The Canadian path from discovery to implementation of personalized medicine approaches

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November 20, 2018
Towards the Personalized Health Initiative

2012

**Personalized Medicine Initiative**
- Enhance health outcomes through patient stratification approaches by integrating evidence-based medicine and precision diagnostics into clinical practice
  - $240M ($85M from CIHR)
  - 110 competition and application partners, including Genome Canada

**eHealth Innovation Initiative**
- Enhance health outcomes and health care delivery, through the implementation, evaluation and scale-up of eHealth innovations
  - $34.4M ($16.2M from CIHR)
  - 77 application partners

2016

**Personalized Health Initiative**
- Drive evidence-based implementation of PH that will identify solutions that can contribute to more cost-effective and sustainable healthcare
- Investments currently planned: $82M ($61M from CIHR)
- Alignment with IC PerMed
Genomics and Personalized Health:
2012 Large-Scale Applied Research Project Competition

CIHR/Genome Canada partnership: One of the most significant public sector investment in PM

• Research projects span various areas including cancer, rare diseases, epilepsy, inflammation, HIV, cardiovascular disease and autism

Investment: over $165M

• $68.8M CIHR/GC investment with more than 1:1 match from outside sources

Projects funded: 17

• 15 large-scale applied research projects with integrated genomics, ethical, environmental, economic, legal, social (GE3LS) components
• 2 large-scale GE3LS research projects

CIHR’s contribution was made available by the following Institutes (IG, ICR, ICRH, INMHA, III, INMD & IHSPR) & Initiatives (HIV/AIDS & Breast Cancer)
Canadian Success Stories

- International consortium grouping samples and expertise on high grade astrocytomas
- Identified recurrent driver mutations affecting DNA structure, now part of WHO test recommendations
- Developed an oncopanel now used in clinical trials and CLIA certified

Recruited over 3000 patients and family members to study
- Studied 637 different rare diseases
- Have provided a diagnosis to over 1000 patients
- Have identified 85 novel rare disease genes
- Are developing three experimental therapies
- Contribute to international data sharing standards
Canadian Success Stories

• PErsonalized Genomics for prenatal Aneuploidy Screening USing maternal blood
• Develop evidence-base to make informed value-based decisions about implementation of genomics-based non-invasive prenatal testing (NIPT)
• Recruited 3,819 pregnant woman
• Demonstrated that implementation of NIPT as a second tier test, followed by amniocentesis if NIPT is abnormal, would be cost neutral in Québec
• Tackled the ethical, social and legal implications of implementing NIPT in Canada
• Collaborated with a working group on NIPT from the government of Québec
Rare Diseases Research Catalyst Network

Creation of a national network in 2014 organized to:

• Identify Canadian model expertise relevant to newly discovered disease genes
  ▪ Funded research projects focus on functional validation
• Enhance clinical translation
  ▪ Develop and implement innovative knowledge translation strategies/activities to link clinical genetics & model research communities

Investment: $2.3 M CIHR-IG in partnership with GC

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<tr>
<th>Principal Investigators</th>
<th>Title of Project</th>
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<tr>
<td>Philip A. Hieter (UBC)</td>
<td>Canadian &quot;Rare Diseases: Models &amp; Mechanisms&quot; Network (RDMM)</td>
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<td>Kym Boycott (CHEO)</td>
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<td>Janet Rossant (SickKids)</td>
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NANS-mediated synthesis of sialic acid is required for brain and skeletal development

van Karnebeek et al, Nature Genetics 2016; 48:777-784
Towards a More Efficient Healthcare Ecosystem

Success will require integrating the perspectives of policy makers, health technology assessments, health care providers, regulators, researchers, and patients.

Opportunities:
- Recognition of the importance and urgency by the advisory panel on health care innovation.
- Awareness of the increasing importance of PM and genomic approaches in several provinces.
- Potential synergies with ongoing and developing initiatives (e.g., epigenetics, eHealth, microbiome, ICT).

Gaps:
- Impact and demonstration of PM value still in its infancy.
- Many biomarkers, “omic” and clinical data are not used.

Foundation:
- Early signs of successful implementation (e.g., specific cancers, rare diseases, pharmacogenetics).
- Strong national and international positioning.

Significant investments made in the past few years by GC/CIHR PM Signature Initiative, SPOR support unit (NL) and through initiatives in several provinces.
Where are we?

