



Genome Québec



solutions are in the genes



SESSION III

POPULATION HEALTH AND GENOMICS: INCREMENTAL IMPLEMENTATION OR RADICAL REFORM?

Washington, November 19, 2015

By Marc LePage, President and CEO, Génome Québec

QUÉBEC: KEY FACTS

- Population: 8,054,756 (2012)
- Main university research centres:
 - Concordia University
 - Université Laval
 - McGill University
 - Université de Montréal
 - Université de Sherbrooke
- Health care expenditures: \$45 billion (75% Public • 25% Private)
- 49% of provincial budget
- One of the lowest per capita expenditure rates in Canada



MCGILL UNIVERSITY AND GÉNOME QUÉBEC INNOVATION CENTRE



- Genomics research centre established in 2002
- High throughput genomics (18 HiSeq, etc.)
- 60 Génome Québec employees and 170 McGill researchers
- \$19 M revenues
- More than 900 teams annually:
Human 2/3 • Non-human 1/3



INTERNATIONAL REACH

Innovation Centre Users International Distribution

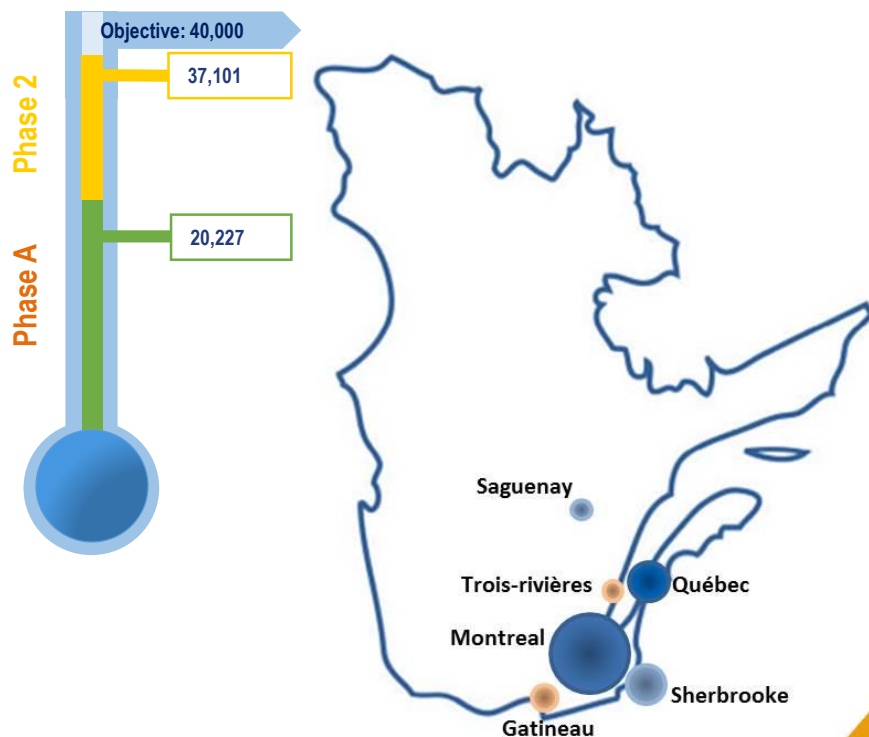


28 countries

Québec 60% • Canada 25% • International 15%

THE CARTaGENE BIOBANK

RECRUITMENT AND SAMPLING



Phase A (Nov 2009-Oct 2010)

- 20,000 participants aged 40-69 years at 12 clinical sites from four “urban” centres
- Health questionnaires + biologicals (PBMCs, serum, plasma, saliva urine, cells) + physical and clinical measures + genealogies
- Nutrition and environment surveys (2011-2012)

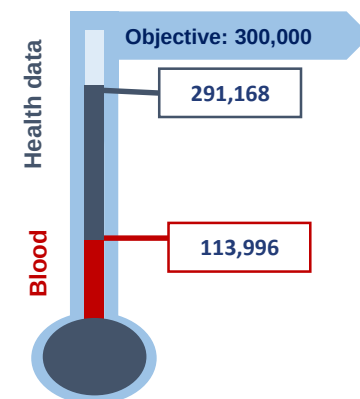
Second wave (Jan 2013 - ongoing)

- 20,000 participants aged 40-69 years
- Health questionnaires + biologicals + genealogy



CPTP Pan-Canadian cohort

* BCGP, TP, OHS, CaG, AP



CPTP – A CANADIAN –WIDE EFFORT

CMAJ

ANALYSIS

The Canadian Partnership for Tomorrow Project: building a pan-Canadian research platform for disease prevention

