Global Alliance
for Genomics & Health

Catalysing genomic and health data sharing

GA4GH 2024 Annual Report

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Letter from the Chair

Dear colleagues,

2024 marked my first year as Chair of GA4GH. I am continuously in awe of the energy and passion of the community, our collaborative efforts to advance genomic data sharing, and the progress we have made together. This report highlights the work of the community, including key updates and success stories. I invite you to explore what we have accomplished together over the past year.

The convening power of GA4GH is one of our community's greatest strengths. From our Plenary and Connect conferences to regular working meetings, we have a platform to cultivate and drive global collaborations. Together, we can build bigger networks of data that deliver new insights into the causes of disease and help us realise the potential of precision medicine — all within an ethical and human rights framework. I look forward to building new connections and welcoming more projects and initiatives into our community.

One of GA4GH's imperatives — and one of my goals as Chair of GA4GH and as a clinical lab director — is to bridge the often disparate worlds of research and healthcare. In 2024, we launched a new Community of Interest, the Clinical Genomics Laboratory Community, to partner more closely with the clinical community. I hope to continue expanding our work in this area to improve the accuracy and efficiency of genetic diagnosis to ultimately improve patient care.

I want to thank our community of contributors, funders, and sponsors for making all of our work possible. I look forward to collaborating with the GA4GH community to continue catalysing data sharing for the betterment of human health and care.

Heidi Rehm

Heidi Rehm

Welcoming new leadership

GA4GH product development is driven by members of the genomics and health community, whose leadership and expertise are crucial to ensuring GA4GH products are fit for purpose and can meet real-world needs. In 2024, we announced the appointment of nine new Co-Leads to help drive our work forward.

Mallory Freeberg
EMBL's European Bioinformatics Institute
GA4GH Implementation Forum Co-Lead
6 March 2024

Alastair Thomson
ARPA-H
GA4GH Implementation Forum Co-Lead
6 March 2024

Jaime Delgado
Universitat Politècnica de Catalunya
Data Security Work Stream Co-Lead
24 May 2024

Benjamin Haibe-Kains
Princess Margaret Cancer Centre and University of Toronto
Cancer Community Co-Lead
18 July 2024

Zinaida Perova
EMBL's European Bioinformatics Institute
Cancer Community Co-Lead
18 July 2024

Bernie Pope
The University of Melbourne
Cancer Community Co-Lead
18 July 2024

Jonathan Lawson
Broad Institute of MIT and Harvard
Data Use & Researcher Identity Work Stream Co-Lead
8 August 2024

Melissa Cline
University of California, Santa Cruz
Genomic Knowledge Standards Work Stream Co-Lead
29 August 2024

Alex Wagner
Nationwide Children's Hospital
Genomic Knowledge Standards Work Stream Co-Lead
29 August 2024

We want to extend a thank you to our exiting Co-Leads, who have been instrumental in driving GA4GH's work forward.

David Bernick
Broad Institute of MIT and Harvard
Data Security Work Stream Co-Lead

Lucila Ohno-Machado
University of California, San Diego
Data Security Work Stream Co-Lead

Andrew Russette
National Center for Biotechnology
Information (NCBI/NLM/NIH)
Data Security Work Stream Co-Lead

David Torrents
Barcelona Supercomputing Center
Cancer Community Co-Lead

Robert Freimuth
Mayo Clinic
Genomic Knowledge Standards
Work Stream Co-Lead

Andy Yates
EMBL's European Bioinformatics
Institute
Genomic Knowledge Standards
Work Stream Co-Lead

In 2024, GA4GH launched its inaugural **Strategic Leadership Committee (SLC)**. The SLC advises the Executive Leadership Committee and the Product Steering Committee to inform GA4GH strategies and advance responsible use of genomic and related health data. SLC members include:

