Laura Arbour

List of Publications by Year
in descending order

105 papers	3,301 citations	172207 29 h-index	189595 50 g-index
105 all docs	105 docs citations	105 times ranked	4283 citing authors

#	Article	IF	Citations
1	Oligonucleotide Microarray Analysis of Genomic Imbalance in Children with Mental Retardation. American Journal of Human Genetics, 2006, 79, 500-513.	2.6	261
2	Genetic polymorphisms in methylenetetrahydrofolate reductase and methionine synthase, folate levels in red blood cells, and risk of neural tube defects. , 1999, 84, 151-157.		252
3	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.	2.6	209
4	Evidence for a familial Wilms' tumour gene (FWT1) on chromosome 17q12â€“q21. Nature Genetics, 1996, 13, 461-463.	9.4	166
5	Rights, interests and expectations: Indigenous perspectives on unrestricted access to genomic data. Nature Reviews Genetics, 2020, 21, 377-384.	7.7	141
6	Genomic Research Through an Indigenous Lens: Understanding the Expectations. Annual Review of Genomics and Human Genetics, 2019, 20, 495-517.	2.5	129
7	Association Between Preeclampsia and Congenital Heart Defects. JAMA - Journal of the American Medical Association, 2015, 314, 1588.	3.8	127
8	Race and ancestry in biomedical research: exploring the challenges. Genome Medicine, 2009, 1, 8.	3.6	106
9	DNA on Loan: Issues to Consider when Carrying Out Genetic Research with Aboriginal Families and Communities. Public Health Genomics, 2006, 9, 153-160.	0.6	90
10	Genetic Testing in the Evaluation of Unexplained Cardiac Arrest. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	76
11	The Gene for Juvenile Hyaline Fibromatosis Maps to Chromosome 4q21. American Journal of Human Genetics, 2002, 71, 975-980.	2.6	71
12	Hypomorphic Temperature-Sensitive Alleles of NSDHL Cause CK Syndrome. American Journal of Human Genetics, 2010, 87, 905-914.	2.6	64
13	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, 271-275.	1.1	62
14	Carnitine palmitoyltransferase 1A (CPT1A) P479L prevalence in live newborns in Yukon, Northwest Territories, and Nunavut. Molecular Genetics and Metabolism, 2010, 101, 200-204.	0.5	62
15	The Shared Pathoetiological Effects of Particulate Air Pollution and the Social Environment on Fetal-Placental Development. Journal of Environmental and Public Health, 2014, 2014, 1-20.	0.4	56
16	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, 1192-1194.	1.8	51
17	Maternal risk factors for gastroschisis in Canada. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 111-118.	1.6	51
18	Confirmation of FWT1 as a Wilmsâ€™ tumour susceptibility gene and phenotypic characteristics of Wilmsâ€™ tumour attributable to FWT1. Human Genetics, 1998, 103, 547-556.	1.8	48

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19	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.	1.2	47
20	Severe maternal morbidity surveillance: Monitoring pregnant women at high risk for prolonged hospitalisation and death. Paediatric and Perinatal Epidemiology, 2020, 34, 427-439.	0.8	44
21	Glutathione deficiency as a complication of methylmalonic acidemia: Response to high doses of ascorbate. Journal of Pediatrics, 1996, 129, 445-448.	0.9	40
22	Prenatally detected trisomy 20 mosaicism. Prenatal Diagnosis, 2005, 25, 239-244.	1.1	38
23	Severe Maternal Morbidity in Canada: Temporal Trends and Regional Variations, 2003-2016. Journal of Obstetrics and Gynaecology Canada, 2019, 41, 1589-1598.e16.	0.3	37
24	Indigenous Genomic Databases: Pragmatic Considerations and Cultural Contexts. Frontiers in Public Health, 2020, 8, 111.	1.3	37
25	Importance of genetic testing in unexplained cardiac arrest. European Heart Journal, 2022, 43, 3071-3081.	1.0	36
26	Characteristics of primary biliary cirrhosis in British Columbia's First Nations population. Canadian Journal of Gastroenterology & Hepatology, 2005, 19, 305-310.	1.8	35
27	Exposure to anesthetic gases and congenital anomalies in offspring of female registered nurses. American Journal of Industrial Medicine, 2011, 54, 118-127.	1.0	34
28	Prenatal diagnosis of apparently isolated unilateral multicystic kidney: implications for counselling and management. Prenatal Diagnosis, 2002, 22, 388-394.	1.1	33
29	Human exposure to environmental contaminants and congenital anomalies: a critical review. Critical Reviews in Toxicology, 2017, 47, 59-84.	1.9	32
30	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, 545-550.	1.1	31
31	Causes and risk factors for infant mortality in Nunavut, Canada 1999â€“2011. BMC Pediatrics, 2012, 12, 190.	0.7	31
32	Detection of pathogenic copy number variants in children with idiopathic intellectual disability using 500 K SNP array genomic hybridization. BMC Genomics, 2009, 10, 526.	1.2	30
33	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.	1.7	30
34	Characterization of a new Xâ€linked mental retardation syndrome with microcephaly, cortical malformation, and thin habitus. American Journal of Medical Genetics, Part A, 2009, 149A, 2469-2478.	0.7	27
35	Variable onset of metachromatic leukodystrophy in a Vietnamese family. Pediatric Neurology, 2000, 23, 173-176.	1.0	25
36	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.	0.4	25

