

GA4GH 11th Plenary Meeting Report

GA4GH 11th Plenary brought together organisations and individuals from the genomics and health community for keynotes, talks, and workshops focused on genomic and clinical data sharing issues that pervade diverse industries, disciplines, and communities.

Attendees had the opportunity to learn about GA4GH's technical standards and policy frameworks and tools, which aim to break down the many barriers to data and knowledge sharing in order to advance genomic research.

The GA4GH 12th Plenary will take place in Melbourne, Australia, from (tentatively) 16 to 20 September 2024.

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Wednesday, 20 September

Opening remarks

Slides • Recording

GA4GH Chair Ewan Birney and 11th Plenary Programme Chair David Glazer welcomed participants in San Francisco, USA, and online. Birney announced that he would become Chair Emeritus in January, and that the GA4GH Inc. Board of Directors had elected Heidi Rehm to succeed as Chair.

A decade of GA4GH

Slides • Recording

CEO Peter Goodhand celebrated a milestone — 10 years of collaborative efforts and groundbreaking advancements within the Global Alliance for Genomics and Health (GA4GH). Over the past decade, GA4GH has emerged as a leading force in driving responsible and effective genomic data sharing to benefit human health. This session took attendees on a journey through the evolution of GA4GH, showcasing its pivotal role in shaping the genomics landscape and vision for the future.



GA4GH in action: real-world implementations and success stories

Slides • Recording

Speakers presented compelling success stories that demonstrate how GA4GH work has translated into transformative solutions for genomics and health. The talks covered tangible outcomes and real-world applications of GA4GH products in Europe, Japan, the United States, and internationally. These implementations facilitated cross-disciplinary collaboration, accelerated discovery, and ultimately improved patient outcomes.

- Genomic Data Infrastructure (GDI) — Dylan Spalding and Melissa Konopko
- Japanese biobanks — Soichi Ogishima
- AnVIL — Rachel Liao
- Autism Sharing Initiative: creating a global federated network for autism and neuroscience — Marc Fiume

In the spotlight: GA4GH technical standards and policies supporting broad and responsible use of genomic data

Slides • Recording

This session showcased several GA4GH technical standards and policy tools and frameworks that have seen significant progress since our last Plenary meeting: Beacon, VRS, refget, and an effort focused on paediatric pharmacogenomics. Presentations and an expert panel explained how these products facilitate data sharing, interoperability, and ethical considerations across diverse sectors.

- Beacon — Jordi Rambla
- VRS — Larry Babb
- refget v2.0 and Sequence Collections — Andy Yates and Nathan Sheffield
- Return of secondary findings in paediatric pharmacogenomics — Vasiliki Rahimzadeh

Thursday, 21 September



The Human Pangenome Project: creating a reference that better represents human global genetic diversity (Karen Miga, Human Pangenome Reference Consortium)

Slides • Recording

Karen Miga discussed the need to modernise the human reference genome and introduced the Human Pangenome Project, a newly-announced GA4GH Driver Project. This federated alliance of global genomics partners focuses on the development, release, global acceptance, and use of a new human pangenome resource and pangenomic tool ecosystem. The project aims to broaden studies of genetic variants that impact the effectiveness of medical treatments, influence the risk of developing diseases, and have the potential to determine personalised medical treatments.

Emerging technologies in genomics

Slides • Recording

This session explored the latest breakthroughs and cutting-edge tools that are reshaping the landscape of genomics. Topics included long-read sequences, machine learning, and artificial intelligence more generally.

- New insights into genomic variation from long-read sequencing in the *All of Us* Research Program — Anjene Musick
- Decoding regulatory DNA and genetic variation by interpreting deep learning models of cell context specific molecular profiling experiments — Anshul Kundaje
- Explainable AI: where we are and how to move forward for biomedicine — Su-In Lee

Fostering collaboration and innovation: exploring industry engagement

Slides • Recording

The panel shared success stories, case studies, and innovative approaches that highlighted the impact of cross-sector partnerships. Speakers explored the evolving landscape of industry engagement in genomics, and the mutual benefits and challenges faced by academia and industry.



