Once **your WGS test results** are back, the doctor who referred you to the study will share the results with you. One of our study genetic counsellors will also be available to discuss the meaning of your results and provide you and your family with additional support.

WHAT WILL THE RESULTS OF THIS STUDY MEAN TO ME?

No one knows whether the results of this study will help you or your family. Some people will receive useful information from this study, while others will not. One of our study genetic counsellors is available to discuss the pros and cons with you in more detail.

It is possible that this study will find a genetic cause for the health condition affecting you and/or your family. Some people find it helpful to learn the reason for their health condition, so they no longer have to wonder and can stop having medical tests to find the cause. If a genetic diagnosis is made, it may help your family and healthcare team learn more about what prognosis to expect, and how to provide the best healthcare for you or your family members.

This study is paid for by research grants from:

GenomeCanada

Genome BritishColumbia
Leading > Investing > Connecting

CIHR IRSC
Canadian Institutes of Health Research / Instituts de recherche en santé du Canada

BC Children's Hospital
An agency of the Provincial Health Services Authority

MICHAEL SMITH FOUNDATION FOR HEALTH RESEARCH
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(in-kind)

WHO DO I CONTACT FOR MORE INFORMATION?

If you would like to hear more about this study, or confirm if you are eligible to join, please contact the study genetic counsellor at the site nearest you:

Victoria – Sarah McIntosh
sarahmc@uvic.ca
toll-free: 1-888-853-8924

or

Vancouver – Karen Jacob
karen.jacob@bcchr.ca
604-875-2000 extension 5271

If you prefer our genetic counsellor to initiate contact with you instead, please tell your referring doctor so he/she can ask us to reach out to you.

v.2, 23 October 2018

PRECISION DIAGNOSIS FOR INDIGENOUS FAMILIES WITH GENETIC CONDITIONS

This research study is one of four activities of the ***Silent Genomes Project*** supported by Indigenous partners: First Nations Health Authority, Inuit Tapiriit Kanatami, Métis National Council and the Assembly of First Nations.

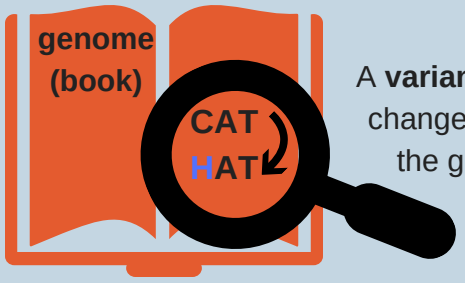
https://bcchr.ca/silent-genomes-project

WHAT

is this study about?

- The goal of the study is to try to find the cause for health conditions where no diagnosis has been made by regular tests available through the medical system.
- In this study, Indigenous families in Canada with an undiagnosed health condition will be offered a state-of-the-art genetic test called **'whole genome sequencing' (WGS)** to check if their condition is caused by changes in **genes.**

Genes are the instructions we inherited from our parents, which tell our bodies how to work. We have about 25,000 different genes, and each gene does specific jobs in our bodies. Genes are made up of **DNA**, a long string of genetic '*letters*' that our bodies can read. Spelling changes in DNA (e.g. when a letter is changed, added, or deleted), are called **variants**. Some variants cause a gene to be misread, and can lead to health problems.



A **variant** (spelling change) found in the genome

The whole package of all the DNA in our body is called the **genome**. The **genome** is the *book of all the DNA letters* in a person.
The **WGS** test searches the entire genome to look for DNA variants that might explain the reason for a health condition.

WHY

am I invited to learn about this study?

- You may be eligible to take part in this research study because you (or someone in your family) has a health condition that is thought to have a genetic cause, but no diagnosis has been made yet.

- Genetic conditions occur in all populations around the world, but we are only inviting families that self-identify as Indigenous (**First Nations, Métis, or Inuit**) into this study in an effort to reduce the barriers that many Indigenous families face when trying to access WGS testing and genetic diagnosis.

WHO

is doing this study?

Dr. Laura Arbour and **Dr. Anna Lehman,** geneticists at the University of British Columbia, are leading this study. Many other scientists and health professionals are involved.

DO I HAVE TO JOIN THIS STUDY?

You do **not** have to join this study if you do not want to. It is up to you. If you choose to join and then change your mind, you can stop being in the study at any time.

Your doctors and other healthcare providers will continue to try to diagnose the health condition in your family, regardless of whether or not you join this study.

IF I JOIN THE STUDY WHAT WILL HAPPEN?

If you are eligible for this study and agree to join, you will be asked to do the following steps:

- **Meet with one of our study genetic counsellors**, who will explain the study, help you understand the pros & cons of having WGS testing, walk you through the consent process, and gather information about the condition in your family.
- **Have a blood sample drawn** (or a saliva or cheek-swab sample) to collect genetic material (called DNA) for WGS testing.