



# The Silent Genomes Project

## Great News

Four First Nations communities have approved the transfer of DNA samples to begin sequencing for the Indigenous Background Variant Library (IBVL).

A variant catalogue pipeline was custom-built for the IBVL. This open access tool (CAFÉ (Cohort Allele Frequency Estimation Pipeline) aims to further reduce barriers for under-represented populations around the world who are building their own variant catalogues, on local servers.

There is still a great deal to be done to build an IBVL that will benefit Indigenous patients, but we are grateful crucial first steps are in place.

## In Summary

- 22 S-GIRDD Meetings
- 2 videos
- 13 Community Visits
- 3 Annual Meetings

## Activity 1

### Summer Internship for Indigenous Peoples' Genomics (SING)

SING Canada 2022 was held July 11-15 at the University of Alberta. Its focus was decolonizing genomics through a critical Indigenous lens. We are excited to announce that SING Canada 2023 will again take place July 15-21, again in Edmonton, Alberta. This year's theme is *#LandBack: Indigenous Peoples, Soil Science and Disruptive Sequencing Technologies*. For further information, please check out [SING Canada](#) or contact the Silent Genomes Project.



*SING staff & students in front of the Native Studies building, July 2022*

### Silent Genomes Indigenous Rare Disease Diagnosis Steering Committee (S-GIRDD)

A 2-day in-person meeting with the S-GIRDD was held in September. The purpose was two-fold: to consider the process of transforming the S-GIRDD steering committee into a governance entity, and to draft a process to review manuscripts for those writing papers using sequencing data from the SGP. It was the first in-person event for many attending and it was inspiring to get a chance to work together and move the above issues, and others forward.



*Dinner at Salmon n' Bannock in Vancouver, BC, with the Silent Genomes team and the S-GIRDD Steering Committee, September 2022.*

## Activity 2

Activity 2 "Precision Diagnosis Study" is now closed for recruitment. While Activity 2 is wrapping up in 2023, the team has been working hard to finish the analysis and return diagnoses and findings to patients, families, and clinicians.

To date, 91 Indigenous patients or families, recruited from 10 different sites across the country, have undergone sequencing through our study. First tier (or "Level 1") genomic analysis has been completed and results returned to 69 families so far. An additional four families have received results after further second tier (or "Level 2") genomic analysis.

We are looking forward to the completion of IBVL which will help us better interpret approximately 2/3 of the variants reported back to Indigenous families in Activity 2, which are currently considered to be uncertain or research candidate findings.

We would like to thank everyone who worked as part of our National Clinical Network of recruitment sites for their hard work these past few years.

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## Activity 3

### The Alliance

The Canadian Alliance for Healthy Hearts and Minds (the Alliance) is made up of seven First Nations communities across Canada who participated in research to address cardiovascular disease and cancer. They collected biological samples and health data for storage at the PHRI in Hamilton for future research important to their own communities. Dr. Anand and the Alliance team are the stewards of the samples and data, which are owned by each community. After many years of consultation, sample transfer has been approved by four communities to send a small amount of their stored DNA samples for sequencing to build the Indigenous Background Variant Library. Additional Alliance communities are also considering participation in the IBVL.



*DNA samples are frozen for storage at the Public Health Research Institute (PHRI) in Hamilton, ON*

### Building Connections

Dr. Wyeth Wasserman, the lead for Activity 3, is currently spending 8 months in Aotearoa (New Zealand), working with *the Variome Project*. This project is the Maori equivalent to the Silent Genomes Project, where Maori People are making their own Indigenous Background Variant Library called *the Variome*. While in New Zealand, Dr. Wasserman is learning about the similarities and differences between the two projects, and looking for ways to improve both background variant libraries and create long-lasting partnerships for the future.



*The Public Health Research Institute (PHRI) in Hamilton, ON. This site contains the biobank of the Alliance samples.*

## Activity 4

During February and March of 2022, Activity 4 held five focus groups and one interview with 24 Indigenous participants. Discussions were held over Zoom with adults living in urban settings around British Columbia to document their preferences and values around the use of genomic testing to diagnose children with suspected genetic conditions. The analysis of focus group data is underway, guided by the Activity 4 Research Advisory Council. Member checking is also ongoing, in which Activity 4's preliminary findings are returned to participants for their review and feedback. We presented our progress at the Canadian Network for Learning Healthcare Systems and Cost-Effective 'Omics Innovation (CLEO)-Sulston Joint Workshop in Vancouver, BC in August of 2022.

*Laurie Kariosta Montour, Dr. Dean Regier, and Morgan Ehman are giving a talk about their focus group progress at the international CLEO-Sulston Joint Workshop in Vancouver, BC in August of 2022.*

