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

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

GLOBAL CHALLENGES ◆ GENOMIC SOLUTIONS DÉFIS MONDIAUX ◆ SOLUTIONS GÉNOMIQUES

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

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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.

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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

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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 GE^{3}LS
- translational GE^{3}LS network
- GA4GH – CanShare project
- big data analysis– B/CB national strategy and competitions

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Genomics and Health

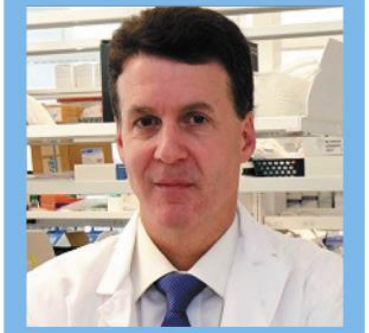
GENOMICS

Prevention

Dr. François Rousseau

Non-invasive prenatal testing

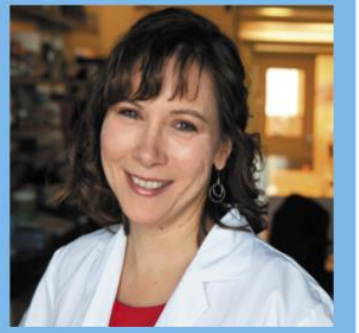
Diagnosis



Dr. Steve Scherer

Autism Spectrum Disorders

Treatment



Dr. Kym Boycott

Care 4 Rare

Prognosis

Dr. Richard Harrigan

Improved HIV testing, surveillance and treatment

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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

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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

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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

SickKids®
THE HOSPITAL FOR SICK CHILDREN

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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

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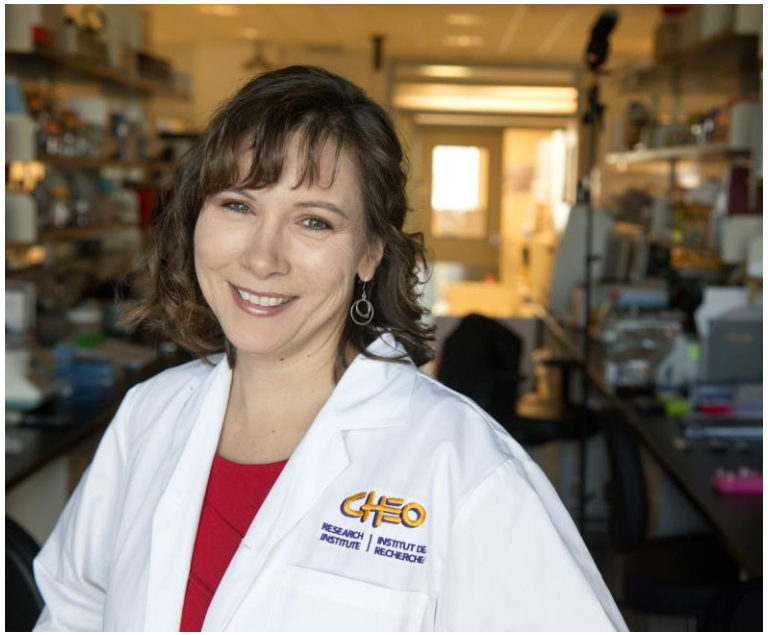
Care 4 Rare

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

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



Dr. Kym Boycott

CHEO Children's Hospital of Eastern Ontario
Centre hospitalier pour enfants de l'est de l'Ontario

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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.

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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

BRITISH COLUMBIA CENTRE *for* EXCELLENCE *in* HIV/AIDS

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Impact to date

- Established real-time system to monitor spread of HIV resistance that is being used by BC Centre fro 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

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Phase 2

- National Personalized Medicine Strategy
  - Survey of Genetic/Genomic Tests across Canada
  - Exemplar Study
- 2016 Genomics and Personalized Health Competition
- Translational Network

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