CIHR IRSC
Canadian Institutes of Health Research
Instituts de recherche en santé du Canada

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

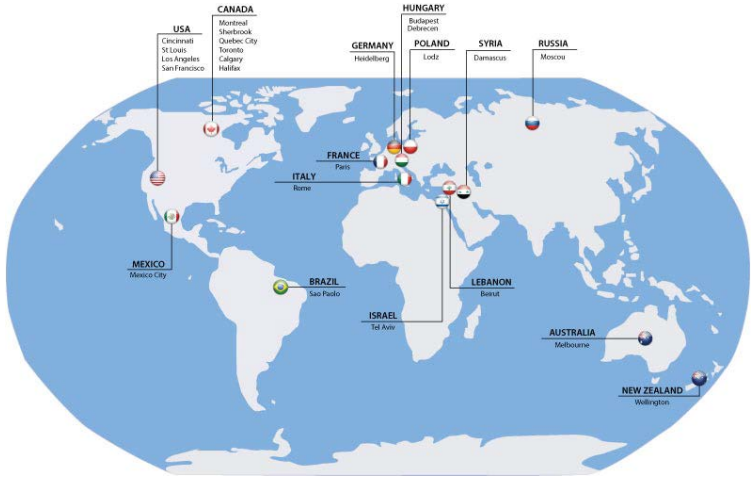
- 15 large-scale applied research projects with integrated genomics, ethical, environmental, economic, legal, social (GE3LS) components
- 2 large-scale GE3LS research projects

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

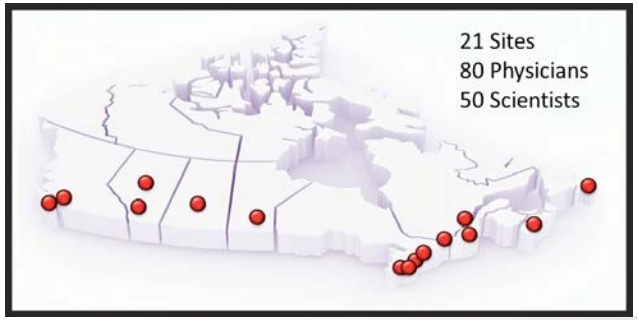
ICHANGE | International Childhood Astrocytoma Novel Genomics and Epigenomics Consortium

- 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



CARE forRARE

- 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:

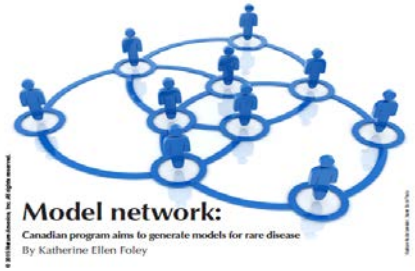
- Identify Canadian model expertise relevant to newly discovered disease genes
  - Funded research projects focus on functional validation
- Enhance clinical translation
  - Develop and implement innovative knowledge translation strategies/activities to link clinical genetics & model research communities

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

Principal Investigators	Title of Project
Philip A. Hieter (UBC) Kym Boycott (CHEO) Janet Rossant (SickKids)	Canadian "Rare Diseases: Models & Mechanisms" Network (RDMM)

http://webapps.cihr-irsc.gc.ca/cfdd/db_search?p_language=E&p_competition=201404RCN

RARE DISEASES
MODELS and MECHANISMS
NETWORK



Nature Medicine 2015; 21: 1242

QUICK STATISTICS

- Genes Added: 6643
- Number of researchers registered: 499
- Number of researchers registered with genes: 323
- Number of genes added unique: 5604
- Number of Connections Supported: 78

FUNDERS

CIHR IRSC
Canadian Institutes of Health Research | Instituts de recherche en santé du Canada

