# Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.

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

# 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.

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**

# 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

# Organizational Milestones

2013

Jan.28
NYC Meeting

Jun.5
White Paper Release

Mar. 4
1st Plenary

2014

Sep. 18
GA4GH Formal Launch

Oct.18
2nd Plenary

Dec. 12
200 Org Members

2015

Jan. 29
SAB Established

Jun. 3
300 Org Members

Jun.10
3rd Plenary

2016

Apr. 13
400 Org Members

Jun. 9
Perspective Paper Published

Oct.18
4th Plenary

# Membership

**470+**
Organizational Members

**1000+**
Individual Members

**75**
Countries

**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%)

*Last Update: January 2017*

# 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
Ontario Institute
for Cancer Research

# Research & Clinic - Historically Two Solitudes

Global Alliance
for Genomics & Health

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

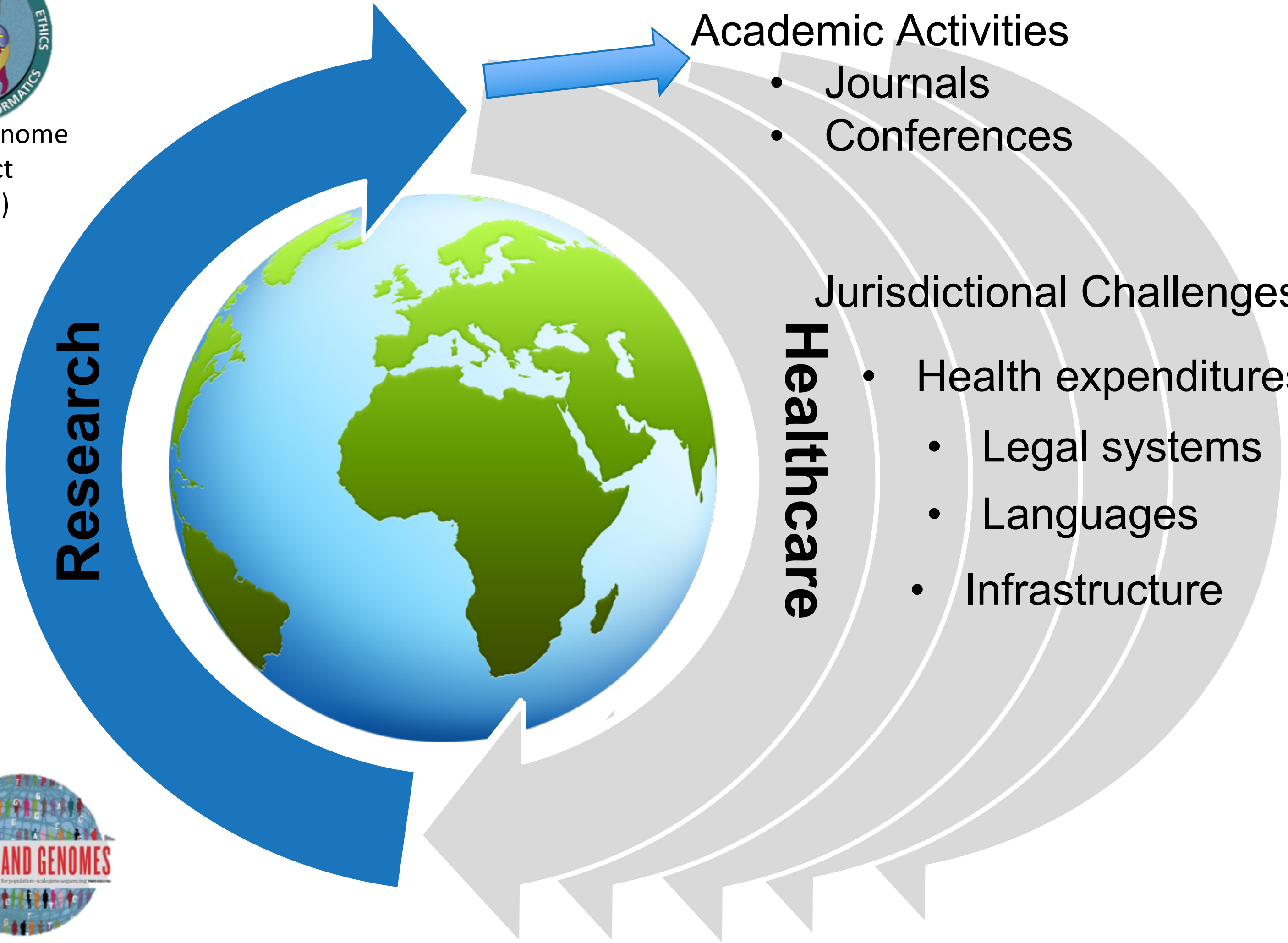
# Global Research, Local Healthcare

Human Genome
Project
(1990)

International HapMap
Project
(2002)

International Cancer Genome
Consortium
(2008)

