

Laura Arbour's Publications & Presentations

2000-2008

Peer-Reviewed Publications

1. **Arbour L**, Silver K, Hechtman P, Treacy E, Coulter-Mackie, M. Variable onset of metachromatic leukodystrophy in Vietnamese sibs with identical novel mutations in the ARSA gene. *Pediatric Neurology* 23(2):173-176, 2000.
2. Rahman L, **Arbour L**, Houlston R, Bonaiti-Pellie C, Abidi F, Tranchemontagne J, Ford D, Narod S, Foulkes W, Pritchard-Jones K, Schwartz C, Stratton M. Penetrance of mutations in the familial wilms tumour gene, FWT1. *JNCI* 92(8):650-652, 2000.
3. Yoshida EM, Caron NR, Buczkowski AK, **Arbour LT**, Scudamore CH, Steinbrecher UP, Erb SR, Chung SW. Indications for liver transplantation in British Columbia's Aboriginal population: a ten year retrospective analysis. *Canadian Journal of Gastroenterology* 14(9):775-779, 2000.
4. Rajcan-Separovic E, Robinson WP, Stephenson M, Pantzar T, **Arbour L**, McFadden D, Guscott J. Recurrent trisomy 15 in a female carrier of der(15)t(Y;15)(q12;p13). *Am J Med Genet* 99(4):320-324, 2001.
5. **Arbour L**, Christensen B, Delormier T, Platt R, Gilfix B, Forbes P, Kovitch I, Morel J, Rozen R. Neural tube defects, folate metabolism, and dietary folate intake in a northern Canadian Aboriginal population. *Int J Circum Health* 61:341-349, 2002.
6. Aubertin G, Coleman G, Shaw D, Van Allen M, **Arbour L**. Prenatal diagnosis and management of unilateral multicystic kidney. *Prenatal Diagnosis* 22:388-394, 2002 (Supervisor for project and corresponding author).
7. Rahman N, Dunstan M, Teare MD, Hanks S, Edkins, SJ, Hughes J, Bignell GR, Mancini G, Kleijer w, Campbell M, keser G, Black C, Williams N, **Arbour L**, Warman M, Superti-Furga A, Futreal PA, Pope FM. The gene for juvenile hyaline fibromatosis maps to chromosome 4q21. *Am J Hum Genet.* 71:975-980, 2002.
8. Hanks S, Adams S, Douglas J, **Arbour L**, Atherton DJ, Balci S, Bode H, Campbell ME, Feingold M, Gökhan K, Kleijer W, Mancini G, McGarth JA, Muntoni F, Nanda A, Teare MD, Warman M, Pope FM, Superti-Furga A, Futreal PA, Rahman N. Mutations in the gene encoding capillary morphogenesis protein 2 cause juvenile hyaline fibromatosis and infantile systemic hyalinosis. *Am J Hum Genet* 73:791-800, 2003.

9. Egeland GM, Berti P, Soueida R, **Arbour L**, Receveur O, Kuhnlein H. Vitamin A intake among Canadian Inuit. *Can J Public Health* 95(6):465-9, 2004.
10. **Arbour L**, Gilpin C, Millor-Roy V, Pekeles G, Egeland GM, Hodgins S, Eydoux P. Heart defects and other malformations in the Inuit of Canada: a baseline study. *Int J Circumpolar Health* 63(3):251-266, 2004.
11. **Arbour L**, Rupps R, Field L, Ross P, Erikson A, Henderson H, Hill W, Yoshida E. Characteristics of primary biliary cirrhosis in British Columbia's First Nations people. *Can J Gastroenterol* (5):305-310, 2005. (Winner of the Thompson Williams award for the most significant contribution to the CJG 2005)
12. Tyson C, Harvard C, Locker R, Friedman JM, Langlois S, Lewis ME, Van Allen M, Somerville M, **Arbour L**, Clarke L, McGillivray B, Yong SL, Siegel-Bartel J, Rajcan-Separovic E. Submicroscopic deletions and duplications in individuals with intellectual disability detected by array-CGH. *Am J Med Genet Part A* 139(3):173-85, 2005.
13. Robinson WP, McGillivray B, Lewis ME, **Arbour L**, Barrett I, Kalousek DK. Prenatally detected trisomy 20 mosaicism. *Prenatal Diagnosis* 25(3):239-244, 2005.
14. Gair JL, **Arbour L**, Rupps R, Jiang R, Bruyere H, Robinson WP. Recurrent trisomy 21: four cases in three generations. *Clin Genet* 68(5):430-435, 2005.
15. **Arbour L**, Cook D. DNA on loan: issues to consider when carrying out genetic research with Canadian Aboriginal populations. *Community Genet* 9(3):153-160, 2006. (invited manuscript)
16. Friedman JM, Baross A, Delaney AD, Ally A, **Arbour L**, Asano J, Bailey DK, Barber S, Birch P, Brown-John M, Cao M, Chan S, Charest DL, Farnoud N, Fernandes N, Flibotte S, Go A, Gibson WT, Holt RA, Jones SJM, Kennedy, Krzywinski M, Langlois S, Li HI, McGillivray BC, Nayar T, Pugh TJ, Rajcan-Separovic E, Schein JE, Schnerch A, Siddiqui A, Van Allen MI, Wilson G, Yong S, Zahir F, Eydoux P, and Marra MA. Oligonucleotide microarray analysis of genomic imbalance in children with mental retardation. *Am. J. Hum. Genet.* 79:500–513, 2006.
17. Yoshida EM, Riley M, **Arbour LT**. Autoimmune liver disease and the Canadian First Nations Aboriginal communities of British Columbia's pacific northwest. *World J Gastroenterol* 12(23):3625-3627, 2006.
18. Altman AD, McLaughlin J, Schellenberg R, Penner C, **Arbour L**, Tsang P, Ballem P, Lim KI. Hereditary angioedema managed with low-dose danazol and C1 esterase inhibitor concentrate: a case report. *J Obstet Gynaecol Can* 28(1):27-31, 2006.

