Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.

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.

Human Rights Foundation

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Universal Declaration of Human Rights (1948)

27(1) "The Right to Science"

"Everyone has the right freely to participate in the cultural life of the community, to enjoy the arts and **to share in scientific advancement and its benefits.** "

27(2) "The Right to Recognition"

"Everyone has the right to the protection of the moral and material interests resulting from any scientific, literary or artistic production of which he is the author."

Framework for Responsible Sharing of Genomic and Health-Related Data

ga4gh.org/framework

FOUNDATIONAL PRINCIPLES

- Respect Individuals, Families and Communities
- Advance Research and Scientific Knowledge
- Promote Health, Wellbeing and the Fair Distribution of Benefits
- Foster Trust, Integrity and Reciprocity

AIMS OF THE FRAMEWORK

- Foster responsible data sharing
- Protect and promote the welfare, rights, and interests of groups and individuals who donate their data
- Provide benchmarks for accountability
- Establish a framework for greater international data sharing, cooperation, collaboration, and governance

How Many Genomes?

RESEARCH

HEALTHCARE

60M individuals
132.5 sequences

CLINICAL TRIALS

2.7-3M individuals

COHORTS

140M individuals

200+ Genomic Data Initiatives Globally

Clinical/Genomic Medicine

40

Initiatives

Research

70

Initiatives

National

21

From 15 Countries

Cohorts

64

Globally

THE OPPORTUNITY...

If we can enable secondary use of clinical genomic data for research, we will have**a virtual cohort of >60 million samples by 2025.**

Global Genomic Data Sharing Can...

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Demonstrate patterns in health & disease

Increase statistical significance of analyses

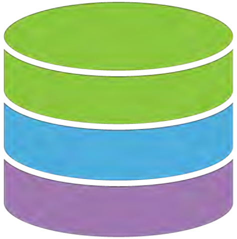
Lead to “stronger” variant interpretations

Increase accurate diagnosis

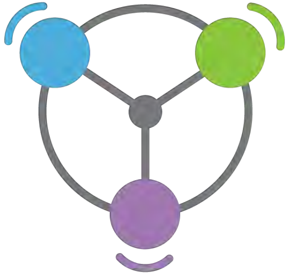
Advance precision medicine

Different Approaches to Data Sharing

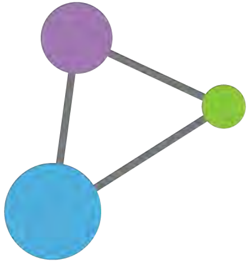
Centralized Genomic Knowledge Bases



Data Commons
Trusted, controlled single repository of multiple datasets



Hub and Spoke Federation
Common data elements, structures, access, and usage rules



Linkage of distributed and disparate datasets

A New Paradigm for Data Sharing

FROM

Data Copying

→

TO

Data Visiting

The Future: Federation

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"A grouping of autonomous organizations and datasets with a centralised control"

Federation allows us to….

1. Move analysis to data , not aggregate data close to each researcher
2. Have broad, reciprocal data access methods which respect national processes and patient consent
3. Transfer methods and skills into the healthcare sector
4. Leverage healthcare data to make more discoveries on humans

Federation

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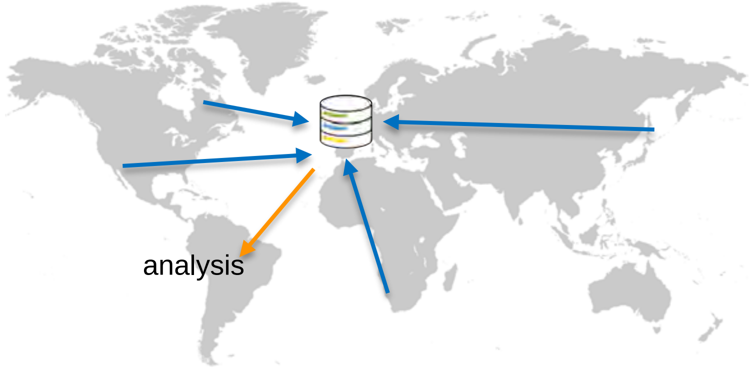
Open Research Data

