# BACKGROUNDER

## Details About the Eight Genomic Research Projects

The eight research projects are part of Genome BC and Genome Alberta's **Healthy Outcomes through Genomic Innovations program**. The key priorities of the program are:

- expanding genomics-based testing and technologies into routine clinical care by addressing system barriers;
- ensuring equitable access to testing in both rural and urban settings; and
- integrating genomics data into digital health infrastructure for better patient management.

Here's a list and the details of the eight research projects:

### Pediatric innovations in genomics: let everyone be tested

**Researchers:** Bruce Carleton (BC); Gregory Guilcher (AB)
**B.C. organizations:** Provincial Health Services Authority (PHSA), BC Children's Hospital, University of British Columbia

***Briefly:*** *This project will expand pharmacogenetic testing to predict severe chemotherapy side effects in children, making the testing widely accessible and improving safety in pediatric oncology. By embedding these advancements into the health-care system, the project aims to enhance treatement safety and long-term outcomes for pediatric cancer patients*.

**Complete summary:** This project aims to develop, validate and implement an expanded pharmacogenetic test panel to predict the risk of seven severe chemotherapy-induced adverse drug reactions (ADRs) in pediatric cancer patients. By integrating precision health strategies into pediatric oncology, the project seeks to improve treatment safety and effectiveness for children undergoing chemotherapy.

Building on previous research, which successfully introduced pharmacogenetic testing for three ADRs at nine pediatric oncology academic health centres across Canada, this initiative will expand the test panel to include additional, highly predictive genetic markers. The project will also assess the impact of this expanded testing with clinicians, patients and families in British Columbia and Alberta.

The project has two primary objectives. The first is to enhance the existing test panel by incorporating newly identified genetic markers that improve the ability to predict chemotherapy-induced ADRs. The second is to develop a long-term sustainability plan to transition these pharmacogenetic tests from research-based tools to standard clinical practice. This transition will

enable routine use by health-care providers, ensuring widespread accessibility for pediatric cancer patients in B.C. and Alberta.

Providing patient-specific ADR risk information is essential for two reasons: it helps families understand their child's individual risk of severe side effects compared to other children receiving the same treatment, and it facilitates informed discussions between parents and clinicians about strategies to prevent or manage these risks.

In addition to expanding the test panel, the project will introduce an education program to support healthcare providers in integrating pharmacogenetic testing into clinical practice. A detailed sustainability plan will outline key steps and timelines to establish pharmacogenetic testing as a standard of care in both provinces.

## Overcoming barriers for rapid adoption of donor-derived cell-free DNA for surveillance of kidney allograft injury in Canada

**Researchers:** James Lan (BC); Paul Keown (BC); Michael Mengel (AB)
**B.C. organizations:** Vancouver Coastal Health, Providence Health Care, University of British Columbia

***Briefly:*** *This project aims to implement a new test for early detection of kidney organ injury in B.C. and Alberta, enhancing the monitoring of transplant health and improving patient care.*

**Complete summary:** Kidney transplantation is the most effective treatment for kidney failure, yet more than 25% of kidney transplants in Canada fail within 10 years due to rejection or other injuries. Currently, doctors monitor kidney health using a blood test that measures creatinine levels, but this method only detects problems once significant damage has already occurred, limiting opportunities for early intervention.

A newer blood test, known as donor-derived cell-free DNA (dd-cfDNA), has shown promise in detecting kidney injury much earlier, before creatinine levels become abnormal. Despite its benefits, this test is currently only available through specialized laboratories in the United States, making it costly and inaccessible for many Canadian patients.

This project aims to bring dd-cfDNA testing to British Columbia and Alberta, improving access to this advanced diagnostic tool. The project team will first work with local transplant laboratories to ensure that the test performs at the same high standard as U.S.-based labs. Once validated, the test will be formally introduced for clinical use in both provinces to support patient care.

To assess the impact of dd-cfDNA testing, the project will evaluate key measures such as the speed of test results, equitable access for patients and the test's influence on treatment decisions. A comprehensive economic analysis will also be conducted to determine the most cost-effective way to implement the test in the healthcare system.

By collaborating with national leaders and patient partners, the project team will share findings and best practices with other transplant centres across Canada. This initiative aims to accelerate the adoption of dd-cfDNA testing nationwide, ultimately improving outcomes for kidney transplant patients by enabling earlier detection and intervention.