Marilyn J. Borugian PhD, Paula Robson PhD, Isabel Fortier PhD, Louise Parker PhD, John McLaughlin PhD, Bartha Maria Knoppers PhD, Karine Bédard MA, Richard P. Gallagher MA, Sandra Sinclair MSc, Vincent Ferretti PhD, Heather Whelan MSc, David Hoskin PhD, John D. Potter MD PhD

300,000 samples collected

CLINICAL PLATFORMS:

- ▶ **GÉNOME QUÉBEC AND CHU SAINTE-JUSTINE INTEGRATED CLINICAL GENOMICS CENTRE IN PEDIATRICS**
- ▶ **CQGC NATIONAL LABORATORY**

7,000 RARE GENETIC DISORDERS



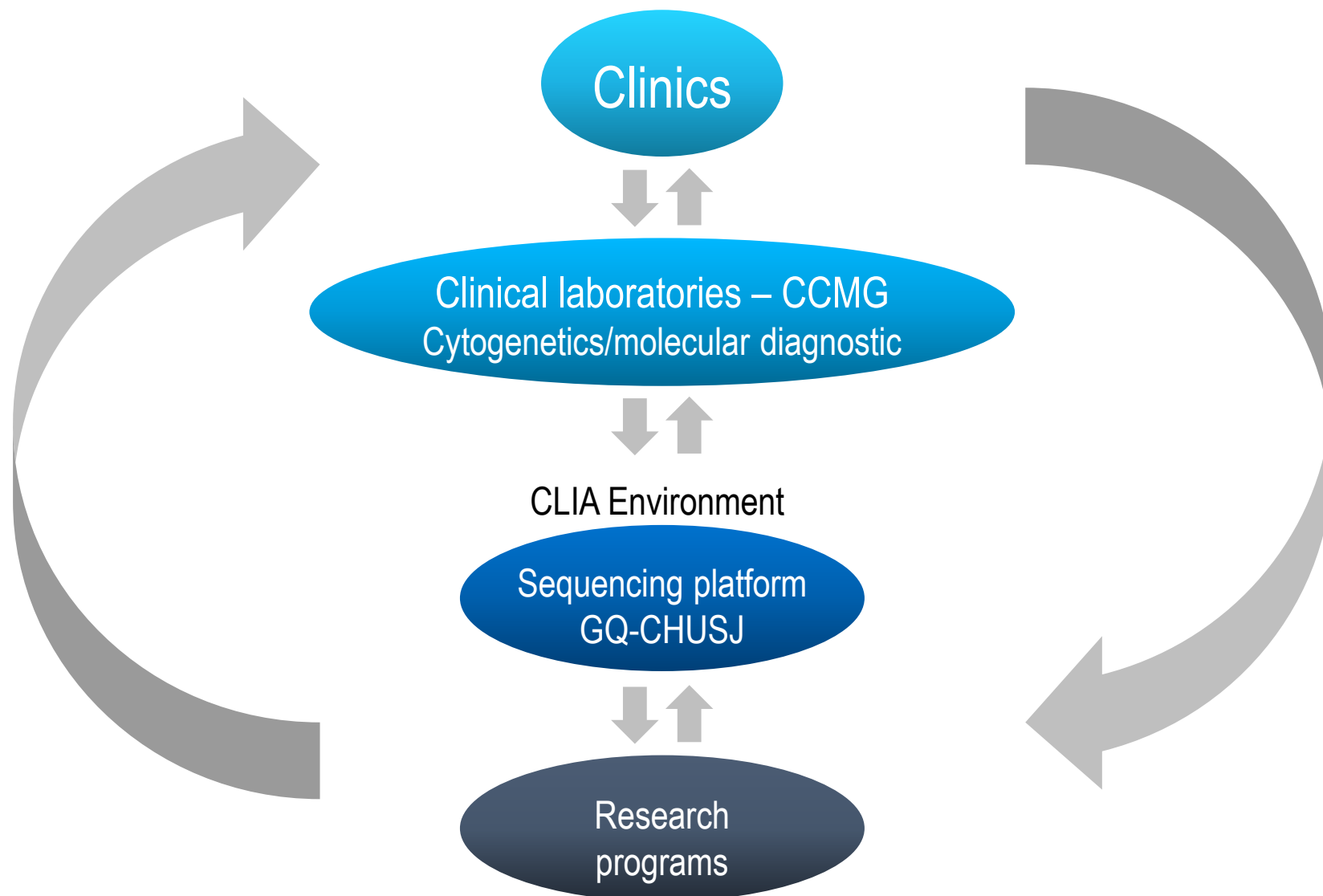
Diagnostic odyssey

500,000 Canadian children

- ▶ 40% wrong diagnosis
- ▶ 50% no diagnosis
- ▶ 25% wait 5 to 30 years

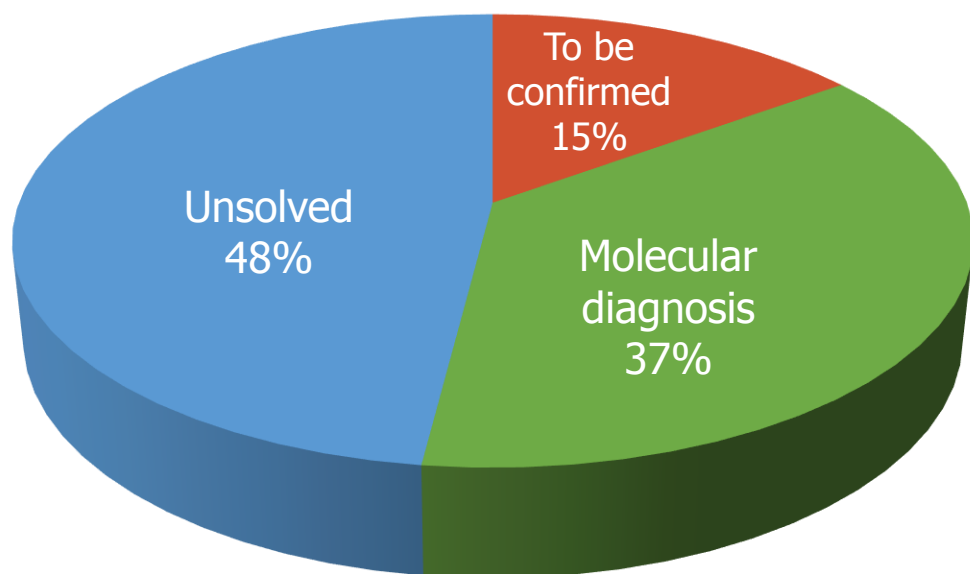
Unmet medical need: Diagnosis is care

CENTRE FOR CLINICAL GENOMIC IN PEDIATRICS



PILOT PROJECT

EXOME SEQUENCING IN THE PROBAND



- 96 children
- Global developmental delay intellectual disability (18/49 • 37%)
- Other conditions (17/47 • 36%)