Michael Baudis
University of Zurich

Tiffany Boughtwood
Australian Genomics

Mélanie Courtot
Ontario Institute for Cancer Research

Robert Freimuth
Mayo Clinic

David Glazer
Verily

Oliver Hofmann
University of Melbourne

Yann Joly
McGill University

Augusto Rendon
Genomics England

Serena Scollen
ELIXIR

Heidi Sofia
National Center for
Biotechnology Information

2024 in numbers

Our community now comprises…

582 Organisational Members
across **53** countries

28 Driver Projects
active in **113** countries

5 Host Institutions
active in **3** countries

4 Strategic Partners committed to responsible
data sharing in the US, UK, Europe, and globally

355 contributors
active across **30** countries

In 2024, we had…

2 — **new products** approved by the GA4GH Product Steering Committee, bringing our total to **40** products

13 — **academic papers** published by Work Stream contributors

6 — **projects** conducted through the Google Summer of Code initiative

12 — **meetings and workshops**

2024 timeline

14 February • National Institutes of Health (NIH) and GA4GH commit to ongoing collaboration

6 April • Health Data Research UK (HDR UK) and GA4GH form a strategic partnership to unite genomic and health data

21 to 24 April • The 2024 April Connect meeting brings the GA4GH community together to advance development and implementation of GA4GH products

28 May • GA4GH submits comments on the World Health Organization’s (WHO) draft principles for human genome access, use, and sharing

13 June • GA4GH introduces the members of its inaugural Strategic Leadership Committee

20 June • The Victor Phillip Dahdaleh Institute of Genomic Medicine at McGill University is formally named a Host Institution of GA4GH

24 June • The Regulatory & Ethics Work Stream defines and promotes consistent usage and application of genetic discrimination

11 July • GA4GH and the Research Data Alliance (RDA) agree to a strategic relationship to advance responsible data sharing

21 August • New Product Development and Approval Process fosters consensus through collaboration and consultation

9 September • Diversity in Datasets product encourages a holistic approach to implementing diversity in research studies

16 to 20 September • The global genomics community comes together for GA4GH 12th Plenary

7 October • GA4GH community members and international leaders in genomics call for action to scale research and clinical genomic data sharing

9 October • Updates to the Variant Call Format (VCF) supports large cohort analysis

19 November • Model Data Access Agreement Clauses are approved as an official GA4GH Product

Global outreach and engagement

GA4GH is committed to cultivating a global community that is open and accessible to people of all perspectives and backgrounds to ensure that everyone benefits from advancements in genomics. GA4GH has outlined several organisational goals to be achieved over the next five years to facilitate and encourage meaningful engagement from the entire global genomics community, with particular emphasis on empowering participation from members of underrepresented communities.

GA4GH's global outreach and engagement efforts centre around three fundamental priority areas that anchor our mission and shape our future initiatives:

PEOPLE. The GA4GH community comprises people from a variety of backgrounds who feel represented, included, and recognised.

PRODUCTS. GA4GH builds products using ethical tools that minimise bias and maximise fairness.

PRACTICE. GA4GH products actively meet the needs of genomics researchers and clinicians around the globe, regardless of resources, geography, and other limiting factors.

GA4GH will work towards these priority areas to foster engagement and greater participation from organisations around the world. This will ensure that our products are relevant and fit for purpose across a wide range of global contexts and that there is a diversity of data — all of which are critical to inform human health.

Updates on GA4GH community imperatives

GA4GH aims to accelerate progress in human health by establishing a shared global approach to responsible, broad, and democratised use of genomic and related health data. The GA4GH community has identified three imperatives that are essential to realising GA4GH's mission: supporting implementation of GA4GH products, improving interoperability between standards, and engaging with healthcare.

2024 HIGHLIGHTS

Supporting implementation of GA4GH products

GA4GH standards provide the blueprint for sharing genomic and health data in secure, responsible, and technology-forward ways. Supporting implementation efforts is vital for broad adoption of GA4GH products.

