Graham Sinclair, Fccmg

List of Publications by Year
in descending order

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Version: 2024-02-01

52 papers	1,964 citations	430874 18 h-index	254184 43 g-index
52 all docs	52 docs citations	52 times ranked	3731 citing authors

#	Article	IF	Citations
1	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. Genetics in Medicine, 2011, 13, 230-254.	2.4	308
2	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	27.0	254
3	Synonymous codon usage bias and the expression of human glucocerebrosidase in the methylotrophic yeast, Pichia pastoris. Protein Expression and Purification, 2002, 26, 96-105.	1.3	140
4	Enhanced interpretation of newborn screening results without analyte cutoff values. Genetics in Medicine, 2012, 14, 648-655.	2.4	117
5	Loss-of-Function and Gain-of-Function Mutations in KCNQ5 Cause Intellectual Disability or Epileptic Encephalopathy. American Journal of Human Genetics, 2017, 101, 65-74.	6.2	99
6	Lysine restricted diet for pyridoxine-dependent epilepsy: First evidence and future trials. Molecular Genetics and Metabolism, 2012, 107, 335-344.	1.1	97
7	Mitochondrial Carbonic Anhydrase VA Deficiency Resulting from CA5A Alterations Presents with Hyperammonemia in Early Childhood. American Journal of Human Genetics, 2014, 94, 453-461.	6.2	82
8	The paradox of the carnitine palmitoyltransferase type Ia P479L variant in Canadian Aboriginal populations. Molecular Genetics and Metabolism, 2009, 96, 201-207.	1.1	72
9	The genotypic and phenotypic spectrum of PIGA deficiency. Orphanet Journal of Rare Diseases, 2015, 10, 23.	2.7	70
10	Treatment of intractable epilepsy in a female with SLC6A8 deficiency. Molecular Genetics and Metabolism, 2010, 101, 409-412.	1.1	66
11	De Novo Mutations in YWHAG Cause Early-Onset Epilepsy. American Journal of Human Genetics, 2017, 101, 300-310.	6.2	65
12	Carnitine palmitoyltransferase 1A (CPT1A) P479L prevalence in live newborns in Yukon, Northwest Territories, and Nunavut. Molecular Genetics and Metabolism, 2010, 101, 200-204.	1.1	62
13	Heparin cofactor IIâ€“thrombin complex: A biomarker of MPS disease. Molecular Genetics and Metabolism, 2008, 94, 456-461.	1.1	54
14	Atypical cerebral palsy: genomics analysis enables precision medicine. Genetics in Medicine, 2019, 21, 1621-1628.	2.4	47
15	Heparin cofactor IIâ€“thrombin complex in MPS I: A biomarker of MPS disease. Molecular Genetics and Metabolism, 2006, 88, 235-243.	1.1	41
16	Generation of a conditional knockout of murine glucocerebrosidase: Utility for the study of Gaucher disease. Molecular Genetics and Metabolism, 2007, 90, 148-156.	1.1	35
17	Infantile cardioencephalopathy due to a COX15 gene defect: Report and review. , 2011, 155, 840-844.		29
18	A Novel Complex Allele and Two New Point Mutations in Type 2 (Acute Neuronopathic) Gaucher Disease. Blood Cells, Molecules, and Diseases, 1998, 24, 420-427.	1.4	22

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19	Defects in fatty acid amide hydrolase 2 in a male with neurologic and psychiatric symptoms. Orphanet Journal of Rare Diseases, 2015, 10, 38.	2.7	19
20	A three-tier algorithm for guanidinoacetate methyltransferase (GAMT) deficiency newborn screening. Molecular Genetics and Metabolism, 2016, 118, 173-177.	1.1	19
21	Carnitine Palmitoyltransferase I and Sudden Unexpected Infant Death in British Columbia First Nations. Pediatrics, 2012, 130, e1162-e1169.	2.1	18
22	Evaluation of two year treatment outcome and limited impact of arginine restriction in a patient with GAMT deficiency. Molecular Genetics and Metabolism, 2012, 105, 155-158.	1.1	18
23	Clinical Impact and Cost Efficacy of Newborn Screening for Congenital Adrenal Hyperplasia. Journal of Pediatrics, 2020, 220, 101-108.e2.	1.8	15
24	Detection of a novel intragenic rearrangement in the creatine transporter gene by next generation sequencing. Molecular Genetics and Metabolism, 2013, 110, 465-471.	1.1	14
25	A girl with developmental delay, ataxia, cranial nerve palsies, severe respiratory problems in infancyâ€"Expanding NDST1 syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 712-715.	1.2	14
26	Pregnant women of South Asian ethnicity in Canada have substantially lower vitamin B₁₂status compared with pregnant women of European ethnicity. British Journal of Nutrition, 2017, 118, 454-462.	2.3	14
27	Secretion of human glucocerebrosidase from stable transformed insect cells using native signal sequences. Biochemistry and Cell Biology, 2006, 84, 148-156.	2.0	12
28	Fish odour syndrome. Cmaj, 2011, 183, 929-931.	2.0	12
29	Performance of serum and dried blood spot acylcarnitine profiles for detection of fatty acid Î²â€oxidation disorders in adult patients with rhabdomyolysis. Journal of Inherited Metabolic Disease, 2014, 37, 207-213.	3.6	11
30	Health economic evaluation of plasma oxysterol screening in the diagnosis of Niemannâ€"Pick Type C disease among intellectually disabled using discrete event simulation. Molecular Genetics and Metabolism, 2015, 114, 226-232.	1.1	11
31	Optic atrophy, cataracts, lipodystrophy/lipoatrophy, and peripheral neuropathy caused by a de novo OPA3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a001156.	1.2	11
32	Protective effects of d-3-hydroxybutyrate and propionate during hypoglycemic coma: Clinical and biochemical insights from infant rats. Molecular Genetics and Metabolism, 2011, 103, 179-184.	1.1	10
33	Acylcarnitine profile in thyroid disease. Clinical Biochemistry, 2013, 46, 180-183.	1.9	10
34	The p.P479L variant in CPT1A is associated with infectious disease in a BC First Nation. Paediatrics and Child Health, 2019, 24, e111-e115.	0.6	10
35	Reference interval of methylmalonic acid concentrations in dried blood spots of healthy, term newborns to facilitate neonatal screening of vitamin B12 deficiency. Clinical Biochemistry, 2016, 49, 973-978.	1.9	8
36	Acute Shoshin beriberi syndrome immediately postâ€"kidney transplant with rapid recovery after thiamine administration. Pediatric Transplantation, 2019, 23, e13493.	1.0	8

