

Allison Matthews

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/11472398/publications.pdf>

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5
papers

173
citations

1684188

5
h-index

2053705

5
g-index

5
all docs

5
docs citations

5
times ranked

663
citing authors

#	ARTICLE	IF	CITATIONS
1	Integration of genomics and metabolomics for prioritization of rare disease variants: a 2018 literature review. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 435-445.	3.6	35
2	Optic atrophy, cataracts, lipodystrophy/lipoatrophy, and peripheral neuropathy caused by a de novo <i>OPA3</i> mutation. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a001156.	1.2	11
3	Novel homozygous PCK1 mutation causing cytosolic phosphoenolpyruvate carboxykinase deficiency presenting as childhood hypoglycemia, an abnormal pattern of urine metabolites and liver dysfunction. <i>Molecular Genetics and Metabolism</i> , 2017, 120, 337-341.	1.1	29
4	Assessment of the ExAC data set for the presence of individuals with pathogenic genotypes implicated in severe Mendelian pediatric disorders. <i>Genetics in Medicine</i> , 2017, 19, 1300-1308.	2.4	58
5	Secondary neurotransmitter deficiencies in epilepsy caused by voltage-gated sodium channelopathies: A potential treatment target?. <i>Molecular Genetics and Metabolism</i> , 2016, 117, 42-48.	1.1	40