1000 Genomes Project
(2008)



Research

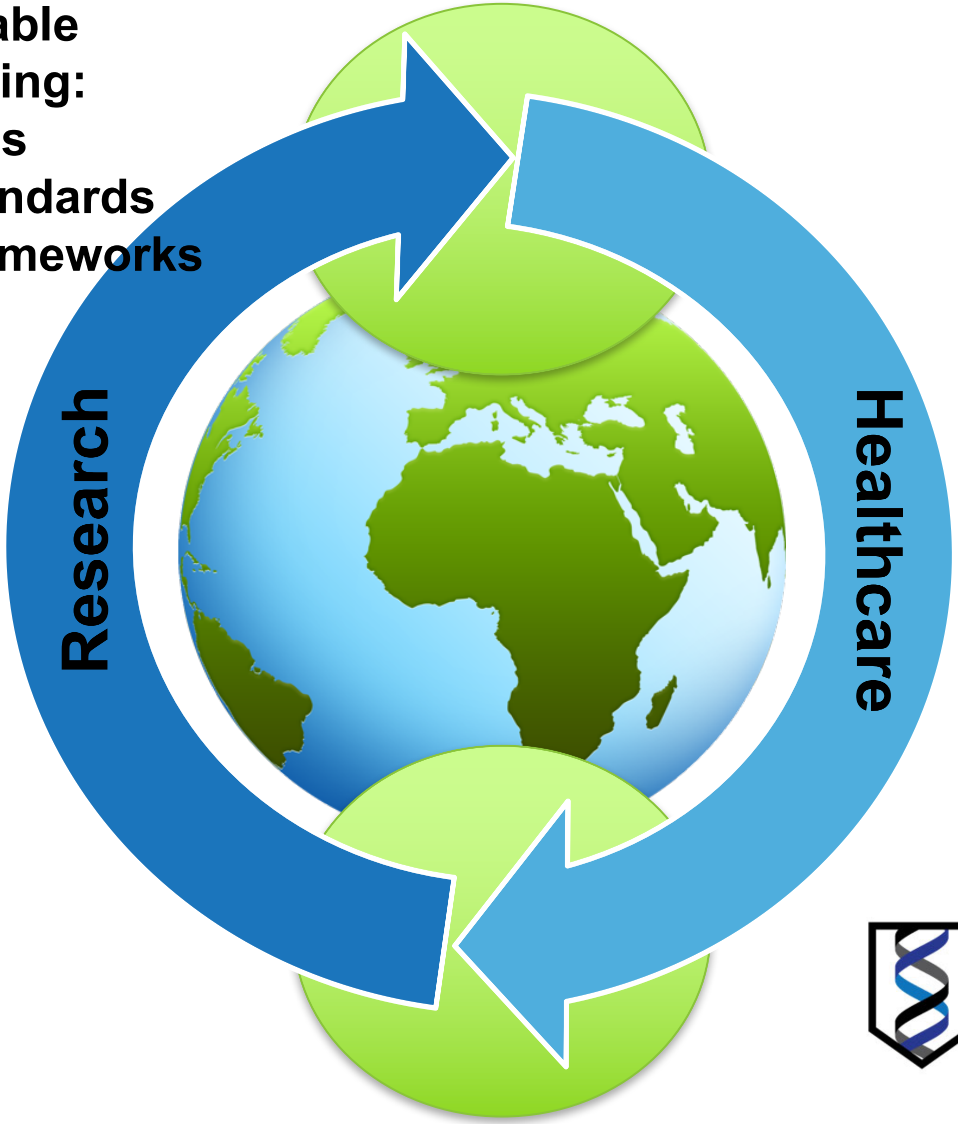
Healthcare

Academic Activities
- Journals
- Conferences

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

# “Global Knowledge, Local Care”

**Interoperable Data Sharing:**
- APIs
- Standards
- Frameworks



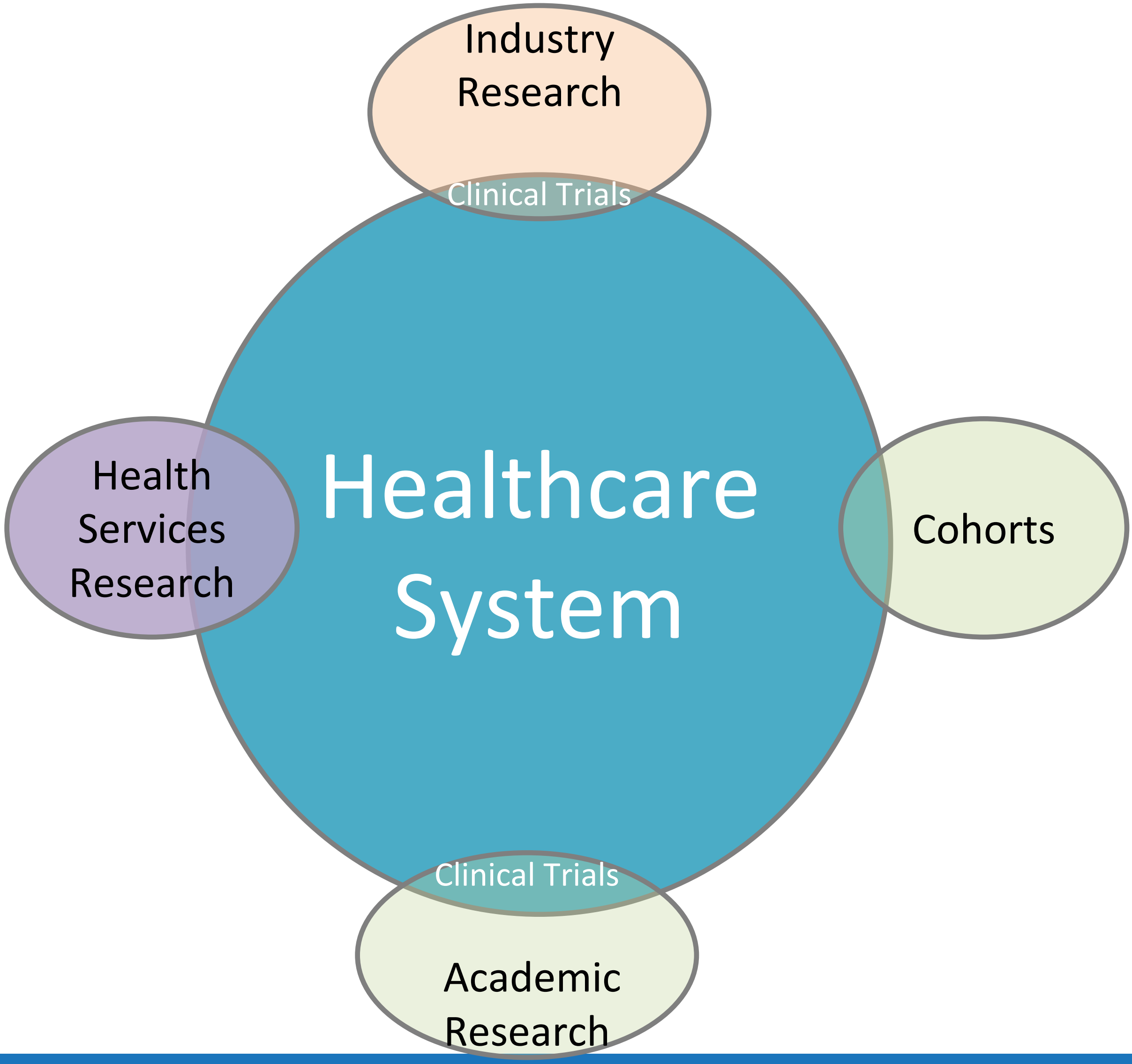
Beacon

BRCA CHALLENGE

**Genomic Knowledge Exchanges**

Matchmaker Exchange

Cancer Gene Trust



Industry Research

Clinical Trials

Health Services Research

Healthcare System

