ALLFORONE HEALTH DATA ECOSYSTEM

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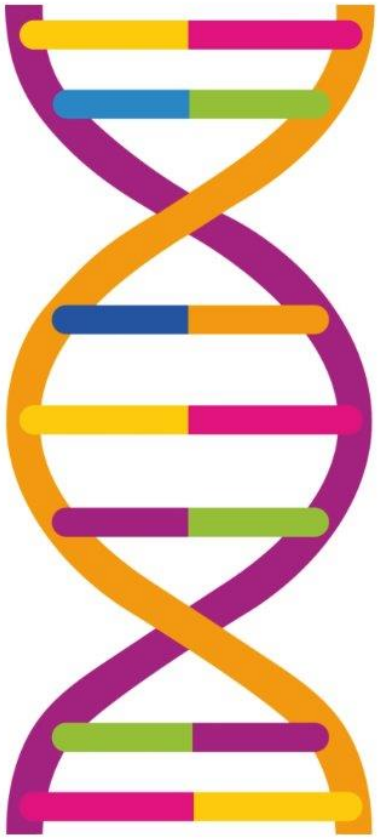
RARE DISEASES

- 1 in 12 Canadians will be affected by a rare disease
- Genetic testing can help in the diagnosis of those patients
- More comprehensive genetic tests are now available, including genome wide sequencing (GWS)
    - Data sharing is important to facilitate molecular diagnosis of patients
- Obtaining a diagnosis can lead to an adjustment in the patient's medical care
    - A treatment may be available for the specific genetic condition
    - Connecting patients with other families, medical specialists, scientists is important
- Family members can also be impacted by a molecular diagnosis