- Powering the local ecosystem through the National Precision Medicine programme — Seow Shih Wee
- Enabling innovation through a European bioinformatics research infrastructure — Melissa Konopko
- International genomic data sharing by health technologies industries: points to consider — Shane Chase and Bartha Knoppers

Keynote session: genomic surveillance and characterisation of microbial threats facilitates early detection and containment of disease outbreaks in West Africa (Christian Happi, Redeemer's University)

Slides • Q&A recording *(due to a technical problem, unfortunately the talk was not recorded)*
Happi discussed preempting pandemics and containing disease outbreak in West Africa. He introduced the African Centre of Excellence for Genomics of Infectious Diseases, based in Nigeria, and presented some of the successes of affiliated researchers. For example, metagenomic surveillance helped uncover outbreaks of yellow fever and other diseases. The centre also builds capacity in genomics, both in careers and facilities.

Towards inclusive research: diverse datasets and cohort representation

Slides • Recording
This session explored the significance of diversity in genomic datasets and the pivotal role of comprehensive cohort representation in shaping research outcomes and healthcare equity. The panel of experts shared insights, best practices, and strategies that promote inclusivity in research.

- REWS considerations for how genetic and genomic researchers should approach thinking about diversity in data — Maili Raven-Adams
- Toward inclusive and equitable biomedical research — Alham Saadat
- Putting epistemic justice as inclusivity at the heart of genomic research — Calvin Ho
- The Silent Genomes Project: addressing genomic inequity for, and with, Indigenous populations of Canada — Laura Arbour



Friday, 22 September

Keynote session: Patrick Sullivan (Team Finn Foundation)

Slides • Recording

Patient advocate Patrick Sullivan discussed how doctors must go out of their way to find personalised solutions for patients with rare disease and/or cancer. He challenged attendees to make personalised care a feature, not a bug, in the system. Sullivan gave examples from his advocacy work and his own family patient story, and advice for researchers who are communicating or engaging with patients and their advocates.

Bridging the gap: integrating genomics into clinical practice for personalised medicine

Slides • Recording

Through presentations and interactive discussion, this session explored the challenges and opportunities faced when integrating genomics into medical workflows. Presenters explained how advancements are driving precision medicine to new heights.

- Bridging the gap: integrating genomics into clinical practice for personalised medicine: the Australian experience — Kathryn North
- Estonian Biobank — Andres Metspalu
- Genomics and clinical practice: ARPA-H Biomedical Data Fabric Toolbox — Alastair Thomson

Bridging the gap: overcoming challenges in translating from research to clinical care

Slides • Recording

In the second part of "Bridging the Gap," new panellists turned to hurdles encountered when bridging the gap between scientific discovery and clinical implementation. The speakers delved into the multifaceted challenges of translating research insights into practical applications within clinical settings. They explained the strategies, innovations, and ethical



considerations that guide this process.

- Bridging the gap from specialty care to preventive care — Nicolas Encina
- From theory to practice: clinical implementation of translational research — Bob Freimuth
- Patient associations' role in translating from research to clinical care: nothing about us without us — Marcela Ciccioli

Keynote session: Susan Gregurick (NIH Office of Data Science Strategy)

Slides • Recording

Gregurick discussed catalysing data science in research through collaborations. She gave many examples of NIH collaborations, including efforts to connect European researchers with access to the *All of Us* Research Program (a GA4GH Driver Project), and work by the NIH Cloud Platform Interoperability effort (a newly-announced GA4GH Driver Project) to incorporate standards from GA4GH, HL7/FHIR, and elsewhere.

GA4GH leadership: past, present, and future

Slides • Recording

This dialogue between two eminent thought leaders in genomics covered the current landscape, challenges, and potential future directions of genomics research, data sharing, and clinical application. Ewan Birney and Heidi Rehm shared their insights into the origins of GA4GH, the impactful initiatives that have shaped its present, and their collective vision for its future impact.

Closing remarks

Slides • Recording

CEO Peter Goodhand thanked speakers, funders, attendees, outgoing Chair Ewan Birney, and GA4GH contributors. He announced the next GA4GH Connect meeting (in Ascona, Switzerland, from 21 to 24 April 2024) and GA4GH 12th Plenary meeting (in Melbourne,



Australia, tentatively from 16 to 20 September).