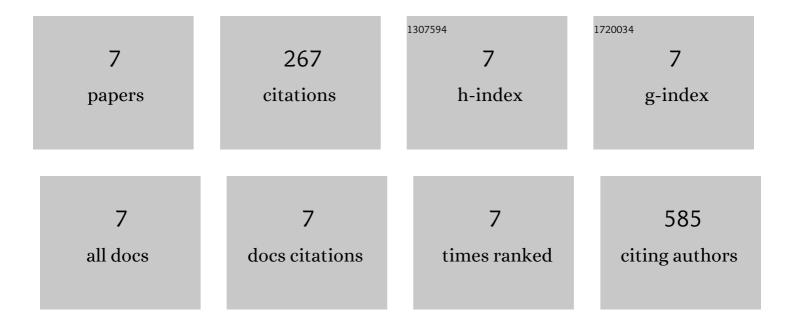
Lisa M Crowther

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/6530372/publications.pdf Version: 2024-02-01



#	Article	IF	CITATIONS
1	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
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	N ⁸ â€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 B ₆ 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 <i>S</i> -depalmitoylase linked to neurodegeneration. Science Advances, 2022, 8, eabj8633.	10.3	12