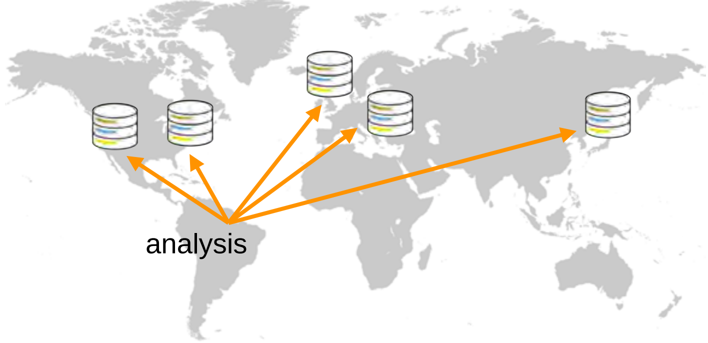


Aggregate data globally

Download, analyze locally

Continues for basic research

Healthcare Data with Research Use



Aggregate data locally (via VMs)

Collate analyses

New approach for research and healthcare

Core Principles of Data Sharing

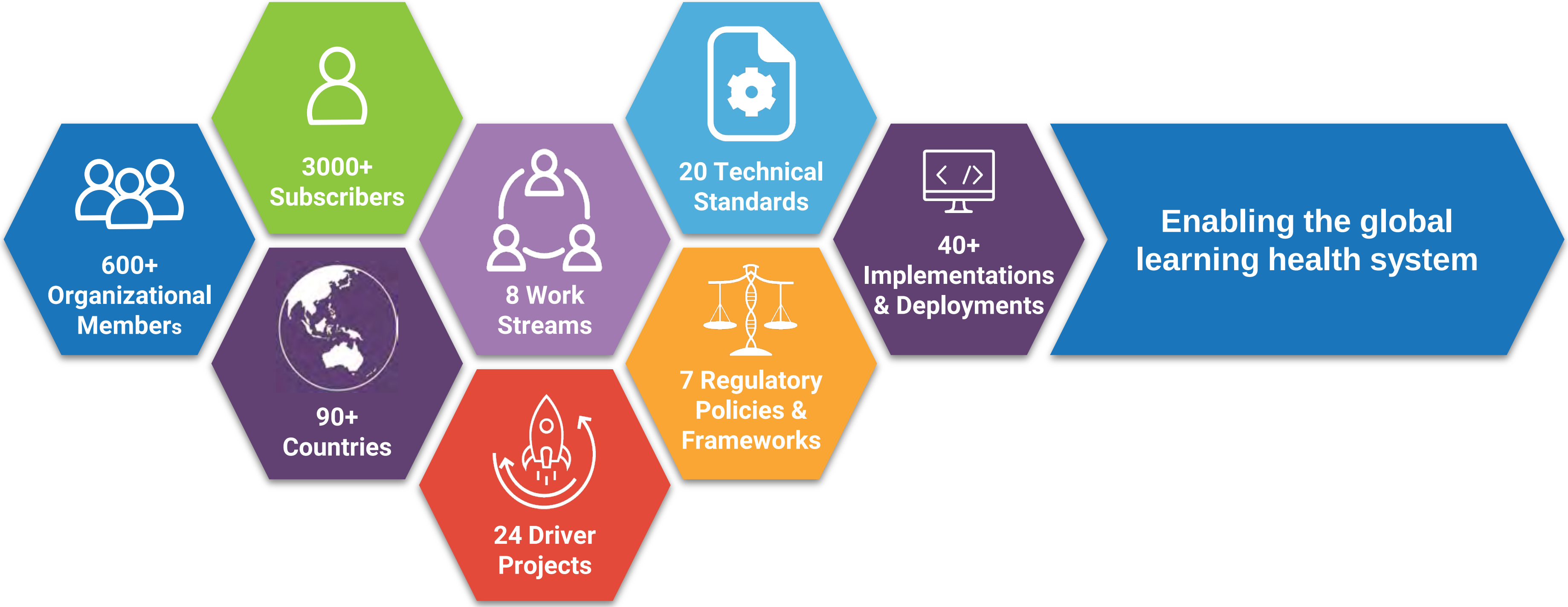
Enable International Data Sharing

Sharing across the translational continuum

Encourage technology - enabled federated approaches

Promote Interoperability

The GA4GH Ecosystem



600+ Organizational Members

3000+ Subscribers

90+ Countries

8 Work Streams

24 Driver Projects

20 Technical Standards

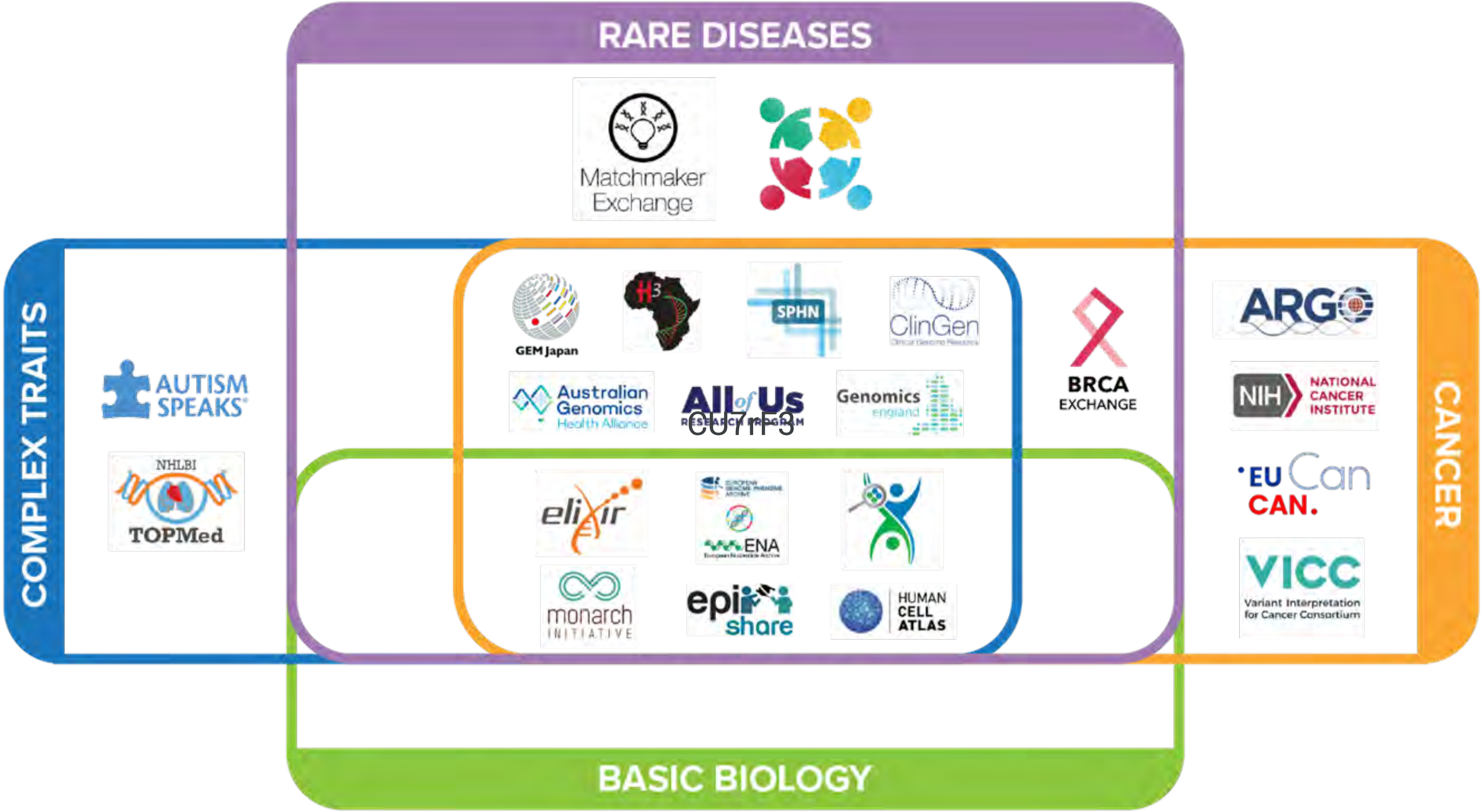
7 Regulatory Policies & Frameworks

40+ Implementations & Deployments

Enabling the global learning health system

Work Streams vs. Driver Projects

Work Streams	Driver Projects
