SILENT GENOMES PROJECT

The Silent Genomes Project was established in 2018 as a national research initiative aimed at increasing access to genomic testing technologies and improving diagnostic outcomes for Indigenous people with genetic conditions. In its current phase, the main focus is building a sustainable path for continued genetic/genomic care. This phase encompasses three overlapping activities (‘Rare Insights’, IBVL Expansion, and Equity, social, and economic implications) with Indigenous engagement, governance, and capacity building integrated throughout.

SILENT GENOMES PROJECT

Indigenous Background Variant Library (IBVL)

Frequently Asked Questions

This booklet provides general information about the Indigenous Background Variant Library (IBVL), including what it is, why it was developed, and how it is used to support genomic diagnosis for Indigenous patients.

Who this booklet is for?

- Individuals and families
- Communities and partners

IBVL OVERVIEW

WHAT IS THE IBVL?

The Indigenous Background Variant Library (IBVL) is a secure genomic database developed through the Silent Genomes Project as a clinical tool to support genomic diagnosis for Indigenous patients. It shows the frequency of each DNA variant found in a population of Indigenous people*. The IBVL contains genomic data from several consented First Nations communities and individuals and is designed to expand over time as more individuals and communities choose to participate.
The IBVL is not a biobank containing human samples, nor will it be used to determine the level of Indigenous ancestry, geographical origins, or to challenge cultural beliefs. It will not be used for commercialization, patenting interests, or to provide 'direct to consumer' genetic test results. The purpose is to provide access to the benefits of genetic testing and diagnosis for Indigenous people.

HOW IS THE IBVL DIFFERENT FROM OTHER BVLs?

The IBVL is a unique BVL in several ways, including having Indigenous governance (see 'Governance Structure') and multiple layers of safeguards with restricted access (see 'Protection and Use of Indigenous Data Within the IBVL'). The intention of the IBVL is to provide access to the benefits of genomic medicine for Indigenous people, while ensuring that the principles of Indigenous Data Sovereignty are respected.

WHY IS AN IBVL NEEDED?

Genomic data are needed to understand which DNA variants are common and which are rare in different populations. This information helps doctors correctly interpret genetic test results, identify variants that may be causing disease, and avoid misdiagnosis. Each population worldwide has genetic variants that are unique to them and are not present in the DNA of other populations, or found at a different frequency. Therefore, having

DNA variant frequency information that is specifically from Indigenous people helps with more accurate genetic diagnosis and better care for Indigenous patients.

WHEN WAS THE IBVL CREATED?

Development of the IBVL occurred over a period of 4 years (2020-2025) and the launch of the IBVL for genetic diagnosis and health research was in January 2025.

PROTECTION AND USE OF INDIGENOUS DATA WITHIN THE IBVL

HOW IS INDIGENOUS DATA WITHIN THE IBVL PROTECTED?

The IBVL is stored on secure servers at the BC Children's Hospital Research Institute in Vancouver, British Columbia. It is protected by multiple layers of technical and governance safeguards, only accessible by authorized personnel, and subject to regular security audits. Access is highly restricted, thoroughly controlled, and limited to approved health care professionals involved in genomic diagnosis, using the data for the purposes of clinical diagnosis. Downloading the full data set from the IBVL database is not possible.
Another key principle of the IBVL is that no individual data is available, and only population frequencies of variants will be included. This means that no one person's data can be recognized or accessed in any way.

WILL INDIGENOUS DATA WITHIN THE IBVL BE ACCESSIBLE FOR RESEARCH?

Yes, Indigenous data within the IBVL ***may*** be accessible for health-related research that is approved by the SG-IBVL Governance Committee. There is no open access for researchers to use the IBVL. Health research has the goal of improving diagnosis and treatment of disease and improving the

health and quality of life of individuals. Researchers who wish to utilize variant frequency data present in the IBVL should submit a *'Clinical Research Review Request'* form, which is then considered by the SG-IBVL Governance Committee.

WHAT ARE THE FACTORS THAT DETERMINE RESEARCH APPROVAL?

The Committee comes together to determine its clinical goal, how exactly the research can benefit Indigenous people, how results will be shared with the community, whether there are cultural or other risks, and how these are addressed. Only if the Committee decides that the research is an acceptable use of the IBVL, then the researcher will be granted access to the data to use for the purposes of their specified project.

GOVERNANCE STRUCTURE

WHO GOVERNS THE IBVL?

The IBVL is governed by the SG-IBVL Governance Committee who provides cultural oversight and strategic advice and decisions in support of the collaborative expansion and utilization of the IBVL, including respectful, culturally safe policies regarding access to data for clinical diagnoses and health research, while engaging future generations of Indigenous scholars and community members.

IS THE SG-IBVL GOVERNANCE COMMITTEE INDIGENOUS-LED?

