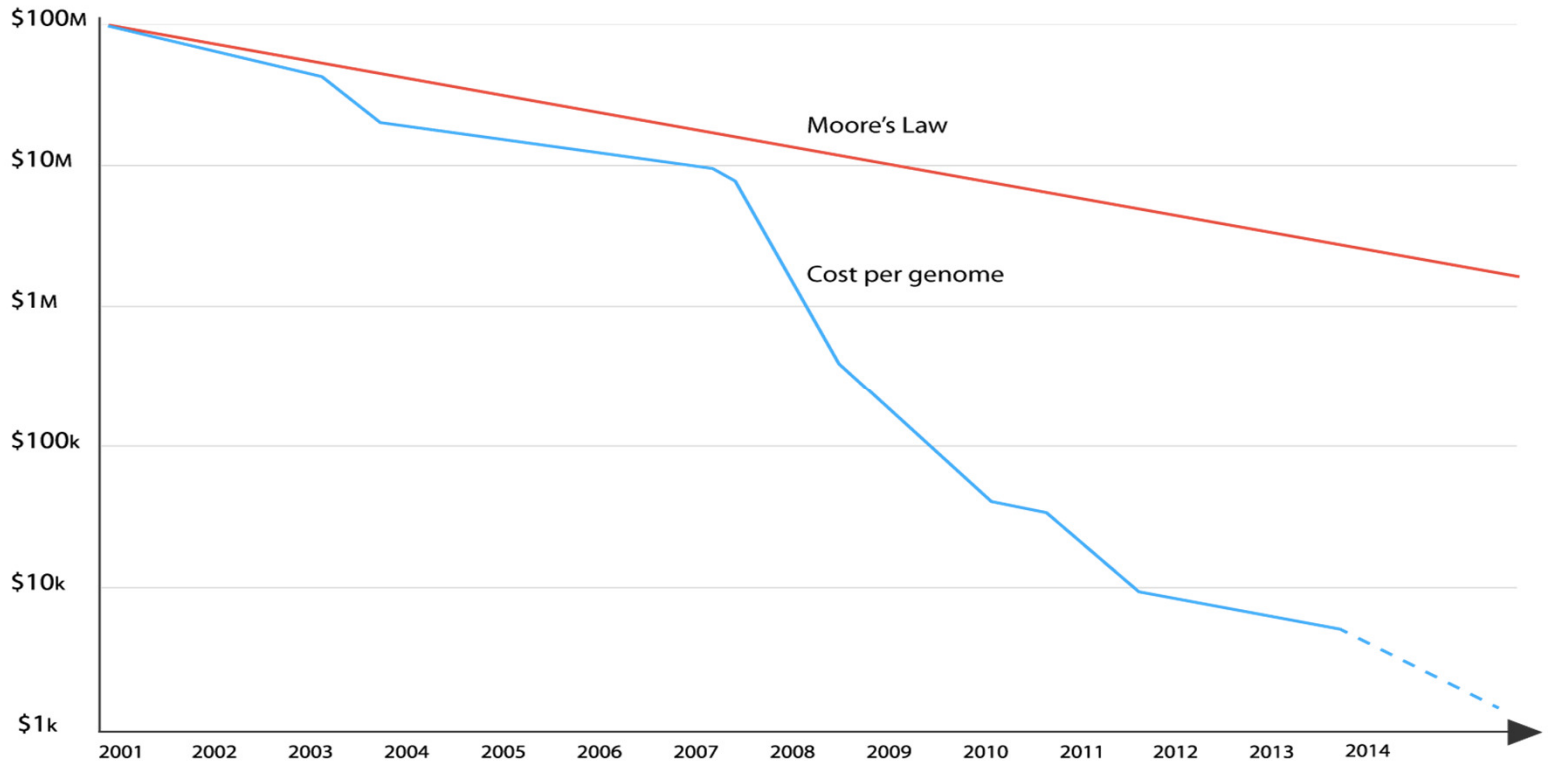
Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.

