



Global Alliance for Genomics & Health

Collaborate. Innovate. Accelerate.

Global Initiatives in Genomics and Health



Peter Goodhand
CEO • GA4GH

Outline

1 Why do we need standards?

2 GA4GH

3 Global Collaborations

4 Japanese Example

5 Getting Involved

Since data is
distributed globally,
we need interoperable
standards to answer
research questions



Standards are essential



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Interoperability is key to data sharing and enabling the necessary ecosystem



What makes a good standard?



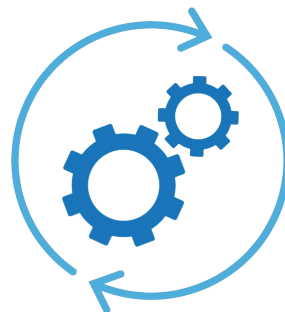
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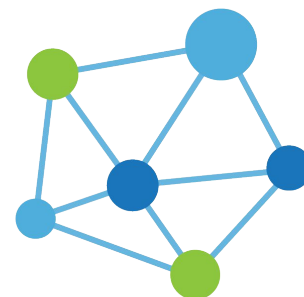
Clear and specific
specification



Meets the needs of
everyone in the
system



Easy to adopt



Works as part of a
broader ecosystem

For standards to help...

...they need to be deployed.

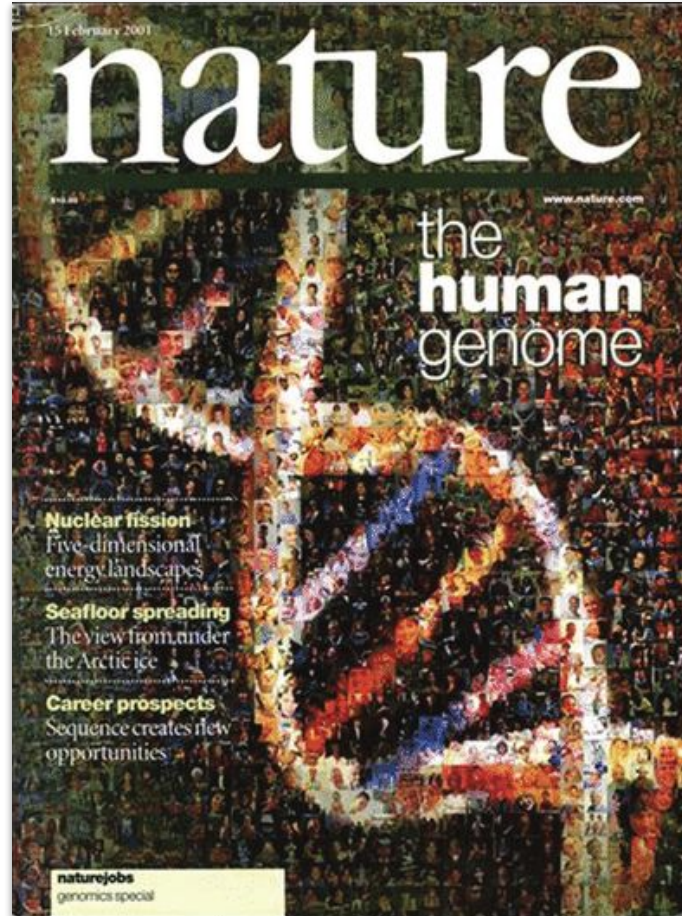
To be deployed...

...they need to be built with
adoption in mind.

To be adopted...

**...we need to check what
people need (everywhere).**

How do we advance genomics globally?



The 1000 Genomes Project ecosystem



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Common consent across
all 26 populations and
5 continents



Common resources:
Cell lines, arrays, exomes
and genomic sequences



File formats (SAM and
VCF) and data sharing
methods



Software: samtools,
FreeBayes, GATK,
VCFtools, tabix, etc.

