GA4GH 8th Plenary Meeting Report

The Global Alliance for Genomics and Health (GA4GH) held its first ever virtual plenary from September 29 to 30, 2020. The 8th Plenary meeting brought together over 770 participants across more than 50 countries for talks and discussions around the importance of diversity in genomic datasets, harmonizing ethical and technical solutions to harness the power of diverse data, and real-world implementations of GA4GH standards. The meeting offered English closed captioning and live interpretation in Spanish, French, German, and Japanese.

At the meeting, GA4GH announced the release of its 2020-2021 Roadmap. This version of the Roadmap consists of two parts: a strategic roadmap with three key community priorities for the coming years, and a product roadmap, which outlines 43 deliverables that address needs in the areas of data use and researcher identities, variation representation and annotation, federated analysis, privacy and security, regulatory and ethics, discovery, clinical and phenotypic data capture, and large scale genomics.

GA4GH also showcased the 2020 Connection Demos—real-world implementations of GA4GH standards working together across institutions. These demos show how researchers can discover controlled access data hosted in one computational environment and analyze them in another, all following best practices in privacy and security.

The GA4GH 8th Plenary Meeting was covered by *Bio-IT World*, *Cambridge Network*, *ClinicalOmics*, DNAStack, EMBL-EBI, ELIXIR, *(GIGA)Blog*, and Terra.

Overview

- Track 1A: The Importance of Diversity in Genomic Datasets
    - Opening Remarks
    - Keynote: “Diversity in Genome Science: A Scientific & Social Justice Imperative” (Charles Rotimi, NIH)
    - Panel: Information Disparities and Implications for Clinical Genetics (Alice Popejoy, Stanford University)
    - Panel The Missing Diversity in Human Genetic Studies, and its Impact on Health (Giorgio Sirugo, University of Pennsylvania)
    - Panel: Diversity in genomics will fail unless we de-biologize race (Consuelo Wilkins, Vanderbilt University)
    - Joint Question & Answer Session
    - Closing Remarks
- Track 1B: Uniting Ethical and Technical Solutions to Harness the Power of Data
    - Opening Remarks

    - Keynote: Building an Intentional Community for Standards Development (Laura Paglione, Spherical Cow Group)
    - Panel: Data Science and the Digital Divide: Public Health Challenges in LMICs (Amel Ghouila, Gates Foundation)
    - Panel: Building Trust: A Pre-Requisite for Global Data-Sharing (Keymanthri Moodley, Stellenbosch University)
    - Panel: Reimagining Data (Jasmine McNealy, University of Florida)
    - Joint Question & Answer Session
    - Closing Remarks
- Track 2A: Real-World Implementations (Europe, Africa, and the Americas)
    - GA4GH Connect: The Story So Far
    - Question & Answer Session with Work Stream Leads
    - Breakout Session: DURI in the Real World
    - Breakout Session: GA4GH Connection 2020
    - Breakout Session: Crypt4GH in the Real World
    - Breakout Session: Regulatory & Ethics Work Stream
    - Real-World Implementations: CanDIG (Shaikh Farhan Rashid, University Health Network)
    - Real-World Implementations: BIPMed (Benilton Carvalho, University of Campinas)
    - Real World Implementations: Genomics England (Augusto Rendon, Genomics England)
    - Joint Question & Answer Session with CanDIG & BIPMed
- Track 2B: Real World Implementations (GEM Japan, Australian Genomics)
    - GA4GH Connect: The Story So Far
    - Q&A with Work Stream Leads
    - Breakout Session: DURI in the Real World
    - Breakout Session: GA4GH Connection 2020
    - Breakout Session: Crypt4GH in the Real World
    - Breakout Session: Regulatory & Ethics Work Stream
    - Opening Remarks: Genomics in Health Implementation Forum (Kathryn North, Australian Genomics)
    - Real-World Implementations: GEM Japan (Takako Takai and Soichi Ogishima, GEM Japan)
    - Real-World Implementations: Australian Genomics (Tiffany Boughtwood and Oliver Hofmann, Australian Genomics)
    - Joint Question & Answer Session with GEM Japan & Australian Genomics

Track 1A: The Importance of Diversity in Genomic Datasets

Opening Remarks

Ewan Birney (EMBL-EBI), Heidi Rehm (MGH/Broad Institute)
Presentation Video | Slides | Animation | Message from the GA4GH Funders

Birney gave an overview of the meeting, highlighting the three key themes: the importance of diversity in genomic datasets, uniting ethical and technical solutions to harness the power of data, and Real-World Implementations of GA4GH Standards.