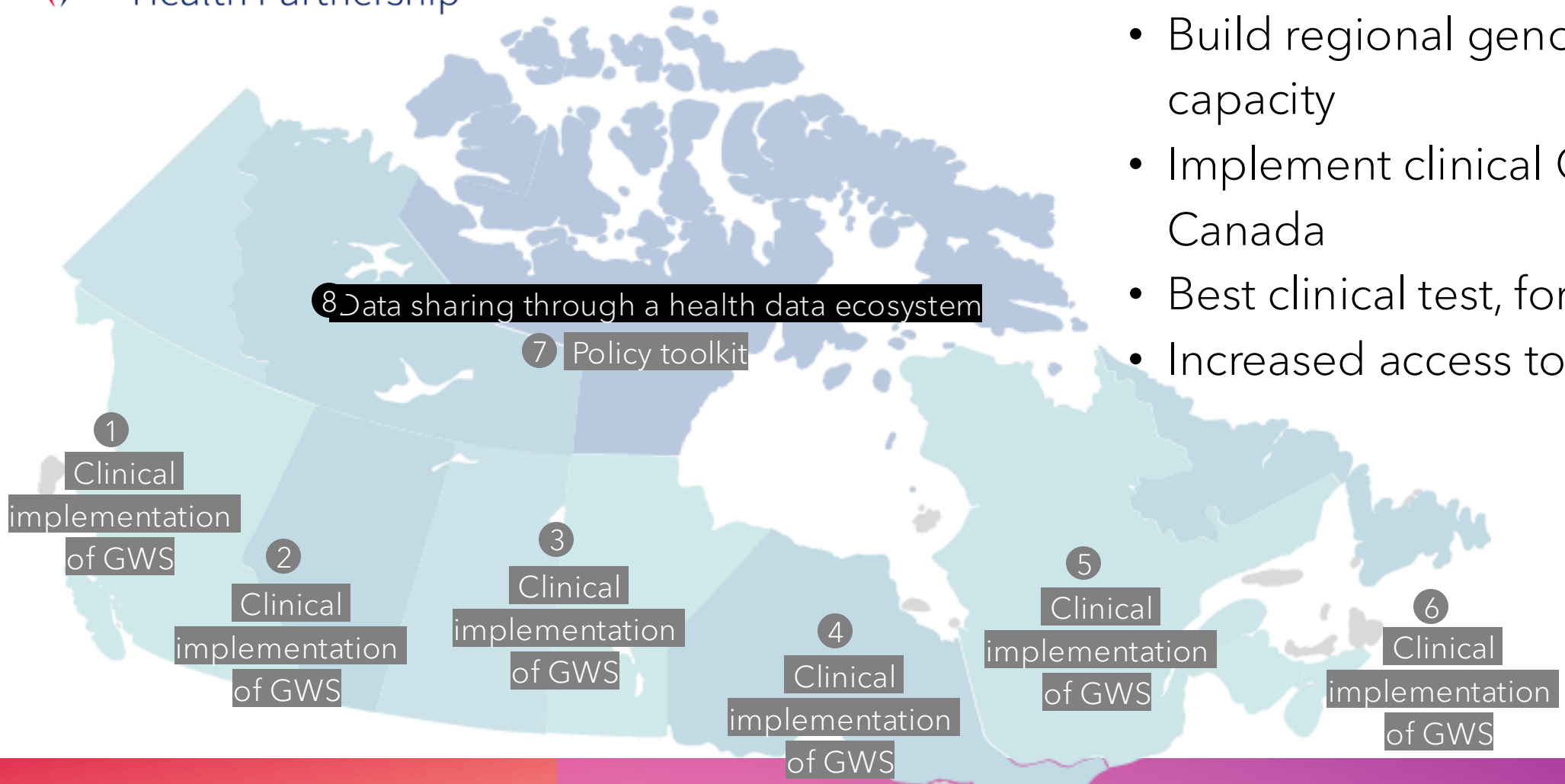


AllforOne
Canada's Precision Health Partnership

2019-2025 (6 YRS)
$41M CAD

GOALS:

- Build regional genomics capacity
- Implement clinical GWS in Canada
- Best clinical test, for all
- Increased access to research



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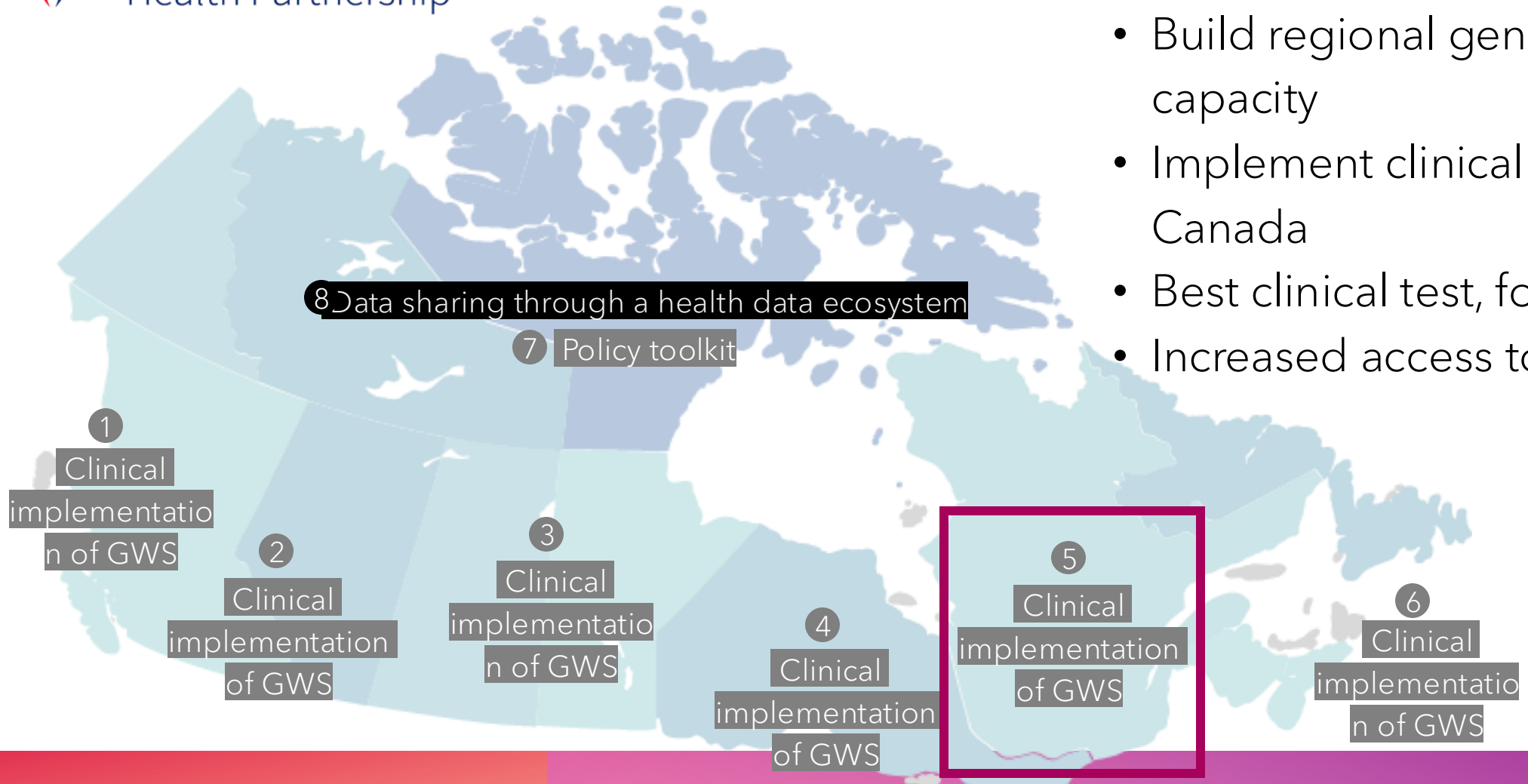


