DesirÃ©e Ec Smith

List of Publications by Year
in descending order

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53 papers	1,728 citations	257101 24 h-index	301761 39 g-index
54 all docs	54 docs citations	54 times ranked	2857 citing authors

#	Article	IF	Citations
1	Dihydrofolate Reductase Deficiency Due to a Homozygous DHFR Mutation Causes Megaloblastic Anemia and Cerebral Folate Deficiency Leading to Severe Neurologic Disease. American Journal of Human Genetics, 2011, 88, 226-231.	2.6	108
2	Adenosine Kinase Deficiency Disrupts the Methionine Cycle and Causes Hypermethioninemia, Encephalopathy, and Abnormal Liver Function. American Journal of Human Genetics, 2011, 89, 507-515.	2.6	104
3	Global DNA methylation measured by liquid chromatography-tandem mass spectrometry: analytical technique, reference values and determinants in healthy subjects. Clinical Chemistry and Laboratory Medicine, 2007, 45, 903-11.	1.4	90
4	Identification and Characterization of an Inborn Error of Metabolism Caused by Dihydrofolate Reductase Deficiency. American Journal of Human Genetics, 2011, 88, 216-225.	2.6	90
5	<i>S</i>-Adenosylmethionine Is Decreased in the Cerebrospinal Fluid of Patients with Alzheimerâ€™s Disease. Neurodegenerative Diseases, 2010, 7, 373-378.	0.8	88
6	5-Methyltetrahydrofolic acid and folic acid measured in plasma with liquid chromatography tandem mass spectrometry: applications to folate absorption and metabolism. Analytical Biochemistry, 2004, 326, 129-138.	1.1	71
7	Cellular folate vitamer distribution during and after correction of vitamin B12 deficiency: a case for the methylfolate trap. British Journal of Haematology, 2006, 132, 623-629.	1.2	62
8	Bi-allelic Mutations in EPRS, Encoding the Glutamyl-Prolyl-Aminoacyl-tRNA Synthetase, Cause a Hypomyelinating Leukodystrophy. American Journal of Human Genetics, 2018, 102, 676-684.	2.6	58
9	Evaluation of the novel folate receptor ligand [18F]fluoro-PEG-folate for macrophage targeting in a rat model of arthritis. Arthritis Research and Therapy, 2013, 15, R37.	1.6	57
10	Quantitative determination of erythrocyte folate vitamer distribution by liquid chromatography-tandem mass spectrometry. Clinical Chemistry and Laboratory Medicine, 2006, 44, 450-9.	1.4	56
11	Homocysteine-Induced Apoptosis in Endothelial Cells Coincides With Nuclear NOX2 and Peri-nuclear NOX4 Activity. Cell Biochemistry and Biophysics, 2013, 67, 341-352.	0.9	54
12	Folic acid supplementation does not reduce intracellular homocysteine, and may disturb intracellular one-carbon metabolism. Clinical Chemistry and Laboratory Medicine, 2013, 51, 1643-1650.	1.4	46
13	Loss of NARS1 impairs progenitor proliferation in cortical brain organoids and leads to microcephaly. Nature Communications, 2020, 11, 4038.	5.8	44
14	Arginine and Mixed Amino Acids Increase Protein Accretion in the Growth-Restricted and Normal Ovine Fetus by Different Mechanisms. Pediatric Research, 2005, 58, 270-277.	1.1	43
15	Bi-allelic TARS Mutations Are Associated with Brittle Hair Phenotype. American Journal of Human Genetics, 2019, 105, 434-440.	2.6	42
16	Homocysteine affects cardiomyocyte viability: concentration-dependent effects on reversible flip-flop, apoptosis and necrosis. Apoptosis: an International Journal on Programmed Cell Death, 2007, 12, 1407-1418.	2.2	41
17	Analysis of polyols in urine by liquid chromatographyâ€“tandem mass spectrometry: A useful tool for recognition of inborn errors affecting polyol metabolism. Journal of Inherited Metabolic Disease, 2005, 28, 951-963.	1.7	40
18	Red blood cell folate vitamer distribution in healthy subjects is determined by the methylenetetrahydrofolate reductase C677T polymorphism and by the total folate status. Journal of Nutritional Biochemistry, 2007, 18, 693-699.	1.9	37