BIG genomic data

Global Alliance for Genomics & Health



Adapted from: http://www.genome.gov/sequencingcost

The opportunity

Global Alliance for Genomics & Health

Unparalleled generation of human genetic data

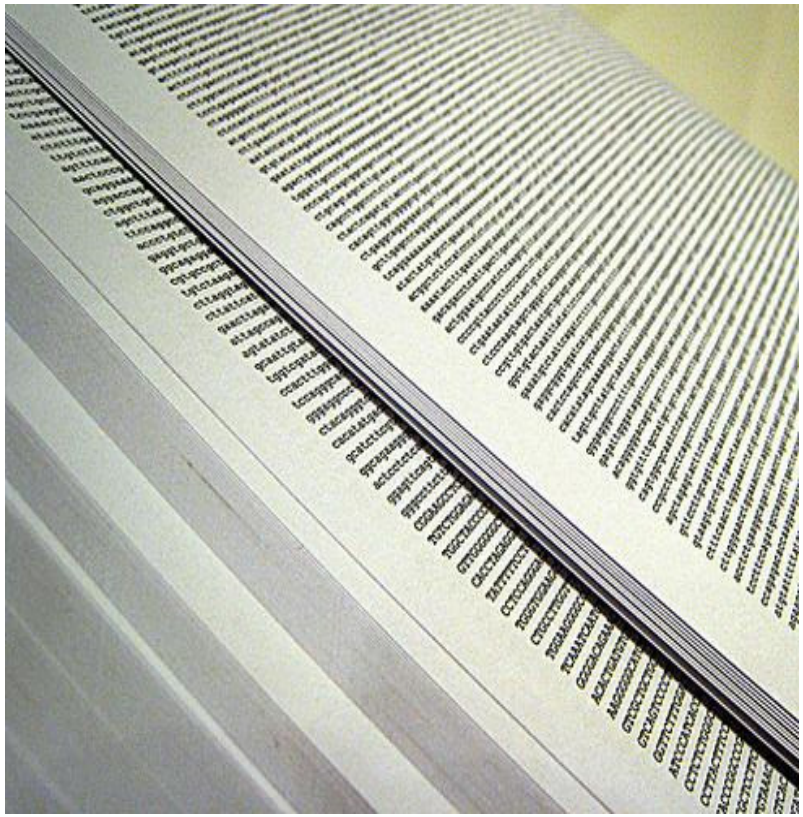
Learning from these data, we should accelerate progress in:

- Cancer outcomes and targeted therapy
- Understanding the basis of inherited disease
- Infectious disease
- Identifying targets for drug development
- Complex common diseases

The Challenge

Global Alliance for Genomics & Health

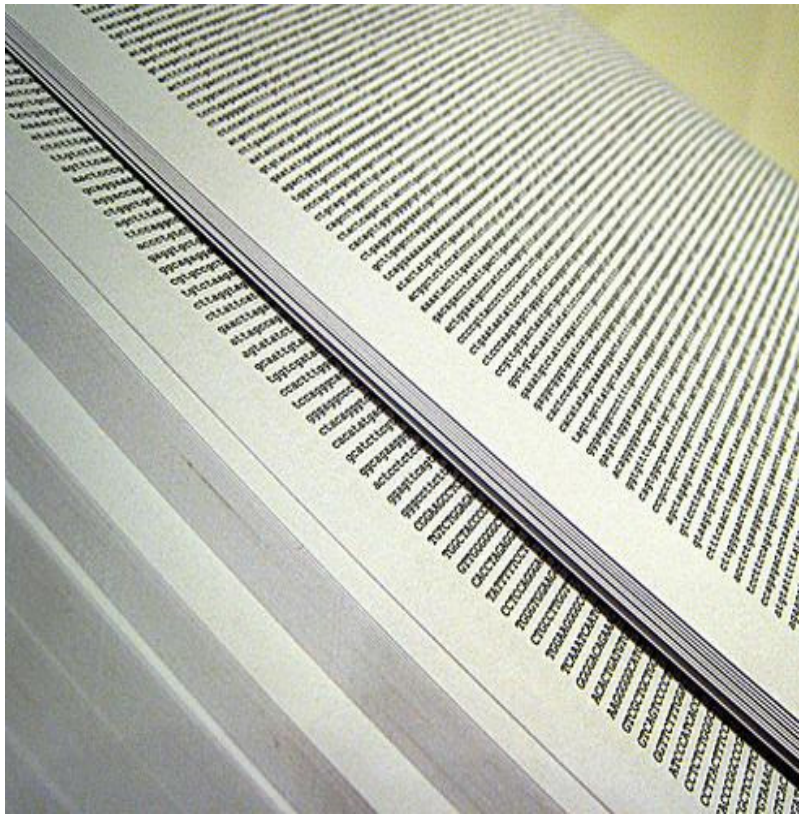
Unparalleled generation of human genetic data



The Challenge

Global Alliance for Genomics & Health

Unparalleled generation of human genetic data



How do we unlock its potential?

The Challenge

Global Alliance for Genomics & Health

In a way that allows data to be shared on a global level

The Challenge

Global Alliance for Genomics & Health

In a way that allows data to be shared on a global level

Thus empowering new knowledge, new diagnostics and new therapeutics for patients and populations

The challenge

Global Alliance for Genomics & Health

Data from **millions of samples** may be needed to achieve results and progress - showing patterns that would otherwise remain obscure.

