Parul Jayakar

List of Publications by Year
in descending order

21	441	840776 11	794594 19
papers	citations	h-index	g-index

21	21	21	930
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	ITSN1: a novel candidate gene involved in autosomal dominant neurodevelopmental disorder spectrum. European Journal of Human Genetics, 2022, 30, 111-116.	2.8	4
2	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	2.5	9
3	Uridineâ€responsive epileptic encephalopathy due to inherited variants in CAD : A Tale of Two Siblings. Annals of Clinical and Translational Neurology, 2021, 8, 716-722.	3.7	6
4	Chromosomal microarray detects genetic risks of neurodevelopmental disorders in newborns with congenital heart disease. Cardiology in the Young, 2021, 31, 1275-1282.	0.8	3
5	Discovery of a neuromuscular syndrome caused by biallelic variants in ASCC3. Human Genetics and Genomics Advances, 2021, 2, 100024.	1.7	1
6	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	2.5	18
7	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. Genetics in Medicine, 2021, 23, 1624-1635.	2.4	7
8	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
9	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
10	Clinical and molecular features of 66 patients with musculocontractural Ehlersâ€'Danlos syndrome caused by pathogenic variants in CHST14 (mcEDS-CHST14). Journal of Medical Genetics, 2021, , jmedgenet-2020-107623.	3.2	18
11	A Case of UDP-Galactose 4â€²-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. Molecular Syndromology, 2020, 11, 320-330.	0.8	5
12	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
13	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	2.8	45
14	Novel Loss of Function in the AGK Gene. JACC: Case Reports, 2019, 1, 11-16.	0.6	2
15	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042.	2.4	23
16	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.2	59
17	KCTD7 deficiency defines a distinct neurodegenerative disorder with a conserved autophagyâ€lysosome defect. Annals of Neurology, 2018, 84, 766-780.	5.3	42
18	Mutations in TBCK, Encoding TBC1-Domain-Containing Kinase, Lead to a Recognizable Syndrome of Intellectual Disability and Hypotonia. American Journal of Human Genetics, 2016, 98, 782-788.	6.2	50

#	Article	IF	Citations
19	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotypeâ€“phenotype correlations. Genetics in Medicine, 2016, 18, 1143-1150.	2.4	64
20	Acquired retinal pigmentary degeneration in a child with 13q deletion syndrome. Journal of AAPOS, 2015, 19, 482-484.	0.3	1
21	PRKAG2 mutation: An easily missed cardiac specific non-lysosomal glycogenosis. Annals of Pediatric Cardiology, 2015, 8, 153.	0.5	17