Yes, the SG-IBVL Governance Committee is Indigenous-led and Indigenous-operated. The Governance Committee works in partnership with the Silent Genomes team and in consultation with Indigenous communities, partners and leaders as the IBVL expands to ensure that participating Indigenous individuals, families and communities have their genomic data securely protected. The Committee relies on the expertise of Knowledge Carriers, individuals with lived experience of genetic conditions,

Indigenous scholars, youth, and emerging leaders. Membership is open to those from First Nations, Métis, and Inuit communities.

WHAT IS THE IBVL'S GROWTH AND FUTURE DIRECTION

The IBVL is intended to be a sustainable, lasting resource for clinical diagnosis, with continued governance through the SG-IBVL Governance Committee. Increased uptake by appropriate healthcare professionals and better representation for all Indigenous people are priorities. The Indigenous Background Variant Library is designed to grow over time as more individuals and communities choose to participate, which will lead to improved frequency information and improved accuracy in diagnosis. As genomic data in the IBVL is only currently representative of some First Nations, engaging with Inuit and Métis communities, as well as expanding to other First Nations communities, is a continued priority.

PARTNERSHIPS

Listed below are research initiatives that have partnered with the IBVL. As the IBVL is envisioned to be a sustainable clinical resource, new partnerships may be formed over time.

CANADIAN ALLIANCE FOR HEALTHY HEARTS AND MINDS – FIRST NATIONS COHORT

Canadian ALLIANCE for Healthy Hearts & Minds

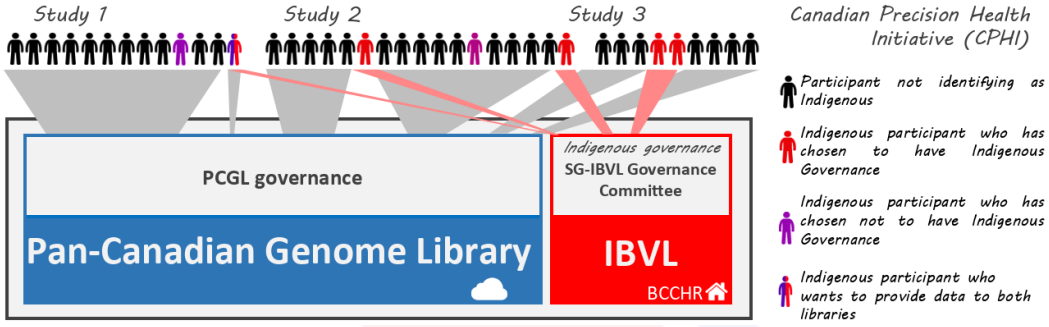
CAHHM is a prospective cohort study designed to investigate the impact of community level factors, individual health behaviours, and access to health services, on cognitive function, subclinical vascular disease, fat distribution, and the development of chronic diseases among adults living in Canada. The First Nations cohort involves a strong partnership between academic institutions and First Nations communities, with a governance model where each community owns their data, controls its dissemination, and decides when and what research is in the best interests of their specific community. Four First Nations communities that are a part of CAHHM decided to take

part in the IBVL. After significant engagement and discussions, 596 samples were transferred from CAHHM to the Silent Genomes Project to build the first version of the IBVL.

PAN-CANADIAN GENOME LIBRARY (PCGL)

Pan-Canadian Genome Library
Bibliothèque génomique pancanadienne

PCGL is a framework for unifying Canada's human genome sequencing efforts in a federated data management system. PCGL has been working in partnership with the Silent Genomes Project and the IBVL, to ensure that Indigenous patients will also benefit from national advances in genomic medicine while their genomic data remains protected under Indigenous-led governance and used only in ways that align with consent, cultural priorities, and data sovereignty principles. Indigenous participants who choose to take part in genomic research may opt to place their data in the IBVL. Their data would then be under the governance, storage, and access limits of the IBVL, in line with OCAP and UNDRIP principles.



WHERE DO I GO FOR MORE INFORMATION?

Useful links:

Silent Genomes Project
https://www.bcchr.ca/silent-genomes-project

IBVL General information and Educational Materials
https://www.bcchr.ca/silent-genomes-project/ibvl/general-information-ibvl-overview

SG-IBVL Governance Committee
https://www.bcchr.ca/silent-genomes-project/our-team/sg-ibvl-governance-committee

CONTACT

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** - "Indigenous peoples" is a collective name for the original peoples of North America and their descendants. The Canadian Constitution recognizes 3 groups of Indigenous peoples: First Nations, Inuit, Metis. These are 3 distinct peoples with unique histories, languages, cultural practices and spiritual beliefs. According to the 2021 Census, more than 1.8 million people in Canada identify as Indigenous, which represents 5% of Canada's total population*
From https:/www.rcaanc-cirnac.gc.ca/eng/1100100013785/1529102490303)