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37	Performance of Immunoglobulin G Serology on Finger Prick Capillary Dried Blood Spot Samples to Detect a SARS-CoV-2 Antibody Response. Microbiology Spectrum, 2022, 10, e0140521.	3.0	8
38	Pyruvate Carboxylase Deficiency Type C: A Rare Cause of Acute Transient Flaccid Paralysis with Ketoacidosis. Neuropediatrics, 2018, 49, 369-372.	0.6	7
39	Reference intervals for serum total vitamin B12 and holotranscobalamin concentrations and their change points with methylmalonic acid concentration to assess vitamin B12 status during early and mid-pregnancy. Clinical Chemistry and Laboratory Medicine, 2019, 57, 1790-1798.	2.3	7
40	Analysis of 2-methylcitric acid, methylmalonic acid, and total homocysteine in dried blood spots by LC-MS/MS for application in the newborn screening laboratory: A dual derivatization approach. Journal of Mass Spectrometry and Advances in the Clinical Lab, 2021, 20, 1-10.	2.4	7
41	Maternal vitamin B₁₂ status in early pregnancy and its association with birth outcomes in Canadian motherâ€“newborn Dyads. British Journal of Nutrition, 2021, 126, 1823-1831.	2.3	6
42	Interference of ketone bodies on laboratory creatinine measurement in children with DKA: a call for change in testing practices. Pediatric Nephrology, 2022, 37, 1347-1353.	1.7	6
43	Heterologous Expression and Characterization of a Rare Gaucher Disease Mutation (c.481C > T) from a Canadian Aboriginal Population Using Archival Tissue Samples. Molecular Genetics and Metabolism, 2001, 74, 345-352.	1.1	5
44	Performance of a Three-Tier (IRT-DNA-IRT) Cystic Fibrosis Screening Algorithm in British Columbia. International Journal of Neonatal Screening, 2020, 6, 46.	3.2	5
45	Integrated Multianalyte Second-Tier Testing for Newborn Screening for MSUD, IVA, and GAMT Deficiencies. FIRE Forum for International Research in Education, 2016, 4, 232640981666629.	0.7	4
46	Serum Betaine and Dimethylglycine Are Higher in South Asian Compared with European Pregnant Women in Canada, with Betaine and Total Homocysteine Inversely Associated in Early and Midpregnancy, Independent of Ethnicity. Journal of Nutrition, 2019, 149, 2145-2155.	2.9	4
47	Diagnostic yield from routine metabolic screening tests in evaluation of global developmental delay and intellectual disability. Paediatrics and Child Health, 2021, 26, 344-348.	0.6	4
48	A Visit with Dr. Louis Woolf, Recognizing His 100th Birthday and His Contributions to the Diagnosis and Treatment of Phenylketonuria. International Journal of Neonatal Screening, 2020, 6, 45.	3.2	3
49	Glycine cleavage enzyme complex: Molecular cloning and expression of the H-protein cDNA from cultured human skin fibroblasts. Biochemistry and Cell Biology, 2011, 89, 299-307.	2.0	2
50	Investigating oxythiamine levels in children undergoing kidney transplantation and the risk of immediate post-operative metabolic and hemodynamic decompensation. Pediatric Nephrology, 2021, 36, 987-993.	1.7	2
51	Vitamin B-12 Biomarkers Reviewed: Holotranscobalamin Allows Identification of Vitamin B12 Deficiency in Children with Short-Bowel Syndrome. Current Developments in Nutrition, 2020, 4, nzaa067_066.	0.3	0
52	A simple method modification to increase separation of 2- and 3-hydroxyglutaric acid by GCâ€“MS for clinical urine organic acids analysis. Clinical Biochemistry, 2022, , .	1.9	0