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37	Barriers and Considerations for Diagnosing Rare Diseases in Indigenous Populations. Frontiers in Pediatrics, 2020, 8, 579924.	0.9	25
38	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.	5.1	23
39	Discordant Measures of Androgen-Binding Kinetics in Two Mutant Androgen Receptors Causing Mild or Partial Androgen Insensitivity, Respectively1. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 805-810.	1.8	22
40	Elevated ambient temperatures and risk of neural tube defects. Occupational and Environmental Medicine, 2017, 74, 315-320.	1.3	22
41	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.	1.4	22
42	Penetrance of Mutations in the Familial Wilms Tumor Gene FWT1. Journal of the National Cancer Institute, 2000, 92, 650-652.	3.0	19
43	The mystery of primary biliary cirrhosis in British Columbiaâ€™s First Nations people. International Journal of Circumpolar Health, 2004, 63, 185-188.	0.5	18
44	Carnitine Palmitoyltransferase I and Sudden Unexpected Infant Death in British Columbia First Nations. Pediatrics, 2012, 130, e1162-e1169.	1.0	18
45	Spatial variability of gastroschisis in Canada, 2006â€“2011: An exploratory analysis. Canadian Journal of Public Health, 2016, 107, e62-e67.	1.1	18
46	The familial Wilms' tumour susceptibility gene, FWT1, may not be a tumour suppressor gene. Oncogene, 1997, 14, 3099-3102.	2.6	17
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.	0.7	17
48	Recurrent trisomy 15 in a female carrier of der(15)t(Y;15)(q12;p13). American Journal of Medical Genetics Part A, 2001, 99, 320-324.	2.4	16
49	<scp>LQTS</scp> in Northern <scp>BC</scp>: homozygosity for <scp>KCNQ1</scp><scp>V205M</scp> presents with a more severe cardiac phenotype but with minimal impact on auditory function. Clinical Genetics, 2014, 86, 85-90.	1.0	16
50	Primary Biliary Cholangitis in British Columbia First Nations: Clinical features and discovery of novel genetic susceptibility loci. Liver International, 2018, 38, 940-948.	1.9	16
51	Autoimmune liver disease and the Canadian First Nations Aboriginal Communities of British Columbiaâ€™s Pacific Northwest. World Journal of Gastroenterology, 2006, 12, 3625.	1.4	16
52	Heart defects and other malformations in the Inuit in Canada: a baseline study. International Journal of Circumpolar Health, 2004, 63, 251-266.	0.5	15
53	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.	0.3	15
54	Maternal and community predictors of gastroschisis and congenital diaphragmatic hernia in Canada. Pediatric Surgery International, 2015, 31, 1055-1060.	0.6	15

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55	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.	1.5	15
56	Bariatric surgery and the risk of congenital anomalies in subsequent pregnancies. American Journal of Clinical Nutrition, 2019, 110, 1168-1174.	2.2	15
57	Spina bifida, folate metabolism, and dietary folate intake in a Northern Canadian aboriginal population. International Journal of Circumpolar Health, 2002, 61, 341-351.	0.5	14
58	The Hearts in Rhythm Organization: A Canadian National Cardiogenetics Network. CJC Open, 2020, 2, 652-662.	0.7	14
59	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, 230-234.	1.1	13
60	Patient Recall, Interpretation, and Perspective of an Inconclusive Long QT Syndrome Genetic Test Result. Journal of Genetic Counseling, 2017, 26, 150-158.	0.9	13
61	Pregnancy outcomes of women with spina bifida. Disability and Rehabilitation, 2019, 41, 1403-1409.	0.9	12
62	Inherited Heart Rhythm Disease: Negotiating the Minefield for the Practicing Cardiologist. Canadian Journal of Cardiology, 2013, 29, 122-125.	0.8	11
63	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.	1.2	11
64	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.	0.2	11
65	Time trends, geographic variation and risk factors for gastroschisis in Canada: A populationâ€based cohort study 2006â€“2017. Paediatric and Perinatal Epidemiology, 2021, 35, 664-673.	0.8	11
66	Recognizing Life-Threatening Causes of Syncope. Cardiology Clinics, 2013, 31, 51-66.	0.9	10
67	The p.P479L variant in CPT1A is associated with infectious disease in a BC First Nation. Paediatrics and Child Health, 2019, 24, e111-e115.	0.3	10
68	Variant Reinterpretation in Survivors of Cardiac Arrest With Preserved Ejection Fraction (the Cardiac) Tj ETQq0 0 0 rgBT /Overlock 10 Tf Laboratories. Circulation Genomic and Precision Medicine, 2021, 14, e003235.	1.6	10
69	The development of a comprehensive maternalâ€“child health information system for Nunavut-Nutaqqavut (Our Children). International Journal of Circumpolar Health, 2011, 70, 363-372.	0.5	9
70	The Genetics of Cardiovascular Disease in Canadian and International Aboriginal Populations. Canadian Journal of Cardiology, 2015, 31, 1094-1115.	0.8	9
71	Preâ€eclampsia and risk of infantile haemangioma. British Journal of Dermatology, 2017, 176, 371-377.	1.4	9
72	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.	1.1	9