That will take new methods and organizational models.

Right now:

- Data is typically in silos: by type, by disease, by country, by institution
- Analysis methods are non-standardized, few at scale
- Approaches to regulation, consent and data sharing limit interoperability

If we don't act: risk an overwhelming mass of fragmented data, such as electronic medical records in many countries

What can we do?

Global Alliance for Genomics & Health

Work together **internationally** to ensure **interoperability** of data and of methods, to **harmonize** approaches to ethics and regulation, and to promote **participant** autonomy

Support pilot **projects** that responsibly and effectively harmonize, analyze and share genomic and clinical data

Engage professional communities and the public; build **trust** and encourage appropriate sharing and learning

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for Genomics & Health

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What is the Global Alliance for Genomics and Health?

2013

Global Alliance for Genomics & Health

January 2013: 50 people from eight countries met in NYC to define the problem and consider solutions

June 2013: after having engaged 80 people in writing a White Paper, we announced the formation of the Alliance with 73 organizations as Partners to take on the challenge

December 2013: four Working Groups up and running; Expanded Steering Committee; Executive Staff at Host Organizations; Progress on governance, branding…

2014

Global Alliance for Genomics & Health

1st plenary meeting of Alliance partners at the Wellcome Trust on March 4th: 180 participants

Increased participation, including patient groups and leading information and life science companies, and expanded global presence

All four Working Groups advanced their topics and task teams

Engaged with 3 key projects for real time- real world implementation

2nd Plenary and 4 working groups met Oct 17-20th in San Diego : 250 + participants

Mission

Global Alliance for Genomics & Health

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

Role

Global Alliance for Genomics & Health

Convene stakeholders

Catalyze sharing of data

Create harmonized approaches

Act as a clearinghouse

Foster innovation

Commit to responsible data sharing

The Global Alliance will NOT *directly*:

Global Alliance for Genomics & Health

Generate or store data

Perform research or care for patients

Interpret genomes

Be exclusive to entities that have and share data

Long-term vision, near-term goals

Global Alliance for Genomics & Health

Long-term

- An ever-growing network
- A learning system in which data and models continuously improve
- The potential to incorporate other types of data

Near-term

- The remainder of 2014 and 2015 is about demonstrating **value**
  - Set and achieve practical working group goals
  - Organize around high priority projects
  - Define an effective sustainable model
  - Increase global engagement
  - Secure funding for the first 3 years
  - Develop a roadmap for 2015 and beyond

Organisational members

Global Alliance for Genomics & Health

Global Alliance members include:

1. Universities and research institutes
2. Academic medical centres and health systems
3. Disease advocacy organizations and patient groups
4. Consortia and professional societies
5. Funders and agencies
6. Life science and information technology companies

Last Update: November 4, 2014

Member location

Global Alliance for Genomics & Health

- Argentina
- Australia
- Austria
- Belgium
- Brazil
- Canada
- China
- Denmark
- Finland
- France
- Germany
- Hungary
- India
- Italy
- Ireland
- Japan
- Luxembourg
- Mexico
- Netherlands
- New Zealand
- Qatar
- Russia
- Singapore
- Spain
- South Africa
- Sri Lanka
- Sweden
- Switzerland
- United Kingdom
- United States

