Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.

Empowering personalised health through data sharing

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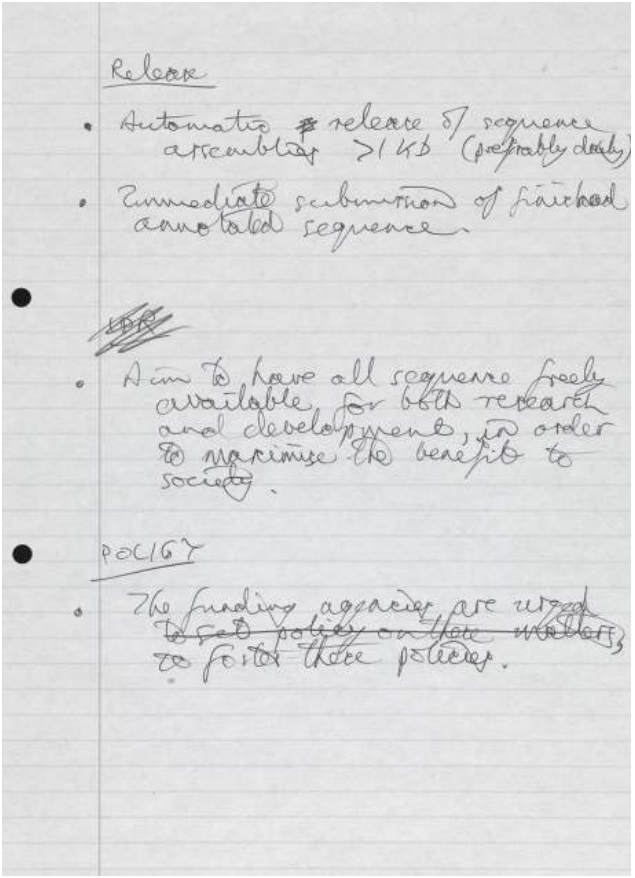
Data sharing



Release

- Automatic release of sequence assemblies >1 kb (preferably daily)
- Immediate submission of finished annotated sequence.

- Aim to have all sequence freely available for both research and development, in order to maximise the benefit to society.

POLICY

- The funding agencies are urged to ~~set policy on these matters~~ to foster these policies.

