



GENOMICS AT WORK FOR

ALBERTA'S HEALTHCARE SYSTEM

What We Heard Workshop Summary

DECEMBER 2023

As a not-for-profit research funding organization, we're here to help initiate, fund and manage genomics partnerships and research. Together, we will catalyze genomics solutions that benefit Albertans and the world.

Genome Alberta is part of a Canadian network, established in 2000, to build international leadership in genomics and other 'omics research, develop specific Life Sciences research platforms and address and support regional priorities.

Since our establishment, Genome Alberta has enabled **\$572.6M** of genomics research and has helped secure **\$126M** of federal research funds for Alberta-led projects, creating thousands of Alberta jobs and 26 spin-off companies.

We add significant value to the provincial innovation ecosystem by:

- Working collaboratively with innovation entities to drive technology development and implementation.
- Securing significant federal funds for Alberta-led research and increasing innovation support for our provincial priorities.
- Ensuring scientific excellence is supported by social sciences research and addressing ethical, legal and environmental barriers.

OUR VISION

To inspire and catalyze genomics solutions that benefit Albertans and the world.

OUR MISSION

To promote and support genomics solutions for end-user needs to create value and investment opportunities through excellent science, technology and application development, collaborations and partnerships.

EXECUTIVE SUMMARY

Genetic tests (including single gene tests, gene panels, and/or whole exome or genome sequencing) play a pivotal role in modern healthcare. The integration of genetic data into routine clinical care is revolutionizing disease management and improving patient outcomes through faster diagnoses, informed decision-making, and the use of targeted preventions and treatments (where available).

Additional benefits of evidence-based and appropriate genomic medicine include improvements in healthcare costs and efficiencies, and advances in scientific discovery and clinical trial enrolment. The challenge that health systems face, is how to scale genetic testing so that it is more accessible to patients.

A recent ‘State of Readiness Progress Report’ (May 2023)¹ evaluated five Canadian healthcare regions with regard to their state of infrastructure, operations and healthcare environment required to implement, deliver and regulate genetic testing. Findings from the report indicate that Alberta has many of the conditions necessary for broader implementation of genetic testing and thus the delivery of ‘genomic-based medicine’ – e.g., the broader incorporation of genetic tests into routine clinical care to improve health outcomes.

On December 7, 2023, Genome Alberta hosted a ‘Genomics at Work for Alberta’s Healthcare System’ workshop that brought together a cohort of professionals working at the interface of science, policy, and healthcare, to discuss Alberta’s state of readiness to broadly scale genetic testing into routine care. Workshop objectives were to:

- Provide a forum to engage the research community, private and public sector representatives to increase awareness about the current state of genetic testing in Alberta’s healthcare system,
- Discuss infrastructure, operations and healthcare environment opportunities and barriers to the broader implementation of genetic testing in Alberta, and
- Consider future research programs that could address such barriers and opportunities, thus helping to inform future research funding programs.

¹ Husereau D, Villalba E, Muthu V, Mengel M, Ivany C, Steuten L, Spinner DS, Sheffield B, Yip S, Jacobs P, et al. Progress Toward Health System Readiness for Genome-Based Testing in Canada. 2023. ISBN 978-1-7389568-0-7.

The workshop provided a valuable forum for improving understanding of the current state of genome-based medicine in Alberta and for engaging in a collaborative, open and frank discussion on the most pressing barriers to overcome for broader adoption of genome-based testing in Alberta's healthcare system. Barriers include work force, policy hurdles, testing knowledge of healthcare providers and patients, cost, and more. A summary of the forward-looking discussions regarding Alberta's readiness for the era of genomic medicine are summarized in the opportunities and challenges sections in the areas of infrastructure, operations and health environment on the following pages.

Genome Alberta will continue to support efforts, in partnership with the broader health innovation community in Alberta, to address the identified barriers through the innovation and research funding programs we deliver. Knowledge and insights gained will help to inform future research and innovation programs that enable greater benefits to Albertans through adoption of genomics medicine.

The workshop provided a valuable forum for improving understanding of the current state of genome-based medicine in Alberta and for engaging in a collaborative, open and frank discussion on the most pressing barriers to overcome for broader adoption of genome-based testing in Alberta's healthcare system.

TABLE OF CONTENTS

- Introduction 4
- Overview - Conditions Required for Delivery of Genomic Medicine 6
- Overview - State of Genetic Testing in Alberta 7
- Workshop Discussions on Improving Alberta’s Readiness for Genomic Medicine 8
- 1. Infrastructure 8**
 - 1.1 Alberta’s Single Integrated Clinical Testing Laboratory..... 8**
 - Opportunities..... 8
 - Challenges..... 9
 - 1.2 Laboratory Resource Planning 9**
 - Opportunities..... 10
 - Challenges..... 10
 - 1.3 Digital Infrastructure..... 11**
 - Opportunities..... 12
 - Challenges..... 12
- 2. Operations..... 14**
 - 2.1 Transparent Test Evaluation and Listing Process 14**
 - Opportunities..... 14
 - Challenges..... 14
 - 2.2 Entry Point for Innovation and Role of Private Sector..... 15**
 - Opportunities..... 15
 - Challenges..... 16
- 3. Health Environment 17**
 - 3.1 Growth and Sustainability of Genomic Medicine 17**
 - Opportunities..... 17
 - Challenges..... 17
 - 3.2 Education of Patients and Care Providers..... 18**
 - Opportunities..... 18
 - Challenges..... 18
- What’s Next 19
- Acknowledgements and List of Participating Organizationsinside back cover





INTRODUCTION

Genetic tests (including single gene tests, gene panels, and/or whole exome or genome sequencing) play a pivotal role in modern healthcare. While such tests are mainstays in cancer diagnosis and management, and in the diagnosis of genetic diseases (cystic fibrosis, Down syndrome, Duchenne muscular dystrophy, ataxia, etc.), evidence supporting the benefits of genetic testing in many other therapeutic areas is rapidly accumulating (e.g., rheumatoid arthritis, multiple sclerosis, Crohn’s disease, neurological conditions, etc.).