Interoperable

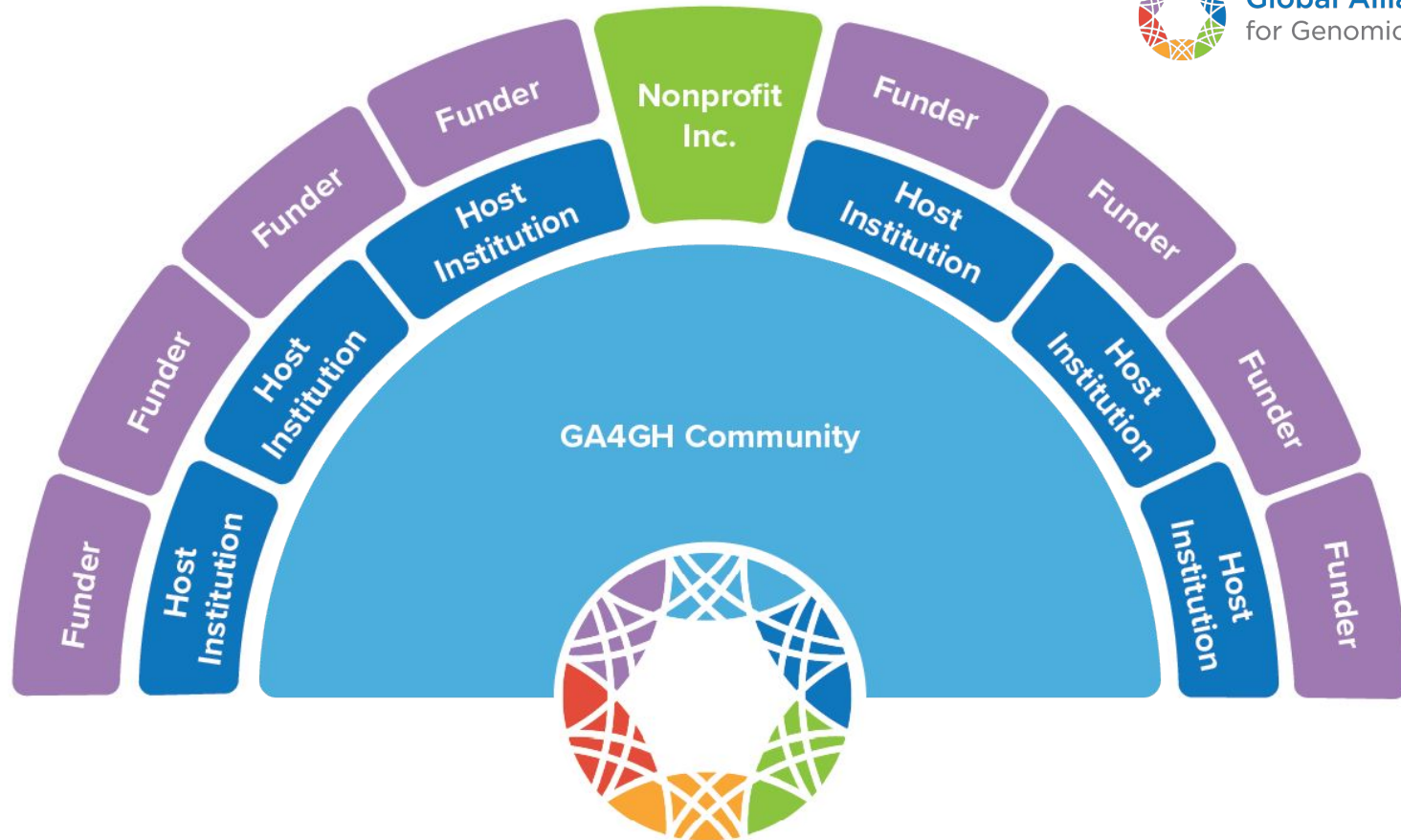
Together, able to address research questions and lay foundation for future projects



- To fully realize the benefits of genomics in health, it is necessary to **work globally**
- **Collaboration** is the only way to build standards and achieve global data sharing

**GA4GH is a
global forum**





THE GA4GH MISSION...

