Primary Biliary Cirrhosis (PBC) in BC Coastal First Nations People: An Update to Participants

March 2011

This is the fifth in a series of update notices sharing the progress of the PBC study with study participants and their families.

Thank you to all of the families who are participating in our study of the genetic causes of primary biliary cirrhosis (PBC) in B.C.'s First Nations People. We hope that this study will help answer the question of why PBC is so much more common in BC Coastal First Nations populations than in non-First Nations people. In addition, we aim to educate health care providers about the higher rate of PBC among First Nations people so that it can be diagnosed promptly and treated at an early stage in the disease. Your help with this study is very much appreciated.

We appreciate the funding we have had for the last 2 years from the Canadian Liver Foundation (CLF). This funding has allowed us the opportunity to get a good start on searching for possible genes that might be increasing the risk for PBC. However, there is more work to be done, and we will continue to search for more funding sources so we can keep moving forward with this important research. Even when our funding ends, we will still be available to answer your questions and enroll any new participants who are interested in taking part in the research. Currently we have 131 participants, including 45 with PBC.

What has been accomplished over the last two years?

1. Genetic Analysis:

The expanded genetic analysis was completed in 2010. This compared the genetic makeup of some participants with PBC to their family members who did not have PBC. The results from this genetic analysis are complex, and are still being summarized by our research associate, Dr. Sirisha Asuri. There were a number of genetic differences found between the group of those with PBC compared to those without. Some of these differences point to interesting genes that may promote the development of PBC. We aim to complete our analysis by late summer. At that time, we will send out the summary of our findings to all participants and write an article for publication in a scientific journal. After this phase of the genetic analysis is complete, we will decide what the next steps should be.

2. Website Development:

We now have a website that highlights the work of our Community Genetics Research Program. This website is under the umbrella of the Division of Medical Sciences at the University of Victoria.

To access our website, go to http://medsci.uvic.ca/faculty/arbour.php

If you scroll down the page, you will see a "research" tab. Here, you can click on the "Primary Biliary Cirrhosis in the First Nations People of BC" to access information about the PBC study, including previous publications, information pamphlets, contact information, etc. We hope that you will find it to be a helpful resource!

3. Updated analysis of the chance for close relatives to develop PBC, and the frequency of other autoimmune conditions in PBC families:

We had the pleasure of working with Valerie Taylor, a UVic Biology Co-op student, last summer. Valerie won both a Kloshe Tillicum Undergraduate Aboriginal Health Research Training award and a BC Clinical Genomics Network summer student award to help us with the PBC project. Specifically, Valerie organized and updated our participant medical information in preparation for the genetic analysis. She also updated and re-analyzed data about how often PBC occurs in close relatives of affected individuals, and how often other autoimmune conditions (such as lupus, rheumatoid arthritis, Sjogren's syndrome, thyroid disease, etc.) occur within PBC families.

Based on information gathered from our study participants, each first-degree relative (parent, full sibling, or child) of someone with PBC has approximately a 5% (1 in 20) chance of developing PBC. This chance rises to 9% if the first-degree relative is *female* (about 1 in 11). The chance for more distant relatives to develop PBC would be much lower. Although these chances are much higher than the average person's risk for developing PBC, it is reassuring to know that most people who have a relative with PBC do NOT go on to develop the disease. Looking at the numbers the other way around, **there is a 95% chance that a first-degree relative of someone with PBC will NOT develop the condition** (91% chance that a *female* first-degree relative will NOT develop PBC). These results are very similar to the analysis carried out earlier in our study (2005) with fewer participants.

Other autoimmune conditions, such as lupus, rheumatoid arthritis, Sjogren's syndrome (dry eyes & mouth), occur more frequently in families with PBC. 56% of our study participants who have PBC were found to have an additional autoimmune condition, while 29% of the unaffected relatives in our study have an autoimmune condition. It is apparent that PBC and certain autoimmune conditions must share some of the same genetic factors and environmental "triggers". Our genetic analysis will help us understand this more fully.

4. PBC Fact Sheet:

We have created an updated fact sheet about PBC, which includes information about the symptoms, diagnosis, and treatment of the condition. We circulated this fact sheet to Fairlie Mendoza, Community Health Nurse at Ts'ewulhtun Health Center in Duncan in the summer, so perhaps some of you have already seen it. We have included a copy with this update letter, and we hope it will help you to inform your relatives, friends, and/or health care providers about PBC.

Where do we go from here?

The study will continue to remain open for enrollment. Anyone of First Nations ancestry who has PBC, or has a relative with PBC, is welcome to join our research study. We are happy to hear from you at any time. Please feel free to contact us if you wish to enroll, or if you have any questions. We are always grateful to receive updates about your health history, as well.

If you have questions about PBC and the work we are doing, or if you are interested in joining the research study, please contact **Samantha Lauson** (genetic counsellor/research assistant) on our toll-free line: **1-888-853-8924** or by e-mail: pbcstudy@uvic.ca.

If you have a specific question for Dr. Laura Arbour (principal investigator), she can be reached at: (250)472-5544, or larbour@uvic.ca

Wishing you and your families all the best in 2011!
Sincerely,

Dr. Laura Arbour and the Community Genetics Research Program team.