Global Alliance for Genomics & Health

GA4GH Connect

2023 Strategic Refresh

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Executive Summary

In 2022, through a series of open-brainstorming sessions chaired by members of the GA4GH executive committee, as well as a written survey, GA4GH solicited more than 300 pieces of feedback from more than 120 individual stakeholders on our current and future ability to achieve the three community imperatives identified during the 2020 Gap Analysis: improve interoperability and alignment with external standards and between GA4GH products, improve implementation support for technical standards, and engage more closely with healthcare and clinical standards. The most recurrent themes among the strategic refresh feedback were recommendations to (i) create a forum for sharing experiences implementing GA4GH standards and mechanisms for implementers to demonstrate usage of GA4GH standards, (ii) work with collaborative projects to identify & advance use cases, and (iii) ensure clinical relevance of GA4GH toolkit before promoting to the clinical community.

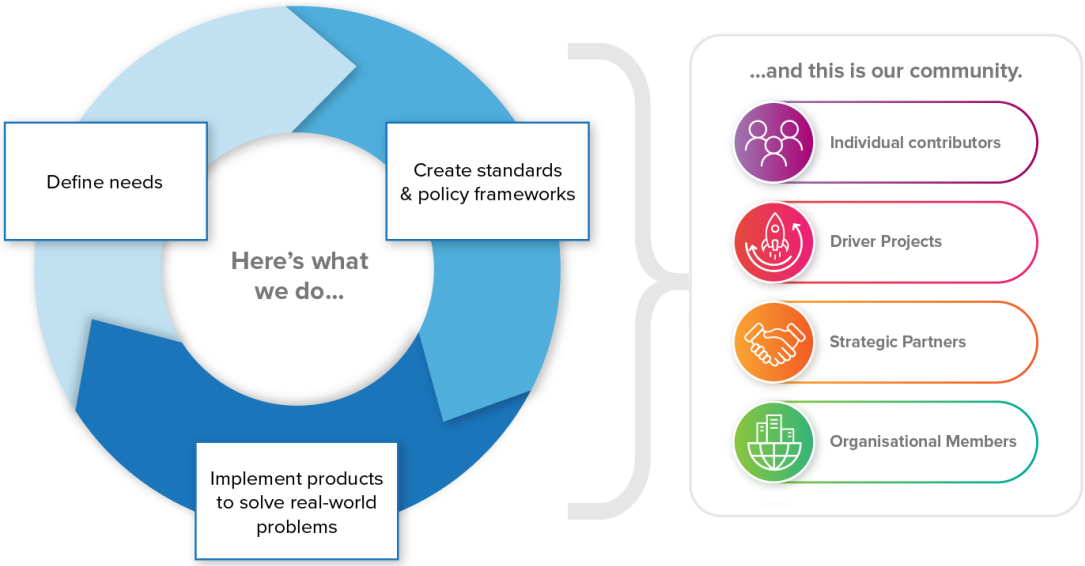
In order to better support these recommendations, GA4GH secretariat herein proposes to formalise — and provide greater resources for — several changes that have naturally emerged over the past five years in both what we do and with whom we engage. For example, GA4GH is no longer a simple matrix of Driver Projects and Work Streams. While this remains the foundational strategy underlying all of our work, we also support activities before and after the work stream development phase and rely on contributions from a much broader community than those reflected solely in the Driver Project cohort.

In this document we present an update matrix that better reflects how we now work as well as introduce a series of new strategies, including (i) term limits for Driver Projects and regular application cycle to bring in new DPs, (ii) more robust guidelines for strategic partnerships, (iii) a definition and process for advancing the previously defined Assigned Experts model, and (iv) a reimagined GA4GH Implementation Forum (GIF), which brings together elements of the former Genomics in Health Implementation Forum (GHIF) and Federated Analysis Systems Project (FASP). We more clearly define the role of National Initiatives within our community and position that work in the context of additional community alignment activities, such as strategic engagement with other Standards Development Organizations (SDOs). We describe an optimised meeting cadence and efforts to drive broader representation among our community. We outline the roles of the various staff teams — project management, technical development, and policy development — and how we aim to deploy them to provide greater support for cross-work-stream communications and technical alignment activities as well as developing robust implementation support resources. Finally, in an appendix, we describe how we believe these updates will directly impact the feedback we gathered in 2022 through the strategic refresh exercise. While some of the more granular details remain to be determined, this document lays out the broad strokes for a refreshed GA4GH strategic plan in 2023.

