Meeting Report

Connect 2019

Global Alliance for Genomics & Health

Opening Remarks

To kick-off the meeting, GA4GH provided organisational updates and an overview of the meeting. Attendees then heard updates and priorities from all Work Streams and existing Driver Projects. With the addition of six new Driver Projects to the GA4GH community this year, the group learned about their mission and goals.

- Work Stream Updates and Priorities
- Existing Driver Project Updates and Priorities
- New Driver Project Updates & Priorities

Working Sessions

Security Review Process
Agenda
The Data Security Work Stream met to review and finalise the GA4GH Security review process for proposed and completed deliverables. This included reviewing the overall process and finalizing forms.

Family History / Pedigree Standard
Agenda
The Pedigree team met to advance and coordinate ongoing project plans for the development of a GA4GH Family History/Pedigree standard.

Intro to Researcher IDs
Agenda
The Data Use and Researcher Identities (DURI) Work Stream met to discuss the Researcher IDs specification, prototype working systems, and identify potential early adopters.

Genomic Knowledge Standards Working Session Kick-Off
Agenda

The Genomic Knowledge Standards (GKS) Work Stream met to continue development of a variation profile design proposal and to review the reference implementation project plan.

Cloud API Review & DRS
The Cloud Work Stream met to review the status of all Cloud APIs and standards and to discuss next steps. The team also focused on the Data Repository Service (DRS) API, looking at demos of DRS v0.1 implementations and identifying issues for development.

RNASeq
Contributors of the Large Scale Genomics (LSG) Work Stream met to discuss the development of the RNAget standard. The team reviewed the product roadmap and heard from Driver Projects.

Schemablocks
Contributors of the Large Scale Genomics (LSG) Work Stream met to discuss the development of the RNAget standard. The team reviewed the product roadmap and heard from Driver Projects.

Regulatory & Ethics Work Stream Meeting
Agenda
The Regulatory & Ethics Work Stream (REWS) held a general meeting to introduce new Driver Projects, share updates on emerging issues, and review REWS policy framework initiatives—including return of results, participant engagement, GDPR forum, and the public interest basis for data sharing.

Acute Paediatric Care Information Models
Agenda
The Clinical and Phenotypic Data Capture (Clin/Pheno) Work Stream met to discuss the current development of a standard using FHIR and terminology for acute paediatric care at the Australian Genomics Health Alliance (AGHA) and Genomics England (GeL).

Connecting DUO and RI
Agenda
The Data Use & Researcher Identities (DURI) Work Stream met to discuss the integration of their two standards, the Data Use Ontology (DUO) and Researcher Identities. The team discussed holding a call for datasets that can be used with DUO for data discovery, exploring ways to encourage adoption of DUO terms, and plans for wider adoption.

Variant Annotation / Variant Representation
Agenda
The Genomic Knowledge Standards (GKS) Work Stream held a working session to dive into the development of the Variant Annotation and Variation Representation standards.

TES & TRS
The Cloud Work Stream met to discuss the Task Execution Service (TES) and the Tool Registry Service (TRS) standards. For TES, the team reviewed the roadmap for development and identified implementations. For TRS, the team discussed direction of the API, integration with other Cloud Work Stream standards, and requests for Driver Project feedback.

Crypt4GH
Contributors of the Large Scale Genomics (LSG) Work Stream met to discuss the Crypt4GH standard, including interaction with other specifications such as htsget and the Data Repository Service (DRS) and exploring final changes to the draft proposal.

JSON Schema Workshop
JSON schema is a vocabulary that allows one to annotate and validate JSON documents. Contributors of the GA4GH community held a workshop to teach the basics of using JSON, share upcoming changes, and provide tips for troubleshooting existing schemas.

Authentication and Authorisation Infrastructure (AAI) & Researcher IDs (RI)
Agenda
Contributors from the Data Security and Data Use & Researcher Identities (DURI) met to find implementers for the AAI and RI standards, precisely explain the security validation process behind the AAI and RI standards, and listen to feedback and concerns that Work Streams and Driver Projects might face in terms of data protection.

WES
The Cloud Work Stream met to discuss the Workflow Execution Service (WES) standard. The team reviewed WES implementations, discussed the design of a WES registry, shared plans for onboarding new users and compliance testing, and dove into WES development questions.

