Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.
Mission

To accelerate progress in human health by helping to establish a common framework of harmonized approaches to enable effective and responsible sharing of genomic and clinical data, and by catalyzing data sharing projects that drive and demonstrate the value of data sharing.
2015/2016 Road Map objectives

Results
Objective 1: Develop priority products
Objective 2: Deliver packaged, working solutions
Objective 3: Support demonstration projects

Relevance
Objective 4: Align with major data collection and sharing efforts
Objective 5: Communicate strategically with key audiences
Objective 6: Establish GA4GH as a thought leader

Sustainability
Objective 7: Build leadership and participation
Objective 8: Expand organizational capacity and funding streams
Global Alliance members include:

1. Universities and research institutes (33%)
2. Academic medical centers and health systems (11%)
3. Disease advocacy organizations and patient groups (5%)
4. Consortia and professional societies (5%)
5. Funders and agencies (6%)
6. Life science and information technology companies (40%)
Countries

56 Countries represented in the Global Alliance
- 370 Organizational Members from 35 countries (in bold)
- 547 Individual Members from 53 countries

Afghanistan  Afghanistan
Argentina    Argentina
Australia    Australia
Austria      Austria
Belgium      Belgium
Brazil       Brazil
Cameroon     Cameroon
Canada       Canada
China        China
Colombia     Colombia
Congo        Congo
Croatia      Croatia
Czech Republic Czech Republic
Denmark      Denmark
Finland      Finland
France       France

Georgia      Georgia
Germany      Germany
Ghana        Ghana
Greece       Greece
Hong Kong    Hong Kong
India        India
Ireland      Ireland
Israel       Israel
Italy        Italy
Japan        Japan
Kenya        Kenya
Korea, Republic of Korea
Luxembourg   Luxembourg
Malaysia     Malaysia
Mexico       Mexico
Morocco      Morocco
Nepal        Nepal
Netherlands  Netherlands
New Zealand  New Zealand
Nicaragua    Nicaragua
Nigeria      Nigeria
Norway       Norway
Peru         Peru
Philippines  Philippines
Portugal     Portugal
Qatar        Qatar
Russia       Russia
Sierra Leone Sierra Leone
Singapore    Singapore
Slovenia     Slovenia
South Africa South Africa
Spain        Spain

Sudan        Sudan
Sweden       Sweden
Switzerland  Switzerland
Taiwan       Taiwan
Turkey       Turkey
Uganda       Uganda
Ukraine      Ukraine
United Kingdom United Kingdom
United States United States
Venezuela    Venezuela
Virgin Islands, U.S. Virgin Islands, U.S.

Last Update: October 29, 2015
Leverage

7.5 professional staff
Support and enable

898 volunteer contributors
### Current Initiatives

<table>
<thead>
<tr>
<th><strong>Clinical</strong></th>
<th><strong>Data</strong></th>
<th><strong>Regulatory and Ethics</strong></th>
<th><strong>Security</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical Cancer Genome - Cancer Community Intro to GA4GH</td>
<td>Beacon Network</td>
<td>Accountability (Policy)</td>
<td>Cloud Security</td>
</tr>
<tr>
<td>Clinical Cancer Genome - Cancer Data Sharing</td>
<td>Benchmarking</td>
<td>Ageing and Dementia</td>
<td>Incident Response</td>
</tr>
<tr>
<td>Clinical Cancer Genome - Actionable Cancer Genome Initiative</td>
<td>Containers and Workflows</td>
<td>BRCA Ethico-Legal and Advocacy</td>
<td>Security Infrastructure</td>
</tr>
<tr>
<td>eHealth - Pedigree Consent</td>
<td>File Formats</td>
<td>Data Protection Regulation</td>
<td>Software Security</td>
</tr>
<tr>
<td>eHealth - Family History</td>
<td>Genotype2Phenotype Association</td>
<td>Data Sharing Lexicon</td>
<td></td>
</tr>
<tr>
<td>eHealth - Federated Queries</td>
<td>Metadata</td>
<td>Ethics Review Equivalency</td>
<td></td>
</tr>
<tr>
<td>eHealth - Catalogue of Activities</td>
<td>Reference Implementation</td>
<td>Individual Access</td>
<td></td>
</tr>
<tr>
<td>eHealth - Data Sharing</td>
<td>Reference Variation</td>
<td>Machine-Readable Consent</td>
<td></td>
</tr>
<tr>
<td>Phenotype Ontologies - Rare Diseases</td>
<td>RNA and Gene Expression</td>
<td>Paediatric</td>
<td></td>
</tr>
<tr>
<td>Phenotype Ontologies - Cancer / Complex Diseases</td>
<td>Variant Annotation</td>
<td>Privacy Breach Notification</td>
<td></td>
</tr>
</tbody>
</table>

