Michael T Geraghty

List of Publications by Year
in descending order

38	1,165	471509 17	395702 33
papers	citations	h-index	g-index

38	38	38	2751
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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
2	Mutations in TRNT1 cause congenital sideroblastic anemia with immunodeficiency, fevers, and developmental delay (SIFD). Blood, 2014, 124, 2867-2871.	1.4	162
3	Identification of the Î±-Aminoadipic Semialdehyde Synthase Gene, Which Is Defective in Familial Hyperlysinemia. American Journal of Human Genetics, 2000, 66, 1736-1743.	6.2	99
4	Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. Cmaj, 2016, 188, E254-E260.	2.0	86
5	Molecular Characterization of Saccharomyces cerevisiae Î”3,Î”2-Enoyl-CoA Isomerase. Journal of Biological Chemistry, 1998, 273, 33184-33191.	3.4	75
6	Congenital Visual Impairment and Progressive Microcephaly Due to Lysylâ€“Transfer Ribonucleic Acid (RNA) Synthetase (KARS) Mutations. Journal of Child Neurology, 2015, 30, 1037-1043.	1.4	47
7	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
8	Disturbed phospholipid metabolism in serine biosynthesis defects revealed by metabolomic profiling. Molecular Genetics and Metabolism, 2018, 123, 309-316.	1.1	38
9	Homozygous mutation in the eukaryotic translation initiation factor 2alpha phosphatase gene,PPP1R15B, is associated with severe microcephaly, short stature and intellectual disability. Human Molecular Genetics, 2015, 24, 6293-6300.	2.9	36
10	Clinical, biochemical, and genetic features of four patients with shortâ€chain enoylâ€CoA hydratase (ECHS1) deficiency. American Journal of Medical Genetics, Part A, 2018, 176, 1115-1127.	1.2	36
11	Characterization of the human gene encoding Î±-aminoadipate aminotransferase (AADAT). Molecular Genetics and Metabolism, 2002, 76, 172-180.	1.1	34
12	Identification of the Î±-Aminoadipic Semialdehyde Dehydrogenase-Phosphopantetheinyl Transferase Gene, the Human Ortholog of the Yeast LYS5 Gene. Molecular Genetics and Metabolism, 2001, 72, 336-342.	1.1	33
13	Congenital sucraseâ€“isomaltase deficiency: identification of a common Inuit founder mutation. Cmaj, 2015, 187, 102-107.	2.0	31
14	Concordance between wholeâ€exome sequencing and clinical Sanger sequencing: implications for patient care. Molecular Genetics & Genomic Medicine, 2016, 4, 504-512.	1.2	30
15	Diagnostic clarity of exome sequencing following negative comprehensive panel testing in the neonatal intensive care unit. American Journal of Medical Genetics, Part A, 2018, 176, 1688-1691.	1.2	28
16	Biotinidase deficiency: Spectrum of molecular, enzymatic and clinical information from newborn screening Ontario, Canada (2007â€“2014). Molecular Genetics and Metabolism, 2015, 116, 146-151.	1.1	22
17	Severe Neonatal Presentation of Mitochondrial Citrate Carrier (SLC25A1) Deficiency. JIMD Reports, 2016, 30, 73-79.	1.5	21
18	Efficacy and safety of D,L-3-hydroxybutyrate (D,L-3-HB) treatment in multiple acyl-CoA dehydrogenase deficiency. Genetics in Medicine, 2020, 22, 908-916.	2.4	19

#	Article	IF	Citations
19	Molecular characterization of a large group of Mucopolysaccharidosis type IIIC patients reveals the evolutionary history of the disease. Human Mutation, 2019, 40, 1084-1100.	2.5	17
20	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <scp>PIGQ</scp>: Report of seven new subjects and review of the literature. Journal of Inherited Metabolic Disease, 2020, 43, 1321-1332.	3.6	15
21	Outcomes in pediatric studies of medium-chain acyl-coA dehydrogenase (MCAD) deficiency and phenylketonuria (PKU): a review. Orphanet Journal of Rare Diseases, 2020, 15, 12.	2.7	15
22	Genotypeâ€“phenotype characterization in 13 individuals with chromosome Xp11.22 duplications. American Journal of Medical Genetics, Part A, 2016, 170, 967-977.	1.2	11
23	Deleterious variants in CRLS1 lead to cardiolipin deficiency and cause an autosomal recessive multi-system mitochondrial disease. Human Molecular Genetics, 2022, 31, 3597-3612.	2.9	11
24	Enantiomerâ€specific pharmacokinetics of D,Lâ€3â€hydroxybutyrate: Implications for the treatment of multiple <scp>acylâ€CoA</scp> dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 926-938.	3.6	10
25	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
26	Severe Neutropenia and Anemia in a Child With Epilepsy and Copper Deficiency on a Ketogenic Diet. Pediatric Neurology, 2017, 76, 93-94.	2.1	8
27	Glutaric Aciduria Type 3: Three Unrelated Canadian Cases, with Different Routes of Ascertainment. JIMD Reports, 2017, 39, 89-96.	1.5	8
28	Channelopathies Are a Frequent Cause of Genetic Ataxias Associated with Cerebellar Atrophy. Movement Disorders Clinical Practice, 2020, 7, 940-949.	1.5	7
29	Health Care for Mitochondrial Disorders in Canada: A Survey of Physicians. Canadian Journal of Neurological Sciences, 2019, 46, 717-726.	0.5	6
30	Carnitine uptake defect due to a 5â€²UTR mutation in a pedigree with false positives and false negatives on Newborn screening. Molecular Genetics and Metabolism, 2020, 129, 213-218.	1.1	6
31	Direct Health Care Costs, Health Services Utilization, and Outcomes of Biliary Atresia. Journal of Pediatric Gastroenterology and Nutrition, 2020, 70, 436-443.	1.8	5
32	Very lateâ€onset Sandhoff disease presenting as Kennedy Disease. Muscle and Nerve, 2015, 52, 1135-1136.	2.2	4
33	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
34	Blindness Caused by a Junk Food Diet. Annals of Internal Medicine, 2020, 172, 575.	3.9	2
35	A recurrent de novo ATP5F1A substitution associated with neonatal complex V deficiency. European Journal of Human Genetics, 2021, 29, 1719-1724.	2.8	2
36	Fatty liver in a non-obese patient. Paediatrics and Child Health, 2017, 22, 59-60.	0.6	1

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37	MG-107â€...Congenital sucrase-isomaltase deficiency: Identification of the common inuit founder mutation. Journal of Medical Genetics, 2015, 52, A1.3-A2.	3.2	0
38	Back Cover, Volume 40, Issue 8. Human Mutation, 2019, 40, ii-ii.	2.5	0