

Canadian Genomics Partnership for Rare Diseases (CGP4-RD)

Mission Statement

Goal

The goal of this Partnership is to ensure access to clinical genome-wide sequencing as a standard of care for Canadians at risk for a serious genetic condition and who would benefit from diagnosis and/or timely and effective intervention. Moreover, the Partnership will enable patients to benefit while helping others through the sharing of their clinical and genomic data within a learning health system¹. It lays the foundation for precision health in Canada.

Principles

- Implementation of cost-effective clinical genome-wide sequencing as a standard of care for patients and families suspected of having a serious genetic disease in Canada can best be achieved by a nation-wide Partnership involving provincial health ministries, health authorities, patient organizations, families, relevant healthcare institutions and providers, and key stakeholder organizations, such as Genome Canada, the regional Genome Centres and the Canadian Institutes of Health Research.
- Clinical genome-wide sequencing should be provided in a manner that:
 - is consistent with Canadian law, ethical principles and values;
 - is responsive to the needs of patients and their families;
 - is accessible through the provincial/territorial healthcare system to all patients who are suspected of having a serious genetic disease and may benefit from such testing, in accordance with current professional guidelines;
 - includes pre-test and post-test genetic counselling and the availability of specialist medical consultation in a system of care that strives to maximize benefit and minimize the chance of harm for patients and their families;
 - ensures that the test and its laboratory and clinical interpretation and reporting quality, meet or exceed provincial, Canadian, and international clinical standards;
 - is efficient and transparent;
 - provides patients and their families with the opportunity to share their clinical and genomic data and to participate in research benefiting themselves and other patients and families; and,
 - aligns with national and international data sharing initiatives, such as the Global Alliance for Genomics and Health.

¹ A learning health system is the concept of capturing, aggregating and analyzing data generated in standard clinical care to create a body of evidence for research so as to continuously improve health care cost, operations and patient outcome.

In accordance with these principles, organizations and individuals subscribing to this Mission Statement agree to:

- collaborate to work towards the provision of genome-wide sequencing as a regular clinical service through the provincial healthcare system, as appropriate in light of current evidence;
- create a sustainable governance structure that will oversee the development of the Partnership;
- participate in the development and implementation of evidence-based Canadian clinical guidelines and care standards for the performance and delivery of clinical genome-wide sequencing for patients suspected of having a serious genetic disease;
- share, in a secure and protected way, individual, family and population clinical and genomic data relevant to understanding genetic variation and disease in accordance with patient/family/community wishes and to the extent permitted by law;
- participate in the development and implementation of a pan-Canadian database to share such data with appropriate clinicians and researchers throughout Canada to enhance the interpretation of genomic variants and further understanding of phenotypic variability and natural history;
- support the development and operation of appropriate clinical and laboratory quality standards, quality assurance and quality improvement activities related to clinical genome-wide sequencing;
- use and develop global data standards and FAIR² principles for purposes of interoperability, harmonization and collaboration.
- collect and analyze data and processes on an ongoing basis to assess the clinical utility and cost effectiveness of clinical genome-wide sequencing and the associated clinical services;
- contribute to the development and deployment of harmonized data collection, sharing and access policies, through a pan-Canadian ethics and governance framework;
- promote the continuing development and use of Canadian technological resources and expertise in clinical genome-wide sequencing, data interpretation, data management, and clinical translation to maximize medical, scientific, educational and economic benefits;
- monitor and regularly report their contributions to the provision of clinical genome-wide sequencing and other agreed-upon issues; and
- review the Mission Statement at least once a year and work to improve it and the Partnership with the goal of making genome-wide sequencing available as a clinical service to all Canadians at risk for a serious genetic disease.

² Principles and associated standards for making data resources findable, accessible, interoperable and reusable: <https://www.force11.org/group/fairgroup/fairprinciples>