Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.

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

2015/2016 Road Map objectives

Global Alliance for Genomics & Health

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

Organizational Members

Global Alliance for Genomics & Health

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%)

Last Update: October 29, 2015

Countries

Global Alliance for Genomics & Health

56 Countries represented in the Global Alliance

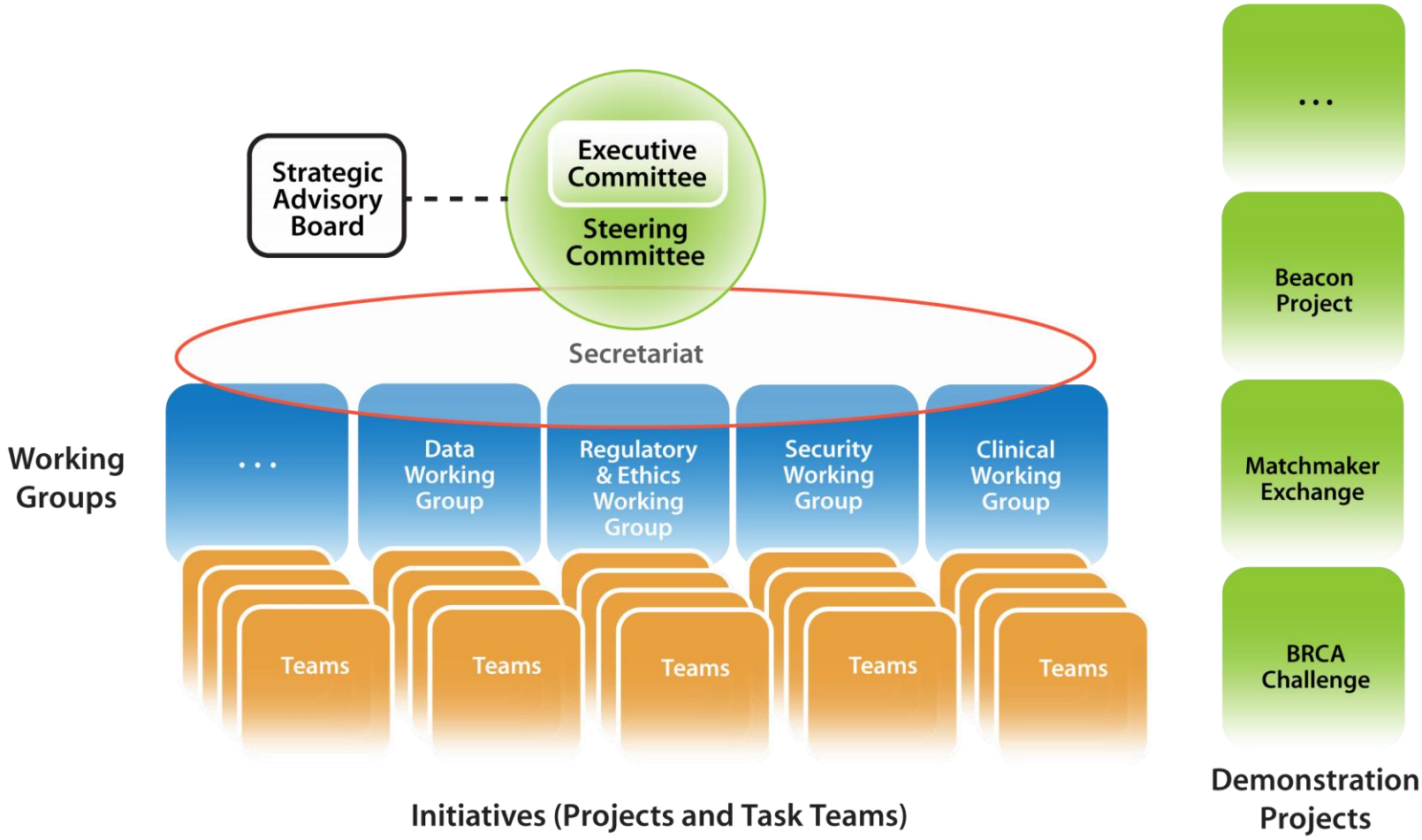
- 370 Organizational Members from 35 countries (in bold)
- 547 Individual Members from 53 countries

Afghanistan	**Georgia**	Nepal	Sudan
Argentina	**Germany**	**Netherlands**	**Sweden**
Australia	Ghana	**New Zealand**	**Switzerland**
Austria	Greece	Nicaragua	Taiwan
Belgium	**Hong Kong**	Nigeria	**Turkey**
Brazil	**India**	**Norway**	Uganda
Cameroon	**Ireland**	Peru	Ukraine
Canada	Israel	Philippines	**United Kingdom**
China	**Italy**	**Portugal**	**United States**
Colombia	**Japan**	**Qatar**	**Venezuela**
Congo	Kenya	**Russia**	Virgin Islands, U.S.
Croatia	Korea, Republic of	Sierra Leone	
Czech Republic	**Luxembourg**	**Singapore**	
Denmark	Malaysia	**Slovenia**	
Finland	**Mexico**	**South Africa**	
France	Morocco	**Spain**	

