

Parul Jayakar

List of Publications by Year in descending order

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Version: 2024-02-01

21
papers

441
citations

840776

11
h-index

794594

19
g-index

21
all docs

21
docs citations

21
times ranked

930
citing authors

#	ARTICLE	IF	CITATIONS
1	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotypeâ€œphenotype correlations. <i>Genetics in Medicine</i> , 2016, 18, 1143-1150.	2.4	64
2	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2018, 102, 985-994.	6.2	59
3	Mutations in TBCK, Encoding TBC1-Domain-Containing Kinase, Lead to a Recognizable Syndrome of Intellectual Disability and Hypotonia. <i>American Journal of Human Genetics</i> , 2016, 98, 782-788.	6.2	50
4	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. <i>European Journal of Human Genetics</i> , 2019, 27, 1611-1618.	2.8	45
5	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. <i>Nature Genetics</i> , 2021, 53, 1006-1021.	21.4	44
6	<i>KCTD7</i> deficiency defines a distinct neurodegenerative disorder with a conserved autophagyâ€œlysosome defect. <i>Annals of Neurology</i> , 2018, 84, 766-780.	5.3	42
7	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. <i>Genetics in Medicine</i> , 2019, 21, 2036-2042.	2.4	23
8	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. <i>Genetics in Medicine</i> , 2020, 22, 1215-1226.	2.4	22
9	Clinical, neuroimaging, and molecular spectrum of <i>TECPR2</i> â€œassociated hereditary sensory and autonomic neuropathy with intellectual disability. <i>Human Mutation</i> , 2021, 42, 762-776.	2.5	18
10	Clinical and molecular features of 66 patients with musculocontractural Ehlersâ€œDanlos syndrome caused by pathogenic variants in CHST14 (mcEDS-CHST14). <i>Journal of Medical Genetics</i> , 2021, , jmedgenet-2020-107623.	3.2	18
11	PRKAG2 mutation: An easily missed cardiac specific non-lysosomal glycogenosis. <i>Annals of Pediatric Cardiology</i> , 2015, 8, 153.	0.5	17
12	Clinicoâ€œradiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. <i>Human Mutation</i> , 2022, 43, 403-419.	2.5	9
13	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. <i>Genetics in Medicine</i> , 2021, 23, 1624-1635.	2.4	7
14	Uridineâ€œresponsive epileptic encephalopathy due to inherited variants in CAD : A Tale of Two Siblings. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 716-722.	3.7	6
15	A Case of UDP-Galactose 4â€œ-Epimerase Deficiency Associated with Dyshematopoiesis and Atrioventricular Valve Malformations: An Exceptional Clinical Phenotype Explained by Altered N-Glycosylation with Relative Preservation of the Leloir Pathway. <i>Molecular Syndromology</i> , 2020, 11, 320-330.	0.8	5
16	ITSN1: a novel candidate gene involved in autosomal dominant neurodevelopmental disorder spectrum. <i>European Journal of Human Genetics</i> , 2022, 30, 111-116.	2.8	4
17	Chromosomal microarray detects genetic risks of neurodevelopmental disorders in newborns with congenital heart disease. <i>Cardiology in the Young</i> , 2021, 31, 1275-1282.	0.8	3
18	Novel Loss of Function in the AGK Gene. <i>JACC: Case Reports</i> , 2019, 1, 11-16.	0.6	2

#	ARTICLE	IF	CITATIONS
19	Acquired retinal pigmentary degeneration in a child with 13q deletion syndrome. Journal of AAPOS, 2015, 19, 482-484.	0.3	1
20	Discovery of a neuromuscular syndrome caused by biallelic variants in ASCC3. Human Genetics and Genomics Advances, 2021, 2, 100024.	1.7	1
21	Case Report: The Association of Wilson Disease in a Patient With Ataxia and GLUT-1 Deficiency. Frontiers in Pediatrics, 2021, 9, 750593.	1.9	1