Executive Summary

Towards the routine use of genome-based testing in Canada's largest regions:
A State of Readiness Progress Report

Why genome-based testing?

High quality care that most benefits patients requires information from testing. Increasingly, laboratory-based biomarkers that measure the expression, function and regulation of genes and gene products are being used for this purpose. These genome-based tests have already become commonplace in oncology and there is expected to be an exponential growth in new tests and test approaches across a number of therapeutic areas in coming years — an era of genomic medicine.

A rapid increase in available innovation and technologic platforms will also lead to an increase in the complexity of implementing genome-based tests and how testing services can be delivered effectively and efficiently. As the structure, remit and organization

of healthcare systems (and the laboratory functions within them) vary, there are likely to be no one-size-fits-all solution; however , some necessary conditions will be required to manage these technologies in a way that benefits patients and is sustainable.

Unlike traditional healthcare technologies, genome-based testing also provides research opportunities beyond healthcare decision making, and can lead to discoveries about the nature of disease or effectiveness of current and future therapies. They will also serve as an important part of implementing future cell and gene therapies and in the diagnosis and monitoring of genetic and rare disease.

What is the impact of system readiness?

Genomic medicine has already improved patient health outcomes through predicting who may benefit (or not be harmed) from therapy, and helping clinical decisions through better prognosis and diagnosis of disease.

However, genomic medicine harbors a number of additional benefits including:

- **Better patient and care provider experiences** – Reducing the need for referrals and other diagnostic tests, and improving time to diagnosis. Improving the state of readiness across Canada will lead to equitable care and access.
- **Better science and economic growth** – Aiding scientific discovery and clinical trial enrollment, creating commercial and investment opportunities as well as future-proofing Canada's healthcare workforce.

- **Healthcare efficiency** – Genomic medicine creates opportunities to reduce healthcare costs while creating the necessary infrastructure for delivering 21st century care.
- **Equitable access to care** – Readiness at a provincial level ensures equitable access to care across Canada and within provinces, including between academic and community settings. Standards for readiness also create opportunities for collaboration across provinces.

Summary

- Genomic medicine harbors the real potential to improve the health and healthcare journey of patients, care provider experiences, and improve health system efficiency – even reducing health care costs. There is expected to be an exponential growth in medically necessary new genome-based tests and test approaches in coming years.
  - ◆ Unlike traditional healthcare technology, genome-based testing can also create scientific research and commercial opportunities beyond healthcare decision-making.
- The State of Readiness Progress Report uses a purposive sample of 5 healthcare regions in Canada: Ontario, Quebec, British Columbia, Alberta, and Nova Scotia, representing >85% of the Canadian population.
  - ◆ While Canada has made some progress, it is far from ready. There are still significant gaps that need to be addressed. These gaps vary by province and include: better informatics/linked data systems; timely, fair and equitable test review processes; navigational and educational supports for care providers; timely and adequate financing for test services; and creating better opportunities for innovation through genomic medicine.
- Further research to fill these gaps should engage patients, care providers, and health systems.
  - ○ Key priorities include implementation research to support innovators, linking research data to health data, and exploring the ethical legal and social implications of testing, particularly in high priorities such as rare disease.
- Further work must also be done to improve patient journeys from diagnosis of disease to receiving timely and high quality care that can benefit their families while advancing our scientific understanding of human health; creating better opportunities for genomic innovation is a clear means of improving the health and welfare of Canadians.

What Does Readiness Mean for Patients?

- **Improving health.** Genome-based testing enables more accurate and timely diagnosis or the use of targeted treatments which can be more effective and less harmful to patients. It may also help patients and care providers better understand the future of their disease, and help patients and their families make more informed decisions.

- **Improving care experiences for patients and their families.** Knowing what genetic mutations are responsible for their loved one's disease may be potentially lifesaving for both patients and family members and could also allow for earlier (and less costly) interventions.

- **Fair treatment.** Patients are concerned that access to valuable testing may be determined by where they live or how well-informed care providers are about the availability or health impacts of testing. Readiness for testing means patients can receive the best possible care, regardless of where they live.

- **Time and support.** Patients, especially those with rare conditions and poor prognoses, don't have time to waste. Genomic medicine can expedite their time to diagnosis and treatment. A ready health system can also offer necessary counselling and psychosocial support for patients and their loved ones to better understand their illness.

Is Canada Ready?

To better understand Canada's state of readiness, a set of necessary conditions was developed in consultation with regional and national experts. These conditions fell into three categories:

1) *Infrastructure* - Health and human resource infrastructure that includes communities of practice, resource planning and a digital infrastructure (informatics).

