



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.



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

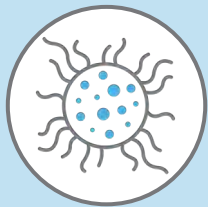
THE OPPORTUNITY...

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



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Centralized Genomic Knowledge Bases



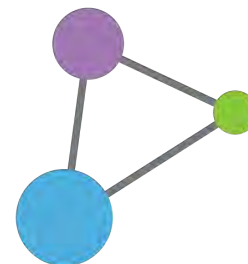
Data Commons

Trusted, controlled
single repository of
multiple datasets



Hub and Spoke Federation

Common data elements,
structures, access, and
usage rules

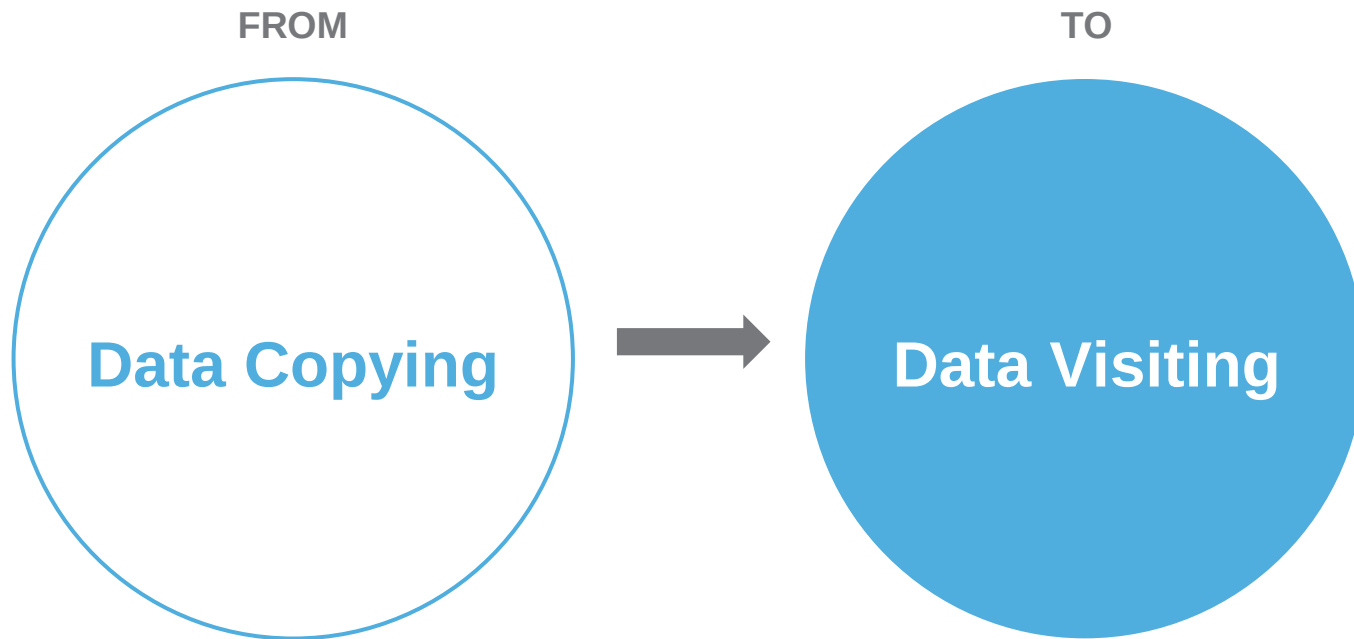


Linkage of distributed and disparate datasets

A New Paradigm for Data Sharing



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“A grouping of autonomous organizations and datasets with a centralised control”

Federation allows us to....