The **Global Alliance for Genomics and Health** aims to accelerate progress in genomic science and human health by developing standards and framing policy for responsible genomic and health-related data sharing.

The GA4GH Ecosystem



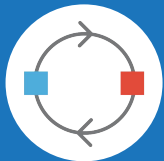
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GA4GH aims to...



Enable **international** data sharing



Promote sharing across the **translational continuum**



Encourage technology-enabled **federated approaches**



Promote **interoperability**

GA4GH achieves this by...



Convening stakeholders



Catalyzing sharing of data



Creating harmonized approaches



Acting as a clearinghouse



Fostering innovation



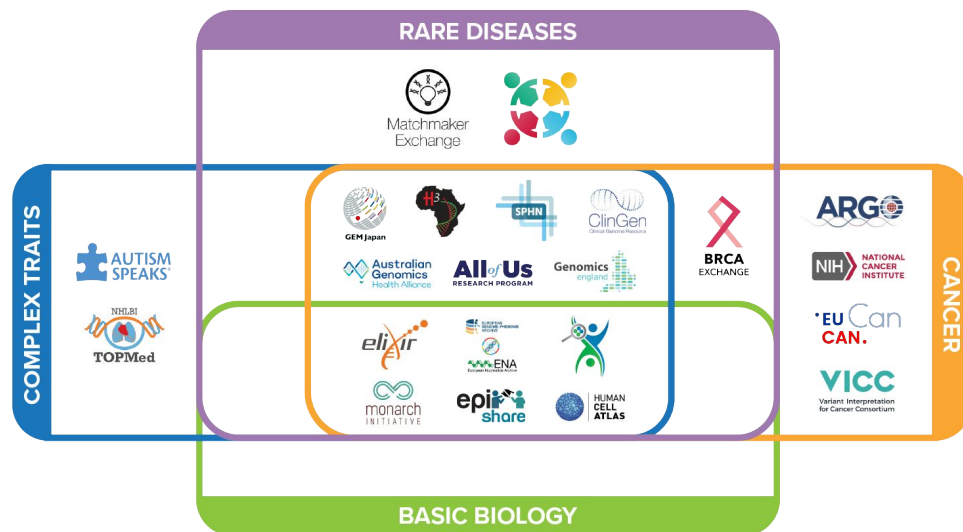
Committing to responsible data sharing

Use cases: what are we trying to do?

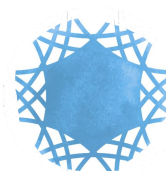


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We have many sources of real world scientific and health use cases



