GA4GH 5th Plenary Meeting Report

The GA4GH 5th Plenary Meeting brought together more than 350 individuals for the launch of GA4GH Connect, a new phase of the organization focused on connecting GA4GH development work to the immediate data sharing needs of the genomics and health communities. An additional 100+ individuals joined the meeting by live stream on Day 3 (see full Day 3 video here). Days 1 and 2 included internal working meetings while Day 3 was focused on launching GA4GH Connect to the broader community. It included a launch event with an international group of speakers discussing the importance of GA4GH for enabling genomic and health-related data sharing as well as a discussion with former and current US NIH directors, Harold Varmus and Francis Collins, on the importance of international open science. GA4GH Connect and the GA4GH 5th Plenary Meeting were covered by *Bio-IT World*, *GenomeWeb*, and *Healthcare Informatics*.

October 15

On Day 1 of the GA4GH 5th Plenary Meeting, Work Streams convened for the first time in person to discuss deliverables and timelines for the 2018 Strategic Roadmap. Within each full-day meeting, a one hour session devoted to Driver Projects brought in the Champions from each real-world genomic data initiative to specify their immediate standards needs in each domain. The full 2018 Strategic Roadmap will be released in January 2018.

October 16

On Day 2 of the meeting, Driver Project Champions presented their needs for genomic data sharing standards and how they plan to work with GA4GH to identify, develop, and pilot those standards. These overviews were followed by presentations from Work Stream Leads on the team's goals, planned deliverables and milestones, and plans for working with Driver Projects to produce workable genomic data sharing standards. Side meetings from the two Foundational Work Streams focused on activating the human right for all to benefit from advances in science, harmonizing data governance across traditional boundaries, and issues of breach response planning in a federated genomic data sharing ecosystem.

Click a Driver Project to download presentation slides:

- All of Us
- Australian Genomics
- BRCA Challenge
- CanDIG
- ClinGen

Click a Work Stream to download presentation slides:

- Clinical & Phenotypic Data Capture
- Data Use & Researcher Identities (DURI)
- Data Security
- Discovery

- ELIXIR Beacon
- ENA/EGA/EVA
- Genomics England
- ICGC-ARGO
- Matchmaker Exchange
- Monarch Initiative
- NCI Genomic Data Commons
- Variant Interpretation for Cancer Consortium
- Cloud
- Genomic Knowledge Standards
- Large Scale Genomics
- Regulatory & Ethics
- Partner Engagement

October 17

GA4GH STRATEGIC PLAN LAUNCH

Introduction to GA4GH Connect

Video (19:00) | Slides

Ewan Birney (EMBL-EBI, UK), Peter Goodhand (Ontario Institute for Cancer Research, Canada)

GA4GH Steering Committee Chair Ewan Birney and Executive Director Peter Goodhand introduce the new 5 year strategic plan, GA4GH Connect. Birney speaks on the growing health care contribution to the world's compendium of genome sequencing data and the resulting opportunity, responsibility, and challenge that these data present to the research and medical communities. Goodhand describes the organizational changes associated with the new launch, including the introduction of 15 new GA4GH Driver Projects, which will work directly with 8 new Work Streams to develop the genomic data sharing standards and frameworks necessary to share data in the real world and the near-term.

Leadership Video

Video (02:44)

Dame Sally Davies (Chief Medical Officer, UK), Jeremy Farrar (Wellcome Trust, UK), Eric Lander (Broad Institute of MIT and Harvard, USA), Sharon Terry (Genetic Alliance, USA)

Four world leaders from the genomics and health communities speak on the importance of genomic data sharing for the advancement of human health and medicine as well as the role that GA4GH plays in enabling responsible sharing of genomic and health related data.

Validators & Champions

Video (28:08) | Slides

Heidi Rehm (Broad Institute of MIT and Harvard), Bartha Knoppers (McGill University, Canada), Kathryn North (Australian Genomics, Australia), Sumit Jamuar (Global Gene Corp, India), Iscia Lopes-Cendes (UniCamp, Brazil), Nicola Mulder (H3Africa, South

Africa), Makoto Suematsu (Japan Agency for Medical Research and Development, Japan), Marc Le Page (Genome Canada, Canada), David Glazer (Verily Life Sciences, USA), Kym Boycott (Children's Hospital of Eastern Ontario, Canada)
A group of nine international leaders in the genomics and health communities deliver 3-minute key messages on why data sharing is necessary to advance human health in the era of genomic medicine. Topics include activating the human right to benefit from the advances of science, international collaboration, the importance of large, genetically diverse datasets for globally accessible precision medicine, and formalizing the historical culture of data sharing in the rare disease community through the development of infrastructure and standards.