The integration of genetic data into routine clinical care is revolutionizing disease management and improving patient outcomes through faster diagnoses, informed decision-making, and the use of targeted preventions and treatments (where available). Additional benefits of evidence-based and appropriate genomic medicine include improvements in healthcare costs and efficiencies, and advances in scientific discovery and clinical trial enrolment. The challenge that health systems face is how to scale genetic testing so that it is more accessible to patients. Barriers include work force, policy hurdles, testing knowledge of healthcare providers and patients, cost, and more.

A recent ‘State of Readiness Progress Report’ (May 2023)² evaluated the five Canadian healthcare regions – British Columbia, Ontario, Alberta, Quebec, and Nova Scotia – with regard to their state of infrastructure, operations and healthcare environment required to implement, deliver and regulate genetic testing. Findings from the report indicate that Alberta has many of the conditions necessary for broader implementation of genetic testing and thus the delivery of ‘genomic-based medicine’ – e.g., the broader incorporation of genetic tests into routine clinical care to improve health outcomes.

On December 7, 2023, Genome Alberta hosted a ‘Genomics at Work for Alberta’s Healthcare System’ workshop that brought together a cohort of professionals working at the interface of science, policy, and healthcare, to discuss Alberta’s state of readiness to broadly scale genetic testing into routine care. The ‘workshop took place at the Life Sciences Innovation Hub in Calgary, Alberta. Forty-three individuals attended the workshop.

² Husereau D, Villalba E, Muthu V, Mengel M, Ivany C, Steuten L, Spinner DS, Sheffield B, Yip S, Jacobs P, et al. Progress Toward Health System Readiness for Genome-Based Testing in Canada. 2023. ISBN 978-1-7389568-0-7

Workshop objectives were to:

- Provide a forum to engage the research community, private and public sector representatives to increase awareness about the current state of genetic testing in Alberta’s healthcare system,
- Discuss infrastructure, operations and healthcare environment opportunities and barriers to the broader implementation of genetic testing in Alberta, and
- Consider future research programs that could address such barriers and opportunities, thus helping to inform future research funding programs.

This report provides an overview of discussions and learnings from the workshop. The workshop agenda included opening remarks by Tammy Hofer, COO at Alberta Precision Laboratories, a keynote presentation by Don Husereau, Adjunct Professor, School of Epidemiology and Public Health, University of Ottawa and two panel sessions.

The topic of panel session 1 was “Alberta’s healthcare system infrastructure and environment for routine genome-based testing” with panelists Dennis Bulman, Medical Scientific Director, Genetics & Genomics, Alberta Precision Laboratories, Kate Harback, CEO and Executive Director, Institute of Health Economics, Aneal Khan, CEO, Discovery DNA and Michael Mengel, Professor, University of Alberta. The topic of panel session 2 was “Alberta’s data management system for routine Genome-based testing” with panelists Kym Boycott, Associate Professor, University of Ottawa, Stafford Dean, Chief Data & Analytics Officer, Alberta Health Services, Erik Nohr, Molecular Pathologist, Alberta Precision Laboratories/University of Calgary, Jeff Round, Chief Scientific Officer, Institute of Health Economics, and Becky Wong, Director, Health System Access, Health Evidence & Innovation, Alberta Health Services. A full list of participating organizations can be found at the end of the report.

This report provides an overview of discussions and learnings from the workshop.

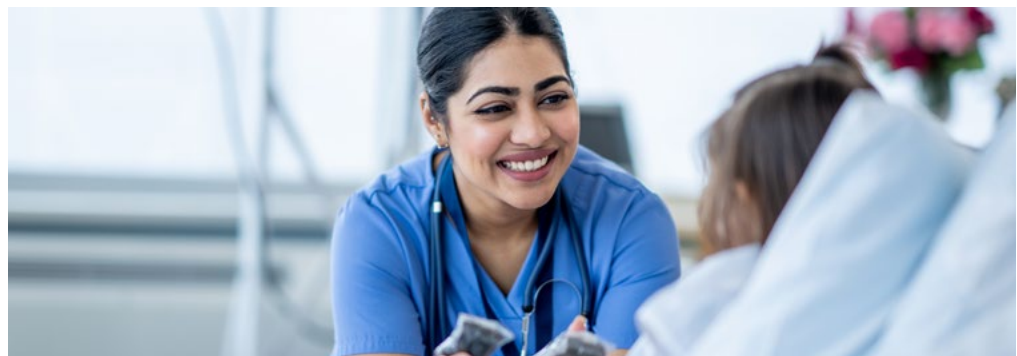
OVERVIEW -

CONDITIONS REQUIRED FOR DELIVERY OF GENOMIC MEDICINE

In consultation with regional and national experts, authors of the ‘State of Readiness Progress Report’ (May 2023)³ proposed a set of necessary conditions to consider for the implementation of genomic medicine into health systems.