Building a global, interoperable federated network

The **Federated European Genome-phenome Archive (FEGA)** is building a global resource to facilitate the discovery and access of human -omics data, with the goal of accelerating health and disease research. To build this infrastructure and governance framework, FEGA has adopted GA4GH standards, including the **Data Use Ontology**. As of December 2024, there are 7 participating countries, or nodes, and 37 datasets available in the **FEGA Network**, which are discoverable on the EGA website. To access data from a node, researchers submit a request to the corresponding data access committee; access is then granted based on each dataset's jurisdiction. By bringing more data together through a federated approach, FEGA aims to maximise data reuse for secondary research. New studies are underway, with the University of Vilnius in Lithuania using a dataset from Poland to investigate the genetic basis of overgrowth syndrome.

Testing federated analysis of datasets in South Africa, Mali, and Uganda

The **eLwazi open data science platform** aims to help users find, access, and run analyses on different datasets. The project is part of the **Data Science for Health Discovery and Innovation in Africa** consortium, which harnesses data science for the generation of health benefits from diverse data types. The platform uses multiple GA4GH standards, including the **Data Repository Service (DRS)**, the **Tool Registry Service**, the **Workflow Execution Service**, and the **Authorisation and Authentication Infrastructure**. Datasets are also curated using the **Data Use Ontology** to describe access restrictions. In recent work, the eLwazi team has been using these GA4GH tools and standards to test federated analysis across three on-premise sites South Africa, Mali, and Uganda. The initial pilot implementation used the **GA4GH WES Starter Kit**. There are plans to transition to the use of **WESkit**, developed by the German Cancer Research Center and the Berlin Institute of Health. Expansions to WESkit, including adding a DRS resolver and integrating **Passports** and **Crypt4GH**, are being explored.

Applying GA4GH regulatory and ethics tools to explore epigenomics data

Contributors from the **Regulatory & Ethics Work Stream** and the Driver Project **EpiShare** published a **paper** detailing their approach to protecting patient privacy in the EpiVar browser — a tool that supports exploration of genetic and epigenetic datasets. GA4GH's ***Framework for responsible sharing of genomic and health-related data*** helped guide these methods. The *Framework* lays out four foundational principles: respect for individuals, families, and communities; advancement of research and scientific knowledge; promotion of health, wellbeing, and the fair distribution of benefits; and fostering trust, integrity, and reciprocity. Recognising a need to balance broad access to data while respecting patient and participant privacy, the EpiVar browser employs a registered access policy to foster secure access. Researchers can use the platform to explore aggregated results with all identifiable information filtered out. This approach can help facilitate data exploration and analysis before researchers begin to navigate the data access approval process.

The GA4GH Framework is guided by Article 27 of the 1948 Universal Declaration of Human Rights.

27(1) **"The Right to Science"**

27(2) **"The Right to Recognition"**

Connecting disparate datasets to advance neuroscience research

DNAstack's Omics AI is a cloud-based software suite that enables federated discovery and analysis of genomics and other -omics data. Data platforms built using Omics AI integrate many GA4GH standards, including **Service Info**, **Service Registry**, **Data Connect**, **Beacon**, **Passports**, the **Data Repository Service**, and the **Workflow Execution Service**.

Since 2024, Omics AI has been powering a number of data sharing collaborations:

- **Target ALS Data Portal**: Launched in May 2024, the Target ALS Data Portal contains unique datasets including whole genome sequencing, spatial and bulk transcriptomics, and semi-quantitative histopathology from more than 250 ALS cases. The portal uses Omics AI to facilitate secure data exploration, with the goal of accelerating the discovery of potential treatments and biomarkers for ALS.
- **MSSNG database**: In May 2024, 2,489 fully sequenced genomes were added to the MSSNG database — the world's largest autism whole-genome database. The data was processed and shared using Omics AI, increasing the availability of accessible data for deeper analysis.
- **Aligning Science Across Parkinson's Collaborative Research Network Cloud (ASAP CRN Cloud)**: Launched in June 2024, the ASAP CRN Cloud provides a dataset of human postmortem derived samples from over 156 donors, with more than 600 donor samples expected in future releases. The data is securely stored in Omics AI for data exploration and analysis. Studies conducted on this dataset have generated new insights into the development, detection, and treatment of Parkinson's disease.
- **HiFi Solves**: Launched in 2024, the HiFi Solves network is a collaboration with PacBio and their global customer base to create a realtime network for sharing insights across organisations leveraging long-read sequencing. More than 10 organisations, spanning Asia, Europe, and North America, have already begun networking their long-read data, with more initiatives expected to join in 2025.