## Comprehensive cancer gene fusion detection for B.C. and Alberta

**Researchers:** Tony Ng (BC); Erik Nohr (AB)
**B.C. organizations:** Vancouver Coastal Health Research Institute, Vancouver General Hospital

***Briefly:*** *This project will roll out new testing in B.C. and AB to better detect fusion genes. This will lead to more accurate, reliable testing and better cancer care.*

**Complete summary:** Fusion genes, formed when two genes merge, are key drivers of cancer growth and serve as important targets for diagnosis and treatment. Identifying these genes is critical, particularly for cancers such as sarcomas and pediatric tumours. The discovery of fusion genes has led to life-saving treatments, including ALK inhibitors for certain lung cancers and NTRK inhibitors for tumours with NTRK fusions.

Traditional testing methods, such as fluorescence in-situ hybridization (FISH), can only detect common fusion genes, often missing rarer but clinically significant ones. More advanced technologies, such as next-generation sequencing (NGS), provide a more comprehensive view, identifying nearly all types of fusion genes. Among these, hybrid capture RNA sequencing is one of the most effective approaches, capable of detecting over 99% of gene fusions.

Currently, testing methods for fusion genes vary between British Columbia and Alberta. In B.C., many laboratories rely on older techniques such as FISH or small-scale NGS tests, which can miss critical fusions, resulting in delays or the need for testing outside the province. In contrast, Alberta uses broader NGS panels, providing more comprehensive detection.

To address these inconsistencies, a new partnership between B.C. and Alberta aims to enhance fusion gene testing in both provinces. Vancouver General Hospital in B.C. will introduce a hybrid capture RNA sequencing test capable of detecting over 500 fusion genes. Alberta Precision Laboratories will expand its NGS capabilities with a custom hybrid capture Pan-Cancer Fusion Panel. Through the exchange of tumour samples, both provinces will ensure their testing methods are accurate, reliable and consistent.

This collaboration will improve the speed, accuracy and accessibility of fusion gene testing for cancer patients in both provinces. By standardizing testing approaches, the project aims to enhance cancer care and foster continued cooperation in cancer diagnostics. The findings will be shared widely to help advance fusion gene testing across Canada and beyond.

## “Mainstreaming” genetic testing for non-ischemic cardiomyopathy in Western Canada

**Researchers:** Thomas Roston (BC); Omid Kiamanesh (AB)
**B.C. organizations:** St. Paul’s Hospital, University of British Columbia, Provincial Health Services Authority, Providence Health Care

***Briefly:*** *This project will empower heart failure cardiologists in B.C. and Alberta to order genetic tests for hereditary heart conditions, improving early diagnosis and treatment for patients and their families. It has the potential to improve heart failure management, reduce wait times and ultimately save lives.*

**Complete summary:** Heart muscle disorders are a leading cause of heart failure, a life-threatening condition that can lead to dangerous abnormal heart rhythms (arrhythmia) and fluid buildup in the body (edema). In British Columbia and Alberta, patients with heart failure receive specialized care in Heart Function Clinics (HFCs), where early diagnosis and treatment can prevent severe complications, disability and death.

In some cases, particularly in younger patients, heart failure is caused by inherited genetic abnormalities in the heart muscle. These conditions can be identified through a simple blood or saliva test, allowing for more precise treatment and the early diagnosis of at-risk family members. However, in B.C. and Alberta, genetic testing for heart failure is only available through highly specialized programs, which require referrals and often have wait times of up to three years. As a result, many HFCs are unaware of these tests, or hesitant to refer patients due to these significant delays.

This project aims to integrate genetic testing directly into HFCs, enabling cardiologists to order genetic tests for their patients without the need for specialized referrals. By equipping HFC providers with the knowledge and resources to implement genetic testing, patients can receive faster diagnoses and earlier treatment. Additionally, at-risk family members will be identified more quickly, allowing for preventive care that could reduce the risk of severe heart failure and sudden cardiac events.

Beyond improving individual patient care, the project will generate evidence to support broader healthcare system changes. By demonstrating the benefits of integrating genetic testing into HFCs, the findings will help inform healthcare leaders and policymakers about the need for expanded access to genetic testing.

