



Turning Genomic Data into Translational Impact: CPHI and the Pan-Canadian Genome Library

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Land Acknowledgment

Genome Canada is located on the unceded traditional land of the Anishinaabe Algonquin People. As a national organization, we support activities located on the traditional territories of many First Nations, Inuit and Métis Peoples.

We recognize past and ongoing injustices perpetrated against Indigenous peoples as part of the colonial project in Canada, including violence, oppression, land theft and harmful attempts to erase culture.

Given Canada's colonial foundation and how it shapes the discourse and practice of science—especially genetics and related health and biomedical research—Genome Canada commits to fostering Indigenous truth, reconciliation and engagement in our programs to address the gaps in the participation of underrepresented equity deserving and Indigenous groups in Canada's genomics research agenda, data sets and governance.

Genome Canada commits to working in co-creative partnership with Indigenous researchers, leaders and communities to develop and support a stand-alone and distinctions-based strategy to bolster Indigenous genomics leadership in Canada.



Canada's precision health landscape

- **Stability through collaboration:** Canada's governance model fosters consensus-building, making it a reliable international partner
- **Strategic opportunities:** Major investments in precision health, genomics infrastructure, and data coordination create immediate collaboration pathways
- **Convening power:** Through ICPeMed, EP PerMed and GA4GH engagement, Canada demonstrates its commitment to multilateral precision health coordination
- **Complementary strengths:** Canada's research excellence, diverse population, universal healthcare system, and trade access make it an ideal partner for translating personalized medicine research into clinical implementation
- **Future-focused:** Despite current global uncertainty, Canada is making bold investments in transformative research programs that align with international precision medicine priorities



Genome Canada

translates genomics-driven biotechnology
breakthroughs into transformative solutions for
Canadians.





Genome Canada aligns Canadian genomics capabilities into national missions that deliver **transformative health, economic and environmental solutions**—tackling challenges too big for any one institution to solve alone.



Health



Forestry



Fisheries and Aquaculture



Agrifood



Mining



Energy



Environment

Our vision

Canada as a **global leader** in
biotechnology innovation and adoption



Our mission

Lead large-scale, mission-driven genomics initiatives that translate research into real-world impact.

Build Canada's genomics capacity through strategic investments in research, technology and talent.

Connect industry, academia and government in powerful cross-sector alliances—aligning federal, provincial and private-sector investments to amplify Canada's genomics impact nationally and globally.



Our approach: Mission-driven research tackling complex problems

- Canada faces complex challenges that require coordinated, multi-disciplinary solutions.
- In 2020, Genome Canada made a strategic decision to transform its approach:

We committed to mission-driven innovation, focusing on delivering tangible solutions to clearly defined problems.

- From disciplinary silos --> integrated solutions
- From individual projects --> coherent portfolios
- From research outputs --> delivered real-world solutions
- From risk aversion --> balanced innovation investment
- From neutral facilitation --> directional national leadership
- From basic coordination --> strategic ecosystem orchestration



Strategic Directions for 2025-2030

Precision health

Sequencing 100,000 human genomics to power AI-driven health-care innovation that delivers more personalized and preventative health care.

Canadian Precision Health Initiative underway

Agriculture and food

Reducing the carbon footprint of food production systems—building their resiliency, environmental sustainability and economic viability.

Climate-Smart Agriculture and Food Systems initiative underway

Bioeconomy and natural resources

Fueling Canadian productivity and economic growth while safeguarding the biodiversity and health of the environment.

New initiative in development

Commercialization and adoption

Transforming Canadian industry across key economic sectors and driving innovation and productivity among homegrown companies.

