Marie Zikanova

List of Publications by Year
in descending order

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28 papers	617 citations	687363 13 h-index	610901 24 g-index
32 all docs	32 docs citations	32 times ranked	682 citing authors

#	Article	IF	Citations
1	Adenylosuccinate lyase deficiency. Journal of Inherited Metabolic Disease, 2015, 38, 231-242.	3.6	119
2	Clinical, biochemical and molecular findings in seven Polish patients with adenylosuccinate lyase deficiency. Molecular Genetics and Metabolism, 2008, 94, 435-442.	1.1	62
3	Mutations of ATIC and ADSL affect purinosome assembly in cultured skin fibroblasts from patients with AICA-ribosiduria and ADSL deficiency. Human Molecular Genetics, 2012, 21, 1534-1543.	2.9	62
4	Lethal Fetal and Early Neonatal Presentation of Adenylosuccinate Lyase Deficiency: Observation of 6 Patients in 4 Families. Journal of Pediatrics, 2007, 150, 57-61.e2.	1.8	58
5	Biochemical and structural analysis of 14 mutant adsl enzyme complexes and correlation to phenotypic heterogeneity of adenylosuccinate lyase deficiency. Human Mutation, 2010, 31, 445-455.	2.5	45
6	CRISPR-Cas9 induced mutations along de novo purine synthesis in HeLa cells result in accumulation of individual enzyme substrates and affect purinosome formation. Molecular Genetics and Metabolism, 2016, 119, 270-277.	1.1	33
7	Prolyl hydroxylase substrate adenylosuccinate lyase is an oncogenic driver in triple negative breast cancer. Nature Communications, 2019, 10, 5177.	12.8	27
8	Familial juvenile hyperuricaemic nephropathy (FJHN): linkage analysis in 15 families, physical and transcriptional characterisation of the FJHN critical region on chromosome 16p11.2 and the analysis of seven candidate genes. European Journal of Human Genetics, 2003, 11, 145-154.	2.8	25
9	Preparation of 5-amino-4-imidazole-N-succinocarboxamide ribotide, 5-amino-4-imidazole-N-succinocarboxamide riboside and succinyladenosine, compounds usable in diagnosis and research of adenylosuccinate lyase deficiency. Journal of Inherited Metabolic Disease, 2005, 28, 493-499.	3.6	22
10	PAICS deficiency, a new defect of de novo purine synthesis resulting in multiple congenital anomalies and fatal outcome. Human Molecular Genetics, 2019, 28, 3805-3814.	2.9	22
11	d-Ribose therapy in four Polish patients with adenylosuccinate lyase deficiency: Absence of positive effect. Journal of Inherited Metabolic Disease, 2008, 31, 329-332.	3.6	20
12	Mass spectrometric analysis of purine de novo biosynthesis intermediates. PLoS ONE, 2018, 13, e0208947.	2.5	16
13	Genetic and metabolomic analysis of AdeD and AdeI mutants of de novo purine biosynthesis: Cellular models of de novo purine biosynthesis deficiency disorders. Molecular Genetics and Metabolism, 2013, 108, 178-189.	1.1	14
14	Attenuated Adenylosuccinate Lyase Deficiency: A Report of One Case and a Review of the Literature. Neuropediatrics, 2014, 45, 050-055.	0.6	13
15	Clinical manifestations and molecular aspects of phosphoribosylpyrophosphate synthetase superactivity in females. Rheumatology, 2018, 57, 1180-1185.	1.9	12
16	The CRISPR-Cas9 crADSL HeLa transcriptome: A first step in establishing a model for ADSL deficiency and SAICAR accumulation. Molecular Genetics and Metabolism Reports, 2019, 21, 100512.	1.1	12
17	Molecular characterization of the AdeI mutant of Chinese hamster ovary cells: A cellular model of adenylosuccinate lyase deficiency. Molecular Genetics and Metabolism, 2011, 102, 61-68.	1.1	9
18	Study of purinosome assembly in cell-based model systems with de novo purine synthesis and salvage pathway deficiencies. PLoS ONE, 2018, 13, e0201432.	2.5	8

#	Article	IF	Citations
19	The need for vigilance: False-negative screening for adenylosuccinate lyase deficiency caused by deribosylation of urinary biomarkers. Clinical Biochemistry, 2013, 46, 1899-1901.	1.9	7
20	Screening for adenylosuccinate lyase deficiency using tandem mass spectrometry analysis of succinylpurines in neonatal dried blood spots. Clinical Biochemistry, 2015, 48, 2-7.	1.9	7
21	The CRISPR-Cas9 crATIC HeLa transcriptome: Characterization of a novel cellular model of ATIC deficiency and ZMP accumulation. Molecular Genetics and Metabolism Reports, 2020, 25, 100642.	1.1	7
22	Pathway-specific effects of ADSL deficiency on neurodevelopment. ELife, 2022, 11, .	6.0	7
23	Combined Targeted and Untargeted Profiling of HeLa Cells Deficient in Purine De Novo Synthesis. Metabolites, 2022, 12, 241.	2.9	3
24	Transcriptome and metabolome analysis of crGART, a novel cell model of de novo purine synthesis deficiency: Alterations in CD36 expression and activity. PLoS ONE, 2021, 16, e0247227.	2.5	2
25	Oligodendroglia from ADSL-deficient patient produce SAICAribotide and SAMP. Molecular Genetics and Metabolism, 2010, 101, 286-288.	1.1	1
26	Age Dependent Progression of Multiple Epiphyseal Dysplasia and Pseudoachondroplasia Due to Heterozygous Mutations in COMP Gene. Prague Medical Report, 2020, 121, 153-162.	0.8	1
27	Biochemicalâ€structuralâ€phenotypic correlation of adenylosuccinate lyase deficiency based on analysis of mutant enzyme complexes. FASEB Journal, 2010, 24, 889.7.	0.5	0
28	Modified HeLa Cells and C.elegans Multicellular Model as Unique Systems for the Study of Purinosome Formation. FASEB Journal, 2018, 32, 527.16.	0.5	0