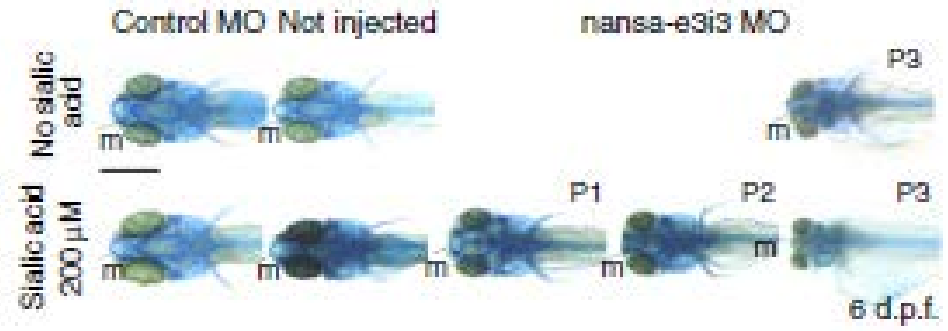
GenomeCanada

Genome BritishColumbia

dravet.ca

NIH Undiagnosed Diseases Program

RARE DISEASES MODELS and MECHANISMS NETWORK



ARTICLES

nature genetics

NANS-mediated synthesis of sialic acid is required for brain and skeletal development

Clara D M van Karnebeek[1,2,28], Luisa Bonafé[3,28], Xiao-Yan Wen[4,5,28], Maja Tarailo-Graovac[2,6], Sara Balzano[7], Beryl Royer-Bertrand[3,7], Angel Ashikov[8], Livia Garavelli[9], Isabella Mammi[10], Licia Turolla[11], Catherine Breen[12], Dian Donnai[12], Valerie Cormier[13], Delphine Heron[13], Gen Nishimura[14], Shinichi Uchikawa[15], Belinda Campos-Xavier[3], Antonio Rossi[16], Thierry Hennet[17], Koroboshka Brand-Arzamendi[4,5], Jacob Rozmus[1], Keith Harshman[18], Brian J Stevenson[19], Enrico Girardi[20], Giulio Superti-Furga[20,21], Tammie Dewan[1], Alissa Collingridge[1], Jessie Halparin[1], Colin J Ross[1,2,6], Margot I Van Allen[6], Andrea Rossi[22], Udo F Engelke[23], Leo A J Kluijtmans[23], Ed van der Heeft[23], Herma Renkema[23], Arjan de Brouwer[24], Karin Huijben[23], Fokje Zijlstra[23], Thorben Heisse[25], Thomas Boltje[25], Wyeth W Wasserman[2,6], Carlo Rivolta[7], Sheila Unger[26], Dirk J Lefeber[8,23], Ron A Wevers[23,29] & Andrea Superti-Furga[3,27,29]

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

Canadian Research Agenda for PM

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)

