Curtis R Coughlin Ii

List of Publications by Citations

57 papers	1,350 citations	21 h-index	36 g-index
67 ext. papers	1,725 ext. citations	4.9 avg, IF	3.97 L-index

#	Paper	IF	Citations
57	Variant non ketotic hyperglycinemia is caused by mutations in LIAS, BOLA3 and the novel gene GLRX5. *Brain*, **2014**, 137, 366-79	11.2	146
56	Pyridoxine-Dependent Epilepsy: An Expanding Clinical Spectrum. *Pediatric Neurology*, **2016**, 59, 6-12	2.9	110
55	An X-linked cobalamin disorder caused by mutations in transcriptional coregulator HCFC1. *American Journal of Human Genetics*, **2013**, 93, 506-14	11	90
54	Lysine restricted diet for pyridoxine-dependent epilepsy: first evidence and future trials. *Molecular Genetics and Metabolism*, **2012**, 107, 335-44	3.7	77
53	Triple therapy with pyridoxine, arginine supplementation and dietary lysine restriction in pyridoxine-dependent epilepsy: Neurodevelopmental outcome. *Molecular Genetics and Metabolism*, **2015**, 116, 35-43	3.7	75
52	Clinical and biochemical characterization of four patients with mutations in ECHS1. *Orphanet Journal of Rare Diseases*, **2015**, 10, 79	4.2	51
51	Mutations in the accessory subunit NDUFB10 result in isolated complex I deficiency and illustrate the critical role of intermembrane space import for complex I holoenzyme assembly. *Human Molecular Genetics*, **2017**, 26, 702-716	5.6	51
50	Mutations in the mitochondrial cysteinyl-tRNA synthase gene, CARS2, lead to a severe epileptic encephalopathy and complex movement disorder. *Journal of Medical Genetics*, **2015**, 52, 532-40	5.8	48
49	Biochemical and molecular predictors for prognosis in nonketotic hyperglycinemia. *Annals of Neurology*, **2015**, 78, 606-18	9.4	47
48	Clinical impact of copy number variation analysis using high-resolution microarray technologies: advantages, limitations and concerns. *Genome Medicine*, **2012**, 4, 80	14.4	44
47	Mitochondrial energy failure in HSD10 disease is due to defective mtDNA transcript processing. *Mitochondrion*, **2015**, 21, 1-10	4.9	40
46	Neurodevelopmental Outcome and Treatment Efficacy of Benzoate and Dextromethorphan in Siblings with Attenuated Nonketotic Hyperglycinemia. *Journal of Pediatrics*, **2016**, 170, 234-9	3.6	39
45	The genetic basis of classic nonketotic hyperglycinemia due to mutations in GLDC and AMT. *Genetics in Medicine*, **2017**, 19, 104-111	8.1	38
44	Evidence for a recurrent microdeletion at chromosome 16p11.2 associated with congenital anomalies of the kidney and urinary tract (CAKUT) and Hirschsprung disease. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2618-22	2.5	38
43	Penicillamine therapy for pediatric cystinuria: experience from a cohort of American children. *Journal of Urology*, **2008**, 180, 2620-3	2.5	34
42	The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy: A common epileptic encephalopathy. *Journal of Inherited Metabolic Disease*, **2019**, 42, 353-361	5.4	33
41	Very long-chain acyl-CoA dehydrogenase deficiency in a patient with normal newborn screening by tandem mass spectrometry. *Journal of Pediatrics*, **2010**, 156, 492-4	3.6	33