Genomic Applications Partnership Program underway



Importance of Diversity for Equitable Health Outcomes

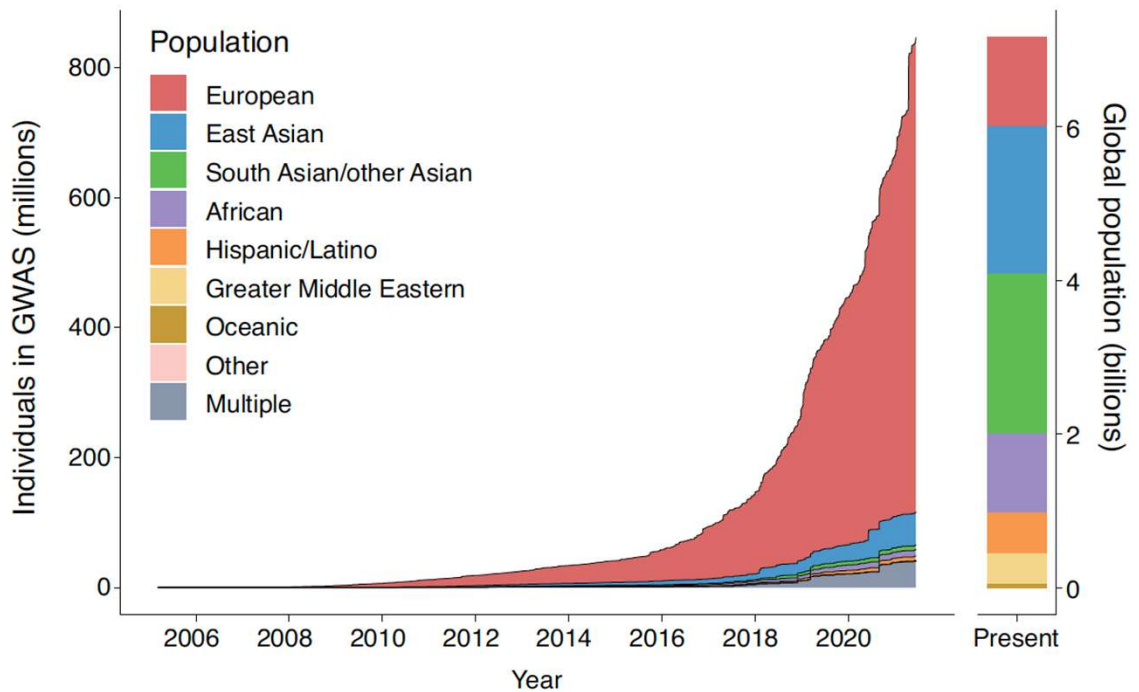


Fig. 1 | The proportion of samples from individuals cumulatively reported by the GWAS Catalog¹ as of 8 July 2021.

Fraction of European genetic ancestry individuals in GWAS :
from 81% in 2016 to 86% in 2021