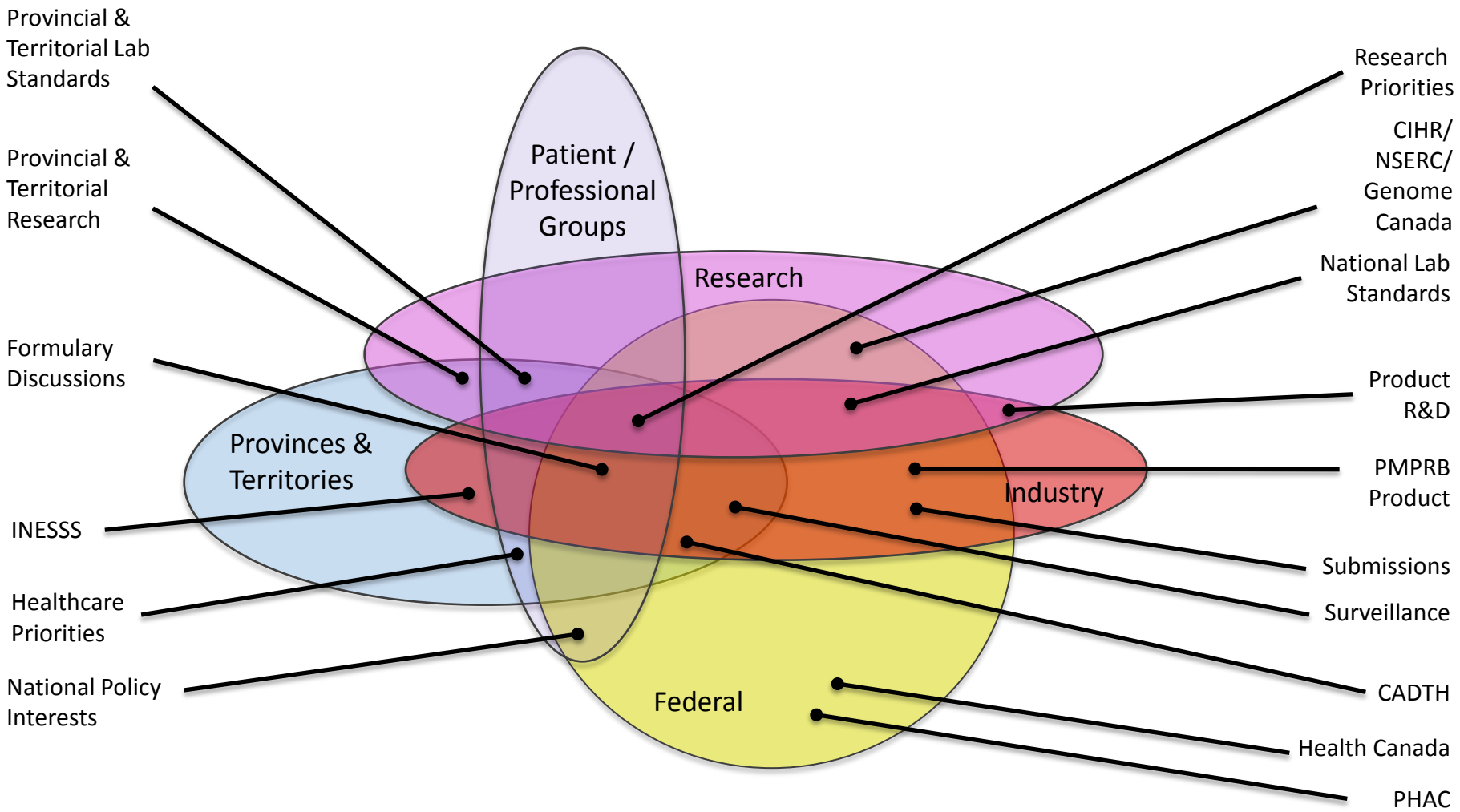
Gaps

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

Foundation

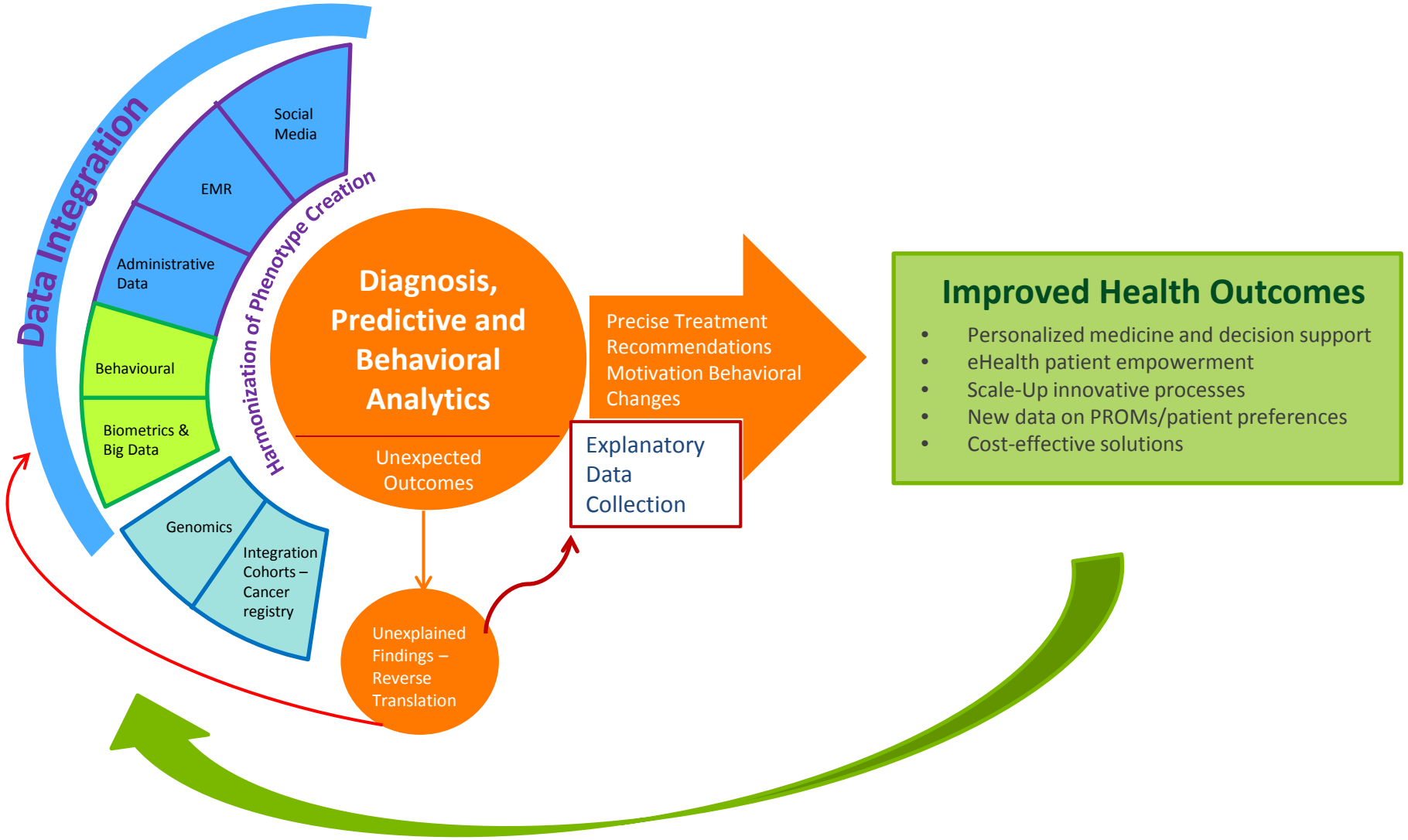
- Early signs of successful implementation (e.g. specific cancers, rare diseases, pharmacogenetics)
- Strong national and international positioning
- Significant investments made in the past few years by GC/CIHR PM Signature Initiative, SPOR support unit (NL) and through initiatives in several provinces

Where are we?



T. Ryan Sigouin, Health Canada adapted by Inga Murawski CIHR-ICR and Etienne Richer CIHR-IG

How bold do we want to be?



