Julian SchrÃ¶ter

List of Publications by Year
in descending order

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9 papers	92 citations	1478280 6 h-index	1588896 8 g-index
9 all docs	9 docs citations	9 times ranked	168 citing authors

#	Article	IF	Citations
1	Refining Genotypes and Phenotypes in KCNA2-Related Neurological Disorders. International Journal of Molecular Sciences, 2021, 22, 2824.	1.8	20
2	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	1.1	19
3	Phenotypic diversity, disease progression, and pathogenicity of <scp>MVK</scp> missense variants in mevalonic aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 1272-1287.	1.7	17
4	Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	1.1	9
5	Complementing the phenotypical spectrum of TUBA1A tubulinopathy and its role in early-onset epilepsies. European Journal of Human Genetics, 2022, 30, 298-306.	1.4	9
6	Cross-sectional quantitative analysis of the natural history of TUBA1A and TUBB2B tubulinopathies. Genetics in Medicine, 2021, 23, 516-523.	1.1	8
7	Succinic Semialdehyde Dehydrogenase Deficiency: In Vitro and In Silico Characterization of a Novel Pathogenic Missense Variant and Analysis of the Mutational Spectrum of ALDH5A1. International Journal of Molecular Sciences, 2020, 21, 8578.	1.8	5
8	Quantitative retrospective natural history modeling of WDR45-related developmental and epileptic encephalopathy â€“ a systematic cross-sectional analysis of 160 published cases. Autophagy, 2022, 18, 1715-1727.	4.3	5
9	Generation of an induced pluripotent stem cell line (DHMCi008-A) from an individual with TUBA1A tubulinopathy. Stem Cell Research, 2022, 62, 102818.	0.3	0