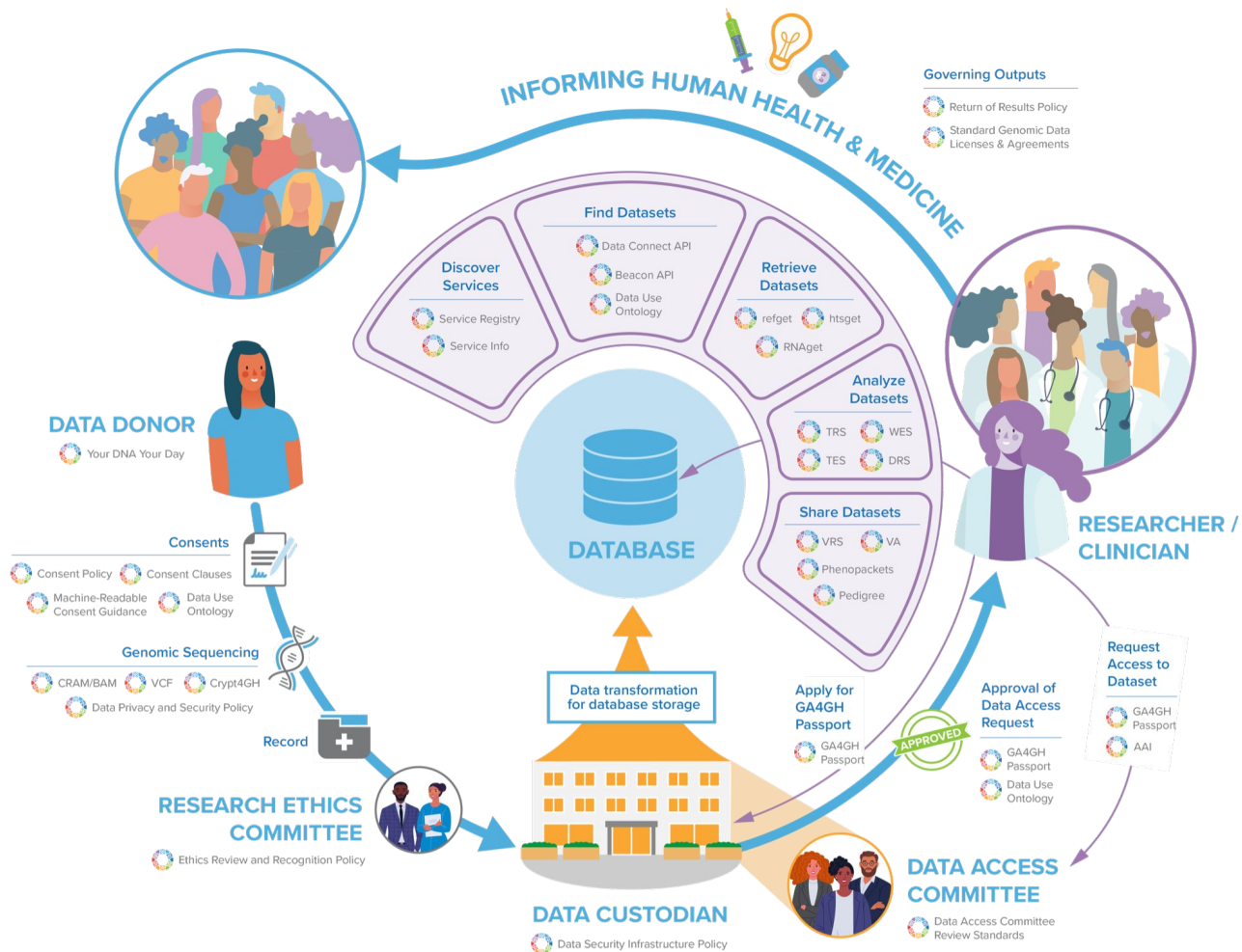
Driver Projects



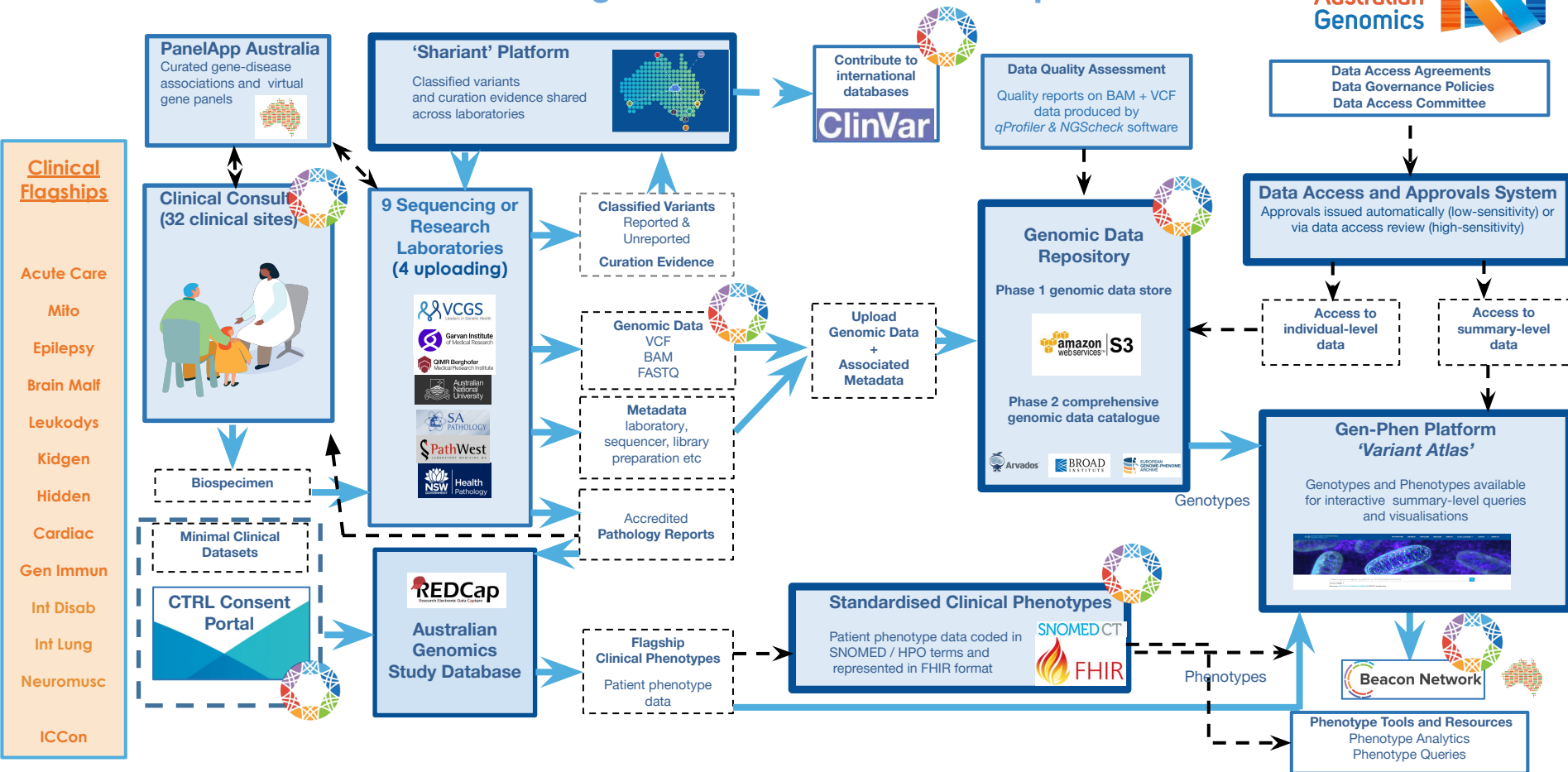
GENOMICS IN HEALTH
IMPLEMENTATION FORUM

Cancer Community

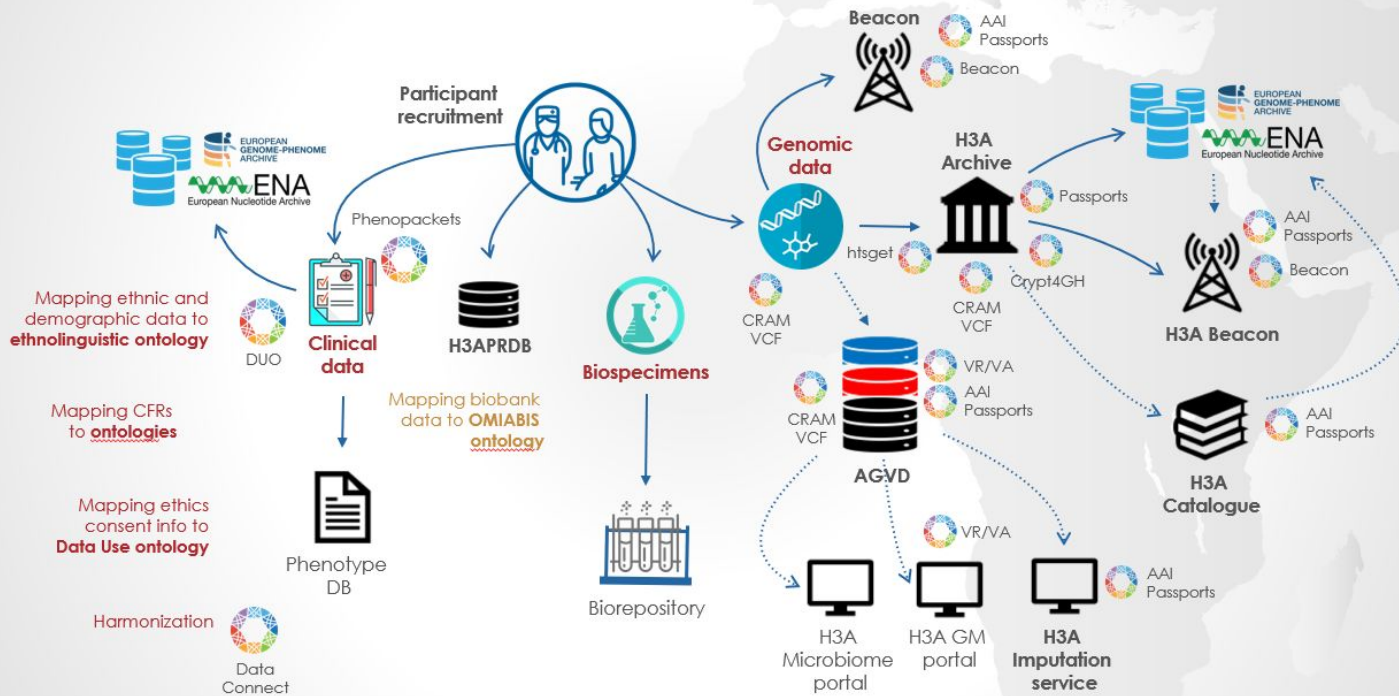
...and many more



Australian Genomics: Data Management Work Flow and Capabilities



H3Africa genomic research ecosystem



Current GHIF Members

Foundation
members:

Australian
Genomics



Genomics
england



GEM Japan



Swiss
Personalized
Health
Network



AllForOne
Canada's Precision
Health Partnership



IndiGen Genomics for
Public Health
in India



Global Genomic