Last Update: November 4, 2014

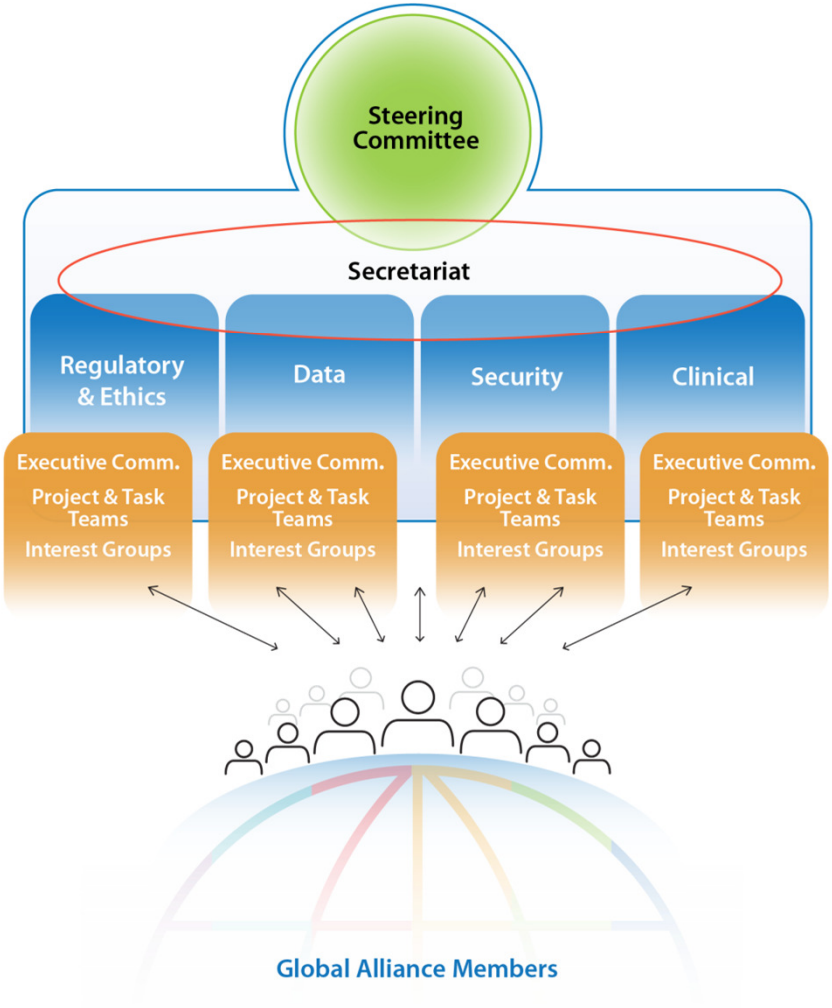
Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.

How does the Global Alliance operate?

Organizational structure

Global Alliance
for Genomics & Health



Leadership and operations

Global Alliance for Genomics & Health

- **Steering Committee**
  - Chair: David Altshuler
  - Vice-Chairs: Martin Bobrow and Kathryn North
  - Executive Director: Peter Goodhand

- **Host institutions**
  - Ontario Institute for Cancer Research
  - Wellcome Trust Sanger Institute
  - Broad Institute of MIT and Harvard

- **Secretariat role**
  - Membership
  - Communications and outreach
  - Web –site development
  - Working Group coordination
  - Project Liaison

- **Funding**
  - Working with major international funders to establish core funding
  - Outreach to other public and philanthropic funders for expanded funding

Four Working Groups

Global Alliance for Genomics & Health

Regulatory and Ethics- Bartha Knoppers, Kazuto Kato

Focuses on ethics and the legal and social implications of the Global Alliance, including harmonizing policies, developing consents, privacy procedures, and guidelines for data privacy, security, governance and transparency.

Data- David Haussler, Richard Durbin

Concentrates on data representation, storage, and analysis, including working with platform development partners and industry leaders to develop standards that will facilitate interoperability.

Security- Paul Flicek, Dixie Baker

Leads the thinking on the technology aspects of data security, user access control, and audit functions; Working to develop or adopt standards for data security, privacy protection, and user/owner access control.

Clinical- Charles Sawyers, Kathryn North

Establishing linkages to phenotypic and clinical (health) informatics data. Rather than invent such standards, this group focuses on aligning genomic data activities with the ongoing international standards initiatives in clinical and health data.

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Our Work in Progress

Progress highlights

Global Alliance for Genomics & Health

Working Group products

- Framework for Responsible Data Sharing
- Genomics API 0.5
- Security Infrastructure document
- Global activity mapping – rare diseases

Projects

- BRCA Challenge
- Matchmaker Exchange
- Beacons

Governance

- Constitution – basis of organizational and individual membership

Framework for responsible sharing of Genomic and Health Related Data

Global Alliance for Genomics & Health

- Existing frameworks founded on principle of protection from harm.
- GA4GH Framework founded on *Article 27* of the ***1948*** **Universal Declaration of Human Rights**. Guarantees the right of every individual in the world "*to share in scientific advancements and its benefits*," including to freely engage in responsible scientific inquiry, and at the same time "*to the protection of the moral and material interests resulting from any scientific production of which the person is the author*."

Genomics Data Working Group

Welcome to the Global Alliance for Genomics and Health development team site. This site provides links to source code and documentation created by and for software developers. If you are interested in general information about the Global Alliance, please visit **GenomicsAndHealth** instead

Our API

Current data sharing projects

Global Alliance for Genomics & Health

Undertaken by the members, not by GA4GH as an organization. Catalyzed and supported by GA4GH coordinators and working groups.

Drive learning, identify requirements, evaluate value, coordinate activity.

- **Matchmaker Exchange**
- **BRCA Challenge**
- **Beacon Project**

More to come!