Patient Video

Video (05:30)
This video highlights the story of Justin Vachon, a young man born in 1997 with a rare neurological disorder. His family diagnostic odyssey was brought to a close 19 years later when they shared his genetic data through Matchmaker Exchange—a GA4GH Connect Driver Project and previous GA4GH Demonstration Project.

GA4GH Connect Q&A

Video (11:09)
Ewan Birney (EMBL-EBI)
Birney leads a community question and answer session on GA4GH Connect, a 5 year strategic plan dedicated to enabling real-world uptake of GA4GH standards and frameworks by 2022.

INTERNATIONAL OPEN SCIENCE

Video (53:01)
Ewan Birney (EMBL-EBI), Francis Collins (NIH), Harold Varmus (Weill Cornell Medical College)
Birney hosts a "fireside chat" on the subject of international open science with Collins (US National Institutes of Health (NIH) Director) and Varmus (former NIH Director and Nobel Laureate). The discussion covers issues of controlled versus open access to data and associated privacy concerns, the importance of international collaboration in developing data sharing infrastructure, encouraging individuals across disparate communities to work together toward a common goal, and promoting uptake of GA4GH standards and frameworks.

ENABLING TECHNOLOGIES AND PLATFORMS

Opening Remarks

Video (02:03)
David Haussler (UCSC)

Haussler delivers opening remarks to the “Enabling Technologies and Platforms” session, noting that deployment of common technology for data sharing within GA4GH Driver Projects holds the opportunity to change the biomedical research ecosystem.

Europe, ELIXIR, and GA4GH

Video (16:00) | Slides

Niklas Blomberg (ELIXIR)

ELIXIR is the European infrastructure for life sciences data. It has 21 Members, which each host an ELIXIR Node, and aims to drive responsible data sharing across Europe by connecting the continent’s national investments in life sciences data management. Many of the Nodes are themselves collaborations, which allows ELIXIR to reach nearly every life science researcher across Europe. Blomberg discusses challenges to enabling international data sharing, including geographic distribution, access to identifiable human data, and meeting the open data mandate of many nations around the globe. He discusses ways that ELIXIR is meeting those challenges and how it is working with GA4GH to establish data sharing standards, particularly in the area of data discovery through its collaboration with the Beacon Project.

The Pan-Cancer Analysis of Whole Genomes Project

Jan Korbel (EMBL Heidelberg)

Content embargoed until publication.

Privacy-Preserving Linkage of Genomic and Clinical Data Sets

Video (13:04) | Slides

Dixie Baker (Martin, Blanck & Associates)

The Privacy-Preserving Record Linkage (PPRL) task team launched in 2016 as a collaboration between GA4GH and the International Rare Diseases Research Consortium (IRDiRC) to address ethical questions, regulatory requirements, and technical approaches around PPRL. The primary motivation for the effort was the belief that having the ability to link data in a privacy preserving way would improve data quality for research and healthcare, while at the same time contributing to privacy risk—therefore requiring careful assessment of technical approaches that will enable global record linkage within the bounds of applicable jurisdictional law. The task team concluded that every technique currently available makes some use of individually identifiable data and that no technical solution alone can both enable data sharing and assure individual privacy—strong privacy policy, security protections, and governance are essential. The team concluded that the European Patient Identity management approach (EUPID) is most closely aligned with the needs of the GA4GH community.

Enabling Technologies and Platforms Panel Discussion

Video (14:18)

David Haussler (UCSC), Niklas Blomberg (ELIXIR), Jan Korbel (EMBL Heidelberg), Dixie Baker (Martin, Blanck & Associates).
Haussler leads a Q&A session following the session on “Enabling Technologies and Platforms,” covering the application of artificial intelligence and machine learning to large genomic datasets as they become available more broadly as well as approaches for protecting data in the cloud, such as homomorphic encryption and federation.

