Ayaz Khan

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4666632/publications.pdf

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7 papers	135 citations	1684188 5 h-index	1720034 7 g-index
7 all docs	7 docs citations	7 times ranked	438 citing authors

#	Article	IF	Citations
1	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328.	8.2	89
2	Homozygous GRID2 missense mutation predicts a shift in the D-serine binding domain of GluD2 in a case with generalized brain atrophy and unusual clinical features. BMC Medical Genetics, 2017, 18, 144.	2.1	21
3	Whole exome sequencing identifies novel variant underlying hereditary spastic paraplegia in consanguineous Pakistani families. Journal of Clinical Neuroscience, 2019, 67, 19-23.	1.5	8
4	Modifier Genes in Microcephaly: A Report on WDR62, CEP63, RAD50 and PCNT Variants Exacerbating Disease Caused by Biallelic Mutations of ASPM and CENPJ. Genes, 2021, 12, 731.	2.4	8
5	Updates on Clinical and Genetic Heterogeneity of ASPM in 12 Autosomal Recessive Primary Microcephaly Families in Pakistani Population. Frontiers in Pediatrics, 2021, 9, 695133.	1.9	5
6	A Homozygous AKNA Frameshift Variant Is Associated with Microcephaly in a Pakistani Family. Genes, 2021, 12, 1494.	2.4	3
7	Identification of a novel variant in GPR56/ADGRG1 gene through whole exome sequencing in a consanguineous Pakistani family. Journal of Clinical Neuroscience, 2021, 94, 8-12.	1.5	1