Canadian Precision Health Initiative

Funding opportunity

Overview

BRINGING TOMORROW'S HEALTH CARE TO CANADIANS TODAY

The future of health is personal and tailored—responsive to each individual's history, environment, genetic makeup and molecular reality. What was once in the realm of science fiction is increasingly part of our reality, with precise molecular diagnostics, customized therapeutics and treatments that are as unique as each of us. This revolution in health and medicine is helping us tackle chronic diseases, rare conditions and global pandemics with new tools that are getting more powerful every day.

Driving all these advances is our ability to combine and compare our individual data with large-scale, relevant population data. Underpinning and driving this data is genomics,[1] the molecular data that shapes who we are and how similar we are (or not) to those around us.

Large-scale genomic health datasets can:

- Yield data-driven insights that tangibly improve health-care delivery and advance research and development.
- Identify genetic predispositions to diseases, enabling proactive and preventive health care.
- Facilitate personalized medicine, ensuring treatments are tailored to individual genetic profiles.
- Enhance the development of new drugs and therapies by identifying novel genetic targets.
- Support public health responses by tracking the spread and evolution of infectious diseases.
- Drive innovation in the biotechnology and health-care industries, fostering economic growth and job creation.
- Improve health outcomes through more accurate and timely interventions.

[1] Genome Canada defines genomics as the comprehensive study, using high-throughput technologies, of the genetic information of a cell or organism and its functions. This study is understood to include work done in related disciplines, such as epigenomics, metabolomics, metagenomics, proteomics, transcriptomics, bioinformatics and synthetic biology, as long as the link to genetic information is clear.

- Promote equity in health care and provide a deeper understanding of the nature of health by ensuring that the benefits of genomics and precision health are accessible to all.
- Foster a more efficient health-care system by reducing trial and error in treatment and minimizing adverse drug reactions.

Integrating genomic data with clinical information, environmental factors and artificial intelligence (AI)-driven analysis will enhance the accuracy, affordability and clinical utility of precision health approaches. This enhancement will not only improve treatment efficacy, but also help prevent diseases by identifying the risk factors specific to an individual's genetic makeup and enabling more proactive, predictive health care. Ultimately, the result will be better outcomes for patients and more efficient use of resources.

Genomics-driven precision health is a multifaceted, multimodal approach that hinges on a number of key elements, including:

- The development of large-scale, diverse, representative, interoperable datasets to ensure all Canadians can receive high-quality, personalized health care.
- Robust infrastructure to support the generation, integration and utilization of this data so it can be used to its full potential in patient care.
- Effective policies and a regulatory framework for the adoption of new genomic tools and technologies that effectively balance the need for privacy, ethical governance and security with the imperative for widespread access to knowledge.
- Responsible approaches that support innovation in the application of large-scale datasets—including by the private sector—for improved diagnostics, therapeutics and analytics.
- Sufficient capacity, capabilities and resources, including a pool of skilled professionals with the expertise to interpret and apply genomic data effectively.
- An interdisciplinary, collaborative culture across fields such as bioinformatics, AI, clinical medicine and public health to drive holistic solutions.
- Public engagement and education to increase understanding, acceptance and informed consent among Canadians.
- Equity and accessibility to ensure access to genomic health care across all demographics, including Indigenous, under-served and in rural communities.
- Global collaboration to share knowledge, data and best practices to advance the field.
- Continuous monitoring and evaluation for ongoing assessment of effectiveness, impact and opportunities for improvement.

Many of these elements are underway across Canada but are fragmented and disconnected across provincial and national health-care and research systems. A co-ordinated, large-scale initiative is needed to bring these pieces together, fill gaps, and build on existing initiatives to realize the potential for transformative change.

Genome Canada, alongside its regional, provincial and national partners, is embarking on the most ambitious genomic health research program in Canada’s history. Together, we aim

to gather, share and make accessible genomic data from 100,000 or more people in Canada to accelerate research, foster innovation and improve health care. Building a diverse data resource that researchers can easily access—while keeping personal information private—will help transform our one-size-fits-all health-care approach into a modern, precision health system that can have enormous impacts.

The Canadian Precision Health Initiative is expected include an investment of $200 million from Genome Canada and its partners. The initiative comprises four integrated, connected pillars of activity:

1. **Generating population-level genomic data**
    - This pillar will focus on the creation of national data assets that are accessible and reflect the diversity of Canada's population. The goal is to enable further research and innovation.
2. **Mobilizing and advancing the utility of genomics data**
    - This pillar will involve working with key end users, including academics and businesses, to ensure the data assets being developed meet their needs and can be used to advance research, develop new AI-powered tools, and create new genomics technologies to improve health outcomes for Canadians. Investments in genomics research and innovation will also be made to maximize impact and enhance the utility and accessibility of the data assets.
3. **Applying data governance and GE^{3}LS^2**
    - This pillar will build on a foundation of ethical, environmental, economic, legal and social principles (GE^{3}LS) to guide the stewardship and use of genomic data.
4. **Assembling an alliance of Canadian partners for genomics in health**
    - To ensure the first three pillars achieve their goals, Genome Canada will assemble an alliance aimed at fostering collaboration and coherence among the various partners and stakeholders across the ecosystem. The alliance will work to align and integrate efforts across other national, regional and provincial initiatives to ensure a cohesive approach. Genome Canada may call on alliance members for advice and guidance throughout the Canadian Precision Health Initiative.