2) *Operations* - the ability to evaluate and , coordinate, and implement testing at a health system level including an entry point for innovation, an evaluative function, a model for coordinating service, and supports for care provider awareness and patient navigation.

3) *Healthcare environment* – the larger healthcare supports required including necessary approaches to financing, integrating innovation, education and training patients and providers, and regulating testing to ensure quality care.

Eleven essential conditions were then mapped to Canada's 4 largest provinces as well as Nova Scotia, the largest province in Canada's Atlantic region. Assessing the state of progress for each of these regions revealed varying states of readiness for genomic medicine.

Important gaps that will need to be addressed in Canada's future include:

- **Improving Informatics** – Is essential for test development, interpretation, and clinical decision support. Ensuring adequate integration of test results into electronic health records will also provide a key resource for real-world monitoring, disease management, quality assessment and assurance, and financing. Most provinces still lack sufficient data integration.

- **Evaluation/Health Technology Assessment (HTA)** – Fit for purpose HTA will be needed to identify high-value testing. Most provinces lack evaluative processes that adhere to HTA principles of timeliness, transparency, and engagement.

- **Navigational Tools** – Effective delivery of genetic testing requires navigation tools for patients and the public including referral guidelines, a test directory, eligibility criteria, tools/ education for ordering genetic testing, and a care clinic directory. Some of the provinces are working on these navigational resources.

- **Financing Approach** – Most provinces lack dedicated funding to facilitate rapid onboarding or a funding formula that supports test development and proficiency testing. The current reliance on the private sector to fund test development may be counterproductive as priorities are influenced by who is paying, rather than unmet need, equity, or efficiency.

- **Engagement** – High performing health systems require broad engagement of those impacted by testing. These include the patients, administrators, IT professionals, implementation and genome scientists, public and private sector innovators and others (scientists, legal and ethics experts, professional organizations, bioethicists, regulators).
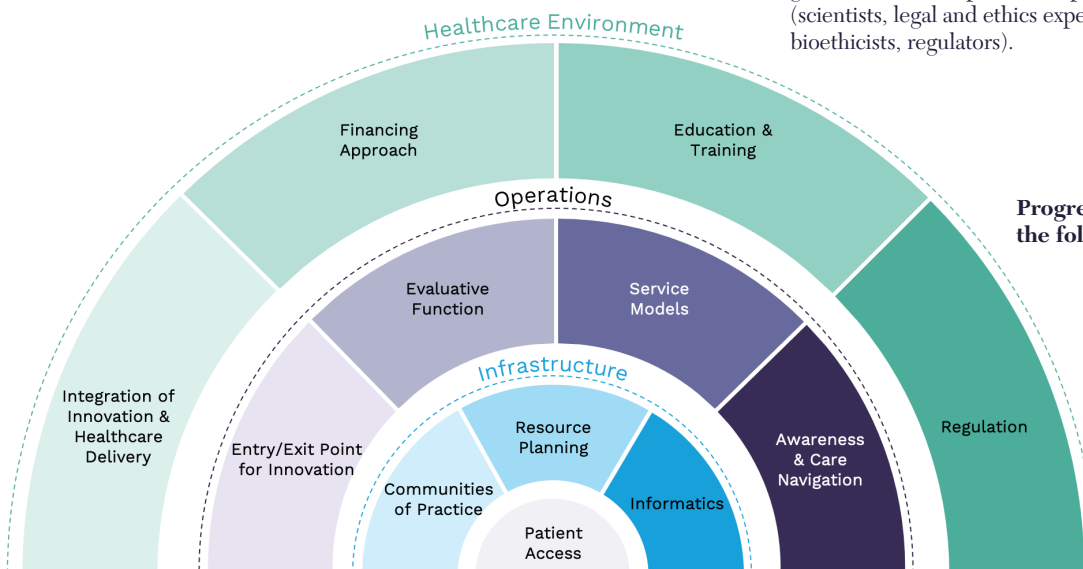
Progress by province is described in the following pages.

Where do the Provinces Stand?