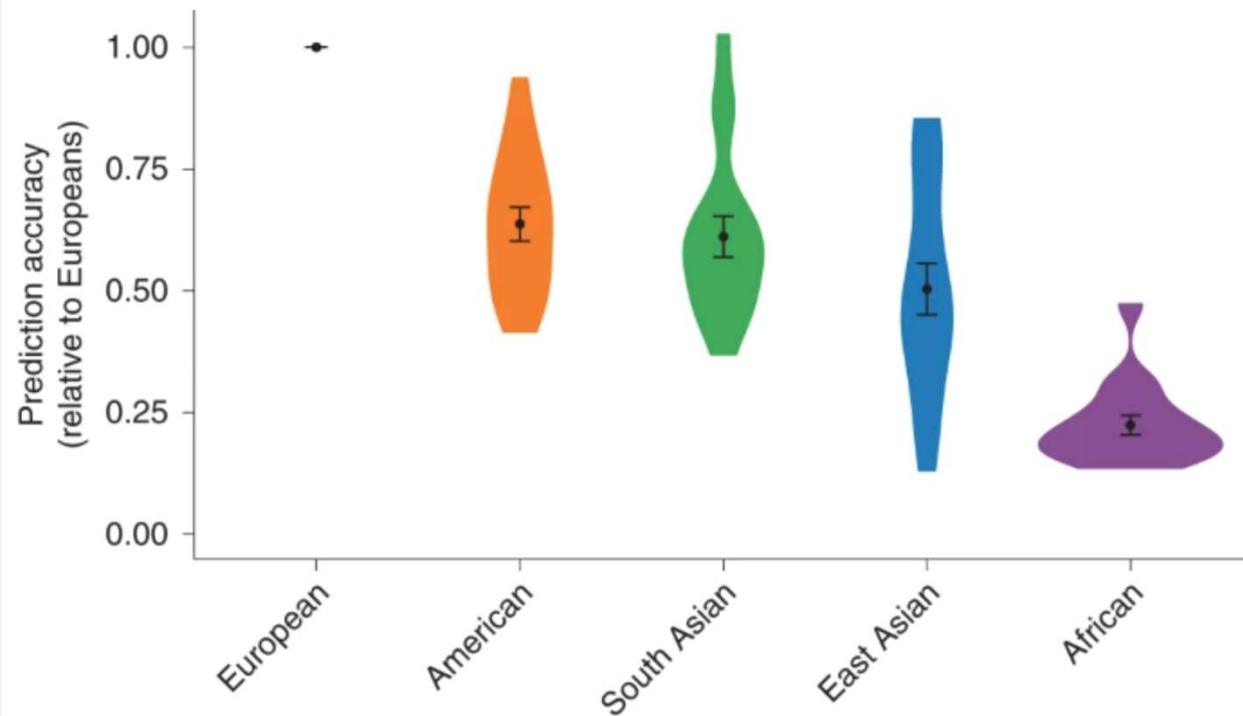
Consequences

- Genetic Misdiagnoses
- Pharmacogenomics gaps
- Genotyping arrays biases
- Polygenic Risks Scores Accuracy

Fatumo et al., 2022, *Nature Medicine*, <https://pubmed.ncbi.nlm.nih.gov/35145307/>

Polygenic Risk Score Accuracy Compared to European-ancestry

Fig. 3: Prediction accuracy relative to European-ancestry individuals across 17 quantitative traits and 5 continental populations in the UKBB.



Canadian Precision Health Initiative

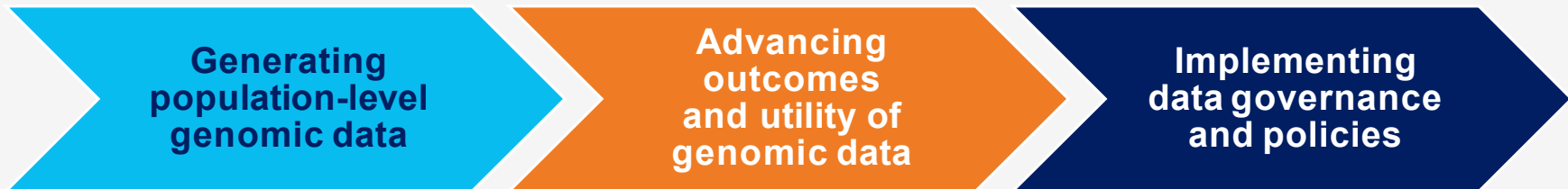
*The future of health care is
personal... and data driven*

100K human genome sequences
in four years | **12** foundational
projects | **\$200M** total investment
expected, including **\$81M** in
federal investment through
Genome Canada and co-funding
through Genome Centres



The Canadian Precision Health Initiative (CPHI): Building a foundational dataset (100K+ genomes)

Building Canada's largest-ever collection of **AI ready human genomic data**—starting with more than 100K new genomes



The CPHI will be fueled by an estimated total investment of \$200M, including \$81M in Government of Canada investment through Genome Canada and co-funding from industry, academia and public sector partners. The initiative will:

Enhance health care options for all Canadians

Enabling more precise, personalized, predictive, preventative and cost-effective healthcare options for Canadians.

Drive economic growth

Empowering Canadian scientists and companies to accelerate development of world-leading, next-generation health solutions and clean economic growth.

Strengthen health security and data sovereignty

Increasing access to Canadian genomic data needed to combat major public health threats, including pandemics.



Indigenous Data Governance

We are taking a distinct approach to Indigenous data

- Guided by the principles of the United Nations Declaration on the Rights of Indigenous Peoples (UNDRIP)
- Upheld through core principles—including [OCAP®](#) and [CARE](#)—alongside other distinctions-based and community-led frameworks
- This approach ensures that genomic activities are led by Indigenous priorities, respects the integrity of biocultural heritage and are rooted in the sovereign right to self-governance
- These commitments requires clearer, earlier and more sustained dialogue with Indigenous leadership and communities, including national and regional organizations
- That framework continues to be developed through ongoing discussion and collaboration across a complex data and research ecosystem