Genome Canada is looking forward to working with diverse partners in funding a portfolio of genomics research and innovation projects that deliver solutions to advance precision health for all Canadians. The initiative will be deployed in two phases. Phase I will focus on establishing an alliance of partners and generating population-level genomic data. Phase II will focus on mobilizing and advancing the utility of genomics data as well as data governance and GE^{3}LS.

[2] The acronym GE^{3}LS stands for genomics and its ethical, environmental, economic, legal and social aspects. However, it should be understood broadly as research into the implications of genomics in society from the perspectives of the social sciences and humanities. Therefore, it is not limited to disciplines that make up the acronym. Rather, it encompasses all those that rely on quantitative and qualitative methodologies to investigate the implications of genomics in society and inform applications, practices and policies.

Objectives

The initiative's key objectives and the steps needed to achieve them are listed here.

1. **Generate a co-ordinated, large-scale, diverse genomic data asset that reflects Canada's population**
    - Develop a ready-to-use data asset of 100,000 genomes to drive genomics-enabled precision health in Canada and address imbalances in existing genomic datasets.
    - Establish standardized protocols for data collection, processing, analysis and governance to ensure reliability and ethical compliance in genomic research.
    - Support and coordinate the development of a trusted research environment for research and innovation using the genomic data asset.
2. **Advance research outcomes and clinical impact for patients**
    - Develop world-leading genomics tools and solutions to detect and diagnose diseases and support the identification of therapeutic targets.
    - Improve patient stratification using genetic profiles derived from a diverse Canadian genomic dataset, accelerating translational research from bench to bedside.
    - Position Canada as a global leader in the application of AI to genomics, fostering advancements in both AI and genomic research.
3. **Accelerate the application and uptake of genomic data**
    - Promote responsible data-sharing and the interoperability of genomic data across Canadian health systems and institutions by engaging with policy and regulatory stakeholders.
    - Collaborate with federal, provincial, territorial and Indigenous health authorities to bring genomic solutions to the clinic to improve health outcomes for all Canadians.
    - Understand the societal and economic impacts of integrating genomic data into Canada's health systems.
4. **Build and coordinate a community to inform precision health in Canada**
    - Establish an alliance of partners to coordinate strategies and health investments across Canada toward shared goals and objectives.
    - Facilitate public engagement and outreach to increase Canadians' understanding and acceptance of genomics and precision health.
    - Uphold Indigenous rights and interests in data governance, ensuring respect for data sovereignty and promoting equitable access and benefits for Indigenous communities in precision health.

Governance

Genome Canada has the ultimate responsibility for the stewardship and success of the Canadian Precision Health Initiative. To manage its governance effectively, Genome Canada will undertake the following tasks:

- Establish an alliance of Canadian partners for genomics in health. This alliance will comprise several "tables" that will provide advice and guidance and serve a coordinating function with other national, regional and provincial initiatives. The tables will include funders, researchers, industry partners and users.
- Work with the regional Genome Centres to understand and reflect provincial priorities, secure co-funding, and develop and manage individual projects, as appropriate.
- Establish a Governance and Review Committee (GRC) composed of national and international experts and leaders who will provide recommendations and advice to Genome Canada to support the execution of the initiative **at a portfolio level**. To ensure the projects collectively deliver on their portfolio-level objectives, the GRC will recommend projects for funding and evaluate their progress against milestones and deliverables. It will also advise Genome Canada, the Genome Centres and other partners on the management of projects. The GRC will advise Genome Canada, where appropriate, to re-profile funding across the portfolio and set conditions that must be met by funded projects. The committee may recommend to reallocate and/or redistribute funding to other projects if progress is severely hampered for any reason. Genome Canada's Board of Directors will retain responsibility for all decisions regarding the allocation of funds administered by Genome Canada on behalf of the Government of Canada.
- Engage sub-committees of the GRC as needed to allow for focused and detailed discussions (e.g., on data solutions, ethics, diversity, end users), leveraging the expertise of GRC members and additional external experts.
- Work (through the alliance of Canadian partners for genomics in health) with other national funders that have existing or projected investments in genomic data, including the Canadian Institutes of Health Research, the Canada Foundation for Innovation, the Digital Research Alliance of Canada, and the Terry Fox Research Institute/Marathon of Hope.
- Work (in collaboration with the regional Genome Centres) with organizations and initiatives that align with the initiative's broader objectives to improve the representation of under-represented groups and Indigenous communities in Genome Canada datasets.
- Ensure (in collaboration with the regional Genome Centres) that the initiative complies with the terms of the Genome Canada agreement with the federal government and provides information and data that will allow for the ongoing assessment of progress, including performance metrics and financial reporting.
- Work with the Global Alliance for Genomics and Health, GA4GH, and international partners to share best practices and co-operate on global data-sharing.