Using Beacons to Discover Data

Repository Discovery

- ***Does the data exist?***
- **Role of Public Beacons**
- Query*: Do you have any genomes with an "A" at position 100,735 on chromosome 3?*
- Reply*: 'Yes' or 'No'*

Data context

- ***Does the data have the properties I require?***
- **Role of Registered Access Beacons**
- Shows details of studies with data and provides link for requesting access

Data Preview

- ***Does the metadata have the properties I require?***
- **Requires approved access**
- Permits users to narrow the search and access the precise sets of data needed.

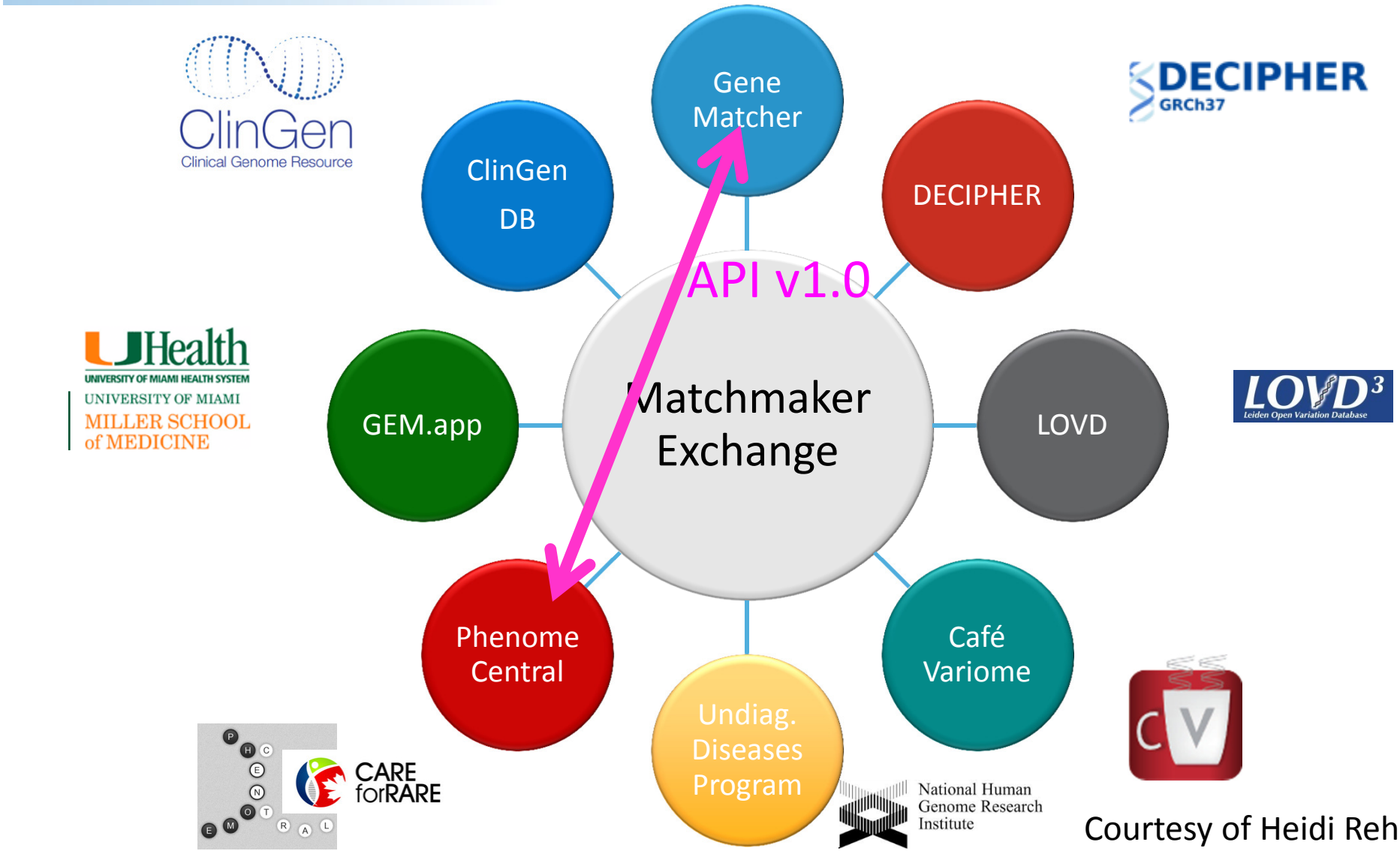
Global Alliance for Genomics and Health

Beacon of Beacons

Query Subscribe new beacon Documentation Source code Feedback

Beacon: all | Chromosome: 9 | Position: 136132908 | Alt: T | Genome: all

Beacon	Result
1000 Genomes Project	No
1000 Genomes Project - Phase 3	No
AMPLab	Yes
Beacon of Beacons	Yes
ClinVar	No
EMBL-EBI	Null
GA4GH Example Data	No
Google Genomics Public Data	No
Illumina Platinum Genomes	No
Known VARiants	Yes
Leiden Open Variation	No
NCBI	Yes
PGP	No
UniProt	No
Wellcome Trust Sanger Institute	No



Courtesy of Heidi Rehm

BRCA Challenge

Global Alliance for Genomics & Health

Idea to leverage existing and emerging BRCA data globally - originated at the March 2014 plenary meeting of the Global Alliance.

