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List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6885682/publications.pdf>

Version: 2024-02-01

8
papers

168
citations

1478458

6
h-index

1588975

8
g-index

8
all docs

8
docs citations

8
times ranked

319
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical pattern, mutations and in vitro residual activity in 33 patients with severe 5, 10 methylenetetrahydrofolate reductase (MTHFR) deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 115-124.	3.6	52
2	Translational Targeted Proteomics Profiling of Mitochondrial Energy Metabolic Pathways in Mouse and Human Samples. <i>Journal of Proteome Research</i> , 2016, 15, 3204-3213.	3.7	40
3	Living on the edge: substrate competition explains loss of robustness in mitochondrial fatty-acid oxidation disorders. <i>BMC Biology</i> , 2016, 14, 107.	3.8	27
4	Neonatal screening for profound biotinidase deficiency in the Netherlands: consequences and considerations. <i>European Journal of Human Genetics</i> , 2016, 24, 1424-1429.	2.8	17
5	Homozygous damaging SOD2 variant causes lethal neonatal dilated cardiomyopathy. <i>Journal of Medical Genetics</i> , 2020, 57, 23-30.	3.2	16
6	Dopaminergic and serotonergic alterations in plasma in three groups of dystonia patients. <i>Parkinsonism and Related Disorders</i> , 2021, 91, 48-54.	2.2	8
7	The Mitochondrial Epigenome: An Unexplored Avenue to Explain Unexplained Myopathies?. <i>International Journal of Molecular Sciences</i> , 2022, 23, 2197.	4.1	7
8	Elevated granulocyte-colony stimulating factor and hematopoietic stem cell mobilization in Niemann-Pick type C1 disease. <i>Journal of Lipid Research</i> , 2022, 63, 100167.	4.2	1