Data sharing: FAIR Principles

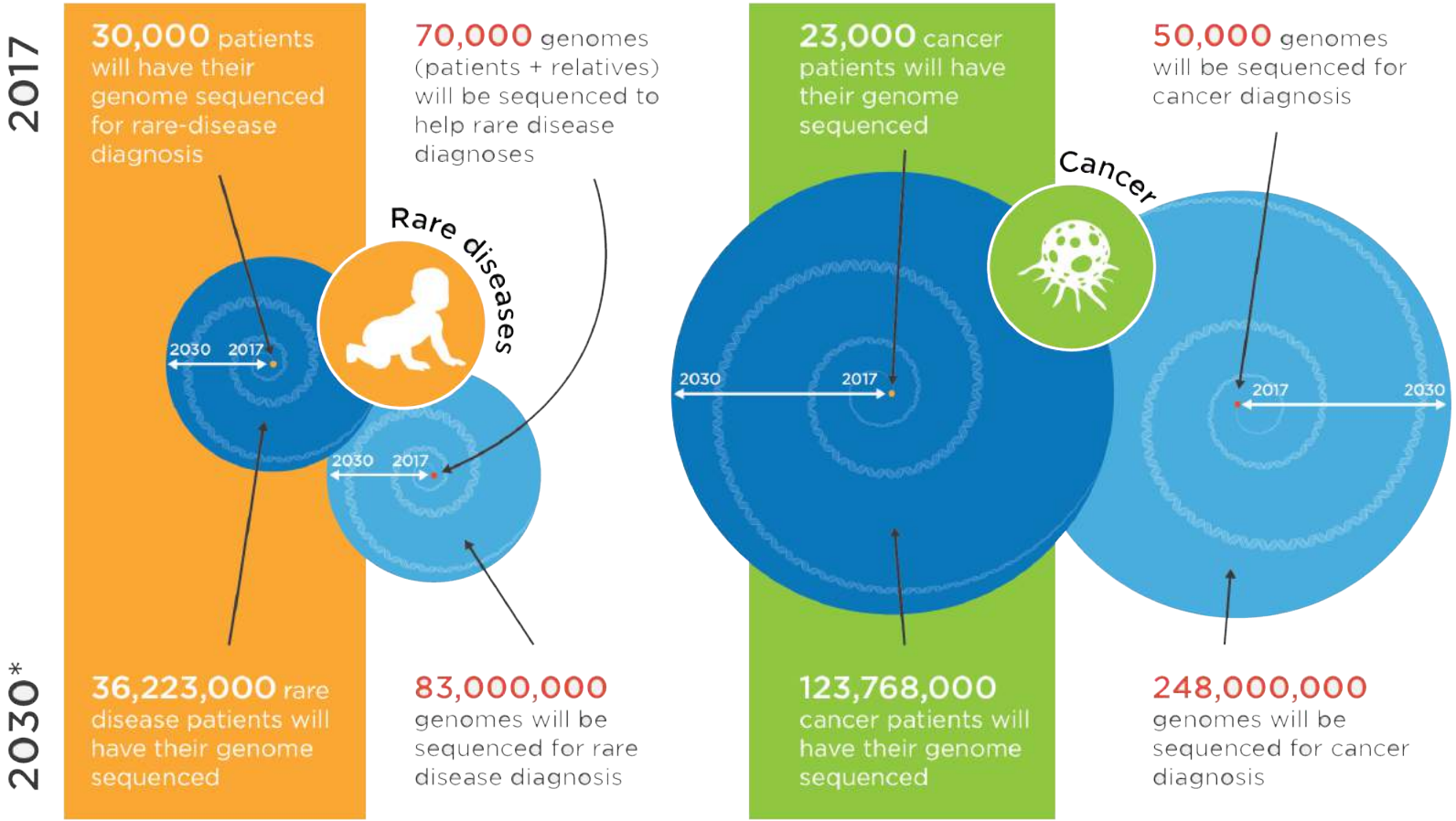
Making data

- Findable
- Accessible
- Interoperable
- Reusable

Motion to add Attributable and provide Recognition

Data sharing

Scale is changing



* Projected figures, based on current data and known status of genomics initiatives worldwide.

Genomics in Cancer

- When cancer patients have genomic sequence this informs clinical decisions ~10% of the time

- The clinical research community is confident this will increase steadily over the coming years

The world is changing

Percentage of whole genomes and exomes that are funded solely by healthcare systems

The GA4GH mission

The Global Alliance for Genomics and Health aims to accelerate progress in genomic science and human health by developing standards and framing policy for responsible genomic and health-related data sharing.

The GA4GH community

Global Alliance members include:

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

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Responsibility
Opportunity
Challenges

Responsibility

- Much of the technical knowhow around genomics is in the research community
- The expertise in clinical features and diagnosis is in the clinical community
- We have a joint responsibility to make this work for patients

Opportunity

- If we can enable secondary use of clinical genomic data for research we will have a >60 million virtual cohort by 2025
- Humans will be the best studied organisms on the planet due to healthcare
- We can improve clinical care and accelerate research for better patient outcomes

The challenges are surmountable

- Healthcare not used to this type/amount of data; need to draw on skills, learnings of research
- Clinical data are not interoperable: cultural, regulatory, technical differences between international healthcare systems. Portable analysis must be developed.
- As genomic data grows from terabyte to petabyte to exabyte scale, the community must re-tool
- Different nations have unique regulatory environments, requiring broad, reciprocal data access methods that are as open as possible while respecting national processes and patient consent.

Federation for global data sharing

It allows us to….

