

Lisa M Crowther

List of Publications by Year in descending order

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Version: 2024-02-01

7
papers

267
citations

1307594

7
h-index

1720034

7
g-index

7
all docs

7
docs citations

7
times ranked

585
citing authors

#	ARTICLE	IF	CITATIONS
1	Cln5 represents a new type of cysteine-based <i>S</i> -depalmitoylase linked to neurodegeneration. Science Advances, 2022, 8, eabj8633.	10.3	12
2	New insights into human lysine degradation pathways with relevance to pyridoxine-dependent epilepsy due to antiquitin deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 620-628.	3.6	45
3	The role of recessive inheritance in early-onset epileptic encephalopathies: a combined whole-exome sequencing and copy number study. European Journal of Human Genetics, 2019, 27, 408-421.	2.8	52
4	Confirmation of mutations in <i>PROSC</i> as a novel cause of vitamin B ₆ -dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814.	3.2	66
5	Plasma metabolomics reveals a diagnostic metabolic fingerprint for mitochondrial aconitase (ACO2) deficiency. PLoS ONE, 2017, 12, e0176363.	2.5	40
6	The value of plasma vitamin B ₆ profiles in early onset epileptic encephalopathies. Journal of Inherited Metabolic Disease, 2016, 39, 733-741.	3.6	19
7	N ⁸ -acetylspermidine as a potential plasma biomarker for Snyder-Robinson syndrome identified by clinical metabolomics. Journal of Inherited Metabolic Disease, 2016, 39, 131-137.	3.6	33