Manju A Kurian

List of Publications by Year
in descending order

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73 papers	4,456 citations	126907 33 h-index	118850 62 g-index
75 all docs	75 docs citations	75 times ranked	8431 citing authors

#	Article	IF	Citations
1	Biallelic <scp>AOPEP</scp> Lossâ€ofâ€Function Variants Cause Progressive Dystonia with Prominent Limb Involvement. Movement Disorders, 2022, 37, 137-147.	3.9	14
2	The dopamine transporter gene SLC6A3: multidisease risks. Molecular Psychiatry, 2022, 27, 1031-1046.	7.9	28
3	Structural mapping of GABRB3 variants reveals genotypeâ€“phenotype correlations. Genetics in Medicine, 2022, 24, 681-693.	2.4	10
4	Relationship of Genotype, Phenotype, and Treatment in Dopaâ€Responsive Dystonia: <scp>MDSGene</scp> Review. Movement Disorders, 2022, 37, 237-252.	3.9	19
5	Comparison of methylation episignatures in KMT2B- and KMT2D-related human disorders. Epigenomics, 2022, 14, 537-547.	2.1	10
6	Highlighting the Dystonic Phenotype Related to <scp>GNAO1</scp>. Movement Disorders, 2022, 37, 1547-1554.	3.9	25
7	<scp>STXBP1</scp> Stopâ€Loss Mutation Associated with Complex Early Onset Movement Disorder without Epilepsy. Movement Disorders Clinical Practice, 2022, 9, 837-840.	1.5	3
8	Freezing of Gait as a Complication of Pallidal Deep Brain Stimulation in <scp>DYTâ€KMT2B</scp> Patients with Evidence of Striatonigral Degeneration. Movement Disorders Clinical Practice, 2022, 9, 992-996.	1.5	1
9	Precision medicine for genetic childhood movement disorders. Developmental Medicine and Child Neurology, 2021, 63, 925-933.	2.1	4
10	Aromatic <scp>l</scp>-amino acid decarboxylase deficiency: a patient-derived neuronal model for precision therapies. Brain, 2021, 144, 2443-2456.	7.6	16
11	A Recurrent <scp>VPS16</scp> p.Arg187* Nonsense Variant in Earlyâ€Onset Generalized Dystonia. Movement Disorders, 2021, 36, 1984-1985.	3.9	7
12	An Update on the Phenotype, Genotype and Neurobiology of <scp>ADCY5â€Related</scp> Disease. Movement Disorders, 2021, 36, 1104-1114.	3.9	24
13	Childhoodâ€Onset Chorea Caused by a Recurrent De Novo DRD2 Variant. Movement Disorders, 2021, 36, 1472-1473.	3.9	6
14	Assessment of intellectual impairment, healthâ€related quality of life, and behavioral phenotype in patients with neurotransmitter related disorders: Data from the <scp>iNTD</scp> registry. Journal of Inherited Metabolic Disease, 2021, 44, 1489-1502.	3.6	7
15	Commentary: Galactosemia Diagnosis by Whole Exome Sequencing Later in Life. Movement Disorders Clinical Practice, 2021, 8, S40-S41.	1.5	0
16	Commentary: <scp>GM1</scp>â€Gangliosidosis Type <scp>III</scp> Associated Parkinsonism. Movement Disorders Clinical Practice, 2021, 8, S24-S25.	1.5	0
17	Consensus clinical management guideline for betaâ€propeller proteinâ€associated neurodegeneration. Developmental Medicine and Child Neurology, 2021, 63, 1402-1409.	2.1	17
18	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, .	8.5	43