Genome BritishColumbia
Leading ▸ Investing ▸ Connecting

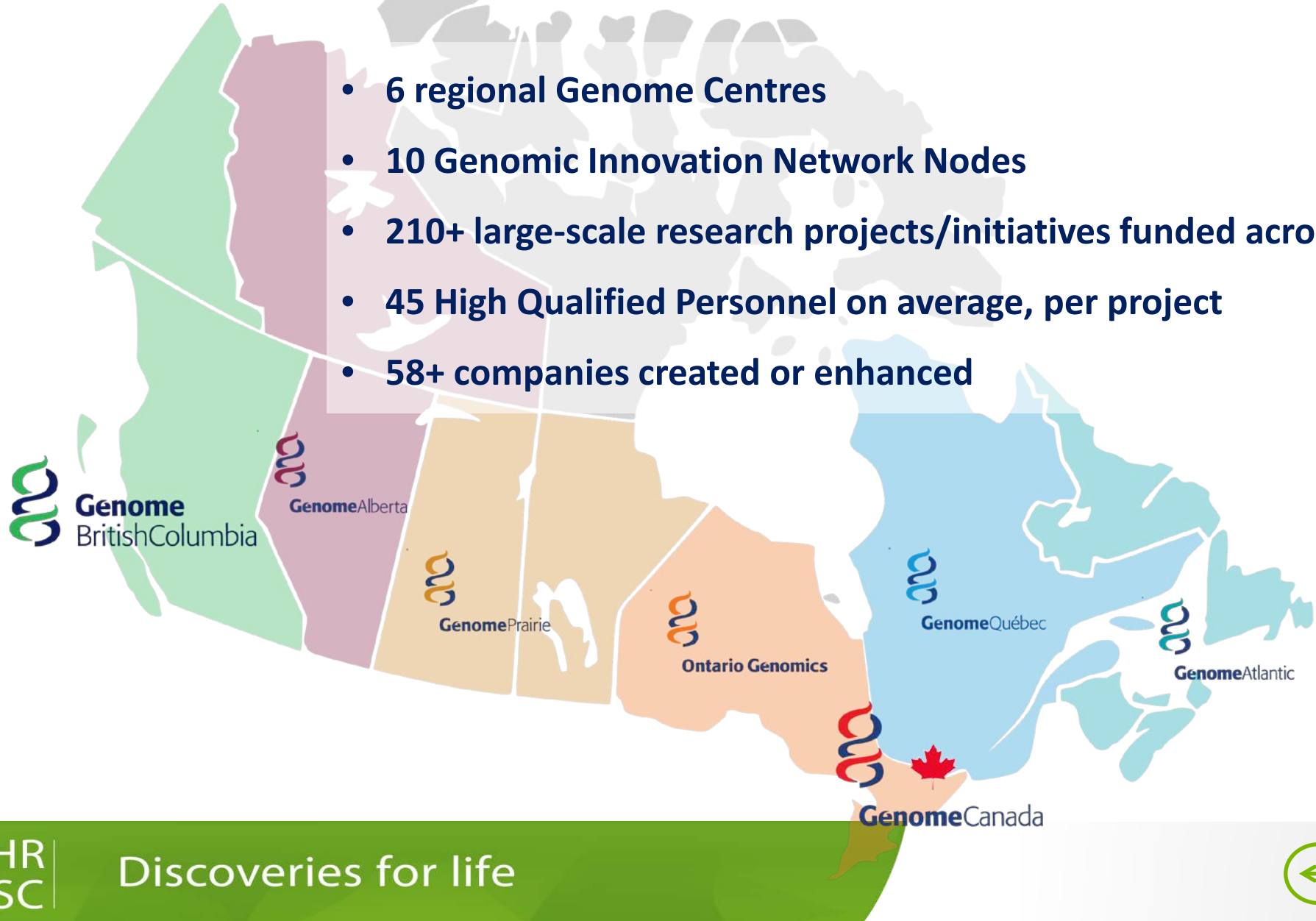
A wave of change:

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 | **Diagnosis** | **Treatment** | **Prognosis**

