

# DesirÃ©e Ec Smith

## List of Publications by Year in descending order

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53  
papers

1,728  
citations

257357  
24  
h-index

302012  
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54  
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54  
docs citations

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times ranked

2857  
citing authors

#	ARTICLE	IF	CITATIONS
1	Infantile Liver Failure Syndrome 1 associated with a novel variant of the <i>LARS1</i> gene: Clinical, genetic, and functional characterization. <i>Clinical Genetics</i> , 2021, 99, 601-603.	1.0	0
2	<i>FARS1</i> -related disorders caused by bi-allelic mutations in cytosolic phenylalanyl-tRNA synthetase genes: Look beyond the lungs!. <i>Clinical Genetics</i> , 2021, 99, 789-801.	1.0	16
3	Protein instability associated with <i>AARS1</i> and <i>MARS1</i> mutations causes trichothiodystrophy. <i>Human Molecular Genetics</i> , 2021, 30, 1711-1720.	1.4	20
4	Expanded phenotype of <i>AARS1</i> -related white matter disease. <i>Genetics in Medicine</i> , 2021, 23, 2352-2359.	1.1	8
5	A bi-allelic loss-of-function <i>SARS1</i> variant in children with neurodevelopmental delay, deafness, cardiomyopathy, and decompensation during fever. <i>Human Mutation</i> , 2021, 42, 1576-1583.	1.1	6
6	<i>RARS1</i> -related hypomyelinating leukodystrophy: Expanding the spectrum. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 83-93.	1.7	18
7	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in <i>LARS1</i> . <i>Genetics in Medicine</i> , 2020, 22, 1863-1873.	1.1	19
8	De Novo and Bi-allelic Pathogenic Variants in <i>NARS1</i> Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. <i>American Journal of Human Genetics</i> , 2020, 107, 311-324.	2.6	32
9	Loss of <i>NARS1</i> impairs progenitor proliferation in cortical brain organoids and leads to microcephaly. <i>Nature Communications</i> , 2020, 11, 4038.	5.8	44
10	Rescue of respiratory failure in pulmonary alveolar proteinosis due to pathogenic <i>MARS1</i> variants. <i>Pediatric Pulmonology</i> , 2020, 55, 3057-3066.	1.0	19
11	Recurrent acute liver failure in alanyl-tRNA synthetase-1 ( <i>AARS1</i> ) deficiency. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100681.	0.4	2
12	Functional analysis of thirty-four suspected pathogenic missense variants in <i>ALDH5A1</i> gene associated with succinic semialdehyde dehydrogenase deficiency. <i>Molecular Genetics and Metabolism</i> , 2020, 130, 172-178.	0.5	8
13	Bi-allelic <i>TARS</i> Mutations Are Associated with Brittle Hair Phenotype. <i>American Journal of Human Genetics</i> , 2019, 105, 434-440.	2.6	42
14	Changes in intracellular folate metabolism during high-dose methotrexate and Leucovorin rescue therapy in children with acute lymphoblastic leukemia. <i>PLoS ONE</i> , 2019, 14, e0221591.	1.1	10
15	Biallelic variants in <i>LARS2</i> and <i>KARS</i> cause deafness and (ovario)leukodystrophy. <i>Neurology</i> , 2019, 92, e1225-e1237.	1.5	32
16	Cysteinyl-tRNA Synthetase Mutations Cause a Multi-System, Recessive Disease That Includes Microcephaly, Developmental Delay, and Brittle Hair and Nails. <i>American Journal of Human Genetics</i> , 2019, 104, 520-529.	2.6	31
17	Bi-allelic Mutations in <i>EPRS</i> , Encoding the Glutamyl-Prolyl-Aminoacyl-tRNA Synthetase, Cause a Hypomyelinating Leukodystrophy. <i>American Journal of Human Genetics</i> , 2018, 102, 676-684.	2.6	58
18	Clinically Distinct Phenotypes of Canavan Disease Correlate with Residual Aspartoacylase Enzyme Activity. <i>Human Mutation</i> , 2017, 38, 524-531.	1.1	18