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73	Recurrent trisomy 21: four cases in three generations. Clinical Genetics, 2005, 68, 430-435.	1.0	8
74	Genetic Research and Culture: Where Does the Offense Lie?. , 0, , 115-139.		8
75	Association of Birth Defects With Child Mortality Before Age 14 Years. JAMA Network Open, 2022, 5, e226739.	2.8	8
76	Stillbirth in Canada: anachronistic definition and registration processes impede public health surveillance and clinical care. Canadian Journal of Public Health, 2021, 112, 766-772.	1.1	7
77	Postoperative dystonia in a female patient with homocystinuria. Journal of Pediatrics, 1988, 113, 863-864.	0.9	6
78	Beckwithâ€“Wiedemann syndrome in sibs discordant for IC2 methylation. American Journal of Medical Genetics, Part A, 2012, 158A, 1662-1669.	0.7	6
79	The Canadian Arrhythmogenic Right Ventricular Cardiomyopathy Registry: Rationale, Design, and Preliminary Recruitment. Canadian Journal of Cardiology, 2016, 32, 1396-1401.	0.8	6
80	Early Repolarization Pattern Inheritance in the Cardiac Arrest Survivors With Preserved Ejection Fraction Registry (CASPER). JACC: Clinical Electrophysiology, 2018, 4, 1473-1479.	1.3	6
81	Maternal proximity to extremely low frequency electromagnetic fields and risk of birth defects. European Journal of Epidemiology, 2019, 34, 689-697.	2.5	6
82	Inflammatory Bowel Disease and Risk of Birth Defects in Offspring. Journal of Crohn's and Colitis, 2020, 14, 588-594.	0.6	6
83	Rare disorders have many faces: in silico characterization of rare disorder spectrum. Orphanet Journal of Rare Diseases, 2022, 17, 76.	1.2	6
84	Lifeâ€history chronicle for a patient with the recently described chromosome 4q21 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2606-2609.	0.7	5
85	The current state of birth outcome and birth defect surveillance in northern regions of the world. International Journal of Circumpolar Health, 2009, 68, 443-458.	0.5	4
86	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.	1.2	4
87	Future risk of cancer in women who have children with birth defects. Annals of Epidemiology, 2019, 37, 57-63.e3.	0.9	4
88	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.	0.3	4
89	Association of first trimester anaesthesia with risk of congenital heart defects in offspring. International Journal of Epidemiology, 2022, 51, 737-746.	0.9	4
90	Association of the CPT1A p.P479L Metabolic Gene Variant With Childhood Respiratory and Other Infectious Illness in Nunavut. Frontiers in Pediatrics, 2021, 9, 678553.	0.9	4

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91	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.	1.2	4
92	Cost Analysis of Patients Referred for Inherited Heart Rhythm Disorder Evaluation. Canadian Journal of Cardiology, 2017, 33, 814-821.	0.8	3
93	Prepregnancy asthma and the subsequent risk of central nervous system defects in offspring. Birth Defects Research, 2019, 111, 254-260.	0.8	3
94	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 fourâ€generation Canadian family. Molecular Genetics & Genomic Medicine, 2020, 8, e1151.	0.6	3
95	Coeliac disease and risk of birth defects in pregnancy. Gut, 2021, 70, 1198-1199.	6.1	3
96	Return of Results Policies for Genomic Research: Current Practices and the Hearts in Rhythm Organization (HiRO) Approach. Canadian Journal of Cardiology, 2022, 38, 526-535.	0.8	3
97	Cascade testing for inherited arrhythmia conditions: Experiences and attitudes of family communication approaches for a Canadian cohort. Journal of Genetic Counseling, 2022, 31, 815-828.	0.9	3
98	Implementation of the BC Congenital Anomalies Surveillance System (BCCASS). Canadian Journal of Public Health, 2022, , 1.	1.1	2
99	Understanding the personal and community impact of long QT syndrome: A perspective from Gitxsan women. Journal of Genetic Counseling, 2020, 29, 562-573.	0.9	1
100	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.	1.2	1
101	Risk of Offspring Birth Defects in Women After Bariatric Surgery. JAMA - Journal of the American Medical Association, 2020, 323, 668.	3.8	1
102	Post-partum Primary Biliary Cholangitis Preceded by Intrahepatic Cholestasis of Pregnancy in Three First Nation Patients. Digestive Diseases and Sciences, 2021, 66, 1367-1369.	1.1	1
103	Observational study of birth outcomes in children with inborn errors of metabolism. Pediatric Research, 2022, , .	1.1	1
104	Inherited heart rhythm disorders: Diagnostic dilemmas after the sudden death of a young family member. Journal of Nursing Education and Practice, 2013, 4, .	0.1	0
105	Preeclampsia and Congenital Heart Defectsâ€"Reply. JAMA - Journal of the American Medical Association, 2016, 315, 1168.	3.8	0