BACKGROUNDER

BC led research to transform early disease detection, treatment and health equity using cutting edge genomic technologies

AI-powered genome sequencing data holds the key to more precise, preventative and cost-effective health care — and Canada's competitive edge in health innovation.

Genome Canada's Canadian Precision Health Initiative (CPHI) aims to gather, share and make genomic data from 100,000+ Canadians accessible to accelerate research, foster innovation and improve health.

The initiative will:

- Deliver the AI-powered, high-quality, large-scale genomic datasets needed to firmly establish Canada as a global leader in precision health, life-sciences innovation and clinical trials
- Transform health care and fuel economic growth in a highly competitive, high-impact sector—enabling scientists and companies to develop more precise molecular diagnostics, tailor-made therapeutics and treatments
- Play a vital role in strengthening Canadian health security, increasing access to and utility of genomic data to fight major public health threats

The five BC-related projects are:

Enhanced Population Cancer Care through Mainstream Genome Sequencing and Parent-of-Origin Detection

Project Leaders: Kasmintan Schrader, Peter Lansdorp, Steven Jones (BC Cancer)
Institution(s): BC Cancer
Genome Centre: Genome British Columbia

This project will improve hereditary cancer care by expanding access to research participation and providing parent-of-origin-aware genetic risk assessments for individuals with pathogenic variants. Knowing which side of the family a variant came from will guide clinical outreach and help identify at-risk family members across Canada, enabling earlier detection of hereditary cancer risk and more timely, personalized care.

Fewer than 10% of Canadians with an inherited cancer risk have been identified. Many eligible patients are not referred for testing, despite increasing clinical need. There is a limited genetics workforce capacity. Furthermore, not enough family members undergo follow up testing when a hereditary cancer risk is discovered.

BC and the Yukon have addressed these challenges with streamilined approaches, including "mainstreamed" genetic testing with a universal multi-gene panel offered by non-genetics providers, centralized review by a lab-based genetic counselling team, and a digital portal to speed

access to testing within the Hereditary Cancer Program. Building on these advances, BC Cancer is introducing clinical genetic testing using a whole genome sequencing (WGS) platform.

This project will allow patients undergoing routine cancer genetic testing to consent to contribute their WGS data to the Pan-Canadian Genome Library, enriching it with diverse, clinically relevant genomes. It will also implement Parent-of-Origin-Aware genomic analysis (POAga) to determine whether a variant was inherited from the mother or father, improving real-time family-based testing and helping over 1000 families better identify at-risk relatives sooner.

By expanding equitable access to state-of-the-art genomics research, this project will improve how hereditary cancer risk is identified in real-time, enabling more Canadians and their families to immediately benefit from timely testing, screening and prevention, while advancing Canada's leadership in genomic medicine.

Longitudinal, Deep-Phenotyped Pediatric Databank of Medical and Drug Therapy Outcomes

Project Leaders: Bruce Carleton (University of British Columbia), Michael Rieder (University of Western Ontario), Maja Krajinovic (Université de Montréal)
Institution(s): University of British Columbia, University of Western Ontario, Université de Montréal
Genome Centres: Genome British Columbia, Genome Alberta, Génome Québec, Ontario Genomics

This project will improve drug safety and effectiveness for children by expanding access to genomic data on how they respond to medications, helping researchers and health agencies reduce harmful drug reactions.

Many children experience serious side effects from medications, but because pediatric diseases like cancer are rare, it is difficult to gather enough data to understand why. Over the past 20 years, the Canadian Pharmacogenomics Network for Drug Safety (CPNDS) has collected DNA and medical data from more than 12,350 patients, tracking their responses to over 100,000 medication uses and 10,000 severe adverse drug reactions. Some patients in this database have more than 40 years of medical history. This project will build on that work by adding a pediatric component to the Pan-Canadian Genomics Library (PCGL)—a national resource that makes genomic and clinical data accessible for research.

The project will reconnect with previously enrolled patients to obtain their consent to add de-identified genomic and clinical data to the PCGL and continue recruiting new patients at 10 existing study sites across Canada. A key focus will be increasing diversity and accessibility to ensure the database better represents all populations. Researchers will also upgrade the data from genome-wide typing (GWAS) to whole genome sequencing (WGS), allowing for a more detailed investigation of drug-related harm.

With these expanded resources, the team will use genomic data to identify biomarkers that predict harmful drug reactions in children with cancer. The insights gained will improve how medications are prescribed, making treatments safer and more effective for children in Canada and beyond.

MOSAIC — Multi-Omics and Sonography to Advance Indigenous and Community Cardiovascular Health

Project Leaders: Teresa Tsang, Anna Lehman, Anna Meredith, Gabrielle Legault, Purang Abolmaesumi (University of British Columbia), Anurag Singh (University of Northern British Columbia)
Institution(s): University of British Columbia, University of Northern British Columbia
Genome Centre: Genome British Columbia

This project will improve the early diagnosis and treatment of cardiovascular disease (CVD), particularly for underserved Indigenous and rural communities, by combining advanced genetic research, cardiac imaging, and artificial intelligence.

