yield of targeted massively parallel sequencing in children with epileptic encephalopathy. Seizure: the Journal of the British Epilepsy Association, 2018, 59, 132-140.	0.9	63
28	Gait patterns of children and adolescents with Charcot-Marie-Tooth disease. Gait and Posture, 2017, 56, 89-94.	0.6	24
29	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.	2.7	39
30	Natural history of Charcotâ€Marieâ€Tooth disease during childhood. Annals of Neurology, 2017, 82, 353-359.	2.8	50
31	Eye movement disorders are an early manifestation of <scp>CACNA</scp>1A mutations in children. Developmental Medicine and Child Neurology, 2016, 58, 639-644.	1.1	58
32	Phenotypic Variability of Childhood Charcot-Marie-Tooth Disease. JAMA Neurology, 2016, 73, 645.	4.5	71
33	Variants in SLC18A3 , vesicular acetylcholine transporter, cause congenital myasthenic syndrome. Neurology, 2016, 87, 1442-1448.	1.5	46
34	Neurophysiological profile of peripheral neuropathy associated with childhood mitochondrial disease. Mitochondrion, 2016, 30, 162-167.	1.6	13
35	Auditory neuropathy in Brownâ€“Vialettoâ€“Van Laere syndrome due to riboflavin transporter RFVT2 deficiency. Developmental Medicine and Child Neurology, 2016, 58, 848-854.	1.1	22
36	Prominent scapulae mimicking an inherited myopathy expands the phenotype of CHD7-related disease. European Journal of Human Genetics, 2016, 24, 1216-1219.	1.4	2

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37	Pathophysiology of motor dysfunction in a childhood motor neuron disease caused by mutations in the riboflavin transporter. Clinical Neurophysiology, 2016, 127, 911-918.	0.7	22
38	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.	1.5	20
39	Systematic review of exercise for Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2015, 20, 347-362.	1.4	51
40	Delayed diagnosis of congenital myasthenia due to associated mitochondrial enzyme defect. Neuromuscular Disorders, 2015, 25, 257-261.	0.3	16
41	Response. Neuromuscular Disorders, 2015, 25, 360.	0.3	0
42	Infectious and Autoantibody-Associated Encephalitis: Clinical Features and Long-term Outcome. Pediatrics, 2015, 135, e974-e984.	1.0	115
43	Reply: The p.Ser107Leu inBICD2is a mutation â€˜hot spotâ€™ causing distal spinal muscular atrophy. Brain, 2015, 138, e392-e392.	3.7	1
44	Phenotypic and molecular insights into spinal muscular atrophy due to mutations in BICD2. Brain, 2015, 138, 293-310.	3.7	82
45	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. Brain, 2014, 137, 44-56.	3.7	143
46	Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. Nature Genetics, 2014, 46, 503-509.	9.4	490
47	Whole exome sequencing identifies three recessive FIG4 mutations in an apparently dominant pedigree with Charcotâ€“Marieâ€“Tooth disease. Neuromuscular Disorders, 2014, 24, 666-670.	0.3	17
48	Randomised controlled trial protocol of foot and ankle exercise for children with Charcot-Marie-Tooth disease. Journal of Physiotherapy, 2014, 60, 55.	0.7	9
49	Mutations in BICD2 Cause Dominant Congenital Spinal Muscular Atrophy and Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 92, 965-973.	2.6	156
50	Rapidly progressive asymmetrical weakness in Charcotâ€“Marieâ€“Tooth disease type 4J resembles chronic inflammatory demyelinating polyneuropathy. Neuromuscular Disorders, 2013, 23, 399-403.	0.3	38
51	Transitioning outcome measures: relationship between the CMTPedS and CMTNSv2 in children, adolescents, and young adults with Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2013, 18, 177-180.	1.4	15
52	Importance and challenge of making an early diagnosis in LMNA -related muscular dystrophy. Neurology, 2012, 78, 1258-1263.	1.5	41
53	Mutations in the RNA exosome component gene EXOSC3 cause pontocerebellar hypoplasia and spinal motor neuron degeneration. Nature Genetics, 2012, 44, 704-708.	9.4	216
54	An inherited TUBB2B mutation alters a kinesin-binding site and causes polymicrogyria, CFEOM and axon dysinnervation. Human Molecular Genetics, 2012, 21, 5484-5499.	1.4	106

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55	Peripheral neuropathy associated with mitochondrial disease in children. Developmental Medicine and Child Neurology, 2012, 54, 407-414.	1.1	27
56	Inherited neuromuscular disorders: Pathway to diagnosis. Journal of Paediatrics and Child Health, 2012, 48, 458-465.	0.4	19