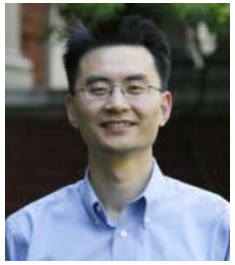
GENOMICS

First Nations Biobank

Prenatal Screening

RapidOMICS

Genomics for Pharmacists



Biomarkers for COPD Management

2017 Large Scale Applied Research Project Competition Stats

6/15

awards went to BC led projects

38.5%

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

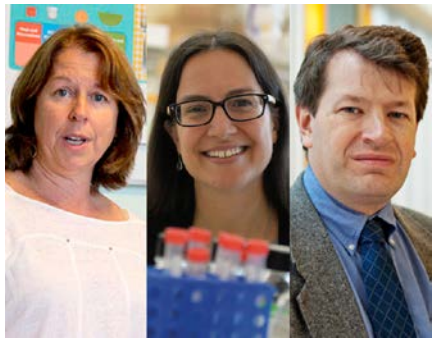
2/15

awards went to BC co-led projects

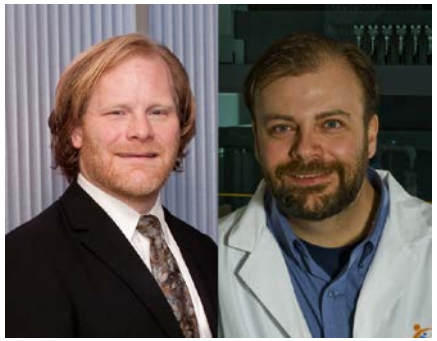
$80.3M

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



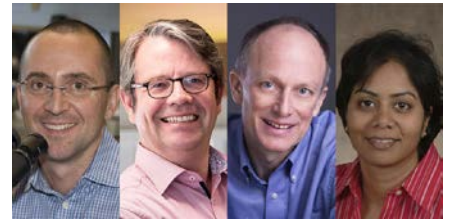
- **Go-PGx: Reducing adverse drug reactions (ADR) for children with cancer**
  Bruce Carleton, Colin Ross
  Total Budget: $10,517,507



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

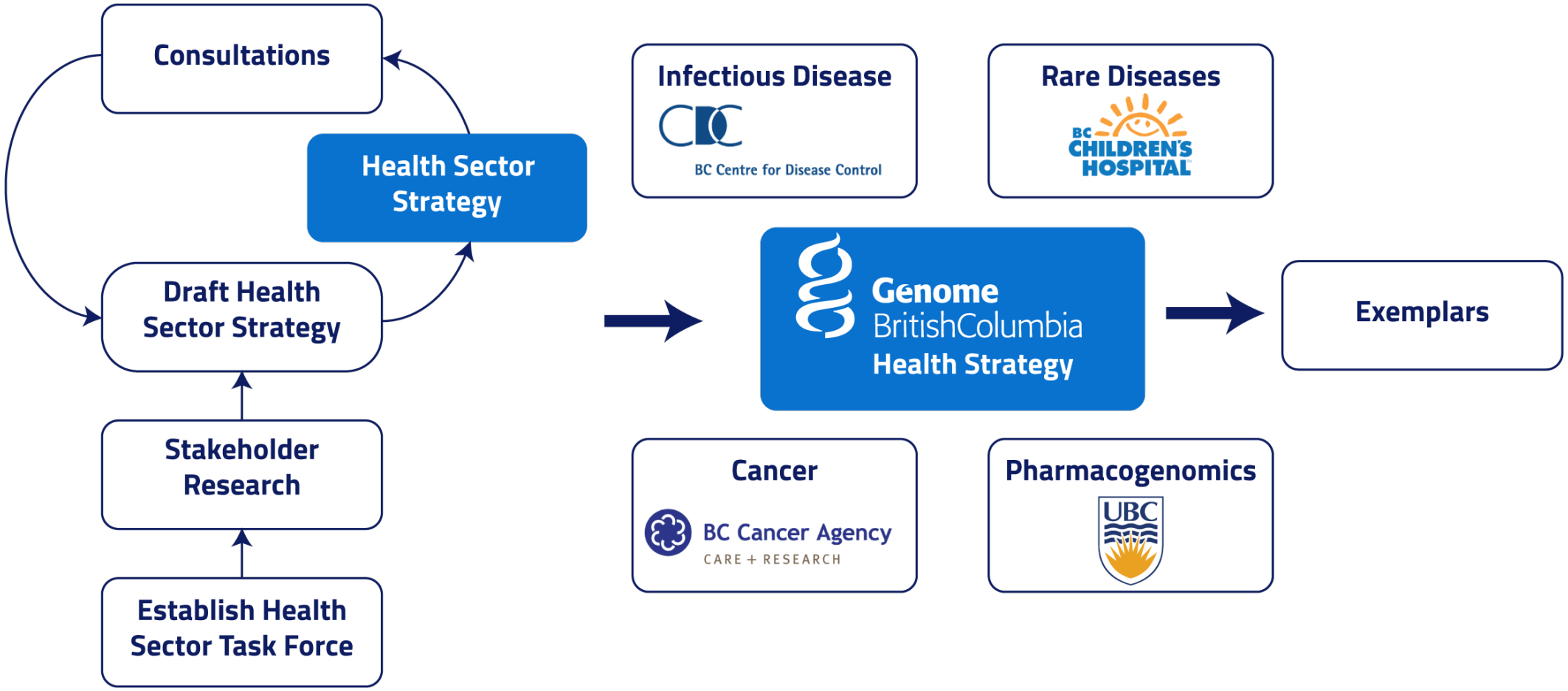
- **Deciphering relapsed lymphoid cancers to improve patient management**
  Christian Steidl, Marco Marra, David Scott
  Total Budget: $11,926,360

