

Hilary D Vallance

List of Publications by Citations

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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

1,153
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	Exome Sequencing and the Management of Neurometabolic Disorders. <i>New England Journal of Medicine</i> , 2016 , 374, 2246-55	59.2	197
56	IDH2 mutations in patients with D-2-hydroxyglutaric aciduria. <i>Science</i> , 2010 , 330, 336	33.3	152
55	Loss-of-function mutations in SCN4A cause severe foetal hypokinesia or classical congenital myopathy. <i>Brain</i> , 2016 , 139, 674-91	11.2	76
54	Mitochondrial carbonic anhydrase VA deficiency resulting from CA5A alterations presents with hyperammonemia in early childhood. <i>American Journal of Human Genetics</i> , 2014 , 94, 453-61	11	66
53	The paradox of the carnitine palmitoyltransferase type Ia P479L variant in Canadian Aboriginal populations. <i>Molecular Genetics and Metabolism</i> , 2009 , 96, 201-7	3.7	56
52	Carnitine palmitoyltransferase 1A (CPT1A) P479L prevalence in live newborns in Yukon, Northwest Territories, and Nunavut. <i>Molecular Genetics and Metabolism</i> , 2010 , 101, 200-4	3.7	55
51	Screening for Fabry disease in patients with chronic kidney disease: limitations of plasma alpha-galactosidase assay as a screening test. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2008 , 3, 139-45	6.9	40
50	Carrier testing for autosomal-recessive disorders. <i>Critical Reviews in Clinical Laboratory Sciences</i> , 2003 , 40, 473-97	9.4	37
49	Limitation of TREC-based newborn screening for ZAP70 Severe Combined Immunodeficiency. <i>Clinical Immunology</i> , 2014 , 153, 209-10	9	30
48	Atypical cerebral palsy: genomics analysis enables precision medicine. <i>Genetics in Medicine</i> , 2019 , 21, 1621-1628	8.1	28
47	Infantile cardioencephalopathy due to a COX15 gene defect: report and review. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 840-4	2.5	25
46	Sialic acid catabolism by N-acetylneuraminase pyruvate lyase is essential for muscle function. <i>JCI Insight</i> , 2018 , 3,	9.9	25
45	A hemizygous SCO2 mutation in an early onset rapidly progressive, fatal cardiomyopathy. <i>Molecular Genetics and Metabolism</i> , 2006 , 89, 129-33	3.7	24
44	RMND1 deficiency associated with neonatal lactic acidosis, infantile onset renal failure, deafness, and multiorgan involvement. <i>European Journal of Human Genetics</i> , 2015 , 23, 1301-7	5.3	20
43	Single point mutation in Rabenosyn-5 in a female with intractable seizures and evidence of defective endocytotic trafficking. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 141	4.2	20
42	Mucopolysaccharidosis type VII (Sly syndrome) presenting as neonatal cholestasis with hepatosplenomegaly. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2001 , 33, 216-20	2.8	20
41	An improved method for quantification of very long chain fatty acids in plasma. <i>Clinical Biochemistry</i> , 1994 , 27, 183-6	3.5	20

40	Prediction of obstetrical risk using maternal serum markers and clinical risk factors. <i>Prenatal Diagnosis</i> , 2014 , 34, 172-9	3.2	17
39	Carnitine palmitoyltransferase I and sudden unexpected infant death in British Columbia First Nations. <i>Pediatrics</i> , 2012 , 130, e1162-9	7.4	17
38	Newborn screening for MCAD deficiency: experience of the first three years in British Columbia, Canada. <i>Canadian Journal of Public Health</i> , 2008 , 99, 276-80	3.2	17
37	Late-onset nonketotic hyperglycinemia caused by a novel homozygous missense mutation in the GLDC gene. <i>Molecular Genetics and Metabolism</i> , 2011 , 103, 193-6	3.7	16
36	A three-tier algorithm for guanidinoacetate methyltransferase (GAMT) deficiency newborn screening. <i>Molecular Genetics and Metabolism</i> , 2016 , 118, 173-177	3.7	13
35	A genomic approach to mutation analysis of holocarboxylase synthetase gene in three Chinese patients with late-onset holocarboxylase synthetase deficiency. <i>Clinical Biochemistry</i> , 2003 , 36, 145-9	3.5	12
34	The Brugada ECG pattern in a neonate. <i>Journal of Cardiovascular Electrophysiology</i> , 2005 , 16, 342-4	2.7	12
33	Pregnant women of South Asian ethnicity in Canada have substantially lower vitamin B12 status compared with pregnant women of European ethnicity. <i>British Journal of Nutrition</i> , 2017 , 118, 454-462	3.6	11
32	Optic atrophy, cataracts, lipodystrophy/lipoatrophy, and peripheral neuropathy caused by a de novo mutation. <i>Journal of Physical Education and Sports Management</i> , 2017 , 3, a001156	2.8	10
31	Mutation-based diagnostic testing for primary hyperoxaluria type 1: survey of results. <i>Clinical Biochemistry</i> , 2008 , 41, 598-602	3.5	10
30	6-pyruvoyl-tetrahydropterin synthase deficiency with mild hyperphenylalaninemia. <i>Annals of Neurology</i> , 2005 , 58, 164-7	9.4	10
29	Delays in diagnosing cystic fibrosis: can we find ways to diagnose it earlier?. <i>Canadian Family Physician</i> , 2008 , 54, 877-83	0.9	10
28	Performance of serum and dried blood spot acylcarnitine profiles for detection of fatty acid oxidation disorders in adult patients with rhabdomyolysis. <i>Journal of Inherited Metabolic Disease</i> , 2014 , 37, 207-13	5.4	9
27	Newborn screening by tandem mass spectrometry: ethical and social issues. <i>Canadian Journal of Public Health</i> , 2007 , 98, 284-6	3.2	9
26	Long-term outcomes of blood phenylalanine concentrations in children with classical phenylketonuria. <i>Molecular Genetics and Metabolism</i> , 2013 , 108, 255-8	3.7	8
25	Oral zinc therapy in the treatment of alpha-mannosidosis. <i>American Journal of Medical Genetics Part A</i> , 1993 , 46, 410-4		7
24	Reference interval of methylmalonic acid concentrations in dried blood spots of healthy, term newborns to facilitate neonatal screening of vitamin B12 deficiency. <i>Clinical Biochemistry</i> , 2016 , 49, 973-8	3.5	7
23	The p.P479L variant in CPT1A is associated with infectious disease in a BC First Nation. <i>Paediatrics and Child Health</i> , 2019 , 24, e111-e115	0.7	6

