

Nutrient/genetic determinants of birth defects in northern Canada: challenges to carrying out research with an aim to improve outcome

Laura Arbour, Department of Medical Genetics, University of British Columbia, Canada

Birth defects are an important worldwide cause of infant morbidity and mortality. The Canadian arctic is no exception, where there is evidence for a disproportionately high rate of congenital heart defects. A baseline study of Inuit births occurring between 1989 and 1994 confirmed that congenital heart defects, specifically ventricular septal defects and atrial septal defects were more than four times more frequent in Nunavik and Nunavut than in other Canadian populations contributing to an overall rate of birth defects in these Arctic locations that was twice that of other Canadian populations. Although genetic predisposition may play a role and is the subject of study, more directly applicable to public health policy is whether there is sufficient folic acid and vitamin A in the 'northern' diet since both are important in septal heart development. Indeed there is evidence to suggest that supplemental folic acid in the periconceptional period is protective for heart defects.

Thus a case control study of mothers of affected children, using food frequency questionnaires and blood vitamin levels is under way on Baffin Island, an arctic island in Nunavut covering 476 070 square kilometres. For this Inuit population, community involvement is essential from the onset. The presumed and real challenges of carrying out this and other studies in remote areas may prohibit such research and contribute to either health policy being adopted without evidence, being irrelevant to remote populations or the issues being ignored altogether. The challenges of implementing this study will be presented, with results that demonstrate the importance of carrying out research relevant to the issues present in rural and remote populations with the aim of improving health outcome with evidence.

PRESENTER

Laura Arbour is a Paediatrician and Clinical Geneticist in the Department of Medical Genetics at the University of British Columbia. Her clinical practice and research focuses on northern and Aboriginal health issues as they pertain to genetics. She is funded by the Canadian Institutes of Health Research from both the Institute of Genetics and the Institute of Aboriginal Peoples Health. Her research focus is on investigation of gene/environment interaction for complex conditions in Canadian Aboriginal populations with an aim to improve health. She is currently investigating the nutrient and genetic basis of congenital heart defects in the Inuit of Baffin Island.