- Communicated to a large cross-section of BRCA specialists globally
- Approach being developed further by steering committee led by Stephen Chanock (NCI) and Sir John Burn (HVP)
- Short term – consolidate largest datasets through submission to existing BRCA databases (ClinVar and LOVD)
- Longer term – expand data sources, expert classification of variants, and functional studies, etc

Overlap between ClinVar, LOVD and UMD

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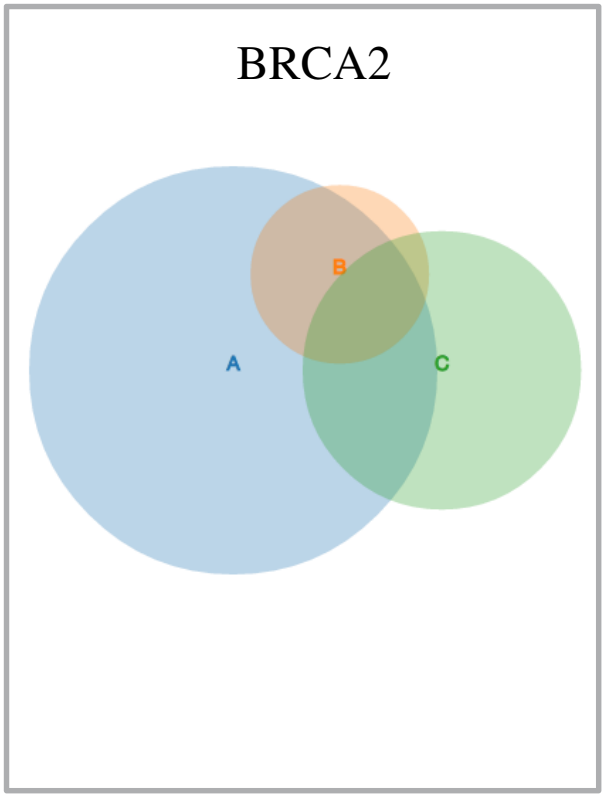