40

petabytes

Did you know? The **Data Repository Service** provides the community with nearly 40 petabytes of data from projects such as the NIH Cloud Platform Interoperability effort, NHLBI BioData Catalyst, Human Cell Atlas, the European Genomic Data Infrastructure, and the ICGC ARGO Data Platform.

Applying Beacon: CSIRO's sBeacon and AskBeacon projects

The **Beacon standard** is a data discovery tool that helps researchers find datasets of interest for analysis while preserving patient privacy and data ownership. In order to accommodate both small and large-scale research needs in an efficient and economical manner, Australia's **Commonwealth Scientific and Industrial Research Organisation (CSIRO)** has developed **Serverless Beacon (sBeacon)** in collaboration with GA4GH Driver Projects **Australian Genomics** and **Genomics England**. A team at the University of Melbourne's Center for Cancer Research has implemented sBeacon to share cancer-related data. By employing a cloud-based framework, sBeacon has shown to be efficient, cost-effective, and scalable.

AskBeacon helps users query and analyse data across the global Beacon network using natural language. While the Beacon standard is a powerful tool to find relevant datasets, it requires programming skills and knowledge of Beacon syntax. AskBeacon simplifies this process by using large language models (LLM), allowing users to ask Beacon a question without needing to write code or know the technical details of the Beacon schema.

Improving accuracy of variant interpretation in databases

The **Genome Aggregation Database (gnomAD)** is composed of exome and genome sequences from large-scale sequencing projects around the world. To improve the accuracy of variant interpretation, the GA4GH **Variation Representation Specification (VRS)** has now been integrated into the entire gnomAD database. This will enable precise, computable identifiers for specific variants within a large-scale resource. In addition, the launch of the Federated gnomAD project will allow contributions of diverse datasets from around the globe to become aggregated with gnomAD and inform genomic interpretation.

MaveDB is a community-driven public repository for multiplexed assays of variant effect (MAVEs), experiments that can generate valuable functional evidence for clinical geneticists and researchers. **VRS IDs have now been used to map MaveDB variants to the human reference genome**, enabling seamless data transfer to other resources that recognise these identifiers.

2024 HIGHLIGHTS

Improving interoperability

Interoperability is key to ensuring GA4GH products can be used together seamlessly. In 2024, product teams made progress to ensure that GA4GH standards can interface with each other, as well as with other standards and tools in the genomics and health landscape.

Forming Strategic Partnerships with HDR UK and RDA

To align and improve interoperability with other standards organisations, GA4GH has formed strategic partnerships with **Health Data Research UK (HDR UK)** and the **Research Data Alliance (RDA)**. HDR UK marks GA4GH's first strategic relationship with a health data research organisation. Both groups aim to expand existing open standards work in the wider health data space.

The RDA aims to build the social and technical bridges that enable researchers and innovators to openly share and reuse data across technologies, disciplines, and countries. Through a Memorandum of Understanding, GA4GH and RDA aim to coordinate efforts, with the goal of enabling responsible data sharing across geographies, institutions, and systems.

Joining forces with ISO

GA4GH has been involved with the **International Standards Organization (ISO)** for many years as a liaison to **ISO Technical Committee (TC) 215**. In July 2022, **Phenopackets** became an approved ISO standard. GA4GH is now bringing forth other products to ISO, such as the **Passports standard** — a data model and protocol for securely encoding information about a researcher and their data access permissions. The team presented Passports at the ISO TC 215 Subcommittee 1 Conference to explore the potential for an open Ad Hoc Group.

Making more cloud workflows and tools available with TRS

The **Cloud Work Stream** aims to support researchers in using any workflow or analysis tool on any dataset. The **Tool Registry Service (TRS)** provides a standard mechanism to list, search, and retrieve tools and workflows across multiple registries. Several repositories now support TRS, including Dockstore, which shares a library of more than 4,300 workflows and tools with the wider research community.

