Heiko Brennenstuhl

List of Publications by Year
in descending order

17	308	1162889 8	940416 16
papers	citations	h-index	g-index

21	21	21	373
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Gene replacement therapy with onasemnogene abeparvovec in children with spinal muscular atrophy aged 24 months or younger and bodyweight up to 15 kg: an observational cohort study. The Lancet Child and Adolescent Health, 2022, 6, 17-27.	2.7	57
2	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
3	Unmet Needs of Parents of Children with Urea Cycle Disorders. Children, 2022, 9, 712.	0.6	4
4	Refining Genotypes and Phenotypes in KCNA2-Related Neurological Disorders. International Journal of Molecular Sciences, 2021, 22, 2824.	1.8	20
5	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
6	Patterns of extreme temperature-related catastrophic events in Europe including the Russian Federation: a cross-sectional analysis of the Emergency Events Database. BMJ Open, 2021, 11, e046359.	0.8	9
7	The â€œYoung Metabolic Societyâ€• An interest group for young professionals in the field of metabolic medicine. Journal of Inherited Metabolic Disease, 2021, 44, 789-789.	1.7	0
8	Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	1.1	9
9	Assessment of intellectual impairment, healthâ€related quality of life, and behavioral phenotype in patients with neurotransmitter related disorders: Data from the <scp>iNTD</scp> registry. Journal of Inherited Metabolic Disease, 2021, 44, 1489-1502.	1.7	7
10	Compound heterozygosis in AADC deficiency: A complex phenotype dissected through comparison among heterodimeric and homodimeric AADC proteins. Molecular Genetics and Metabolism, 2021, 134, 147-155.	0.5	10
11	High throughput newborn screening for aromatic ÊŸâ€aminoâ€acid decarboxylase deficiency by analysis of concentrations of 3â€Oâ€methyldopa from dried blood spots. Journal of Inherited Metabolic Disease, 2020, 43, 602-610.	1.7	26
12	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
13	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
14	Rescue of respiratory failure in pulmonary alveolar proteinosis due to pathogenic MARS1 variants. Pediatric Pulmonology, 2020, 55, 3057-3066.	1.0	19
15	Semi-quantitative detection of a vanillactic acid/vanillylmandelic acid ratio in urine is a reliable diagnostic marker for aromatic L-amino acid decarboxylase deficiency. Molecular Genetics and Metabolism, 2020, 131, 163-170.	0.5	8
16	Succinic Semialdehyde Dehydrogenase Deficiency: An Update. Cells, 2020, 9, 477.	1.8	24
17	Inherited Disorders of Neurotransmitters: Classification and Practical Approaches for Diagnosis and Treatment. Neuropediatrics, 2019, 50, 002-014.	0.3	55