These conditions formed the basis for discussions at the Genome Alberta-hosted workshop and are outlined as follows:

- **Infrastructure** - the health and human resource infrastructure required for genomic medicine that includes communities of practice, the ability to resource plan around genetic testing, and a digital infrastructure (informatics) system that incorporates genetic testing results into medical records and accommodates test interpretation and clinical decision-making.
- **Operations** - the ability to evaluate, coordinate, and implement genetic testing at a health system level including an entry point for innovation, a transparent evaluative function that includes fit for purpose health technology assessment (HTA) of tests and outcomes, and a model for coordinating services and supports for care provider awareness and patient navigation.
- **Healthcare Environment** – the larger healthcare supports required for genomic medicine that includes approaches to integrated care pathways and system financing, integrating innovation, education and training of patients and providers, and regulating testing to ensure quality of care.



³ IBID

OVERVIEW -

STATE OF GENETIC TESTING IN ALBERTA

Alberta Precision Laboratories (APL) is the provider of all publicly funded laboratory service operations across the province, including those in hospitals, urgent care centres, and in community care.

APL's provincial Genetics and Genomics program (formerly Genetic Lab Services) consists of laboratories in Edmonton and Calgary and provides highly specialized genetic testing for a significant number of medical conditions. Like other provinces, APL also accesses out-of-province genetic testing (from private and public entities) when there are capacity constraints, a lack of test availability in Alberta, or when the quality of testing depends on an adequate number of referrals. Coordinated outsourcing allows the purchase of bundled test volumes at reduced costs.

In Alberta, genetic tests are available for prenatal and newborn screening (along with other biochemical tests that detect inborn errors of metabolism), predictive disease testing, and late-onset diagnostic disease testing. Thus, testing is available for a variety of germline and somatic genetic tests, including Next Generation Sequencing (NGS) panels (i.e., targeted panel sequencing interrogating tens to hundreds of targeted genes) for cancer, cardiac, connective tissue, vascular, and endocrine disorders, as well as whole exome sequencing, which interrogates the coding region of every gene, for genetic disorders of unknown cause.

Genetic tests approved for Alberta's test menu are covered by Alberta's provincial health plans for those patients who meet test-specific conditions, and where a recommendation for testing has been made by a physician. In some instances, private genetic testing services are purchased by individuals paying the cost out-of-pocket, or in some cases, through private or employer-based insurance programs. Also, Alberta may provide and pay for testing (in-house or externally) for genetic tests that are not on Alberta's test formulary but have been requested by a physician, where the request has been approved through laboratory services. Finally, some drug companies offer select genetic testing to aid in therapeutic decisions.

As evidence accumulates to support the benefits of genetic testing in healthcare, health systems around the world are incorporating large-scale genetic testing into routine clinical care. While Alberta offers many advantages required for implementation of genomic medicine (e.g., health system infrastructure, data integration infrastructure, test evaluation processes), further investigations and planning are required to enable full implementation of genomic medicine into Alberta's healthcare system.

Genetic tests approved for Alberta's test menu are covered by Alberta's provincial health plans for those patients who meet test-specific conditions, and where a recommendation for testing has been made by a physician.

WORKSHOP DISCUSSIONS -

IMPROVING ALBERTA'S READINESS FOR GENOMIC MEDICINE

Topics not intended for discussion at the workshop included health system restructuring and historical issues. Forward-looking discussions regarding Alberta's readiness for the era of genomic medicine are summarized in the opportunities and challenges in the following sections.

1. INFRASTRUCTURE

1.1 Alberta's Single Integrated Clinical Testing Laboratory

Alberta Precision Laboratories (APL), a not-for-profit subsidiary of Alberta Health Services (AHS), provides province-wide clinical laboratory services to Albertans. The pan-provincial approach ensures that province-wide accreditation, proficiency, and analytical standards exist across individual labs, including those that provide genetic testing. Through the provincial Laboratory Formulary Committee, APL coordinates an evaluative process for new test proposals requested from stakeholders (care providers, patients, innovators), via a single point of access/entry located on the APL website. APL thus provides oversight and approval functions for new tests and oversees their implementation. A key strength in Alberta is AHS's **Strategic Clinical Networks (SCNs)**. The SCNs provide a system of clinicians, researchers, patients, and community representatives for APL to engage with regarding appropriate test utilization and test adoption to move evidence into practice. APL recently completed the implementation of a single integrated laboratory information system (EpicBeaker) for test ordering and reporting that is fully integrated with AHS's clinical information system (ConnectCare), thus allowing patient test results to be transmitted to the patient portal (with the exception of predictive genetic tests for conditions such as Huntington's Disease). Senior leadership in APL provides oversight to resource planning for testing, and are the point of engagement with clinical stakeholders, patients, government, and industry, as it pertains to testing.

Alberta Precision Laboratories (APL), a not-for-profit subsidiary of Alberta Health Services (AHS), provides province-wide clinical laboratory services to Albertans.

OPPORTUNITIES

- Having a single, publicly funded entity (APL) that provides clinical laboratory services across the province contributes to the coordination, nimbleness, and standardization of testing and analytics across Alberta. This was demonstrated during the recent COVID-19 pandemic, where APL's governance, leadership and communication channels facilitated deployment of high volumes of testing across the province.
- APL delivers laboratory services via a province-wide laboratory information system (EpicBeaker) that is fully integrated with AHS's clinical information system (Connect Care)

where test results are transmitted into the patient portal. The integrated health record ensures robust standardization and documentation with respect to test ordering and reporting.

- AHS's SCNs provide a network of care providers to work with APL in terms of identifying which tests should be adopted, and how tests are appropriately utilized in the right clinical context.
- Laboratories and sequencing equipment for genetic testing are established in Edmonton and Calgary and will expand with the opening of the **Arthur J. E. Child Comprehensive Cancer Centre**, in Calgary.