Medicine Collaborative

Nordic Alliance
for Clinical Genomics



عضو في مؤسسة قطر
Member of Qatar Foundation



ClinGen
Clinical Genome Resource



PRECISE
Precision Health Research
Singapore



estonian genome center
university of tartu



Brazilian Initiative on
PRECISION MEDICINE



International HundredK+
Cohorts Consortium (IHCC)



CINECA



DANISH NATIONAL
GENOME CENTER

Collaborations



CINECA

Common Infrastructure for National Cohorts in Europe, Canada, and Africa



International 100K Cohort Consortium



Global Genomic
Medicine Collaborative

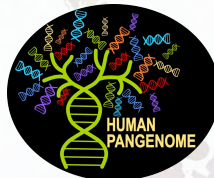


INTERNATIONAL
COMMON DISEASE
ALLIANCE

Maps to
Mechanisms to
Medicine



Beyond One Million
Genomes Project



Human Pangenome
Project (HPP)



International COVID-19
Data Alliance



WORLD
ECONOMIC
FORUM



Science Council
World Health
Organization

THE MEDICAL GENOME
INITIATIVE



PUBLIC HEALTH ALLIANCE FOR
GENOMIC EPIDEMIOLOGY

HUMAN GENOME ORGANISATION

HUGO

Human Genome Organisation

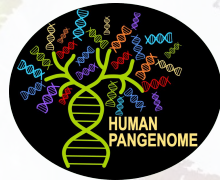


HUMAN GENOME
VARIATION SOCIETY



THE
HUMAN VARIOME
PROJECT

Global Collaborations



Human Pangenome
Project (HPP)



