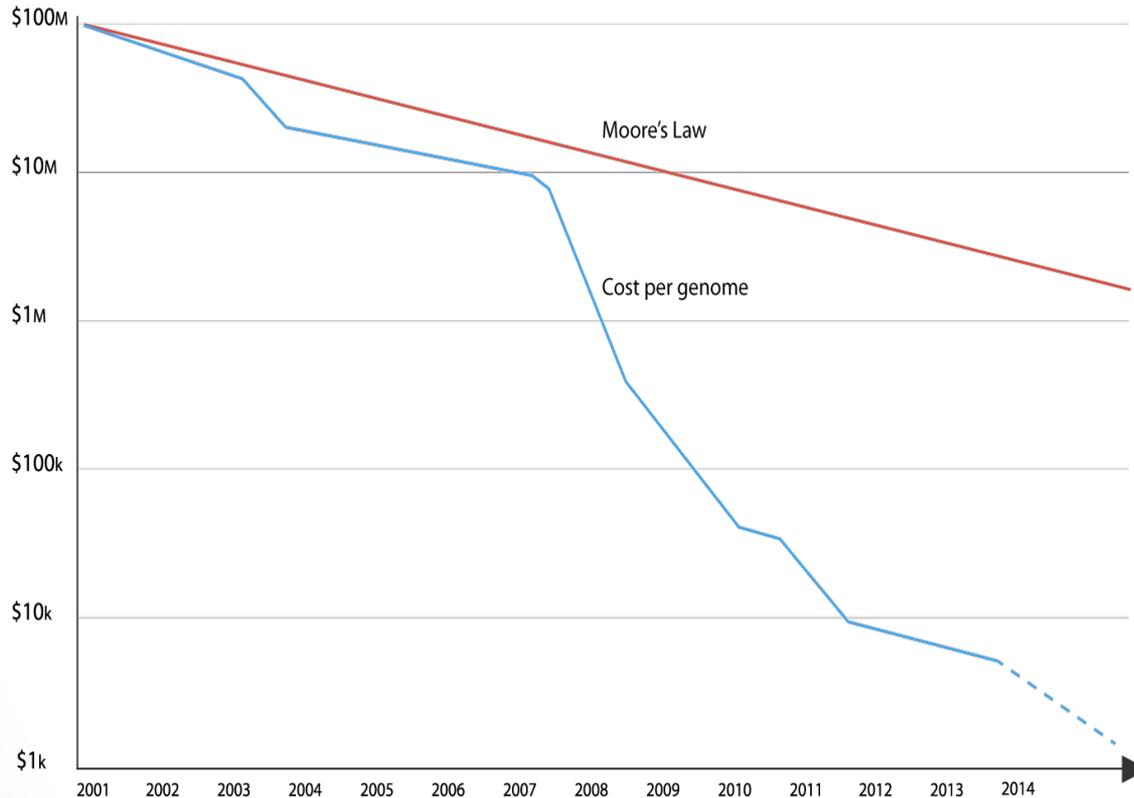




Global Alliance
for Genomics & Health

The Cause of "big genomic data"



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

The opportunity

An explosion of genomic information from individuals with known clinical characteristics and disease outcomes

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

Enable clinical interpretation of individual genome sequences

The challenge

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 institution
- Analysis methods are non-standardized, few at scale
- Approaches to regulation, consent and data sharing limit interoperability

If we don't act: risk a hodge-podge of Balkanized data, such as electronic medical records in the USA

What can we do?

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

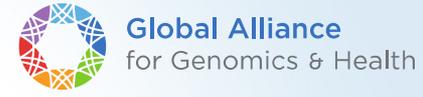
Engage professional communities and the public; build trust and encourage appropriate sharing and learning



Global Alliance
for Genomics & Health

What is the Global Alliance for
Genomics and Health?

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

Theory of change

After discussion and feedback by partners, it was concluded that the Global Alliance has the best chance to succeed and achieve our mission if we:

- Gather a broad and diverse network of stakeholders
- Publicly commit to advancing progress in data sharing
- Establish a common frameworks of approaches to this goal
- Catalyze interactions and shared activity among the members: in particular data sharing pilot projects that drive the work of and are supported by the Alliance

Roles in the biomedical ecosystem

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*~~:

Generate or store data

Perform research or care for patients

Interpret genomes

Be exclusive to entities that have and share data

A fast paced non-profit
start-up...

Starting in 2013

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...