Alberta and Quebec have created more of the necessary conditions for readiness, largely in part to the earlier establishment of single, laboratory service organizations and programs that provide the necessary infrastructure for coordination and planning as well as necessary operational conditions. In Nova Scotia, a higher level of coordination and planning is achieved due to lower levels of service demand and the ability of the government to work directly with the individual teaching hospitals who provide province-wide testing. However, many of the operational and evaluative processes are informal, and not public facing. The opposite is true in Ontario, which is challenged with much higher levels of demand for service, a complex web of formal evaluative processes, and until recently, a highly decentralized health system.
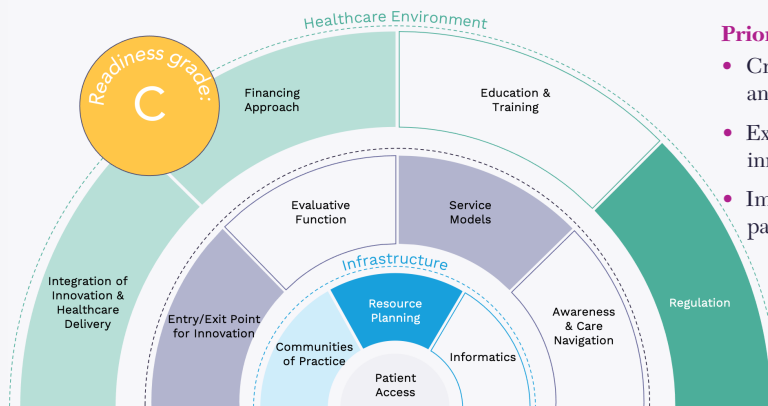
British Columbia

Strengths:

- Single service organization (Provincial Laboratory Medicine Services) that establishes a community of practice and supports resource planning.
- Single point of entry with explicit timelines for evaluation and coordination across service providers.
- Some integration of innovative testing.

Weaknesses:

- Lack of integration of laboratory information systems.
- Limited engagement and involvement of broader stakeholder community.
- Substantial opportunities to improve care navigation.

Priority Actions

- Create a cross-regional integrated laboratory information system and a plan for integration into electronic health records.
- Expand networks to include broader members of the innovation community,
- Improving the processes of navigation for care providers and patients and develop standards for education and training.

Readiness grade: C

Its current state of readiness has earned
British Columbia a grade of C

Alberta

Strengths:

- Single service organization (Alberta Precision Laboratories) that provides oversight and resource planning.
- Integration of laboratory information across province is established.
- Integration and exchange with innovators through dedicated translational research programs, open application processes and mainstream use of investigational testing.

Weaknesses:

- The test review process, timelines and criteria are not publicly available.
- There are still opportunities to improve care navigation and education standards for patients and providers.

Priority Actions

- Expand opportunities for engagement with broader members of the innovation community to improve healthcare planning and foster innovation.
- Improve the process of deliberation that surrounds the consideration and adoption of tests.
- Improving the financing approach to include funding for test development and to account for capital infrastructure, human resources, and other associated costs of testing.
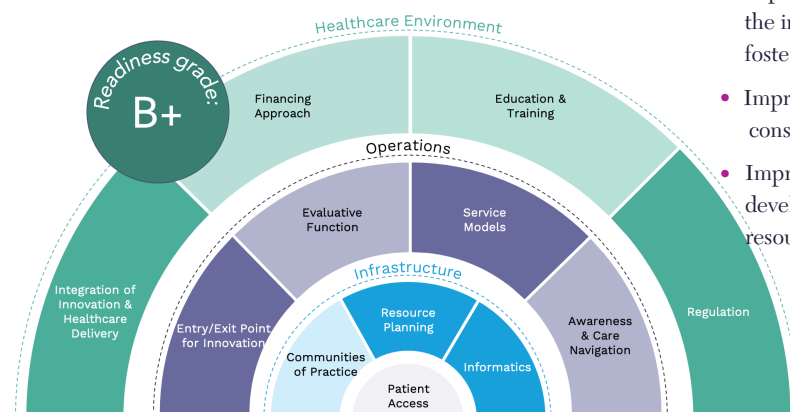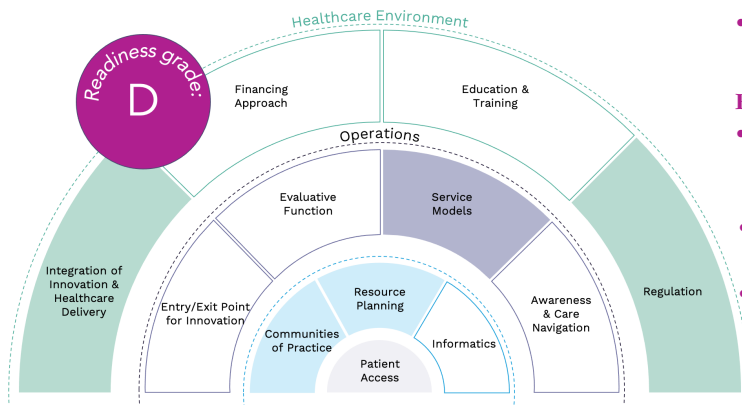

Readiness grade: B+

Its current state of readiness has earned
Alberta a grade of B+

Needs Improvement
Partially Established
Established

Ontario

Strengths:

- Recently created single service organization (Provincial Genetics Program).
- Clear standards for accreditation and proficiency.

