Purvi Majethia

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9762678/publications.pdf

Version: 2024-02-01

8	34	2257263 3	2053342 5
papers	citations	h-index	g-index

8	8	8	22
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Multilocus disease-causing genomic variations for Mendelian disorders: role of systematic phenotyping and implications on genetic counselling. European Journal of Human Genetics, 2021, 29, 1774-1780.	1.4	7
2	NAD(P)HX dehydratase (NAXD) deficiency due to a novel biallelic missense variant and review of literature. European Journal of Medical Genetics, 2021, 64, 104266.	0.7	6
3	Expanding the electro-clinical phenotype of CARS2associated neuroregression. Epilepsy and Behavior Reports, 2021, 16, 100485.	0.5	5
4	Biallelic start loss variant, c.<scp>1A</scp>Â>â€‰G in <scp>GCSH</scp> is associated with variant nonketotic hyperglycinemia. Clinical Genetics, 2021, 100, 201-205.	1.0	4
5	Further evidence of affected females with a heterozygous variant in FGF13 causing X-linked developmental and epileptic encephalopathy 90. European Journal of Medical Genetics, 2022, 65, 104403.	0.7	4
6	Wiedemannâ€“Rautenstrauch syndrome in an Indian patient with biallelic pathogenic variants in POLR3A. American Journal of Medical Genetics, Part A, 2021, 185, 1602-1605.	0.7	3
7	Further evidence of muscle involvement in neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy. Annals of Human Genetics, 2022, 86, 94-101.	0.3	3
8	Second report of SHMT2 related neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities. European Journal of Medical Genetics, 2022, 65, 104481.	0.7	2