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19	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila. American Journal of Human Genetics, 2021, 108, 1669-1691.	6.2	23
20	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nature Communications, 2021, 12, 5529.	12.8	21
21	RHOBTB2 Mutations Expand the Phenotypic Spectrum of Alternating Hemiplegia of Childhood. Neurology, 2021, 96, e1539-e1550.	1.1	15
22	The role of manganese dysregulation in neurological disease: emerging evidence. Lancet Neurology, The, 2021, 20, 956-968.	10.2	51
23	Towards Precision Therapies for Inherited Disorders of Neurodegeneration with Brain Iron Accumulation. Tremor and Other Hyperkinetic Movements, 2021, 11, 51.	2.0	7
24	Biallelic Mutations of TBC1D24 in Exerciseâ€Induced Paroxysmal Dystonia. Movement Disorders, 2020, 35, 372-373.	3.9	8
25	The expanding spectrum of movement disorders in genetic epilepsies. Developmental Medicine and Child Neurology, 2020, 62, 178-191.	2.1	31
26	Recent genetic advances in early-onset dystonia. Current Opinion in Neurology, 2020, 33, 500-507.	3.6	5
27	Lossâ€ofâ€Function Variants in <scp>HOPS</scp> Complex Genes <scp>VPS16</scp> and <scp>VPS41</scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. Annals of Neurology, 2020, 88, 867-877.	5.3	70
28	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	7.6	57
29	<scp>AADC</scp> deficiency from infancy to adulthood: Symptoms and developmental outcome in an international cohort of 63 patients. Journal of Inherited Metabolic Disease, 2020, 43, 1121-1130.	3.6	59
30	<scp>DNAJC6</scp> Mutations Disrupt Dopamine Homeostasis in Juvenile <scp>Parkinsonismâ€Dystonia</scp>. Movement Disorders, 2020, 35, 1357-1368.	3.9	22
31	KIF1Aâ€related disorders in children: A wide spectrum of central and peripheral nervous system involvement. Journal of the Peripheral Nervous System, 2020, 25, 117-124.	3.1	40
32	Autosomal dominant mitochondrial membrane proteinâ€associated neurodegeneration (MPAN). Molecular Genetics & Genomic Medicine, 2019, 7, e00736.	1.2	40
33	Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A singleâ€center cohort study. Movement Disorders, 2019, 34, 1516-1527.	3.9	55
34	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45
35	Yield of comparative genomic hybridization microarray in pediatric neurology practice. Neurology: Genetics, 2019, 5, e367.	1.9	4
36	Phenotypes, genotypes, and the management of paroxysmal movement disorders. Developmental Medicine and Child Neurology, 2018, 60, 559-565.	2.1	31

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37	Neurodegeneration with brain iron accumulation. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 293-305.	1.8	153
38	Clinical and molecular characterization of KCNT1 -related severe early-onset epilepsy. Neurology, 2018, 90, e55-e66.	1.1	89
39	Spectrum of movement disorders and neurotransmitter abnormalities in paediatric POLG disease. Journal of Inherited Metabolic Disease, 2018, 41, 1275-1283.	3.6	12
40	Postsynaptic movement disorders: clinical phenotypes, genotypes, and disease mechanisms. Journal of Inherited Metabolic Disease, 2018, 41, 1077-1091.	3.6	24
41	GNAO1 encephalopathy. Neurology: Genetics, 2017, 3, e143.	1.9	84
42	Consensus clinical management guideline for pantothenate kinase-associated neurodegeneration (PKAN). Molecular Genetics and Metabolism, 2017, 120, 278-287.	1.1	64
43	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
44	Not all SCN1A epileptic encephalopathies are Dravet syndrome. Neurology, 2017, 89, 1035-1042.	1.1	97
45	Utility of Induced Pluripotent Stem Cells for the Study and Treatment of Genetic Diseases: Focus on Childhood Neurological Disorders. Frontiers in Molecular Neuroscience, 2016, 9, 78.	2.9	29
46	Improving diagnosis and broadening the phenotypes in early-onset seizure and severe developmental delay disorders through gene panel analysis. Journal of Medical Genetics, 2016, 53, 310-317.	3.2	191
47	Delineation of the movement disorders associated with FOXG1 mutations. Neurology, 2016, 86, 1794-1800.	1.1	55
48	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	6.2	96
49	Biallelic Mutations in PDE10A Lead to Loss of Striatal PDE10A and a Hyperkinetic Movement Disorder with Onset in Infancy. American Journal of Human Genetics, 2016, 98, 735-743.	6.2	65
50	Psychiatric disorders, myoclonus dystonia and <scp>SGCE</scp> : an international study. Annals of Clinical and Translational Neurology, 2016, 3, 4-11.	3.7	43
51	GABRB3 mutations: a new and emerging cause of early infantile epileptic encephalopathy. Developmental Medicine and Child Neurology, 2016, 58, 416-420.	2.1	56
52	<scp>RARS</scp>2 mutations in a sibship with infantile spasms. Epilepsia, 2016, 57, e97-e102.	5.1	23
53	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	6.2	99
54	The clinical syndrome of dystonia with anarthria/aphonia. Parkinsonism and Related Disorders, 2016, 24, 20-27.	2.2	10