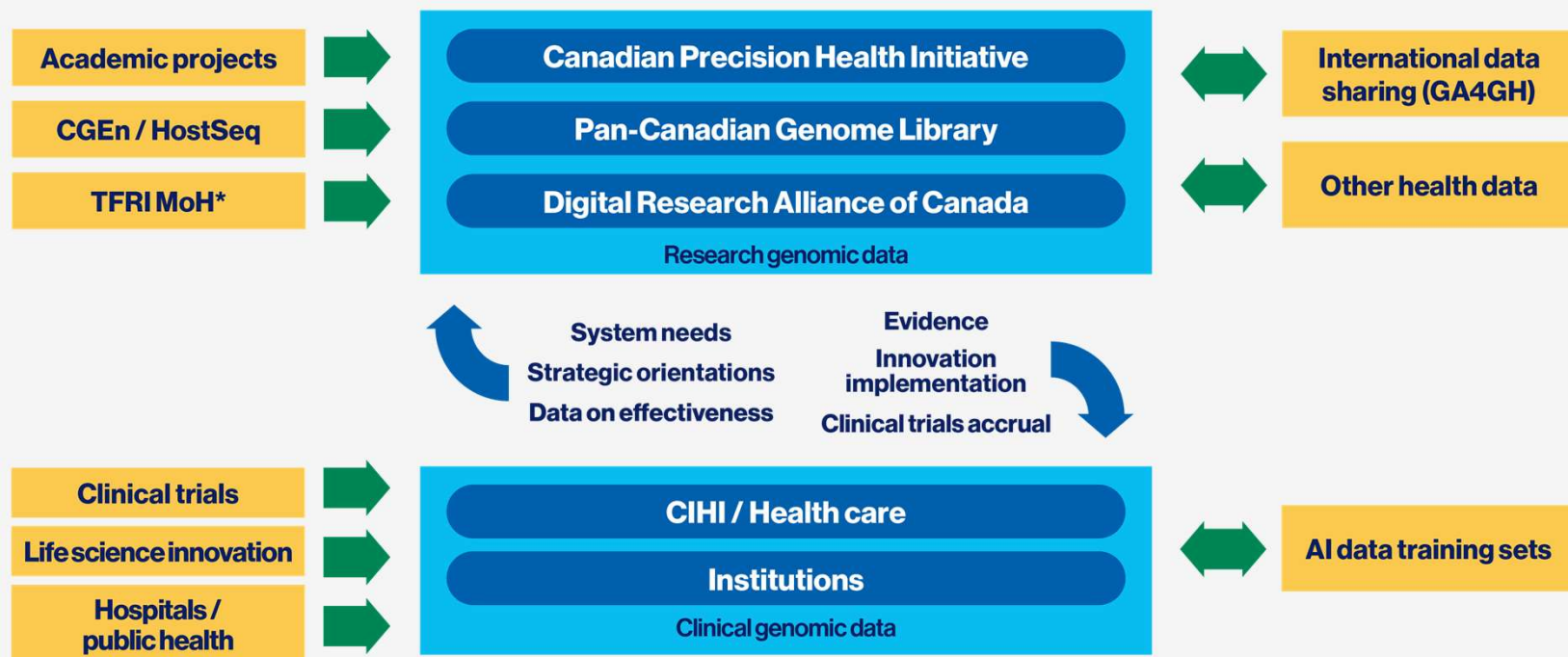


Canadian Precision Health Initiative portfolio

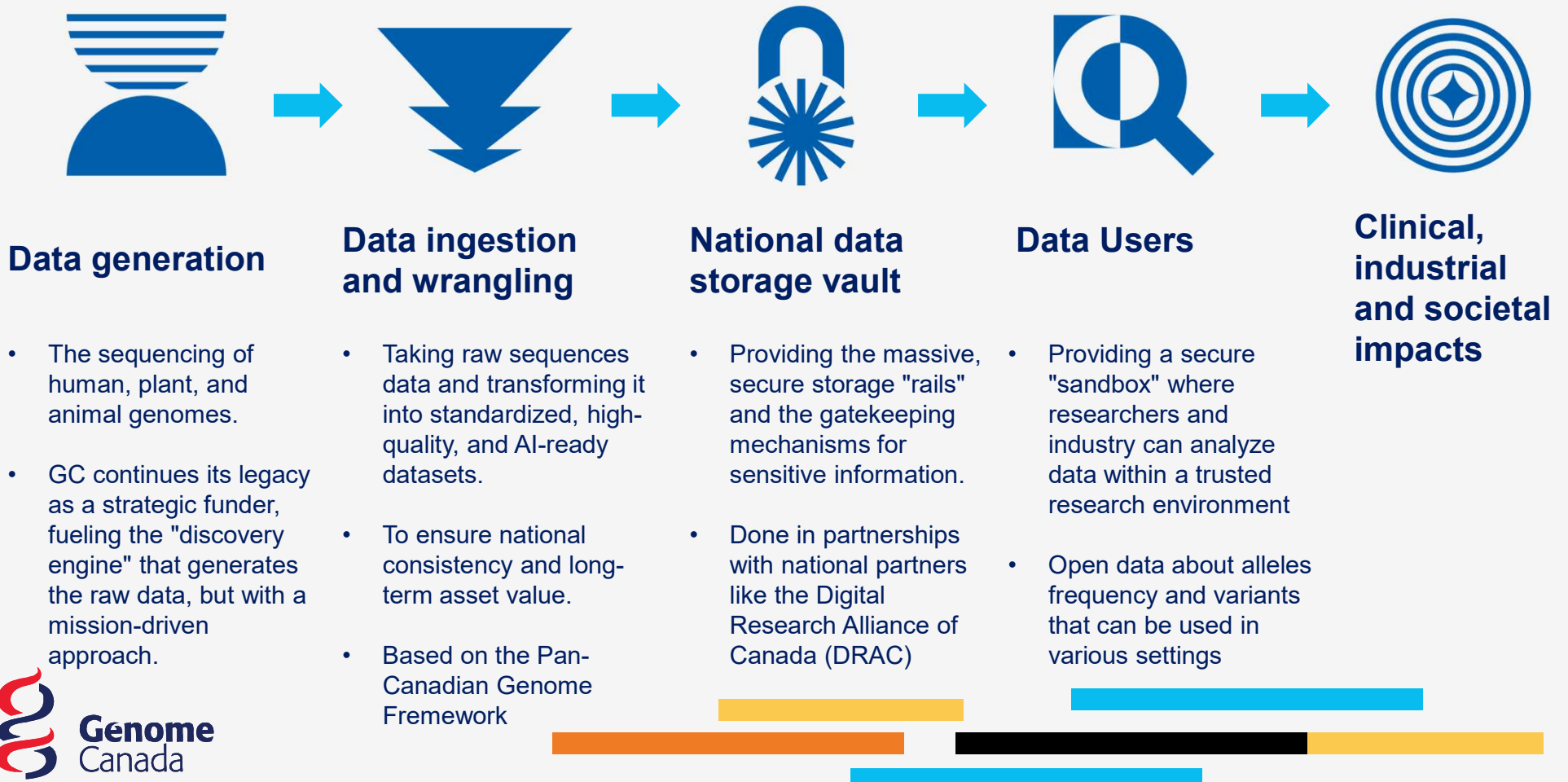
- Neurological disorders
- Rare diseases
- Pediatric cohort
- Cerebrovascular accident
- Oncology
- Cohort of Black individuals
- Newborn sequencing
- Cardiovascular
- Precision child health
- Neurodevelopment, mental health and brain injury
- Children's mental health
- Multi-omics data



Canadian health genomics coordination: Supported by the Canadian Genomic and AI Strategies



Genomic research data stewardship approach



Towards CPHI Phase2

Goal

- Demonstrate and maximize the value and application of the CPHI data and Indigenous genomic data

Approach

- Work with key end users, both from the public and private sectors, communities, and decision-makers to plan the next phase of investments

Objectives (under development)

- Invest in projects that can bring a solution or support a clearly identified needs
- Optimize the translation of innovations for direct impacts
- Capitalize on the diversity of the CPHI data representing Canada's population
- Catalyze the creation and development of new genomics tools and technologies
- Support Indigenous-led genomic projects

Tentative timeline

- Consultation: Q2-Q3 2026-27
- Launch of FO: Q4 2026-27 or Q1 2027-28
- Funding start date: Q2-Q3 2028-29



Growing the dataset :

