Lisa M Crowther

List of Publications by Year
in descending order

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	Confirmation of mutations inPROSCas a novel cause of vitamin B_₆-dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814.	3.2	66
2	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
3	New insights into human lysine degradation pathways with relevance to pyridoxineâ€dependent epilepsy due to antiquitin deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 620-628.	3.6	45
4	Plasma metabolomics reveals a diagnostic metabolic fingerprint for mitochondrial aconitase (ACO2) deficiency. PLoS ONE, 2017, 12, e0176363.	2.5	40
5	N⁸â€acetylspermidine as a potential plasma biomarker for Snyderâ€Robinson syndrome identified by clinical metabolomics. Journal of Inherited Metabolic Disease, 2016, 39, 131-137.	3.6	33
6	The value of plasma vitamin B₆ profiles in early onset epileptic encephalopathies. Journal of Inherited Metabolic Disease, 2016, 39, 733-741.	3.6	19
7	Cln5 represents a new type of cysteine-based S -depalmitoylase linked to neurodegeneration. Science Advances, 2022, 8, eabj8633.	10.3	12