## Development and testing of an at-home lung cancer-screening test

**Researchers:** David Wishart (AB), William Lockwood (BC)
**B.C. organizations:** BC Cancer, Provincial Health Services Authority

***Briefly:*** *This project will develop and validate an affordable, at-home lung cancer screening test. By making diagnostics more accessible and reliable, this test will enable early detection and save lives.*

*The long-term vision is to integrate this screening test into the diagnostic framework for Alberta Health Services and the B.C. health-care system, improving access for high-risk populations, including Indigenous, rural and remote communities. Transitioning from low-dose CT (LDCT) scans to this metabolomics-based test could result in significant healthcare cost savings—potentially over $1.5 billion per year—while saving more than 15,000 Canadian lives annually.*

**Complete summary:** Lung cancer remains one of the deadliest cancers in Canada, with nearly 50% of cases diagnosed at Stage IV, where survival rates are extremely low. Early detection is critical to improving patient outcomes, yet current screening methods are costly, require specialized imaging and are not easily accessible to all Canadians.

This project aims to develop and validate a blood plasma-based laboratory-developed test (LDT) for early lung cancer detection and create an affordable, at-home plasma collection kit to enable mail-in screening. The proposed test aims to measure 10 specific metabolites in a process that takes just five minutes per sample and is designed to be cost-effective, with an estimated price of less than $35 per test. Ensuring the stability of plasma samples collected at home will be a key focus to guarantee reliable results comparable to those obtained in a clinical setting.

Key project milestones include:

- developing a mass-spectrometry (MS)-based LDT for detecting early-stage lung cancer;
- designing and testing an at-home plasma collection and stabilization kit for mail-in screening;
- Demonstrating that biomarker readings remain consistent between mailed-in samples and fresh venous blood plasma collected in a clinical setting;
- conducting a pilot study with 260 at-home lung cancer screening kits to evaluate feasibility; and
- taking the first steps toward Health Canada approval for a plasma-based lung cancer screening test.

By offering an omics-based, patient-centred, and cost-effective screening solution, this initiative aligns with the Healthy Outcomes Program's goals, providing a transformative approach to early lung cancer detection and treatment across Canada.

## Translation of BSIDx - a rapid diagnostic platform for bloodstream infections

**Researchers:** Ian Lewis (AB); Michael Mengel (AB)
**B.C. contributor:** Marthe Charles, Vancouver Coastal Health
**B.C. organizations:** Vancouver Coastal Health, Provincial Health Services Authority

***Briefly:*** *This project aims to evaluate and implement a rapid diagnostic test for bloodstream infections in Alberta and B.C., reducing diagnostic turnaround time to enhance treatment and save lives.*

*The BSIDx test is a fast lab test that helps identify infections in the blood and determines which medications can effectively treat an infection. By reducing diagnostic wait times by more than 30 hours, BSIDx has the potential to significantly improve treatment decisions for life-threatening infections. If implemented nationwide, this innovation could save over 2,250 lives each year while also reducing health-care costs associated with prolonged hospital stays and inappropriate antibiotic use.*

**Complete summary:** Bloodstream infections (BSIs) are a major health threat, with more than 500,000 cases and over 90,000 deaths annually in North America. Each hour of delay in administering the correct antibiotics increases a patient's risk of death by 7%. However, current diagnostic methods take 24 to 48 hours—or longer—to identify which antibiotics will be effective, leaving clinicians to make critical prescribing decisions with limited lab data. Faster diagnostic tools are needed to reduce errors in antibiotic selection and save lives.

To address this issue, researchers at the University of Calgary and Alberta Precision Laboratories have developed BSIDx, a metabolomics-based diagnostic tool that can provide test results in just five hours. This project aims to evaluate the performance of BSIDx in provincial laboratories and collect the data necessary to support its adoption in Alberta and British Columbia.

Key objectives of the project include:

- refining the BSIDx design to align with the specific needs of provincial healthcare systems;
- conducting direct comparisons between BSIDx and current testing methods to assess accuracy, sensitivity, specificity, and turnaround time in clinical lab settings; and
- performing economic and health impact analyses to evaluate the potential benefits of BSIDx adoption.