Rehm then announced the release of the GA4GH 2020-2021 Roadmap. Part I: The Strategic Roadmap presented outcomes of the 2020 Gap Analysis, which identified three community imperatives for GA4GH to focus on in 2021 and beyond: 1) Interoperability & Alignment, 2) Implementation Support, and 3) Engaging with Healthcare & Clinical Standards. Part II: The Product Roadmap presents 43 deliverables to be developed or expanded across the eight GA4GH Work Streams over the next two years.

Rehm dove into efforts underway on the first strategic focus area, with the announcement of the GA4GH 2020 Connection Demos—examples of interoperability which showcase GA4GH standards working together to power scientific research. A short animation provided a conceptual perspective of the future that these demos are aiming to enable.

Opening remarks concluded with a short Message from the GA4GH Funders.

Keynote: Diversity in Genome Science: A Scientific & Social Justice Imperative

Charles Rotimi (National Institutes of Health)
Presentation Video | Slides

Rotimi discussed the importance of diversity in genomics through a scientific and social justice lens. From a scientific standpoint, diversity in genomics is key to understanding global distribution of genetic variation, which can inform human migrational history, biology, susceptibility to disease, and variable drug response. Rotimi also discussed how diversity in genomics is a social justice imperative to prevent health and economic inequalities and to ensure that we can improve effective clinical care for all people.

Panel Presentation: Information Disparities and Implications for Clinical Genetics

Alice Popejoy (Stanford University)

Presentation Video | Slides

Popejoy discussed the ways in which information disparities, biases in research, and lack of guidance on the use of diversity measures have impacted clinical genetics. Popejoy discussed the need to increase diversity in genomics research and reference databases, quantify "uncertainty" in clinical genetics, and engage stakeholders across multiple disciplines and continents to develop guidelines for diversity measures in clinical genetics.

Panel Presentation: The Missing Diversity in Human Genetic Studies, and its Impact on Health

Giorgio Sirugo (University of Pennsylvania)
Presentation Video | Slides

Sirugo discussed the ways in which a lack of diversity in human genomics research can impact human health. These include exacerbating health inequalities; incomplete or mistaken translation of genetic research into clinical practice or public health policy; inaccurate assessments arising from estimates of genetic risk based on European-based studies; and inaccurate assessment of pathogenic variants in clinical genomics studies.

Panel Presentation: Diversity in genomics will fail unless we de-biologize race

Consuelo Wilkins (Vanderbilt University Medical Center)
Presentation Video

Wilkins discussed several key topics that we must consider in order to de-biologize race in the context of diversity in genomics. Wilkins emphasized the importance of determining what kind of diversity is greatly needed when we talk about diversity in genomics; how to disentangle race and ancestry; and including standards related to the inclusion of race, ethnicity, and social determinants of health within genomic research.

Track 1A Joint Q&A

Q&A Video

Charles Rotimi (National Institutes of Health), Alice Popejoy (Stanford University), Giorgio Sirugo (University of Pennsylvania), and Consuelo Wilkins (Vanderbilt University Medical Center) answered questions from the audience during the Q&A session.

Track 1A Closing Remarks

Meg Doerr (Sage Bionetworks)
Presentation Video

Doerr, Chair of the 8th Plenary Programme Committee Chair, gave closing remarks for the Track 1A session.

Track 1B: Uniting Ethical and Technical Solutions to Harness the Power of Data

Opening Remarks

Link to Summary

Keynote: Building an Intentional Community for Standards Development

Laura Paglione (Spherical Cow Group)
Presentation Video

Paglione discussed the importance of building intentional communities to develop technical and policy solutions that will work in the real world, across multiple contexts and environments. Paglione shared examples of this principle in action, including through her work as co-lead of the GA4GH Equity, Diversity, and Inclusion (EDI) group.