19. Ko HH, Chung H, McMahon B, Hurlburt K, **Arbour L**, Yoshida E. Liver disease in the indigenous communities of the arctic, sub-arctic and pacific northwest: an approach to investigations of Alaska Natives and the First Nations of British Columbia. *BC Medical Journal* 48(5):216-221, 2006.
20. Chung H, Riley M, Ho JK, Leung B, Jevon PG, **Arbour LT**, Barker C, Schreiber R, Yoshida EM. Retrospective review of pediatric and adult autoimmune hepatitis in two quaternary care centres in British Columbia: increased prevalence seen in British Columbia's First Nations. *Can J Gastroenterology* 21(9):565-568, 2007.
21. Godwin K, Sibbald B, Lowry RB, Bedard T, Kuzeljevic B, **Arbour L**. Is folic acid fortification changing the frequencies of congenital anomalies other than neural tube defects? *Canadian Journal of Public Health* (In press. Accepted for publication February, 2008)
22. **Arbour L**, Rezazadeh S, Eldstrom J, Weget-Simms G, Rupps R, Dyer Z, Tibbits G, Accili E, Casey B, Kmetc A, Sanatani S, Fedida D. 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* (In press. Accepted for publication March, 2008)

Books and Book Chapters

1. Oyvind J, **Arbour L**. Maternal child health in circumpolar nations. In: Young KT (ed). *Arctic Populations in Transition*. University of Toronto Press, Toronto, Canada. 2008

Other

1. Shieh JTC, Hoyme HE, **Arbour L**. Inherited Systemic Hyalinosis. *Gene Reviews* (an on-line peer reviewed genetics resource). Accessed at: <http://www.geneclinics.org>. February 27, 2008.

Published Abstracts

1. **Arbour L**, Pekeles G, Hodgins S. Birth Defects in the Inuit of Baffin Island and Arctic Quebec, International Congress on Circumpolar Health, Harstad, Norway, June 2000.
2. **Arbour L**, Reilly C, McGillivray B, Prendiville J, Dimmick J. Infantile Systemic Hyalinosis: A rare syndrome of progressive, painful contractures with peculiar hyperpigmentation and death in infancy. The David W. Smith Workshop on Malformations and Morphogenesis, August 2000, La Jolla, California. Proc Greenwood Genet 20: 2001.
3. Power P, **Arbour L**, Ives, E. Pericentric Inversion of Chromosome 12 in a Large Newfoundland Family. ASHG Annual Meeting, San Diego, California, AJHG 69(4)296, 2001.
4. Sicilano D, Lim K, **Arbour L**. Ultrasound markers after a positive triple screen: How is individual risk modified? ASHG Annual Meeting, San Diego, California, AJHG 69(4) 671, October, 2001 .
5. Aubertin G, Coleman G, Shaw D, Van Allen M, **Arbour L**. Prenatal Diagnosis and management of unilateral multicystic kidney. ASHG Annual Meeting, AJHG 69(4) 660, 2001.
6. **Arbour L**, Yoshida E, Field L. Primary Biliary Cirrhosis in BC's First Nations People. International Congress of Circumpolar Health, Nuuk Greenland, September 2003.
7. Egeland G, Soueida R, Kuhnlein H, **Arbour L**. Vitamin A Intake in Inuit Women in Canada. International of Congress of Circumpolar Health, Nuuk Greenland, September 2003.
8. **Arbour L**. Nutrient/Genetic Determinants of Birth Defects in Northern Canada: Challenges to Carrying Out Research With an Aim to Improve Outcome. 8th Australian Rural and remote Health Conference, Alice Springs, Australia, March 2005.
9. **Arbour L**. Will Folic Acid Fortification Have an Effect on the High Rate of Septal Heart Defects in the Inuit of Nunavut? International Meeting on Inuit and Native American Health, Seattle, Washington, May 2005.
10. **Arbour L**, Rupp R, Field L, Egeland G. Genetic Nutrient Determinants of Heart Defects in the Inuit of Baffin Island. 26th Annual David W Smith Workshop on Malformations and Morphogenesis, Iowa City, Iowa August 2005.
11. Riley M, Ho JK, Schreiber R, Yoshida E, **Arbour L**. PBC and AIH in a First Nations Mother-Daughter Pair. Can J Gastroenterol 19:191, 2005.