Integrating commonly used hospital data models in Beacon

Hospitals house useful data for research studies. However, many of these institutions lack technical resources needed to implement new systems or technologies. To lower the barrier to implementation, the **Discovery Work Stream** is exploring ways to deploy the **Beacon** standard within hospital systems. Conversations have started to integrate existing hospital data models, such as OMOP CDM or cBioPortal, directly with Beacon, thus eliminating the need to transform data. This method would allow for more flexible use of data and complements Beacon's existing interoperability with Fast Healthcare Interoperability Resources (FHIR) standards.

Expanding the volume of computable phenotypic data to learn more about rare diseases

Developed by the **Clinical & Phenotypic Data Capture Work Stream**, the **Phenopackets** standard provides a method to structure clinical and phenotypic information in a human and machine-readable way. Used across a variety of systems and applications, Phenopackets has helped standardise and facilitate the sharing of more clinical data. In 2024, the team developed **Phenopacket-store** — a repository representing the first large-scale collection of Phenopackets from individuals diagnosed with a rare disease condition. Standardising data from 959 publications, Phenopacket-store contains 6,668 Phenopackets representing 475 Mendelian and chromosomal diseases associated with 423 genes and 3,834 unique pathogenic alleles. This data collection aims to support rare disease diagnostics, machine learning analysis, and global computational analysis for precision health applications.

Supporting harmonisation and interoperability of GA4GH products: updates from TASC

The **Technical Alignment Subcommittee (TASC)** supports the harmonisation, interoperability, and technical alignment of GA4GH products. In 2024, the group progressed key issues, including technical recommendations concerning a **standard approach to pagination**, establishing a consistent method for **citing GA4GH standards**, and beginning the development of a process for when products move into maintenance mode and assigning product maintainers. A **new web page** about TASC is now available on the GA4GH website, listing ongoing projects, the scope of TASC, and current contributors.

2024 HIGHLIGHTS

Engaging healthcare

In order to unlock the power of genomic and health data, connecting with clinical communities is vital. In 2024, GA4GH made strides to engage these communities and support clinical adoption of GA4GH products.

Launching the Clinical Genomics Laboratory Community of Interest

GA4GH has formed several Communities of Interest to support the implementation of GA4GH products within specific genomic and health domains. In 2024, GA4GH partnered with ClinGen to launch the **Clinical Genomics Laboratory Community**, which aims to foster collaboration among globally distributed laboratories, develop standards and best practices for clinical genomics, and facilitate knowledge exchange. The community is advancing the pilot of the ACMG/AMP/CAP/ClinGen Sequence Variant Classification v4.0 standards. Future sessions will focus on the development of gene-level guidance for variant interpretation, best practices for variants of unknown significance (VUS), and the use of variant identifiers and Matched Annotation from NCBI and EBI (MANE) select transcripts.

Accelerating paediatric cancer research

Members of the **Genomic Knowledge Standards Work Stream** are building standards and tools to ingest, integrate, and exchange genomic knowledge to address paediatric cancer. Through the **Molecular Characterisation Initiative (MCI)**, 3,723 patients have enrolled and consented to have their genomic data studied. One implementation underway at Nationwide Children's Hospital in the US is capturing curated variant-level information from the MCI clinical interpretation pipeline for downstream dissemination and reuse, implementing the **Variation Representation Specification** and the emerging **Variant Annotation specification**. The related **Categorical Variation Representation Specification** has also been used to aggregate and harmonise content from several public cancer variant knowledgebases including **ClinVar**, **CIViC**, and the **Molecular Oncology Almanac** in support of this effort. Looking forward, the team is actively working across multiple community resources — including **gnomAD**, **MaveDB**, and cohort data from the **NIH AnVIL platform** — to create a federated genomic knowledge ecosystem, in which standardised evidence can be readily generated and shared across resources used in variant interpretation.