CHALLENGES

- Equal access to genetic testing expertise and genetic counselling regarding test results (e.g., through specialty care) is a challenge across the province (e.g., for rural and other underserved populations).
- Although APL has a large menu of approved tests, there are cases where tests are not on the test formulary because there are no care pathways in place to address test results. If there is no actionable treatment for a particular condition, testing is not usually justified.
- Laboratory services are on a 'fixed budget' thus a challenge is to determine where to spend laboratory dollars, e.g., what brings the greatest value from a health and economic perspective. For every dollar spent on a specific clinical area, a dollar is not invested in a different clinical area.
- With the significant and steady growth in genetic testing needs, ongoing investment in state-of-the-art equipment and training of highly qualified personnel is required.

1.2 Laboratory Resource Planning

APL is responsible for resource planning for all laboratory services and receives an annual funding envelope from AHS to do so. Unlike traditional tests, funding formulas for genetic testing must consider the need for additional resources (human capital) associated with development and proficiency testing, as well as the education of care providers required to interpret the test. If more genetic tests are adopted in Alberta, these secondary costs would need to be incorporated into the lab services budget. Alberta's reliance on research funding and/or private sector investment to develop and evaluate new genetic tests also limits their adoption into the health system. For example, a project funded by Genome Canada/Genome Alberta and Alberta Innovates (hosted through the University of Calgary) entitled 'Translational Implementation of Genomics for Rare Diseases' (TIGeR) is enabling the repatriation of exome and genome sequencing into the APL 'Genetics and Genomics' laboratories. While research funding for projects like this and other private investment is a good starting point, an integrated model comprising dedicated provincial funding for test development, would stimulate faster adoption of emerging genomics-enabled technologies that address gaps and find efficiencies to improve the quality of care in Alberta.

An integrated model comprising dedicated provincial funding for test development, would stimulate faster adoption of emerging genomics-enabled technologies that address gaps and find efficiencies to improve the quality of care in Alberta.

Additional genetic testing would require an increase in the numbers of clinical and laboratory geneticists, genetic counselors, and technologists trained to work in genetic testing labs.

Further, APL currently has a shortage of laboratory technicians, as well as other key laboratory personnel required for genetic testing. Additional genetic testing would require an increase in the numbers of clinical and laboratory geneticists, genetic counselors, and technologists trained to work in genetic testing labs. Resources would also need to be devoted to educating care providers and patients regarding test results and interpretation. Importantly, there is a national paucity of key people who interpret genetic data and develop genetic laboratory reports which needs to be addressed.

OPPORTUNITIES

- APL has strong leadership and vision for the implementation of genomic medicine.
- Alberta could meet growing test demands via increased equipment automation and adoption of artificial intelligence/machine learning (AI/ML) tools to help with testing and the analysis of data. Menial, repetitive tasks could be completed using automated technology, thereby enabling more effective utilization of employees' skills.
- APL services all of Alberta with tests offered to rural and urban communities alike. Genetic testing is centralized to the Edmonton and Calgary laboratory locations, however, samples from across the region are collected, providing access to testing for all Albertans.

CHALLENGES

- A challenge is to ensure that all care providers order genetic tests in the appropriate context. Care providers must be able to use the right test, in the right patient, and at the right time; be able to interpret test results; and then act appropriately to ensure optimal health outcomes and stewardship of valuable resources. One model would be to have every genetic test on the formulary, accompanied with evidence-based information regarding when to use the test and how to interpret it. Caregivers would need to be upskilled to be able to do this for genetic tests.
- Currently, only specialists (e.g., oncologists, cardiologists, geneticists) can order genomic tests thus creating a bottleneck in the system for test access. Additional training is necessary to allow other professions (primary care physicians, others) to order genetic tests. Further, it is recommended that the College of Physicians & Surgeons of Alberta (CPSA) make mandatory the ability for primary care (and other) physicians to order genetic tests, by ensuring that they meet competencies in these areas of practice. Medical schools must also incorporate greater training across all clinical areas regarding genomic medicine.
- Not enough clinical geneticists and clinical laboratory geneticists are trained in Canada, thus there is a shortage of the expertise necessary to deliver a comprehensive model of genomic medicine in Alberta.

- Accreditation bodies should acknowledge that all clinical genetic laboratories be supervised by board certified clinical laboratory geneticists (Canadian College of Medical Geneticists/American College of Medical Genetics and Genomics) and that this should be mandated as part of the certification process for all laboratory genetic labs in Alberta. All genetic laboratory reports should be interpreted and written by personnel trained on interpreting genetic results, thereby reducing patient harm and interpretation errors generated by labs with unqualified staff. There is also a shortage of genetic counselors, genetic nurses, metabolic dietitians, and other genetic health-care professionals.

1.3 Digital Infrastructure

As noted elsewhere, APL has implemented a pan-provincial electronic record for streamlining laboratory operations. Laboratory test results are integrated into Connect Care (clinical information system) and are available in the patient portal (except for predictive genetic tests). ‘Decision Support Tools’ guiding test ordering, test interpretation and clinical decision making would be a valued addition to Connect Care. As health systems move toward a model of personalized medicine, the incorporation of other patient specific “omic” information (e.g. proteomics, metabolomics) into a patient’s electronic medical record would also add value. This would allow the province and researchers to study patient populations from a broad perspective, as well as to understand in-depth phenotypes of individual patients, relating clinical presentation to molecular signatures – each approach with the goal to optimize care.