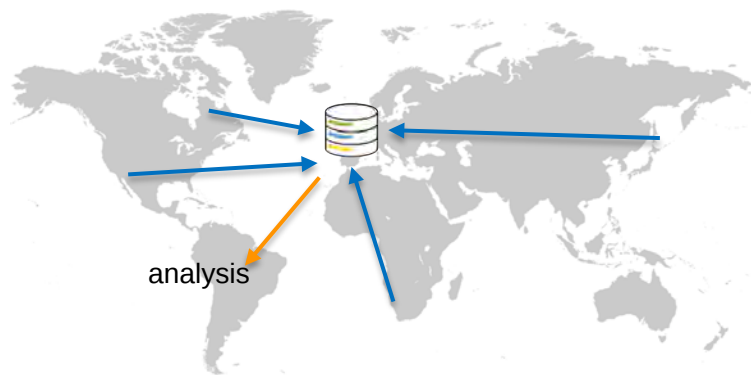
1. **Move analysis to data**, not aggregate data close to each researcher
2. **Have broad, reciprocal data access methods** which respect national processes and patient consent
3. Transfer methods and skills into the **healthcare sector**
4. **Leverage healthcare data** to make more discoveries on humans

Federation



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Open Research Data

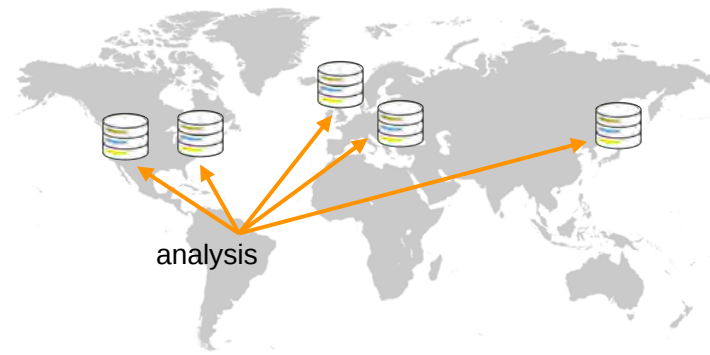


Aggregate data globally

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

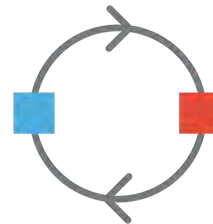
Core Principles of Data Sharing



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Enable International
Data Sharing