Cardiovascular disease remains one of the leading causes of death in Canada, but current methods for predicting risk often fail to account for diverse populations. Many genetic studies focus on people of European ancestry, meaning they do not fully reflect the genetic and environmental factors affecting Indigenous and other underrepresented groups. At the same time, people in rural and remote areas often lack access to heart specialists, diagnostic imaging, and early screening, leading to later-stage diagnoses and poorer health outcomes.

The MOSAIC Initiative will help close these gaps by creating a more inclusive approach to assessing cardiovascular risk. The project will collect genetic, clinical, and lifestyle data from a diverse group of 8,000 participants across British Columbia and Yukon. It will also improve access to diagnostic imaging by using point-of-care ultrasound (POCUS) in under-resourced areas, allowing healthcare providers to conduct heart assessments closer to home. Advanced artificial intelligence (AI) models will be developed to analyze genetic and imaging data, helping doctors make faster, more accurate decisions about a patient's heart health.

By working with all regional health authorities in BC, Indigenous representatives, rural physicians, and government stakeholders, this project will ensure that personalized, genomics-based cardiac care is more equitable and accessible. The knowledge gained from this study will help create a scalable model for cardiovascular risk assessment, improving heart health for all Canadians.

*Pan-Canadian Initiative Linking Genomic, Environmental and Mental Health Data in Children and Youth (Pan-GEM)

Project Leaders: Paul Arnold, Chad Bousman, Gina Dimitropoulos, Jennifer Zwicker (University of Calgary)
Institution(s): University of Calgary
Genome Centres: Genome Alberta, Genome British Columbia

This project will help improve early diagnosis and treatment of mental health disorders in children and youth by identifying genetic and environmental risk factors that contribute to these conditions.

Mental health disorders (MHDs) often begin early in life, with 75% of cases emerging before age 25. In Canada, about 1 in 5 children and youth—or 1.6 million young people—are affected. While mental health disorders often run in families, there are still significant gaps in understanding the genetic and environmental factors that contribute to them. Current research is limited by small

sample sizes, inconsistent data collection methods, and a lack of representation from diverse populations, which can lead to inequities in care.

The Pan-GEM Initiative will address these challenges by creating Canada's largest research collaboration focused on the genetics of youth mental health. The project will sequence the genomes of 5,250 children and youth, with an additional 1,200 long-read genome sequences if funding allows. Researchers will also collect environmental and social health data to explore how genetics and life experiences interact to influence mental health. Special attention will be given to recruiting participants from diverse backgrounds to ensure the research benefits all communities.

By bringing together experts in psychiatric genetics, data science, health policy, and clinical care, Pan-GEM will develop new tools to analyze, standardize, and share mental health data. The team will also work with Indigenous and community partners to develop culturally appropriate recruitment, consent, and knowledge-sharing strategies. The insights gained from this project will help shape new guidelines and policies that ensure genomic research is applied ethically and effectively in mental health care.

By removing barriers to research and ensuring diverse representation, Pan-GEM will help advance precision medicine, leading to more accurate diagnoses and better, more personalized treatments for young Canadians with mental health disorders.

*PrairieGen: A Multi-Omics Approach to Advancing Data Integration from Manitoba and Saskatchewan Populations into the Pan-Canadian Genome Library

Project Leaders: Cheryl Rockman-Greenberg, Athanasios Zovoilis (University of Manitoba), Spencer Zwarych (University of Saskatchewan), Donna Turner (CancerCare Manitoba)
Institution(s): University of Manitoba, University of Saskatchewan, CancerCare Manitoba
Genome Centre: Genome Prairie, Genome British Columbia

This project will map the unique genetic makeup of populations in Manitoba and Saskatchewan to improve healthcare efficiency, lower costs, and enhance patient outcomes in the Prairie provinces.

Many existing genomic studies in Canada have focused on larger urban populations, leaving gaps in knowledge about the genetic diversity of Manitoba and Saskatchewan. These gaps make it harder to develop personalized treatments and policies that address the specific health needs of people in the Prairies. PrairieGen will fill this gap by studying the genetics of people from Manitoba and Saskatchewan, linking multiple sources of health and genomic data to build a more complete picture of how genetic and environmental factors contribute to disease.

This project will integrate genetic and health data from multiple disease cohorts, including patients with multiple sclerosis, schizophrenia, cancer, inflammatory bowel disease, and metabolic disorders, as well as participants in long-term health studies like the Manitoba Tomorrow Project. Using advanced genomic techniques, researchers will analyze whole genome sequencing (WGS), DNA methylation, RNA sequencing, and protein/metabolite data to uncover key genetic markers and risk factors unique to these populations.

By building a secure, single-source genomic database, PrairieGen will help guide public healthcare investments and policy decisions in Manitoba, Saskatchewan, and beyond. The project will also refine standards for data governance, privacy protection, and patient consent, ensuring responsible use of genetic information.

By strengthening research capacity in the Prairie provinces, this initiative will empower local clinicians and policymakers to develop precision medicine approaches tailored to regional populations, ultimately improving healthcare delivery and patient outcomes in Manitoba and Saskatchewan.

**15.25% of the total budget for these projects is for activities within British Columbia, primarily through Canada's Michael Smith Genome Sciences Centre at BC Cancer (GSC).*