Canada’s Genomics and Personalized Health Initiative

Global Leaders in Genomic Medicine Meeting

Dr. Cindy Bell, Interim President CEO, Genome Canada
November 6, 2015 - Singapore
The Canadian Context

- Publically funded health-care system
- Provincially delivered
- Costs the country ~$200B/year
- Growth in cost is around 3% annually (NOT sustainable)
- Biomedical research is strong in Canada
- Strong clinical networks across the country and – for some diseases – has among the best outcomes in the world
Our Challenges

• Our ability to move the latest technology into the health-care system is traditionally low and the way technology is assessed across the country is heterogeneous.

• New technologies are often seen as just an added cost and economic analyses performed are not convincing enough for the payers.
Genomics and Personalized Health

• A Genome Canada program, in partnership with the Canadian Institutes of Health Research (CIHR)

• $65 million from Genome Canada & CIHR leveraged to $150 million through partnerships (regional Genome Centres, industry, health authorities, international organizations)

• 17 large-scale applied research projects selected for funding in 2013; ~$10M over 4 years

• Outcomes of the research must include concrete deliverables with clinical utility or other applications that would allow for subsequent translation into the health-care system
Key Attributes

*Demonstration of potential for impact*
- an economic analysis and rationale for how each project will bring value to the health-care system.
- a detailed development plan for integration into the health-care system, including:
  - demonstration of engagement by end-user(s)
  - consideration of regulatory frameworks

*Addressing barriers to translation and uptake into the health-care system*
- integrated and stand-alone GE³LS
- translational GE³LS network
- GA4GH – CanShare project
- big data analysis– B/CB national strategy and competitions
Genomics and Health

**GENOMICS**

**Prevention**
- Dr. François Rousseau
  - Non-invasive prenatal testing

**Diagnosis**
- Dr. Steve Scherer
  - Autism Spectrum Disorders

**Treatment**
- Dr. Kym Boycott
  - Care 4 Rare

**Prognosis**
- Dr. Richard Harrigan
  - Improved HIV testing, surveillance and treatment
Non-invasive prenatal testing

• Goal: A safer prenatal screening solution
  - Each year 10,000 amniocentesis for T21 with the loss of 70 healthy fetuses.

• Independent study that validates performance and utility of NIPT – an evidence-based cost-effective approach for implementation of this new technology into the Canadian health care system.

Dr. François Rousseau

Université Laval
Impact to date

- Implementation of NIPT into a number of provincial laboratories and development of common bioinformatics pipeline
- Decision making tools to help families make informed decisions are being tested
- Educational tools for health-care professionals are being tested
Autism Spectrum Disorders

- Identify genetic heterogeneity in ASD using WGS
- Integrate genetic information into feedback to families and possibly lead to behavioural interventions that can have tremendously positive impacts on development
- Cost-effective analysis outlining direct and indirect costs of WGS in Autism

Dr. Steve Scherer
Impact to date

- So far, 1000 Canadian families are fully sequenced (whole genome) – in ~40% a genetic explanation found and is helping with medical management of children

- New gene discoveries also identifying new targets for novel drug development
Care 4 Rare

• Improve the diagnosis of rare diseases – halt the diagnostic odyssey:
  o 25% wait 5 to 30 years
  o 40% initial wrong diagnosis
  o 50% no diagnosis

• Identify novel therapies for rare diseases currently without effective interventions:
  o only 5% have access to a proven treatment

Dr. Kym Boycott
Impact to date

• 3000 patients and family members recruited worldwide (20 countries)

• 637 disorders studied

• > 300 Canadian families provided a diagnosis

• **81 novel rare disease genes identified**

• 3 experimental therapies being developed in the lab

• Policy: Position Statement of the Canadian College of Medical Geneticists

Sienna Knapp has a rare genetic disease that gives her seizures. She's helped by her dog, Jedi, trained to alert Sienna's parents when the girl has a seizure.
Improved HIV testing, surveillance and treatment

- Develop an improved HIV drug-resistance test
- Implement real-time HIV drug resistance surveillance
- Develop and validate improved methods for individualised treatment of HIV based on each patient’s unique DNA.

Dr. Richard Harrigan
Impact to date

• Established real-time system to monitor spread of HIV resistance that is being used by BC Centre for Disease Control & BC Ministry of Health

• Helped prompt outbreak investigations, intervention, and monitoring

• Created software for HIV drug resistance genotyping that is much faster than traditional methods
Phase 2

• National Personalized Medicine Strategy
  • Survey of Genetic/Genomic Tests across Canada
  • Exemplar Study

• 2016 Genomics and Personalized Health Competition

• Translational Network