1. move analysis to data, not aggregate data close to each researcher
2. have broad, reciprocal data access methods, as open as possible while still respecting national processes and patient consent
3. transfer methods and skills into the healthcare sector
4. leverage healthcare data to make more discoveries on humans

The GA4GH data sharing toolkit

Genomic Data

- GA4GH Streaming API
- Genomics API
- File Formats Standards
- Standard for DNA sequence reads

Regulatory & Ethics

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

Data Security

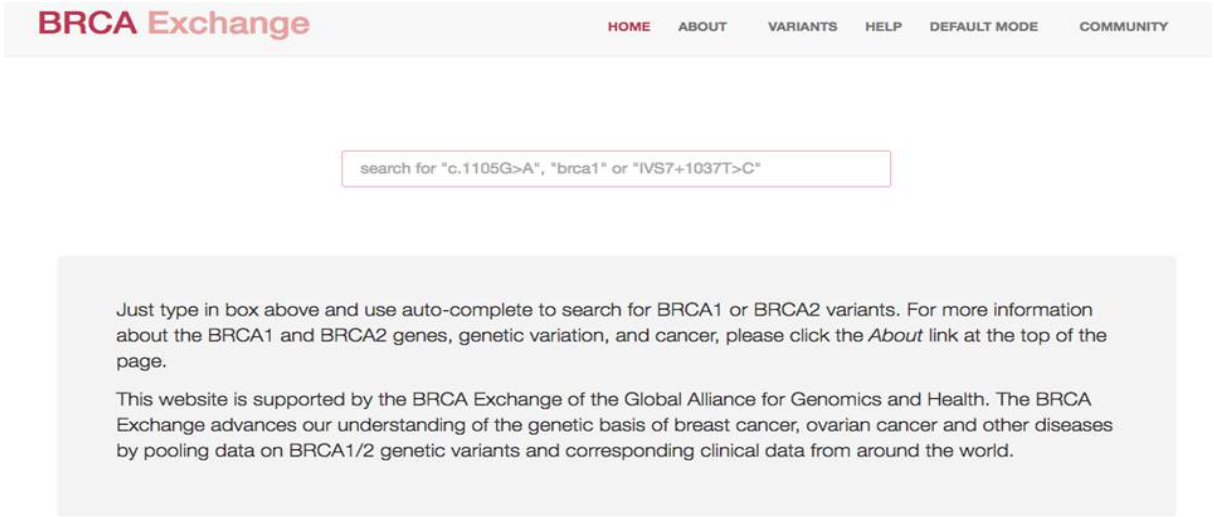
- Security Infrastructure

The BRCA Challenge

To **improve care of patients** at risk of breast / ovarian cancer by creating a **decentralized, global public repository** to assist ***BRCA1 BRCA2 analysis***

1. Share *BRCA1* and *BRCA2* variant data publicly
2. Create an environment for collaborative variant curation with access to evidence (phenotypes, family history, genetic data, and functional studies)
3. Create 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, and engage with patient advocacy organizations
5. Create a model for all genes

The BRCA Challenge



BRCA CHALLENGE

brcaexchange.org

GA4GH standards and frameworks will...

- Enable interoperable tools and exchange of data between organisations and countries
- Advance research and clinical management for the benefit of patients with rare disease, cancer, and complex diseases
- Enable an ecosystem of data stewards to collaboratively use, analyse, and store the data sets that drive precision medicine
- Facilitate real-world evidence generation and continuous learning among diverse healthcare and research stakeholders, contributing to better patient outcomes globally

GA4GH Connect: 2017 Driver Projects

All of Us Research Program
United States

Australian Genomics
Australia

BRCA Challenge
International

CanDIG
Canada

ClinGen
United States

ELIXIR Beacon
Europe

ENA / EVA / EGA
Europe

Genomics England
United Kingdom

Human Cell Atlas
International

ICGC-ARGO
International

Matchmaker Exchange
International

Monarch Initiative
International

National Cancer Institute Genomic Data Commons
United States

TOPMed
United States

Variant Interpretation for Cancer Consortium
International

By 2022 we expect...

- 10s of millions of genome sequences to be available from research / clinical care, spanning many nationalities, ethnicities
- majority of genomic data to be collected in the clinical setting, rather than in research
- massive increase in both spending and sequencing output per $/£/€/¥, enabling a new era of genomic medicine
- clinical genomic data to feed back into research, and vice versa, enabling a global ‘learning health system’
- every qualified clinician, researcher worldwide will be able to responsibly share and access federated databases

Thank you

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