Global Alliance
for Genomics & Health
Collaborate. Innovate. Accelerate.

Peter Goodhand, Executive Director GA4GH
President Ontario Institute for Cancer Research
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.

Historically:
- 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

**Don’t act:** an overwhelming mass of fragmented data, such as electronic medical records in many countries

**Collective Action:** achieve the interoperability of the www or global telecommunications – Smart phones.

**LAUNCH A GLOBAL ALLIANCE – 2013**
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.
Organizational Milestones

- **Jan.28**: NYC Meeting
- **Jun.5**: White Paper Release
- **Sep. 18**: GA4GH Formal Launch
- **Jan. 29**: SAB Established
- **Jun. 9**: Perspective Paper Published

### 2013
- **Mar. 4**: 1st Plenary
- **Jun. 5**: White Paper Release
- **Oct. 18**: 2nd Plenary

### 2014
- **Dec. 12**: 200 Org Members
- **Jun. 10**: 3rd Plenary

### 2015
- **Jun. 3**: 300 Org Members

### 2016
- **Apr. 13**: 400 Org Members
- **Oct. 18**: 4th Plenary
Membership

Global Alliance members include
1. Life science and information technology companies (43%)
2. Universities and research institutes (32%)
3. Academic medical centers and health systems (10%)
4. Disease advocacy organizations and patient groups (5%)
5. Consortia and professional societies (5%)
6. Funders and agencies (5%)

470+ Organizational Members

1000+ Individual Members

75 Countries
Next Steps

• 5-year Strategic Roadmap
  • GA4GH vision to enable genomic data sharing
  • Cross-cutting ‘product lines’ to implement goals
  • Development of GA4GH Toolkit

• National Initiatives
  • Engaging national precision medicine and genomics initiatives
  • Preliminary meeting: 8 countries (Oct, 2016)
  • Next meeting (co-hosted by Genomics England and AGHA): 20+ countries (May, 2017)

• 5th Plenary
  • Open meetings to engage community, advance projects
  • Oct 15-17, 2017 (Orlando, Florida)
Coordination with G2MC

- G2MC: *implementation* of genomic medicine into clinical care

- GA4GH: enabling international genomic and health data sharing within and across the research-care continuum

- GA4GH-G2MC collaboration on health informatics

- G2MC secretariat co-localized with GA4GH at OICR
## Research & Clinic - Historically Two Solitudes

<table>
<thead>
<tr>
<th>Genomic Research</th>
<th>Genetic Health Care</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Subjects, papers, global</td>
<td>• Patients, providers, local (national or state/system)</td>
</tr>
<tr>
<td>• Rapid change of scale and scope of NGS due to decreased costs and increased compute power</td>
<td>• Change from single gene to panels</td>
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<tr>
<td>• Data science primarily academic developed/used</td>
<td>• Data science developed and delivered by HIT providers</td>
</tr>
<tr>
<td>• BAM/SAM/CRAM, VCF, Aligners, variant callers, pipelines</td>
<td>• EHR, EMR, HL7, FHIR, SMART</td>
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</tbody>
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Global Research, Local Healthcare

Academic Activities:
- Journals
- Conferences

Jurisdictional Challenges:
- Health expenditures
- Legal systems
- Languages
- Infrastructure

Human Genome Project (1990)
International HapMap Project (2002)
International Cancer Genome Consortium (2008)
1000 Genomes Project (2008)

Research

Healthcare

Global Research, Local Healthcare

1000 Genomes Project (2008)
International Cancer Genome Consortium (2008)
International HapMap Project (2002)
Human Genome Project (1990)

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Research

Healthcare
“Global Knowledge, Local Care”

Interoperable Data Sharing:
• APIs
• Standards
• Frameworks

Research

Healthcare

Genomic Knowledge Exchanges
Healthcare System

- Industry Research
- Clinical Trials
- Health Services Research
- Cohorts
- Academic Research

Global Alliance for Genomics & Health
Tremendous opportunity in digitized data

- Power in numbers
- Dynamic
- Interoperable
- Linkage of clinical experience & research data over time

