Laura Arbour

List of Publications by Citations

99 papers	2,369 citations	25 h-index	46 g-index
105 ext. papers	2,808 ext. citations	5.6 avg, IF	4.47 L-index

#	Paper	IF	Citations
99	Oligonucleotide microarray analysis of genomic imbalance in children with mental retardation. *American Journal of Human Genetics*, **2006**, 79, 500-13	11	247
98	Genetic polymorphisms in methylenetetrahydrofolate reductase and methionine synthase, folate levels in red blood cells, and risk of neural tube defects. *American Journal of Medical Genetics Part A* , **1999**, 84, 151-7		210
97	Mutations in the gene encoding capillary morphogenesis protein 2 cause juvenile hyaline fibromatosis and infantile systemic hyalinosis. *American Journal of Human Genetics*, **2003**, 73, 791-800	11	180
96	Evidence for a familial WilmsStumour gene (FWT1) on chromosome 17q12-q21. *Nature Genetics*, **1996**, 13, 461-3	36.3	145
95	Race and ancestry in biomedical research: exploring the challenges. *Genome Medicine*, **2009**, 1, 8	14.4	81
94	Association Between Preeclampsia and Congenital Heart Defects. *JAMA - Journal of the American Medical Association*, **2015**, 314, 1588-98	27.4	80
93	DNA on loan: issues to consider when carrying out genetic research with aboriginal families and communities. *Public Health Genomics*, **2006**, 9, 153-60	1.9	63
92	The gene for juvenile hyaline fibromatosis maps to chromosome 4q21. *American Journal of Human Genetics*, **2002**, 71, 975-80	11	62
91	Rights, interests and expectations: Indigenous perspectives on unrestricted access to genomic data. *Nature Reviews Genetics*, **2020**, 21, 377-384	30.1	60
90	Genomic Research Through an Indigenous Lens: Understanding the Expectations. *Annual Review of Genomics and Human Genetics*, **2019**, 20, 495-517	9.7	59
89	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
88	Changes in frequencies of select congenital anomalies since the onset of folic acid fortification in a Canadian birth defect registry. *Canadian Journal of Public Health*, **2008**, 99, 271-5	3.2	49
87	Hypomorphic temperature-sensitive alleles of NSDHL cause CK syndrome. *American Journal of Human Genetics*, **2010**, 87, 905-14	11	46
86	An autosomal dominant form of familial persistent hyperinsulinemic hypoglycemia of infancy, not linked to the sulfonylurea receptor locus. *Journal of Clinical Endocrinology and Metabolism*, **1997**, 82, 1192-4	5.6	46
85	Genetic Testing in the Evaluation of Unexplained Cardiac Arrest: From the CASPER (Cardiac Arrest Survivors With Preserved Ejection Fraction Registry). *Circulation: Cardiovascular Genetics*, **2017**, 10,		43
84	Confirmation of FWT1 as a WilmsStumour susceptibility gene and phenotypic characteristics of WilmsStumour attributable to FWT1. *Human Genetics*, **1998**, 103, 547-56	6.3	42
83	The shared pathoetiological effects of particulate air pollution and the social environment on fetal-placental development. *Journal of Environmental and Public Health*, **2014**, 2014, 901017	2.6	38