Introduction

The Global Alliance for Genomics and Health (GA4GH) was launched in 2013 with a mission to to accelerate progress in genomic research and human health by cultivating a common framework of standards and harmonised approaches for effective and responsible genomic and health-related data sharing. In 2017, the GA4GH Connect five-year strategic plan refocused the organisation on technical standards development and policy framing, introducing the concepts of Work Streams and Driver Projects — real-world clinical and research initiatives working in genomics to support real world genomic data interoperability. An accompanying product roadmap was released in 2018, outlining the development plans for more than 23 standards and policy frameworks across eight Work Streams. In 2020, GA4GH underwent a Gap Analysis led by Heidi Rehm (Mass General Brigham; Broad Institute) and Andrew Morris (Health Data Research UK), leading to the identification of three community imperatives upon which GA4GH members believed the organisation should focus its efforts over the coming years: improve interoperability and alignment with external standards and between GA4GH products, improve implementation support for technical standards, and engage more closely with healthcare and clinical standards. In 2022, GA4GH Secretariat oversaw a "strategic refresh" to reflect on our current and future ability to meet the mission, vision, and goals outlined previously.

In this document, we share the feedback gathered during the strategic refresh and introduce a series of process and structural updates that we believe will be critical to the long term success of the organisation and its ability to achieve the original mission and goals, as well as the 2020 community imperatives. To help orient readers, we organise our thinking around the two axes of the GA4GH community: who we are and what we do.



Part I: Strategic Refresh Feedback on the 2020 Community Imperatives

In the following three sections, we review the expectations set in 2020 around each of the three community imperatives and summarise the feedback we received in 2022 on our current and future ability to achieve them.

Interoperability & Alignment

In 2020, the GA4GH gap analysis report stated that effectively driving uptake of GA4GH standards depended upon the development of "an interconnected suite of standards that are compatible and interoperable with each other and hardened for real-world use." We sought to identify alignment opportunities and to support a centralised forum for discussing all ongoing GA4GH technical details. These activities primarily manifested in the Federated Analysis Systems Project (FASP) and the Technical Alignment Subcommittee (TASC), respectively.

Through the 2022 strategic refresh, we learned that more support and broader engagement were needed for both FASP and TASC to achieve their goals. We also heard new ideas, such as the suggestion to bring together groups of disparate Driver Projects to formally work together to implement standards to share data, particularly sensitive human data. Others urged the GA4GH Work Streams to place a greater emphasis on ingesting external use cases, to add formal alignment requirements to the product development and approval process, and to create an issue board for GA4GH-wide, cross-work stream concerns. There was also a suggestion to regularly convene Work Stream leadership to identify integration and alignment challenges and opportunities, perhaps through the existing TASC framework.

Implementation Support

In 2020, GA4GH stated that implementations of standards — particularly those that serve the high priority needs of the community — would be critical to informing the development and "hardening" of existing technical standards. Implementations would also serve to instantiate standards by driving awareness and encouraging adherence through the need to enable downstream, interconnected functions.

To drive broader uptake, 2022 strategic refresh participants suggested we encourage leading stakeholders to set the tone for the community by supporting and promoting GA4GH standards. Many felt that GA4GH should create a forum for implementers to share their experiences engaging with our resources, both to learn from one another as well as to drive improvements in the standards and policies. Journals should be encouraged to include GA4GH in their author guidelines and funders should create specific GA4GH-focused requests for proposals. Another recurring theme in our discussions was the need for a mechanism for users to both test the

success of their implementations and to share their uptake through an associated badge program. We also learned of several improvements we can make to our working practices, including more comprehensive documentation (which should be consistently translated into multiple languages), better processes for users to provide ongoing feedback, and more rigorous review and testing of deliverables prior to approval.

Clinical Engagement

In 2020 we learned that the community wanted us to better engage both the clinical community as well as the standards organisations that support it. While our connection to the research side of the "learning health system"[1] was strong, we acknowledged that the diversity of the global clinical community had limited our ability to interface with the healthcare side of that cycle. We identified several specific barriers standing in our way, including resourcing needs, regulatory limitations, and difficulty in finding the right points of engagement (eg., clinical vendors / healthcare industry vs. practising clinicians).