World Health
Organization

Science Council



International 100K Cohort Consortium



Global Genomic
Medicine Collaborative



PUBLIC HEALTH ALLIANCE FOR
GENOMIC EPIDEMIOLOGY



Human Genome Organisation



Alignment with Other Standards Organizations



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Research

Clinical Research

Clinical Care



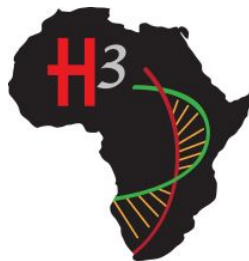
Recent implementation updates



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GEM Japan



National Heart, Lung,
and Blood Institute



IRUD



PRECISE
Precision Health Research
Singapore

Watch these implementer talks from 9th Plenary: bit.ly/9thplenary-videos

Example

Melissa Cline & Japan



CIHR COVID-19 Interoperability Initiative



CanCOGeN 



COVID Cloud



Public Health Alliance for
Genomic Epidemiology



COVID-19
GENOMICS
UK CONSORTIUM



COVID-19 *Data Portal*

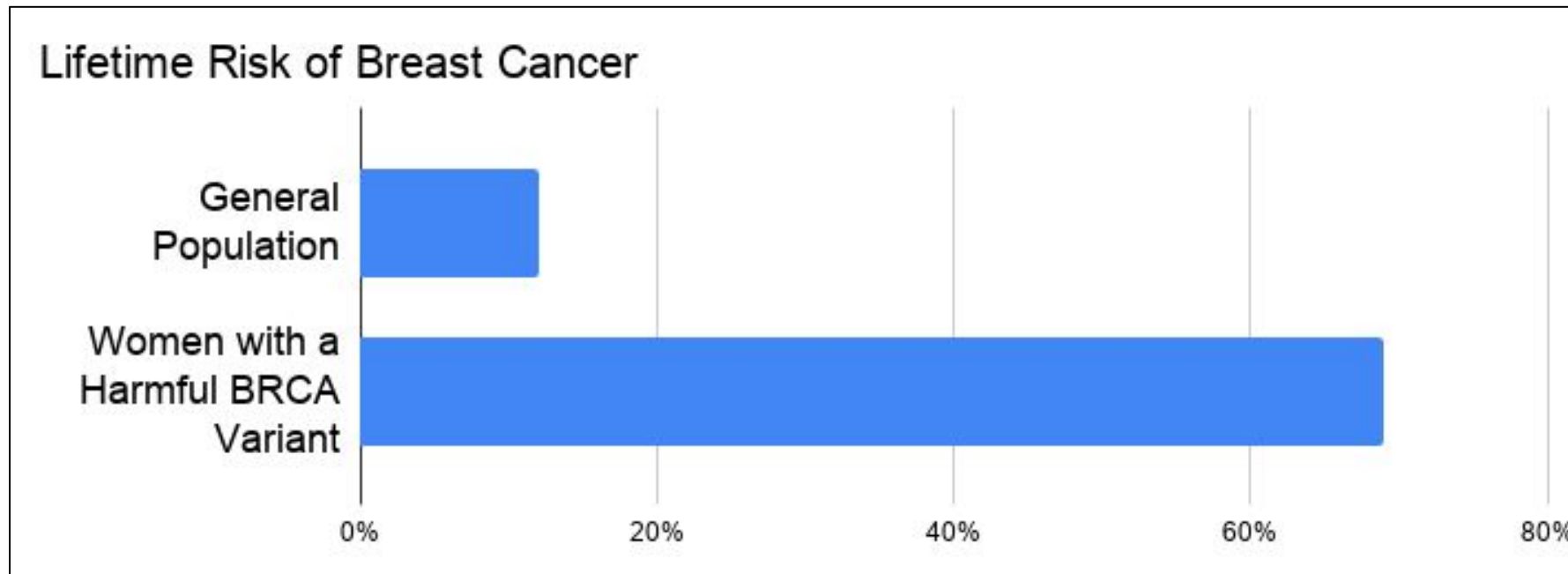


COVID-19 hg

BRCA Variation and Cancer Risk



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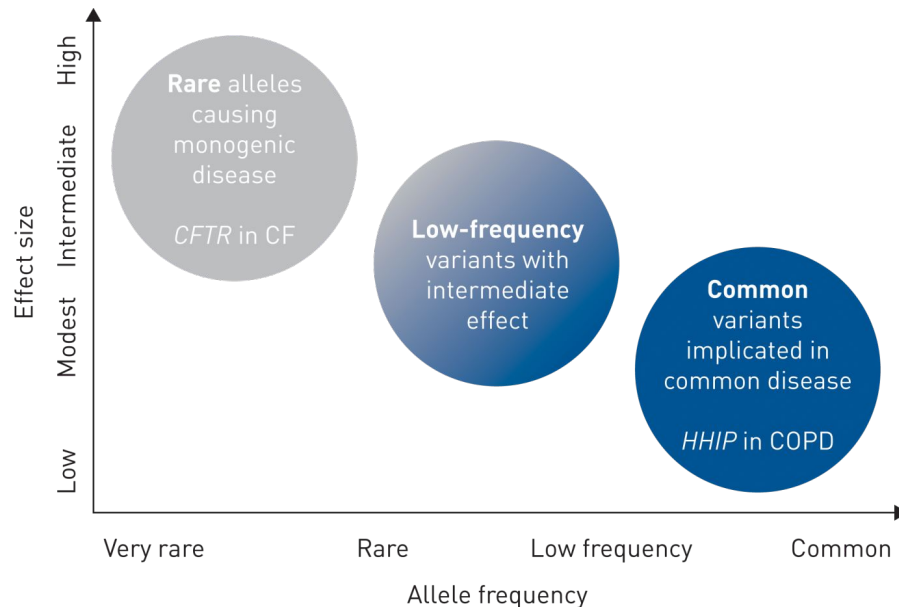


