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## TOWARDS IMPLEMENTING PERSONALIZED MEDICINE IN CANADA

CSA PerMed Workshop 1  
Berlin, Germany

March 27-28, 2014

*Pierre Meulien, Ph.D.  
President and CEO, Genome Canada*

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April-7-14

## PRESENTATION TODAY



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- Introduction to the Canadian healthcare environment
- Genomics in Canada
- New program design in Canada for large scale research program in **Personalized Health** (launched in January 2012)
- Some conclusions and looking ahead

## CANADIAN ENVIRONMENT



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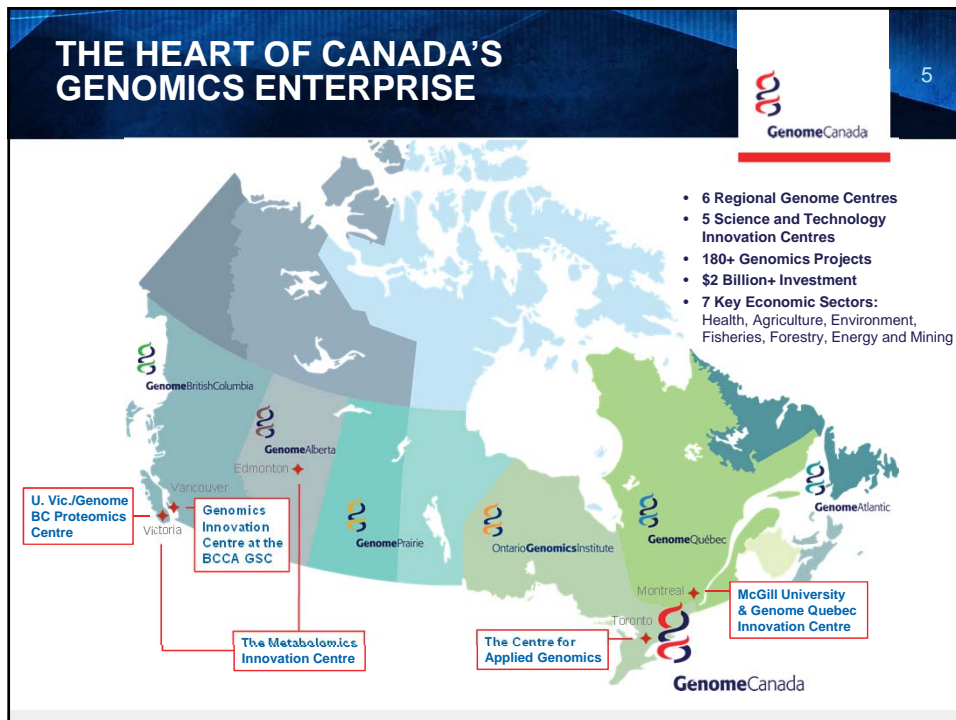
- Publically funded health care system
- Provincially delivered (Regional Health Authorities)
- Costs the country around \$220 Billion per year
- Growth in cost is around 3% annually (NOT sustainable)
- Biomedical research very strong in Canada
- Canada spends about 2% of government funded global research but produces 4% of the highest impact factor publications
- Strong clinical networks across the country and - for some diseases - has among the best outcomes in the world

## CANADIAN ENVIRONMENT (CONT'D)



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- However, our ability to move the latest technology into healthcare delivery is traditionally poor and the way technology is assessed across the country is very 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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**Since 2000, Genome Canada (and partners) has invested over \$1.2 billion in human health related genomics research**