In 2022, we are still facing the same challenges. Strategic refresh participants told us that the ultimate targets of implementation — individuals with clinical backgrounds and clinical genomic experience in the areas of patient care, clinical laboratory roles, and clinical research — are still underrepresented within the community. They urged us to introduce more clinically-focused Driver Projects, to align with other clinical Standards Development Organisations (SDOs) and professional organisations (such as ACMG, NSGC, ASHG and Medical Genome Initiative), and to work with stakeholders to identify real-world clinical interoperability use cases. Trusted Research Environments (TREs), which provide a safe haven for research use of clinically generated data, arose as important touchpoints. We were encouraged to work with key clinical genomics labs to identify appropriate standards and applications, and with EHR systems to encourage adoption of GA4GH standards (VRS, vcf, etc). Participants asked us to create implementation guidelines for practical use in clinical environments and training materials targeted at clinical stakeholders. Finally, a very specific suggestion was to leverage and build on ClinGen's database of Genomic Analysis Software Platforms Meeting Minimum Requirements for Data Sharing to Support Quality Assurance.

Responding to the Feedback

While we hope to eventually follow up on each of the suggestions that came in through the 2022 Strategic Refresh, we must acknowledge that some will require a longer term horizon than others, some fall outside the scope of GA4GH's remit, and still others would require resources not yet within our reach.

[1] The learning health system refers to a virtuous cycle in which secondary use of patient data feeds into research, and the learnings from research reciprocally inform medical care.

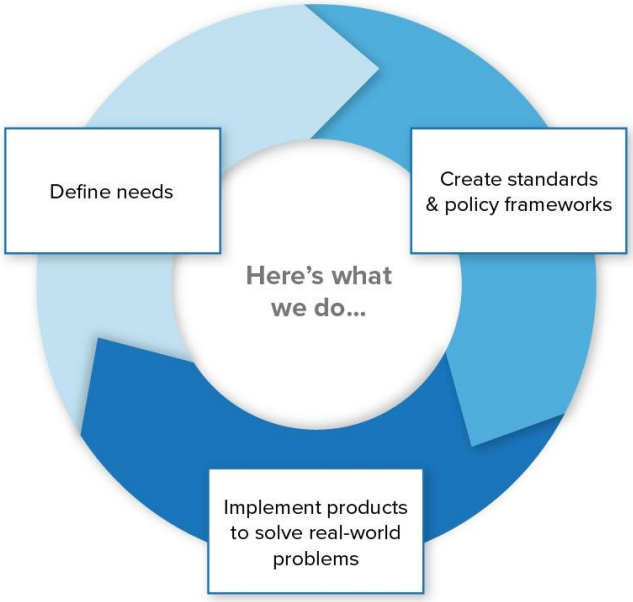
In the rest of this document, we propose a series of organisational updates that we hope will address the most prominent recommendations that continually recurred throughout our 2022 discussions:

- Create a forum for sharing experiences implementing GA4GH standards
- Create mechanisms for implementers to demonstrate usage of GA4GH standards
- Work with collaborative projects to identify & advance use cases
- Ensure clinical relevance of GA4GH toolkit before promoting to the clinical community

Through these changes, we hope that several other recommendations will also be in a better position to succeed. We highlight how each suggestion is related to specific pieces of feedback in Appendix II: How this proposal will impact the community imperatives.

Part II: Updates to What We Do

In 2017, the GA4GH Connect Strategic Plan introduced a matrix organisational structure that connected eight Work Streams to 24 real-world Driver Projects. Work Streams were the spaces within which GA4GH activities were to take place, namely the development of technical standards and policy frameworks. Driver Projects were the real-world initiatives that would use them.