Provincial & Territorial Lab Standards

Provincial & Territorial Research

Formulary Discussions

INESSS

Healthcare Priorities

National Policy Interests

Patient / Professional Groups

Research Priorities

CIHR/NSERC/Genome Canada

National Lab Standards

Product R&D

PMPRB Product

Submissions

Surveillance

CADTH

Health Canada

PHAC

Federal

Indirect

Provinces & Territories

T. Ryan Sigouin, Health Canada adapted by Inga Murawski CIHR-ICR and Etienne Richer CIHR-IG
How bold do we want to be?

**Improved Health Outcomes**
- Personalized medicine and decision support
- eHealth patient empowerment
- Scale-Up innovative processes
- New data on PROMs/patient preferences
- Cost-effective solutions

**Data Integration**
- Social Media
- EMR
- Administrative Data
- Biometrics & Big Data
- Genomics

**Harmonization of Phenotype Creation**
- Emphasis on integration of various data sources to facilitate better outcomes.

**Diagnosis, Predictive and Behavioral Analytics**
- Unexplained Findings – Reverse Translation
- Unexpected Outcomes

**Explanatory Data Collection**
- Precise Treatment Recommendations
- Motivation Behavioral Changes

**Behavioural Biometrics & Big Data**
- Improved Health Outcomes

**Discoveries for life**
- CIHR IRSC

A wave of change:
Implementing Precision Medicine in BC

Dr. Catalina Lopez-Correa
Chief Scientific Officer and VP Sectors
The genomics enterprise in Canada

- 6 regional Genome Centres
- 10 Genomic Innovation Network Nodes
- 210+ large-scale research projects/initiatives funded across all sectors
- 45 High Qualified Personnel on average, per project
- 58+ companies created or enhanced
Precision Health Care

Genome BC cumulative investment in **145 projects** • **53 active**
Total **$352.5M**: Genome BC **$69.3M** with co-investment **$283.2M**

**Prevention**
- First Nations Biobank

**Diagnosis**
- Prenatal Screening
- RapidOMICS

**Treatment**
- Genomics for Pharmacists

**Prognosis**
- Biomarkers for COPD Management

DISCOVERIES FOR LIFE
2017 Large Scale Applied Research Project Competition Stats

6/15 awards went to BC led projects

2/15 awards went to BC co-led projects

38.5% of funds invested by Genome Canada and Canadian Institutes for Health Research

$80.3M Total investments when combined with Genome BC's contribution
• Silent Genomes: Improving diagnostics for Indigenous children
  Laura Arbour, Nadine Caron, Wyeth Wasserman
  Total Budget: $10,399,812

• Go-PGx: Reducing adverse drug reactions (ADR) for children with cancer
  Bruce Carleton, Colin Ross
  Total Budget: $10,517,507

• CanPREVENT: Preventing rejection and premature kidney transplant loss
  Paul Keown, Ruth Sapir-Pichhadze, Timothy Caulfield, Stirling Bryan
  Total Budget: $9,671,756

• Deciphering relapsed lymphoid cancers to improve patient management
  Christian Steidl, Marco Marra, David Scott
  Total Budget: $11,926,360

• Childhood asthma and the microbiome: The CHILD*study
  Stuart Turvey, Michael Kobor, Brett Finlay, Padmaja Subbarao
  Total Budget: $9,142,486

• GenCOUNSEL: Optimizing genetic counselling for clinical implementation
  Alison Elliott, Bartha Knoppers, Larry Lynd, Jehannine Austin
  Total Budget: $4,237,284
Genome BC Health Strategy

- Consultations
- Health Sector Strategy
- Draft Health Sector Strategy
- Stakeholder Research
- Establish Health Sector Task Force
- Infectious Disease
  - BC Centre for Disease Control
- Rare Diseases
  - BC Children’s Hospital
- Genome British Columbia Health Strategy
- Exemplars
- Cancer
  - BC Cancer Agency
  - C4R
- Pharmacogenomics
  - UBC
BC pharmacists leading precision medicine

Researcher: Corey Nislow

Approximately 50% of all emergency department visits each year are due to adverse reactions to medications in adults aged 50 and over.

Across BC, 33 community pharmacies have taken part in North America’s first project to implement pharmacogenomics allowing them to:

- Extract DNA from saliva, sequence and analyze the DNA
- Prescribe the right drug to the right patient at the right time and the right dose based on their genomic information
Transformational potential of Pharmacogenomics

Clinician Centered

Data belongs to HCS
Access to care
Regulatory
Payments

Health care system transformation

Patient Centered

Data belongs to patient
APPs/Mobile devices
Informed consent
Prevention
Crosscutting areas to accelerate implementation

1. Education of Health Care Providers
   - Cancer
   - Infections Disease
   - Rare Diseases
   - Pharmacogenomics

2. Big Data
   - Data integration
   - Data sharing

3. Capacity
   - Biobanking
   - Sequencing
   - HQP

4. Access
   - Policy
   - Reimbursement
   - Economics
   - Equity

Clinical Implementation
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