A=ClinVar B=LOVD C=UMD courtesy of Xin Feng

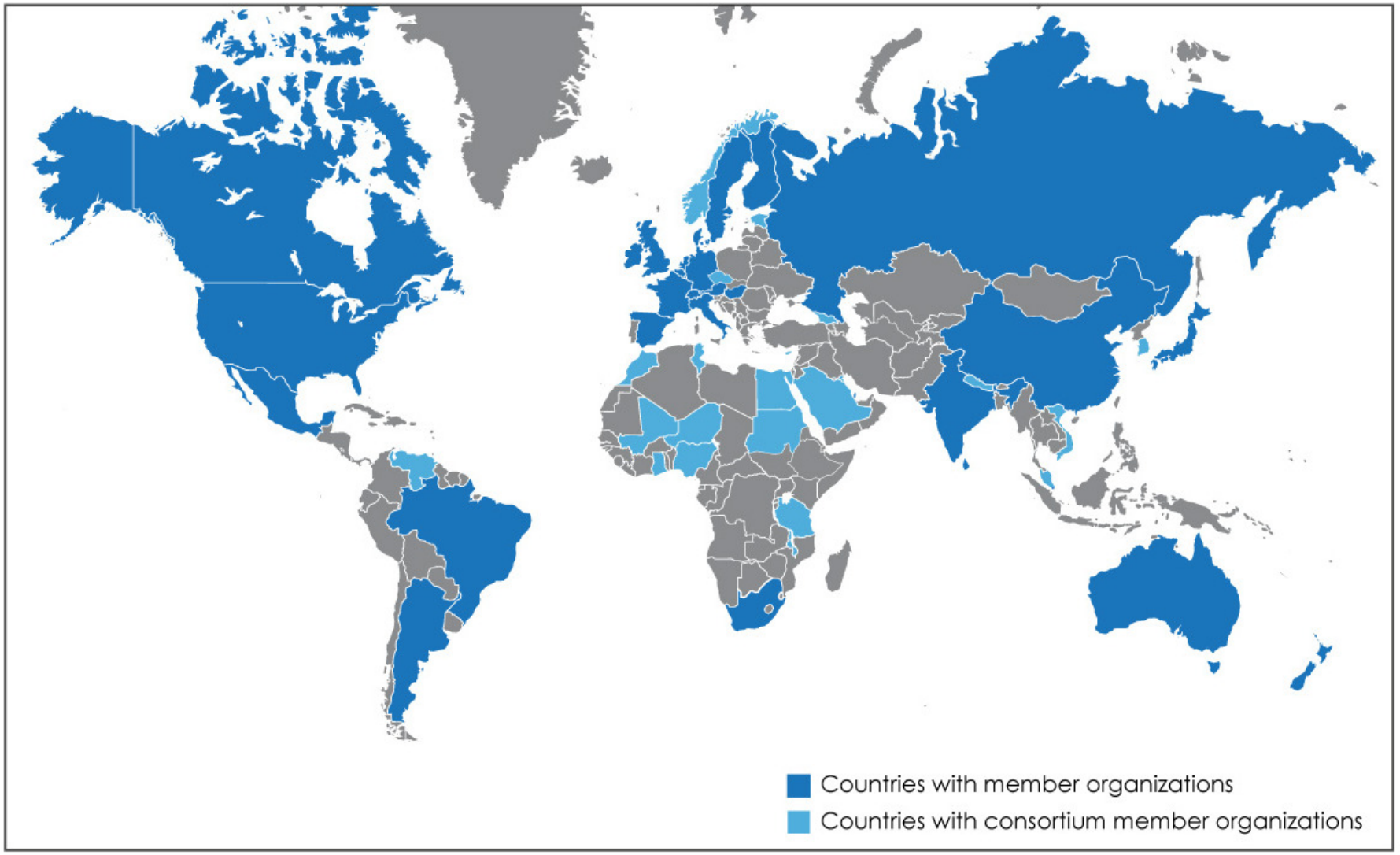
Culture and incentives

Global Alliance for Genomics & Health

- International data sharing can be achieved by federation and use of metadata while respecting national and regional restrictions
- To realize the benefits a new and widespread willingness to share data for the greater good, and to learn from data is required.
- Public attitudes towards personal data differ between countries and are changing
  - increasing awareness of the benefit of sharing,
  - increasing attention to privacy
- Collaborate on INTEROPERABILITY – Compete on Implementation