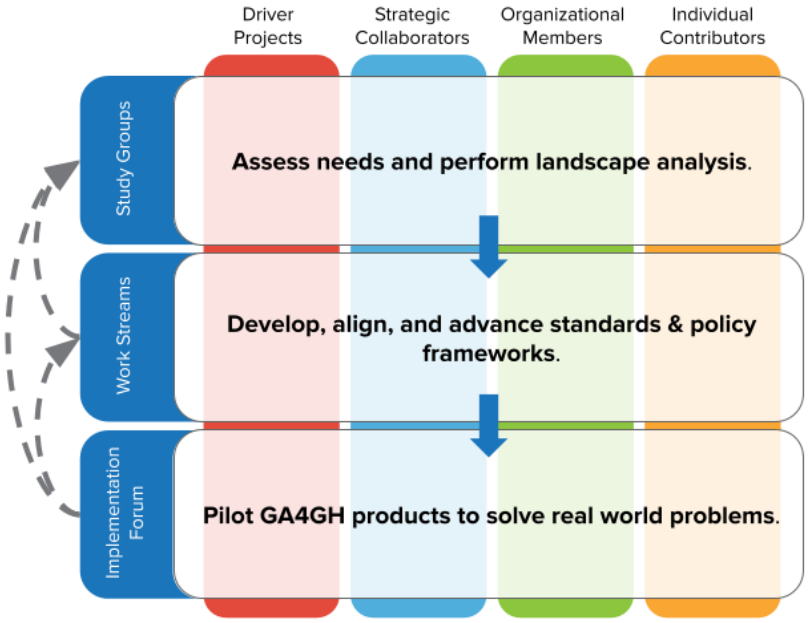
Since that time, the repertoire of activities in which GA4GH stakeholders engage has expanded considerably. While we have focused discussion of how we work on a matrix structure in which Driver Projects are focused on developing standards and policy frameworks in the context of technical and foundational Work Streams, our work now includes activities that come before development commences and after initial approval. In 2023, we aim to better communicate how we work by formally adding two more horizontal categories to the GA4GH matrix: Study groups and the GA4GH Implementation Forum. Together with Work Streams, these spaces will be populated by individuals from Driver Projects, Strategic Partners, and Organisational Members, as well as individual contributors (see Updates to Our Community). We must also emphasise that these three activities feed into one another in a continuous feedback cycle.

Study Groups

As described in the [DRAFT] 2022 update to the GA4GH product development and approval process, study groups are formed to investigate whether a new GA4GH product is needed, could be developed, and would fulfil a need not met by any existing efforts. Study group activities include broad outreach, stakeholder engagement, consultation with the Regulatory-Ethics and Security Work Streams, landscape analysis, use case development, and defining a problem statement. In some cases, Study Groups will be focused on whether or not to update an existing product or to formally recognize work that has already taken place outside of GA4GH.

Study Group Participants

Study group participants include stakeholders from a broad cross-section of the community, including developers and users (eg., researchers, bioinformaticians, clinical stakeholders, and more). Many members of a study group will often go on to form the seed of a Work Stream sub-group.



Work Streams

GA4GH Work Streams develop standards and policy frameworks that are designed by the community to overcome technical and regulatory hurdles to international genomic data-sharing and interoperability.

Foundational Work Streams

Foundational Work Streams provide guidance to Technical Work Streams, Driver Projects, and National Initiatives in the areas of regulation, ethics, and data security in genomics. They both develop standards and policy frameworks identified by the community as immediately relevant, as well as independently establish policies and guidance documents that cut across the organisation and have the potential to shape the field more broadly.

Technical Work Streams

Technical Work Streams develop technical standards that are of most relevance to the practising genomic medicine and research communities. These groups work to harden existing working standards and to create entirely new standards where needed.

Technical Alignment Sub-Committee

Based on feedback from the strategic refresh consultation period, we aim to put more resources and support behind this group to create a space for effective interoperability testing and ensure that GA4GH standards work together in practice. Some of this effort has previously been carried out within the context of the Federated Analysis Systems Project (FASP) and some of it has taken place within the Technical Alignment Subcommittee (TASC).

Both established in 2019, TASC aims to provide a mechanism for generating internal consistency across work streams and deliverables and FASP set out to demonstrate real world interoperability using GA4GH standards in practice.

In the original TASC charter document, the group identified three areas of focus: (i) defining and maintaining core GA4GH "brand" elements (e.g. namespacing, w3id.org, domains); (ii) providing mechanisms to lower the barriers for alignment and improve interoperability such as the creation of a repository of common schema elements, a GA4GH terminology glossary, and mechanisms to enhance portability; and (iii) developing coherent support and resolution procedures for technical issues affecting the whole of GA4GH including GitHub structure and Docker related issues.