Sharing across the
translational
continuum



Encourage technology -
enabled federated
approaches

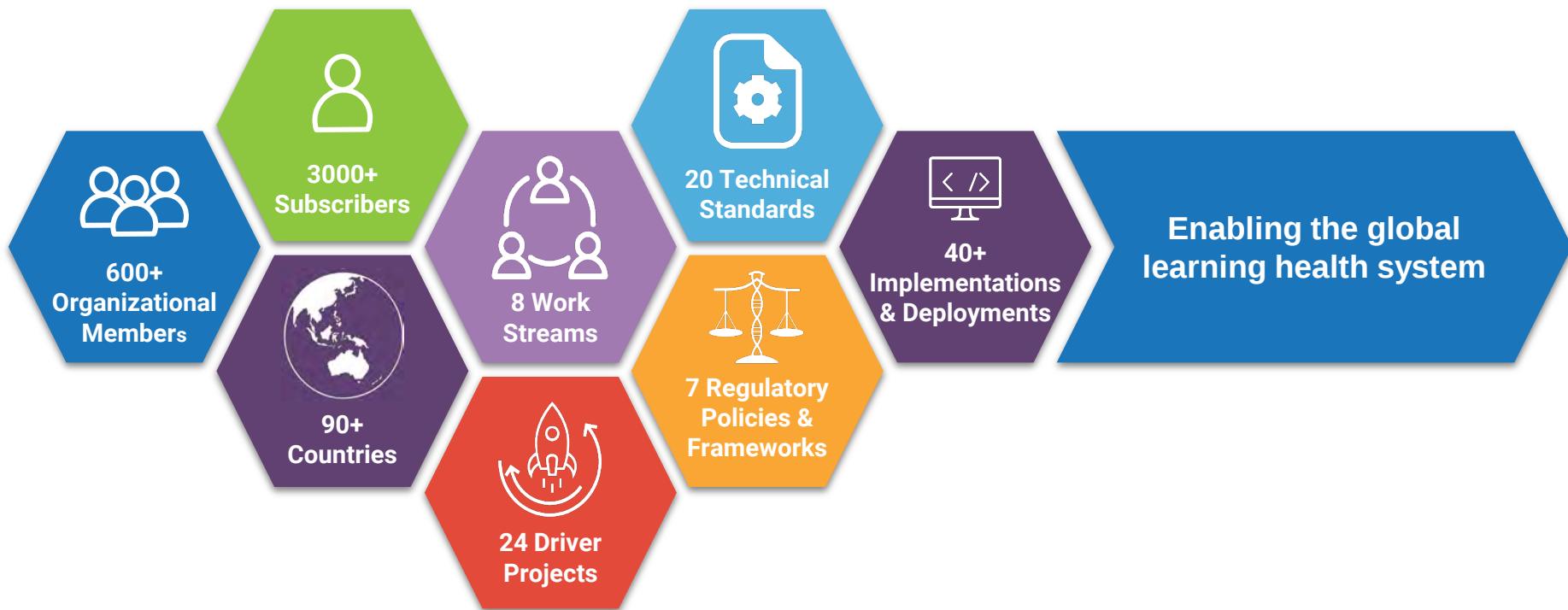


Promote
Interoperability

The GA4GH Ecosystem



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Work Streams vs. Driver Projects



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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 • <i>Example</i> 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 • <i>Examples</i>  Accelerating Research in Genomic Oncology International Cancer Genome Consortium  BRCA CHALLENGE  Australian Genomics Health Alliance

