David S Sinasac

List of Publications by Year
in descending order

18	792	759233 12	839539 18
papers	citations	h-index	g-index

18	18	18	872
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The gene mutated in adult-onset type II citrullinaemia encodes a putative mitochondrial carrier protein. Nature Genetics, 1999, 22, 159-163.	21.4	392
2	Slc25a13 -Knockout Mice Harbor Metabolic Deficits but Fail To Display Hallmarks of Adult-Onset Type II Citrullinemia. Molecular and Cellular Biology, 2004, 24, 527-536.	2.3	69
3	Citrin/Mitochondrial Glycerol-3-phosphate Dehydrogenase Double Knock-out Mice Recapitulate Features of Human Citrin Deficiency. Journal of Biological Chemistry, 2007, 282, 25041-25052.	3.4	65
4	PISDis a mitochondrial disease gene causing skeletal dysplasia, cataracts, and white matter changes. Life Science Alliance, 2019, 2, e201900353.	2.8	41
5	Pyruvate ameliorates the defect in ureogenesis from ammonia in citrin-deficient mice. Journal of Hepatology, 2006, 44, 930-938.	3.7	32
6	Metabolomic analysis reveals hepatic metabolite perturbations in citrin/mitochondrial glycerol-3-phosphate dehydrogenase double-knockout mice, a model of human citrin deficiency. Molecular Genetics and Metabolism, 2011, 104, 492-500.	1.1	30
7	Characterization of the C584R variant in the mtDNA depletion syndrome gene FBXL4, reveals a novel role for FBXL4 as a regulator of mitochondrial fusion. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 165536.	3.8	25
8	SS-31 Peptide Reverses the Mitochondrial Fragmentation Present in Fibroblasts From Patients With DCMA, a Mitochondrial Cardiomyopathy. Frontiers in Cardiovascular Medicine, 2019, 6, 167.	2.4	24
9	ALG9-CDG: New clinical case and review of the literature. Molecular Genetics and Metabolism Reports, 2017, 13, 55-63.	1.1	21
10	Effects of supplementation on food intake, body weight and hepatic metabolites in the citrin/mitochondrial glycerol-3-phosphate dehydrogenase double-knockout mouse model of human citrin deficiency. Molecular Genetics and Metabolism, 2012, 107, 322-329.	1.1	20
11	A New Patient With Intermediate Severe Salla DiseaseÂWith Hypomyelination: A Literature Review for Salla Disease. Pediatric Neurology, 2017, 74, 87-91.e2.	2.1	18
12	Transiently elevated plasma methionine, S â€adenosylmethionine and S â€adenosylhomocysteine: Unreported laboratory findings in a patient with NGLY1 deficiency, a congenital disorder of deglycosylation. JIMD Reports, 2019, 49, 21-29.	1.5	16
13	Oral aversion to dietary sugar, ethanol and glycerol correlates with alterations in specific hepatic metabolites in a mouse model of human citrin deficiency. Molecular Genetics and Metabolism, 2017, 120, 306-316.	1.1	10
14	Mechanism for increased hepatic glycerol synthesis in the citrin/mitochondrial glycerol-3-phosphate dehydrogenase double-knockout mouse: Urine glycerol and glycerol 3-phosphate as potential diagnostic markers of human citrin deficiency. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 1787-1795.	3.8	9
15	Pivotal role of inter-organ aspartate metabolism for treatment of mitochondrial aspartate-glutamate carrier 2 (citrin) deficiency, based on the mouse model. Scientific Reports, 2019, 9, 4179.	3.3	8
16	Phenotype and pathology of the dilated cardiomyopathy with ataxia syndrome in children. Journal of Inherited Metabolic Disease, 2022, 45, 366-376.	3.6	7
17	Hybrid gel electrophoresis using skin fibroblasts to aid in diagnosing mitochondrial disease. Neurology: Genetics, 2019, 5, e336.	1.9	4
18	<scp>3â€Hydroxyisobutyric</scp> acid dehydrogenase deficiency: Expanding the clinical spectrum and quantitation of Dâ€and <scp>Lâ€3â€Hydroxyisobutyric</scp> acid by an <scp>LCâ€“MS</scp> / <scp>MS</scp> method. Journal of Inherited Metabolic Disease, 2022, , .	3.6	1