12th Plenary

Meeting Report

The global genomics community convened in Melbourne, Australia and online for the GA4GH 12th Plenary meeting from 16 to 20 September 2024. Participants engaged in collaborative workshopping, discussed equity and inclusion in the field of genomics, and explored emerging technologies to scale genomic data sharing and advance human health.



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Experiments Metadata Standard

Agenda • Slides • Recording

The Experiments Metadata Standard product team developed a core set of properties to properly describe a sequencing experiment and a list of sequencing experiment categories to characterise in more detail the various types of sequencing experiments. Attendees presented the results of their work to date and collected additional feedback from the community.

Key takeaways

- Participants provided multiple suggestions to improve the first draft of the core properties checklist. These included adding a date field, making the sequence_protocol field mandatory, removing the checklist_group field, and adding metadata about sequencing instruments.
- The team should begin to determine its approach to embedding metadata in identifiers, particularly in the context of prioritising either human-readable labels or machine-readable identifiers for better clarity and functionality.
- There is currently no linkage to a specific library prep ID. Clarification is needed on whether the "library_name" field should be included in the identifiers tab, as well as what is meant by "library_name."

Next steps

- Implement proposed changes to the core properties checklist based on participant feedback.
- Continue discussions on complex issues such as identifier strategies and library prep IDs in future meetings.
- Interested in learning more or getting involved? Contact Neerjah Skantharajah.

Regulatory & Ethics Work Stream (REWS) general meeting

Agenda • Slides • Recording

Members of the Regulatory & Ethics Work Stream (REWS) and the broader GA4GH community met to discuss current REWS initiatives, provide a background on REWS products, and identify opportunities for future collaboration.

Key takeaways

- The Ethical Provenance Model Data Access Agreement (DAA) clauses were passed unanimously.
- The Newborn Sequencing Study Group is in the process of becoming a product team.
- The community had the opportunity to hear from Kimberly Ray from the Neuroscience Community of Interest.
- REWS looks forward to working with more Driver Projects, Communities of Interest, and Work Streams in the months ahead.

Next steps

- Ethical Provenance Model DAA clauses will be presented for final approval at the upcoming Product Steering Committee meeting in October 2024.
- REWS is updating its content on the GA4GH website to better reflect the range of products developed over the years.
- For those interested in joining any of the REWS groups, contact Jessica Seegobin.

Mad GKS v2.0: the Implementation Warrior

Agenda • Slides • Recording

In this session, implementation project representatives and session participants came together to discuss implementing Genomic Knowledge Standards (GKS) Work Stream products. This included raising concerns, identifying needs in both modelling and tooling, and discussing potential solutions.

Key takeaways

- The group identified 13 key areas for discussion. The top topics include:
    - Is there a recommended standard human-readable format for VRS data?
    - Is Cat-VRS the right solution for tying amino acid variants to genomic variants?
    - How can GKS build adoption confidence in the planning and delivery of its products?
    - How can we leverage elements from across GA4GH for better synergies?
    - Given the importance of normalisation, how do we ensure that all the normalisation pieces of code are aligned in the VRS python package?
    - Modelling Multiplexed Assays of Variant Effects (MAVE) and other functional assay data using Variant Annotation (VA).
- Aligning common data elements and entities across other GA4GH standards, such as Phenopackets and Beacon, should be a key focus.
    - Facilitating discussions among product teams and improving visibility will be vital. That will seed common understanding for all alignment efforts.
- Sharing and representing what models are being used in implementations will better support interoperability.

Next steps

- Prioritise putting together a process for transparently moving projects through the product development and approval process.
- Compile a list of items to understand what is preventing implementers from moving forward in their projects.
- Interested in getting involved in GKS? Contact Beatrice Amos.

What's new in VCF v4.5

Agenda • Slides • Recording

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During this session, attendees were introduced to the new features in the latest version of VCF v4.5. Particular areas of improvement include enhanced VCF scalability and methylation support.

Key takeaways

- VCF now supports methylation and scaling to larger cohorts.
- VCF supports many use cases that are currently implementing custom file formats.
- Widespread adoption of VCF v4.4 and v4.5 features requires support from key libraries such as htslib.

Next steps

- The Future of VCF team is gathering feedback and best practices on scaling VCF to handle large cohorts. Share your perspective and ideas via a brief questionnaire.
- The VCF specification team aims to:
    - continue work on updating the VCF validator to support VCF v4.5;
    - consider developing examples and recommended encodings for complex variant types in VCF.
- Interested in becoming a VCF specification maintainer or contributing to the specifications and test suite? Reach out to Reggan Thomas.

Advancing Variant Level Matching (VLM) and phenotype data sharing: building a federated network

Agenda • Slides • Recording

This interactive session of the GA4GH Implementation Forum (GIF) project, Federated Variant Level Matching, tackled pivotal topics related to implementing variant level matching and querying. The group discussed enhancing compatibility across platforms by integrating phenotype data sharing using Phenopackets and optimising variant query and representation using Beacon and the Variation Representation Specification (VRS). Attendees also discussed the design of a federated network and whether centralised

hubs are needed, in addition to evaluating the role of Passports in ensuring secure database connections within the federated VLM network. Learn more about this project here.

Key takeaways

- There is a strong push to improve variant-level data sharing across multiple datasets, moving beyond gene-level matching. This involves federated querying to facilitate one-sided matching, where researchers can search for any variant and retrieve phenotypic data from other databases.
- Standardising the APIs used across data-sharing networks is essential for ensuring efficient querying and data interoperability. A unified specification will simplify integration, reduce redundancy, and facilitate future scalability, though some flexibility is needed to accommodate diverse systems.
- The group will explore the feasibility of a centralised aggregation model or broker-based system to streamline data queries and manage format conversions. This could simplify interactions and provide a more scalable solution than the current decentralised model, though funding and technical challenges remain.

Next steps

- Collaborate with the VLM Network! The team is looking for funders, developers, and project representatives to support centralised platform expansion. To get involved, reach out to vlm@ga4gh.org.
- Work towards developing a unified API specification, with examples for better integration.
- Further investigate the feasibility and potential benefits of a centralised or broker-based system for managing queries and data formats.
- Continue working on a reference model for API and system interoperability, particularly in handling diverse data formats and variant types.
- Everyone is encouraged to attend monthly meetings to contribute to network development and data integration strategies. To get involved, reach out to vlm@ga4gh.org.

Mad GKS: beyond maturity

Agenda • Slides • Recording

The GKS maturity model is a fundamental component of the GKS product development and approval process. It is essential for ensuring community-driven, consistent, and methodical evolution of Variant Annotation (VA), Variation Representation Specification (VRS), and Categorical Variation Representation Specification (Cat-VRS) and their underlying data models. These developing technical specifications have a collection of data classes that will evolve and mature at varying rates, based on product implementer commitment to applying them. This session reviewed lessons learned from trialling the GKS product development and maintenance processes for the VRS specification, and provided an overview of how these processes provide a sustainable and agile mechanism that integrates with the GA4GH Product Development and Approval Process.