COMPLEX TRAITS



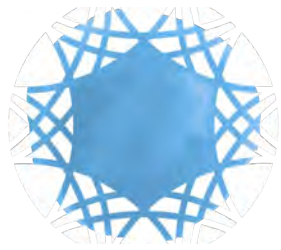
RARE DISEASES



CANCER

BASIC BIOLOGY





GENOMICS IN HEALTH IMPLEMENTATION FORUM



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Global Collaboration



Alignment with Other Genomics Standards Organizations

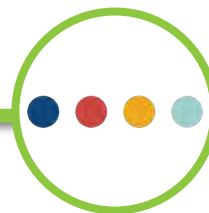


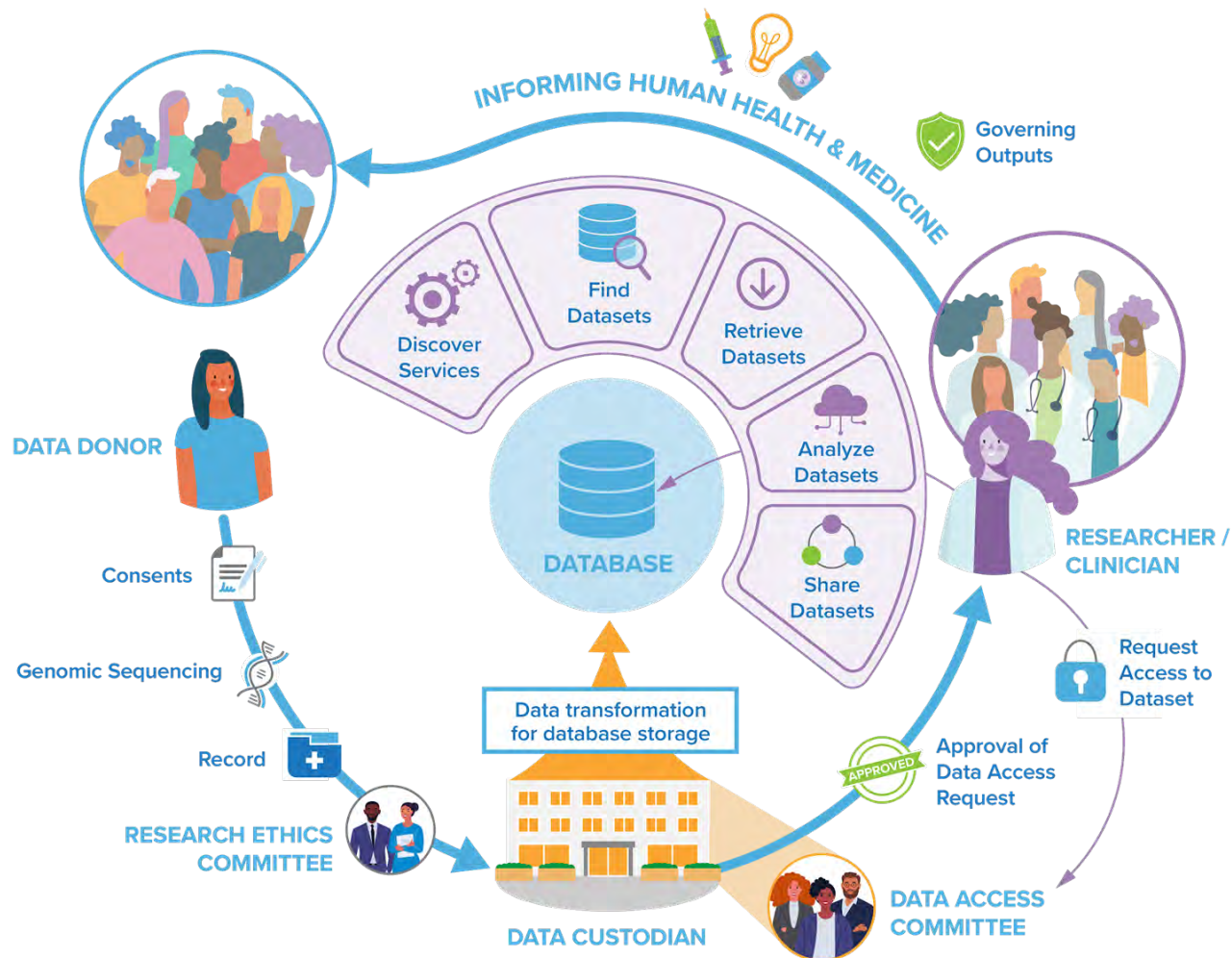
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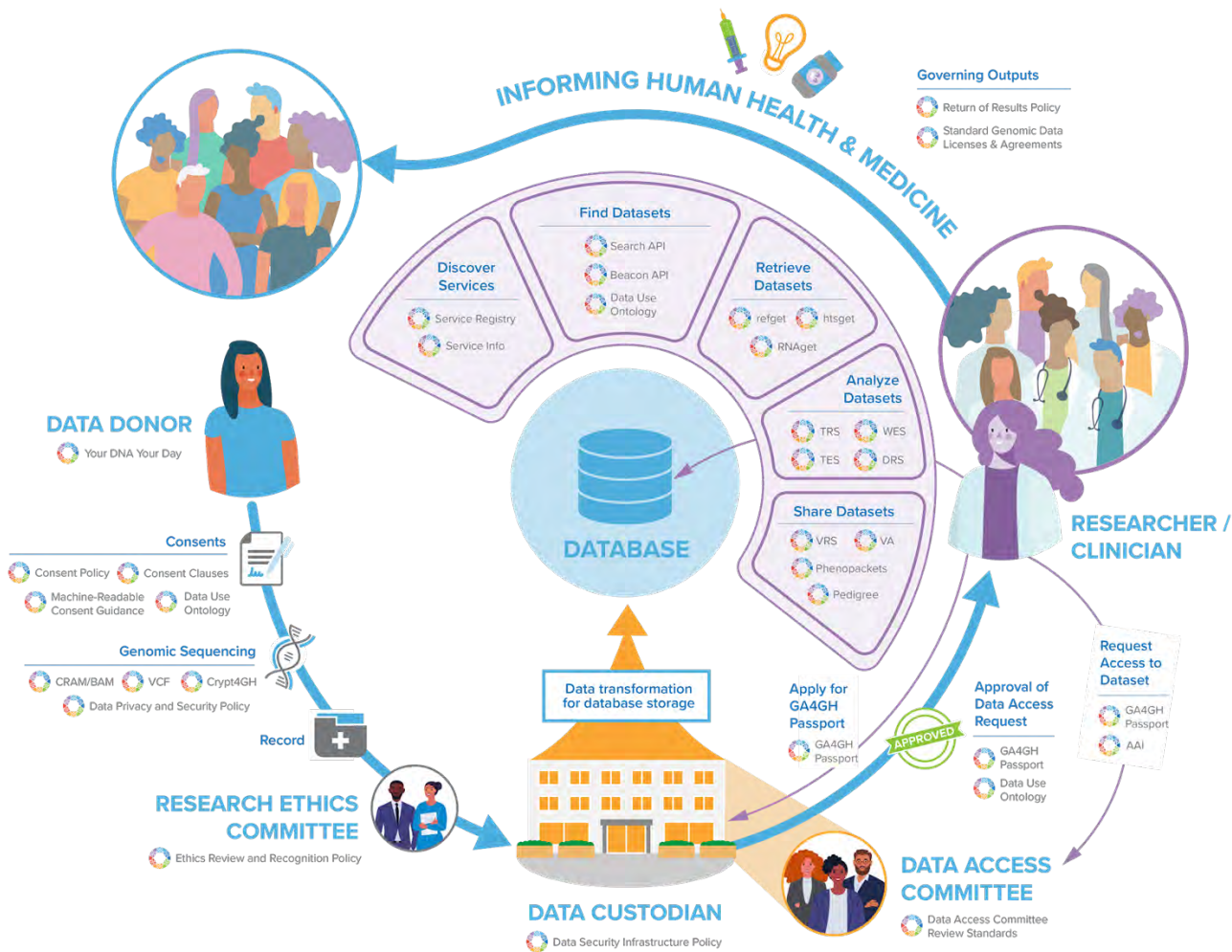
Research

