

The research study

Dr. Laura Arbour's research team led a study to understand the genetic causes of the high rate of PBC in BC First Nations. Altogether, the research study included 105 individuals, comprised of 44 people with PBC and 61 unaffected relatives from 32 First Nations families. Family history and the presence of other autoimmune diseases were documented. The medical records and available biopsies of the participants were reviewed. Genetic 'linkage' testing was carried out.

Key findings

- ❖ Our genetic studies support that no one *single* gene is responsible for the high rate of PBC in BC First Nations families.
- ❖ Our research showed that a combination of *several* small 'genetic signals' may increase the risk for PBC. Some of these signals have been detected in previous studies of other PBC populations.
- ❖ Why it is more common in BC First Nations families is still unknown, but it is possible that certain environmental factors, combined with multiple small genetic factors, increases the risk.

For more information, contact us at:

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Additional information about PBC:

- ❖ PBCer's organization:
www.pbcers.org
- ❖ Canadian Liver Foundation:
www.liver.ca
- ❖ The PBC Patient Support Network: www.pbcpatsup.net

Island Medical Program



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**Primary Biliary
Cholangitis (PBC)
Among Coastal First
Nations in British
Columbia**

**Our research is
showing that many
small genetic
factors may be
contributing to
PBC in First
Nations of British
Columbia.**

What is PBC?

- ❖ Primary Biliary Cholangitis (PBC) is a chronic autoimmune disease that attacks the small bile ducts in the liver.
- ❖ In PBC, bile and other substances build up over time and eventually cause damage to the liver (cirrhosis).

How frequent is PBC?

- ❖ PBC is a rare disease in non-First Nations populations, but more frequent in First Nations of British Columbia, especially coastal First Nations.
- ❖ It has been estimated that PBC among these First Nations could be as much as 8 times more common than in the non-First Nations populations.
- ❖ PBC predominantly affects adult women with a female: male ratio of 10:1.

Stigma

PBC used to refer to Primary Biliary *Cirrhosis*; however, the word cirrhosis was replaced in an attempt to avoid stigmatization. The word cirrhosis refers to scarring of the liver. **Although alcohol is a common cause of liver cirrhosis it is not the cause of liver damage in PBC.**

Symptoms

- ❖ PBC can have non-specific symptoms like fatigue and itchiness of the skin.
- ❖ Other possible symptoms include jaundice (yellowing of skin and eyes), swelling of the ankles or abdomen, and easy bruising and bleeding.
- ❖ Patients with PBC are also more likely to have other autoimmune conditions like rheumatoid arthritis, thyroid disease and lupus.

Diagnosis

- ❖ Diagnosis of PBC is confirmed by specific blood tests, such as positive antimitochondrial antibody (AMA) and liver enzyme tests (e.g. high alkaline phosphatase).
- ❖ In some cases, a liver biopsy is needed to confirm the diagnosis.

Treatments

- ❖ Ursodeoxycholic acid is the most common and effective treatment. Obeticholic acid is also under study.
- ❖ Although these treatments slow the progression of PBC, some patients eventually require a liver transplant.

Prognosis

- ❖ The outcome of PBC varies greatly from one person to another.
- ❖ PBC usually progresses slowly. Many patients lead active and productive lives and have few symptoms for 10-20 years after their diagnosis.
- ❖ However, in some patients the condition progresses more rapidly, and liver failure may occur within a few years.