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List of Publications by Year in descending order

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9
papers

179
citations

1478280

6
h-index

1372474

10
g-index

11
all docs

11
docs citations

11
times ranked

160
citing authors

#	ARTICLE	IF	CITATIONS
1	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies. Orphanet Journal of Rare Diseases, 2020, 15, 126.	1.2	85
2	Analysis of Catecholamines and Pterins in Inborn Errors of Monoamine Neurotransmitter Metabolismâ€”From Past to Future. Cells, 2019, 8, 867.	1.8	22
3	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nature Communications, 2021, 12, 5529.	5.8	21
4	Brain <scp>MR</scp> patterns in inherited disorders of monoamine neurotransmitters: An analysis of 70 patients. Journal of Inherited Metabolic Disease, 2021, 44, 1070-1082.	1.7	13
5	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
6	Pregnancy management and outcome in patients with four different tetrahydrobiopterin disorders. Journal of Inherited Metabolic Disease, 2018, 41, 849-863.	1.7	6
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	Integrative Approach to Predict Severity in Nonketotic Hyperglycinemia. Annals of Neurology, 2022, 92, 292-303.	2.8	3
9	Volumetric study of brain MRI in a cohort of patients with neurotransmitter disorders. Neuroradiology, 0, , .	1.1	1