Hilary D Vallance

List of Publications by Year in Descending Order

57 papers	1,153 citations	17 h-index	32 g-index
62 ext. papers	1,365 ext. citations	5.3 avg, IF	3.46 L-index

#	Paper	IF	Citations
57	Diagnostic yield from routine metabolic screening tests in evaluation of global developmental delay and intellectual disability. *Paediatrics and Child Health*, **2021**, 26, 344-348	0.7	0
56	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		0
55	Maternal vitamin B status in early pregnancy and its association with birth outcomes in Canadian mother-newborn Dyads. *British Journal of Nutrition*, **2021**, 126, 1823-1831	3.6	2
54	Performance of a Three-Tier (IRT-DNA-IRT) Cystic Fibrosis Screening Algorithm in British Columbia. *International Journal of Neonatal Screening*, **2020**, 6, 46	2.6	1
53	Clinical Impact and Cost Efficacy of Newborn Screening for Congenital Adrenal Hyperplasia. *Journal of Pediatrics*, **2020**, 220, 101-108.e2	3.6	6
52	Evaluation of the quality of clinical data collection for a pan-Canadian cohort of children affected by inherited metabolic diseases: lessons learned from the Canadian Inherited Metabolic Diseases Research Network. *Orphanet Journal of Rare Diseases*, **2020**, 15, 89	4.2	4
51	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	4.1	1
50	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	5.9	5
49	The p.P479L variant in CPT1A is associated with infectious disease in a BC First Nation. *Paediatrics and Child Health*, **2019**, 24, e111-e115	0.7	6
48	Health services use among children diagnosed with medium-chain acyl-CoA dehydrogenase deficiency through newborn screening: a cohort study in Ontario, Canada. *Orphanet Journal of Rare Diseases*, **2019**, 14, 70	4.2	3
47	A mitochondrial DNA D loop insertion detected almost exclusively in non-replicating tissues with maternal inheritance across three generations. *Mitochondrion*, **2019**, 46, 298-301	4.9	1
46	Atypical cerebral palsy: genomics analysis enables precision medicine. *Genetics in Medicine*, **2019**, 21, 1621-1628	8.1	28
45	Sialic acid catabolism by N-acetylneuraminate pyruvate lyase is essential for muscle function. *JCI Insight*, **2018**, 3,	9.9	25
44	Optic atrophy, cataracts, lipodystrophy/lipoatrophy, and peripheral neuropathy caused by a de novo mutation. *Journal of Physical Education and Sports Management*, **2017**, 3, a001156	2.8	10
43	Pregnant women of South Asian ethnicity in Canada have substantially lower vitamin B12 status compared with pregnant women of European ethnicity. *British Journal of Nutrition*, **2017**, 118, 454-462	3.6	11
42	A three-tier algorithm for guanidinoacetate methyltransferase (GAMT) deficiency newborn screening. *Molecular Genetics and Metabolism*, **2016**, 118, 173-177	3.7	13
41	Loss-of-function mutations in SCN4A cause severe foetal hypokinesia or 'classical' congenital myopathy. *Brain*, **2016**, 139, 674-91	11.2	76

40	Integrated Multianalyte Second-Tier Testing for Newborn Screening for MSUD, IVA, and GAMT Deficiencies. *FIRE Forum for International Research in Education*, **2016**, 4, 232640981666629	1.4	0
39	Exome Sequencing and the Management of Neurometabolic Disorders. *New England Journal of Medicine*, **2016**, 374, 2246-55	59.2	197
38	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, 973-8	2.5	7
37	Unravelling 5-oxoprolinuria (pyroglutamic aciduria) due to bi-allelic OPLAH mutations: 20 new mutations in 14 families. *Molecular Genetics and Metabolism*, **2016**, 119, 44-9	3.7	6
36	RMND1 deficiency associated with neonatal lactic acidosis, infantile onset renal failure, deafness, and multiorgan involvement. *European Journal of Human Genetics*, **2015**, 23, 1301-7	5.3	20
35	Mitochondrial carbonic anhydrase VA deficiency resulting from CA5A alterations presents with hyperammonemia in early childhood. *American Journal of Human Genetics*, **2014**, 94, 453-61	11	66
34	Performance of serum and dried blood spot acylcarnitine profiles for detection of fatty acid -oxidation disorders in adult patients with rhabdomyolysis. *Journal of Inherited Metabolic Disease*, **2014**, 37, 207-13	5.4	9
33	Limitation of TREC-based newborn screening for ZAP70 Severe Combined Immunodeficiency. *Clinical Immunology*, **2014**, 153, 209-10	9	30
32	Prediction of obstetrical risk using maternal serum markers and clinical risk factors. *Prenatal Diagnosis*, **2014**, 34, 172-9	3.2	17
31	Single point mutation in Rabenosyn-5 in a female with intractable seizures and evidence of defective endocytotic trafficking. *Orphanet Journal of Rare Diseases*, **2014**, 9, 141	4.2	20
30	Three years experience with dried blood spot -glucosidase screening for Pompe disease in British Columbia, Canada. *BMC Musculoskeletal Disorders*, **2013**, 14,	2.8	1
29	Long-term outcomes of blood phenylalanine concentrations in children with classical phenylketonuria. *Molecular Genetics and Metabolism*, **2013**, 108, 255-8	3.7	8
28	A variant of unknown significance in the GLA gene causing diagnostic uncertainty in a young female with isolated hypertrophic cardiomyopathy. *Gene*, **2012**, 497, 320-2	3.8	4
27	Carnitine palmitoyltransferase I and sudden unexpected infant death in British Columbia First Nations. *Pediatrics*, **2012**, 130, e1162-9	7.4	17
26	Levator palpebrae biopsy and diagnosis of progressive external ophthalmoplegia. *Canadian Journal of Neurological Sciences*, **2012**, 39, 520-4	1	2
25	Late-onset nonketotic hyperglycinemia caused by a novel homozygous missense mutation in the GLDC gene. *Molecular Genetics and Metabolism*, **2011**, 103, 193-6	3.7	16
24	Infantile cardioencephalopathy due to a COX15 gene defect: report and review. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 840-4	2.5	25
23	IDH2 mutations in patients with D-2-hydroxyglutaric aciduria. *Science*, **2010**, 330, 336	33.3	152