Global participation

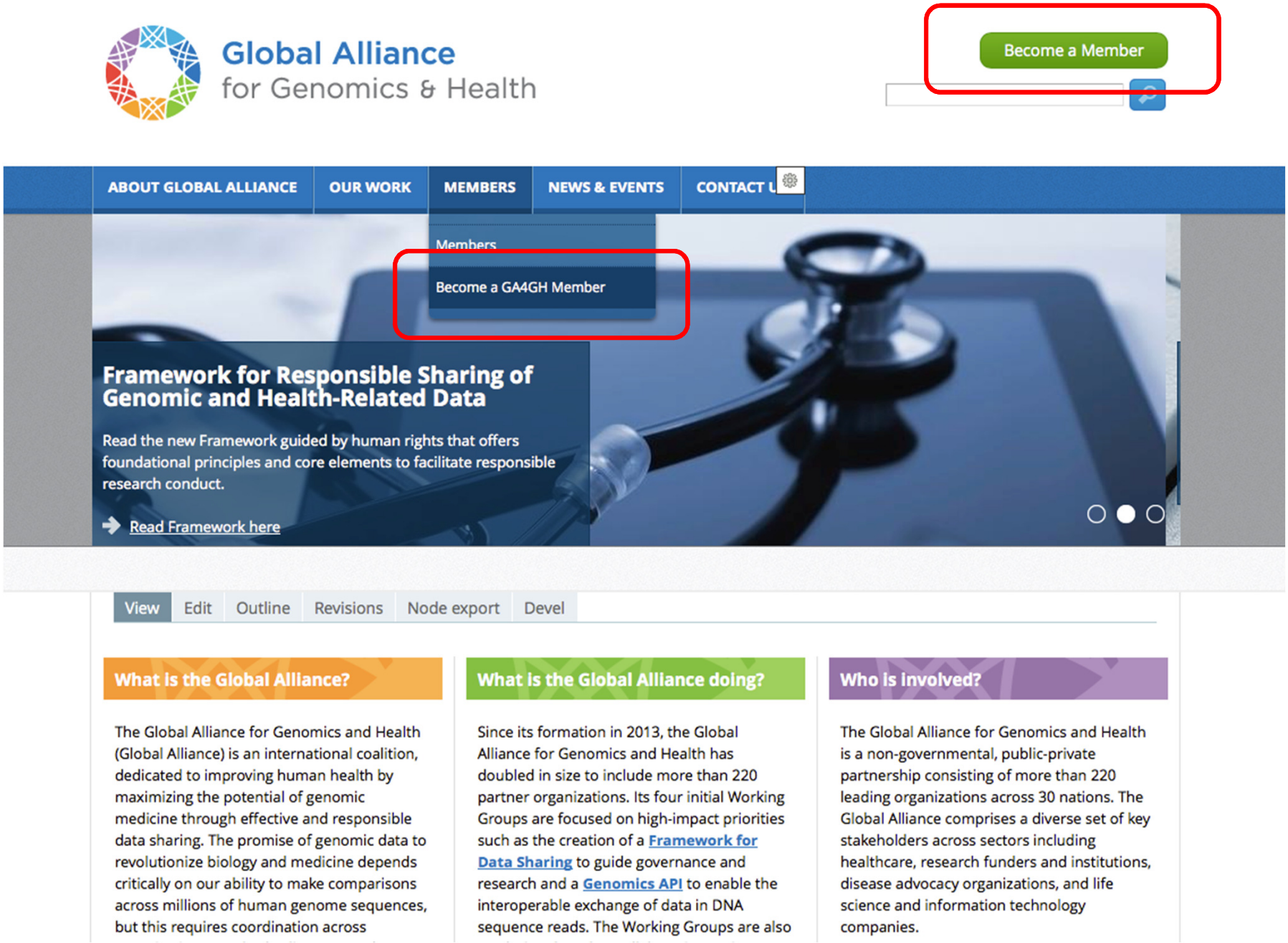
Global Alliance for Genomics & Health



GLOBAL ENGAGEMENT

Global Alliance for Genomics & Health

- We need to fully engage with individuals and organisations in all continents to be truly global
- Individuals are KEY to creating the new tools, frameworks, enablers, projects and opportunities
- Organizations are KEY to ensuring the adoption of best practices and support/reward of responsible data sharing
- We will be stronger and more effective with your participation


Global Alliance
for Genomics & Health
Become a Member
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OUR WORK
MEMBERS
NEWS & EVENTS
CONTACT U
Members
Become a GA4GH Member
Framework for Responsible Sharing of Genomic and Health-Related Data
Read the new Framework guided by human rights that offers foundational principles and core elements to facilitate responsible research conduct.
Read Framework here
View
Edit
Outline
Revisions
Node export
Devel
What is the Global Alliance?
The Global Alliance for Genomics and Health (Global Alliance) is an international coalition, dedicated to improving human health by maximizing the potential of genomic medicine through effective and responsible data sharing. The promise of genomic data to revolutionize biology and medicine depends critically on our ability to make comparisons across millions of human genome sequences, but this requires coordination across
What is the Global Alliance doing?
Since its formation in 2013, the Global Alliance for Genomics and Health has doubled in size to include more than 220 partner organizations. Its four initial Working Groups are focused on high-impact priorities such as the creation of a Framework for Data Sharing to guide governance and research and a Genomics API to enable the interoperable exchange of data in DNA sequence reads. The Working Groups are also
Who is involved?
The Global Alliance for Genomics and Health is a non-governmental, public-private partnership consisting of more than 220 leading organizations across 30 nations. The Global Alliance comprises a diverse set of key stakeholders across sectors including healthcare, research funders and institutions, disease advocacy organizations, and life science and information technology companies.

Members

- Become a GA4GH Member
  - Organizational Membership
  - Individual Membership
  - Membership FAQs

Become a GA4GH Member

The Global Alliance for Genomics and Health invites organizations and individuals to become Members of GA4GH. See the Membership FAQs.

As a member, you may join Working Groups and contribute to Initiatives. You may also register to:

- Receive the latest Global Alliance updates
- Receive the latest Working Group updates through mailings and webinars
- Download documents and presentations
- Participate in regular meetings of Global Alliance Membership

We have two types of Memberships

Organizational Membership

Submit information about your organization and review the Member Agreement

Become an Organizational Member

Individual Membership

Submit contact information, select your area(s) of interest and review the Member Agreement

Become an Individual Member

The Signup Process

Applicant submits basic information and agrees to the Member Agreement.

Global Alliance reviews the submission. Membership Criteria »

Applicant is notified that they have been approved and will create a profile to customize the membership information.

In closing..

Global Alliance for Genomics & Health

Please join our Global Alliance for Genomics and Health

THANK YOU