Key takeaways

- GKS is addressing variant knowledge fragmentation through the standardisation of three GKS products: VRS, VA, and Sequence Annotation (SA).
- Community feedback is critical, but survey fatigue and communication barriers need to be addressed, possibly through a centralised feedback portal.
- The GKS model will be piloted to show its potential to the broader GA4GH development community, but more collaboration across GA4GH product teams is needed for a standardised approach.
- Maintaining momentum is key, with pre-releases and trial use helping to incrementally develop specifications without long delays.
- Feedback from registered implementers indicates a need for stable, adoptable data formats that won't change drastically in the short term.

Next steps

- Are you implementing a GKS standard? Receive the latest updates and share your feedback directly with the development team to help improve the product. Register your implementation.

- Generate community awareness and input on the GKS Technical Specification Maintenance — a guidance document for maintaining and extending GKS technical specifications using a maturity model.
- Further feedback has been requested on timelines for GKS specification development.
- Feedback is encouraged on VRS v2.0 from all implementations.
- Interested in getting involved in GKS? Contact Beatrice Amos.

Identifying challenges to interoperability and gaps in GA4GH standards

Agenda • Slides • Recording

This session identified opportunities to improve interoperability between implementations of GA4GH standards. The workshop aimed to uncover areas where additional standardisation might be required to help drive interoperability and adoption. Through the workshop, participants shared their experiences of implementing GA4GH standards, challenges encountered along the way, and opportunities for improving interoperability of GA4GH standards. The whiteboard and unconference format helped gather feedback from participants on key areas of focus and possible solutions. In addition, this session expanded upon some of the topics raised at the 2024 ELIXIR All-Hands meeting in June, during which a similar session took place. These exercises helped the community further explore and understand the gaps and challenges in interoperability.

Key takeaways

- Interoperability continues to be a focus area for the GA4GH community and beyond, and the challenges faced in achieving interoperability needs further deliberation and discussion.
- Audience members responded to the question, “What are the challenges to achieving interoperability to, from, and in-between GA4GH standards (and their various implementations and clients)?” The ideas were captured on the Figma board and voted for importance, with the most voted item being a lack of specification maintainers.

Next steps

- Review the ideas shared on the Figma board based on the thematic area of the interoperability question, and evaluate if any of the ideas needs to be prioritised for the GA4GH tech team.
- Prioritised ideas would be funnelled into the GA4GH tech team's backlog, and outcomes will be shared at the next meeting.

Towards GA4GH-powered SPEs/TREs

Agenda • Slides • Recording

In this session, attendees discussed a possible GA4GH Implementation Forum (GIF) Project around data security: a toolbox of privacy-enhancing technologies for GA4GH-powered Secure Processing Environments (SPEs) and Trusted Research Environments (TREs).

Key takeaways

- Middlewares, gateways, and side cars seem to be useful patterns for granting GA4GH products with security and other extra features, in a reusable manner.
- There is a need to share credentials across GA4GH products in a unified way.

Next steps

- Apply to become a GIF project.
- Develop the White Paper "Universal Toolbox for Trust-Enhancing Technologies" operationalised through GA4GH products.
- Interested in getting involved in this initiative? Contact Reggan Thomas.

Cancer Data Commons: insights gained and future directions

Agenda • Slides • Recording

This session aimed to identify the critical elements and distinctive features of data commons while highlighting relevant GA4GH technologies that benefit these platforms. Additionally, it shared insights and lessons learned from existing data commons and similar initiatives, covering concepts like user requirements, access tiers, and minimal data models.

Key takeaways

- Data commons promote open science, high-quality data curation, and streamlined access through cloud-based platforms, accelerating cancer research.
- To improve data access and integration, cancer data initiatives have future goals of moving towards single multi-purpose user portals, like ZERO Childhood Cancer. Likewise, the National Cancer Institute (NCI) Cancer Research Data Commons (CRDC) is currently developing a centralised submission portal and data discovery dashboard.
- Platforms like Biomedical Research Hub (BRH), powered by Gen3, support cross-repository data exploration through FAIR APIs, enabling interaction between various cancer data commons.
- Future data commons will likely incorporate artificial intelligence (AI) models, aiming to enhance cancer research through tools like retrieval augmented generation (RAG).

Next steps

- Sign up here to get involved with the Cancer Community. Stay tuned for announcements from the Cancer Community, as we discuss next steps to continue the discussion on Cancer Data Commons!
- Join the next Cancer Community meeting on 11 November at 13:00 UTC to hear from Marie Wong-Erasmus, who will be presenting on the ZERO Childhood Cancer Beacon implementation. Email Neerjah Skantharajah or Yasmeen Kurdi to get meeting details.

File Formats Working Practices

Agenda • Slides • Recording

The File Formats group maintains a series of specifications, including the SAM/BAM/CRAM, VCF/BCF, and BED formats. The canonical implementations have historically been in C and Java, and, more recently, Rust. Some of these implementations and formats are underrepresented in the File Formats working group, making it hard to consider the needs of those implementations and slowing progress in those formats. During the session, attendees discussed proposals and brainstormed ways to improve working practices, including strategies for both offline work and ways of conducting the monthly meetings.

Key takeaways

- Define procedures for facilitating timely progress of simple, uncontroversial changes.
- Consider sharing the workload of preparing for and leading meetings by rotating chairing amongst interested regular attendees.
- Determine how to foster wider interest and participation — achieving a critical mass of audience size could change the status quo of off-line discussions tending to diminish through inactivity.

Next steps

- Monthly meeting time is valuable, but not always effectively used — the team will revisit meeting agendas, considering deep dives into particular topics and other more intentional uses of the time.
- Interested in getting involved in the file formats group? Contact Reggan Thomas.

GA4GH testbeds: a system to ensure conformance of implementations with GA4GH standards

Agenda • Slides • Recording

The GA4GH testbed is intended to aggregate results from multiple standards and invert control of running and reporting results onto individual deployments of GA4GH standards. In addition, the testbed also forms part of a new effort from GA4GH called "GA4GH Verified Implementations," which seeks to identify and catalogue open implementations of GA4GH standards. This workshop introduced the testbed

infrastructure and the GA4GH Verified Implementations effort, as well as educated attendees on how to submit a report and how to write new test suites. In addition, the session highlighted how existing tooling can be adapted to provide compatible submissions with the GA4GH testbed.