40	Lysine-Restricted Diet as Adjunct Therapy for Pyridoxine-Dependent Epilepsy: The PDE Consortium Consensus Recommendations. *JIMD Reports*, **2014**, 15, 1-11	1.9	32
39	Human recombinant arginase enzyme reduces plasma arginine in mouse models of arginase deficiency. *Human Molecular Genetics*, **2015**, 24, 6417-27	5.6	28
38	Developing a conceptual, reproducible, rubric-based approach to consent and result disclosure for genetic testing by clinicians with minimal genetics background. *Genetics in Medicine*, **2019**, 21, 727-735	8.1	26
37	Pathogenic variants in SQOR encoding sulfide:quinone oxidoreductase are a potentially treatable cause of Leigh disease. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1024-1036	5.4	22
36	The 22q11 low copy repeats are characterized by unprecedented size and structural variability. *Genome Research*, **2019**, 29, 1389-1401	9.7	21
35	Identification of a novel biomarker for pyridoxine-dependent epilepsy: Implications for newborn screening. *Journal of Inherited Metabolic Disease*, **2019**, 42, 565-574	5.4	21
34	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. *Annals of Neurology* , **2019**, 86, 116-128	9.4	20
33	Genotype-phenotype correlations: sudden death in an infant with very-long-chain acyl-CoA dehydrogenase deficiency. *Journal of Inherited Metabolic Disease*, **2010**, 33 Suppl 3, S129-31	5.4	20
32	Low bone mineral density is a common finding in patients with homocystinuria. *Molecular Genetics and Metabolism*, **2016**, 117, 351-4	3.7	17
31	Consensus guidelines for the diagnosis and management of pyridoxine-dependent epilepsy due to α-aminoadipic semialdehyde dehydrogenase deficiency. *Journal of Inherited Metabolic Disease*, **2021**, 44, 178-192	5.4	15
30	Brain imaging in classic nonketotic hyperglycinemia: Quantitative analysis and relation to phenotype. *Journal of Inherited Metabolic Disease*, **2019**, 42, 438-450	5.4	13
29	Distinctive pattern of restricted diffusion in a neonate with molybdenum cofactor deficiency. *Pediatric Radiology*, **2013**, 43, 882-5	2.8	13
28	Dihydropteridine reductase deficiency and treatment with tetrahydrobiopterin: a case report. *JIMD Reports*, **2013**, 10, 53-6	1.9	13
27	Somatic mosaicism for PDHA1 mutation in a male with pyruvate dehydrogenase complex deficiency. *Molecular Genetics and Metabolism*, **2010**, 100, 296-9	3.7	12
26	Developing interactions with industry in rare diseases: lessons learned and continuing challenges. *Genetics in Medicine*, **2020**, 22, 219-226	8.1	12
25	Discovery of a potentially deleterious variant in TMEM87B in a patient with a hemizygous 2q13 microdeletion suggests a recessive condition characterized by congenital heart disease and restrictive cardiomyopathy. *Journal of Physical Education and Sports Management*, **2016**, 2, a000844	2.8	11
24	Mild orotic aciduria in UMPS heterozygotes: a metabolic finding without clinical consequences. *Journal of Inherited Metabolic Disease*, **2017**, 40, 423-431	5.4	10
23	X-Linked Cobalamin Disorder (HCFC1) Mimicking Nonketotic Hyperglycinemia With Increased Both Cerebrospinal Fluid Glycine and Methylmalonic Acid. *Pediatric Neurology*, **2017**, 71, 65-69	2.9	9

22	The 22q11 low copy repeats are characterized by unprecedented size and structure variability		5
21	Conducting an investigator-initiated randomized double-blinded intervention trial in acute decompensation of inborn errors of metabolism: Lessons from the N-Carbamylglutamate Consortium. *Translational Science of Rare Diseases*, **2018**, 3, 157-170	3.3	5
20	The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy: a common epileptic encephalopathy. *Journal of Inherited Metabolic Disease*, **2018**,	5.4	4
19	Abnormal expression of GABA receptor subunits and hypomotility upon loss of in zebrafish. *Biology Open*, **2020**, 9,	2.2	4
18	Inconsistencies in the Nutrition Management of Glutaric Aciduria Type 1: An International Survey. *Nutrients*, **2020**, 12,	6.7	3
17	Cognitive and neurological outcome of patients in the Dutch pyridoxine-dependent epilepsy (PDE-ALDH7A1) cohort, a cross-sectional study. *European Journal of Paediatric Neurology*, **2021**, 33, 112-120	3.8	3
16	REVIEW: Practical strategies to maintain anabolism by intravenous nutritional management in children with inborn metabolic diseases. *Molecular Genetics and Metabolism*, **2021**, 133, 231-241	3.7	3
15	Genetic Testing: Consent and Result Disclosure for Primary Care Providers. *Medical Clinics of North America*, **2019**, 103, 967-976	7	3
14	Lethal neonatal hyperammonemia in severe ornithine transcarbamylase (OTC) deficiency compounded by large hepatic portosystemic shunt. *Journal of Inherited Metabolic Disease*, **2017**, 40, 159-160	5.4	2
13	Nonketotic Hyperglycinemia (Glycine Encephalopathy) and Lipoate Deficiency Disorders **2016**, 349-356		2
12	Timing of therapy and neurodevelopmental outcomes in 18 families with pyridoxine-dependent epilepsy.. *Molecular Genetics and Metabolism*, **2022**, 135, 350-356	3.7	2
11	Application of a framework to guide genetic testing communication across clinical indications. *Genome Medicine*, **2021**, 13, 71	14.4	1
10	Genomic regions associated with microdeletion/microduplication syndromes exhibit extreme diversity of structural variation. *Genetics*, **2021**, 217,	4	1
9	Development and application of an ethical framework for pediatric metabolic and bariatric surgery evaluation. *Surgery for Obesity and Related Diseases*, **2021**, 17, 425-433	3	0
8	Comment on Late-Onset Nonketotic Hyperglycinemia With a Heterozygous Novel Point Mutation of the GLDC Gene. *Pediatric Neurology*, **2018**, 79, e1	2.9	
7	Child neurology: a case illustrating the role of imaging in evaluation of sudden infant death. *Neurology*, **2009**, 73, e54-6	6.5	
6	Laboratory Evaluations in Inherited Metabolic Diseases **2015**, 75-85		
5	Glutaric Acidemia Type I: Diagnosis and Management **2015**, 203-209		

List of Publications

4	Pyridoxine-dependent epilepsy is more than just epilepsy. *Developmental Medicine and Child Neurology*, **2020**, 62, 268	3.3
3	Disorders of Glycine Metabolism **2022**, 469-478	
2	Fatty Acid Oxidation Disorders **2022**, 309-323	
1	Laboratory Evaluations in Inherited Metabolic Diseases **2022**, 85-96	