**GC has a vested interest in seeing some of the results of this investment put into practice**

## HOW DO WE TRANSLATE WHEN WE NEED TO CONSIDER A LOT OF COMPLEX ISSUES?



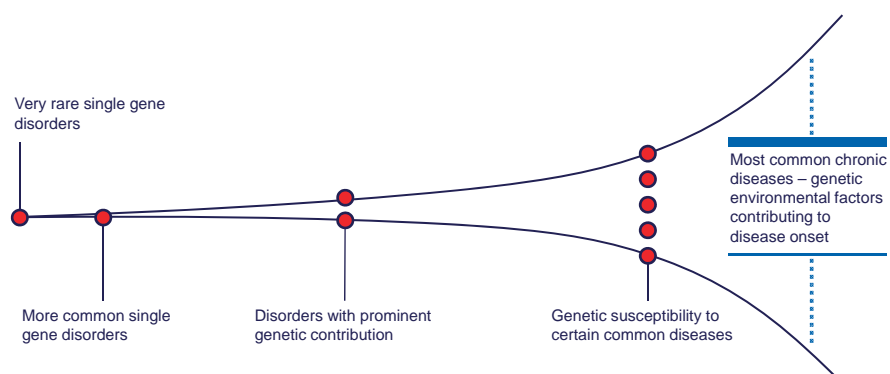
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- How good is the technology? (clinical validation)
- In a fast moving field, when do we decide that “now is the time for transfer”
- Is it easy to adapt existing clinical laboratory structures?
- Who will be making these decisions? (and based on what criteria?)
  - Technology assessment based on sound economics and clinical benefit?
- Who will pay?


## SPECTRUM OF GENETIC CONTRIBUTION TO DISEASE




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**FORGE CANADA CONSORTIUM**



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*A nation-wide effort to identify genes causing rare pediatric disorders*




**Kym Boycott, PhD, MD, FRCPC, FCCMG**


Clinical Geneticist, Department of Genetics Investigator, Children's Hospital of Eastern Ontario Research Institute Associate Professor, Department of Pediatrics, University of Ottawa









**FORGE CLINICAL NETWORK**


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**SITE CHAMPIONS**



## HOW MANY HUMAN SINGLE GENE DISORDERS REMAIN TO BE DISCOVERED?

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The image shows an iceberg floating in the ocean. The tip of the iceberg is above the water, and the much larger base is submerged. Red arrows point from text labels to different parts of the iceberg.

Category	Count
Gene known	~3500
Gene unknown	~1700
Suspected single gene disorders	~2000

## 1 STORY... UNDIAGNOSED NEURODEGENERATION

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A photograph of two young boys. The boy on the left is wearing a grey hoodie with 'DESIGNER' written on it. The boy on the right is wearing a blue and grey striped sweater. They are standing indoors, possibly in a hallway or room.

<http://www.ottawacitizen.com/technology>  
December 4, 2011

## FORGE PROGRESS— 24 MONTHS—OVER 150 CASES SOLVED

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Gene known  
~3500

Gene unknown  
~1700

Suspected single  
gene disorders  
~2000

- ACTH Resistance
- Hermansky-Pudlak like
- Hawk Junction Microcephaly
- Perrault-Syndrome
- Severe Combined Immunodeficiency
- AD Retinitis Pigmentosa
- Hadju-Cheney Syndrome
- Metaphyseal dysplasia, maxillary hypoplasia

- Leber Congenital Amaurosis
- Floating Harbor syndrome
- French Canadian Joubert syndrome
- Chudley McCullough syndrome
- Weaver syndrome
- Hyper IgM
- Megalencephaly Capillary Malformation
- Nager syndrome

- Microcephaly Capillary Malformation
- Mandibulofacial dysostosis with Microcephaly
- Jeune-Joubert syndrome
- Short stature, cataracts, peripheral neuropathy
- Intestinal pseudo-obstruction with sick sinus

## WHAT WE NEED NOW

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So...Genomics works for rare genetic diseases...  
what about other more common diseases?

- Demonstrations that the technology can deliver real value to patients with more common disorders
- Demonstrations that integrating the technology within the healthcare system will be cost effective

## NEW PROGRAM DESIGN – ROLE OF FUNDERS



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- **How do we encourage....**
  - The right team formation
  - The right types of deliverable are achieved (timely impact on patients)
  - That true demonstrations of value are obtained
- **Importance of the right peer review process**

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## GENOME CANADA - CANADIAN INSTITUTES OF HEALTH RESEARCH (CIHR)

2012 LARGE SCALE APPLIED RESEARCH COMPETITION  
GENOMICS AND PERSONALIZED HEALTH



## GENOMICS AND PERSONALIZED HEALTH



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- Program partnered 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 partnerships)
- Requirement for teams to provide an **economic analysis and rationale** for why their particular application will demonstrate value to the health system
- Required relevant Economic, Environmental, Ethical, Legal and Social (GE<sup>3</sup>LS) research

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

## GE<sup>3</sup>LS - GENOMICS AND ITS ETHICAL, ENVIRONMENTAL, ECONOMIC, LEGAL AND SOCIAL ASPECTS



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- Genomics-related research undertaken from the perspective of the social sciences and humanities
- In the context of this RFA it was extended to cover researchers in the fields of health administration, health management, health services research, health economics, health technology assessment, evaluation and comparative effectiveness studies.