Key takeaways

- The community is engaged in the development of the GA4GH centralised testbed and would possibly like to use it for conformance testing of their implementations.
- There is interest in expanding the scope of the testbed to not only focus on API-based standards — for example, testing data structures. Another key feature is security through the implementation of authentication-authorisation mechanisms.

Next steps

- Interested parties are encouraged to submit their ideas on what features should be included in the testbed. These ideas will be reviewed, prioritised, and included in the GA4GH tech team's backlog.
- As the testbed evolves, all the enhancements will be presented to the community at the next Connect meeting for feedback.

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Beacon down under: pioneering practical cohort building in genomic research

Agenda • Slides • Recording

This session focused on advancing Beacon capabilities, from theoretical discussions to practical cohort building. Building upon insights from the April Connect 2024 meeting, attendees explored tangible ways to enhance Beacon's functionality by empowering users to actively engage with its capabilities in real-world scenarios. The session delved into the evolving landscape of Beacon technologies, particularly addressing the transition from conceptual frameworks to practical implementation strategies. We considered the potential for next steps in this journey. A key focal point was our alignment with the Beacon Scout group's efforts on "Aggregation & Cohorts." This group is tackling

challenges closely related to the Federated Cohort Building product team. By synergising efforts, the groups aim to explore collaborative opportunities that could lead to a unified approach.

Key takeaways

- The Beacon Scout Working Group is actively engaging stakeholders to define rules for data access and querying.
- Privacy and data-sharing challenges persist, particularly around managing sensitive cohort information.

Next steps

- Continue to gather community feedback on Beacon v2.0 features, specifically pre-emptive filtering counts.
- Continue exploring how to harmonise cohort filtering across different systems and networks.
- Investigate the development of a more sophisticated query language for the Beacon API.
- Engage contributors with expertise in linked data and semantic technologies for API improvements.
- Interested in getting involved in Beacon? Contact Beatrice Amos.

Public Perspectives on Genetic Discrimination

Agenda • Slides • Recording

Genetic Discrimination (GD) is a multifaceted phenomenon that can infringe upon human rights and erode trust in genomic research. Cognisant of the growing occurrence of GD in various fields, policymakers have adopted various policy approaches to address the phenomenon. Despite these efforts, public anxiety around GD persists. This working session brought together the Regulatory & Ethics Work Stream (REWS) Public Attitudes and GD product teams to brainstorm the development of a study that captures public perceptions around GD.

Key takeaways

- Public concerns and fear around GD exist worldwide and the meeting provided an opportunity to discuss these in the context of insurance and to hear from experts with examples in the Australian context.

Next steps

- The group will write a blog post around the A-GLIMMER project in Australia.
- Interested in getting involved in this initiative? Contact Jessica Seegobin.

DRS v1.6 and beyond: DRS access options for compute tools

Agenda • Slides • Recording

In this session, attendees scoped out features and improvements for the Data Repository Service (DRS) v1.6, to be released by the 2025 Plenary meeting. In particular, the group explored ways to improve access options for systems that compute on DRS objects.

Key takeaways

- The benefits and challenges of cold storage data access methods were explored, including direct cloud API access methods and direct copy of the data from cold storage to the compute environment.
- Participants stressed the importance of metadata management and the potential for a central registry to optimise data access and storage.

Next steps

- Implement a mechanism to encode geolocation and cloud constraints in DRS metadata.
- Explore adding object creation functionality to the DRS API.
- Investigate options for managing trust relationships between DRS servers and clients.
- Coordinate integration of Crypt4GH support in DRS.
- Encourage Driver Projects, implementers, and other interested parties to vote on DRS v1.5 PR #408.

- Explore options for adding defrosting mechanisms to DRS for deep cold storage objects.
- Interested in getting involved in DRS or other Cloud Work Stream projects? Contact Reggan Thomas.

Collaborative development of a FHIR-based family health history standard: leveraging KIN Ontology and Phenopackets with HL7, G2MC, and the US CDC/NCI

Agenda • Slides • Recording

This session introduced a new multi-organisation project focused on family health history (FHH) using FHIR (Fast Healthcare Interoperability Resources) standards. This project aims to leverage the KIN ontology and incorporate a Phenopackets use case, with input and participation commitments from various Driver Projects, including the NIH Cloud Platform Interoperability (NCPI) initiative. This session discussed and defined the scope and goals of this collaborative FHH FHIR project. Partners include Health Level Seven (HL7), Global Genomic Medicine Consortium (G2MC), the US Centers for Disease Control and Prevention (CDC) National Family Health History group, and the US National Cancer Institute. The organisations involved have tried out different representations for FHH in FHIR and are now looking to standardise. Stakeholders from these organisations, along with other interested parties, are encouraged to participate. This session's output included a comprehensive road map outlining the project's objectives, milestones, and implementation strategies, including the development of an open-source FHH tool to be used as a reference for other implementations. Pilot implementations were identified to test the proposed standards and approaches. This will lay the groundwork for a cohesive effort to standardise and enhance FHH data exchange, ultimately benefiting the global genomics community.

Key takeaways

- There are significant barriers to fully integrating family health history into electronic health records (EHRs), including limited data models and brief family history collection during clinical visits.
- A universal family health history data model, integrated with existing standards like HL7 FHIR, could enable better clinical decision-making and support precision medicine.

- The tool that this group is developing has global applications, particularly for rare disease advocacy groups and low-resource settings, with customisation options for specific conditions like cancer and pharmacogenomics.

Next steps

- Encourage more collaboration to progress the development of the group's new FHH tool.
- Continue refining the FHIR-based data model and ensure it encompasses comprehensive family relationships.
- Await feedback on Grant Wood's funding proposal for the Alpha-1 disease project, aiming for collaborative input from various organisations.
- Investigate opportunities for creating FHH tools tailored to regions with less developed healthcare infrastructure, utilising mobile technology.
- Maintain ongoing communication about project developments, inviting feedback and engagement from stakeholders.
- Interested in getting involved in this initiative? Contact Beatrice Amos.

Cloud Work Stream in 2025

Agenda • Slides • Recording

In this session, attendees explored the evolution of the Cloud Work Stream and goals for 2025. This included setting high-level priorities for each of the APIs — the Workflow Execution Service (WES), Task Execution Service (TES), Tool Registry Service (TRS), and Data Repository Service (DRS). The group also looked at potential new standards work, such as mesh cards.

Key takeaways

- The TRS team aims to continue maintaining support of its current version and explore new directions, such as AI, usage statistics reporting, and federated indexing or sharing mechanisms.