82	Maternal risk factors for gastroschisis in Canada. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2015**, 103, 111-8		37
81	Heavy smoking during pregnancy as a marker for other risk factors of adverse birth outcomes: a population-based study in British Columbia, Canada. *BMC Public Health*, **2012**, 12, 102	4.1	36
80	Glutathione deficiency as a complication of methylmalonic acidemia: response to high doses of ascorbate. *Journal of Pediatrics*, **1996**, 129, 445-8	3.6	35
79	Prenatally detected trisomy 20 mosaicism. *Prenatal Diagnosis*, **2005**, 25, 239-44	3.2	32
78	Human exposure to environmental contaminants and congenital anomalies: a critical review. *Critical Reviews in Toxicology*, **2017**, 47, 59-84	5.7	27
77	Exposure to anesthetic gases and congenital anomalies in offspring of female registered nurses. *American Journal of Industrial Medicine*, **2011**, 54, 118-27	2.7	27
76	Detection of pathogenic copy number variants in children with idiopathic intellectual disability using 500 K SNP array genomic hybridization. *BMC Genomics*, **2009**, 10, 526	4.5	27
75	Characteristics of primary biliary cirrhosis in British Columbia's First Nations population. *Canadian Journal of Gastroenterology & Hepatology*, **2005**, 19, 305-10		26
74	The reduction of birth weight by fine particulate matter and its modification by maternal and neighbourhood-level factors: a multilevel analysis in British Columbia, Canada. *Environmental Health*, **2016**, 15, 51	6	25
73	Causes and risk factors for infant mortality in Nunavut, Canada 1999-2011. *BMC Pediatrics*, **2012**, 12, 190	2.6	25
72	A KCNQ1 V205M missense mutation causes a high rate of long QT syndrome in a First Nations community of northern British Columbia: a community-based approach to understanding the impact. *Genetics in Medicine*, **2008**, 10, 545-50	8.1	24
71	Prenatal diagnosis of apparently isolated unilateral multicystic kidney: implications for counselling and management. *Prenatal Diagnosis*, **2002**, 22, 388-94	3.2	23
70	Variable onset of metachromatic leukodystrophy in a Vietnamese family. *Pediatric Neurology*, **2000**, 23, 173-6	2.9	23
69	Characterization of a new X-linked mental retardation syndrome with microcephaly, cortical malformation, and thin habitus. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2469-78	2.5	21
68	Submicroscopic deletions of 11q24-25 in individuals without Jacobsen syndrome: re-examination of the critical region by high-resolution array-CGH. *Molecular Cytogenetics*, **2008**, 1, 23	2	21
67	Discordant measures of androgen-binding kinetics in two mutant androgen receptors causing mild or partial androgen insensitivity, respectively. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 805-10	5.6	21
66	Severe Maternal Morbidity in Canada: Temporal Trends and Regional Variations, 2003-2016. *Journal of Obstetrics and Gynaecology Canada*, **2019**, 41, 1589-1598.e16	1.3	18
65	Carnitine palmitoyltransferase I and sudden unexpected infant death in British Columbia First Nations. *Pediatrics*, **2012**, 130, e1162-9	7.4	17

64	Congenital microcephaly in Quebec: baseline prevalence, risk factors and outcomes in a large cohort of neonates. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **2018**, 103, F167-F172	4.7	15
63	The familial WilmsStumour susceptibility gene, FWT1, may not be a tumour suppressor gene. *Oncogene*, **1997**, 14, 3099-102	9.2	15
62	Penetrance of mutations in the familial Wilms tumor gene FWT1. *Journal of the National Cancer Institute*, **2000**, 92, 650-2	9.7	15
61	Indigenous Genomic Databases: Pragmatic Considerations and Cultural Contexts. *Frontiers in Public Health*, **2020**, 8, 111	6	14
60	LQTS in Northern BC: homozygosity for KCNQ1 V205M presents with a more severe cardiac phenotype but with minimal impact on auditory function. *Clinical Genetics*, **2014**, 86, 85-90	4	14
59	The mystery of primary biliary cirrhosis in British Columbia's First Nations people. *International Journal of Circumpolar Health*, **2004**, 63 Suppl 2, 185-8	1.7	14
58	Recurrent trisomy 15 in a female carrier of der(15)t(Y;15)(q12;p13). *American Journal of Medical Genetics Part A*, **2001**, 99, 320-4		14
57	Severe maternal morbidity surveillance: Monitoring pregnant women at high risk for prolonged hospitalisation and death. *Paediatric and Perinatal Epidemiology*, **2020**, 34, 427-439	2.7	14
56	Hereditary angioedema managed with low-dose danazol and C1 esterase inhibitor concentrate: a case report. *Journal of Obstetrics and Gynaecology Canada*, **2006**, 28, 27-31	1.3	13
55	Spatial variability of gastroschisis in Canada, 2006-2011: An exploratory analysis. *Canadian Journal of Public Health*, **2016**, 107, e62-e67	3.2	13
54	Maternal and community predictors of gastroschisis and congenital diaphragmatic hernia in Canada. *Pediatric Surgery International*, **2015**, 31, 1055-60	2.1	12
53	Autoimmune liver disease and the Canadian First Nations Aboriginal Communities of British Columbia's Pacific Northwest. *World Journal of Gastroenterology*, **2006**, 12, 3625-7	5.6	12
52	KCNQ1 p.L353L affects splicing and modifies the phenotype in a founder population with long QT syndrome type 1. *Journal of Medical Genetics*, **2017**, 54, 390-398	5.8	11
51	Primary Biliary Cholangitis in British Columbia First Nations: Clinical features and discovery of novel genetic susceptibility loci. *Liver International*, **2018**, 38, 940-948	7.9	11
50	Heart defects and other malformations in the Inuit in Canada: a baseline study. *International Journal of Circumpolar Health*, **2004**, 63, 251-66	1.7	11
49	Spina bifida, folate metabolism, and dietary folate intake in a Northern Canadian aboriginal population. *International Journal of Circumpolar Health*, **2002**, 61, 341-51	1.7	11
48	Patient Recall, Interpretation, and Perspective of an Inconclusive Long QT Syndrome Genetic Test Result. *Journal of Genetic Counseling*, **2017**, 26, 150-158	2.5	10
47	Exome sequencing identified a de novo mutation of PURA gene in a patient with familial Xp22.31 microduplication. *European Journal of Medical Genetics*, **2019**, 62, 103-108	2.6	10

