Stephen D Cederbaum

List of Publications by Year
in descending order

31	1,021	394421	434195
papers	citations	19 h-index	31 g-index

32	32	32	1135
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Intermittent lipid nanoparticle mRNA administration prevents cortical dysmyelination associated with arginase deficiency. Molecular Therapy - Nucleic Acids, 2022, 28, 859-874.	5.1	9
2	Arginine to ornithine ratio as a diagnostic marker in patients with positive newborn screening for hyperargininemia. Molecular Genetics and Metabolism Reports, 2021, 27, 100735.	1.1	6
3	Confidential genetic testing and electronic health records: A survey of current practices among Huntington disease testing centers. Molecular Genetics & Genomic Medicine, 2020, 8, e1026.	1.2	8
4	Myopathy, lactic acidosis and sideroblastic anemia 1 (MLASA1): A 25-year follow-up. Molecular Genetics and Metabolism Reports, 2019, 21, 100517.	1.1	7
5	Lipid nanoparticle-targeted mRNA therapy as a treatment for the inherited metabolic liver disorder arginase deficiency. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 21150-21159.	7.1	86
6	Hepatic arginase deficiency fosters dysmyelination during postnatal CNS development. JCI Insight, 2019, 4, .	5.0	14
7	Human hepatocyte transplantation corrects the inherited metabolic liver disorder arginase deficiency in mice. Molecular Genetics and Metabolism, 2018, 124, 114-123.	1.1	7
8	Is it time to retire fragile X testing as a first-tier test for developmental delay, intellectual disability, and autism spectrum disorder?. Genetics in Medicine, 2017, 19, 1380-1381.	2.4	15
9	Restoring Ureagenesis in Hepatocytes by CRISPR/Cas9-mediated Genomic Addition to Arginase-deficient Induced Pluripotent Stem Cells. Molecular Therapy - Nucleic Acids, 2016, 5, e394.	5.1	30
10	Rescue of the Functional Alterations of Motor Cortical Circuits in Arginase Deficiency by Neonatal Gene Therapy. Journal of Neuroscience, 2016, 36, 6680-6690.	3.6	16
11	Interpreting sequence variants in a clinical context. Genetics in Medicine, 2015, 17, 1012-1012.	2.4	4
12	Myocyte-mediated Arginase Expression Controls Hyperargininemia but not Hyperammonemia in Arginase-deficient Mice. Molecular Therapy, 2014, 22, 1792-1802.	8.2	24
13	Lethal phenotype in conditional late-onset arginase 1 deficiency in the mouse. Molecular Genetics and Metabolism, 2013, 110, 222-230.	1.1	29
14	Long-term Survival of the Juvenile Lethal Arginase-deficient Mouse With AAV Gene Therapy. Molecular Therapy, 2012, 20, 1844-1851.	8.2	40
15	Diffusion Tensor Imaging in Arginase Deficiency Reveals Damage to Corticospinal Tracts. Pediatric Neurology, 2010, 42, 49-52.	2.1	27
16	Guanidino compound levels in blood, cerebrospinal fluid, and post-mortem brain material of patients with argininemia. Molecular Genetics and Metabolism, 2010, 100, S31-S36.	1.1	45
17	Short-term Correction of Arginase Deficiency in a Neonatal Murine Model With a Helper-dependent Adenoviral Vector. Molecular Therapy, 2009, 17, 1155-1163.	8.2	29
18	Increased plasma and tissue guanidino compounds in a mouse model of hyperargininemia. Molecular Genetics and Metabolism, 2008, 93, 172-178.	1.1	37

#	Article	IF	Citations
19	New frontiers in hereditary metabolic disease: An historical perspective. Molecular Genetics and Metabolism, 2006, 87, 184-189.	1.1	1
20	Arginase deficiency with lethal neonatal expression: Evidence for the glutamine hypothesis of cerebral edema. Journal of Pediatrics, 2003, 142, 349-352.	1.8	44
21	Mouse Model for Human Arginase Deficiency. Molecular and Cellular Biology, 2002, 22, 4491-4498.	2.3	152
22	Phenylketonuria: an update. Current Opinion in Pediatrics, 2002, 14, 702-706.	2.0	33
23	Carnitine membrane transporter deficiency: a long-term follow up and OCTN2 mutation in the first documented case of primary carnitine deficiency. Molecular Genetics and Metabolism, 2002, 77, 195-201.	1.1	58
24	Expression of arginase isozymes in mouse brain. Journal of Neuroscience Research, 2001, 66, 406-422.	2.9	71
25	VOIDING DYSFUNCTION IN A MOTHER AND DAUGHTER WITH MITOCHONDRIAL CYTOPATHY. Journal of Urology, 1998, 160, 830-830.	0.4	1
26	Delivery of cytosolic liver arginase into the mitochondrial matrix space: A possible novel site for gene replacement therapy. Somatic Cell and Molecular Genetics, 1996, 22, 489-498.	0.7	3
27	Identification of mutations (D128G, H141L) in the liver arginase gene of patients with hyperargininemia. Human Mutation, 1994, 4, 150-154.	2.5	36
28	Arginase deficiency manifesting delayed clinical sequelae and induction of a kidney arginase isozyme. Human Genetics, 1993, 91, 1-5.	3.8	50
29	Deletion in Blood Mitochondrial DNA in Kearns-Sayre Syndrome. Pediatric Research, 1992, 31, 557-560.	2.3	29
30	Hyperammonemia following allogeneic bone marrow transplantation. American Journal of Hematology, 1991, 38, 140-141.	4.1	31
31	Sensitivity to Carbohydrate in a Patient with Familial Intermittent Lactic Acidosis and Pyruvate Dehydrogenase Deficiency. Pediatric Research, 1976, 10, 713-720.	2.3	79