Jaya Punetha

List of Publications by Year
in descending order

31 papers	1,289 citations	394421 19 h-index	454955 30 g-index
31 all docs	31 docs citations	31 times ranked	2652 citing authors

#	Article	IF	Citations
1	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	2.5	7
2	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
3	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	2.4	22
4	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. American Journal of Human Genetics, 2021, 108, 1981-2005.	6.2	38
5	Novel Biallelic Variants in KIF21A Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects. , 2021, 52, .		0
6	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	7.6	33
7	Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network. Molecular Genetics & Genomic Medicine, 2020, 8, e1397.	1.2	16
8	<scp>Wolffâ€“Parkinsonâ€“White</scp> syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. American Journal of Medical Genetics, Part A, 2020, 182, 1387-1399.	1.2	14
9	Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Annals of Clinical and Translational Neurology, 2020, 7, 610-627.	3.7	15
10	Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1395-1406.	3.7	20
11	The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. American Journal of Human Genetics, 2019, 105, 132-150.	6.2	74
12	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056.	6.2	30
13	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	6.2	24
14	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
15	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
16	The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. Human Molecular Genetics, 2018, 27, 2064-2075.	2.9	16
17	Identification of a pathogenic PMP2 variant in a multi-generational family with CMT type 1: Clinical gene panels versus genome-wide approaches to molecular diagnosis. Molecular Genetics and Metabolism, 2018, 125, 302-304.	1.1	13
18	Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. Journal of Pediatric Genetics, 2018, 07, 164-173.	0.7	15

#	Article	IF	Citations
19	Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles. American Journal of Human Genetics, 2018, 103, 171-187.	6.2	160
20	Novel Col12A1 variant expands the clinical picture of congenital myopathies with extracellular matrix defects. Muscle and Nerve, 2017, 55, 277-281.	2.2	31
21	Targeted Re-Sequencing Emulsion PCR Panel for Myopathies: Results in 94 Cases. Journal of Neuromuscular Diseases, 2016, 3, 209-225.	2.6	18
22	Association Study of Exon Variants in the NF-ÎºB and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	6.2	71
23	Diagnosis and etiology of congenital muscular dystrophy: We are halfway there. Annals of Neurology, 2016, 80, 101-111.	5.3	57
24	Somatic mosaicism due to a reversion variant causing hemi-atrophy: a novel variant of dystrophinopathy. European Journal of Human Genetics, 2016, 24, 1511-1514.	2.8	8
25	Genetic modifiers of ambulation in the cooperative international Neuromuscular research group Duchenne natural history study. Annals of Neurology, 2015, 77, 684-696.	5.3	111
26	Exome Sequencing Identifies DYNC1H1 Variant Associated With Vertebral Abnormality and Spinal Muscular Atrophy With Lower Extremity Predominance. Pediatric Neurology, 2015, 52, 239-244.	2.1	27
27	Recessive ACTA1 variant causes congenital muscular dystrophy with rigid spine. European Journal of Human Genetics, 2015, 23, 883-886.	2.8	23
28	A novel mutation expands the genetic and clinical spectrum of MYH7-related myopathies. Neuromuscular Disorders, 2013, 23, 432-436.	0.6	35
29	Mutations in GDP-Mannose Pyrophosphorylase B Cause Congenital and Limb-Girdle Muscular Dystrophies Associated with Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 93, 29-41.	6.2	197
30	Short Read (Next-Generation) Sequencing. Circulation: Cardiovascular Genetics, 2013, 6, 427-434.	5.1	23
31	Facile synthesis of nucleoside 5â€²-(Î±-P-seleno)-triphosphates and phosphoroselenoate RNA transcription. Rna, 2011, 17, 1932-1938.	3.5	24