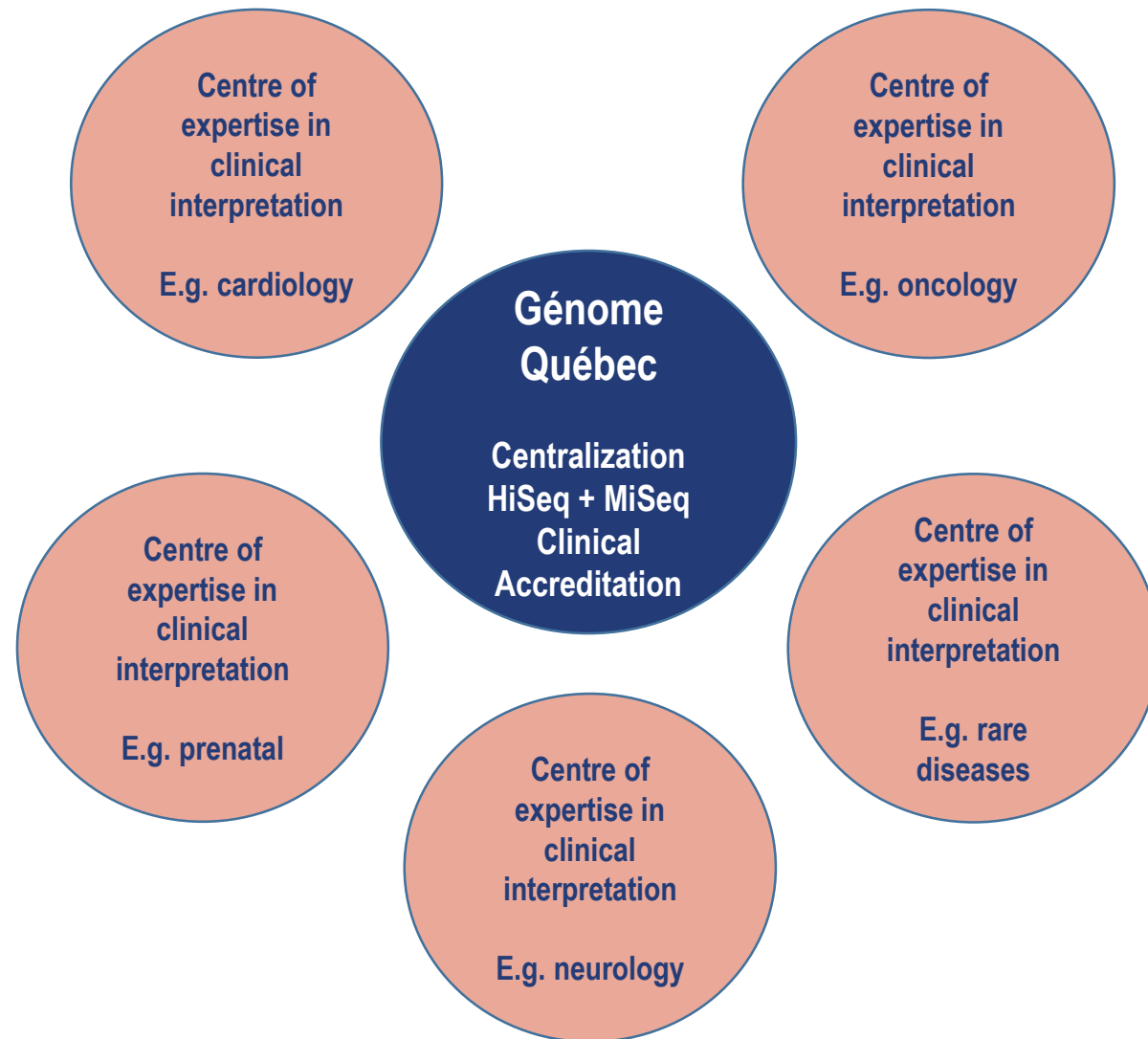


Genome Québec

CENTRE QUÉBÉCOIS DE GÉNOMIQUE CLINIQUE (CQGC)

NATIONAL LABORATORY MODEL

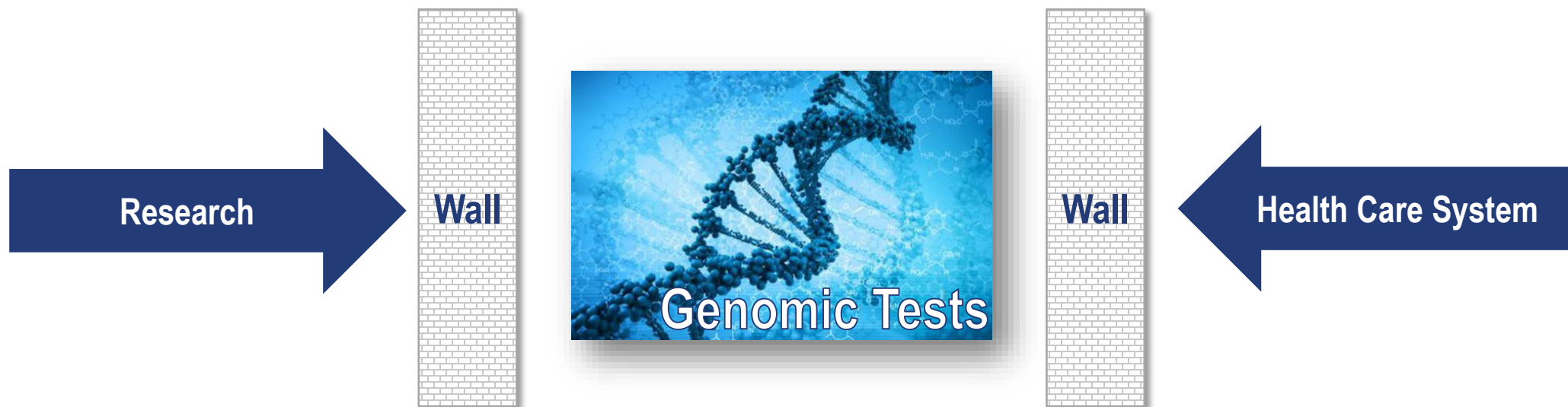
STRUCTURE



Centres of expertise: - MiSeq and validation with Sanger
- Development of new tests



MIND THE GAP PROGRAM ALIGNMENT





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Making **Genomics** Work for Citizens and Society