In 2023, we aim to provide additional secretariat support to the TASC and to expand its membership. The original charter stated that the group would consist of "at least one" member from each Work Stream; going forward we would like to considerably increase engagement.

FASP will be retired as a name, however its participants and activities will largely continue under the new GA4GH Implementation Forum (GIF). Some of its activities, such as developing implementation support resources, will become the remit of the staff Technical Team and Assigned Experts.

Work Stream Participants

All members of the broad genomics community — whether or not they are affiliated with a GA4GH Stakeholder Organization — who have an interest in using or helping to develop GA4GH standards or policy frameworks are invited to participate in the GA4GH Work Streams. Participation includes attendance at meetings and discussion on the associated Slack channels or Google Group listservs. Participants help ensure development is proceeding in the appropriate fashion to meet users' needs. Often, Work Stream participants will have also engaged in study groups specific to the Work Stream's deliverables. All work streams must have representation from at least two GA4GH stakeholder groups at all times, including at least one Driver Project.

- **Work Stream Leads**
  Each Work Stream has two Work Stream Leads (WSLs). These are individuals with the technical capacity, leadership skills, "community-mindedness", and bandwidth to ensure delivery of agreed-upon milestones at the expected rate. WSLs serve at least one two-year renewable term. (Read more.)

- **Product Leads**
  Each product developed by a work stream will have at least one product lead (PL). These are individuals with both technical expertise as well as bandwidth to ensure timely delivery of product-level milestones. PLs are expected to attend both WS meetings as well as subgroup meetings focused on specific products, which may be more frequent than WS meetings. PLs serve for the duration of the product's initial development or until they choose to step down.

- **Work Stream Contributors**
  Work Stream Contributors (WSCs) are individuals with deep technical backgrounds and sufficient time/resources to actively contribute to Work Streams and subgroups in the development of GA4GH products. WSCs may be participating on behalf of any GA4GH stakeholder (see below) or as an individual contributor unaffiliated with a member organisation.

Advancing products within a Work Stream

The [DRAFT] 2022 update to the GA4GH product development and approval process outlines a complete product development process, beginning with a formal proposal and ending with review and approval by the GA4GH Standards Steering Committee, the Regulatory & Ethics and Security Work Streams, and a specially convened Product Review Committee specific to the product in question. The process gives clear direction around the criteria for decision making at various time points in the development cycle and puts the onus of technical content review on those with appropriate expertise, while relying on the Standards Steering Committee (SSC) to ensure that the process is followed appropriately.

The Shifting Landscape of GA4GH Work Streams

If a Study Group identifies a product that does not have a clear home within an existing Work Stream, it is acceptable to propose the creation of a new one. Such an addition should be discussed with the GA4GH leadership and confirmed by the SSC during the product proposal period.

It is also important to note that the role of a Work Stream may shift over time. In such circumstances, it may be appropriate to introduce new leadership better suited for the current priorities. Similarly, there is no expectation that WSs should persist in perpetuity; if the group decides that it makes more sense to evolve or dissolve, GA4GH will support this so long as a product maintenance plan is well-defined.

Finally, if a Work Stream completes its original remit to develop one or more technical standards or policy frameworks but feels more work is needed to promote uptake or perform necessary testing, but the current Work Stream members and processes do not support these new needs, the GA4GH Secretariat, particularly the Work Stream Managers, will work with existing leadership to develop a transition plan for the Work Stream.

Cross-Work Stream Communication

A major point of discussion within the Strategic Refresh consultation period was the need for more formalised mechanisms for Work Streams to (i) share current and future work and experiences from past work, (ii) ensure harmonisation between standards and policy frameworks, and (iii) ensure technical alignment between technical standards. The TASC is well positioned to address the latter of these requirements, however additional opportunities are needed for the

first two. The GA4GH secretariat staff will aim in 2023 to create more opportunities for cross-pollination between the work streams to ensure discussions do not proceed in silo. GA4GH will hold four hybrid or virtual meetings each year, each of which will emphasise cross-WS coordination to varying degrees. Additionally, we will strive to provide more support for asynchronous cross-work stream discussions as well as regular communications on the website and the GA4GH news blog. Finally, the Best Practices for Working Effectively effort aims to identify opportunities for Work Streams to better engage the international genomics community and we strongly encourage all Work Stream Members to engage with this activity.