Variant Annotation / Variant Representation
Agenda
The Genomic Knowledge Standards (GKS) Work Stream held a working session to dive into the development of the Variant Annotation and Variation Representation standards.

Search API
The Discovery Work Stream met to review pilot implementations for the Search API and discuss considerations such as query language, referring to GA4GH and non-GA4GH data models, and building Search applications.

Read File Formats
The Large Scale Genomics (LSG) Work Stream met to review progress on and next steps for SAM, BAM, and CRAM.

FHIR / Phenopackets

Agenda

The Clinical & Phenotypic Data Capture(Clin/Pheno) Work Stream met to discuss and prepare the Phenopackets standard for the GA4GH product approval process.

Privacy & Security Policy

Agenda

The Regulatory & Ethics Work Stream (REWS) and the Data Security Work Stream held a joint session to discuss updates to the GA4GH policy approval process, changes to the Privacy & Security Policy to comply with the General Data Protection Regulation (GDPR), updates to the GA4GH Security Infrastructure, and ways to collaborate for the GenoPri conference.

Cloud Work Stream

The Cloud Work Stream held a session to dive into incomplete items in need of further discussion and brainstorming.

Future Plans for DURI

Agenda

The Data Use & Researcher Identities (DURI) Work Stream met to recap progress made at the meeting and discuss future plans. The team heard updates from the Authentication & Authorisation Infrastructure/Search meetings, learned about requirements from new Driver Projects, and held a presentation on ORCID. Next, the DURI Work Stream reviewed their Roadmap, explored plans for integrating the Data Use Ontology (DUO) and Researcher Identities (RI), and made plans to collaborate with the Clinical & Phenotypic Data Capture (Clin/Pheno) Work Stream and the Regulatory & Ethics Work Stream (REWS) to explore if RI could handle clinical data in upcoming versions.

GKS Hackathon

Agenda

The Genomic Knowledge Standards (GKS) Work Stream held a hackathon to explore the SEPIO model as a basis for demonstrating Variant Annotation ideas.

Beacon API

The Discovery Work Stream met to discuss the Beacon API and collaborations with ELIXIR.

VCF

The Large Scale Genomics (LSG) Work Stream met to discuss the next version of VCF.

Phenopackets

Agenda

The Clinical & Phenotypic (Clin/Pheno) Work Stream met to continue work on Phenopackets.

IP Policy
Agenda
Contributors across GA4GH met to discuss core elements of a draft GA4GH Intellectual property policy, which will outline guidelines for collaborating to develop open technical standards.

Data Security Document Updates & Future Planning
Agenda
The Data Security Work Stream met to discuss updates on their Security Technology Document and review their product development roadmap.

Cloud Work Stream
The Cloud Work Stream recapped progress made during the meeting and reviewed their product development roadmap.

Researcher IDs Claims Lab
Agenda
The Data Use & Researcher Identities (DURI) Work Stream met discuss the creation of a Researcher IDs Claims lab and to discuss the collection of claims.

Genomic Knowledge Standards Future Planning
Agenda
The Genomic Knowledge Standards (GKS) Work Stream recapped progress made during the meeting. The team heard from new Driver Projects and their needs, reviewed the GKS strategy, and updated their product roadmap.

Discovery Work Stream Moving Forward / Discovery Networks
The Discovery Work Stream recapped progress made during the meeting and discussed future plans. In particular, the team reviewed plans for the Service Info and Service Registry standards, and a potential collaboration with the Cloud Work Stream on the Data Repository Service (DRS) API.

htsget / refget recap
The Large Scale Genomics (LSG) Work Stream reviewed progress on the htsget and refget APIs and discussed plans to build out reference implementations and add support for Crypt4GH.

Open Terminology, FHIR Terminology & Wrap-up
Agenda
The Clinical & Phenotypic Data Capture (Clin/Pheno) Work Stream met to discuss using a FHIR Terminology Server with REDCap to collect phenotypic data and standardise data.

Work Stream Report Backs

Each Work Stream reported back on progress made during the meeting.

- Clinical & Phenotypic Data Capture (Clin/Pheno) Work Stream
- Cloud Work Stream
- Data Use & Researcher Identities (DURI) Work Stream
- Discovery Work Stream
- Genomic Knowledge Standards (GKS) Work Stream
- Large Scale Genomics (LSG) Work Stream
- Data Security Work Stream
- Regulatory & Ethics Work Stream (REWS)