Additional committees or working groups may be established to provide advice and guidance on key initiative activities. These groups could comprise members of the project teams as well as external advisors with the required expertise.

AN ECOSYSTEM APPROACH

The initiative will intentionally create a portfolio of projects that synergize the deliverables of individual projects toward broader national impacts and generate value beyond the deliverables and outcomes of each individual project. This crafted portfolio will accelerate the use and uptake of precision health in Canada by connecting to other initiatives in the ecosystem, allowing better coordination of health data. The portfolio must collectively represent Canada's diverse demographic and ethno-racial makeup and contribute to the development of equitable precision health tools, technologies and solutions.

Pillar 1 – Generating population-level genomic data

The goal of Genome Canada's planned investment in Pillar 1 of the initiative is to sequence the genomes of at least 100,000 Canadians, reflecting Canada's unique and diverse population. This will provide the rich dataset needed to advance research, drive innovation and improve health-care outcomes for all Canadians.

AVAILABLE FUNDING AND TERM

Whole-genome sequencing data generation using short-read technology:

- There is approximately $60 million investment available from Genome Canada for short-read data generation using Illumina technology.
- Genome Canada's contribution to an approved project can be up to $6 million.
- A project's eligible costs must be co-funded from eligible sources such that the co-funding is at least equal to the Genome Canada contribution. See the Genome Canada **Guidelines for Funding** for more details.
- Successful teams must have completed all project activities by March 31, 2029. The final report (including financial reconciliation) must be provided by June 30, 2029.

Funding for the generation of long-read genome sequencing is also available to enhance the utility of the dataset generated. Please note that this additional funding will be available only to projects that are approved for funding for short-read, whole-genome sequencing data generation.

Whole-genome sequencing data generation using long-read technologies:

TWO OPTIONS ARE AVAILABLE FOR THE GENERATION OF ADDITIONAL LONG-READ WHOLE GENOME SEQUENCING DATA:

Option 1: Long-read genome sequencing using Oxford Nanopore Technologies

- Oxford Nanopore Technologies must be used to generate additional long-read genome sequences.
- Approximately $10 million is available from Genome Canada.
- Projects can apply for any amount up to $10 million.
- The costs associated with this option must be co-funded from eligible sources such that the co-funding is at least equal to the Genome Canada contribution.

Option 2: Long-read genome sequencing using PacBio Technology

- PacBio technology must be used to generate additional long-read genome sequences.
- Approximately $10 million is available from Genome Canada.
- Projects can apply for any amount up to $10 million.
- Costs associated with this option must be co-funded from eligible sources such that the co-funding is at least equal to the Genome Canada contribution.

FUNDING ALLOCATION AND RELEASE

- It is expected that at least 45 per cent of the total project budget will be used to cover costs associated with generating short-read genome sequencing data through Genome Canada–approved sequencing centres (SCs). A list of approved centres will be published on Genome Canada's website at the time of launch.
- Funding for each project will be distributed primarily through the six regional **Genome Centres**.

In addition, note that the release of funding to each project will be conditional on adherence to planned project deliverables, including the deposition of genome sequences and associated metadata with appropriate consent into a national databank through the Pan-Canadian Genome Library (PCGL). Only projects that have received approval from their research ethics boards to collect data will be able to send their data to PCGL. Participants must have consented to:

- Sharing genetic and clinical data nationally and internationally.
- Making aggregate data discoverable through search engines nationally and internationally.

- Having future health research conducted responsibly on their data, including by industry researchers.

Please note that: There is no established limit on how long project data can be stored. The data will be stored in the PCGL, which is hosted in a secure environment. The library will implement open, registered and controlled access. Requests for controlled access data will go through the library's Data Access Committee Office (DACO), after which they will be forwarded to its Data Access Committee (DAC) for adjudication (whether through a central PCGL committee or in coordination with other DACs, where necessary).

ELIGIBILITY CRITERIA AND PORTFOLIO CONSIDERATIONS

Project eligibility criteria

- Each funded project must generate a minimum of 2,000 genomes. The GRC may consider projects with a smaller number in specific circumstances where need and statistical power at the project level can be justified.
- Whole-genome sequencing must be completed by a Genome Canada–approved SC. Material transfer agreements must be in place before samples can be sent to an SC. Project teams must be able to operate and execute the sequencing program that allows the validation of deposited whole-genome sequences into the national databank to provide payments.
- Projects must include self-identified race, ethnicity and ancestry data in their depositions and are required to report and quantitatively characterize this data. Additionally, the methodology used for collecting and analyzing this data must be clearly documented in the project protocols.
- Research Ethics Board approval must be obtained and shared with Genome Canada before biological samples are shared with the SC.

