If a definite (or possible) disease-causing gene change is discovered, your test results will be given back to you by the doctor who referred you, and you will able to discuss the meaning of your results with a genetic counsellor.

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. The study genetic counsellor is available to discuss the potential 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 done to try 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.

WHO DO I CONTACT FOR MORE INFORMATION ABOUT THE STUDY?

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

Brenda McInnes, MSc ,CCGC

Alberta Children's Hospital

brenda.mcinnes@ahs.ca

403-955-7028

If you prefer the study genetic counsellor/genetic associate to initiate contact with you instead, please let your referring doctor know, so your doctor can let us know to reach out to you.

UNIVERSITY OF CALGARY

This study has been approved by the University of Calgary Conjoint Health Research Ethics Board

REB#18-1847

PRECISION DIAGNOSIS FOR INDIGENOUS FAMILIES WITH GENETIC CONDITIONS

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 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 like instructions that tell our bodies how to work. We each have about 25,000 different genes which are passed down from our parents. Genes are made up of letters and words, similar to a book.

If there is a spelling difference in a gene (called a **"gene change"**), the gene may be misread and can lead to health problems. The WGS test searches the entire set of genes at one time, to look for gene changes that might explain the reason for health conditions.

WHY

am I being 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?

This study is paid for by research grants from many organizations, including Genome Canada and the Canadian Institutes for Health Research.

The doctors leading this study in British Columbia are **Dr. Laura Arbour** (based in Victoria) and **Dr. Anna Lehman** (based in Vancouver). There are many other scientists and health professionals involved in this study at both sites.

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 take care of you as they have in the past, 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 a study genetic counsellor**, who will explain the study to you, help you to 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 possibly a saliva or cheek-swab sample instead) to collect genetic material (called 'DNA') for WGS testing.