Implementation Forum

The 2020 product roadmap included more than two dozen deliverables that were identified as high priority by the current GA4GH Driver Projects. As of 2022, most of these have now been released in V1.0 or greater and GA4GH has specified the necessary elements for a minimal viable protocol for federation: htsget for presenting data with access control; WES and TES for encapsulating and running analyses; DRS for abstracting the data's location; AAI/ Passports for federated authorization and authentication; and Beacon for discovering datasets of interest. While more ambitious goals are on the horizon (eg., matching requests, analyses, and datasets; describing phenotypes; reporting on variants), the 2020 GA4GH Connection Demos, developed by the Federated Analysis Systems Project (FASP) laid the groundwork for federated analysis.

Now, through the launch of the GA4GH Implementation Forum, we aim to identify a series of multi-stakeholder use cases focused on implementing multiple production-ready standards to advance a real-world scientific or clinical problem. GA4GH will support 3-4 flagship use cases at a time that have the following characteristics:

1. A clear remit focused on genomic data interoperability
2. Dedicated resources from at least three distinct global initiatives
3. At least two GA4GH products needed to achieve the stated goal

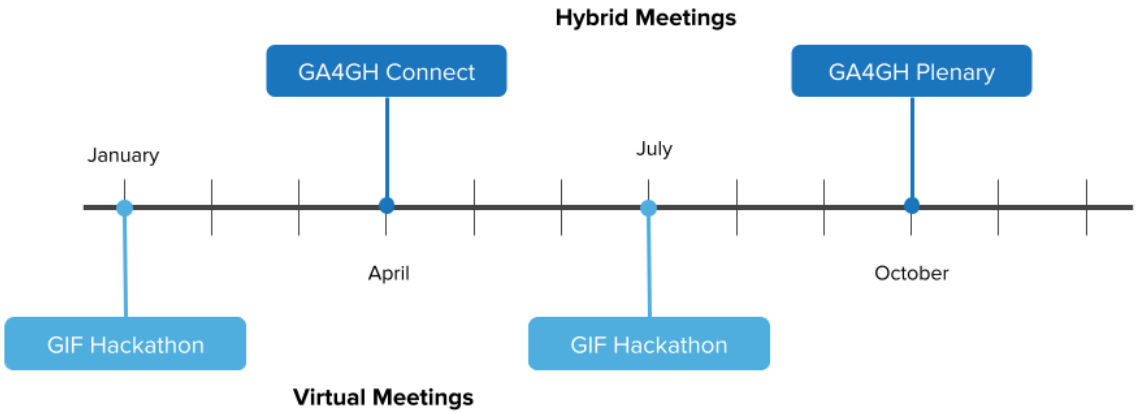
GIF use cases will be identified in multiple venues within the GA4GH community, such as the National Initiatives Forum, Work Streams, GA4GH meetings, and Communities of Interest.

GIF use cases will be advanced using the framework developed for the 2020 Connection Demos, which focused on enabling "third parties to perform real-world, scientific use-cases by connecting GA4GH standards." This framework has three key outputs:

1. Resources to enable the broader genomics community to solve real-world challenges using GA4GH products
2. Demonstration of GA4GH-supported interoperability
3. Insights about using the products in practice, which can be fed back to those developing and/or maintaining them (eg., the GA4GH Work Streams).

GIF Meetings

Two hybrid GIF Meetings will be held each year, in conjunction with GA4GH Plenary and GA4GH Connect, our annual hybrid working meeting focused on cross-workstream development. A bi-annual GIF Hackathon will allow engineers to advance use cases in a virtual setting, between annual meetings. Individual use case teams will define the frequency and regularity with which they meet. GIF use cases will also provide valuable content for the annual GA4GH Plenary Meeting, as evidence of GA4GH standards being used in real-world settings. GIF milestones will be aligned with these four annual meetings in order to drive activity.



GIF Leadership

Each GIF use case will have its own champions and engineers, as defined by the use case stakeholders. A GIF steering committee (SC) of 5-6 individuals will be nominated by the community and appointed by the GA4GH Executive Committee.