...gaining momentum in 2014



First face-to-face meeting of Alliance partners at the Wellcome Trust on March 4th: included 180 participants from 100 partner organizations and 17 countries

Increased membership to over 190 partner organisations in 28 countries, including leading information and life science companies

All four Working Groups are advancing their topics, new Working Groups are in formation, task and project teams launched



Global Alliance partners 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

Partner overview



28
Countries

- Argentina
- Australia
- Austria
- Belgium
- Brazil
- Canada
- China
- Denmark
- Finland
- France
- Germany
- Hungary
- India
- Italy
- Ireland

Long-term vision, near-term goals

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 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



Global Alliance
for Genomics & Health

How does the Alliance operate?

Core principles



Respect for the data sharing and privacy preferences of participants

Transparency of governance and operations

Accountability to best practices in technology, ethics, and public outreach

Inclusivity by partnering and building

Lean, distributed operation



Staff at multiple Host Institutions – not an incorporated entity

- Ontario Institute for Cancer Research
- Wellcome Trust Sanger Institute
- Broad Institute / Brigham and Women's Hospital

Transitional Steering Committee, 12 Members

Four Initial Working Groups to advance work on specific topics

- Regulatory and Ethics
- Data
- Security
- Clinical

Global Alliance for Genomics and Health Leadership



David Altshuler
Broad Institute, Massachusetts
General Hospital
Chair



Martin Bobrow
University of Cambridge
Vice-Chair



Kathryn North
Murdoch Childrens
Research Institute
Vice-Chair



Peter Goodhand
Global Alliance for Genomics
and Health
Member and Executive Director



Paul Flicek
European Bioinformatics Institute
Member and Co-chair,
Security Working Group



David Haussler
University of California, Santa Cruz
Member and Co-chair, Data
Working Group



Thomas J. Hudson
Ontario Institute for Cancer
Research
Member



Kazuto Kato
Osaka University
Member and Co-chair, Regulatory
and Ethics Working Group



Bartha Maria Knoppers
McGill University
Member and Chair, Regulatory
and Ethics Working Group



Brad Margus
Genome Bridge
Member



Elizabeth Nabel
Brigham and Women's Hospital
Member



Charles Sawyers
Memorial Sloan Kettering Cancer Center
Member and Co-chair, Clinical Working
Group



Partha P Majumder
National Institute of Biomedical
Genomics
Co-chair, Regulatory and Ethics
Working Group



Dixie B. Baker
Consultant to Genetic Alliance
Co-chair, Security Working Group



Richard Durbin
Wellcome Trust Sanger Institute
Co-chair, Data Working Group



Regulatory and Ethics- Bartha Knoppers, Kazuto Kato, Partha Majumder

Focused 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

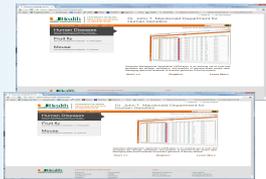
Concentrates on data representation, storage, and analysis, including working with

Our work model

- Initial **Working Groups** with active membership
- Developed initial work plan and priorities
- Now evolving into steering committees supported by professional staff coordinators
- Creating **Task Teams** and **Project Teams** to develop work products to address priority needs
- Distribute initial drafts for comment to **Interest Groups** from partner organizations
- Take comments on-board, iterate and distribute for broad comment
- Final work product for reference implementation and adoption by partner organizations

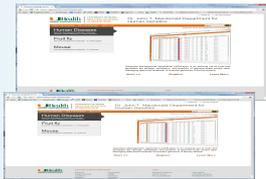


Multiple disconnected solutions





API v1.0
Multiple
disconnected
solutions



Driver Project for Global Alliance Global Alliance for Genomics & Health

- Success highly dependent on international effort
- Critical need for standards
- Activity spans multiple workgroups
 1. Clinical (phenotyping and matching algorithms)
 2. Data (data format and interfaces)
 3. Security (patient privacy)
 4. Regulatory and Ethics (patient consent)

Genomic Data and EHR's

Currently being explored by a sub-group of the Alliance CWG – led by Andrew Morris (Univ of Dundee and the Farr Institute)