- TES now has five ecosystem servers supporting it. The team aims to focus on encouraging broader adoption of the standard, continuing support of federated and distributed compute, and exploring security needs for enterprise scale compute.
- The concept of mesh cards was introduced as a potential solution for describing interconnected systems in a machine-readable and human-friendly format.
- Multiple systems support data sharing with DRS, including NIH cloud platforms, with around 27 petabytes of data from around two million participants. The team is drafting a v1.5 release and is working towards a publication.
- Discussions covered the need for better security, authentication, and federated computing, as well as exploring the potential for using large language models to automate workflow generation and improve documentation.

Next steps

- Explore the "mesh card" concept further in the Cloud Work Stream and get feedback from Driver Projects.
- Look into providing a standard for reporting metrics and provenance data across the different Cloud Work Stream APIs. The team plans to look at what htsget has done.
- Reach out to end-users to create a reference platform highlighting known product implementations and showcasing the integration of the different Cloud Work Stream APIs.
- Improve commonality between the Cloud Work Stream APIs where possible, such as common return or configuration types in TES and WES.
- Interested in getting involved in the Cloud Work Stream? Contact Reggan Thomas.

Generative AI for genomic data sharing session

Agenda • Slides • Recording

With the current rise of generative artificial intelligence (AI) tools, promising opportunities for the streamlining of access procedures and data querying are foreseeable. Conversational AI capabilities based on large language models have the potential to

facilitate large-scale genomic data sharing from multiple available open access datasets online. If this technology is to be rolled out in a responsible manner, however, guidance on the way these technologies can better account for attribution, privacy, bias, and accountability will be necessary. This working session brought together members of the GA4GH community and the REWS Generative AI Study Group to brainstorm points to be included in a guidance document that can promote the responsible use of conversational AI in the context of large-scale, open access genomic databases.

Key takeaways

- AI legislation is context specific; there is a need for refining guidance points around responsible usage with feedback from real world implementation and development.

Next steps

- An AI and machine learning publication is in the process of being drafted, with revisions based on comments from the session.
- Interested in getting involved in this initiative? Contact Jessica Seegobin.

Passports and AAI

Agenda • Slides • Recording

Attendees discussed what is next for the GA4GH Passports and AAI specifications by considering how these specifications are used for standards-based secure data sharing and data use. This session explored the concept of data visiting and where secure data sharing and data use happen across institutions, enabled by establishing trust relationships and security policy. Several AAI and Passports ecosystems were showcased, including ELIXIR and NIH RAS. Session attendees learned how to build and grow ecosystems using the Fake Passport Ecosystem to demonstrate trust relationships and data sharing. Lastly, the group discussed plans for the next version of Passports and AAI and showed drafts and proof of concepts around new specification features that are taking shape, such as smaller tokens and downscoping.

Key takeaways

- There is a need to reassess assumptions around “genomic exceptionalism” within high-priority use cases to ensure the group’s focus is relevant and facilitates appropriate decision-making.
- It is important to clearly articulate the unique community challenges and barriers that exist, while evaluating the applicability of broader approaches versus genomic-specific solutions.
- It is essential to maintain documentation comparing alternative technologies with current approaches in Passports.
- Further exploration is required into the scalability issues with large Passports due to the lack of clarity around scale.
- Collaboration with other organisations and Driver Projects is important to define and address challenges.
- There is a need to explore the potential for a server-based approach with a call back mechanism for requesting specific authorisations in lieu of using large Passports.

Next steps

- Establish the higher-level requirements and use cases for AAI in distributed systems as documented in the IAM & AAI proposal.
- Maintain a living document with an analysis and list of alternatives, emerging technologies, and components of genomic exceptionalism to distil information from use cases.
- Create a closing document to capture agreements and decisions made, including the reasoning behind decisions.
- Collate proposals and additions to the Data Security Work Stream (DSWS) road map into one consolidated place using the DSWS Project Backlog Document.
- Interested in getting involved in this initiative? Contact Jessica Seegobin.

Bridging challenges and new implementations in GA4GH Clin/Pheno standards

Agenda • Slides • Recording

This session focused on addressing the challenges faced by the GA4GH Clinical & Phenotypic Data Capture (Clin/Pheno) Work Stream and introducing new implementers of GA4GH Clin/Pheno standards. This session was designed for stakeholders who are actively involved in clinical and phenotypic data integration and standardisation, and those with ideas for new GA4GH activities or challenges with existing ones. Session participants discussed current obstacles in implementing Clin/Pheno standards, shared insights from various use cases, and explored potential solutions. New implementers had the opportunity to present their experiences and provide feedback on the adoption process. The goal of this session was to foster collaboration, share best practices, and develop a strategic road map to overcome existing challenges and enhance the implementation of GA4GH Clin/Pheno standards across diverse settings. This collaborative effort aimed to drive the advancement of clinical and phenotypic data interoperability, ultimately benefiting the global genomics community.

Key takeaways

- A vision for an integrated Family Health History system was presented, utilising a central repository for genomic, molecular, and family history data. The development of KIN is underway to fill gaps, including non-gender-specific terms and IVF-related relationships.
- The GA4GH Rare Disease Community is focusing on reconciling clinical data, standardising phenotype collection, and improving data analysis. The group is working on building a GA4GH rare disease portfolio to align clinical labs with global standards.
- The challenges in defining cohorts for data sharing was discussed, emphasising the need for a standardised phenotype library and a query language to make inclusion and exclusion criteria computable.
- Ongoing work using ontologies like Mondo and Human Phenotype Ontology (HPO) for clinical data representation was discussed, focusing on challenges with mapping existing cohorts and data liftover using Large Language Models (LLMs).
- Ongoing collaborations for improving the Phenopackets format and integrating environmental and ethnicity data was discussed.

Next steps

- Finalise the Pedigree standard by merging existing frameworks (e.g., HL7 FHIR) and address remaining gaps. Develop a universal implementation guide in collaboration with GA4GH, HL7, and other stakeholders.
- Expand the GA4GH Rare Disease Community, with a focus on increasing membership, adoption of standards, and promoting collaborative research and development.
- Reboot the computable cohorts initiative, focusing on creating standardised cohort definitions and developing query tools to make clinical trial criteria computable.
- Address the challenges of ontology mapping for clinical data, particularly in regions where data is coded in non-standard formats. Explore the use of LLMs for data liftover.
- Resume Phenopackets meetings to advance the work on integrating Phenopackets with HL7 and FHIR standards through community hackathons. Develop the translation software for EHR data to be used in genomic diagnostic tools, aiming for smoother data exchange.
- Strengthen collaborations with Driver Projects and emphasise Indigenous genomics and family relationship data. Preserve Indigenous sovereignty in data representation and explore connections between Indigenous health and genomics research.
- Develop a structured approach for handling extensions in standards to avoid divergence and maintain interoperability.
- Interested in getting involved in Clin/Pheno? Contact Beatrice Amos.