12. Nuk J, Pearson M, Atkinson J, Kelly K, Farquharson D, **Arbour L**. Prenatal testing and Screening in Northern British Columbia: A Question of Access or Personal Preference? American Society of Human Genetics (ASHG), New Orleans, October 2006.
13. Godwin K, **Arbour L**. Reduction in Non-Neural Tube Defects Since Folate Fortification. ASHG, New Orleans, October 2006.
14. **Arbour L**. Genetic and Nutrient Determinants of Congenital Heart Defects in the Inuit. International Congress on Circumpolar Health, Novosibirsk, Russia, June 2006.
15. Yoshida EM, Riley M, **Arbour LT**. Autoimmune liver disease and the Canadian First Nations Aboriginal communities of British Columbia's Pacific Northwest. *World J Gastroenterol* 12(23):3625-3627, 2006.
16. **Arbour L**, Osborne G, Rupps R, Forth M, Nowdlak M, Egeland G, Field L, Rozen R. Genetic and Nutrient Determinants of Congenital Heart Defects in Nunavut. ASHG, San Diego, California, October 2007.
17. Eldstrom J, Rezazadeh S, Rupps R, Sanatani S, Casey B, Tibbits G, Fedida D, **Arbour L**. KCNQ1 V205M Missense Mutation Causes LQTS in a Northern Canadian Community. ASHG, San Diego, California, October 2007.

Non Refereed Publications

Proceedings

1. **Arbour L**. Genetic research in Inuit and 1st Nations communities: experiences from Baffin Island, northern Quebec and British Columbia. In: Glass K, Kaufert J (eds). *Research Involving Aboriginal Individuals and Communities: Genetics as a Focus*. Proceedings of a workshop of the Consent Committee, The National Council on Ethics in Human Research (NCEHR), 2001.
2. **Arbour L**. The researcher's perspective: is there a research imperative? In: Glass K, Kaufert J (eds). *Continuing the Dialogue: Genetic Research with Aboriginal Individuals and Communities*. Proceedings of a workshop sponsored by the Canadian Commission for the United Nations Educational, Scientific and Cultural Organization (UNESCO), Health Canada and the National Council on Ethics in Human Research, 2002.
3. **Arbour L**, Field L, Erikson A, Yoshida E. The mystery of primary biliary cirrhosis in British Columbia's First Nations people. Proceedings of the 12th International Congress on Circumpolar Health. Nuuk, Greenland, September 2003. *Int J Circumpolar Health* 63(Suppl 2):185-188, 2004.

4. **Arbour L**, Rupps R, Field L, Egeland G. Genetic/ nutrient determinants of heart defects in the Inuit of Baffin Island. 26th Annual David W Smith Workshop on Malformations and Morphogenesis. Iowa City, Iowa, August 2005.
5. **Arbour L**, Osborne G, Sobol I, Rupps R, Field LL, Egeland G. Folic acid and birth defects in the north: are we doing enough? Proceedings of the ICCH, 2006. Int J Circumpolar Health 66(Suppl 2), 2007.