CD’s vs digital music streaming!
Variant Modelling Collaboration

- Gil Alterovitz, Harvard Medical School, FHIR Genomics
- Larry Babb, Sunquest, ClinGen
- Karen Eilbeck, University of Utah
- Gaston Fiore, Boston Children’s Hospital, FHIR Genomics
- Bob Freimuth, Mayo Clinic, ClinGen, HL7/FHIR
- Reece Hart, Invitae, GA4GH
- David Kreda, Harvard Medical School, FHIR Genomics
- Jennifer Lee, NCBI, ClinVar
- Peter Robinson, Jackson Labs

Courtesy of Reece Hart
Variant Interpretation for Cancer Consortium (VICC)

Building on the work of Genotype-to-Phenotype (G2P) Task Team led by Adam Margolin

VICC Co-chairs:
- Obi Griffith
- Nuria Lopez-Bigas
- David Tamborero
- Malachi Griffith

Goals/Principles:
- Clinical cancer variant interpretation
- Standards and guidelines
- Open content
- Interoperability

Variation Interpretation for Cancer
- Gene
- Variant
- Cancer subtype
- Clinical implication: drug sensitivity, drug resistance, adverse response, diagnostic, or prognostic
- Source (e.g., PubMed identifier)
- Curation group

http://ga4gh.org/#/vic

ga4gh-dwg-vic@genomicsandhealth.org
GA4GH & eHealth

• eHealth task team (Chair: John Mattison)
• Objectives:
  • Support the discovery, access, and sharing of genomic and clinical information within and among EHR systems
  • Survey current activities in eHealth and promote best practices
  • Harmonize efforts where possible
• Deliverables to date:
  • Catalogue of 85 global resources for sharing clinical and genomic eHealth data
  • Catalogue of 23 family health history tools
  • Family history collection tools: statement of best practice
• Possible focus areas for 2017-2018:
  • Resources to support users in assessing/evaluating products for clinical care
  • Collaborations HL7-FHIR, SMART on FHIR, Sync for Genes
Coordinating with external e-health initiatives (Lead: Grant Wood)

- HL7 Clinical Genomics workgroup (FHIR)
- SNOMED
- DIGITizE, IGNITE, EMERGE
- ClinGen, ClinVar
- Farr Institute, Swiss Institute of Bioinformatics
- Healthcare Services Platform Consortium
- Genetic Alliance (S) and other patient groups
- National Precision Medicine Initiatives
- HUGO, HVP/GV, HGVS
Aligning Data in Research and Health

- Variant Modelling
- Variant Interpretation, Clinical Actionability
- Phenotype Ontologies
- Genotype, Phenotype, “Envirototype”
- Pathogenicity Classifications – 5 vs 2
- Rare/Undiagnosed disease: discovery and matching underway (MME)– P&F facebook, facial recognition
- Cancer: pre-disposition in germline; hetrogeneity, clonal evolution, primary, mets, recurrence in somatic
Genomic data in Healthcare Systems

• In the point of care EMR
• In the integrated/federated EHR
• Data Repositories for Genomic Data - GACS
• Direct and Immediate CDS
• Warnings, Alerts, Updates – New Knowledge
• System responsibilities for security and privacy of genomic data
• Patient/family role in use of their data for research
Many Questions

Research Perspective
How can we gain access to well annotated clinically generated genomes?
How can we interrogate and learn from longitudinal data in the health record - with genomic data?
How can we connect with patients (families) to engage them directly in the research process?
How can we leverage the investment of industry in Clinical Trials?

Clinical Perspective
How can we share clinically generated data in a way that will benefit the care we deliver?
How will complex and at times evolving genomic knowledge be made accessible to my practice?
What should I advise my patients regarding participation in research, incidental findings, return of results?
How should we regard traditional RCTs vs RWD?
What are the issues in your country (state/province/system/region) to accomplish the goals of a longitudinal genomic patient records?