Improving data interoperability across GA4GH: Data Model and Schema Consensus (DaMaSC) Study Group

Agenda • Slides • Recording

This session was an active listening session to gather community input on DaMaSC's two primary initiatives: Best Practices and Schema Registry. The session Co-Chairs reviewed the mission of the DaMaSC product team, describing its two primary initiatives, before moving on to the listening session.

Key takeaways

- Interoperability and data sharing require common standards in ontologies and schema representation.
- A centralised schema registry is essential for organising and accessing GA4GH schemas.
- Flexibility in schema formats (e.g. JSON) and metadata practices will ensure broader adoption.

Next steps

- Continue to refine the schema registry proposal with community feedback, focusing on API development and governance.
- Continue gathering input through the ongoing community survey on data models, ontologies, and schema registry needs.
- Explore best practices for metadata and ontologies.
- Interested in getting involved in DaMaSC? Contact Beatrice Amos.

GA4GH Product Lifecycle

Agenda • Slides • Recording

The Product Development and Approval Process describes GA4GH's approach to developing and approving standards, but it omits the later stages of a product's life cycle, namely what happens when a product moves into maintenance mode or needs to be retired and will no longer be actively developed. Work Streams have flagged a number of products and standards that are approaching this phase. This session brought together interested parties to explore this space and work towards developing a set of criteria and overall process that can be applied to all GA4GH products. The output of this session will not be the final process, but a seed for further discussion. Those who engaged in this

session or are interested in participating are encouraged to continue engaging in the process.

Key takeaways

- A whiteboard exercise was undertaken to explore when or why a product might move into maintenance mode, the types of maintenance mode, and challenges associated with reactivating contributor networks.

Next steps

- Review existing points of contact for all GA4GH products. If no one is assigned, make a note of the gap and flag for recruitment.
- Create a strategy for reviewing standards and processes, and include mechanisms to drive project engagement and insert accountability.
- Define the essential activities needed for projects in "maintenance mode."
- Develop a project management job description, listing the responsibilities of the role.

Towards a GA4GH Rare Diseases portfolio

Agenda • Slides • Recording

As a leading genomic health standards body, GA4GH has published a significant number of standards to support the operationalisation of genomic health implementations, advancing the field of genomic medicine. GA4GH standards support a wide variety of horizontal (domain-agnostic) use cases, but their extensive and overlapping scopes make it challenging to apply them cohesively to vertical domains, such as rare disease. This session aimed to take the first step towards creating a portfolio of GA4GH products and associated descriptions of their recommended usage in the rare disease domain.

Key takeaways

- Working towards a GA4GH Rare Disease Portfolio will require the development of concrete and tangible outputs, cross-Work-Stream collaboration, and the inclusion of patient partners in shaping best practices and applications of GA4GH products for rare disease research.

- Current GA4GH standards that have been identified as relevant to the rare disease domain include:
    - Variation Representation Specification (VRS), Categorical Variation Representation Specification (Cat-VRS), and Variant Annotation (VA) from the Genomic Knowledge Standards (GKS) Work Stream;
    - Model consent clauses for Rare Disease Research and the Diversity in Datasets Policy from the Regulatory & Ethics Work Stream (REWS);
    - Phenopackets from the Clinical & Phenotypic Data Capture Work Stream;
    - Passports and AAI from the Data Security Work Stream.

Next steps

- Each Work Stream will nominate a delegate to continue to build the Rare Disease Portfolio collaboratively through regular meetings, led by Rare Disease Community leadership.
- Stay tuned for announcements from the Rare Disease Community, as we discuss next steps to build this portfolio!
- Interested in getting involved in the Rare Disease Community? Contact Yasmeen Kurdi.

Global Beacon showcase: advancing data discoverability across platforms

Agenda • Slides • Recording

This GA4GH session was dedicated to showcasing the new diverse array of Beacon implementations revolutionising genomic and phenoclinic data discoverability worldwide. The session featured presentations on pioneering initiatives such as the ELIXIR Beacon Network. We explored developments from Beacon projects including the serverless Beacon by Australian BioCommons, designed to optimise scalability and efficiency. There were presentations from the European Open Science Cloud for Cancer (EOSC4Cancer) Beacon, tailored to enhance cancer research. Beacon integrated with cBioPortal, offering interoperability for oncogenomic data exploration, was also discussed. This session discussed the TRE-FX platform from Nottingham, which enables federated analysis and

learning (FX) across Trusted Research Environments (TREs). Experts and developers driving these advancements shared insights into implementing the standard, overcoming challenges, and the future potential of Beacon technologies. This session aimed to foster collaboration and ignite discussions on advancing data discoverability across global research landscapes.

Key takeaways

- Beacon is a lightweight, scalable interface for querying and aggregating data across various systems and implementations.
- Serverless Beacon solutions are efficient and cost-effective for handling large datasets without extensive infrastructure.
- Integration of Beacon with existing data models and systems is crucial but poses challenges that need to be addressed.
- Performance monitoring and scalability are critical for effective data querying and network performance.
- Standardising phenotype definitions and improving community collaboration are essential for advancing Beacon's capabilities and adoption.

Next steps

- Develop and test integration methods for Beacon with existing hospital data models like OMOP and cBioPortal.
- Enhance performance monitoring for Beacon systems, particularly for SQL queries and large datasets.
- Establish a centralised phenotype library to standardise definitions and queries across different systems.
- Promote community engagement by inviting more Beacon implementers to contribute and maintain the network.
- Interested in learning more about or getting involved in Beacon? Contact Beatrice Amos.

Plenary • 18 September 2024

Welcome to Country Ceremony

Plenary began with an acknowledgement and performance by Djirri Djirri Cultural dancer Kiera Hunter, a member of the Traditional Owners of Narrm (Melbourne), the Wurunderji Woi Wurrung people of the Kulin Nations.

Plenary Opening Remarks

Slides • Recording

Plenary Programme Committee Chairs Tiffany Boughtwood and Oliver Hofmann welcomed conference attendees to Plenary. They expressed that with the conference held in Australia, there was a great diversity in conference attendees, which fosters a global, collaborative approach to addressing the GA4GH vision.

Alex Brown (Australian National University) keynote: “Towards a genomics equity agenda: lessons from Indigenous Australians”

Chair: Ewan Birney (EMBL-EBI)

Slides • Recording

Including Indigenous communities and other diverse communities in genomics research is imperative to support the development of tools that can accurately and appropriately address the needs of those communities. Much of genomics research has centred on people of European descent, but Brown noted that it is important to include the uniqueness of stories of Indigenous and ancestrally diverse communities to better understand and care for the health and wellbeing of all of humanity.