22	Carnitine palmitoyltransferase 1A (CPT1A) P479L prevalence in live newborns in Yukon, Northwest Territories, and Nunavut. *Molecular Genetics and Metabolism*, **2010**, 101, 200-4	3.7	55
21	The paradox of the carnitine palmitoyltransferase type Ia P479L variant in Canadian Aboriginal populations. *Molecular Genetics and Metabolism*, **2009**, 96, 201-7	3.7	56
20	Screening for Fabry disease in patients with chronic kidney disease: limitations of plasma alpha-galactosidase assay as a screening test. *Clinical Journal of the American Society of Nephrology: CJASN*, **2008**, 3, 139-45	6.9	40
19	Newborn screening for MCAD deficiency: experience of the first three years in British Columbia, Canada. *Canadian Journal of Public Health*, **2008**, 99, 276-80	3.2	17
18	Mutation-based diagnostic testing for primary hyperoxaluria type 1: survey of results. *Clinical Biochemistry*, **2008**, 41, 598-602	3.5	10
17	Delays in diagnosing cystic fibrosis: can we find ways to diagnose it earlier?. *Canadian Family Physician*, **2008**, 54, 877-83	0.9	10
16	Newborn screening by tandem mass spectrometry: ethical and social issues. *Canadian Journal of Public Health*, **2007**, 98, 284-6	3.2	9
15	Common HEXB polymorphisms reduce serum HexA and HexB enzymatic activities, potentially masking Tay-Sachs disease carrier identification. *Molecular Genetics and Metabolism*, **2006**, 87, 122-7	3.7	6
14	A hemizygous SCO2 mutation in an early onset rapidly progressive, fatal cardiomyopathy. *Molecular Genetics and Metabolism*, **2006**, 89, 129-33	3.7	24
13	Neonatal hyperphenylalaninemia, perinatal hemochromatosis, and renal tubulopathy: a unique patient or a novel metabolic disorder?. *Molecular Genetics and Metabolism*, **2005**, 86 Suppl 1, S148-52	3.7	3
12	6-pyruvoyl-tetrahydropterin synthase deficiency with mild hyperphenylalaninemia. *Annals of Neurology*, **2005**, 58, 164-7	9.4	10
11	The Brugada ECG pattern in a neonate. *Journal of Cardiovascular Electrophysiology*, **2005**, 16, 342-4	2.7	12
10	Laboratory Testing of Infants and Children **2005**, 1729-1739		
9	Biochemical approach to the investigation of pediatric mitochondrial disease. *Pediatric and Developmental Pathology*, **2004**, 7, 633-6	2.2	4
8	A genomic approach to mutation analysis of holocarboxylase synthetase gene in three Chinese patients with late-onset holocarboxylase synthetase deficiency. *Clinical Biochemistry*, **2003**, 36, 145-9	3.5	12
7	Carrier testing for autosomal-recessive disorders. *Critical Reviews in Clinical Laboratory Sciences*, **2003**, 40, 473-97	9.4	37
6	Mucopolysaccharidosis type VII (Sly syndrome) presenting as neonatal cholestasis with hepatosplenomegaly. *Journal of Pediatric Gastroenterology and Nutrition*, **2001**, 33, 216-20	2.8	20
5	Identification of 6 new mutations in the iduronate sulfatase gene. Mutation in brief no. 233. Online. *Human Mutation*, **1999**, 13, 338	4.7	4

4	DNA-based diagnosis of arylsulfatase A deficiencies as a supplement to enzyme assay: a case in point. *Clinical Biochemistry*, **1997**, 30, 57-61	3.5	5
3	Are patients with homocystinuria being missed?. *European Journal of Pediatrics*, **1995**, 154, 589	4.1	3
2	An improved method for quantification of very long chain fatty acids in plasma. *Clinical Biochemistry*, **1994**, 27, 183-6	3.5	20
1	Oral zinc therapy in the treatment of alpha-mannosidosis. *American Journal of Medical Genetics Part A*, **1993**, 46, 410-4		7