Meeting Report
Virtual Connect 2021

This year, the 2021 GA4GH Connect meeting was held entirely virtually in response to global health concerns related to COVID-19. The meeting focused on two major areas: (1) collaboration across Work Streams and (2) development of the 2020 GA4GH Strategic Roadmap.

OPENING REMARKS

Slides | Recording

During Opening Remarks, participants received an overview of each Work Stream, along with their goals for the meeting. Initiatives external to GA4GH — including the Medical Genome Initiative, the Human Pangenome Reference Consortium, and the International Hundred-thousand Cohorts Consortium — also presented opportunities to collaborate with GA4GH.

WORKING SESSIONS

VRS 1.3 Planning and Implementation Guidelines

Agenda & Slides | Recording

In this session, the VRS team shared a progress report and discussed future plans. The team focused on key challenges and solutions for data exchange with VRS, using examples from VICC and ClinGen.

Genetic Discrimination Observatory

Agenda & Slides | Recording

In this session, collaborators discussed how the Genetic Discrimination Observatory (GDO) — a global initiative represented in more than 21 countries — can contribute to GA4GH.

Cloud Work Stream

Agenda & Slides | Recording

The Cloud Work Stream gathered together to hear updates from Driver Projects and discuss challenges in implementing GA4GH cloud standards.

Data Access Committee Review Standards (DACReS)

Agenda & Slides | Recording

Participants from the Regulatory & Ethics Work Stream (REWS) and the Data Use & Researcher Identities (DURI) Work Stream met to discuss progress on a new deliverable, the Data Access Committee Review Standards (DACReS), and to actively draft several key sections of the standard.

VCF/VRS/refget Alignment Meeting

Agenda & Slides | Recording

In this session, collaborators from the Large Scale Genomics (LSG) and Genomic Knowledge Standards (GKS) Work Streams aimed to identify barriers preventing interoperability between VCF, refget, and the Variation Representation Specification (VRS).

Federated Analysis Systems Project (FASP)

Agenda & Slides | Recording

In this session, three collaborators shared use cases for the Federated Analysis Systems Project (FASP). The group also discussed future goals and how to make it easier for new people to get involved.

Variant Annotation

Agenda & Slides | Recording

The Variant Annotation team discussed the plan to deliver a v0 specification for a Variant Annotation standard.

Regulatory & Ethics Work Stream

Agenda & Slides | Recording

The Regulatory & Ethics Work Stream (REWS) heard updates from Driver Projects and collected information on the implementation of REWS policy frameworks and tools.

Discovery Work Stream

Agenda & Slides | Recording

The Discovery Work Stream shared product updates and their roadmap for 2021. The team also discussed alignment of Discovery products, heard from Driver Projects, and collaborations with other Work streams.

Large Scale Genomics Work Stream

Agenda & Slides | Recording

The Large Scale Genomics (LSG) Work Stream shared updates from LSG projects, including file formats, htsget, refget, RNAget, Sequence Annotation, and Crypt4GH. The Work Stream also discussed the future of VCF project and other future plans

Beacon v2

Agenda & Slides | Recording

The Beacon team discussed key updates and features that will be incorporated into the v2.0 release. These include more robust filtering, the representation of structural variants, and a cohort model for Beacon.

Key Management in the Cloud

Agenda & Slides | Recording

The Crypt4GH team discussed ways to enable Crypt4GH-encrypted files to be processed in the cloud natively, ideally with standards developed by the GA4GH Cloud Work Stream.

Phenopackets and VA/VR Integration

Agenda & Slides | Recording

The Clinical and Phenotypic Data Capture (Clin/Pheno) and Genomic Knowledge Standards (GKS) Work Stream met to define the scope and requirements for GKS standards within the Phenopacket schema.

Regulatory & Ethics Work Stream

Agenda & Slides | Recording

The Regulatory & Ethics Work Stream heard from Driver Projects and received updates on ongoing projects, including the Genetic Discrimination Observatory (GDO), Data Access Committee Review Standards (DACReS), and Consent Toolkit.

REWS Return of Results Policy

Agenda & Slides | Recording

The Regulatory & Ethics Work Stream (REWS) met to discuss progress on the Policy on Clinically Actionable Genomic Research Results, including points of contention and other open questions.

EDI Workshop: Building a Diverse and Inclusive GA4GH

Agenda & Slides | Recording

At the EDI Workshop, the team presented the system of EDI initiatives within GA4GH. These include an Onboarding program to welcome newcomers, an initiative to encourage deeper engagement within the Work Streams, and a methods for integrating EDI into the standards development process.

DRS & Passports Alignment

Agenda & Slides | Recording

The Data Repository Service (DRS) and Passports team discussed ways to integrate DRS into the Passports specifications, and the potential changes needed to support this initiative.

Phenopackets & Pedigree Integration with Beacon and Data Connect (formerly Search) APIs

Agenda & Slides | Recording

The Clinical & Phenotypic Data Capture Work Stream met with the Discovery Work Stream to discuss integration of their APIs. The teams provided an overview of Phenopackets, Pedigree, Beacon, and Data Connect, and discussed potential approaches for integrating the standards.

Data Security: Federated Analytics and Cloud Security

Agenda & Slides | Recording

The Data Security and Cloud Work Streams discussed the current landscape of data analysis in the cloud. As computing platforms process highly sensitive data, they are moving from centralized to decentralized architectures to deal with privacy requirements. The group discussed how the GA4GH Cloud APIs must evolve to follow this trend, and key considerations that the APIs must take into account.

Sequence Annotation

Agenda & Slides | Recording

The Large Scale Genomics (LSG) and Genomic Knowledge Standards (GKS) Work Streams met to discuss the creation of a Sequence Annotation standard. The group brainstormed what features are needed and how to describe them.

DRS Alignment with Beacon and Data Connect (formerly Search)

Agenda & Slides | Recording

The Cloud and Discovery Work Streams met to discuss how the Data Repository Service (DRS) API can align and integrate with the Beacon and Data Connect APIs.

Schemablocks

Agenda & Slides | Recording

Contributors met to discuss Schemablocks, an effort to create a “super ontology reference” that spans across GA4GH Work Streams and products. The group discussed use cases from Data Connect (formerly Search) and Phenopackets.

Data Security Work Stream

Agenda & Slides | Recording

The Data Security Work Stream met to discuss ongoing projects, including the development of a Breach Response protocol, guidelines for federated analytics in the cloud, and malfeasance rules.

REPORT BACKS

Slides | Recording

During the Report Backs, each session shared their key accomplishments from the meeting and next steps.