Equity, diversity, and sovereignty: advances in Indigenous genomics

Chair: Anjene Musick (NIH)

Leadership and participation from Indigenous communities are crucial to ensuring equity of access to, and delivery of, the benefits that genomics can offer. This session focuses on collaborative approaches for building resources and systems that embody the perspectives and priorities of Indigenous communities.

Jimmy Breen (Black Ochre Data Labs, Telethon Kids Institute and Australian National University), "Building data systems for Indigenous Australians"

Slides • Recording

Breen discussed the importance of building genomic data systems for Indigenous Australians to bridge healthcare inequalities in Australia. This can be made possible by implementing consistent, culturally appropriate, and ethical national genomic data storage, management, and sharing practices through Indigenous community input to advance precision public health impact.

Azure Hermes (Australian National University — National Centre for Indigenous Genomics), "Building resources the right way: reflecting Indigenous community perspectives and priorities."

Slides • Recording

Partnership with Indigenous communities is imperative to guide the development of culturally informed and appropriate genomic resources that are best positioned to reflect and meet community perspectives and priorities. Hermes discussed how this can be done through dialogue that promotes listening to the priorities of Indigenous communities, collaborative co-creation of resources, and tool sharing that encourages evaluation of the resources' efficacy and impact.

Nadine Caron (University of British Columbia), "The ongoing chase for equity: one Indigenous Canadian's perspective on the potential of genomics"

Slides • Recording

There is currently a scarcity of Indigenous identifiers in biobanks, Indigenous researchers and healthcare professionals, and Indigenous leadership in research foundations and agencies that grant funding. Caron discussed the importance of enhancing inclusion and representation of diverse populations in research to advance culturally appropriate precision healthcare.

Community-driven genomic implementation

Chair: Tiffany Boughtwood (Australian Genomics)

In this session, speakers shared experiences and strategies for implementing genomics projects that are driven by and for communities, ensuring that local and patient needs and values are reflected in genomic research and healthcare.

Tamra Lysaght (University of Sydney), "Engaging diverse communities on the ethics and governance of genome data sharing: experiences from Singapore"

Slides • Recording

Lysaght discussed the importance of engaging, including, and building trust among diverse communities in genomics. Lysaght presented a case study of Singapore's National Precision Medicine Strategy (NPM), which included survey results on public attitudes around research and willingness to share data, with a key finding that relationships are crucial to establishing trust.

Sean Murray (Mito Foundation), "Mito Foundation"

Slides • Recording

Through a series of stories about patients who have been impacted by mitochondrial diseases, Murray presented the work of the Mito Foundation to share Mito community insights, which can advance research, health innovation, and community advocacy.

Catalina Lopez-Correa (Genome Canada), "Evidence based genomic testing: a scientist and patient perspective"

Slides • Recording

The existing precision health landscape is limited and siloed in Canada. Lopez-Correa explained that a Canada-wide Precision Health Initiative requires experience coordination, collaboration between partner organisations and funders, and a prioritisation of data quality, while ensuring data privacy and security. By sharing about her own patient journey with breast cancer, Lopez-Correa emphasised the importance of patient centred data management.

Phillip Wilcox keynote: “With, not for: increasing participation of Indigenous populations in medical genomics research by prioritising their ethical frameworks”

Chair: John Clarkson

Slides • Recording

Phillip Wilcox, experienced in developing tikanga (Māori ethics)-based frameworks for researchers, emphasised that researchers must prioritise the ethical frameworks of underrepresented communities when seeking to include them in genomics research. Engagement with Indigenous knowledge, co-design of research, and establishment of trust, honesty, and transparency are crucial to ensure community health outcomes.

The changing face of genomic technologies

Genomics has the potential to improve human health at scale. In this session, speakers discussed advancements in genomic technology and how these approaches encourage new understandings of human health and disease treatment.

Joseph Powell (Garvan Institute of Medical Research), "Transitioning cellular genomics into the clinic"

Slides • Recording

Powell discussed clinical implementation of cellular genomics for healthcare use across a patient’s lifecycle. Research is being undertaken to determine feasibility of inclusion in clinical care and identify the potential for early adoption, aided by engaging with advocacy groups, leveraging data standards, and partnering with organisations to advance implementation.

Eimear Kenny (Icahn School of Medicine at Mount Sinai), "Population genetics in an era of genomic health"

Slides • Recording

Kenny discussed the power of biobanks to strengthen precision medicine and predictive genomics in clinical care. She introduced the Human Pangenome Project, which is working to enhance the representation of global genomic diversity through many reference genomes (or a pangenome) to advance precision medicine

Jianjun Liu (Genome Institute of Singapore), "A comprehensive exploration of a fully phased diploid T2T Indian genome: towards the creation of Asian Pangenome Reference"

Slides • Recording

Asian populations have not been historically represented in the development of Human Genome References. Liu discussed projects to capture the genetic diversity of Asian Populations through the Asian Reference Genome (ARG) Project.

Alan Rubin (Walter and Eliza Hall Institute), "MaveDB: An international community database for functional assay data"

Slides • Recording

MaveDB, launched in 2019, is a community database for multiplexed functional data with millions of variants. It implements GA4GH tools and standards, including the Variation Representation Specification (VRS) to map genomic variants and Variant Annotation Specification (VA-Spec) to build functional data models.

Evening talk show: AI in genomics

Recording

Through a talk show format hosted by science journalist Bianca Nogrady, Enirco Coiera (MacQuarie University), Daniel MacAruther (Garvan Institute of Medical Research), and Jeannie Marie Paterson (University of Melbourne) discussed the challenges and opportunities in implementing AI in genomic research and healthcare.

Plenary • 19 September 2024

Sue Hill (NHS England)keynote: “The NHS Genomic Medicine Service — advancements, challenges, and future trends”

Chair: Kathryn North (Murdoch Children’s Research Institute)

Slides • Recording

Hill discussed key advancements in implementing genomics at scale in the UK, including significant investment in the 100,000 Genomics Project, the establishment of the Genomic Research Library at Genomics England, the launch of the NHS itself in 2018, and the publication of its first genomics strategy in 2022. Next, Hill dove into the key priorities of the strategic plan — 1) embedding genomics into the NHS service model; 2) delivering equitable genomic testing for improved prevention, diagnosis, and precision medicine; and 3) enabling genomics to be at the forefront of the data and digital revolution — and the challenges and future trends of each.

Advances in health genomic translation: new frontiers of clinical implementation

Chair: Heidi Rehm (Massachusetts General Hospital, Broad Institute of MIT and Harvard)

Representing genomic initiatives from Indonesia, Singapore, and Saudi Arabia, the speakers in this session shared their experiences in implementing genomics into clinical practice to improve patient outcomes and realise the promise of precision medicine.