Developing a federated variant-level matching platform for rare disease evidence building

The **Federated Variant Level Matching (VLM) project** aims to support a global federated network of genomic knowledge bases to improve diagnosis, treatment, and knowledge of rare diseases. Building off of the successful efforts of the **Matchmaker Exchange**, this project is leveraging the **Beacon standard**, which enables discovery of datasets across a federated network. To date, three nodes have been connected, including Variant Matcher, Franklin, and Geno2MP/MyGene2. In addition, matchmaking has been launched across thousands of users of the *seqr* platform, with a novel gene-disease discovery already made, and with connection to the VLM network in progress.

Regulatory and ethics updates

The **Regulatory & Ethics Work Stream (REWS)** develops community-driven policies, tools, and standards that address regulatory and ethical considerations in genomic research and international data sharing. View 2024 highlights and updates from the Work Stream.

Developing new policy tools and resources for the GA4GH community

Diversity in Datasets shares actionable recommendations to promote a more holistic approach to diversity in genomic research.

Genetic Discrimination definition proposes a nuanced and inclusive definition of genetic discrimination, keeping in mind our evolving social context and scientific advancement.

Model Data Access Agreement Clauses provide template clauses that can be used to draft data access agreements.

Data Visiting Lexicon defines key concepts to establish a common ground for discussing data visiting — a form of data sharing in which analysis is performed on shared data within a provider's computing environment.

Expanding the scope and rebranding the GDPR Forum

Since 2018, the **GDPR and International Health Data Sharing Forum** has published over 40 “GDPR Briefs,” helping to demystify the General Data Protection Regulation (GDPR) and its impact on genomic and health data sharing. In an effort to expand the scope of the forum to include discussions about relevant and important topics in the privacy space, the forum has assumed the new name: Health Data Sharing, Privacy, and Regulatory Forum. Under this new scope, the group will publish regular policy briefs on evolving global laws and regulations and how they impact genomic and health data protection and sharing. The policy briefs published in 2024 can be found below.

When are synthetic health data personal data?

By Elizabeth Redrup Hill and Colin Mitchell

11 April

The European Health Data Space — from approval to national implementation

By Julian Sellner, Adrian Thorogood, and Fruzsina Molnar-Gabor

5 September

Navigating the data sharing implications of national security policies

By Yunhe Xue, Diya Uberoi, and Yann Joly

7 November

Sharing new perspectives through the Public Attitudes for Genomics and Policy Forum

REWS publishes blog posts under the **Public Attitudes for Genomic Policy forum** to share findings that relate to studies around public attitudes about how researchers collect, use, and share genomic and health data.

Genomic data sharing in Singapore

By Marthe Smedinga

2 July

Publishing key insights and resources

REWS published several academic papers in 2024, sharing key insights and resources with the genomics and health community.

- **Exploring the ethical, legal, and social issues regarding the return of secondary pharmacogenomic variants in children with a serious disease or developmental disorder**
- **Developing a proposal for an inclusive working definition of genetic discrimination**
- **Responding to the WHO’s request for comment on draft principles for human genome access, use, and sharing**
- **Defining diverse data and ways to pursue diversity goals in research studies**
- **Identifying key elements of an optimal genetic nondiscrimination policy to inform policy frameworks**

Events

April Connect 2024

GA4GH gathered for the 2024 **April Connect meeting** in Ascona, Switzerland and online from 21 to 24 April. The GA4GH Connect meetings provide an opportunity for contributors to advance the **GA4GH Road Map**, showcase GA4GH standards and policies in action, and gather feedback on product development and community needs. The meeting brought together 103 in-person attendees and 312 virtual attendees for updates from Work Streams and Driver Projects, breakout sessions, and workshops.