• Internal to GA4GH • Deliver standards and policy frameworks based on the Strategic Roadmap • Run by 2 volunteer leads within the community • Contributors come from a variety of projects and organizations • *Example*: Data Use & Researcher Identities	• External to GA4GH • Provide input towards the Strategic Roadmap and standards development • Contribute FTE resources to Work Streams for standards development • Pilot implementations for new standards • *Examples*: ARGO Accelerating Research in Genomic Oncology International Cancer Genome Consortium BRCA CHALLENGE Australian Genomics Health Alliance


RARE DISEASES
Matchmaker Exchange
COMPLEX TRAITS
AUTISM SPEAKS
NHLBI
TOPMed
GEM Japan
H3
SPHN
ClinGen
Clinical Genome Resource
Australian Genomics
Health Alliance
All of Us
RESEARCH PROGRAM
CU7rF3
Genomics england
BRCA EXCHANGE
ARGO
NIH NATIONAL CANCER INSTITUTE
EU Can CAN.
VICC
Variant Interpretation for Cancer Consortium
CANCER
elixir
EUROPEAN GENOME-PHENOME ARCHIVE
ENA
European Nucleotide Archive
monarch INITIATIVE
epi share
HUMAN CELL ATLAS
BASIC BIOLOGY

GENOMICS IN HEALTH
IMPLEMENTATION FORUM

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H3

gms

IndiGen Genomics for Public Health in India

All of Us
RESEARCH PROGRAM

Australian
Genomics
Health Alliance

BIG

BIPMed Brazilian Initiative on PRECISION MEDICINE

DANISH NATIONAL
GENOME CENTER

GEM Japan

Genomics
england

elixir

قطر جينوم
QATAR GENOME
عضو في مؤسسة قطر
Member of Qatar Foundation

FINNGEN

Nordic Alliance
for Clinical Genomics

SPHN
Swiss
Personalized
Health
Network

Global Collaboration

Global Alliance
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INTERNATIONAL COMMON DISEASE ALLIANCE | Maps to Mechanisms to Medicine