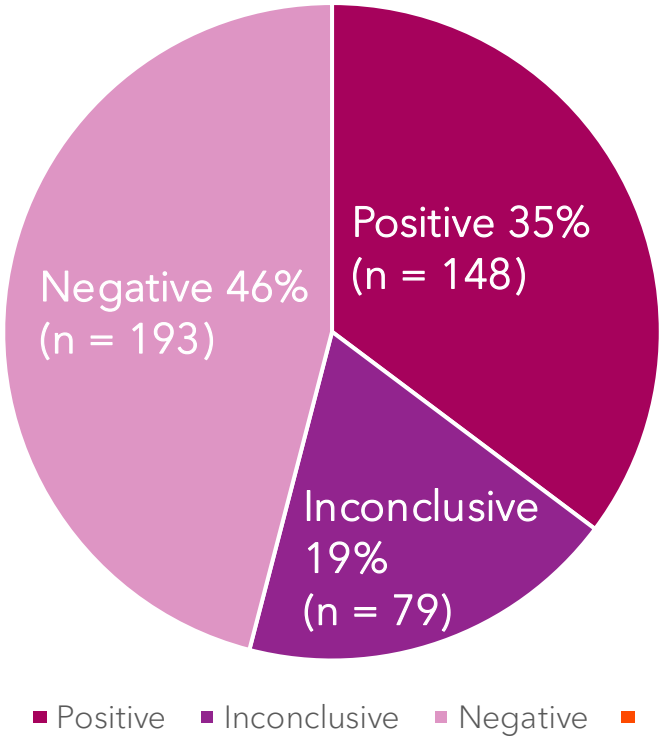
PRAGMATIQ

- Pediatric RApid GenoMIcs in Quebec
- Three-year study
- Implementation of rapid WGS for hospitalized patients in one of the four pediatric hospitals in Quebec (CHUSJ, CHUS, CHUQ, CUSM)
- Suspicion of a genetic condition by a medical geneticist

PRAGMATIQ

By October 1st, 2024
420 patients completed
Diagnostic yield 35%

Diagnostic yield



Clinical impact in
29% of all patients
66% of patients with a molecular diagnosis
(e.g. specialist consultation, medication, comfort care, diet/formula, surgery/procedure, etc.)