Panel: Data Science and the Digital Divide: Public Health Challenges in LMICs

Amel Ghouila (Exaptive, Inc. for the Bill and Melinda Gates Foundation)
Presentation Video | Slides

Amel discussed the potential impacts and challenges of harnessing the power of data science to inform public health in low to middle-income countries (LMICs). While data science methods and AI hold a lot of promise in delivering healthcare—from predicting susceptibility of disease to informing healthcare policy—implementing these approaches requires a thorough assessment of context and understanding of the digital divide in order to develop inclusive, multidisciplinary strategies that would avoid potential harm.

Panel: Building Trust: A Pre-Requisite for Global Data-Sharing

Keymanthri Moodley (Stellenbosch University)
Presentation Video

Moodley discussed the importance of building trust as a prerequisite for global data sharing. Placing this key concept in the context of the COVID-19 pandemic, Moodley discussed how the pandemic has brought to the forefront public health ethics and the need to place public interest and common good at the center of genomics research in order to protect the privacy and consents of data donors.

Panel: Reimagining Data

Jasmine McNealy (University of Florida)
Presentation Video

McNealy discussed the importance of reimagining data as infrastructure—and by extension, people as infrastructure. Due to the interconnected relationship between data and people, McNealy discussed the critical need for laws, policies, regulations, and guidelines to ensure that people are protected from the harms and abuses of data.

Q&A with Track 1B

Q&A Video

Laura Paglione (Spherical Cow Group), Amel Ghouila (Exaptive, Inc. for the Bill and Melinda Gates Foundation), and Jasmine McNealy (University of Florida) answered questions from the audience during the Q&A session.

Closing Remarks

Meg Doerr (Sage Bionetworks)
Presentation Video

Doerr, Chair of the 8th Plenary Programme Committee, gave closing remarks for the Track 1B session.

Track 2A: Real-World Implementations

GA4GH Connect: The Story So Far

Presentation Video

The GA4GH Work Stream Managers discussed how GA4GH standards can help enable a global learning health system.

Learn about specific updates from the GA4GH Work Streams:

Clinical & Phenotypic Data Capture | Cloud | Data Security | Data Use & Researcher Identities | Discovery | Genomic Knowledge Standards | Large Scale Genomics | Regulatory & Ethics

Q&A with Work Stream Leads

Q&A Video

Breakout Session: DURI in the Real World

Presentation Video

In this breakout session, the Data Use & Researcher Identities (DURI) Work Stream put the spotlight on the global implementations of two of their standards: the Data Use Ontology (DUO) and GA4GH Passports. The session featured talks from the NIH, EGA, Wellcome Sanger Institute, GEM Japan, H3Africa, and Signatu, who shared their successes and challenges with implementing DUO and Passports.

Breakout Session: GA4GH Connections Demos

Presentation Video

In this breakout session, members of the Federated Analysis Systems Project (FASP) demonstrated GA4GH standards working together in real-world scenarios: 1) the “horizontal” demo showed multiple vendors executing analyses on the 1000 genomes dataset, demonstrating reproducibility of results; 2) the “vertical demo” showed one vendor utilizing multiple GA4GH standards in a full end-to-end use case; and 3) the “cross-platform” demo laid out a framework for accessing and working with multiple datasets across multiple platforms.

Breakout Session: Crypt4GH in the Real World

Presentation Video

In this breakout session, the Crypt4GH team shared current challenges in the secure sharing of genomic data files, along with the benefits of using Crypt4GH—a GA4GH standard that allows genomic data files to always remain encrypted, whether in storage or in transit. The team also showcased two demos of Crypt4GH in action: 1) distributing EGA files to the Barcelona Supercomputing Center; and 2) running a CWL Analysis Pipeline.

Breakout Session: Regulatory & Ethics Work Stream

Presentation Video

This breakout session provided an overview of the Regulatory & Ethics Work Stream (REWS), including newly-approved policies, initiatives such as the GDPR Briefs, and updates on current

and future projects. The session dove deeper into specific areas of focus, including the Your DNA Your Say global survey, the GA4GH Consent Toolkit, and the coming Participant Patient Public Engagement (PPPE) Policy. There was also an overview of the REWS' perspective article, *Responsible Data Sharing to Respond to the COVID-19 Pandemic: Ethical and Legal Considerations*.

