FREQUENTLY ASKED QUESTIONS

SILENT GENOMES RESEARCH PROJECT - INDIGENOUS BACKGROUND DNA VARIANT LIBRARY

WHAT IS THE SILENT GENOMES RESEARCH PROJECT?

It is a project aimed at reducing health care disparities and improving diagnostic success for Indigenous patients with suspected genetic disorders.

WHAT IS THE PURPOSE OF THIS PART OF THE SILENT GENOMES PROJECT?

There is a lack of genomic reference data for Indigenous peoples. The intent is to build and manage an Indigenous background DNA variant library (IBVL) with Indigenous peoples in Canada. It is a tool to help get a diagnosis, which is a first step towards treatment and management of what are usually complex health conditions.

WHAT IS DNA (deoxyribonucleic acid)?

DNA is like a blueprint of instructions for your body. A gene is a piece of DNA with a code for a specific instruction. It is written in a special alphabet only four letters long.

I-A-M-W-O-N-D-E-R-F-U-L

T-C-G-G-C-T-A-A-T-C-G-G

HOW CAN THE PROPOSED IBVL BE USED?

Rare genetic conditions affect at least 1 in 50 people in Canada. They can affect anyone, and are often difficult to diagnose. It is estimated that about 30,000 Indigenous peoples in Canada are affected. The IBVL will be used to identify these rare genetic conditions.

WHAT IS AN *INDIGENOUS* BACKGROUND VARIANT LIBRARY?

A list of DNA variants seen in a population of Indigenous peoples. In our case, those who are not known to be affected by severe childhood onset genetic disorders.

HOW DOES A BVL WORK?

Each person carries 3.5-5 million variants, yet only 1 or 2 of those may be relevant to a diagnosis. BVLs show which variants in a population are common and unlikely to be disease-causing, and allow health care practitioners to quickly eliminate them to narrow the search for other variant suspects that may be causing their patient's genetic condition. A variant with a high frequency means it is fairly common; a low frequency variant in a population is considered rare.

WHY DO WE NEED AN INDIGENOUS BACKGROUND VARIANT LIBRARY?

At this time, there are genomic databases throughout the world, but they have little to no variants from Indigenous peoples. In fact, very little is known about Indigenous background DNA variation. Without it, Indigenous peoples may get left behind. The hope is that our own, more specialized IBVL will be used well into the future as a clinical tool to improve the diagnosis of genetic conditions and health care management for Indigenous patients and their families.

WHAT WOULD PARTICIPATION INVOLVE?

After appropriate consent, DNA would be collected through blood from Indigenous people not known to have a genetic disorder.

The DNA variants would be analyzed and the information stored in the IBVL.

WHAT ARE THE LIMITS? The IBVL is expected to immediately increase diagnostic success by about 30% and will increase as more DNA variants are added. Beware though, that the IBVL will not lead to diagnosis for all patients. The IBVL may also be used for research that directly helps patients, and only for our own benefit.

HOW WOULD MY IDENTITY AND DATA BE PROTECTED?

Your DNA sample will be de-identified, meaning your name and other identifying information will not be attached to it.

The IBVL database will be protected by a constantly monitored multi-layered security system and a back-up site in case of catastrophe. Human errors are minimized through a tightly monitored security system limited to those who need access to the data directly for patient diagnosis.

WHAT WOULD I GET OUT OF IT? By loaning your DNA, you are helping to reduce the inequality in health care for Indigenous peoples. You are supporting an Indigenous–controlled project. Participating is no cost to you or to Indigenous patients and families participating in the Project. While there is no immediate help to you or your family, genetic conditions can happen in any family. Your loan can help future generations.

WHO CONTROLS THE IBVL? Indigenous peoples guide how the IBVL is developed and used. The Silent Genomes Project is guided by Indigenous staff, Indigenous principal investigators, an international Indigenous advisory team, and a planned Canadian Indigenous steering committee. We are part of a larger team of committed non-Indigenous scientists and other experts who want to make this project work for us. Our focus is on the protection of rights, safety, property and legitimate uses of your DNA that you decide upon for yourself.

For example, *DNA on Loan* is a policy adopted by the Silent Genomes Project which means that you loan your DNA for the purpose of the research for which your free, prior and informed consent was obtained. Your DNA cannot be used for any other purpose.

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SILENT GENOMES RESEARCH PROJECT

This is one of a series of educational materials developed by primary author Laurie Kariiosta Montour, Kanien'kehá:ka, with support from the Silent Genomes Team

WHERE DO I GO FOR MORE INFORMATION? 1(888)853-8924 (ext. 2) or bcchr.ca/silent-genomes-project

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