Familial impact in
12% of all families
34% of families with a molecular diagnosis
(e.g. cascade testing, medical follow-up added/avoided, prenatal diagnosis)

PRAGMATIQ – WHAT'S NEXT?

- Move from experimental and research uses to real-world clinical use to accelerate access to care and treatment
- Discussion with the government to make rWGS available to hospitalized patients in Quebec

Genomics Applications Partnership Program (GAPP) projects – A4O Health data ecosystem

- Data sharing between across Canadian diagnostic molecular laboratories to improve molecular diagnosis of patients with a rare disease – Clinical Network
- Connect patients and families to researchers to increase access to research – Connect Registry

HEALTH DATA ECOSYSTEM

Clinical Network

National network of diagnostic laboratories

Facilitate high-quality clinical GWS for Canadians with RD

Connect Registry

National registry for those willing to be contacted directly about research

Provide equitable access to precision health research for Canadians with RD

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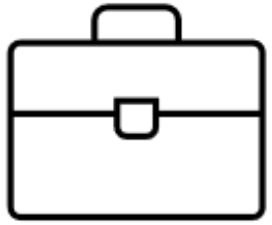
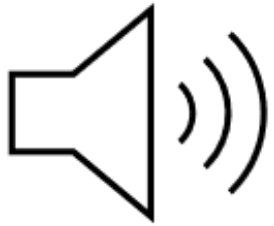
SOME THINGS TO KNOW ABOUT GWS AS A DIAGNOSTIC TOOL



There is overlap in work between labs

But most operate in siloes, as healthcare is administered regionally



Volume is important

Small population, even smaller when divided by province, region or laboratory

Populations are important

Unique populations (e.g., Indigenous, Hutterite, French Canadian) are underrepresented in public databases and regionally

Benefits:

- Test equity
- Consistency between labs
- Increased lab efficiency
- Faster time to diagnosis

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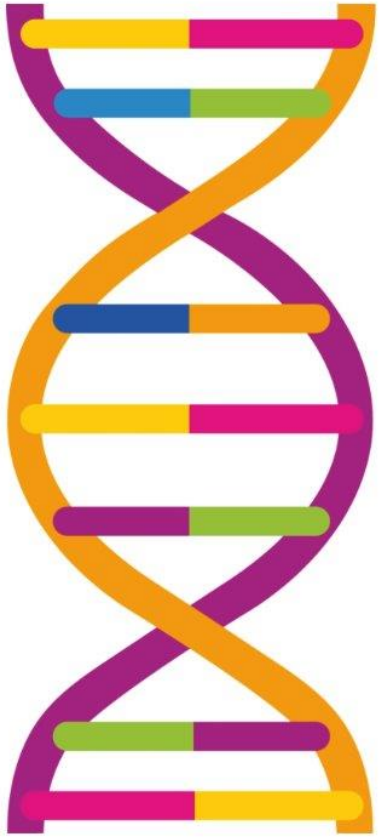
CLINICAL GWS → RESEARCH



- Diagnosis
- Help diagnose others
- Evidence for novel disease gene
- Symptom management
- Understanding for the family or others
- Informatic tool development
- Natural history
- Clinical trials
- Understanding of the human genome
- Precision medicine
- AI

WHY SHOULD ALL FAMILIES WITH RARE DISEASE HAVE ACCESS TO RESEARCH?

Undiagnosed

- Diagnosis
- Candidate
- Evidence for novel disease gene

Diagnosed

- Symptom management
- Natural history studies
- Clinical trials

Everyone

- Help diagnose others
- Understanding for the family or others

WHY DON'T ALL FAMILIES WITH RARE DISEASE HAVE ACCESS TO RESEARCH?

Identification

- Researcher unaware of eligible participants
- Clinic unaware of research opportunity

Connection

- Limited recruitment opportunities
- Geography

Support

- Consenting
- Data entry
- Ethics applications

- Travel
- Time

ACKNOWLEDGMENTS

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with any feedback or questions

THANK YOU!