Working together to achieve global genomic data sharing

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Regional Meeting
Human Genomics for Health: Enhancing the Impact of Effective Research
THE GA4GH MISSION...

The Global Alliance for Genomics and Health aims to accelerate progress in human health by establishing a shared global approach to responsible, broad, and democratised use of genomic and related health data.
GA4GH ecosystem and outputs

- 44 GA4GH products
- 200+ implementations & deployments
- 100+ countries
- 500+ Organisational Members
- 28 Driver Projects
- 450+ active contributors
GA4GH: A decade of collaboration

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<th>Assigned Expert Funders/Employers</th>
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Driver Projects
- All of Us
- Autism Speaks
-vements
- epiShare
- NHLBI BioData
- Catalyst

Strategic relationships
- elixir
- NIH
- HDR UK
- NCPI
- Cancer

Core Funders
- CIHR IRSC
- Medical Research Council
- Genome Canada
- NIHR National Institute for Health Research
- NHGRI National Human Genome Research Institute
- Broad Institute
- EMBL-EBI
- Illumina
- Roche

Host Institutions
- OICR
- Sanger Institute
- Catalyst

Supporting Funders
- abbvie
- Genentech
- Nature Publishing Group
- EuroBiomed
GA4GH Communities of Interest

Domain-specific groups promoting global cooperation, data sharing and collaborative research through identifying the need for new standards, and implementing existing GA4GH standards.

- Cancer
- Infectious Disease
- Rare Disease
- Neurosciences

Opportunities for collaboration
Learn from others’ implementations
Develop a study group
Propose a GIF project
In 2020, we identified three strategic imperatives...

**Interoperability & alignment**
Truly interoperable standards will enable solutions that can encompass multiple components of a pipeline and multiple platforms and use cases.

**Implementation support**
Driver projects and the community are able to quickly adopt and implement GA4GH standards, driving broad uptake and interoperability.

**Engaging with healthcare and clinical standards**
Ability to efficiently and effectively respond to the needs of clinical stakeholders, developing standards that support the healthcare and clinical communities.
INFORMING HUMAN HEALTH & MEDICINE

DATABASE

Find Datasets
- Data Connect API
- Beacon API
- Data Use Ontology

Retrieve Datasets
- mfget
- Xdeg

Analyze Datasets
- TRS
- WES
- TES
- ORS

Share Datasets
- VAF
- Pedigree

Governing Outputs
- Return of Results Policy
- Standard Genetic Data Licences & Agreements

DATA DONOR
- Your DNA: Your Day

Consents
- Consent Notice
- Consent Process
- Machine-Readable Consent Guidance

Genomic Sequencing
- CRAM/BIOT
- VCF
- CryptoGH
- Data Privacy and Security Policy

RESEARCH ETHICS COMMITTEE
- Ethics Review and Recognition Policy

RESEARCHER / CLINICIAN

Apply for GA4GH Passport
- GA4GH Passport
- GA4GH Passport

Approval of Data Access Request
- GA4GH Passport
- GA4GH Passport

REDACTION COMMITTEE
- Data Access Committee Review Standards

DATA STEWARD
- Data Security Infrastructure Policy

Record

Data transformation for database storage
Combining genomic data and health data at scale...

...requires a paradigm shift in data sharing
A new paradigm for data sharing
Global collaborations

- International HundredK+ Cohorts Consortium
- Global Genomic Medicine Consortium
- Public Health Alliance for Genomic Epidemiology
- Global Alliance for Genomics & Health
- World Health Organization
- TAG-G
- INTERNATIONAL COMMON DISEASE ALLIANCE
- THE MEDICAL GENOME INITIATIVE
- ICODA
- Research Data Alliance
Thank you!

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