BRCA variation can also lead to ovarian, prostate, and pancreatic cancers



Proof of Concept with BioBank Japan

- **Rationale:** Disease-causing cancer variants are rare by definition
- If a variant is frequent in some continental population, it can be classified as benign with no further evidence



Federated analysis of BioBank Japan Data

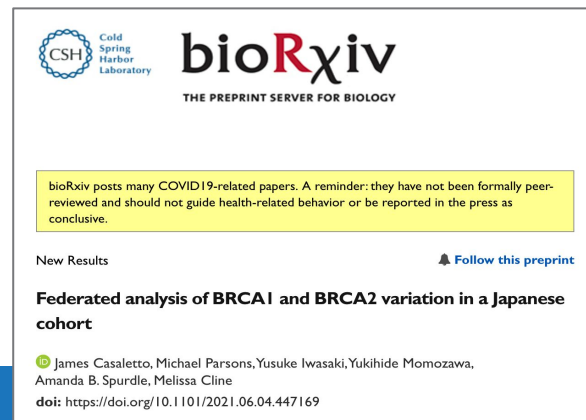


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- RIKEN holds a large cohort of cancer patients and controls from BioBank Japan, which they cannot share directly.
- We shared a Docker container with them to analyze their patient cohort for variant co-occurrences and allele frequencies
- The container generated variant-level data, which we are now using together with the ENIGMA Consortium to interpret BRCA variants!



Yukihide Momozawa, RIKEN





Acknowledgements



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GA4GH Core Funders



GA4GH Supporting Funders



GA4GH Staff and Contributors

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