GOVERNING AND USING HEALTH DATA

Opening Remarks

Video (00:38)
Bartha Knoppers (McGill)
Knoppers opens the session on Governing and Using Health Data.

OECD Recommendation on Health Data Governance 2017

Video (14:36) | Slides
Jennifer Stoddart (McGill)
In 1981, the Organisation for Economic Co-operation and Development (OECD) published the world’s first set of privacy and security guidelines for international data sharing. In 2011, the OECD began a series of studies on health care and health information. As the former Privacy Commissioner of Canada, Stoddart discusses the persistent barriers to data sharing and efforts to reconcile the 1981 guidelines with the urgent need to share health data more widely for the economic and medical betterment of OECD citizens. As a result of this work, the OECD released the Recommendation on Health Data Governance in 2017 with two important themes: (1) that all OECD countries establish a national health data governance framework, and (2) that these frameworks harmonize with other OECD countries. All OECD health ministers adopted the recommendation. Stoddart notes that this is a powerful policy instrument through which GA4GH can pursue its mission to enable a cultural shift toward data sharing which respects security and privacy.

Facilitating Data Sharing through Data Protection: Towards a Code of Conduct

Video (15:29) | Slides
David Townend (Maastricht University)
Townend argues that the requirements of the European Union General Data Protection Regulation can and should be interpreted to facilitate research rather than to create barriers to it. He discusses the potential for GA4GH to participate in developing an international Code of Conduct on processing genomic and health data for research under the GDPR. He argues that the Code of Conduct should (1) include a definition of informed consent that allows for broad foreseeable uses of health data, (2) facilitate research as a compatible purpose for the secondary (unforeseen) processing of already gathered personal data, and (3) establish

guidance on producing effective firewalls between de-identified and identifiable health data for research. Townend argues that a Code of Conduct for health and life science research under the EU GDPR must be constructed so as to ensure and contribute to an international governance structure in this area of international research.

Your DNA, Your Say

Video (19:07) | Slides

Anna Middleton (Wellcome Genome Campus)

Your DNA, Your Say is a project of the GA4GH Participant Values Task Team, a subgroup of the Regulatory and Ethics Work Stream. It developed a survey that probes public attitudes on sharing genomic information across 14 languages including Japanese, Arabic and Icelandic, and covers every continent except Antarctica. Because the majority of the public are unfamiliar even with the term "genomics," the survey uses a series of videos that break down the issues into non-threatening, easy to understand messages. Middleton presents early results from the English speaking survey. For instance, those most in favor of sharing their data are those most familiar with the concept and those most familiar are patients. They also found that while the public can well articulate their fears about data sharing, those fears do not impact their willingness to do so and neither do socio-demographic differences among the respondents. The primary fear was associated with messages propagated by popular culture, suggesting that this may be itself a springboard for education.

Governing and Using Health Data Panel Discussion

Video (10:10)

Bartha Knoppers (McGill), Jennifer Stoddart (McGill), David Townend (Maastricht University), Anna Middleton (Wellcome Genome Campus)

Knoppers leads a panel discussion following the "Governing and Using Health Data" session, fielding questions from the audience and covering topics such as the cultural aspects of international health data governance and public perspectives on data sharing.

GA4GH SUPPORTING GENOMICS AND HEALTH

Opening Remarks

Video (01:57)

Eric Green (NIH)

Green opens the session on GA4GH Supporting Genomics and Health, including discussions of the GA4GH Connect Work Streams and 2017 Driver Projects.

Creating Technical Tools for Sharing Genomic Data

Video (14:58) | Slides

David Glazer (Verily Life Sciences)

Glazer discusses how the collective approaches of the GA4GH Technical Work Streams will come together to enable data interoperability in order to achieve “big health.” A set of standardized methods for accessing and analysing standardized data will make it possible for different researchers working in different computing environments around the globe to create knowledge that is portable and interoperable. All of this will need to be developed in a secure, privacy protected manner. Glazer notes that while genomic data initiatives around the globe are evolving toward a convergent architectural approach, those architectures are not yet interoperable with one another. He outlines the idea of a “data biosphere for biomedical research” that (1) is standards based, (2) allows for the maximum amount of open sharing, (3) is community driven, and (4) is modular and portable. He points out that he many millions of genomic data points soon to be available will be sufficient incentive to attract the best computer scientists to work on these challenges.