46	Elevated ambient temperatures and risk of neural tube defects. *Occupational and Environmental Medicine*, **2017**, 74, 315-320	2.1	9
45	Bariatric surgery and the risk of congenital anomalies in subsequent pregnancies. *American Journal of Clinical Nutrition*, **2019**, 110, 1168-1174	7	9
44	Air pollution, neighbourhood and maternal-level factors modify the effect of smoking on birth weight: a multilevel analysis in British Columbia, Canada. *BMC Public Health*, **2016**, 16, 585	4.1	9
43	Canadian Alliance for Healthy Hearts and Minds: First Nations Cohort Study Rationale and Design. *Progress in Community Health Partnerships: Research, Education, and Action*, **2018**, 12, 55-64	1.2	9
42	Recognizing life-threatening causes of syncope. *Cardiology Clinics*, **2013**, 31, 51-66	2.5	8
41	Explaining the variability in cardiovascular risk factors among First Nations communities in Canada: a population-based study. *Lancet Planetary Health, The*, **2019**, 3, e511-e520	9.8	8
40	Rates of congenital anomalies and other adverse birth outcomes in an offspring cohort of registered nurses from British Columbia, Canada. *Canadian Journal of Public Health*, **2010**, 101, 230-4	3.2	7
39	Recurrent trisomy 21: four cases in three generations. *Clinical Genetics*, **2005**, 68, 430-5	4	7
38	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
37	Barriers and Considerations for Diagnosing Rare Diseases in Indigenous Populations. *Frontiers in Pediatrics*, **2020**, 8, 579924	3.4	6
36	Inherited heart rhythm disease: negotiating the minefield for the practicing cardiologist. *Canadian Journal of Cardiology*, **2013**, 29, 122-5	3.8	6
35	Beckwith-Wiedemann syndrome in sibs discordant for IC2 methylation. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1662-9	2.5	6
34	Red blood cell folate levels in Canadian Inuit women of childbearing years: influence of food security, body mass index, smoking, education, and vitamin use. *Canadian Journal of Public Health*, **2018**, 109, 684-691	3.2	5
33	Pre-eclampsia and risk of infantile haemangioma. *British Journal of Dermatology*, **2017**, 176, 371-377	4	5
32	Genetic Research and Culture: Where Does the Offense Lie?115-139		5
31	Postoperative dystonia in a female patient with homocystinuria. *Journal of Pediatrics*, **1988**, 113, 863-4	3.6	5
30	Maternal proximity to extremely low frequency electromagnetic fields and risk of birth defects. *European Journal of Epidemiology*, **2019**, 34, 689-697	12.1	4
29	The Genetics of Cardiovascular Disease in Canadian and International Aboriginal Populations. *Canadian Journal of Cardiology*, **2015**, 31, 1094-115	3.8	4