The Health Information Act (HIA) protects individuals’ health information and governs the collection, use and disclosure of an individual’s health data. With genomic medicine, tremendous value exists in the ability to analyze genetic/genomic data at a population and/or patient cohort level. However, access to patient data by researchers and innovators remains challenging in Alberta (and elsewhere), especially where patients have not provided consent to the use of their health data for research purposes. The HIA is a complex regulatory document and challenges lie in its interpretation and application. A recent report⁴ by the Alberta Virtual Care Coordinating Body, addresses the issue, advocating for health data interoperability and exchange standards for linking and using health data more effectively.

The ‘All for One: Canada’s Precision Health Partnership’, funded by Genome Canada and led by the six genome centres is designed to build regional genomics capacity, promote the equitable and ethical uptake of precision health tools, and address barriers to data sharing across provinces. A key component of the ‘All for One’ initiative is the development of a ‘Pan-Canadian Health Data Ecosystem’. Plans are for provinces to share clinical Genome-Wide Sequencing (GWS) data for complex rare diseases, noting that this project aligns with the previously mentioned ‘TiGeR’ project designed to repatriate genetic testing in Alberta for rare diseases. The ecosystem will enable data sharing, while respecting patient data and privacy, between institutions, across jurisdictional boundaries and between clinical and research settings, thus defining solutions to data sharing barriers and setting standards around GWS testing and analysis.

⁴ Affleck, E, Murphy, T, Williamson, T, Price, R, Wolfaardt, U, Price, T, Layton, A, Hamilton, B, Dean, S, Frazer, C, Chapman, A, Shute, R, West, Denman, M, Golonka, R, & Lindeman, C. (2023). Interoperability Saves Lives. (www.albertavirtualcare.org).

The addition of full genetic/genomic information on individuals would add value in terms of understanding disease and optimizing health for individuals and populations.

OPPORTUNITIES

- Lessons learned from the ‘All for One’ initiative, particularly around GWS data standardization and data sharing, can be applied to Alberta. The project is establishing data sharing agreements across different data custodians, and a federated database that will allow partners to view all GWS data (noting that patients are consented for such). There will be a clinical GWS database and a registry of GWS data for research (secondary use) purposes. An ‘ethical uptake’ of precision health tools will evolve from this initiative.
- Alberta has phenomenally rich health data assets across AHS and the Ministry of Health, on 4.5 million people (and more if one includes historical data). The addition of full genetic/genomic information on individuals would add value in terms of understanding disease and optimizing health for individuals and populations. It would have a significant impact on the health of Albertans in the future.
- AHS is currently moving from an Oracle data storage system, to a Cloud-based data storage system. This will increase capacity and robustness for data storage and will likely provide the storage capacity needed to support genetic data on patients.
- Tools for the analysis of ‘big data’ exist in AHS, including those that use AI/ML approaches. AHS recently invested in ‘Snowflake’ – a data software process that allows multiple autonomous databases to function as one ‘data federation’. This provides a single point of access to many data sources for front-end applications while retaining data ownership with the original data custodians. Data for research purposes is also de-identified in this software process. Snowflake will thus improve data sharing capabilities for both clinical and research purposes.
- Agencies like the Institute of Health Economics (IHE), and individual physicians and clinics, generally do not want to be the custodians of patient genetic data. They would rather have access through ‘secondary use’ regulations enacted by AHS and consistent with the HIA. Snowflake, AHS’ new software system, will likely be able to facilitate such.
- In the near future, Connect Care will more fully link with community and extended care data systems across Alberta (this is in the process). Challenges remain however, with linking Connect Care to primary care EMRs.

CHALLENGES

- Although genetic test results are available in Connect Care (with the exception of predictive genetic tests), more ‘test information’ needs to be provided in the system – e.g., including full testing data/results, information regarding test interpretation, clinical care decision support tools, supporting literature, and more.
- Storage capabilities required for GWS data on Alberta patients requires evaluation. Does Alberta have enough computing and storage power to undertake a broader model

of genomic medicine? AHS's new cloud-based data storage system may resolve this potential challenge, however more information is required.

- Sharing clinical and genetic data for research purposes remains a challenge, primarily because of patient privacy/security issues. This is in part due to cautious interpretations of the HIA. In addition, gene sequence data provides patient identifiable information, thus magnifying concerns around privacy. Better guidance on interpreting the HIA is needed, as well guidance on the pros and cons of implementing 'opt-out' consent processes ('opt-out' consent assumes consent is given unless there is active withdrawal from the patient). Federated databases, similar to the Canadian Distributed Infrastructure for Genomics (CanDIG) platform⁵, provide clinicians and researchers secure access to data, and may represent an opportunity to address data security issues.
- Current interpretations of the HIA seem to preclude the use of AI/ML analyses. This is because the HIA states that only minimal amounts of patient data can be used for research purposes. With AI/ML, the approach and robustness of the research requires that all available data be incorporated into the analyses. The addition of other provincial datasets to AHS and the Ministry of Health's data is an opportunity for Alberta.
- Although AHS has dedicated data analysts for research purposes, and supports aspects of external (e.g., academic) research, the delivery of care and the research analytics required for such, is AHS's priority. More human research capacity is required in AHS if/when patient genetic testing becomes more mainstream.
- 'Marrying' older datasets in AHS with new data remains a challenge, as does integrating and analyzing genetic data with clinical data.
- Understanding 'what the data means' can be challenging. However, building common data modules is helping.
- Patient apprehension about genetic testing related to data privacy concerns cause patients to decline genetic testing recommended by their physicians. Canada's Genetic Non-Discrimination Act⁶ prohibits non-consensual uses of genetic test results in a broad range of circumstances, such as use by employers or insurance companies, and should be communicated to the patients through healthcare providers.
- Public opinions and preferences regarding genetic data storage, ownership and sharing require investigation. For example, sharing genetic data with the private sector, and for research purposes, will be challenging. Public/patient engagement is also important in determining which genetic tests should be offered, and the impact that testing has on the lives of patients and their families.
- Disease codes in Connect Care do not capture all diseases and syndromes and are thus not always accurate. Physicians also code medical cases differently – thus there is a lack of disease coding standardization in some cases, thus affecting analyses.