Real-World Implementations: Canadian Distributed Infrastructure for Genomics (CanDIG)

Shaikh Farhan Rashid (University Health Network)
Presentation Video

Rashid shared CanDIG's experience implementing GA4GH standards, beginning with an overview of health research data in Canada, followed by CanDIG's federation model. Next, Rashid dove into the GA4GH standards that CanDIG has implemented, including the Beacon API, htsget API, RNAget API, Workflow Execution Service API, Data Repository Service API, Data Use Ontology, Phenopackets, Passports, and Authentication and Authorization Infrastructure—all of which build on GA4GH's *Framework for Responsible Sharing of Genomic and Health-Related Data* and data security best practices.

Real-World Implementations: Brazilian Initiative on Precision Medicine (BIPMed)

Benilton Carvalho (University of Campinas)
Presentation Video

Carvalho shared BIPMed's experience implementing GA4GH standards, beginning with an overview of BIPMed, whose databases served as the first product released to fulfill the increasing need for publicly available genetic and genomic information on the Brazilian population. He discussed a variety of tools and technologies for serving genotypic and phenotypic data, including the Beacon API, and new custom tools, such as the BIPMed Variant Explorer (BraVE), which supports authenticated access and provides a user-friendly interface for users to query, manage, and share variant data.

Real-World Implementations: Genomics England

Augusto Rendon (Genomics England)
Presentation Video

Rendon shared Genomics England's experience in implementing GA4GH standards in order to improve genomic healthcare and accelerate research. Rendon described Genomics England's healthcare loop, where they are actively using the CRAM file format for genomic data compression and htsget API, and are working towards implementing the Task Execution

Service (TES) API, the Workflow Execution Service (WES) API, the Data Repository Service (DRS) API, and Variation Representation Specification.

Real-World Implementations Q&A

Q&A Video

Shaikh Farhan Rashid (CanDIG) and Benilton Carvalho (University of Campinas) answered questions from the audience during the Q&A session.

Track 2B: Real-World Implementations

GA4GH Connect: The Story So Far

Presentation Video

The GA4GH Work Stream Managers discussed how GA4GH standards can help enable a global learning health system.

Learn about specific updates from the GA4GH Work Streams:
Clinical & Phenotypic Data Capture | Cloud | Data Security | Data Use & Researcher Identities | Discovery | Genomic Knowledge Standards | Large Scale Genomics | Regulatory & Ethics

Q&A with Work Stream Leads

Q&A Video

Breakout Sessions

Opening Remarks

Kathryn North (Australian Genomics)
Presentation Video

North gave opening remarks, highlighting the Genomics in Health Implementation Forum (GHIF) as a forum for collective learning and sharing best practices for implementing genomics in health and clinical care.

Real-World Implementations: GEM Japan

Takako Takai (Japan Agency for Medical Research and Development) & Soichi Ogishima (Tohoku Medical Megabank Organization, Tohoku University)
Presentation Video

Takai and Ogishima shared their experiences in implementing GA4GH standards within GEM Japan. Takai began with an overview of GEM Japan's involvement in the GA4GH Work Streams, and shared a major achievement from this year—the release of the GEM Japan Whole Genome Aggregation, the first-of-its-kind open-access variant frequency panel of 7,609 Japanese whole genome sequences. Ogishima then dove into GA4GH standards that have been implemented within the organization, including the Data Use Ontology, Machine-Readable Consent Guidance, Phenopackets, and the Variation Representation Specification.

Real-World Implementations: Australian Genomics

Oliver Hofmann (University of Melbourne Center for Cancer Research)
Presentation Video

Hofmann shared his experiences in implementing GA4GH Standards within Australian Genomics (AGHA). He described the tools and standards used at each step of the AGHA data flow model, including the Data Use Ontology, Crypt4GH, Variation Representation Specification, and Beacon API. Hofmann concluded with a forecast of the future AGHA data flow model, which will include the Data Repository Service API, the htsget API, and a user authentication system.

Real-World Implementations Q&A

Q&A Video

Takako Takai (Japan Agency for Medical Research and Development), Tiffany Boughtwood (Australian Genomics), Oliver Hofmann (University of Melbourne Center for Cancer Research), and Soichi Ogishima (Tohoku Medical Megabank Organization, Tohoku University) answered questions from the audience during the Q&A session.