CINECA

HDRUK
Health Data Research UK

Global Genomic Medicine Collaborative

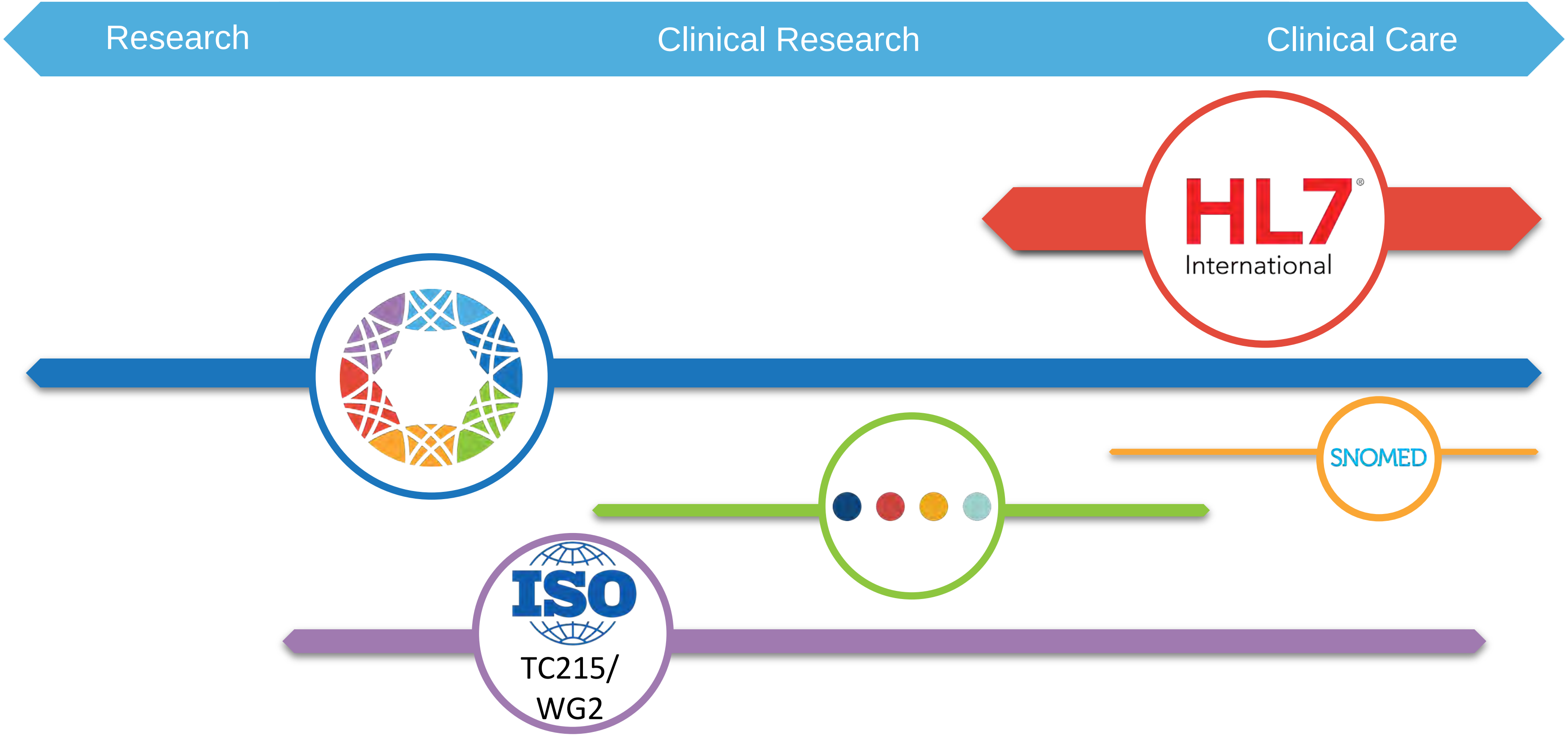
HGVS
HUMAN GENOME VARIATION SOCIETY

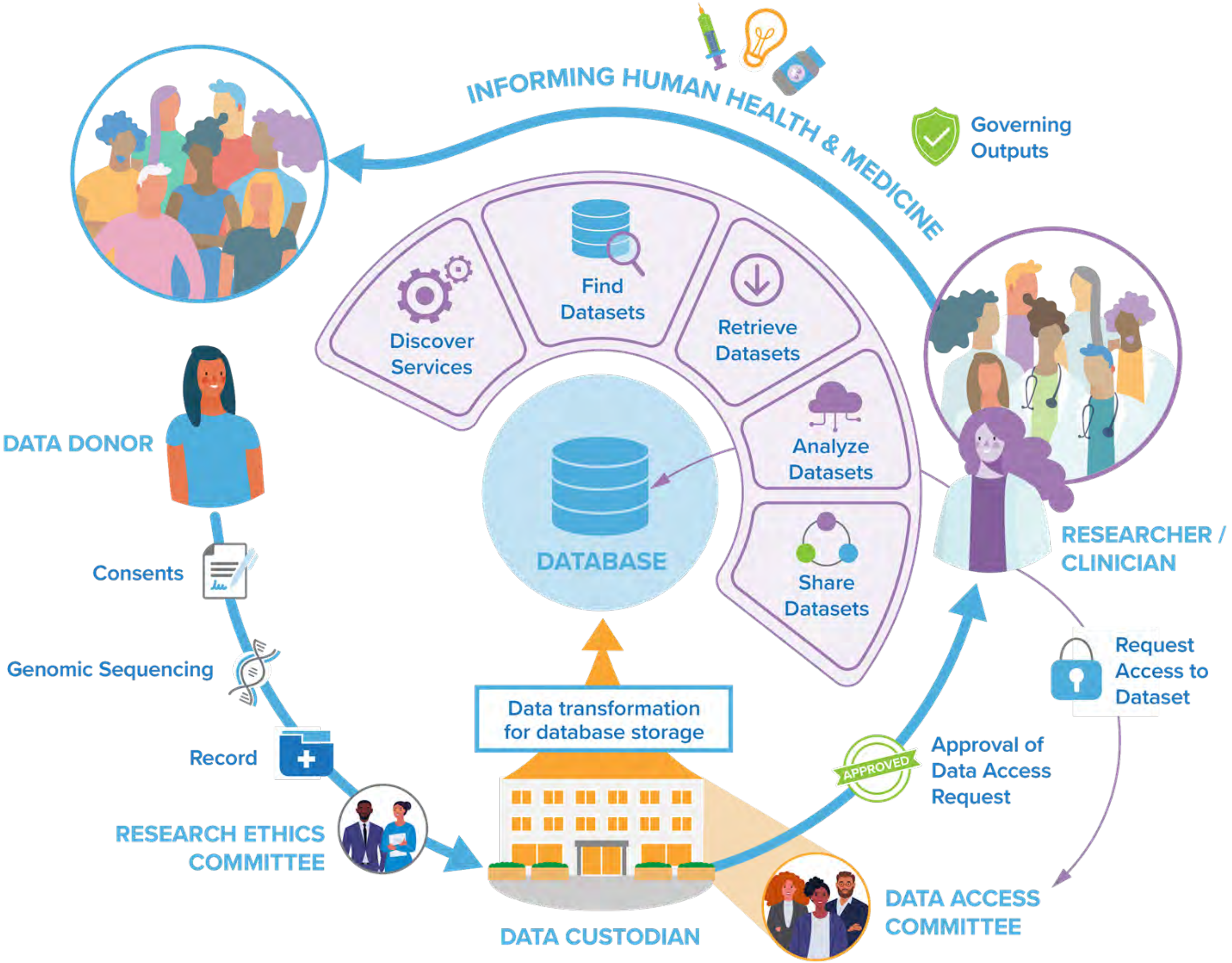
WORLD ECONOMIC FORUM

THE HUMAN VARIOME PROJECT

HUMAN GENOME ORGANISATION
HUGO

Alignment with Other Genomics Standards Organizations





INFORMING HUMAN HEALTH & MEDICINE