The study's primary outcomes will include a comprehensive assessment of BSIDx performance and an evaluation of its impact on patient care and healthcare costs. A key deliverable will be an evidence package for submission to the diagnostic test review committees in Alberta and BC, which oversee the approval of new diagnostic technologies.

## Closing GAPS: Genome approach to preventing spread of healthcare-associated infections through innovation and economics

**Researchers:** Matthew Croxen (AB), Linda Hoang (BC)
**B.C. organizations:** BC Centre for Disease Control Public Health Laboratory, University of British Columbia

***Briefly:*** *This project will assess and compare reactive versus proactive genomic surveillance of multi-drug-resistant bacteria in B.C. and Alberta, evaluating the cost-benefit and effectiveness of early intervention strategies.*

*By assessing the costs and benefits of a proactive genomics-based surveillance strategy, the project will provide critical data to support more effective infection control policies. The findings will not only inform best practices for CPO surveillance in Alberta and B.C. but also contribute to national and international efforts to combat drug-resistant infections more effectively.*

**Complete summary:** Multi-drug resistant infections are a growing global health crisis, responsible for millions of deaths worldwide. In Canada alone, an estimated 14,000 deaths in 2018 were linked to drug-resistant bacterial infections. Hospital-associated infections, particularly those caused by drug-resistant bacteria, pose a significant burden on healthcare systems, leading to severe complications, increased mortality and rising health-care costs.

Among the most concerning drug-resistant bacteria are carbapenemase-producing organisms (CPOs), which can cause life-threatening infections such as pneumonia, meningitis and sepsis. The World Health Organization (WHO) has identified CPOs as a critical global health threat due to their rapid spread and limited treatment options. Unlike many other hospital-acquired infections, CPOs can persist in healthcare environments, such as sinks and drains, making their spread difficult to control through traditional infection prevention measures.

Both the Alberta Public Health Laboratory (Alberta ProvLab) and the BC Centre for Disease Control Public Health Laboratory (BCCDC PHL) have established routine genomic surveillance programs to track and better understand CPO transmission. This project will evaluate these surveillance programs from a health economics perspective, comparing the current reactive approach—responding to individual cases as they arise—to a proactive model that detects early transmission events and prevents further spread.

## Pharmacogenetics data standardization to enhance findability, accessibility, interoperability, and reusability (PGx-EFAIR)

**Researchers:** Chad Bousman (AB)
**B.C. contributors:** J9 Austin, Stirling Bryan, Louisa Edwards, Dan Simic, Ifan Kuo
**B.C. organizations:** Provincial Health Services Authority, University of British Columbia, Vancouver Coastal Health Research Institute, Ministry of Health

***Briefly:*** *This initiative will standardize pharmacogenetics data reporting in Alberta and B.C., improving its integration into electronic medical records to support personalized medicine and broader adoption of genetic testing.*

**Complete summary:** Pharmacogenetics (PGx) testing enables health-care providers to personalize medications and dosages based on a patient's genetic profile, improving treatment effectiveness and reducing side effects. Despite its clinical and economic benefits, PGx testing has not been widely adopted, which is partly due to challenges in standardizing how genetic test results are reported and integrating these results into electronic medical records (EMRs).

Laboratories use different reporting formats, making it difficult for healthcare providers to access and interpret PGx data in a consistent and meaningful way.

The PGx-EFAIR project aims to address this issue by developing solutions to standardize and integrate PGx data into EMRs in Alberta and British Columbia. By collaborating with provincial governments, health authorities and industry partners, the project will create a unified format for reporting PGx test results, automate data standardization and establish machine-readable messaging protocols to ensure seamless integration with health-care systems. These efforts will allow PGx data to be easily shared and applied in clinical settings.

This initiative aligns with Canada's Pan-Canadian Interoperability Roadmap and legislative efforts such as Bill C-72, which focus on improving data exchange in healthcare. By building on existing digital health initiatives in Alberta and B.C., the project will ensure that PGx data is structured, accessible and interoperable across health-care systems.

The project will establish a standardized format for PGx test results, develop software to automate the translation of genetic test data and validate data exchange protocols for compatibility with EMRs. These advancements will improve the accessibility and usability of PGx data, accelerating its adoption in clinical care. While initially focused on Alberta and B.C., the project has the potential to serve as a model for integrating PGx testing into healthcare systems across Canada and internationally, supporting the broader adoption of precision medicine.

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