Last Update: October 29, 2015

Organizational model

Global Alliance for Genomics & Health



Last Update: September 3, 2015

Leverage

Global Alliance for Genomics & Health

7.5 professional staff
Support and enable

898 volunteer contributors

Last Update: September 18, 2015

Current Initiatives

Global Alliance for Genomics & Health

Clinical

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

Data

Beacon Network

Benchmarking

Containers and Workflows

File Formats

Genotype2Phenotype Association

Metadata

Reference Implementation

Reference Variation

RNA and Gene Expression

Variant Annotation

Completed:

- Reads

Regulatory and Ethics

Accountability (Policy)

Ageing and Dementia

BRCA Ethico-Legal and Advocacy

Data Protection Regulation

Data Sharing Lexicon

Ethics Review Equivalency

Individual Access

Machine-Readable Consent

Paediatric

Privacy Breach Notification

Registered Access (Tool)

Completed:

- *Framework for Responsible Sharing of Genomic and Health-Related Data*
- Consent Policy
- Privacy and Security Policy

Security

Cloud Security

Incident Response

Security Infrastructure

Software Security

Completed:

- Data Safe Havens

Last Update: October 29, 2015

CWG Initiatives

Global Alliance for Genomics & Health

- 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

Global Alliance for Genomics & Health

- 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

Matchmaker Exchange

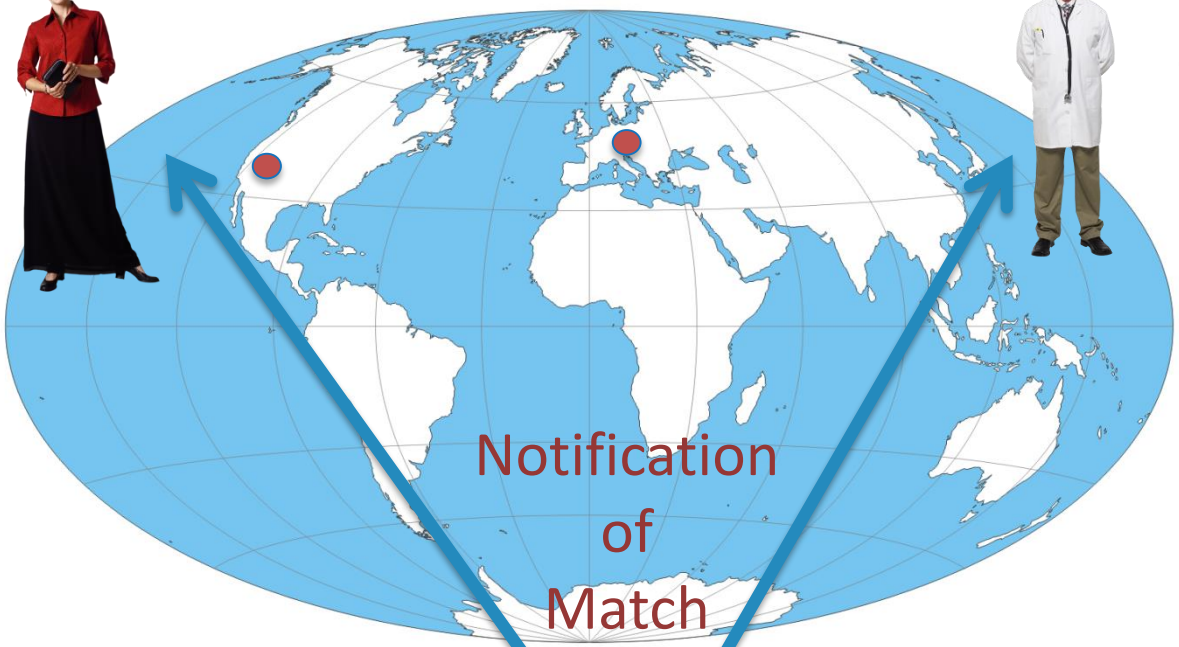
BRCA CHALLENGE

BRCA Challenge

Genomic Matchmaker

Global Alliance for Genomics & Health

Patient #1
Clinical Geneticist #1



Patient #2
Clinical Geneticist #2

Phenotypic Data	Genotypic Data
Feature 1	Gene A
Feature 2	Gene B
Feature 3	Gene C
Feature 4	**Gene D**
Feature 5	Gene E
	Gene F