Curating and Discovering the Clinical Genome: GA4GH Efforts to Engage and Support

Video (17:09) | Slides

Heidi Rehm (Broad Institute of MIT and Harvard)

Rehm highlights GA4GH Driver Projects that represent the clinical genomics community and ways that the Work Streams will be supporting them. She payed particular attention to ClinGen, the Variant Interpretation for Cancer Consortium (VICC), the BRCA Challenge, ELIXIR Beacons, Matchmaker Exchange, and the Monarch Initiative, which are all focused on curating and organizing knowledge and evidence. The work of the Clinical & Phenotypic Data Capture (CPDC) Work Stream, the Genomic Knowledge Standards (GKS) Work Stream, and the Discovery Work Stream will be critical for these groups to share data. CPDC will establish standard ontologies and information models to describe clinical phenotypes; GKS will develop and adapt standards to exchange genomic knowledge with a focus on those that integrate genomic and translational knowledge with clinical practice; Discovery will continue to develop APIs to facilitate genomic information exchange, with its first project under GA4GH Connect to focus on data sharing for ClinVar submissions.

Implementing genomics into clinical practice

Video (19:38) | Slides

Kathryn North (Australian Genomics)

North discusses the challenge of implementing genomic data sharing standards and tools in clinical practice across the globe. Taking a country-level approach is the most efficient way to create a global data sharing network, North believes, since each health system is unique. To this end, GA4GH is convening a consortium of the existing National Initiatives, which has so far brought together nearly 20 countries with nationwide clinical genomics programs to discuss challenges and advances, to learn from one another and share resources. For example, in Australia, the implementation of genomic medicine has focused on the “federation” of existing state-based services, while leveraging national infrastructure and actively engaging both state

and federal governments in the development of a National Health Genomics Policy Framework. Prospective data from the Melbourne-based demonstration project provide strong evidence that genomic sequencing for childhood syndromes markedly increases diagnostic yield, and reduces 'cost-per-diagnosis', while improving short-term patient management. These data can be used to engage government(s) to obtain sustainable and equitable funding for genomic testing.

Establishing a National Genomic Medicine Service

Video (21:44) | Slides

Sue Hill (UK National Health Service)

Genomics England's 100,000 Genomes Project, a Driver Project of GA4GH Connect, was established in 2015 to implement genomic medicine in routine health care through the UK's National Health Service. NHS England has established 13 Genomic Medicine Centers across 90 of the country's 200 hospitals. It has so far collected more than 62,000 samples, and with the full roll-out planned for the end of 2018, will eventually reach three to seven million individuals. Currently, more than 36,000 whole genome sequences are available through the program, the bulk of which represent the rare disease patient community. The work has already demonstrated a diagnostic yield of 20 – 25% above the current standard of care within the NHS. Hill notes that as the world shifts towards a learning health system, it will be crucial for the broad community to be involved, including academics, clinicians, patients, and industry professionals working collaboratively across nations to change practice.

GA4GH Supporting Genomics and Health Panel

Video (07:14)

Eric Green (NIH), David Glazer (Verily Life Sciences), Heidi Rehm (Broad Institute of MIT and Harvard), Kathryn North (Australian Genomics), Sue Hill (UK National Health Service)

Green leads a panel discussion following the session on "GA4GH Supporting Genomics and Health," fielding audience questions about strategies for returning secondary information to participants and how to increase integration between the clinical and research communities within national genome sequencing initiatives like Australian Genomics and Genomics England.

CLOSING: GA4GH LOOKING FORWARD

Video (4:04)

Ewan Birney (EMBL-EBI)

Birney closes the 5th Plenary Meeting of the Global Alliance for Genomics and Health, noting that while there is much more work ahead to ensure that genomic and health-related data are routinely shared according to GA4GH standards, the community has already come a long way since the organization began in 2013.

5TH PLENARY PROGRAMME COMMITTEE

Thank you to the Programme Committee who made this meeting possible:
Heidi Rehm (Chair)
Peter Goodhand
Bartha Knoppers
Nicola Mulder
Kathryn North

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