

# Curtis R Coughlin II

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Disorders of Glycine Metabolism <b>2022</b> , 469-478		
56	Timing of therapy and neurodevelopmental outcomes in 18 families with pyridoxine-dependent epilepsy.. <i>Molecular Genetics and Metabolism</i> , <b>2022</b> , 135, 350-356	3.7	2
55	Fatty Acid Oxidation Disorders <b>2022</b> , 309-323		
54	Laboratory Evaluations in Inherited Metabolic Diseases <b>2022</b> , 85-96		
53	Application of a framework to guide genetic testing communication across clinical indications. <i>Genome Medicine</i> , <b>2021</b> , 13, 71	14.4	1
52	Cognitive and neurological outcome of patients in the Dutch pyridoxine-dependent epilepsy (PDE-ALDH7A1) cohort, a cross-sectional study. <i>European Journal of Paediatric Neurology</i> , <b>2021</b> , 33, 112-120	3.8	3
51	REVIEW: Practical strategies to maintain anabolism by intravenous nutritional management in children with inborn metabolic diseases. <i>Molecular Genetics and Metabolism</i> , <b>2021</b> , 133, 231-241	3.7	3
50	Consensus guidelines for the diagnosis and management of pyridoxine-dependent epilepsy due to $\epsilon$ -aminoadipic semialdehyde dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , <b>2021</b> , 44, 178-192	5.4	15
49	Development and application of an ethical framework for pediatric metabolic and bariatric surgery evaluation. <i>Surgery for Obesity and Related Diseases</i> , <b>2021</b> , 17, 425-433	3	0
48	Genomic regions associated with microdeletion/microduplication syndromes exhibit extreme diversity of structural variation. <i>Genetics</i> , <b>2021</b> , 217,	4	1
47	Inconsistencies in the Nutrition Management of Glutaric Aciduria Type 1: An International Survey. <i>Nutrients</i> , <b>2020</b> , 12,	6.7	3
46	Pathogenic variants in SQOR encoding sulfide:quinone oxidoreductase are a potentially treatable cause of Leigh disease. <i>Journal of Inherited Metabolic Disease</i> , <b>2020</b> , 43, 1024-1036	5.4	22
45	Pyridoxine-dependent epilepsy is more than just epilepsy. <i>Developmental Medicine and Child Neurology</i> , <b>2020</b> , 62, 268	3.3	
44	Abnormal expression of GABA receptor subunits and hypomotility upon loss of in zebrafish. <i>Biology Open</i> , <b>2020</b> , 9,	2.2	4
43	Developing interactions with industry in rare diseases: lessons learned and continuing challenges. <i>Genetics in Medicine</i> , <b>2020</b> , 22, 219-226	8.1	12
42	The 22q11 low copy repeats are characterized by unprecedented size and structural variability. <i>Genome Research</i> , <b>2019</b> , 29, 1389-1401	9.7	21
41	Identification of a novel biomarker for pyridoxine-dependent epilepsy: Implications for newborn screening. <i>Journal of Inherited Metabolic Disease</i> , <b>2019</b> , 42, 565-574	5.4	21

40	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. <i>Annals of Neurology</i> , <b>2019</b> , 86, 116-128	9.4	20
39	The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy: A common epileptic encephalopathy. <i>Journal of Inherited Metabolic Disease</i> , <b>2019</b> , 42, 353-361	5.4	33
38	Brain imaging in classic nonketotic hyperglycinemia: Quantitative analysis and relation to phenotype. <i>Journal of Inherited Metabolic Disease</i> , <b>2019</b> , 42, 438-450	5.4	13
37	Developing a conceptual, reproducible, rubric-based approach to consent and result disclosure for genetic testing by clinicians with minimal genetics background. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 727-735	8.1	26
36	Genetic Testing: Consent and Result Disclosure for Primary Care Providers. <i>Medical Clinics of North America</i> , <b>2019</b> , 103, 967-976	7	3
35	Comment on Late-Onset Nonketotic Hyperglycinemia With a Heterozygous Novel Point Mutation of the GLDC Gene. <i>Pediatric Neurology</i> , <b>2018</b> , 79, e1	2.9	
34	The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy: a common epileptic encephalopathy. <i>Journal of Inherited Metabolic Disease</i> , <b>2018</b> ,	5.4	4
33	Conducting an investigator-initiated randomized double-blinded intervention trial in acute decompensation of inborn errors of metabolism: Lessons from the N-Carbamylglutamate Consortium. <i>Translational Science of Rare Diseases</i> , <b>2018</b> , 3, 157-170	3.3	5
32	The genetic basis of classic nonketotic hyperglycinemia due to mutations in GLDC and AMT. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 104-111	8.1	38
31	Mild orotic aciduria in UMPS heterozygotes: a metabolic finding without clinical consequences. <i>Journal of Inherited Metabolic Disease</i> , <b>2017</b> , 40, 423-431	5.4	10
30	X-Linked Cobalamin Disorder (HCFC1) Mimicking Nonketotic Hyperglycinemia With Increased Both Cerebrospinal Fluid Glycine and Methylmalonic Acid. <i>Pediatric Neurology</i> , <b>2017</b> , 71, 65-69	2.9	9
29	Lethal neonatal hyperammonemia in severe ornithine transcarbamylase (OTC) deficiency compounded by large hepatic portosystemic shunt. <i>Journal of Inherited Metabolic Disease</i> , <b>2017</b> , 40, 159-160	5.4	2
28	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. <i>Human Molecular Genetics</i> , <b>2017</b> , 26, 702-716	5.6	51
27	Pyridoxine-Dependent Epilepsy: An Expanding Clinical Spectrum. <i>Pediatric Neurology</i> , <b>2016</b> , 59, 6-12	2.9	110
26	Neurodevelopmental Outcome and Treatment Efficacy of Benzoate and Dextromethorphan in Siblings with Attenuated Nonketotic Hyperglycinemia. <i>Journal of Pediatrics</i> , <b>2016</b> , 170, 234-9	3.6	39
25	Nonketotic Hyperglycinemia (Glycine Encephalopathy) and Lipoate Deficiency Disorders <b>2016</b> , 349-356		2
24	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. <i>Journal of Physical Education and Sports Management</i> , <b>2016</b> , 2, a000844	2.8	11
23	Low bone mineral density is a common finding in patients with homocystinuria. <i>Molecular Genetics and Metabolism</i> , <b>2016</b> , 117, 351-4	3.7	17