Readiness grade: D

Healthcare Environment
Financing Approach
Education & Training
Operations
Evaluative Function
Service Models
Integration of Innovation & Healthcare Delivery
Entry/Exit Point for Innovation
Resource Planning
Communities of Practice
Informatics
Patient Access
Awareness & Care Navigation
Regulation

Weaknesses:

- Funding not timely or transparent; no funding for test development or human resources .
- No integration of laboratory information.
- Multiple evaluative frameworks.
- Limited engagement and involvement of broader stakeholder community.
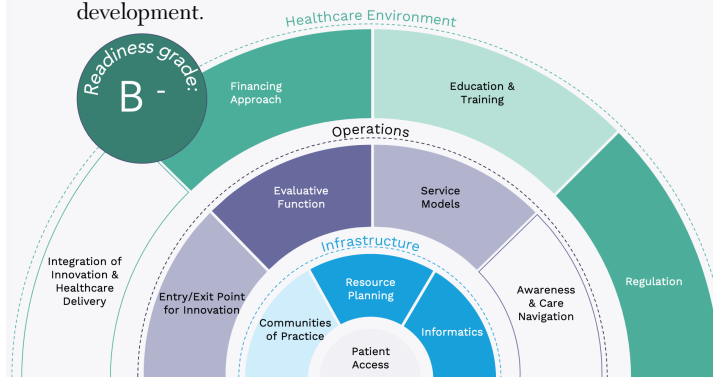
Priority Actions

- Shift away from the Ministry acting as a decision-maker for the funding of individual tests, and toward a system of Ministry as a steward.
- Consolidate evaluation processes and adopt a single-entry approach, supported by horizon scanning.
- Create a laboratory information system integrated with clinical health records.

Its current state of readiness has earned Ontario a grade of D

Quebec

Strengths:

- Single service organization (Direction de la biovigilance et de la biologie médicale).
- Single point of entry and somewhat transparent evaluation.
- Nimble financing approach with funding available for test development.

Readiness grade: B -

Healthcare Environment
Financing Approach
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Weaknesses:

- Navigation and education for care providers and patients limited.
- Limited integration of innovation and healthcare delivery.
- Limited engagement and involvement of broader innovation community.
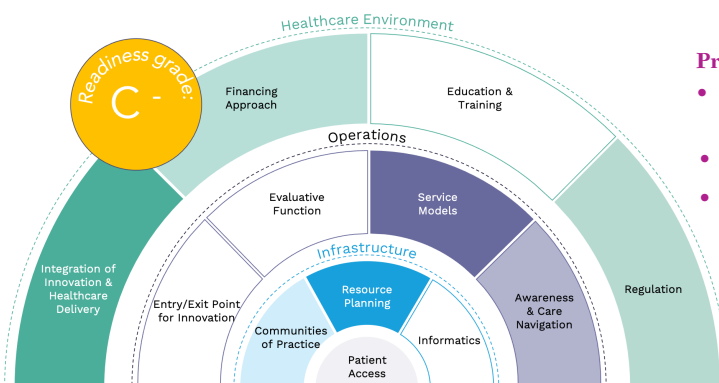
Priority Actions

- The DBBM should consider a separate advisory council for commercial innovators.
- Improve the processes of navigation for care providers and patients; develop standards for education and training.
- Further integrate innovative testing into the mainstream delivery of care, consistent with Quebec's goals within its Bureau of Innovation.

Its current state of readiness has earned Quebec a grade of B -

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Nova Scotia

Strengths:

- Dedicated program (Pathology and Laboratory Medicine Program) that provides oversight and resource planning through key teaching hospitals.
- High level of service coordination.
- Integration of innovative testing.

Readiness grade: C -

Healthcare Environment
Financing Approach
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Regulation

Weaknesses:

- No single entry point, explicit review process, timelines or criteria used to consider new tests.
- Lack of integration of laboratory information across centres.
- Limited engagement and involvement of broader stakeholder community.

Priority Actions

- Create a a transparent evaluation process and a single-entry approach, supported by horizon scanning.
- Integrate laboratory and clinical information systems.
- Expand engagement with broader members of the healthcare / innovation community, particularly commercial innovators.

Its current state of readiness has earned Nova Scotia a grade of C -

Needs Improvement
Partially Established
Established

More information about the State of Readiness Progress Report for Genomic Testing in Canada can be found here: TBD