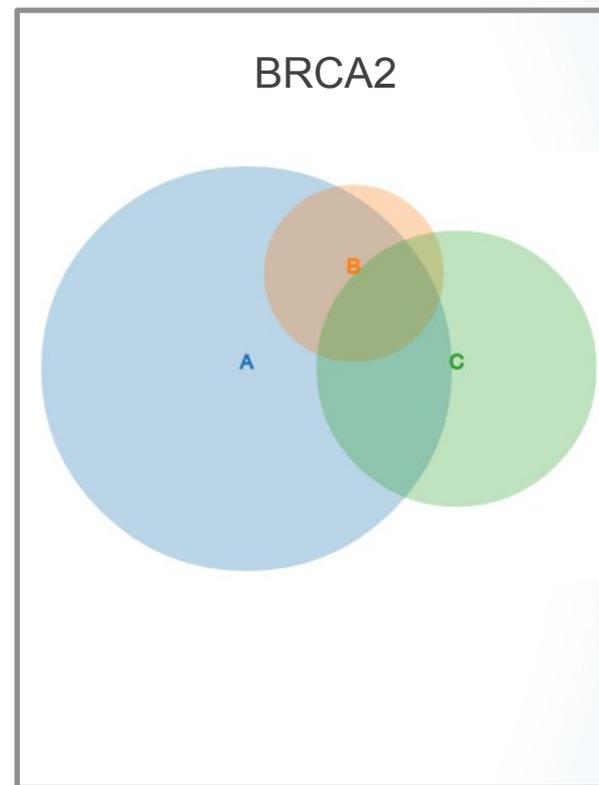
Mapping existing/emerging initiatives

- Global Genomic Medicine Collaboration (G2MC)
- Institute of Medicine (Ginsburg)
- Clinical Genomics group of HL7
- American College of Medical Genetics
- EMERGE
- Stanford/UCSD/New York
- Genomics England

BRCA Challenge

- Idea originated at the Global Alliance March 4th at the Wellcome Trust
- Written up by Stephen Chanock of NCI and John Burn from University of Newcastle
- Communicated to a large cross-section of BRCA specialists globally
- Advanced during a Breast Cancer Interest Group meeting at HVP meeting in April
- Approach being developed further by small leadership group
- Initial thrust - link existing BRCA data bases
- Longer term - strengthen quality and facilitate analysis

Overlap between LOVD, UMD and ClinVar



A=ClinVar B=LOVD C=UMD

A few other initiatives

- Beacons (EBI, NCBI, Berkeley, Sanger)
- Genomics APIs (read and variant)
- Regulatory and Ethics Framework
- Data Safe Havens
- Common Elements of International Consents
- Security Framework
- Phenotype Ontologies

- ICGC Pan-Cancer Analysis

Website

http://genomicsandhealth.org



Global Alliance
for Genomics & Health

The screenshot shows a web browser window displaying the homepage of the Global Alliance for Genomics & Health. The browser's address bar shows the URL genomicsandhealth.org. The website header includes a navigation menu with links for [Home](#), [Global Alliance for Genomics and Health](#), and [Log In](#). The main content area features the organization's logo and a "Sign up for updates" button. Below this is a blue navigation bar with links for [ABOUT GLOBAL ALLIANCE](#), [OUR WORK](#), [PARTNERS](#), [NEWS & EVENTS](#), and [CONTACT US](#). A large banner image shows a woman presenting to a group of people in a meeting room. A dark blue overlay on the banner contains the text: **GA4GH Holds First Partner Meeting.** Below this, it says "View presentations from our first partner meeting, held at the Wellcome Trust on March 4th, 2014." and includes a link: [Learn More About the Meeting](#). The footer consists of three columns with the following headings: **What Is the Global Alliance?**, **What Is the Global Alliance doing?**, and **Who Is Involved?**. Each column contains a short paragraph of text.

Home | Global Alliance for Genomics and Health

genomicsandhealth.org

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[ABOUT GLOBAL ALLIANCE](#) [OUR WORK](#) [PARTNERS](#) [NEWS & EVENTS](#) [CONTACT US](#)

GA4GH Holds First Partner Meeting.

View presentations from our first partner meeting, held at the Wellcome Trust on March 4th, 2014.

[Learn More About the Meeting](#)

What Is the Global Alliance?

The Global Alliance for Genomics and Health (Global Alliance) is an international coalition, formed to enable the sharing of genomic and clinical data. Genomic information offers great potential to inform the understanding

What Is the Global Alliance doing?

The Global Alliance for Genomics and Health has doubled in size since its formation and the four initial Working Groups are focused on high-impact priorities for 2014. The Alliance held its first plenary meeting at the

Who Is Involved?

The Global Alliance for Genomics and Health (Global Alliance) is a broad and inclusive organization that includes over 170 of the world's leading organizations including healthcare providers, research funders,

Need More Information?

Visit our website for relevant materials and updates:

List of Partner Organizations

Working Group Priorities Document

Working Group Work Products

Sign Up for Updates



Global Alliance
for Genomics & Health