Genomic Matchmaker

Genotypic Data	Phenotypic Data
Gene D	**Feature 1**
Gene G	**Feature 3**
Gene H	**Feature 4**
	Feature 5
	Feature 6

Courtesy of Joel Krier

Database connections through common API

Last Update: June 4th 2015

www.matchmakerexchange.org

GA4GH and BRCA Challenge

Global Alliance for Genomics & Health

- 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

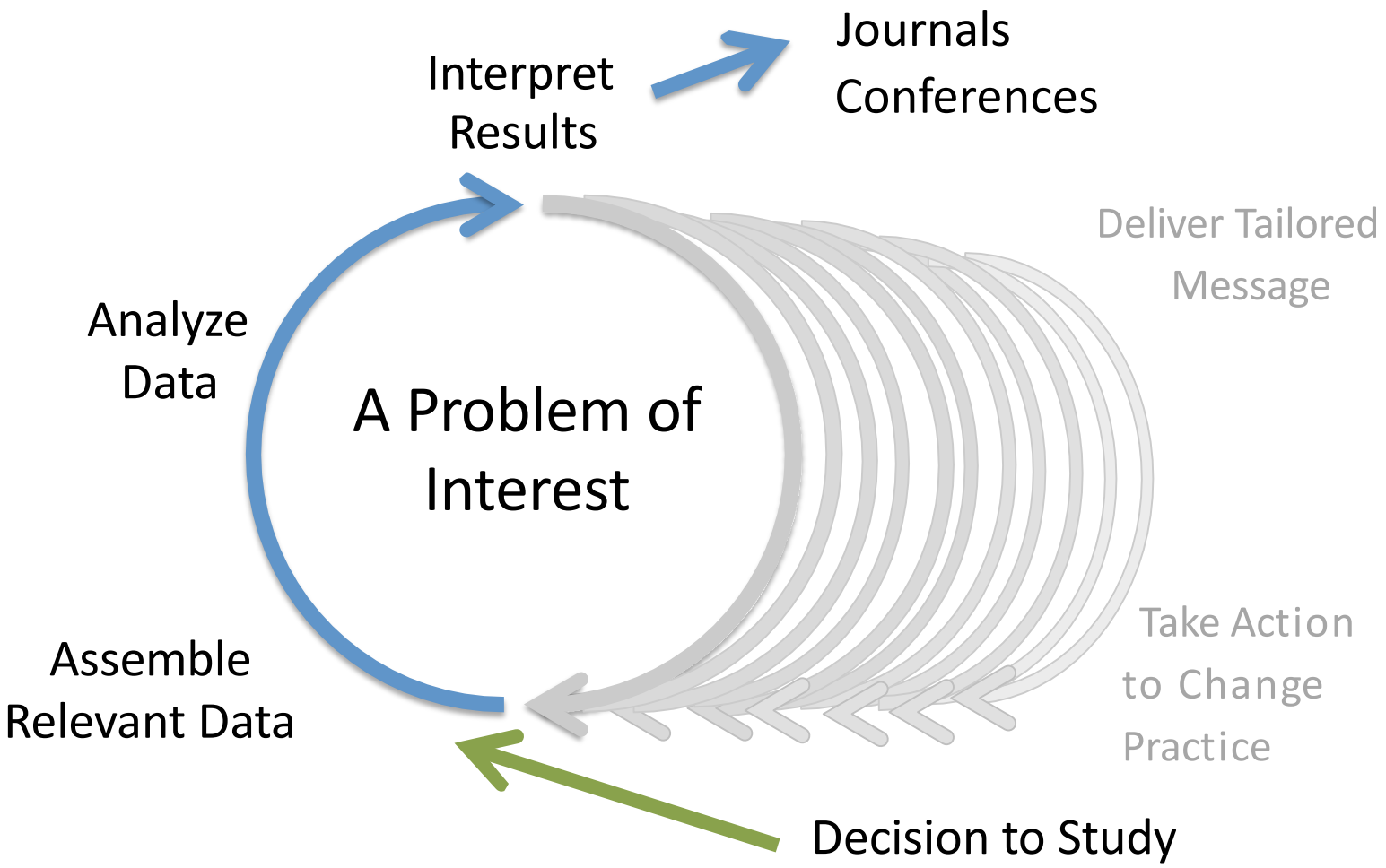
To improve the care of patients at risk of breast and ovarian cancer using global data sharing and collaboration in the analysis of *BRCA1* and *BRCA2*

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



GA4GH Outreach

Global Alliance for Genomics & Health

- 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

Global Alliance for Genomics & Health

- **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