Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.
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.
“Everyone has the right to the protection of the moral and material interests resulting from any scientific, literary or artistic production of which he is the author.”
Framework for Responsible Sharing of Genomic and Health-Related Data

ga4gh.org/framework

FOUNDATIONAL PRINCIPLES

• Respect Individuals, Families and Communities
• Advance Research and Scientific Knowledge
• Promote Health, Wellbeing and the Fair Distribution of Benefits
• Foster Trust, Integrity and Reciprocity

AIMS OF THE FRAMEWORK

• Foster responsible data sharing
• Protect and promote the welfare, rights, and interests of groups and individuals who donate their data
• Provide benchmarks for accountability
• Establish a framework for greater international data sharing, cooperation, collaboration, and governance
How Many Genomes?

RESEARCH

HEALTHCARE
60M individuals
132.5 sequences

CLINICAL TRIALS
2.7-3M individuals

COHORTS
140M individuals
200+ Genomic Data Initiatives Globally

- Clinical/Genomic Medicine: 40 Initiatives
- Research: 70 Initiatives
- National: From 15 Countries
- Cohorts: Globally 64
THE OPPORTUNITY...

If we can enable secondary use of clinical genomic data for research, we will have a virtual cohort of >60 million samples by 2025.
Global Genomic Data Sharing Can…

- Demonstrate patterns in health & disease
- Increase statistical significance of analyses
- Lead to “stronger” variant interpretations
- Increase accurate diagnosis
- Advance precision medicine
Different Approaches to Data Sharing

Centralized Genomic Knowledge Bases

Data Commons
Trusted, controlled single repository of multiple datasets

Hub and Spoke Federation
Common data elements, structures, access, and usage rules

Linkage of distributed and disparate datasets
A New Paradigm for Data Sharing

Data Copying → Data Visiting
The Future: Federation

“A grouping of autonomous organizations and datasets with a centralised control”

Federation allows us to….

1. **Move analysis to data**, not aggregate data close to each researcher

2. **Have broad, reciprocal data access methods** which respect national processes and patient consent

3. Transfer methods and skills into the **healthcare sector**

4. **Leverage healthcare data** to make more discoveries on humans
Federation

Open Research Data

- Aggregate data globally
- Download, analyze locally
- Continues for basic research

Healthcare Data with Research Use

- Aggregate data locally (via VMs)
- Collate analyses
- New approach for research and healthcare
Core Principles of Data Sharing

1. Enable International Data Sharing
2. Encourage technology-enabled federated approaches
3. Sharing across the translational continuum
4. Promote Interoperability
The GA4GH Ecosystem

- 3000+ Subscribers
- 600+ Organizational Members
- 90+ Countries
- 24 Driver Projects
- 8 Work Streams
- 20 Technical Standards
- 7 Regulatory Policies & Frameworks
- 40+ Implementations & Deployments

Enabling the global learning health system
### Work Streams vs. Driver Projects

<table>
<thead>
<tr>
<th>Work Streams</th>
<th>Driver Projects</th>
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<tbody>
<tr>
<td>• Internal to GA4GH</td>
<td>• External to GA4GH</td>
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<tr>
<td>• Deliver standards and policy frameworks based on the Strategic Roadmap</td>
<td>• Provide input towards the Strategic Roadmap and standards development</td>
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<td>• Run by 2 volunteer leads within the community</td>
<td>• Contribute FTE resources to Work Streams for standards development</td>
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<td>• Contributors come from a variety of projects and organizations</td>
<td>• Pilot implementations for new standards</td>
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<td>• <em>Example</em>: Data Use &amp; Researcher Identities</td>
<td>• <em>Examples</em></td>
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**Examples**

![ARG Logo](image1.png)

![BRCA Logo](image2.png)

![Australian Genomics Health Alliance Logo](image3.png)
Alignment with Other Genomics Standards Organizations

- Research
- Clinical Research
- Clinical Care

- HL7 International
- SNOMED
- ISO TC215/WG2
Public willingness to share DNA*

Would you donate your anonymous DNA information and medical information for use by:

Medical doctors - 50-60% (Japan and Egypt lowest)
Non-profit researchers – 40-50% (Japan, Egypt and India lowest)
For-profit researchers – 20-40% (Germany, Japan and UK lowest)

*summarized from “Your DNA Your Say” survey led by Dr Anna Middleton, Cambridge UK

21 countries representing 3 billion people X 20% = ?
Global Advancement for Health

Interoperable APIs, standards & policy frameworks support global data sharing across sectors

Health data sharing across traditional silos supports advancement across all relevant sectors
COVID-19 Distractions & Opportunities

COVID-19 challenged regular work processes, availability and priorities

But from this, there are opportunities:

1. Increased awareness of the need to share data at scale and across boundaries

2. Benefits of a virtual meeting – greater participation (700+ registrants)

3. Tools to enable global collaboration