Manoj P Menezes

List of Publications by Year
in descending order

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56 papers	2,381 citations	279701 23 h-index	214721 47 g-index
71 all docs	71 docs citations	71 times ranked	4487 citing authors

#	Article	IF	Citations
1	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
2	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
3	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
4	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. Brain, 2014, 137, 44-56.	3.7	143
5	Infectious and Autoantibody-Associated Encephalitis: Clinical Features and Long-term Outcome. Pediatrics, 2015, 135, e974-e984.	1.0	115
6	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
7	Phenotypic and molecular insights into spinal muscular atrophy due to mutations in BICD2. Brain, 2015, 138, 293-310.	3.7	82
8	Phenotypic Variability of Childhood Charcot-Marie-Tooth Disease. JAMA Neurology, 2016, 73, 645.	4.5	71
9	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
10	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
11	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
12	Systematic review of exercise for Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2015, 20, 347-362.	1.4	51
13	Natural history of Charcotâ€Marieâ€Tooth disease during childhood. Annals of Neurology, 2017, 82, 353-359.	2.8	50
14	Variants in SLC18A3 , vesicular acetylcholine transporter, cause congenital myasthenic syndrome. Neurology, 2016, 87, 1442-1448.	1.5	46
15	Nusinersen for SMA: expanded access programme. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 937-942.	0.9	46
16	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
17	Importance and challenge of making an early diagnosis in LMNA -related muscular dystrophy. Neurology, 2012, 78, 1258-1263.	1.5	41
18	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

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19	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	3.7	39
20	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
21	Prenusinersen economic and health-related quality of life burden of spinal muscular atrophy. Neurology, 2020, 95, e1-e10.	1.5	30
22	Peripheral neuropathy associated with mitochondrial disease in children. Developmental Medicine and Child Neurology, 2012, 54, 407-414.	1.1	27
23	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. Brain, 2018, 141, 3319-3330.	3.7	25
24	Gait patterns of children and adolescents with Charcot-Marie-Tooth disease. Gait and Posture, 2017, 56, 89-94.	0.6	24
25	Genetic, Radiologic, and Clinical Variability in Brown-Vialetto-van Laere Syndrome. Seminars in Pediatric Neurology, 2018, 26, 2-9.	1.0	24
26	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
27	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
28	Balance impairment in pediatric charcotâ€“marieâ€“tooth disease. Muscle and Nerve, 2019, 60, 242-249.	1.0	22
29	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
30	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
31	Inherited neuromuscular disorders: Pathway to diagnosis. Journal of Paediatrics and Child Health, 2012, 48, 458-465.	0.4	19
32	The First Case of Riboflavin Transporter Deficiency in sub-Saharan Africa. Seminars in Pediatric Neurology, 2018, 26, 10-14.	1.0	19
33	Surgical outcomes of cavovarus foot deformity in children with Charcot-Marie-Tooth disease. Neuromuscular Disorders, 2019, 29, 427-436.	0.3	18
34	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
35	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. Brain Communications, 2020, 2, fcaa178.	1.5	17
36	Delayed diagnosis of congenital myasthenia due to associated mitochondrial enzyme defect. Neuromuscular Disorders, 2015, 25, 257-261.	0.3	16

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37	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
38	Neurophysiological profile of peripheral neuropathy associated with childhood mitochondrial disease. Mitochondrion, 2016, 30, 162-167.	1.6	13
39	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
40	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
41	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
42	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
43	Clinical, Genetic, and Disability Profile of Pediatric Distal Hereditary Motor Neuropathy. Neurology, 2021, 96, e423-e432.	1.5	5
44	Yield of comparative genomic hybridization microarray in pediatric neurology practice. Neurology: Genetics, 2019, 5, e367.	0.9	4
45	Psychiatric comorbidity is common in dystonia and other movement disorders. Archives of Disease in Childhood, 2021, 106, 62-67.	1.0	4
46	Unique clinical and neurophysiologic profile of a cohort of children with CMTX3. Neurology, 2018, 90, e1706-e1710.	1.5	3
47	Established and novel measures of upper limb impairment in children with Charcot‹b›â€‹/b›Marie‹b›â€‹/b›tooth disease type 1A and riboflavin transporter deficiency type 2. Journal of the Peripheral Nervous System, 2018, 23, 29-35.	1.4	3
48	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
49	Association Between Body Mass Index and Disability in Children With Charcot-Marie-Tooth Disease. Neurology, 2021, 97, e1727-e1736.	1.5	2
50	Reply: The p.Ser107Leu inBICD2is a mutation â€˜hot spotâ€™ causing distal spinal muscular atrophy. Brain, 2015, 138, e392-e392.	3.7	1
51	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
52	Response. Neuromuscular Disorders, 2015, 25, 360.	0.3	0
53	Reliability and sensitivity of radiographic measures of hip dysplasia in childhood Charcot-Marie-Tooth disease. HIP International, 2021, , 112070002110275.	0.9	0
54	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

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55	Paediatric neurocysticercosis in high income countries. European Journal of Paediatric Neurology, 2022, 39, 88-95.	0.7	0
56	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