22	Mutations in the mitochondrial cysteinyl-tRNA synthase gene, CARS2, lead to a severe epileptic encephalopathy and complex movement disorder. <i>Journal of Medical Genetics</i> , <b>2015</b> , 52, 532-40	5.8	48
21	Human recombinant arginase enzyme reduces plasma arginine in mouse models of arginase deficiency. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 6417-27	5.6	28
20	Biochemical and molecular predictors for prognosis in nonketotic hyperglycinemia. <i>Annals of Neurology</i> , <b>2015</b> , 78, 606-18	9.4	47
19	Clinical and biochemical characterization of four patients with mutations in ECHS1. <i>Orphanet Journal of Rare Diseases</i> , <b>2015</b> , 10, 79	4.2	51
18	Triple therapy with pyridoxine, arginine supplementation and dietary lysine restriction in pyridoxine-dependent epilepsy: Neurodevelopmental outcome. <i>Molecular Genetics and Metabolism</i> , <b>2015</b> , 116, 35-43	3.7	75
17	Mitochondrial energy failure in HSD10 disease is due to defective mtDNA transcript processing. <i>Mitochondrion</i> , <b>2015</b> , 21, 1-10	4.9	40
16	Laboratory Evaluations in Inherited Metabolic Diseases <b>2015</b> , 75-85		
15	Glutaric Acidemia Type I: Diagnosis and Management <b>2015</b> , 203-209		
14	Lysine-Restricted Diet as Adjunct Therapy for Pyridoxine-Dependent Epilepsy: The PDE Consortium Consensus Recommendations. <i>JIMD Reports</i> , <b>2014</b> , 15, 1-11	1.9	32
13	Variant non ketotic hyperglycinemia is caused by mutations in LIAS, BOLA3 and the novel gene GLRX5. <i>Brain</i> , <b>2014</b> , 137, 366-79	11.2	146
12	Distinctive pattern of restricted diffusion in a neonate with molybdenum cofactor deficiency. <i>Pediatric Radiology</i> , <b>2013</b> , 43, 882-5	2.8	13
11	An X-linked cobalamin disorder caused by mutations in transcriptional coregulator HCFC1. <i>American Journal of Human Genetics</i> , <b>2013</b> , 93, 506-14	11	90
10	Dihydropteridine reductase deficiency and treatment with tetrahydrobiopterin: a case report. <i>JIMD Reports</i> , <b>2013</b> , 10, 53-6	1.9	13
9	Lysine restricted diet for pyridoxine-dependent epilepsy: first evidence and future trials. <i>Molecular Genetics and Metabolism</i> , <b>2012</b> , 107, 335-44	3.7	77
8	Clinical impact of copy number variation analysis using high-resolution microarray technologies: advantages, limitations and concerns. <i>Genome Medicine</i> , <b>2012</b> , 4, 80	14.4	44
7	Genotype-phenotype correlations: sudden death in an infant with very-long-chain acyl-CoA dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , <b>2010</b> , 33 Suppl 3, S129-31	5.4	20
6	Somatic mosaicism for PDHA1 mutation in a male with pyruvate dehydrogenase complex deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2010</b> , 100, 296-9	3.7	12
5	Very long-chain acyl-CoA dehydrogenase deficiency in a patient with normal newborn screening by tandem mass spectrometry. <i>Journal of Pediatrics</i> , <b>2010</b> , 156, 492-4	3.6	33

4	Evidence for a recurrent microdeletion at chromosome 16p11.2 associated with congenital anomalies of the kidney and urinary tract (CAKUT) and Hirschsprung disease. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 2618-22	2.5	38
3	Child neurology: a case illustrating the role of imaging in evaluation of sudden infant death. <i>Neurology</i> , <b>2009</b> , 73, e54-6	6.5	
2	Penicillamine therapy for pediatric cystinuria: experience from a cohort of American children. <i>Journal of Urology</i> , <b>2008</b> , 180, 2620-3	2.5	34
1	The 22q11 low copy repeats are characterized by unprecedented size and structure variability		5