Public opinions and preferences regarding genetic data storage, ownership and sharing require investigation.

⁵ <https://www.distributedgenomics.ca/> (accessed January 29, 2024)

⁶ Cowan JS, Kagedan BL, Graham GE, Heim-Myers B, Bombard Y. Health care implications of the Genetic Non-Discrimination Act: Protection for Canadians' genetic information. *Can Fam Physician*. 2022 Sep;68(9):643-646.

There could be more transparency regarding the process, criteria and outcomes for individual test assessments requested by physicians or by 3rd party entities.

2. OPERATIONS

2.1 Transparent Test Evaluation and Listing Process

In partnership with the IHE, APL developed and implemented a robust process and criteria for determining which clinical genetic tests (and other tests) are on the publicly funded test formulary. The ‘Rapid Health Technology Assessment (HTA) Prioritization Framework’ (available online)⁷ aims to formalize the existing provincial ‘Lab Formulary Committee’ (LFC) decision process and integrates a rapid HTA into the process with the intent to adopt publicly funded laboratory tests that have demonstrated strong clinical and economic value. The prioritization framework describes the main factors considered by the committee in a particular decision and provides a structured decision process to guide the deliberations. The assessment is individualized for each test, and the timeline and specific test reviews are not available publicly. Further, APL’s ‘Test Directory’ (available online)⁸ provides detailed information, guides and references for many of APL’s tests. This includes test and result codes, specimen collection requirements, specimen transport considerations, and test methodology. This information is for the ordering physician.

To request a test for inclusion on the formulary (and thus approval for test funding by AHS) the physician, patient, or private sector innovator fills out an intake form. The review process results in a recommendation and advice to AHS regarding funding of the test. APL’s review process also functions to decommission tests that are no longer warranted.

OPPORTUNITIES

- Alberta’s single point of entry into the testing system (e.g., APL) and its ‘Rapid Health Technology Assessment (HTA) Prioritization Framework’ is a plus for the province. With scarce resources (staffing, testing capacity), HTA is essential to optimizing resource use.
- Alberta currently runs multiple Molecular Pathology NGS panels (up to 130 genes per panel) for cancer patients yet these panels could be expanded for improved coverage and efficiencies. Some of the genes on the cancer panel signal treatment sensitivity for various cancer drugs on the drug formulary (e.g., drug and companion diagnostics).
- APL utilizes brokers to purchase tests that are not delivered in-house – e.g., Mayo Clinic has a relationship with APL. Bundling tests also affords cost efficiencies where possible.

CHALLENGES

- There could be more transparency regarding the process, criteria and outcomes for individual test assessments requested by physicians or by 3rd party entities. Performing HTA for drugs is more straightforward than for tests, as there is a more direct association with a drug and an outcome versus a test and an outcome. The economic and health

⁷ <https://www.albertaprecisionlabs.ca/tc/Page13931.aspx> (accessed January 16, 2024).

⁸ <https://www.albertaprecisionlabs.ca/> (accessed January 16, 2024).

value chain for genetic testing is complicated and requires ongoing consideration/ reassessment (e.g., there is a continuous ongoing ‘life cycle’ assessment of genetic (and other) tests). Fully understanding where the health gains lie is a challenge. Although complicated, the incorporation of other large data sets (e.g., socioeconomic data, ancestry) in the HTA process would help with creating greater testing equity and access for Albertans.

- The test evaluation process (e.g., to determine if a test will be included on the formulary) sometimes takes longer than what is optimal – for example, the process can take up to 1 to 2 years to complete and this is often not ideal for the patient. Implementing an expedited review stream would be beneficial.
- New FDA regulations for tests now require that test interpretation information is included in the regulatory approval request (aka in addition to technical information regarding the test).
- In Canada, it is encouraged, but not formally regulated, that provincial laboratory accreditation bodies and standards ensure that genetic tests give the right diagnosis through all steps of technical proficiency and interpretation, and non-accredited laboratories should not be reporting on genetic results.

2.2 Entry Point for Innovation and Role of Private Sector

Testing services offered by private sector facilities create opportunities for APL/AHS to access genetic tests and/or infrastructures that are not available in-house. As noted previously, APL does source some genetic (and other) tests from other health systems and/or private entities. Senior leadership in APL provides the point of contact for industry partners regarding such discussions/negotiations. Further, industry partners can request that their genetic tests be included on Alberta’s test formulary, via the test approval framework described above.

APL also partners with a platform – called the ‘Alberta Diagnostic Ecosystem Platform for Translation’ (ADEPT)⁹ - that provides a pathway for technology developers to access clinical samples and related clinical data, and for technology and process development and product validation. These assets are valuable for Alberta’s emerging health technology companies where sample access and product prototyping, trialing and validation are often the biggest barriers to commercial success. These resources also facilitate a pathway for the adoption of new technologies in Alberta, if warranted.

Senior leadership in APL provides the point of contact for industry partners regarding such discussions/negotiations.

⁹ <https://www.albertalabdiagnostics.ca/> (accessed January 16, 2024).

There are local (provincial) private sector opportunities for APL/AHS to consider working with, in order to increase capacities for genetic testing.