Gunadi (Universitas Gadjah Mada), "Implementation of genomic medicine in low-middle income countries — lesson learned from Indonesia"

Slides • Recording

Underrepresentation of Asian genomes, including Indonesia, has hindered population and medical genetics research on Asians, leading to population disparities in genomic medicine. Established in 2022, the Indonesian Precision Medicine Initiative aims to implement genomic medicine In Indonesia through a three-pronged approach: personalised therapy, disease diagnosis and screening, and pharmacogenomics.

Ee Shien Tan (KK Women's and Children's Hospital), "Harnessing genomics to impact patient care — the Singapore experience"

Slides • Recording

Tan shared progress and insights from Singapore's National Precision Medicine (NPM) Programme. Currently in its second phase, the programme includes common platforms to support clinical implementation, clinical implementation pilot projects, and economy and policy analysis. Tan emphasised that interdisciplinary collaborations, integration of genomics into healthcare systems, regulatory oversight, ethical frameworks,

infrastructure, and education and awareness of the benefit of genomics and genomic medicine are all critical factors to impacting patient care.

Lama AlAbdi (King Faisal Specialist Hospital and Research Centre), "Founder variants in Arabia: catalysts of precision medicine and beyond"

Slides • Recording

Founder variants play an important role in causing genetic diseases in Saudi Arabia. AlAbdi discussed how the study of Arab founder variants can improve the sensitivity and specificity of the clinical genome, and how the study of Arab founder variants can accelerate genome research and public health solutions, based on disease burden estimation and screening programs. Arab founder variants can also help inform population history and supplement principal component analyses.

GA4GH standards, products, and initiatives

Chair: Laszlo Radvanyi (Ontario Institute for Cancer Research)

GA4GH standards, products, and initiatives aim to support the global genomics community, facilitate responsible data use, and promote interoperability. Speakers in this session shared updates on projects from the Regulatory & Ethics, Genomic Knowledge Standards, and Discovery Work Streams.

Dianne Nicol (University of Tasmania), "Measuring the uptake of ethics and data governance policies in genomics"

Slides • Recording

Nicol shared the aims of the GA4GH Regulatory & Ethics Work Stream (REWS), which takes a human rights approach to fostering responsible data sharing for the public good and equitable healthcare. An overview of key REWS activities was shared, including the move towards clinical implementation through the newborn screening group and the clinical data sharing group. One challenge that Nicol emphasised is measuring the impact of policy tools and resources, as well as methods towards reaching that goal.

Alex Wagner (Nationwide Children's Hospital), "GA4GH genomic knowledge standards: advancing scalable (re)interpretation of paediatric cancer variants driven by FAIR representation of genomic evidence"

Slides • Recording

Paediatric cancer is the leading cause of death by disease in the developed world in children over one year of age. To increase our understanding of paediatric cancer variants, it is vital to integrate knowledge from multiple sources. To address this challenging endeavour, Wagner discussed key products developed by the GA4GH Genomic Knowledge Standards (GKS) Work Stream, including the Variation Representation Specification (VRS), Variant Annotation (VA) and Sequence Annotation (SA), which aim to provide a standard way to structure and share knowledge.

Jordi Rambla (Centre for Genomic Regulation), "GA4GH Beacon v2.0: an example of how the GA4GH community contributes to improve health globally"

Slides • Recording

Rambla discussed how the GA4GH Beacon model can be used to discover and query datasets that are part of a Beacon network. Beacon describes both a model for how different pieces of information relate to each other, and a framework for requests and responses. The presentation concluded with the strengths and weaknesses of Beacon, along with example use cases.

Herawati Sudoyo keynote: “Genomic landscape of the Indonesian Archipelago: unveiling diversity from West to East”

Chair: Rob Annan (Genome Canada)

Slides • Recording

With over 16,000 islands home to around 275 million people representing 483 ethnic groups and 726 languages, Indonesia is a diverse and dynamic theatre for human development and migration. To address wider questions in population genetics and functional genomics, deep whole genome and transcriptome sequencing was adopted in 2018. Local collaborations and community engagement led to the collection of over 3,500 samples from more than 100 communities. Sudoyo emphasised that it is essential to expand beyond DNA to have a full definition of health — including information such as phenotypic data, socio-economic status, and lifestyle.

The progress and power in understudied populations

Chair: Étienne Richer (Canadian Institutes of Health Research)

Featuring speakers from the Philippines, Brazil, Malaysia, and Japan, this session focused on transformative initiatives that are examining understudied populations to uncover new insights and promote global health equity.

Eva Maria Cutiongco-de la Paz (University of the Philippines Manila), "Strategies, standards, and sustainable policy frameworks for the FILIPINOme: The Filipino Genome Sequencing Initiative"

Slides • Recording

The FILIPINOme is a project aiming to sequence 10,000 Filipino genomes. Cutiongco-de la Paz discussed data governance and enterprise architecture strategies, interoperability and data privacy and security standards, and the FILIPINOme's GA4GH 4 PH initiative — all in the effort to develop a secure, integrated, and accessible Filipino reference genome to advance precision medicine globally.

Tábita Hünemeier (University of Sao Paulo), "Exploring Brazilian genomic admixture for medical and evolutionary research"

Slides • Recording

Existing genomic databases are not representative of the world's population. Hünemeier discussed the DNA do Brasil project, which aims to reconstruct the Brazilian population histories by analysing whole-genome sequencing data from 12,000 individuals from all regions of Brazil to investigate the impact of demography, admixture, and selection on the health of Brazilians.

Syahril Abdullah (Malaysia Genome and Vaccine Institute), "MyGenom: establishing a reference genome for precision medicine in Malaysia"

Slides • Recording

Abdullah discussed the MyGenom project, Malaysia's embarkation in population genomics to provide the genomic baseline for future precision public health implementation. The project aims to sequence 10,000 genomes across the country, including both healthy and unhealthy individuals. The project begins this year, 2024, with the sequencing of 2,400 genomes.

Kenjiro Kosaki (Keio University), "Japan's national projects for the implementation of genomic medicine"

Slides • Recording

Kosaki discussed progress made in several of Japan's nationwide genome research projects: the Initiative on Rare and Undiagnosed Diseases (IRUD), the Tohoku Medical Megabank, and the "Action Plan for Whole Genome Analysis" supported by the Health Policy Bureau of the Ministry of Health, Labour, and Welfare. IRUD now contains 7,318 deeply phenotyped undiagnosed patients and families; the Tohoku Medical Megabank contains a healthy cohort of 150,000 people, as well as 54,302 samples of normal Japanese variant allele frequencies; and the "Action Plan for Whole Genome Analysis" is undergoing its preparatory phase, with a focus on cancer and rare disease.