**Completed:**
- Reads
- Framework for Responsible Sharing of Genomic and Health-Related Data
- Consent Policy
- Privacy and Security Policy
- Data Safe Havens

Last Update: October 29, 2015
CWG Initiatives

• Clinical Cancer Genome - Cancer Community Intro to GA4GH
• Clinical Cancer Genome - Cancer Data Sharing
• Clinical Cancer Genome - Actionable Cancer Genome Initiative
• eHealth - Pedigree Consent
• eHealth - Family History
• eHealth - Federated Queries
• eHealth - Catalogue of Activities
• eHealth - Data Sharing
• Phenotype Ontologies - Rare Diseases
• Phenotype Ontologies - Cancer / Complex Diseases
Current demonstration projects

- Undertaken by the members, not by GA4GH as an organization
- Catalyzed and supported by GA4GH coordinators and Working Groups
- Their purpose: to drive learning, to identify requirements, to evaluate value and to coordinate activity

Beacon Project

Matchmaker Exchange

BRCA Challenge
Genomic Matchmaker

Genotypic Data
- Gene A
- Gene B
- Gene C
- Gene D
- Gene E
- Gene F

Phenotypic Data
- Feature 1
- Feature 2
- Feature 3
- Feature 4
- Feature 5

Genotypic Data
- Gene D
- Gene G
- Gene H

Phenotypic Data
- Feature 1
- Feature 3
- Feature 4
- Feature 5
- Feature 6

Notification of Match

Courtesy of Joel Krier
Database connections through common API

Last Update: June 4th 2015

www.matchmakerexchange.org
GA4GH and BRCA Challenge

• Began at the first Global Alliance plenary meeting in London, with leadership from Stephen Chanock and Sir John Burn
• Goals were defined at a Paris meeting several months later with GA4GH, and HVP at UNESCO
• Work was incubated in GA4GH’s Clinical Working Group until early 2015
• Ongoing work is coordinated by GA4GH and overseen by the Steering Committee
• BRCA has its own leadership executive leadership and sub groups focussed on evidence gathering, interpretation, classification, the regulatory and ethical aspects and engaging with patients.
Goals of the Challenge

To improve the care of patients at risk of breast and ovarian cancer using global data sharing and collaboration in the analysis of BRCA1 and BRCA2

1. Share BRCA1 and BRCA2 variants publically
2. Create an environment for collaborative variant curation with access to evidence (e.g. phenotypes, family history, genetic data, and functional studies)
3. Create a curated list of BRCA variants, interpreted by expert consensus, to enable, without dictating, accurate clinical care
4. Address the social, ethical, and legal challenges to global data sharing
5. Create a model for all genes
Current Steps

1. Define data elements to capture, iterate BRCA instantiation of GA4GH API with the help of the Data Working Group

2. Identify and aggregate interpreted variants

3. Solicit and deposit variants not yet shared

4. Formalize federated database system with regard to tiered access for sharing patient data and other protected datasets, microattribution, and ongoing variant curation

5. Establish consensus on rules and terminology for variant classification
Global Understanding: Local Implementation

1. Decision to Study
2. Assemble Relevant Data
3. Analyze Data
4. Interpret Results
5. Journals
6. Conferences
7. Deliver Tailored Message
8. Take Action to Change Practice
9. Decision to Study
GA4GH Outreach

- Mexican Association of Human Genetics meeting
- H3 Africa Meeting in Tanzania
- Qatar Foundation Conference - QBRI
- Multiple meetings in Japan
- HUGO meeting in Kuala Lumpur, Malaysia
- Multi-regional Clinical Trials Group meeting – Cambridge MA
- UK members meeting and leveraging EBI and ELIXIR
- Netherlands - International Mutation Conference
- Brazilian Society of Medical Genetics
- ESHG – Glasgow
- Brazilian Society of Genetics
2015 and beyond

- **Individuals** are key to **creating** the new tools, frameworks, enablers, solutions and opportunities.

- **Organizations** are key to ensuring the **dissemination and adoption** of best practices and to support and reward responsible data sharing.

- We need to **fully engage** with individuals and organizations in **all continents** to be truly global.