OPPORTUNITIES

- Working with ADEPT and APL early in the process of test development provides an opportunity to optimize tests for Alberta's needs and environment, thus increasing the potential for adoption of such tests into the system.
- Alberta funds both well-established and investigational genetic tests (e.g., from industry and/or academia) for some conditions. APL can release some funds for tests once an adoption decision is made, although tests with a large budgetary impact require further consideration by AHS.
- There are local (provincial) private sector opportunities for APL/AHS to consider working with, in order to increase capacities for genetic testing. In situations where APL sources genetic tests from accredited laboratories outside of the province, the quality of such tests have been validated 'head-to-head' against other laboratories, and the personnel interpreting the tests are board certified geneticists.
- There are private Alberta-based laboratories with CPSA accreditation that provide genome and exome testing. Local genetic testing companies may have testing roles as follows:
 - While it is not economically feasible to do genome/exome testing on every Albertan, patients can source and pay for a genome/exome test if they choose to do so (some health benefits programs will cover this).
 - In cases where publicly paid tests have not provided an answer and an expanded analysis is warranted, or in cases where results are required immediately (e.g., exome and pharmacogenomics testing on a child with seizures), local testing may provide value. In such cases, the above criteria (documented comparative test quality/robustness, and test interpretation by a board-certified geneticist) will need to be met in order for APL/AHS to use private services for clinical care.

CHALLENGES

- Test results generated from 'patient pay' in private sector labs are not always transferred into the provincial electronic health record and therefore they are not available to all care providers who care for the patient and act on the test results.
- Greater information sharing between industry and APL is required such that industry can be made aware of opportunities with APL. Information regarding what innovations would provide value to APL/AHS is lacking. This makes it difficult for innovators to plan their own development portfolios or provide useful information that might benefit healthcare planning and priority setting. A framework for bi-directional industry/APL engagement would add value in Alberta.

3. HEALTH ENVIRONMENT

3.1 Growth and Sustainability of Genomic Medicine

In an era of genomic medicine, healthcare supports need to be in place to finance genomic-based testing, integrate testing into clinical workflows, finance testing, and train providers. Alberta has already effectively integrated genetic testing in certain subdisciplines, such as the adoption of pharmacogenomics testing for KRAS mutations (implicated in lung cancer) and HLA genetic testing (implicated in epilepsy), providing a model for integration of larger-scale NGS. However, key barriers to the adoption of large-scale NGS (e.g., whole exome) into mainstream medical practice prevail, including results reproducibility; lack of robust evidence supporting clinical utility and cost expenditures in some disease areas; lack of standardized reporting for test results; and general affordability. In Alberta (and elsewhere) affordability issues are linked to the substantial human resources required to deliver broader genetic testing for the province. Gaps in the workforce needed to deliver a model of genomic medicine are described in section 1.2. Understanding the cost savings associated with genetic testing will help inform integration strategies.

OPPORTUNITIES

- Places where cost savings in the delivery of genomic medicine can be leveraged include:
 - Develop and implement large NGS panels and automate (as much as possible) the technical side of sequencing. AI tools are able to process complex genomic datasets including variant calling, genome annotation and variant classification, and phenotype-to-genotype correspondence, reducing the time commitment of specialists.
 - Ensure appropriate stewardship of resources via the test ordering process by educating physicians regarding test use criteria and including decision support tools in the electronic system to guide genetic testing (note: some tests do have decision support tools to accompany their use).
 - Build and adhere to ‘life cycle assessment’ models for genetic tests that define who can receive the test, criteria for testing, and the desired outcomes and targets. This can be done within APLs software systems and should be done for non-genetic tests, to reduce wastage in the system due to inappropriate test ordering.
- Undertake a comprehensive literature analysis of healthcare costs (savings and expenditures) associated with genetic testing to direct the appropriate context of use for each genetic test.

CHALLENGES

- Alberta’s specialized infrastructure, workforce, and bioinformatics needs will continue to climb as the province undertakes more genetic testing.

In an era of genomic medicine, healthcare supports need to be in place to finance genomic-based testing, integrate testing into clinical workflows, finance testing, and train providers.

Alberta may benefit by developing province-wide education standards for genetic testing, across a variety of disciplines.

3.2 Education of Patients and Care Providers

A key barrier to the full adoption of genomic medicine is the training (up-skilling) and resources required across all “touch points” that genetic testing affects (as noted elsewhere). This includes training of primary care physicians, nurses, nurse practitioners, physician assistants, clinical laboratory scientists and technicians, social workers, and more. Currently, the volume of genetic testing in the field of oncology dwarfs that for other disease areas, noting that different clinical specialties have different requirements/needs for genetic testing. Alberta may benefit by developing province-wide education standards for genetic testing, across a variety of disciplines. As noted above, care providers need to understand the appropriateness of genetic testing, and then how to interpret and action the test results.

OPPORTUNITIES

- Develop and implement educational programs for care providers regarding genetic tests – including information regarding the science, testing appropriateness, and the interpretation and actions to undertake based on test results. The SCN’s are a framework that can be leveraged to roll out such programming across disease specialty areas.
- Work with Alberta’s medical schools and regulatory bodies to ensure that the medical school curriculum adequately addresses training on genomic medicine.
- Develop educational materials and opportunities for patients, informing them about genetic tests including how to navigate access to such tests in Alberta’s health system.

CHALLENGES

- Recent studies have demonstrated cost savings and beneficial health outcomes from pharmacogenomics testing for depression with predicted savings of \$956 million over 20 years to BC’s healthcare system.¹⁰ Pharmacogenomics examines a patient’s genetics to predict drug responses and inform treatment management. Medical students, primary care physicians, pharmacists and the public all require education on this topic. Education needs to introduce broad concepts related to pharmacogenomics with subspecialists (i.e., pharmacists) having a deeper understanding around the pharmacokinetics and pharmacodynamics of drugs.

