

Michael T Geraghty

List of Publications by Year in descending order

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Version: 2024-02-01

38
papers

1,165
citations

471509

17
h-index

395702

33
g-index

38
all docs

38
docs citations

38
times ranked

2751
citing authors

#	ARTICLE	IF	CITATIONS
1	Deleterious variants in <i>CRLS1</i> lead to cardiolipin deficiency and cause an autosomal recessive multi-system mitochondrial disease. <i>Human Molecular Genetics</i> , 2022, 31, 3597-3612.	2.9	11
2	Enantiomer-specific pharmacokinetics of D,L-3-hydroxybutyrate: Implications for the treatment of multiple acyl-CoA dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 926-938.	3.6	10
3	A recurrent de novo ATP5F1A substitution associated with neonatal complex V deficiency. <i>European Journal of Human Genetics</i> , 2021, 29, 1719-1724.	2.8	2
4	Efficacy and safety of D,L-3-hydroxybutyrate (D,L-3-HB) treatment in multiple acyl-CoA dehydrogenase deficiency. <i>Genetics in Medicine</i> , 2020, 22, 908-916.	2.4	19
5	Carnitine uptake defect due to a 5'UTR mutation in a pedigree with false positives and false negatives on Newborn screening. <i>Molecular Genetics and Metabolism</i> , 2020, 129, 213-218.	1.1	6
6	Direct Health Care Costs, Health Services Utilization, and Outcomes of Biliary Atresia. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2020, 70, 436-443.	1.8	5
7	Blindness Caused by a Junk Food Diet. <i>Annals of Internal Medicine</i> , 2020, 172, 575.	3.9	2
8	Channelopathies Are a Frequent Cause of Genetic Ataxias Associated with Cerebellar Atrophy. <i>Movement Disorders Clinical Practice</i> , 2020, 7, 940-949.	1.5	7
9	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <i>PIGQ</i> : Report of seven new subjects and review of the literature. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1321-1332.	3.6	15
10	Outcomes in pediatric studies of medium-chain acyl-coA dehydrogenase (MCAD) deficiency and phenylketonuria (PKU): a review. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 12.	2.7	15
11	Health Care for Mitochondrial Disorders in Canada: A Survey of Physicians. <i>Canadian Journal of Neurological Sciences</i> , 2019, 46, 717-726.	0.5	6
12	Back Cover, Volume 40, Issue 8. <i>Human Mutation</i> , 2019, 40, ii-ii.	2.5	0
13	Molecular characterization of a large group of Mucopolysaccharidosis type IIIc patients reveals the evolutionary history of the disease. <i>Human Mutation</i> , 2019, 40, 1084-1100.	2.5	17
14	Clinical, biochemical, and genetic features of four patients with short-chain enoyl-CoA hydratase (ECHS1) deficiency. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1115-1127.	1.2	36
15	Disturbed phospholipid metabolism in serine biosynthesis defects revealed by metabolomic profiling. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 309-316.	1.1	38
16	Diagnostic clarity of exome sequencing following negative comprehensive panel testing in the neonatal intensive care unit. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1688-1691.	1.2	28
17	Severe Neutropenia and Anemia in a Child With Epilepsy and Copper Deficiency on a Ketogenic Diet. <i>Pediatric Neurology</i> , 2017, 76, 93-94.	2.1	8
18	Glutaric Aciduria Type 3: Three Unrelated Canadian Cases, with Different Routes of Ascertainment. <i>JIMD Reports</i> , 2017, 39, 89-96.	1.5	8

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19	Spinocerebellar ataxia type 29 due to mutations in ITPR1: a case series and review of this emerging congenital ataxia. Orphanet Journal of Rare Diseases, 2017, 12, 121.	2.7	42
20	Fatty liver in a non-obese patient. Paediatrics and Child Health, 2017, 22, 59-60.	0.6	1
21	Genotype-phenotype characterization in 13 individuals with chromosome Xp11.22 duplications. American Journal of Medical Genetics, Part A, 2016, 170, 967-977.	1.2	11
22	Unravelling 5-oxoprolinuria (pyroglutamic aciduria) due to bi-allelic OPLAH mutations: 20 new mutations in 14 families. Molecular Genetics and Metabolism, 2016, 119, 44-49.	1.1	9
23	Concordance between whole-exome sequencing and clinical Sanger sequencing: implications for patient care. Molecular Genetics & Genomic Medicine, 2016, 4, 504-512.	1.2	30
24	Severe Neonatal Presentation of Mitochondrial Citrate Carrier (SLC25A1) Deficiency. JIMD Reports, 2016, 30, 73-79.	1.5	21
25	Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. Cmaj, 2016, 188, E254-E260.	2.0	86
26	MG-107...Congenital sucrose-isomaltase deficiency: Identification of the common inuit founder mutation. Journal of Medical Genetics, 2015, 52, A1.3-A2.	3.2	0
27	Very late-onset Sandhoff disease presenting as Kennedy Disease. Muscle and Nerve, 2015, 52, 1135-1136.	2.2	4
28	Congenital sucrose-isomaltase deficiency: identification of a common Inuit founder mutation. Cmaj, 2015, 187, 102-107.	2.0	31
29	The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists. Journal of Medical Genetics, 2015, 52, 431-437.	3.2	187
30	Congenital Visual Impairment and Progressive Microcephaly Due to Lysyl-Transfer Ribonucleic Acid (RNA) Synthetase (<i>KARS</i>) Mutations. Journal of Child Neurology, 2015, 30, 1037-1043.	1.4	47
31	Homozygous mutation in the eukaryotic translation initiation factor 2alpha phosphatase gene, <i>PPP1R15B</i> , is associated with severe microcephaly, short stature and intellectual disability. Human Molecular Genetics, 2015, 24, 6293-6300.	2.9	36
32	Biotinidase deficiency: Spectrum of molecular, enzymatic and clinical information from newborn screening Ontario, Canada (2007-2014). Molecular Genetics and Metabolism, 2015, 116, 146-151.	1.1	22
33	Mutations in TRNT1 cause congenital sideroblastic anemia with immunodeficiency, fevers, and developmental delay (SIFD). Blood, 2014, 124, 2867-2871.	1.4	162
34	Discrepant DNA analysis in three patients with inherited arrhythmia: Molecular genetic test results deserve a second glance. American Journal of Medical Genetics, Part A, 2008, 146A, 1466-1469.	1.2	2
35	Characterization of the human gene encoding L-lysine aminoadipate aminotransferase (AADAT). Molecular Genetics and Metabolism, 2002, 76, 172-180.	1.1	34
36	Identification of the L-Amino adipic Semialdehyde Dehydrogenase-Phosphopantetheinyl Transferase Gene, the Human Ortholog of the Yeast LYS5 Gene. Molecular Genetics and Metabolism, 2001, 72, 336-342.	1.1	33

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37	Identification of the ϵ -Aminoadipic Semialdehyde Synthase Gene, Which Is Defective in Familial Hyperlysinemia. <i>American Journal of Human Genetics</i> , 2000, 66, 1736-1743.	6.2	99
38	Molecular Characterization of <i>Saccharomyces cerevisiae</i> ϵ -3, ϵ -2-Enoyl-CoA Isomerase. <i>Journal of Biological Chemistry</i> , 1998, 273, 33184-33191.	3.4	75