Project participant inclusion criteria

- Each participant must provide a biological sample and their consent (or their guardian's consent) for the performance of whole-genome sequencing and the use of the results in future research.
- All core elements of the consent form provided in Appendix 1 must be included in the consent provided by participants. An arms-length third party will review and validate the consent forms used by the projects as part of its review process.
- Participants must provide the following permissions:
    - permission to link with and update data from health records and registries
    - permission to deposit data in a cloud-based, access-controlled databank and to share data with academic and industry researchers
    - permission to contact and obtain assent from study participant (when appropriate)

- Projects must be able to share the minimum required clinical data for all sequenced participants (see Appendix 2: Case report form sample).

Project participant exclusion criteria

- Consent was not obtained.
- Consent does not include the participant's agreement to have whole-genome sequencing performed and their data banked, or to be re-contacted for future research, and/or participant does not approve access to their data.
- The individual has already enrolled in the study and is included in the national databank. Minimum clinical data (as per Appendix 2) cannot be obtained.

Sample requirements

- All samples collected for sequencing must originate from within Canada.
- Samples are expected to nominally conform to standard sample quality parameters, as required by the approved SCs.

Data requirements

- Whole-genome sequencing data and associated minimal metadata (see Appendix 2) are to be made available for deposition to the national databank for curation and stewardship, as outlined by Genome Canada. Only de-identified metadata will be submitted.
- Genome sequencing data and metadata must conform, to the degree possible, to international standards—such as the Global Alliance for Genomics and Health—and to best practices for sequence data generation, description and reporting.
- Each project leader must comply with the applicable laws and regulations with respect to protecting privacy and access to information.
- Sequence data and associated metadata residing in the national databank will be stored in a dedicated Canadian cloud that conforms to all national laws and policies governing privacy laws. Access will be provided through application to—and permission will be provided by—a dedicated Data Access Compliance Office.
- Projects must submit plans that explain how they will manage, preserve and share the scientific data and resources generated by the project, if applicable. These plans must comply with Genome Canada's **Data Release and Sharing Policies**. Revisions to teams' data plans may be required to further align with the portfolio of funded projects.

Note: Applicants are encouraged to include additional metadata and multi-omics data that may substantially enhance Canada's ability to perform downstream research activities focused on advancing the utility of genomic data and accelerating the development of diagnostics and new therapeutics. Genome Canada supports the harmonization, storage and stewardship of such enhanced metadata, consistent with Canada's privacy laws and other relevant international regulations and best practices related to managing identifiable information.

The inclusion of additional sequencing approaches (e.g., long-read sequencing) and/or multi-omic datasets is encouraged to enrich the dataset generated.

Applicants are also encouraged to include explicit objectives and metrics (if applicable) to support the creation of an ancestrally and demographically diverse dataset that reflects the diversity of the Canadian population.

Portfolio considerations for Pillar 1

The GRC will use a portfolio approach to review both the letter of intent (LOI) and full proposal to recommend project funding.

The GRC will prioritize projects for funding and inclusion in a portfolio based on the following criteria:

- The population cohort of the national databank is reflective of the diversity of the Canadian population.
- The portfolio includes a combination of disease-specific and targeted cohorts, large population and hypothesis-free cohorts, and cohorts of under-represented groups, including Indigenous populations, to create a large, diverse genomic dataset.
- The resulting national databank has the potential to advance precision health and address the pressing health needs of unique regional populations and priorities.
- The national databank has the potential to advance the utility of genomic data for industry and other stakeholders, increasing the impact of the research through follow-up activities, such as translation, implementation and commercialization.
- The portfolio should encompass partners and project teams that embody Canada's diversity and uphold Genome Canada's values regarding inclusion, diversity, equity, and accessibility (IDEA), as well as its commitment to fulfill the **Truth and Reconciliation Commission's Calls to Action** and adhere to the **United Nations Declaration on the Rights of Indigenous Peoples**.

The portfolio will prioritize projects that make a clear and compelling case for creating short-term impact that can benefit Canadians' health.

Given the portfolio considerations listed above, we encourage the regional Genome Centres and their applicants to consult with each other and with Genome Canada to maximize potential synergies, minimize duplication, and overlap and maximize potential impact and benefits to Canada.

IDEA and Indigenous truth, reconciliation and engagement

Genome Canada is committed to creating a diverse and inclusive environment and ensuring equitable participation by people who live with diverse visual, motor, auditory, learning and cognitive abilities. We are acting on the evidence that achieving a more equitable, diverse and inclusive Canadian research enterprise is essential to creating the excellent, innovative and impactful research needed to advance knowledge and understanding and respond to local, national and global challenges.

Genome Canada encourages partners and applicants to increase the inclusion and advancement of equity-deserving and under-represented communities in leadership positions to enhance excellence in research and training. These communities can include

Indigenous Peoples, people of African descent, members of other racialized groups, women, persons with disabilities, members of 2SLGBTQ+ communities, and early-career researchers. IDEA should be key considerations for team management and composition.

Genome Canada is committed to Indigenous truth, reconciliation and engagement and the right of self-determination as set out in the United Nations Declaration on the Rights of Indigenous Peoples. This commitment is reflected in our support for Indigenous data governance principles that are people- and purpose-oriented and that recognize the crucial role of data in advancing Indigenous innovation and self-determination.