- **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



BC pharmacists leading precision medicine

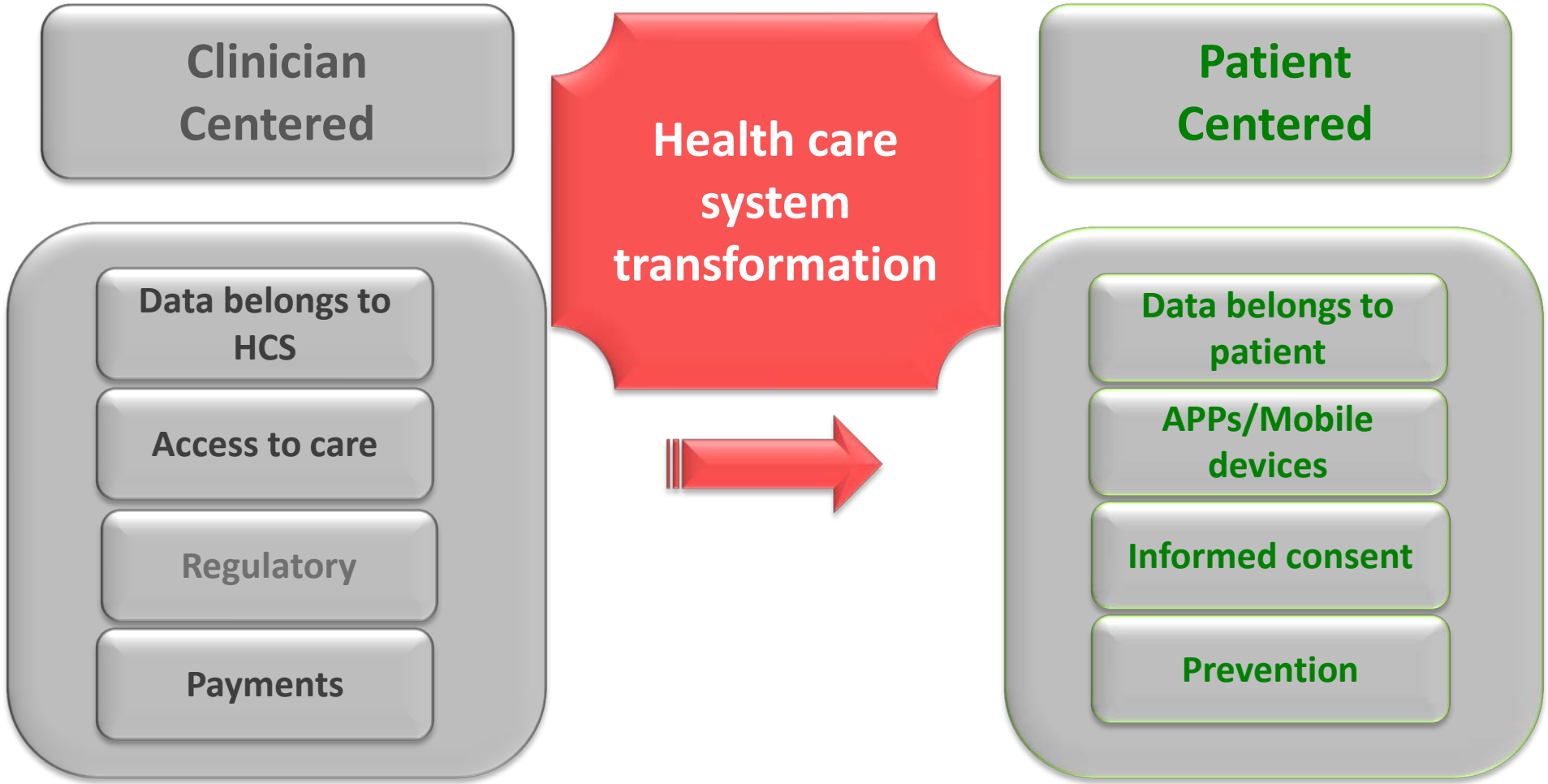
Researcher: Corey Nislow

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

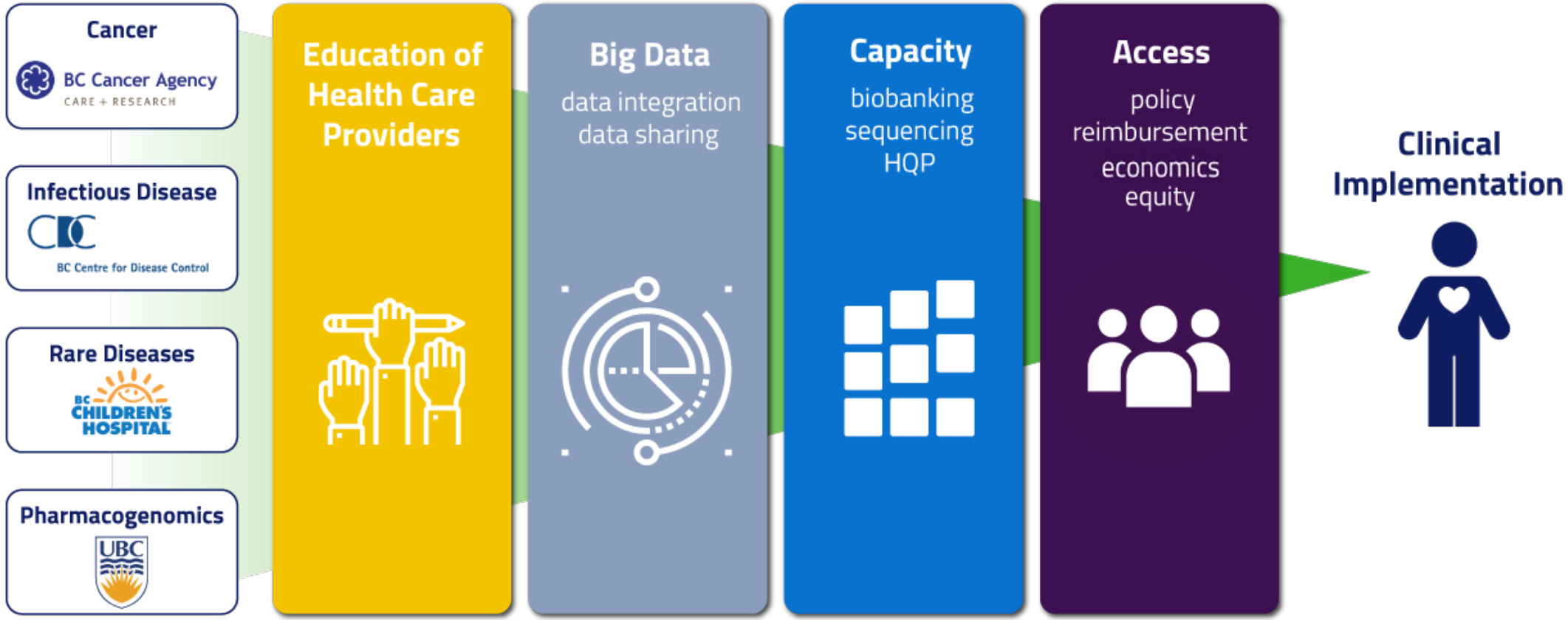
Across BC, 33 community pharmacies have taken part in North America's first project to implement pharmacogenomics allowing them to:

- 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



Crosscutting areas to accelerate implementation



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