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19	Simultaneous determination of asymmetric and symmetric dimethylarginine, l-monomethylarginine, l-arginine, and l-homoarginine in biological samples using stable isotope dilution liquid chromatography tandem mass spectrometry. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2012, 900, 38-47.	1.2	37
20	Homocysteine clearance and methylation flux rates in health and end-stage renal disease: association withS-adenosylhomocysteine. American Journal of Physiology - Renal Physiology, 2004, 287, F215-F223.	1.3	36
21	Detection of transaldolase deficiency by quantification of novel seven-carbon chain carbohydrate biomarkers in urine. Journal of Inherited Metabolic Disease, 2007, 30, 735-742.	1.7	34
22	Biallelic variants in LARS2 and KARS cause deafness and (ovario)leukodystrophy. Neurology, 2019, 92, e1225-e1237.	1.5	32
23	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	2.6	32
24	Cysteinyl-tRNA Synthetase Mutations Cause a Multi-System, Recessive Disease That Includes Microcephaly, Developmental Delay, and Brittle Hair and Nails. American Journal of Human Genetics, 2019, 104, 520-529.	2.6	31
25	[6S]5-methyltetrahydrofolate or folic acid supplementation and absorption and initial elimination of folate in young and middle-aged adults. European Journal of Clinical Nutrition, 2005, 59, 1409-1416.	1.3	26
26	Determinants of the essential one-carbon metabolism metabolites, homocysteine, S-adenosylmethionine, S-adenosylhomocysteine and folate, in cerebrospinal fluid. Clinical Chemistry and Laboratory Medicine, 2012, 50, 1641-7.	1.4	26
27	Hyperhomocysteinemia in Alzheimer's Disease: The Hen and the Egg?. Journal of Alzheimer's Disease, 2013, 33, 1097-1104.	1.2	25
28	A pilot study to estimate incidence of guanidinoacetate methyltransferase deficiency in newborns by direct sequencing of the GAMT gene. Gene, 2016, 575, 127-131.	1.0	24
29	Plasma choline and betaine correlate with serum folate, plasma S-adenosyl-methionine and S-adenosyl-homocysteine in healthy volunteers. Clinical Chemistry and Laboratory Medicine, 2013, 51, 683-92.	1.4	22
30	Reduced response of Cystathionine Betaâ€Synthase (CBS) to Sâ€Adenosylmethionine (SAM): Identification and functional analysis of CBS gene mutations in Homocystinuria patients. Journal of Inherited Metabolic Disease, 2014, 37, 245-254.	1.7	21
31	Protein instability associated with AARS1 and MARS1 mutations causes trichothiodystrophy. Human Molecular Genetics, 2021, 30, 1711-1720.	1.4	20
32	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	1.1	19
33	Rescue of respiratory failure in pulmonary alveolar proteinosis due to pathogenic MARS1 variants. Pediatric Pulmonology, 2020, 55, 3057-3066.	1.0	19
34	Clinically Distinct Phenotypes of Canavan Disease Correlate with Residual Aspartoacylase Enzyme Activity. Human Mutation, 2017, 38, 524-531.	1.1	18
35	RARS1â€related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93.	1.7	18
36	Intrauterine valproate exposure is associated with alterations in hippocampal cell numbers and folate metabolism in a rat model of valproate teratogenicity. Seizure: the Journal of the British Epilepsy Association, 2017, 46, 7-12.	0.9	17

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37	Folic Acid Impairs the Uptake of 5-Methyltetrahydrofolate in Human Umbilical Vascular Endothelial Cells. Journal of Cardiovascular Pharmacology, 2017, 70, 271-275.	0.8	16
38	<scp>FARS1</scp>â€related disorders caused by biâ€allelic mutations in cytosolic phenylalanylâ€<scp>tRNA</scp> synthetase genes: Look beyond the lungs!. Clinical Genetics, 2021, 99, 789-801.	1.0	16
39	Insights into the Regulatory Domain of Cystathionine Beta-Synthase: Characterization of Six Variant Proteins. Human Mutation, 2014, 35, 1195-1202.	1.1	15
40	Homocysteine-induced cardiomyocyte apoptosis and plasma membrane flip-flop are independent of S-adenosylhomocysteine: a crucial role for nuclear p47phox. Molecular and Cellular Biochemistry, 2011, 358, 229-239.	1.4	13
41	Methionine metabolism in an animal model of sepsis. Clinical Chemistry and Laboratory Medicine, 2008, 46, 1398-402.	1.4	11
42	Changes in intracellular folate metabolism during high-dose methotrexate and Leucovorin rescue therapy in children with acute lymphoblastic leukemia. PLoS ONE, 2019, 14, e0221591.	1.1	10
43	Methylation metabolism in sepsis and systemic inflammatory response syndrome. Scandinavian Journal of Clinical and Laboratory Investigation, 2013, 73, 368-372.	0.6	8
44	Small aminothiol compounds improve the function of Arg to Cys variant proteins: effect on the human cystathionine Î²-synthase p.R336C. Human Molecular Genetics, 2015, 24, 7339-7348.	1.4	8
45	Functional analysis of thirty-four suspected pathogenic missense variants in ALDH5A1 gene associated with succinic semialdehyde dehydrogenase deficiency. Molecular Genetics and Metabolism, 2020, 130, 172-178.	0.5	8
46	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	1.1	8
47	A liquid chromatography mass spectrometry method for the measurement of cystathionine Î²-synthase activity in cell extracts. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2012, 911, 186-191.	1.2	7
48	Post-transcriptional regulation of the creatine transporter gene: Functional relevance of alternative splicing. Biochimica Et Biophysica Acta - General Subjects, 2014, 1840, 2070-2079.	1.1	7
49	A biâ€allelic lossâ€ofâ€function SARS1 variant in children with neurodevelopmental delay, deafness, cardiomyopathy, and decompensation during fever. Human Mutation, 2021, 42, 1576-1583.	1.1	6
50	The ratio of S-adenosylmethione and S-adenosyl-homocysteine is increased in the brains of newborn rats in a model of valproic acid teratogenicity. Toxicology, 2012, 293, 132-133.	2.0	4
51	Recurrent acute liver failure in alanyl-tRNA synthetase-1 (AARS1) deficiency. Molecular Genetics and Metabolism Reports, 2020, 25, 100681.	0.4	2
52	No effect of ornithine alphaketoglutarate on nitrogen excretion or urea synthesis rate in healthy male subjects. European E-journal of Clinical Nutrition and Metabolism, 2007, 2, 75-80.	0.4	0
53	Infantile Liver Failure Syndrome 1 associated with a novel variant of the <scp>LARS1</scp> gene: Clinical, genetic, and functional characterization. Clinical Genetics, 2021, 99, 601-603.	1.0	0