Governing Outputs

DATA DONOR

Consents

Genomic Sequencing

Record

RESEARCH ETHICS COMMITTEE

Discover Services

Find Datasets

Retrieve Datasets

Analyze Datasets

Share Datasets

DATABASE

Data transformation for database storage

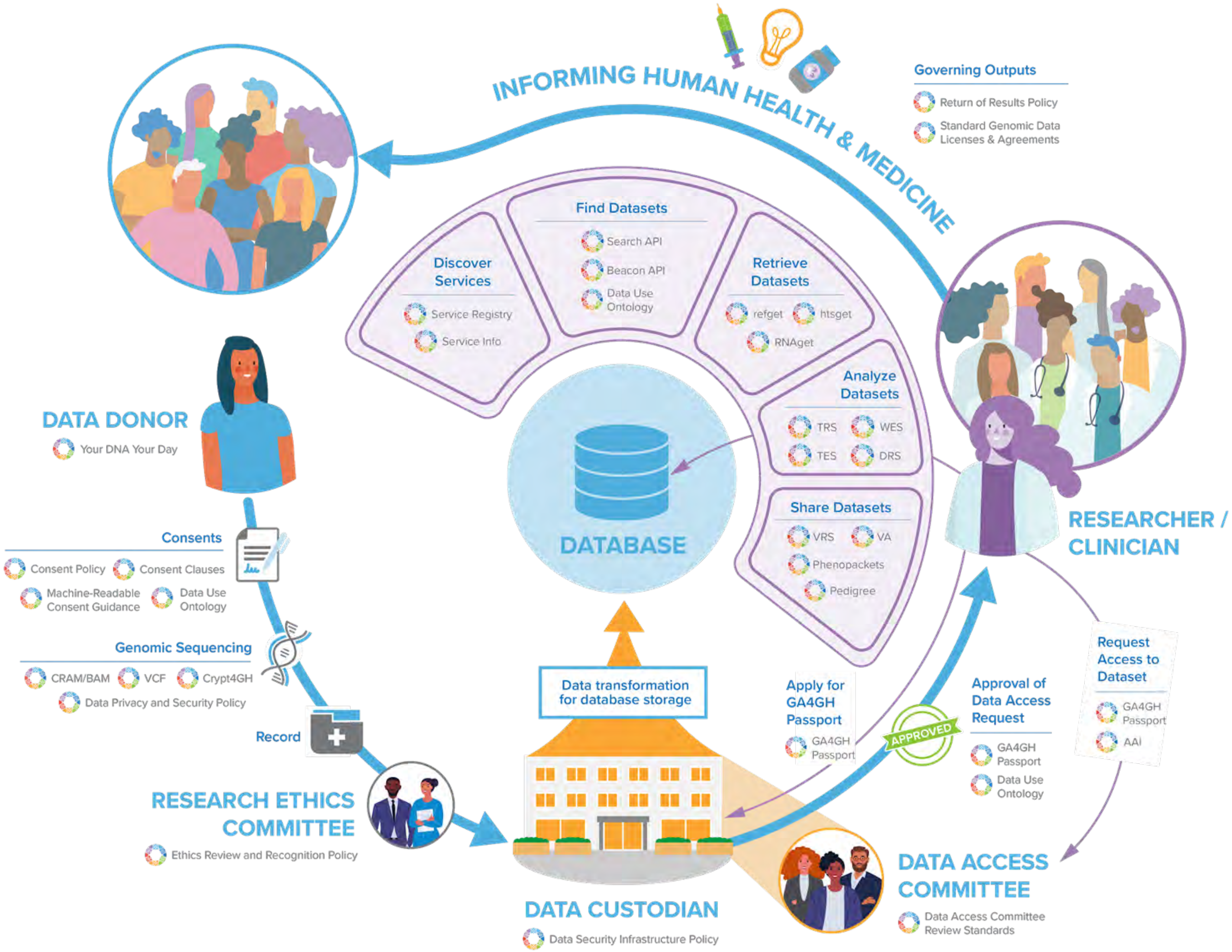
DATA CUSTODIAN

RESEARCHER / CLINICIAN

Request Access to Dataset

Approval of Data Access Request

DATA ACCESS COMMITTEE



INFORMING HUMAN HEALTH & MEDICINE

Governing Outputs
- Return of Results Policy
- Standard Genomic Data Licenses & Agreements

DATA DONOR
- Your DNA Your Day

Consents
- Consent Policy
- Consent Clauses
- Machine-Readable Consent Guidance
- Data Use Ontology

Genomic Sequencing
- CRAM/BAM
- VCF
- Crypt4GH
- Data Privacy and Security Policy