Historically, research done with Indigenous communities, on Indigenous lands, and/or incorporating Indigenous knowledge has been under-recognized, under-valued, under-funded and often conducted in a culturally insensitive manner. Genome Canada recognizes that Indigenous communities have unique approaches to research that are rooted in their unique experiences and relationships with the natural world.

Research projects that address issues of relevance to Indigenous Peoples are expected to include a plan to engage Indigenous Peoples, including First Nations, Métis and Inuit, in research design and practice. The plan should specify how Indigenous groups will participate on the research team and/or as users of the research, as well as how Indigenous knowledge systems will co-exist with and complement the project's other activities. Projects involving Indigenous research should be conducted with sensitivity and only after carefully considering who will conduct the research and why and how. The research should be conducted in line with **the Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans Chapter 9: Research Involving the First Nations, Inuit and Métis Peoples of Canada,** and **the First Nations principles of ownership, control, access and possession (OCAP®).**

TIMELINE

End of July 2024	Genome Canada shares draft guidelines with Genome Centre partners.
August 2024	Genome Canada holds coordination meeting(s) with partners in the initiative.
Early October 2024	Genome Canada launches Pillar 1: Generating population-level genomic data.
By November 01, 2024	Applicants submit LOIs.
Week of November 04, 2024	Genome Canada completes eligibility checks.
Week of November 11, 2024	Genome Canada's GRC meets to discuss the eligibility of the submitted LOIs and complete a portfolio review.
November 15, 2024	Genome Canada notifies applicants of the results of the LOI.

December 15, 2024	This is the first deadline for applicants to submit their full proposal.*
January 2025	Independent experts conduct technical reviews of each project.
Mid-January 2025	The GRC meets to complete a portfolio review based on the proposal submissions.
End of January 2025	Genome Canada informs regional Genome Centres, partners and the broader community of funding decisions made by the GRC. With help from the GRC, Genome Canada works to identify gaps in the portfolio and, where possible, add projects to the portfolio to fill any identified gaps (with remaining funds).
March 15, 2025	This is the second deadline for applicants to submit their full proposal.* A quarterly review by GRC and notice of decisions will follow in April 2025.
Starting June 1, 2025	If there are unallocated funds or opportunities to strengthen the portfolio, Genome Canada will welcome additional applications.

* Full proposals will be accepted only from applicants who submit an LOI before the November 1, 2024 deadline.

APPLICATION PROCESS

Genome Canada requires applicants to submit both an LOI and, later in the process, a full proposal. Applicants are required to apply through Genome Canada's Proposal Central Portal, usually through a regional Genome Centre.

Letter of intent

Each applicant will use a brief LOI to indicate their interest in applying for Genome Canada funding under Pillar 1 of the initiative. The LOI will enable an eligibility check by Genome Canada to ensure the LOI meets the requirements listed in the "Eligibility criteria and portfolio considerations" section of this document. LOIs will also undergo an initial selection process by the GRC based on portfolio considerations. Only LOIs that show potential for inclusion will be invited to submit a full proposal.

In the LOI**,** applicants will be required to briefly describe:

- The proposed project plan.
- The expected deliverables.

- The planned number of genomes to be sequenced per annum.
- How the genomic data generated will represent Canada's diversity and/or address significant population-level gaps in the country's reference genomic library.
- The potential clinical impacts and benefits to Canada's health-care systems.

Projects must also confirm that they meet the eligibility criteria of the population-level genomic data generation pillar. If they do not, they must provide an overview of how they intend to meet the criteria by the start date of the project.

Applicants must also provide a summary of their budget and co-funding plan.

Full proposal

Applicants who succeed at the LOI stage will be asked to submit a full proposal. Full proposals must address the evaluation criteria for individual projects established for the funding opportunity (see Appendix 3).

A technical review of the proposals received will be completed by a small panel of international, diverse and independent experts. It is expected that the technical review will be completed as an independent, at-home review.

All full proposals will then be reviewed and discussed by the GRC. The technical reviews will be used, in part, to assess whether the applications meet the initiative's objectives and will add value to the portfolio of projects and contribute to the expected outcomes. The GRC will include broad expertise in health research and innovation, portfolio design and management, health policies, GE^{3}LS, and the management of large-scale data assets.

The GRC will use the approved applications to monitor progress and provide advice and guidance to partners and project teams to help achieve the objectives of the initiative.

These evaluation processes may be adjusted, where warranted, by the complexity or number of applications received or by other relevant factors. Any changes will be communicated through Genome Canada's website.

PROJECT MANAGEMENT AND OVERSIGHT

All funded projects must adhere to Genome Canada's **Guidelines for Funding.**

All funded projects must meet the conditions of funding by the following dates:

- Full proposals submitted by the first deadline on December 16, 2024, must meet all conditions of funding by March 21, 2025.
- Full proposals submitted by the second deadline on March 15, 2025, must meet all conditions of funding by July 1, 2025.