415 participants

38 sessions

View meeting Report >

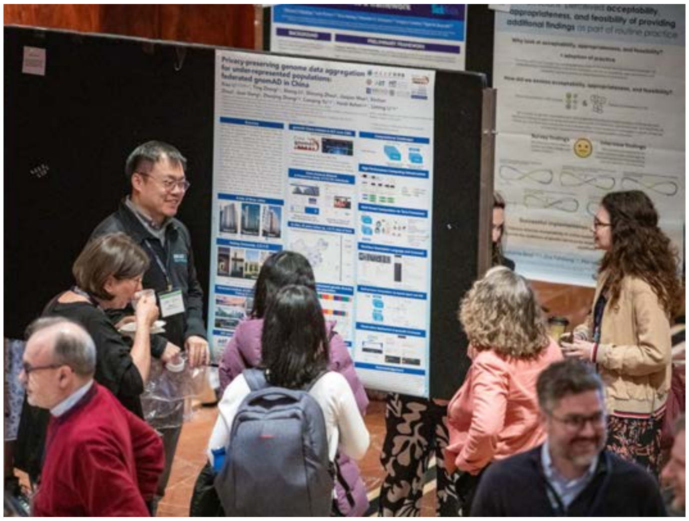
12th Plenary

The global genomics community convened in Melbourne, Australia and online for the **12th Plenary meeting** from 16 to 20 September 2024. Participants engaged in collaborative workshopping, discussed equity and inclusion in the field of genomics, and explored emerging technologies to scale genomic data sharing and advance human health. The **National Initiatives Forum** convened on the last day to share progress and best practices for adding genomics into clinical practice. The conference brought together 294 in-person participants and 417 virtual attendees.

711 participants

50 sessions

View Meeting Report >



Webinars and workshops

2 February

Fireside Chat with Heidi Rehm and Geoff Ginsburg

Heidi Rehm, Chair of GA4GH, and Geoff Ginsburg, Chief Medical and Scientific Officer of the *All of Us* Research Program, discussed progress, highlights, and challenges at the *All of Us* Research Program, as well as methods and approaches to promoting responsible data use.

20 February

Grand challenges in rare diseases webinar

The GA4GH Rare Disease Community hosted a webinar on Rare Disease Day to discuss the current challenges in rare disease research and management and how GA4GH can support advances in these areas. Featured guest speakers included Matt Bolz-Johnson (EURORDIS) and Marc Hanauer (Orphanet).

27 February

National Initiatives Forum workshop — lessons from implementation: sharing variants

The GA4GH National Initiatives Forum (NIF) brings together national programmes focused on advancing genomics at scale. NIF aims to support more accurate data interpretation, diagnosis, and innovative solutions through global cooperation and clinical implementation of genomics.

Sharing information about variants is one of the most powerful tools available to improve the accuracy of genomic data interpretation. But how can this work at scale? NIF convened to share knowledge and lessons learned from variant sharing. Attendees heard from Shariant, Clinical Variant Ark, and the All4One Health Data Ecosystem, and discussed ways to collaborate nationally and internationally.

25 June

Fireside Chat with Kathryn North and Daniel MacArthur

Kathryn North, Vice-Chair of GA4GH and Director of the Murdoch Children's Research Institute, and Daniel MacArthur, Director of the Centre for Population Genomics, based jointly at the Garvan Institute of Medical Research and the Murdoch Children's Research Institute, discussed the implementation of genomics in clinical care.

17 July

Introduction to the GA4GH Variation Representation Specification (VRS) at ASHG

The GA4GH VRS team led a one-hour introductory bioinformatics webinar on the standard, delving into its features and how VRS can be used in the computational exchange of variant annotation and interpretation data.

ASHG
American Society of Human Genetics

4 to 7 August

Cancer Genomics Consortium

GA4GH contributors convened for an "unconference" ahead of the Cancer Genomics Consortium (CGC) 2024 meeting. Hosted by the GA4GH Driver Project Variant Interpretation for Cancer Consortium (VICC), in partnership with CGC (also a GA4GH Organisational member), the Clinical Interpretation of Variants in Cancer, and ClinGen, the meeting aimed to develop community consensus on data standards for the interpretation of cancer variants and to enhance interoperability resources. Learn more in a GA4GH guest blog post by VICC lead Alex Wagner, and view the event meeting minutes.

CANCER
GENOMICS
CONSORTIUM

23 September

BRIDGE Global Workshop

The Neuroscience Community hosted its second meeting at the BRIDGE Global Workshop in Austin, Texas, on 23 September. This event provided an opportunity for theme teams to present project updates, engage with the community, and outline future plans. Access the event meeting summary and recording.