28	The Canadian Arrhythmogenic Right Ventricular Cardiomyopathy Registry: Rationale, Design, and Preliminary Recruitment. *Canadian Journal of Cardiology*, **2016**, 32, 1396-1401	3.8	4
27	Life-history chronicle for a patient with the recently described chromosome 4q21 microdeletion syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2606-9	2.5	4
26	The development of a comprehensive maternal-child health information system for Nunavut-Nutaqqavut (Our Children). *International Journal of Circumpolar Health*, **2011**, 70, 363-72	1.7	4
25	Congenital Anomalies in the Offspring of Nurses: Association with Area of Employment During Pregnancy. *International Journal of Occupational and Environmental Health*, **2011**, 17, 195-201		4
24	Pregnancy outcomes of women with spina bifida. *Disability and Rehabilitation*, **2019**, 41, 1403-1409	2.4	4
23	Cost Analysis of Patients Referred for Inherited Heart Rhythm Disorder Evaluation. *Canadian Journal of Cardiology*, **2017**, 33, 814-821	3.8	3
22	The Hearts in Rhythm Organization: A Canadian National Cardiogenetics Network. *CJC Open*, **2020**, 2, 652-662	2	3
21	Risk of central nervous system defects in offspring of women with and without mental illness. *Archives of Women's Mental Health*, **2018**, 21, 437-444	5	3
20	Early Repolarization Pattern Inheritance in the Cardiac Arrest Survivors With Preserved Ejection Fraction Registry (CASPER). *JACC: Clinical Electrophysiology*, **2018**, 4, 1473-1479	4.6	3
19	Cardiac arrest in a mother and daughter and the identification of a novel RYR2 variant, predisposing to low penetrant catecholaminergic polymorphic ventricular tachycardia in a four-generation Canadian family. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1151	2.3	2
18	The current state of birth outcome and birth defect surveillance in northern regions of the world. *International Journal of Circumpolar Health*, **2009**, 68, 443-58	1.7	2
17	Stillbirth in Canada: anachronistic definition and registration processes impede public health surveillance and clinical care. *Canadian Journal of Public Health*, **2021**, 112, 766-772	3.2	2
16	Variant Reinterpretation in Survivors of Cardiac Arrest With Preserved Ejection Fraction (the Cardiac Arrest Survivors With Preserved Ejection Fraction Registry) by Clinicians and Clinical Commercial Laboratories. *Circulation Genomic and Precision Medicine*, **2021**, 14, e003235	5.2	2
15	Future risk of cancer in women who have children with birth defects. *Annals of Epidemiology*, **2019**, 37, 57-63.e3	6.4	2
14	Prepregnancy asthma and the subsequent risk of central nervous system defects in offspring. *Birth Defects Research*, **2019**, 111, 254-260	2.9	2
13	Understanding the personal and community impact of long QT syndrome: A perspective from Gitxsan women. *Journal of Genetic Counseling*, **2020**, 29, 562-573	2.5	1
12	Congenital anomalies in the offspring of nurses: association with area of employment during pregnancy. *International Journal of Occupational and Environmental Health*, **2011**, 17, 195-201		1
11	Inflammatory Bowel Disease and Risk of Birth Defects in Offspring. *Journal of Crohn's and Colitis*, **2020**, 14, 588-594	1.5	1

10	Neonatal hypoglycemia and the CPT1A P479L variant in term newborns: A retrospective cohort study of Inuit newborns from Kivalliq Nunavut. *Paediatrics and Child Health*, **2021**, 26, 218-227	0.7	1
9	Coeliac disease and risk of birth defects in pregnancy. *Gut*, **2021**, 70, 1198-1199	19.2	1
8	Time trends, geographic variation and risk factors for gastroschisis in Canada: A population-based cohort study 2006-2017. *Paediatric and Perinatal Epidemiology*, **2021**, 35, 664-673	2.7	1
7	Implementation of the BC Congenital Anomalies Surveillance System (BCCASS).. *Canadian Journal of Public Health*, **2022**, 1	3.2	0
6	Risk of Offspring Birth Defects in Women After Bariatric Surgery. *JAMA - Journal of the American Medical Association*, **2020**, 323, 668	27.4	0
5	Association of the p.P479L Metabolic Gene Variant With Childhood Respiratory and Other Infectious Illness in Nunavut. *Frontiers in Pediatrics*, **2021**, 9, 678553	3.4	0
4	Rare disorders have many faces: in silico characterization of rare disorder spectrum.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 76	4.2	0
3	Association of Birth Defects With Child Mortality Before Age 14 Years.. *JAMA Network Open*, **2022**, 5, e226739	10.4	0
2	Preeclampsia and Congenital Heart Defects--Reply. *JAMA - Journal of the American Medical Association*, **2016**, 315, 1168-9	27.4	
1	Post-partum Primary Biliary Cholangitis Preceded by Intrahepatic Cholestasis of Pregnancy in Three First Nation Patients. *Digestive Diseases and Sciences*, **2021**, 66, 1367-1369	4	