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19	Intrauterine valproate exposure is associated with alterations in hippocampal cell numbers and folate metabolism in a rat model of valproate teratogenicity. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2017, 46, 7-12.	0.9	17
20	Folic Acid Impairs the Uptake of 5-Methyltetrahydrofolate in Human Umbilical Vascular Endothelial Cells. <i>Journal of Cardiovascular Pharmacology</i> , 2017, 70, 271-275.	0.8	16
21	A pilot study to estimate incidence of guanidinoacetate methyltransferase deficiency in newborns by direct sequencing of the GAMT gene. <i>Gene</i> , 2016, 575, 127-131.	1.0	24
22	Small aminothiol compounds improve the function of Arg to Cys variant proteins: effect on the human cystathionine $\beta$ -synthase p.R336C. <i>Human Molecular Genetics</i> , 2015, 24, 7339-7348.	1.4	8
23	Insights into the Regulatory Domain of Cystathionine Beta-Synthase: Characterization of Six Variant Proteins. <i>Human Mutation</i> , 2014, 35, 1195-1202.	1.1	15
24	Reduced response of Cystathionine Beta-Synthase (CBS) to S-Adenosylmethionine (SAM): Identification and functional analysis of CBS gene mutations in Homocystinuria patients. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 245-254.	1.7	21
25	Post-transcriptional regulation of the creatine transporter gene: Functional relevance of alternative splicing. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 2014, 1840, 2070-2079.	1.1	7
26	Evaluation of the novel folate receptor ligand [18F]fluoro-PEG-folate for macrophage targeting in a rat model of arthritis. <i>Arthritis Research and Therapy</i> , 2013, 15, R37.	1.6	57
27	Plasma choline and betaine correlate with serum folate, plasma S-adenosyl-methionine and S-adenosyl-homocysteine in healthy volunteers. <i>Clinical Chemistry and Laboratory Medicine</i> , 2013, 51, 683-92.	1.4	22
28	Homocysteine-Induced Apoptosis in Endothelial Cells Coincides With Nuclear NOX2 and Peri-nuclear NOX4 Activity. <i>Cell Biochemistry and Biophysics</i> , 2013, 67, 341-352.	0.9	54
29	Folic acid supplementation does not reduce intracellular homocysteine, and may disturb intracellular one-carbon metabolism. <i>Clinical Chemistry and Laboratory Medicine</i> , 2013, 51, 1643-1650.	1.4	46
30	Methylation metabolism in sepsis and systemic inflammatory response syndrome. <i>Scandinavian Journal of Clinical and Laboratory Investigation</i> , 2013, 73, 368-372.	0.6	8
31	Hyperhomocysteinemia in Alzheimer's Disease: The Hen and the Egg?. <i>Journal of Alzheimer's Disease</i> , 2013, 33, 1097-1104.	1.2	25
32	Determinants of the essential one-carbon metabolism metabolites, homocysteine, S-adenosylmethionine, S-adenosylhomocysteine and folate, in cerebrospinal fluid. <i>Clinical Chemistry and Laboratory Medicine</i> , 2012, 50, 1641-7.	1.4	26
33	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. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2012, 900, 38-47.	1.2	37
34	A liquid chromatography mass spectrometry method for the measurement of cystathionine $\beta$ -synthase activity in cell extracts. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2012, 911, 186-191.	1.2	7
35	The ratio of S-adenosylmethionine and S-adenosyl-homocysteine is increased in the brains of newborn rats in a model of valproic acid teratogenicity. <i>Toxicology</i> , 2012, 293, 132-133.	2.0	4
36	Identification and Characterization of an Inborn Error of Metabolism Caused by Dihydrofolate Reductase Deficiency. <i>American Journal of Human Genetics</i> , 2011, 88, 216-225.	2.6	90

37	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
38	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
39	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
40	&lt;i>&lt;i>&lt;i>-Adenosylmethionine Is Decreased in the Cerebrospinal Fluid of Patients with Alzheimerâ€™s Disease. Neurodegenerative Diseases, 2010, 7, 373-378.	0.8	88
41	Methionine metabolism in an animal model of sepsis. Clinical Chemistry and Laboratory Medicine, 2008, 46, 1398-402.	1.4	11
42	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
43	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
44	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
45	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
46	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
47	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
48	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
49	[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
50	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
51	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
52	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
53	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