Each project team must have human and financial resources allocated to the budget to successfully manage the funded project and help ensure coordination across the portfolio, to support portfolio cross-cutting activities, and to inform and develop portfolio-relevant activities.

Each project must have an individual responsible for data and metadata deposition in the national databank as well as follow-up work. This individual can be a project manager or other individual. It is imperative that projects can rapidly deposit their contributions to the databank to confirm that deliverables are being met and in order for projects to access ongoing project funding.

Project reporting will take the form of regular progress reports as well as a final report following project completion. Requests for no-cost extensions will not be considered.

The GRC will ensure cohesive and co-ordinated oversight of the projects within the portfolio.

Mechanisms will also be put into place to ensure that partners and project teams working on funded projects in the portfolio regularly connect, convene and learn from each other and that the group is collectively enabled to support the goals of the initiative.

PILLARS 2 AND 3: MOBILIZING AND ADVANCING THE UTILITY OF GENOMIC DATA AND APPLYING DATA GOVERNANCE AND GE^{3}LS

The initiative will leverage the diverse genomic datasets generated as part of Pillar 1 by fostering collaborations and partnerships between researchers, end users and industry partners. The focusing will be on bridging the gap between genomic data collection and real-world application, ensuring that the insights gained translate into improved health outcomes for Canadians.

By advancing the utility and application of genomic data and the national databank, Genome Canada aims to catalyze the creation of new genomics tools and technologies that can diagnose, treat and prevent diseases more effectively. This will not only enhance Canada's position as a leader in genomic research, but also stimulate economic growth. Pillar 2 of the initiative will continue to build on the successes of the genomic data generated in Pillar 1 and foster an environment where innovation can thrive.

To mobilize and advance the utility of genomic data, Genome Canada will support its partners in the discovery and development of new genomic solutions, tools and technologies as well as in the integration of AI to enhance the analysis and application of genomic data. This, in turn, will enable further innovation and lead to the availability and implementation of precision health solutions for all Canadians.

GE^{3}LS will also be an integral part of this pillar, emphasizing the importance of addressing the broader implications of genomics research in health. By incorporating GE^{3}LS, the initiative acknowledges the multifaceted impact of genomics on society and commits to advancing research and the implementation of new genomic solutions and technologies in a responsible and ethically sound manner. This comprehensive approach ensures that the benefits of genomic advancements will be realized for all Canadians.

Pillars 2 and 3 are expected to launch by early 2026. Further details will be posted to Genome Canada’s website.

Appendix 1: Required core elements of a participant informed consent form

Participant informed consent is a crucial component of the Canadian Precision Health Initiative because it ensures that individuals are fully aware of how their genetic information will be used, stored and shared.

The following core elements must be included in the participant informed consent form used by the project. An arms-length third party will review and validate the consent forms used by the projects as part of the project review process to ensure they include:

- permission to link with and update data from health records and registries
- permission to deposit data in a cloud-based, access-controlled databank
- permission to share data with national and international researchers from academic, charitable organizations, hospitals and for-profit private companies
- permission to re-contact and obtain assent from a study participant, when appropriate
- ability to share minimum required clinical data for all sequenced participants (see Appendix 2: Case report form sample)

Appendix 2: Case report form

Field	Field description	Type	Permissible values
Study			
study_id	Unique identifier of the study.	Text	Unique identifier of the CPHI study.
name	Study name	Text	Study name
description	Information about the study	Text	An in-depth description of the study, including its overall purpose, goals, scope or nature.
consent	Was informed consent obtained?	Text	Yes No
Participant			
submitter_participant_id	Unique identifier of the participant within the study, assigned by the data provider.	Text	Values must meet the regular expression ^[A-Za-z0-9\-\._]{1,64}} Examples: 90234, BLD_donor_89, AML-90
sex_at_birth	**Refers to sex assigned at birth. Sex at birth is typically assigned based on a person's reproductive system and other physical characteristics. The provided values are based on the categoried defined by Statistics Canada**	Text	Male Female Missing Not collected Not provided Restricted access Not applicable

Field	Field description	Type	Permissible values
vital_status	Participant's last known state of living or deceased.	Text	Alive Deceased Missing Not collected Not provided Restricted access Not applicable
date_of_birth	Indicate participant's date of birth.	Text	Format YYYY-MM-DD
date_of_death	Indicate participant's date of death.	Text	Format YYYY-MM-DD Not applicable
geographical_origin	Forward Sortation Area, part of the postal code indicating the geographic area	Text	Examples: H4A
height	Partcipant's height	Text	cm
weight	Partcipant's weight	Text	kg
gender	**Refers to an individual's personal and social identity as a man, woman or non-binary person (a person who is not exclusively a man or a woman). The provided values are based on the categories defined by Statistics Canada**	Text	Man Woman Non-binary person Missing Not collected Not provided Restricted access Not applicable