AI revolutionizing health care

Clinical predictive models

Pediatric generational cohort

Clinical integration with EHRs

AI patterns mining of EHRs

Driving innovation and economic growth

Genomic-based clinical trials

AI-based therapies

Enhanced drug discovery, diagnostics and screening

Deployment of AI health solutions

Integrating across sectors

Agri-food security and resilience

Natural resources and climate change

Defence and biosecurity



Canada's genomics advantage

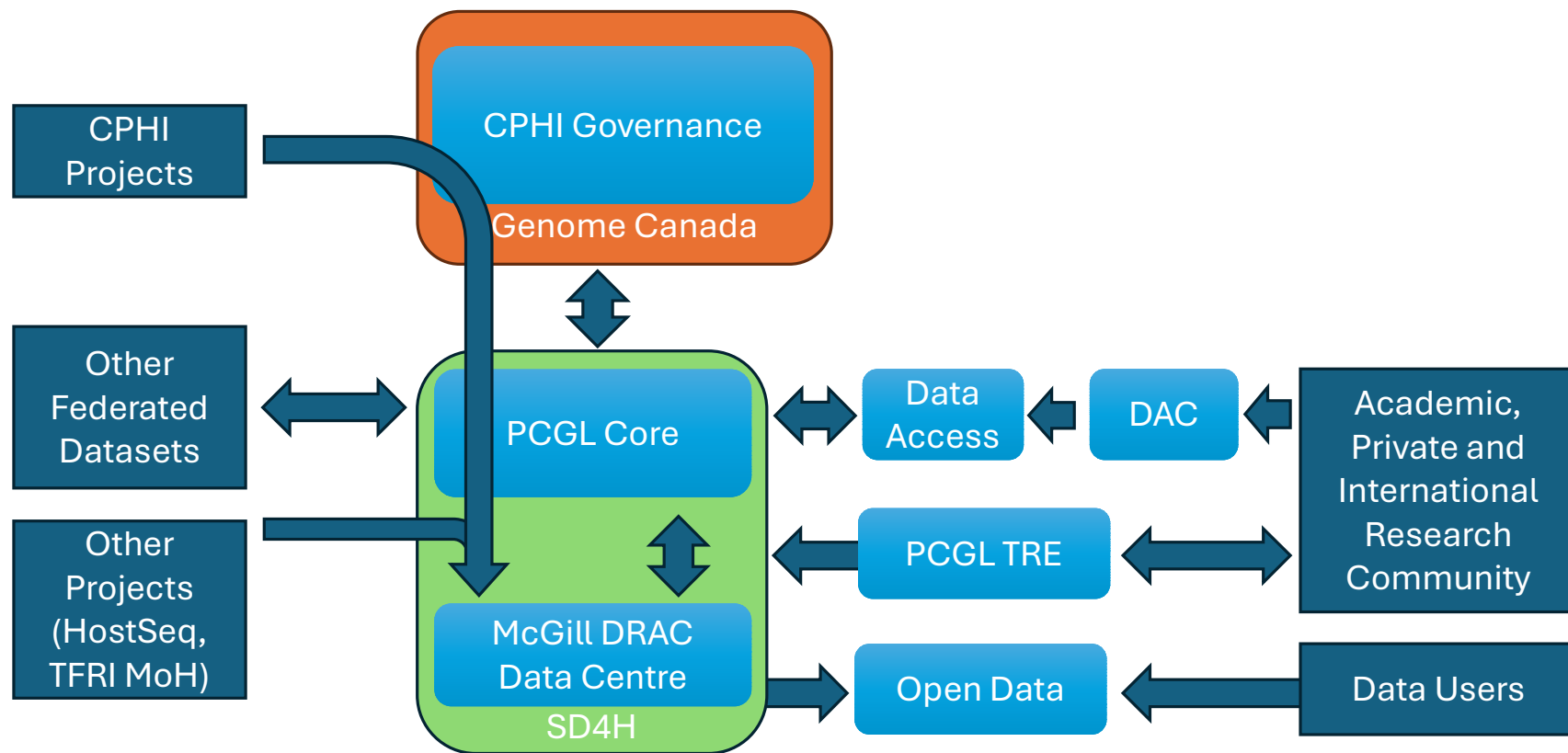
A world-class AI ecosystem fueled by *high-quality big data*

Canada's AI ecosystem—anchored by companies such as **Cohere** and institutions like **AMII**, the **Vector Institute**, and **Mila**—is globally recognized. **Canada ranks 8th in the world for AI capacity** (Tortoise Global AI Index), and this strength is increasingly integrated into genomics research. Cohere

Large-scale collaborations like the Genome Canada-led **Canadian Precision Health Initiative (CPHI)** are building the data infrastructure to power AI-driven health innovation, with over 100,000 genomes being sequenced in four years.