22	Clinical Impact and Cost Efficacy of Newborn Screening for Congenital Adrenal Hyperplasia. <i>Journal of Pediatrics</i> , 2020 , 220, 101-108.e2	3.6	6
21	Common HEXB polymorphisms reduce serum HexA and HexB enzymatic activities, potentially masking Tay-Sachs disease carrier identification. <i>Molecular Genetics and Metabolism</i> , 2006 , 87, 122-7	3.7	6
20	Unravelling 5-oxoprolinuria (pyroglutamic aciduria) due to bi-allelic OPLAH mutations: 20 new mutations in 14 Families. <i>Molecular Genetics and Metabolism</i> , 2016 , 119, 44-9	3.7	6
19	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. <i>Clinical Chemistry and Laboratory Medicine</i> , 2019 , 57, 1790-1798	5.9	5
18	DNA-based diagnosis of arylsulfatase A deficiencies as a supplement to enzyme assay: a case in point. <i>Clinical Biochemistry</i> , 1997 , 30, 57-61	3.5	5
17	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. <i>Orphanet Journal of Rare Diseases</i> , 2020 , 15, 89	4.2	4
16	A variant of unknown significance in the GLA gene causing diagnostic uncertainty in a young female with isolated hypertrophic cardiomyopathy. <i>Gene</i> , 2012 , 497, 320-2	3.8	4
15	Biochemical approach to the investigation of pediatric mitochondrial disease. <i>Pediatric and Developmental Pathology</i> , 2004 , 7, 633-6	2.2	4
14	Identification of 6 new mutations in the iduronate sulfatase gene. Mutation in brief no. 233. Online. <i>Human Mutation</i> , 1999 , 13, 338	4.7	4
13	Health services use among children diagnosed with medium-chain acyl-CoA dehydrogenase deficiency through newborn screening: a cohort study in Ontario, Canada. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 70	4.2	3
12	Neonatal hyperphenylalaninemia, perinatal hemochromatosis, and renal tubulopathy: a unique patient or a novel metabolic disorder?. <i>Molecular Genetics and Metabolism</i> , 2005 , 86 Suppl 1, S148-52	3.7	3
11	Are patients with homocystinuria being missed?. <i>European Journal of Pediatrics</i> , 1995 , 154, 589	4.1	3
10	Levator palpebrae biopsy and diagnosis of progressive external ophthalmoplegia. <i>Canadian Journal of Neurological Sciences</i> , 2012 , 39, 520-4	1	2
9	Maternal vitamin B status in early pregnancy and its association with birth outcomes in Canadian mother-newborn Dyads. <i>British Journal of Nutrition</i> , 2021 , 126, 1823-1831	3.6	2
8	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. <i>Journal of Nutrition</i> , 2019 , 149, 2145-2155	4.1	1
7	Performance of a Three-Tier (IRT-DNA-IRT) Cystic Fibrosis Screening Algorithm in British Columbia. <i>International Journal of Neonatal Screening</i> , 2020 , 6, 46	2.6	1
6	A mitochondrial DNA D loop insertion detected almost exclusively in non-replicating tissues with maternal inheritance across three generations. <i>Mitochondrion</i> , 2019 , 46, 298-301	4.9	1
5	Three years experience with dried blood spot α -glucosidase screening for Pompe disease in British Columbia, Canada. <i>BMC Musculoskeletal Disorders</i> , 2013 , 14,	2.8	1

4	Diagnostic yield from routine metabolic screening tests in evaluation of global developmental delay and intellectual disability. <i>Paediatrics and Child Health</i> , 2021 , 26, 344-348	0.7	○
3	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. <i>Journal of Mass Spectrometry and Advances in the Clinical Lab</i> , 2021 , 20, 1-10		○
2	Integrated Multianalyte Second-Tier Testing for Newborn Screening for MSUD, IVA, and GAMT Deficiencies. <i>FIRE Forum for International Research in Education</i> , 2016 , 4, 232640981666629	1.4	○
1	Laboratory Testing of Infants and Children 2005 , 1729-1739		