Part III: Updates to Our Community

Just as the repertoire of GA4GH activities has expanded since 2017, so has the stakeholder community. The Driver Project model of selecting a small number of projects to serve as proxies for the wider community is — while still critical — no longer sufficient to meet all of our goals. In order to reach a broader cross section of the community through a more scalable mechanism, GA4GH now seeks to add more strategic partnerships similar to that formed with ELIXIR in 2017. These two models (Driver Projects and Strategic Partners) are mirrored by our broader, extended community of Individual Contributors and Organisational Members, whose great value we wish to better and more formally recognize going forward.

Below we describe the characteristics of each group and outline the criteria, terms, and personnel associated with each. We also discuss the different types of individual contributors that make our work possible.



Driver Projects

Driver Projects (DPs) are the primary stakeholders from which GA4GH Work Streams take their direction. Work Streams must ensure that at least two DPs have expressed interest in a new deliverable before it will be added to the roadmap. DPs are real-world initiatives that work in the international genomics landscape and have a vested interest in advancing genomic interoperability.

GA4GH will issue an open call for new DPs annually, beginning in late 2022 / early 2023. Driver Project applications will be evaluated against the following six criteria:

1. Focused on academic or clinical genomic research
2. Mission and goals that depend on real-world genomic interoperability
3. Located in a geographic region that is not yet well represented within GA4GH

4. Scientific merit and representative of the broader community's current foci
5. Dedicated funding
6. Capacity to contribute to GA4GH standards development

Driver Projects will serve at least one three-year term. To be approved for a subsequent term, Driver Projects must submit a letter of intent describing the reasons why the collaboration remains necessary and beneficial to both parties, according to the current year's application criteria.

GA4GH Driver Projects help steer the direction of international genomic data sharing through their central role in GA4GH Work Stream activities. With this benefit comes the responsibility of accurately representing the needs of the broader genomics community.

Each Driver Project is allotted ONE (1) vote on the GA4GH Standards Steering Committee (SSC).

Driver Project Champions

Driver Project Champions are individuals with domain knowledge and a strong understanding of the deliverables needed by their organisation to enable data sharing. Each Driver Project must contribute at least two and not more than four Driver Project Champions (DP Champions), who hold leadership positions within their projects and are able to support the implementation of completed GA4GH tools therein. Driver Project Champions are expected to:

1. Act as 'team leaders' overseeing the Driver Project's participation with GA4GH;
2. Participate in two in-person meetings per year, where they may be asked to present information about the DP and its progress and to provide input on the development of Work Stream deliverables;
3. Participate in quarterly SSC meetings (two in-person, two virtual), and present DP updates on one SSC meeting;
4. Provide high-level, expert guidance and feedback on relevant GA4GH products to ensure they meet the DP's needs;
5. Appoint Driver Project Contributors to Work Streams of particular interest to the DP, and engage those Contributors for updates on progress.

Driver Project Contributors

Driver Project Contributors (DP Contributors) are Work Stream Members who represent a particular DP. DP Contributors have deep technical, ethical, or regulatory backgrounds and sufficient time and resources to actively contribute to Work Streams in the development of GA4GH products. Driver Project Contributors are expected to:

1. Join Work Stream calls or asynchronously represent the Driver Project's needs/priorities (frequency will vary across Work Streams);
2. Provide active, hands-on effort in the framing and development of standards and policy frameworks;
3. Review Work Stream deliverables and give input;

4. Act as an intermediary between the granular work of the Work Streams and the high-level guidance of the DP Champions;
5. Keep DP Champions informed of Work Stream development activities as needed.

Ensuring diversity and representation

In order to meet the expectations set by the 2022 strategic refresh participants, GA4GH will strive for an equal balance between academic and clinically focused research projects in its DP cohort. We will also strive for broader global representation, reserving slots for projects located in areas of the globe that are under-resourced in genomics and Low and Middle-Income Countries (LMICs).

Strategic Partners

GA4GH Strategic Partners are sustainably-funded, broad-based consortia with an aligned mission to that of GA4GH. They are distinct from Driver Projects in that they provide a single touchpoint into a wider network, with the potential to increase GA4GH's global reach. Strategic Partners also provide GA4GH with insights into their network's standards rollout plans and assurance that standards and tools are fit for use within that network.

GA4GH Strategic Partners have the opportunity to help steer the direction of international genomic data sharing, in particular by providing direct, strategic input to the GA4GH leadership. The relationship also provides the Strategic Partner the ability to drive standards enhancements that align with their institutional goals.