Invited Presentations

1. January 2002: Vancouver, BC. National Council on Ethics in Human Research Regional Ethics Update – ‘Genetic Research in Aboriginal Populations: What Should an REB Know?’
2. November 2002: Ottawa, ON. National Aboriginal Health Policy Conference – ‘Genetic Research in Aboriginal Populations’.
3. February 2003: Montreal, QC. International GELS Symposium - ‘Genetic Research With Canadian Aboriginal Populations’.
4. March 2003: Stellenbosch, South Africa. The African Genomic Initiative - ‘*DNA on Loan*: Canadian Discussions on Genetic Research in Aboriginal Populations’.
5. May 2003: Vancouver, BC. National Aboriginal Conference on Hepatitis C – ‘Beyond Hepatitis C, the Mystery of Primary Biliary Cirrhosis in BC’s Aboriginal Population’.
6. May 2003: Prince George, BC. University of Northern BC Northern Doctors Day – ‘Genetics and Genomics Research: Will It Make a Difference to Health Care?’
7. April 2004: Vancouver, BC. Provincial Aboriginal Health Conference - ‘*DNA on Loan*: Can Genetic /Biomedical Research be Carried Out in a Participatory Manner?’
8. May 2004: Prince George BC. University of Northern BC Northern Doctors Day – ‘Finding Solutions to Accessing Prenatal and Cancer Genetic Services’.
9. November 2004: Edmonton, AB. University of Alberta - ‘Primary Biliary Cirrhosis in BC’s First Nations Population’.
10. January 2005: Ottawa, ON. Canadian Congenital Anomalies Surveillance Network (CCASN) – ‘The Impact of Folic Acid Fortification in Nunavut: Is Enough Being Done to Reduce the Rate of Birth Defects?’
11. June 2005: Vancouver, BC. 2005 SADS Foundation Conference – ‘The Genetics of Long QT Syndrome’.

12. June 2005: Fort Yates, ND. Northern Plains Epidemiology Center – ‘Canadian Experiences Carrying Out Genetic Research with Aboriginal Communities’. (key note)
13. January 2006: Seattle, WA. Institute for Public Health Genetics Seminar Series, University of Washington – ‘*DNA on Loan: Approaches to Carrying Out Genetic Research with Aboriginal Families and Communities*’.
14. March 2006: Anchorage, AK. Alaska Native Health Conference – ‘Genetic Research with Canadian Aboriginal Populations’. (key note)
15. November 2006: Vancouver, BC. BC First Nations Health Directors Conference – ‘Genetic Research in BC First Nations People’.
16. November 2006: Ottawa, ON. Canadian Congenital Anomalies Surveillance Network (CCASN) – ‘The Development of a Tri-territorial Birth Outcomes Registry’. (published through Health Canada)
17. April 2007: St. Saviour, QC. Canadian Genetics Diseases Network (Knowledge Translation) – ‘Off the Beaten Path: Relevant Genetic Research with Inuit and First Nations People’.
18. April 2007: Banff, AB. Circumpolar Research Symposium – ‘The Development of a Tri-territorial Birth Outcomes and Child Health Surveillance System’.
19. May 2007: Santa Cruz, CA. University of Southern California – ‘Genomics and Justice: Genetics Research with Canadian Aboriginal People’. (published on line)
20. November 2007: Toronto, ON. CIHR Institute of Genetics New PI Meeting – ‘The Study of Select Genetic Conditions in Canadian Aboriginal Populations’.

Most Significant Contributions:

1) The study of folic acid metabolism, in cases and controls of a Cree Community with a high rate of Spina Bifida was completed. (**Arbour L**, et al. Neural Tube Defects, Folate Metabolism, and Dietary Folate Intake in a Northern Canadian Aboriginal Population. *Int J Circum Health* 61:341-349, 2002). Community impact was through concurrent public health efforts that raised the awareness of the importance of Folic Acid in preventing birth defects.

2) Reviewed 2500 births in Arctic Quebec and Baffin Island to determine the rates of Birth Defects from 1988-1993. The rates of Birth Defects (in particular Congenital Heart defects) were more than twice that of the general population. The Health Care community was made aware of the concern to encourage early detection. **L. Arbour et al**, Increased incidence of heart defects and other malformations in the Inuit of Baffin Island and Arctic Quebec: a baseline study. *IJCH* 63(3)251-266, 2004. A follow-up case control study on pathogenesis (genetic/nutrient determinants) has now been completed and publication is underway (see abstract ASHG October 2007). A follow-up study of 2000 births on Baffin Island has just been completed to determine if rates are as high post fortification.