Clinical Research

Clinical Care









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)

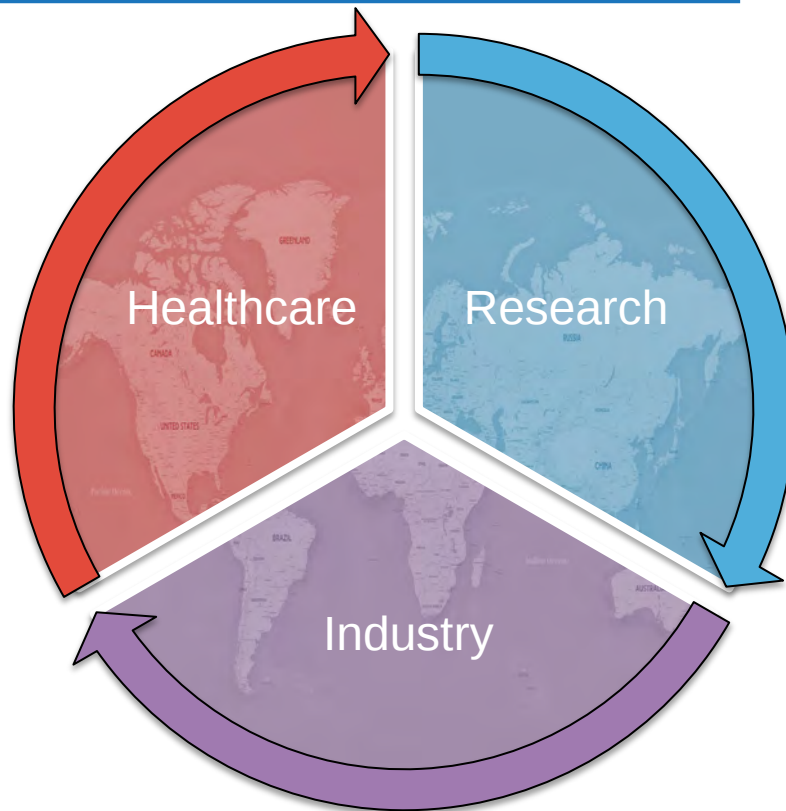
*summarized from “Your DNAYour Say” survey led by Dr Anna Middleton, Cambridge UK

21 countries representing 3 billion people X 20% = ?

Global Advancement for Health



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**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