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## GENOMICS AND PERSONALIZED HEALTH



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- Teams were to provide detailed **development plan for integration** into the health care system
- Demonstrated buy-in from the payer and clinicians
- Considered the regulatory frameworks existing in Canada
- Projects were reviewed by ~40 international translational researchers, social scientists and **health economists**
- Panel Chair: Raju Kucherlapati (HMS)



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## GENOMICS AND PERSONALIZED HEALTH



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- Review was in two phases:
  - Pre-application (146 submissions)
  - Full proposals (40 submissions)
- Review of full proposals took place over 3 days with face to face meetings between members of the review panel and project team applicants
- 17 projects are now launched (average \$8.8 million)



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## SOME EXAMPLES OF APPROVED PROJECTS



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### Overarching theme:

Inform decision making re: effectiveness of drugs, adverse drug reactions, intervention strategies and disease management.

- Epilepsy
- Autism
- Lymphoma, Breast Cancer, Glioblastoma, and other Cancers
- Rare Diseases
- Stroke
- HIV
- Inflammatory Bowel Disease
- Cardiovascular Disease

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## VIRAL AND HUMAN GENETIC PREDICTORS OF RESPONSE TO HIV THERAPIES – RICHARD HARRIGAN AND JULIO MONTANER UBC/ ST PAUL'S HOSPITAL, VANCOUVER



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### Primary Research Objective

- Optimize HIV therapy, improve HIV suppression and reduce HIV drug resistance

### Key Specific Objectives

1. Implement a multi-class HIV drug resistance test based on next-generation sequencing
2. *Establish a program for real-time investigation of HIV resistance incidence and prevalence*
3. Validating additional human genomic tests to guide HIV therapy (expansion program)

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## OPTIMAL BREAST CANCER SCREENING AND TREATMENT STRATEGY - PROF. JACQUES SIMARD, UNIVERSITÉ LAVAL - \$11.5 M



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- Today: systematic screening for women over the age of 50
- But nearly a quarter of cases affects women under 50
- 2.5 million women aged 40-49 currently not screened effectively
- Personalized risk stratification will extend current screening programs in a cost-beneficial manner
- Early detection and follow-up will improve



Optimize the screening strategy, improve prognostic and treatment, and reduce the costs on the health care system



## PERSONALIZED TREATMENT OF LYMPHOID CANCER: BRITISH COLUMBIA AS MODEL

JOSEPH CONNORS, BC CANCER AGENCY;  
MARCO MARRA, RANDY GASCOYNE



To demonstrate the use of genomics in diagnosis of lymphoid cancers in a large population (province), with potential for scale up nationally and internationally.



## PERSONALIZED GENOMICS FOR PRENATAL ANEUPLOIDY SCREENING USING MATERNAL BLOOD – PEGASUS

FRANÇOIS ROUSSEAU, UNIVERSITÉ LAVAL; SYLVIE LANGLOIS



To provide Canada with evidence to make informed decisions about implementation of genomics-based non-invasive prenatal testing (NIPT) for aneuploidies



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## CONCLUSIONS AND FUTURE CHALLENGES

## INTEGRATION OF GENOMICS INTO THE HEALTHCARE SYSTEM



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- Develop receptor capacity for technology pull (capacity for clinical and translational research)
- Involvement of the private sector
- Educate and train healthcare professionals to be proficient users of the technology
- Ensure information systems are state of the art and harmonize e-patient records
- Increase the role of patients and advocacy groups in demanding evidence based medicine
- Apply robust technology assessments focused on improvement on clinical outcomes and economic benefit analyses

## THE FUTURE?







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- This is the beginning of something not the end
- Knowledge base will be totally different in 5 years (again!)
- We will be layering proteomics, epigenomics and microbiome data on top of our personal genome sequence data
- In ten years, technology will allow us to do things unimaginable today

**ACKNOWLEDGEMENTS**

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 **GenomeAtlantic**       **GenomeQuébec**       **OntarioGenomicsInstitute**

 **GenomePrairie**       **GenomeAlberta**       **GenomeBritishColumbia**

**ACKNOWLEDGEMENTS**

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**THANK YOU**