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55	LYRM7mutations cause a multifocal cavitating leukoencephalopathy with distinct MRI appearance. Brain, 2016, 139, 782-794.	7.6	51
56	The genetic landscape of the epileptic encephalopathies of infancy and childhood. Lancet Neurology, The, 2016, 15, 304-316.	10.2	474
57	The novel R347g pathogenic mutation of aromatic amino acid decarboxylase provides additional molecular insights into enzyme catalysis and deficiency. Biochimica Et Biophysica Acta - Proteins and Proteomics, 2016, 1864, 676-682.	2.3	15
58	Faulty cardiac repolarization reserve in alternating hemiplegia of childhood broadens the phenotype. Brain, 2015, 138, 2859-2874.	7.6	30
59	Neurodegeneration with Brain Iron Accumulation: Genetic Diversity and Pathophysiological Mechanisms. Annual Review of Genomics and Human Genetics, 2015, 16, 257-279.	6.2	195
60	Monoamine neurotransmitter disordersâ€"clinical advances and future perspectives. Nature Reviews Neurology, 2015, 11, 567-584.	10.1	221
61	The clinical and genetic heterogeneity of paroxysmal dyskinesias. Brain, 2015, 138, 3567-3580.	7.6	129
62	Benign Hereditary Chorea: An Update. Tremor and Other Hyperkinetic Movements, 2015, 5, 314.	2.0	30
63	Genetic disorders of thyroid metabolism and brain development. Developmental Medicine and Child Neurology, 2014, 56, 627-634.	2.1	19
64	Atypical PLA2G6â€Associated Neurodegeneration: Social Communication Impairment, Dystonia and Response to Deep Brain Stimulation. Movement Disorders Clinical Practice, 2014, 1, 128-131.	1.5	13
65	A type I interferon signature identifies bilateral striatal necrosis due to mutations inADAR1. Journal of Medical Genetics, 2014, 51, 76-82.	3.2	118
66	Exome Sequence Reveals Mutations in CoA Synthase as a Cause of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2014, 94, 11-22.	6.2	176
67	Severe infantile epileptic encephalopathy due to mutations in <scp>PLCB</scp>1: expansion of the genotypic and phenotypic disease spectrum. Developmental Medicine and Child Neurology, 2014, 56, 1124-1128.	2.1	21
68	What is new for monoamine neurotransmitter disorders?. Journal of Inherited Metabolic Disease, 2014, 37, 619-626.	3.6	40
69	Pantothenate Kinase-Associated Neurodegeneration (PKAN) and PLA2G6-Associated Neurodegeneration (PLAN). International Review of Neurobiology, 2013, 110, 49-71.	2.0	68
70	Beta-propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. Brain, 2013, 136, 1708-1717.	7.6	203
71	Migrating partial seizures of infancy: expansion of the electroclinical, radiological and pathological disease spectrum. Brain, 2013, 136, 1578-1591.	7.6	144
72	Exome Sequencing Reveals De Novo WDR45 Mutations Causing a Phenotypically Distinct, X-Linked Dominant Form of NBIA. American Journal of Human Genetics, 2012, 91, 1144-1149.	6.2	309

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73	Niemannâ€“Pick type C disease as proofâ€ofâ€concept for intelligent biomarker panel selection in neurometabolic disorders. Developmental Medicine and Child Neurology, 0, , .	2.1	6