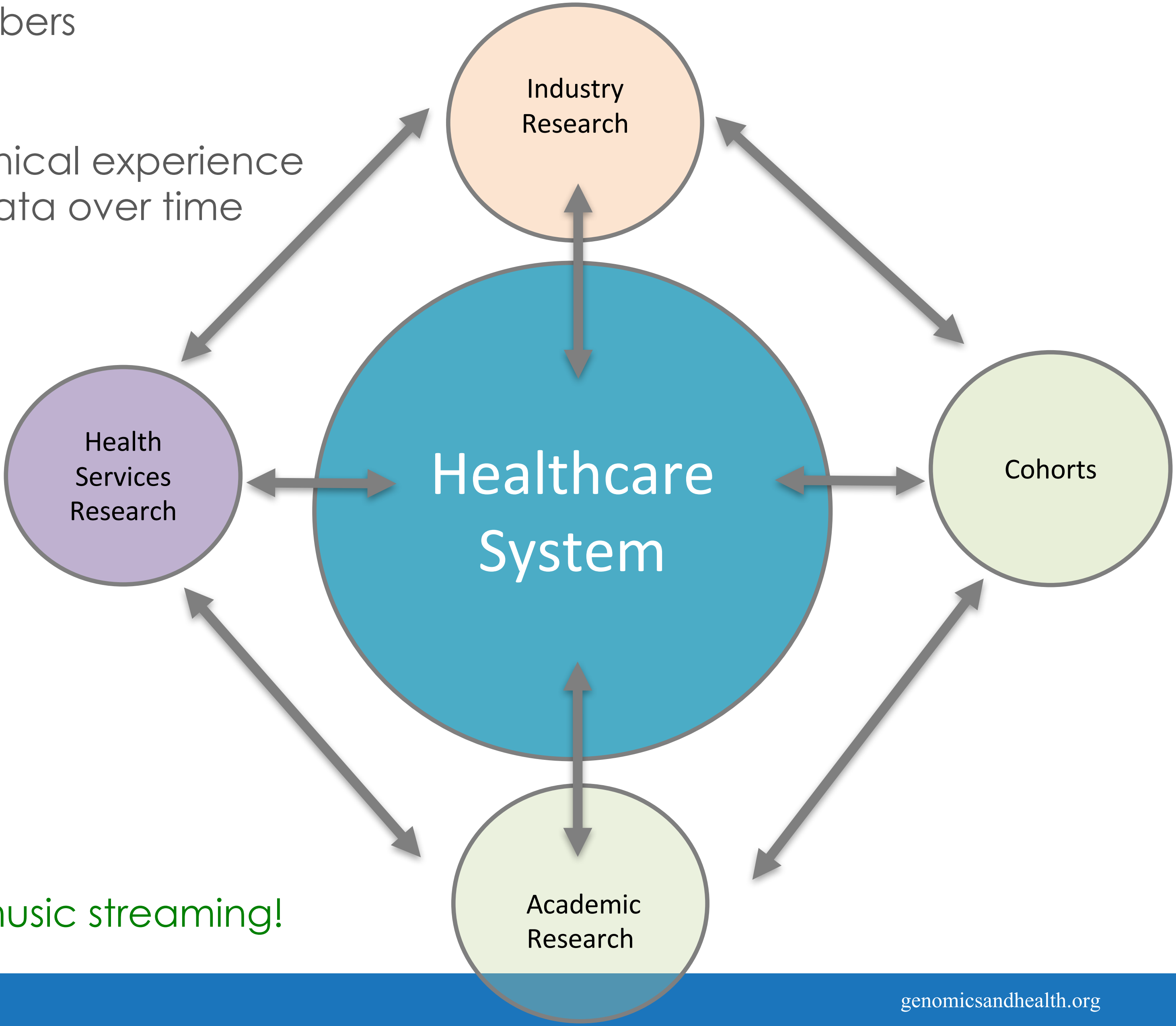
Cohorts

Clinical Trials

Academic Research

# Tremendous opportunity in digitized data

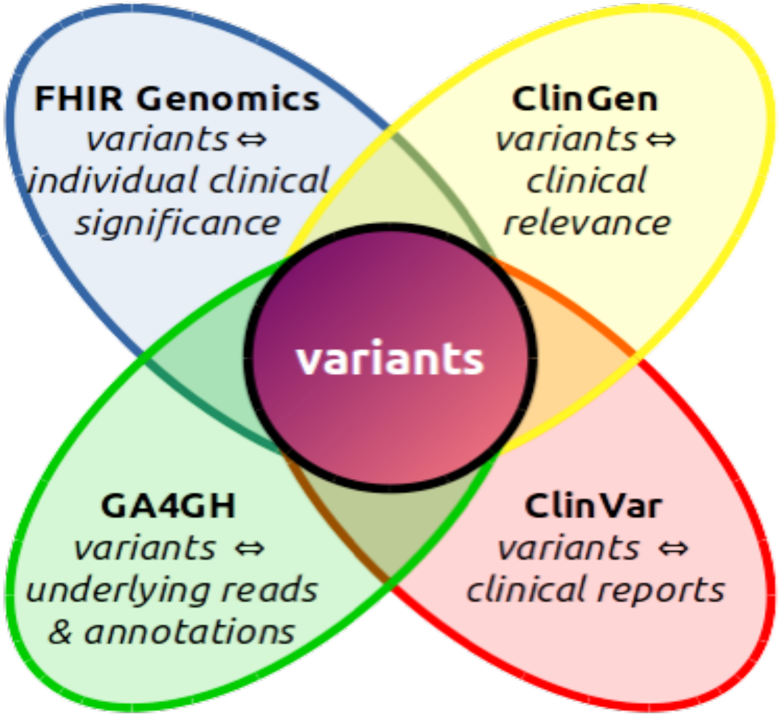
- Power in numbers
- Dynamic
- Interoperable
- Linkage of clinical experience & research data over time



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



**FHIR Genomics**
*variants ⇔ individual clinical significance*

**ClinGen**
*variants ⇔ clinical relevance*

**variants**

**GA4GH**
*variants ⇔ underlying reads & annotations*

**ClinVar**
*variants ⇔ clinical reports*

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

OncoKB
Precision Oncology Knowledge Base
Annotation of Somatic Mutations in Cancer

Precision Medicine KnowledgeBase
Weill Cornell Medicine

CIViC
Clinical Interpretations of Variants in Cancer

Cancer bioMarkers-db

Cancer Genome Interpreter

…

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Variant 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

**VICC Co-chairs:**

- Obi Griffith
- Nuria Lopez-Bigas
- David Tamborero
- Malachi Griffith

**Goals/Principles:**

- Clinical cancer variant interpretation
- Standards and guidelines
- Open content
- Interoperability

# 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

# GA4GH & G2MC +++++

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, “Envirotype”
- 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?

## Discussion Topic

What are the issues in your country (state/province/system/region) to accomplish the goals of a longitudinal genomic patient records?