BRiDGE

4 November

Unconference at ASHG

The GA4GH Genomic Knowledge Standards (GKS) Work Stream hosted an unconference at the American Society of Human Genetics (ASHG) meeting on 4 November. The unconference brought together interested individuals to collaboratively develop tools and features using GKS specifications, which aim to standardise how we describe, share, and apply genomic knowledge to improve patient health outcomes. View the event summary.

ASHG
American Society of Human Genetics

7 November

NIF Workshop: Measuring the value of genomics in healthcare

Demonstrating the value of genomic testing is key to informing health technology assessments (HTAs), clinical practice, and policy decisions. Yet standardised outcome measures and frameworks are still in their infancy. The GA4GH National Initiatives Forum (NIF) hosted a workshop on the strategies, tools, and experiences in measuring the value of genomic testing in healthcare. A panel of speakers featuring Robin Hayeems (Canada), Hadley Stevens Smith (US), Ilias Goranitis (Australia), James Buchanan (UK), and Brian Chung and Claudia Chung (Hong Kong) discussed their experiences measuring value in rare disease genomic diagnostics and screening.
Access the meeting summary and recording.

25 November

Fireside Chat: Bartha Knoppers and Gerardo Jiménez Sánchez

GA4GH Inc. Board members Bartha Knoppers, Distinguished James McGill Professor Emerita and Founder of the Centre of Genomics and Policy at McGill University, and Gerardo Jiménez Sánchez, CEO of Genómica Médica and founder director of the Mexican National Institute of Genomic Medicine, engaged in a virtual Fireside Chat. They discussed how the field of genomics has transformed public health in Mexico and Latin America, the importance of diversity in genomic datasets, and building trust with communities.

Thank you for being a part of the GA4GH community

Thank you to all the Work Stream contributors, participants in Communities of Interest and the National Initiatives Forum, members of Driver Projects or Strategic Partner organisations, and to all those who have provided feedback and input on GA4GH products. Our work to advance responsible genomic and health data sharing is made possible through your contributions and partnership. We look forward to another year of inspiring and collaborative work!

Thank you to our funders

We want to extend a thank you to the funders and sponsors who fuel the work of GA4GH. From supporting our core staff to sponsoring our in-person meetings where crucial progress and connections are made, your commitment to GA4GH has made an important impact on our community. We thank you and look forward to your continued partnership.

Core Funders

CIHR IRSC Canadian Institutes of Health Research Instituts de recherche en santé du Canada
GenomeCanada
UKRI Medical Research Council
NIH National Human Genome Research Institute
NIHR | National Institute for Health Research
W wellcome

Host Institutions

BROAD INSTITUTE
EMBL-EBI
McGill UNIVERSITY VICTOR PHILLIP DAHDALEH INSTITUTE OF GENOMIC MEDICINE AT MCGILL UNIVERSITY
OICR Ontario Institute for Cancer Research
wellcome sanger institute

Supporting Funders

abbvie
aws
DNASTACK
illumina
ONTARIO BRAIN INSTITUTE INSTITUT ONTARIEN DU CERVEAU
Roche

Assigned Expert Funders and Employers

NIH National Human Genome Research Institute
UNIVERSITY OF CAPE TOWN IYUNIVESITHI YASEKAPA • UNIVERSITEIT VAN KAAPSTAD

Publications

Baek, Jinyoung, et al. “Investigating the Roles and Responsibilities of Institutional Signing Officials After Data Sharing Policy Reform for Federally Funded Research in the United States: National Survey.” *JMIR Formative Research*, vol. 8, no. 1, Mar. 2024, p. e49822. *formative.jmir.org*, https://doi.org/10.2196/49822.

Danis, Daniel, et al. “A Corpus of GA4GH Phenopackets: Case-Level Phenotyping for Genomic Diagnostics and Discovery.” *medRxiv*, May 2024, p. 2024.05.29.24308104. *PubMed Central*, https://doi.org/10.1101/2024.05.29.24308104.

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