National Initiatives Forum • 20 September 2024

The National Initiatives Forum (NIF) meeting featured world-class experts leading dynamic presentations and discussions on the most pressing issues facing national genomic initiatives. Thought leaders explored topics including advanced data sharing, the integration of clinical genomics into routine practice, and strategies for securing essential funding. The meeting dove into the future of genomic newborn screening and explored how initiatives in this space are connecting with the public, participants, and professionals. Meeting participants were at the forefront of genomic advancements and gained insights to drive impactful change in their national initiatives.

Kathryn North (Murdoch Children's Research Institute), "NIF Opening Remarks, Vision, Mission, and NIF Charter"

Slides • Recording

North presented an overview of NIF and its integration within the broader GA4GH framework. The session included a review of the updated NIF charter, highlighting key changes made following the NIF Strategic Workshop held in May.

David Glazer (Verily), "Data Sharing: a call to action"

Slides • Recording

Glazer described the current data sharing landscape, identifying existing bottlenecks, and outlining twelve strategic principles and steps to systematically scale data sharing, all

aimed at enhancing health outcomes. The work presented was from an in press paper co-authored by members of NIF leadership and their collaborators.

Heidi Rehm (Massachusetts General Hospital, Broad Institute of MIT and Harvard), “Network of Global GnomADS”

Slides • Recording

Rehm introduced the federated gnomAD project, which seeks to diversify genomic datasets. Participating groups currently working to develop their own instances of gnomAD include China, Singapore, Canada, and Qatar. Monthly meetings are held to share progress updates and foster community engagement.

Christian Marshall (Hospital for Sick Children and University of Toronto), “International Precision Child Health Partnership (IPCHiP): a model for international data sharing”

Slides • Recording

Marshall shared insights from the IPCHiP flagship project, Gene-STEPS, including sample queries clinicians wish to perform. A data platform task force was established to identify and evaluate software technologies that can accelerate IPCHiP’s research. These learnings may also benefit other National Initiatives facing similar challenges.

Robert Freimuth (Mayo Clinic), “Data standards for clinical genomics: GA4GH on FHIR”

Slides • Recording

Limitations of FHIR in representing complex genomic data was used as a case study to illustrate how GA4GH can support the broader vision of aligning information models and ultimately enabling seamless data exchange.

Tiffany Boughtwood (Australian Genomics), “WHO TAG-G: global vision and regional impact”

Slides • Recording

Boughtwood introduced the development and structure of the WHO Technical Advisory Group on Genomics (TAG-G), which will guide progress on implementing the recommendations from the WHO Science Council report, “Accelerating access to genomics for global health: promotion,

implementation, collaboration, and ethical, legal, and social issues," with a focus on low- and middle-income countries (LMICs).

Jordan Lerner-Ellis (Mount Sinai Hospital), "The evolution of health data ecosystems"

Slides • Recording

Lerner-Ellis provided a current state analysis conducted under the Canadian All for One initiative to develop health data ecosystems (HDEs). This included an overview of funding, scope, and mandate of HDEs in centralised versus decentralised systems, alongside other key comparisons.

Zornitza Stark (Australian Genomics), "Introduction: implementation of clinical genomics — making the case for funding"

Slides

Stark introduced the challenges of implementing clinical genomics, emphasising the necessity of a whole-system approach that encompasses education and training, fosters innovative working methods, and establishes the infrastructure and processes required for successful implementation.

Ilias Goranitis (University of Melbourne), "The economic value of genomics"

Slides • Recording

Goranitis consolidated findings from various research papers to construct a compelling case for the value of genomics. This includes not only its significance in direct health outcomes but also its contributions to diagnostic, clinical, personal, and overall process utility.

Michael Mackley (Hospital for Sick Children, University of Toronto), "Defining, refining and reimagining the Clinical Genetics Service in the genomics era"

Slides • Recording

Mackley discussed recognising the need for change in the delivery of genetic services and examining the evolving roles of clinical geneticists. This exploration includes identification of the key factors that make it challenging to effectively demonstrate their value to governments and funding bodies.

Sue Hill (NHS England), “NHS England Genomic Medicine Service: approach to commissioning genomic tests”

Slides • Recording

Hill presented the UK National Health Service (NHS) National Genomic Test Directory, highlighting its innovative commissioning approach and detailing the processes around making updates. A digital Test Directory, strategically aligned with clinical research, is also on the horizon, promising to enhance accessibility and integration in genomic healthcare.

Rob Annan (Genome Canada), “Canadian Precision Health Initiative”

Slides • Recording

Highlighting Genome Canada's efforts to advance genomics research in Canada, focusing on the soon-to be-launched Canadian Precision Health Initiative. Annan emphasised building a large-scale genomic data asset that reflects the country's diverse population, integrating AI, and ensuring ethical governance, with the ultimate goal of transforming healthcare delivery and fostering global collaborations.

Sue Hill (NHS England), “Generation Study”

Slides • Recording

An overview of the Genomics England Generation Study, covering its initial vision, co-design process, regulatory review, and current status within the NHS-embedded research study was shared. The findings of this study will play a critical role in determining the development of a newborn screening program.

Zornitza Stark (Australian Genomics), “GenSCAN and BabyScreen+”

Slides • Recording

GenSCAN includes five research projects on newborn screening, including BabyScreen+. This presentation outlined the project's goals, engagement methods for patients, the public, and healthcare professionals, and insights on key topics such as funding, the inclusion of untreatable conditions in screening results, consent, etc.

Ainsley Newson (University of Sydney), “gEnomics4newborns”

Slides • Recording

Another initiative under GenSCAN, gEnomics4newborns aims to explore the legal, ethical, equity, and economic implications of genomics in newborn screening. Newson detailed its work packages with a particular emphasis on engagement, specifically through community juries and yarning circles.

Kathryn Noth (Murdoch Children’s Research Institute), “Wrap up and next steps”

Slides • Recording

The meeting concluded with a focus on outlining clear, actionable steps for the NIF group to make tangible progress over the next year. Emphasis was placed on the importance of ongoing community engagement, ensuring alignment with community expectations, and supporting project development. The discussion covered strategies for integrating NIF with GA4GH, including the creation of a "Concierge Service" and the revitalization of the NIF Toolbox. Key upcoming initiatives include developing a Maturity Model for national efforts, exploring governance design patterns, and advancing NIF pilot projects such as federated gnomAD and IPCHiP. The importance of identifying concrete steps for GA4GH integration and selecting strategic pilot projects was underscored as crucial to future progress.

Plenary posters

The posters shared at 12th Plenary showcased the work of GA4GH and other related activities that aim to enable the responsible, voluntary, and secure sharing of genomic and health-related data for the broader genomics and health community.

The Plenary Poster Guide contains a full list of poster titles, authors, and abstracts.