Field	Field description	Type	Permissible values
ethnicity	Refers to the ethnic or cultural origins of a person's ancestors. The provided values are based on the list of ethnic or cultural origins 2021 defined by Statistics Canada	Text	North American European Caribbean Latin, Central and South American African Asian Oceanian Other ethnic and cultural Missing Not collected Not provided Restricted access Not applicable
race	A social construct used to judge and categorize people based on perceived differences in physical appearance in ways that create and maintain power differentials within social hierarchies. There is no scientifically supported biological basis for discrete racial groups. The provided values are based on race-based data standard defined by CIHI guidance	Text	Black East Asian Indigenous (First Nations, Inuk/Inuit, Métis) Latin American Middle Eastern South Asian Southeast Asian White Another race category Do not know Prefer not to answer Missing Not collected Not provided Restricted access Not applicable

Sample			
submitter_sample_id	Unique identifier of the sample within the study, assigned by the data provider.	Text	Values must meet the regular expression ^[A-Za-z0-9\-\._]{1,64} Examples: hnc_12, CCG_34_94583, BRCA47832-3239
site_ID	Name of the organization performing whole genome sequencing	Text	Name of approved sequencing centre (Site ID assigned by Genome Canada)
genomic_reference_sequence_ID	Identifier for the genomic reference sequence used	Text	Examples: short read, long read
date_sample_collected	Date sample was collected	Text	Format YYYY-MM-DD
sample_type_collected	Description of the type of tissue/sample collected.	Text	Examples: Blood, bone marrow, tissue sample
report_received	Date the whole genome sequence report was received	Text	Format YYYY-MM-DD
Disease			
clinical_status	Status of the patient's conditions, whether active or inactive	Text	Active Inactive Resolved Remission Unknown Not applicable
clinical_diagnoses	Current medical diagnoses of the patient	Text	ICD-10 codes (e.g., E11 for Type 2 diabetes mellitus) Not Applicable

date_of_diagnoses	Date when the patient was diagnosed with the conditions	Text	Format YYYY-MM-DD Not applicable
disease_progression	Information on how the disease has progressed over time	Text	Stable Improving Worsening Remission Relapse Unknown Not applicable

Appendix 3: Application and portfolio evaluation criteria

Genome Canada uses a rigorous, independent peer review process to assess the merit of proposals, as well as their potential for impacts and benefits for Canada, and to ensure that sound management and financial practices are implemented.

Proposals that are not considered meritorious during the first round of submissions will not be reviewed in the second round.

ELIGIBILITY CRITERIA

Each proposal will be reviewed for eligibility at each stage of the application process using the criteria listed in the "Eligibility criteria and portfolio considerations" section of the funding opportunity document.

FULL PROPOSAL TECHNICAL REVIEW CRITERIA

Full proposals will be reviewed using criteria from the following five categories:

1. Project plan
2. Health equity
3. Impact and benefits to Canada
4. Comprehensiveness of the data strategy
5. Budget justification and fiscal responsibility

1. Project plan

- **Project feasibility:** How well-defined and actionable is the project plan in terms of governance, management and the strategic realization of goals?
- **Timeline:** To what extent does the project present a clear and realistic timeline, with specific milestones and deliverables that are achievable within the proposed time frame?
- **Methodology:** How well does the proposed methodology align with the project's objectives, demonstrating sound scientific and technical approaches to achieving the desired outcomes?
- **Risk management:** Does the plan include a comprehensive risk management strategy that anticipates potential challenges and outlines mitigation measures to ensure project success?
- **Management expertise:** How well does the management plan address project governance, personnel accountabilities and decision-making processes? Are the

project leaders equipped with a robust management structure to effectively oversee the project?

- **Team expertise and IDEA integration:** Does the project team possess the necessary expertise? Are there concrete plans to ensure that inclusion, diversity, equity and accessibility (IDEA) principles are central to team management and composition?

2. Health equity

- **Ethical considerations:** How effectively does the project address ethical issues, including informed consent, data privacy and adherence to relevant ethical guidelines, ensuring the protection and respect of all participants?

- **Enhancement of genomic data diversity**: Does the project increase the volume and diversity of the genomic data available on under-represented groups by sequencing genomes, generating, linking and facilitating better access to data from these populations?

- **Reduction of health disparities:** Will the project contribute to reducing health disparities and promoting equity in genomics and precision health?

- **Community engagement:** Do the project's design and implementation and dissemination plans include robust mechanisms for engaging diverse communities, ensuring their perspectives and needs are integrated?

- **Indigenous data sovereignty:** How well does the project uphold Indigenous rights in data governance, promoting respect for data sovereignty and ensuring equitable access and benefits for Indigenous communities?

3. Impact and benefits for Canada

Deliverables include:

- **Data solutions:** Will the project not only generate innovative, interoperable genomic data that address critical scientific or health challenges in Canada, but also enhance data integration across diverse platforms, contributing to a more robust and unified national health data landscape?

- **Healthcare impacts:** Will the project deliver outcomes that drive significant impacts in health care, enhance value in health-care delivery, and contribute to the sustainability of Canada's health system?

- **Industry value creation:** Does the project have the potential to create value for industry by fostering partnerships, advancing technologies or opening new market opportunities?