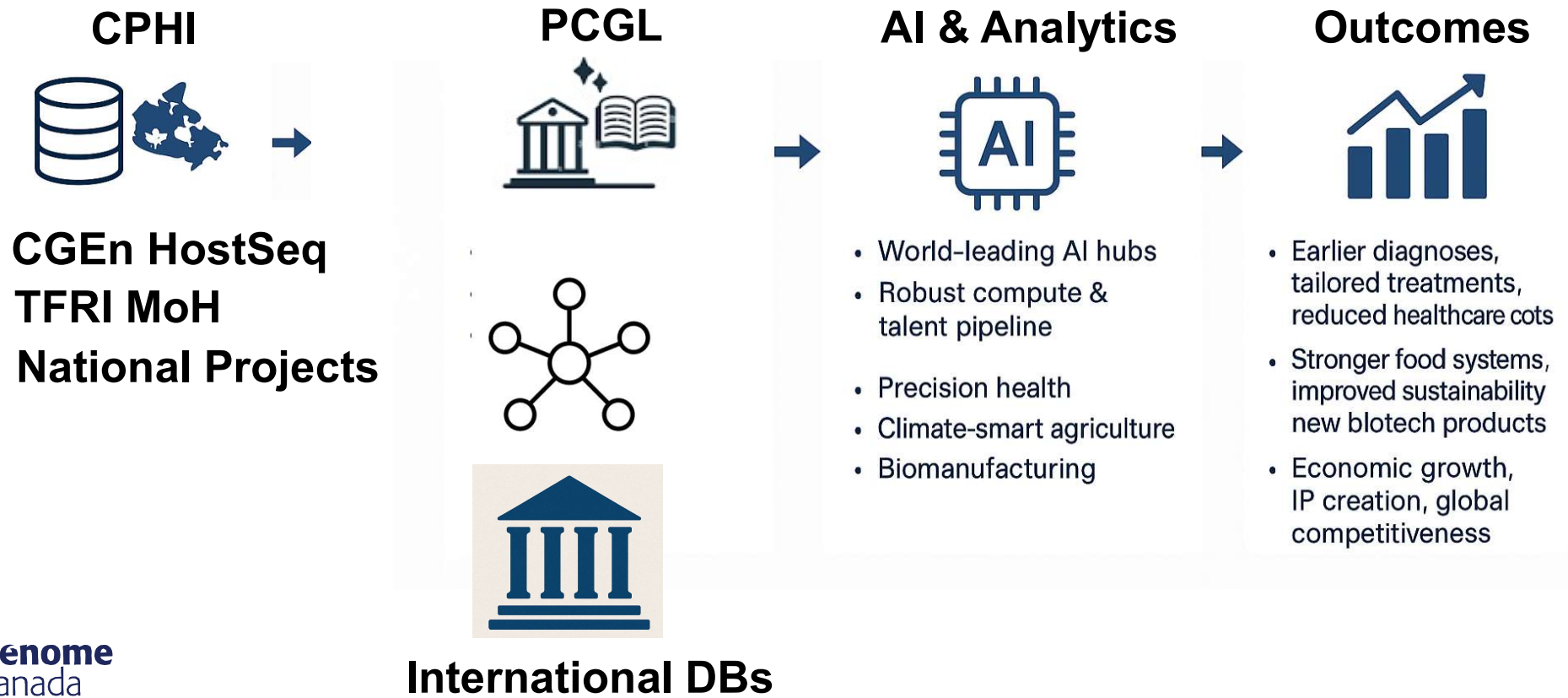


PCGL CPHI Data Governance Model



GA4GH harnessing genomics and AI power internationally

Turning data into health, economic and societal impact



Genome Canada's main office is located on the unceded traditional land of the **Anishinabe Algonquin Nation**

Canada^{ca}

Genome Canada gratefully acknowledges the support of the Government of Canada, and specifically Innovation, Science and Economic Development Canada (ISED), the lead investor in our challenge-driven genomics initiatives.