Record

RESEARCH ETHICS COMMITTEE
- Ethics Review and Recognition Policy

DATA CUSTODIAN
- Data Security Infrastructure Policy

Data transformation for database storage

DATABASE

Discover Services
- Service Registry
- Service Info

Find Datasets
- Search API
- Beacon API
- Data Use Ontology

Retrieve Datasets
- refget
- htsget
- RNAget

Analyze Datasets
- TRS
- WES
- TES
- DRS

Share Datasets
- VRS
- VA
- Phenopackets
- Pedigree

RESEARCHER / CLINICIAN

Request Access to Dataset
- GA4GH Passport
- AAI

Approval of Data Access Request
- GA4GH Passport
- Data Use Ontology

Apply for GA4GH Passport
- GA4GH Passport

DATA ACCESS COMMITTEE
- Data Access Committee Review Standards

Public willingness to share DNA*

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Would you donate your anonymous DNA information and medical information for use by:

Medical doctors - 50-60% (Japan and Egypt lowest)

Non-profit researchers – 40-50% (Japan, Egypt and India lowest)

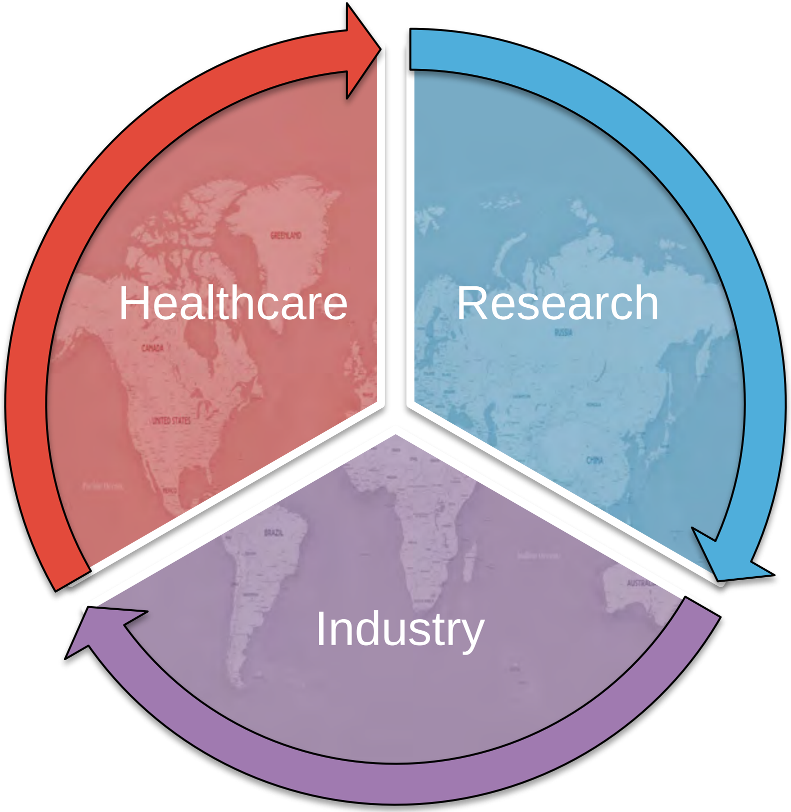
For-profit researchers – 20-40% (Germany, Japan and UK lowest)

*summarized from “Your DNA Your Say” survey led by Dr Anna Middleton, Cambridge UK

21 countries representing 3 billion people X 20% = ?

Global Advancement for Health

Health data sharing across traditional silos supports advancement across all relevant sectors



Interoperable APIs, standards & policy frameworks support global data sharing across sectors

COVID-19 Distractions & Opportunities

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COVID-19 challenged regular work processes, availability and priorities

But from this, there are opportunities:

1. Increased awareness of the need to share data at scale and across boundaries
2. Benefits of a virtual meeting – greater participation (700+ registrants)
3. Tools to enable global collaboration