¹⁰ Ghanbarian S, Wong GWK, Bunka M, Edwards L, Cressman S, Conte T, Price M, Schuetz C, Riches L, Landry G, Erickson D, McGrail K, Peterson S, Vijn R, Hoens AM, Austin J, Bryan S. Cost-effectiveness of Pharmacogenomic-Guided Treatment for Major Depression. CMAJ. 2023 Nov 14;195(44):E1499-E1508.

WHAT'S NEXT

The Chief Scientist at the World Health Organization (WHO) described genomic technologies as driving some of the most ground-breaking research happening today¹¹.

According to the former President of the Canadian Medical Association, Dr Cindy Forbes, six of every ten Canadians will be affected during their life by a health problem that is genetic in whole or in part¹². Indeed, revealing individual genomic sequences and genetic variations has the potential to improve patient health care through faster diagnoses and targeted preventions and treatments. The 'State of Readiness in Canada Progress Report' (May 2023)¹³ that evaluated five Canadian healthcare regions (BC, AB, IN, QC, NS) for their readiness to implement, deliver and regulate genetic testing, ranked Alberta ahead of other jurisdictions. Although ranked above other jurisdictions, Alberta requires additional conditions to optimize genome-based testing services in our healthcare system. There is broad interest from a range of stakeholders in helping Alberta realize the benefits to patients from the adoption of broader genetic testing. The December 2023 workshop 'Genomics at Work for Alberta's Healthcare System' was an opportunity to engage representatives from across Alberta's health delivery and innovation ecosystem to build a better understanding about the current state of genome-based testing as well as to discuss both opportunities and barriers in the broader adoption of genome-based testing into Alberta's routine care.

In many regards, Alberta's healthcare system is positioned to adopt a more comprehensive model of genetic testing into routine clinical practice. Alberta's single public laboratory services provider (APL), along with its formalized process for evaluating and incorporating tests into the system are an advantage. Further, Alberta's pan-provincial data infrastructure is evolving to a stage where the incorporation of patients' genetic data is feasible and could be leveraged to improve care and costs (in clinical areas where evidence supports it). Areas that require effort and resources include establishing the human resource capacity and expertise necessary for genetic testing (clinical geneticists, laboratory geneticists and technologists, genetic counsellors, etc.) and educational programming regarding genetic testing for care providers and patients. These can be costly endeavors, and the challenge is compounded by the national shortage of workforce available with genetic expertise (both clinical and laboratory). Further, while APL is progressive in its innovative culture, there is an opportunity to establish a bidirectional model for engagement with private industry partners which could create benefits for both parties and ultimately for Albertans. Finally, data privacy and sharing for research purposes continues to be a challenge that will require continued attention and effort to mitigate.

¹¹ World Health Organization, 2022 Report: <https://www.who.int/news/item/12-07-2022-who-science-council-launches-report-calling-for-equitable-expansion-of-genomics>.

¹² Evidence: Bill S-201, an Act to prohibit and prevent genetic discrimination. 42nd Parliament, 1st Sess., meeting no. 37. Ottawa, ON: House of Commons Standing Committee on Justice and Human Rights; 2016. Available from: <https://www.ourcommons.ca/DocumentViewer/en/42-1/just/meeting-37/evidence>. (accessed January 31, 2024)

¹³ Husereau D, Villalba E, Muthu V, Mengel M, Ivany C, Steuten L, Spinner DS, Sheffield B, Yip S, Jacobs P, et al. Progress Toward Health System Readiness for Genome-Based Testing in Canada. 2023. ISBN 978-1-7389568-0-7.

The Genomics at Work for Alberta's Healthcare System workshop provided a valuable forum for improving understanding of the current state of genome-based medicine in Alberta. Participants representing a broad range of health innovation system players engaged in a collaborative, open and frank discussion on the most pressing barriers to overcome for broader adoption of genome-based testing in Alberta's healthcare system. Genome Alberta will continue to support efforts, in partnership with the broader health innovation community in Alberta, to address the identified barriers through the innovation and research funding programs we deliver. Knowledge and insights gained through the workshop will help to inform future research and innovation programs that enable greater benefits to Albertans through adoption of genomics medicine.

Knowledge and insights gained through the workshop will help to inform future research and innovation programs that enable greater benefits to Albertans through adoption of genomics medicine.

ACKNOWLEDGEMENTS

Genome Alberta would like to thank Don Husereau for his time and expertise in chairing the workshop and facilitating the discussion.

We also would like to express our appreciation to the panelists and those who attended and contributed their knowledge and insights on how to move forward. Additionally, we would like to acknowledge and thank Deborah James, eDJe Consulting Inc. for capturing the summary of the workshop discussion in the first draft of the report.

LIST OF PARTICIPATING ORGANIZATIONS

Alberta Centre for Advanced Diagnostics
Alberta Diagnostics Ecosystem Platform for Translation
Alberta Health
Alberta Health Services
Alberta Innovates
Applied Pharmaceutical Innovation
Alberta Precision Laboratories
AstraZeneca
BioAro Inc.
Discovery DNA
eDJe Consulting Inc.
Genome Alberta
Government of Alberta
Institute of Health Economics
OncoHelix
Nanostics Inc.
Roche
Sequence2Script Inc.
Thermo Fisher Scientific
University of Alberta
University of Calgary
University of Ottawa



403.210.5275
info@genomealberta.ca
3512-33 Street NW Suite 200
Calgary, Alberta T2L 2A6 Canada