Expected benefits are:

- **Public health impact:** To what extent will the project improve public health outcomes, particularly in areas relevant to Canadian populations?

- **Societal impact:** Will the project promote societal benefits, such as enhancing inclusion, diversity, equity and accessibility in Canada's health-care system?

- **Research innovation:** Will the project introduce novel approaches or technologies to advance genomic research, promote collaboration and strengthen Canada's leadership in the field?

4. **Comprehensive data strategy**

    - **Data generation, management and analysis:** Are the plans for data generation, management and analysis appropriate and feasible within the project's scope?

    - **Data-sharing and accessibility:** How robust and timely are the plans for sharing data and resources with the national databank to maximize accessibility and utility?

    - **Inclusion of clinical phenotypes and metadata:** Does the project include comprehensive clinical phenotypes and metadata that enhance the relevance and applicability of the genomic data collected?

    - **Data governance and stewardship:** Are the data governance and stewardship practices, including ethical considerations, addressed adequately and aligned with recognized standards (e.g., Global Alliance for Genomics & Health standards)? Note that:
        - The plans must comply with **Genome Canada's Data Release and Sharing Policies.**
        - The team must share and preserve genomic data in the national databank and any accompanying metadata using existing repositories rather than keeping them solely with the researcher or institution and must provide these upon request.

    - **Commitment to data preservation and compliance:** Is there a clear commitment to preserving and sharing data with the national databank to ensure long-term accessibility and compliance with Genome Canada's policies?

5. **Budget justification and fiscal responsibility:**

    - **Budget alignment with project goals:** Is the budget reasonable and proportional to the project's goals, considering the level of effort required?

    - **Documentation and compliance of expenditures:** Are the proposed expenditures thoroughly documented, transparent and compliant with funding guidelines?

- **Fiscal oversight and management:** To what extent does the proposal include robust mechanisms for ongoing fiscal oversight, ensuring that project expenditures will be closely monitored and managed effectively throughout the project's duration?

- **Co-funding documentation:** To what extent is the proposed co-funding plan well documented, eligible and feasible?
- **Co-funding alignment with project goals:** Does the proposed co-funding plan clearly support the project's objectives?
- **Co-funding feasibility:** How likely is it that the project will be able to secure at least 75 per cent of the co-funding for eligible costs before the deadline for the release of funds?

PORTFOLIO CRITERIA

The portfolio criteria below are listed in order of priority.

Criteria	Qualifier		
	Green	Yellow	Red
Genomes sequenced*	100,000 or more genomes will be generated	80,000 to 100,000 genomes will be generated	Fewer than 80,000 genomes will be generated
Population diversity*[3]	Diversity is prioritized in the cohort and is representative of the population of Canada, including Indigenous populations	Diversity is prioritized in the cohort and is mostly representative of the population of Canada, but excludes unique regional populations	The dataset is not representative of the population of Canada
Clinical utility and impact*	The project will generate clinically impactful outcomes for both Canadian patients and health-care systems in the near term (i.e., in the next 5 years)	Some clinically impactful outcomes will be generated in the next 5 years	Clinically impactful outcomes are unlikely to be generated in the next 5 years

[3] Population diversity refers to the inclusion of varied genetic backgrounds from different demographic groups, ensuring representation across ethnicities, regions and ancestries.

Criteria	Qualifier		
	Green	**Yellow**	**Red**
Data quality*	The generated dataset includes metadata in addition to the minimum required clinical data, multi-omics data, and data from additional sequencing approaches	The generated dataset includes metadata in addition to the minimum clinical data required, multi-omics data, and data from additional sequencing approaches, but may not enrich the genomic dataset generated	Metadata, in addition to the minimum required clinical data, multi-omics data, and data from additional sequencing approaches, are not well presented in the dataset generated
Pan-Canadian benefits	A broad Canadian geography is represented and benefits are expected nationwide	Benefits are expected nationwide, and a broad Canadian geography is represented, but unique regional populations are not included	A broad Canadian geography is not represented, and nationwide benefits are not expected
Health equity	The portfolio prioritizes the reduction of health disparities and promotes equitable access and outcomes for marginalized communities	The portfolio includes diverse populations, but will have limited impact on improving equitable access and outcomes for marginalized communities	The portfolio is unlikely to improve equitable access and outcomes for marginalized communities
Research innovation	The portfolio pushes boundaries by introducing novel approaches, technologies or methodologies, fostering interdisciplinary collaboration, and enhancing Canada's leadership in genomics	The portfolio incorporates some innovative elements, but these may not significantly advance current practices or methodologies in genomic research	The portfolio lacks substantial innovation and relies primarily on established approaches, limiting its potential impact

Criteria	Qualifier		
	Green	**Yellow**	**Red**
Industry value creation	The genomic data generated are structured to support uptake by a broad group of industry stakeholders	The genomic data generated support uptake by industry stakeholders, but key industry groups are not represented	The genomic data generated are unlikely to support uptake by industry stakeholders

* Equally weighted criteria.