3) Analysis of a large kindred with several cases of Wilms tumour, This lead to molecular collaboration for the delineation of the clinical and molecular genetic features of familial Wilms Tumour. See: *JNC1* 92(8):650-2, 2000, also see: *Nature Genetics* 13, 461-463, 1996; *Oncogene* 14:3099-102, 1997; and *Human Genetics* 103 (5):547-556, 1998)

4) A high rate of primary biliary cirrhosis in Pacific Northwest First Nations people has been identified, leading to a demographic, clinical and genetic study (linkage currently underway). Arbour et al “Characteristics of Primary Biliary Cirrhosis in British Columbia’s First Nations People. *Can J Gastroenterol* (5):305-310, 2005.) **Winner of Thompson-Williams award February 2006.** This study has lead to the recognition that the rate is also high in South East Alaskans. An international collaboration is underway to explore the environmental and genetic determinants of PBC in Pacific Northwest Native Peoples. A successful collaborative workshop was held in August 2005 “Without Borders: Primary Biliary Cirrhosis and other Autoimmune Disease in American Indians and First Nations People of the Pacific Northwest with the support of a CIHR International Opportunities Program Grant.

5) Participated in five international workshops, (primary organizer of 3) held in Canada to explore ethically acceptable ways to carry out genetic research in aboriginal populations. These workshops have been presented in summary at international meetings such as the GEL³S International Conference held in Montreal and The African Genome initiative held in South Africa. They have also contributed to the development of the CIHR Guidelines for carrying out research with Aboriginal communities. A manuscript published in: *Community Genetics* Arbour L, Cook D “DNA on Loan: Issues to consider when carrying out research with aboriginal

communities” From this and above background work, I have been able to establish a “Community Genetics Research Program” where conditions affecting Aboriginal people disproportionately are being addressed. Current Projects Include Long QT Syndrome in the North of British Columbia, Birth defects in Nunavut, CPT1 deficiency in Northern aboriginal infants, epilepsy in Northern British Columbia and the Development of a comprehensive birth outcomes and child health surveillance system.

Activities and Contributions for L Arbour

Committees:

- 1) September 2000- September 2003- Chair- BC Children's Ethics Committee
- 2) September 2000- 2004- UBC Residency/Fellowship Program Director Medical Genetics, UBC
- 3) June 1998 - 2004 Canadian Pediatrics Society Ethics Committee.
- 4) May 2003- 2006t CIHR- Scientific Advisory Committee on Northern and Rural Health Research
- 5) July 2003-present *Chair* of the Birth Defects Working Group: IUCH
- 6) September 2003-2006 (CCMG): Chair of Ethics and Public Policy committee
- 7) September 2003- 2006t. *Secretary* Canadian Society of Circumpolar Health.
- 8) September 2003-2006 *Treasurer* International Union of Circumpolar Health
- 9) June 2006-present Secretary/Treasurer of the International Network of Circumpolar Health Researchers:
- 10) April 2004-2007 CIHR IAPH Aboriginal Ethics Working Group
- 11) January 2004-present Consultant for the BC Vital Statistics Birth Defects Registry
- 12) May 2004-present CCMG Liaison for the CPS Surveillance System
- 13) September 2006-present Institute Advisory Board member for the CIHR Institute for Human Development, Child and Youth Health

Peer Review:

Grant reviews: 2007- present CIHR Aboriginal Health Peer Review committee

External reviewer: Canadian Diabetes Foundation, May 2004; Canadian Breast Cancer Foundation, May 2005; NSERC, July 2005; CIHR External reviewer (four grants) 2005-2007; Alberta Heritage Foundation, September 2005; Australian Fellowship 2007

Journals: Editorial Board (associate): International Journal of Circumpolar Health

Others: Oncology-Hematology, 2005; Canadian Medical Association Journal 2000, 2007; Developing World Bioethics 2004; IJCH 2007

Career Development:

In 2003, I was successful in competing for an innovative Clinical Investigatorship through the CIHR Institute of Genetics which provided substantial clinical relief to allow me to develop a research career focusing on genetic conditions that contribute to health disparity in Canadian Aboriginal populations. This was renewed in 2005, and was followed by a Michael Smith Foundation Health Research Award that commenced in 2007. These opportunities have allowed my research program to flourish, including the development of research applied for in this grant.

In order to refresh research skills, I chose to concentrate on the analysis of complex traits, and therefore participated in the training opportunities listed below. All projects have a direct knowledge translation component.

May 2004: Duke University Short Course in Complex Trait Analysis

May 2005 6 week study leave to Duke University (CIHR Travel Grant) Complex Trait Analysis

July 2006: Wellcome Trust Advanced Course on Multifactorial Analysis: Cambridge University, UK