The first SP — ELIXIR, the European infrastructure for life sciences — was added in 2017. In 2023, we aim to add more Strategic Partners as necessary on a rolling basis. Below we provide clarity around the definition of and process for becoming a GA4GH SP.

Strategic Partners must have the following characteristics:

- Has secure, long-term funding and/or a robust sustainability model
- Provides services to an international genomics and health community
- Is philosophically aligned with GA4GH's mission and vision
- Participates in GA4GH activities (Study Groups, Work Streams, GA4GH Implementation Forum)

Each Strategic Partner is allotted ONE (1) vote on the GA4GH Standards Steering Committee (SSC). If the Strategic Partner is associated with one or more GA4GH Driver Projects (DPs), the SP vote is additional to those DP vote(s).

Each Strategic Partner will define the structure and level of formality of the relationship with GA4GH Inc. Strategic Partners will aim to serve in their capacity for at least five years and will strive to promote GA4GH standards uptake across their networks, such as through internal training activities.

To initiate a strategic collaboration, the leadership of the Strategic Partner and of GA4GH must agree on the need for the collaboration and then work together to draft a public document outlining the relationship.

Organisational Members

All organisations that participate in GA4GH are expected to sign on as Organisational Members. The application is lightweight and only requires that the organisation agree to the terms of the GA4GH Constitution and the *Framework for Responsible Sharing of Genomic and Health-related Data*, as described in the Membership Agreement.

GA4GH Organisational Members must have all of the following characteristics:

- Mission, operations, and public statements are consistent with the GA4GH Constitution and Member Agreement
- Is an established entity (e.g., corporation, not-for-profit Organisation, partnership)
- Has an open and professional presence (e.g., website)
- Is active in the field of genomic research/medicine or related activity
- Ownership, governance, funding, and leadership are clearly defined and accessible

While organisational members are not represented on the Standards Steering committee, all products are made available for open comment for one month prior to the voting period and all GA4GH stakeholders are encouraged to share their feedback during that time. Additionally, Driver Projects are intended to serve as proxies for this broader community.

All Driver Projects, and Strategic Partners must first be GA4GH Organisational Members before being approved for the additional credential.

Contributors, Implementers, and Observers

Organisational members fall into one of three categories: implementer, contributor/ implementer, or observer. Implementers are those who wish to adopt GA4GH standards but are not able to participate in their development. Contributor/implementers both participate in the development of standards and policy frameworks, as well as implement them in real-world practice. Observers are those who wish to support the mission and vision of GA4GH but are not in a position to either contribute to or implement GA4GH deliverables. By definition, only implementers and contributor/implementers are able to participate in GA4GH activities. ***In order to encourage broader participation among our Organisational Members, we will aim in 2023 to recognize those that are actively contributing or implementing GA4GH products on our website.***

Individual Contributors

The GA4GH contributor community currently consists of ~1,400 individuals, who can be characterised as Work Stream Members, Work Stream Contributors, Funded Contributors, and Assigned Experts, as described below. By definition, Work Stream Participants and Driver Project

Champions are all considered Work Stream contributors at a minimum; some may also be funded contributors depending on their individual circumstances.

Work Stream Members

Work Stream Members are individuals that have expressed interest in GA4GH development activities and have been added to a GA4GH Google Group.

Recognition: Work Stream members are not recognized on the GA4GH website.

Work Stream Contributors

Work Stream Contributors are Work Stream Members who have made meaningful contributions to GA4GH standards or policies, for example by joining Work Stream calls, reviewing or developing documents, or developing code.

Recognition: Active and former Work Stream Contributors are listed as "contributors" on the GA4GH webpage(s) for the product(s) to which they have contributed.

Funded Contributors

Funded Contributors are Active Contributors who have received funding to support their work on GA4GH standards development.

Recognition: Funded contributors are listed as "contributors" on the webpage(s) for the product(s) to which they have contributed. Funders of Funded Contributors are not recognized on the GA4GH website.

Assigned Experts

Assigned Experts are funded contributors who have been specifically assigned by their employer (at a level of 30% personnel effort or more) to contribute to GA4GH standards development (see below for more details).

Recognition: As members of the GA4GH Funders Forum, funders of Assigned Experts are recognized on the GA4GH website. Assigned Experts are listed as "Assigned Experts" on the webpage(s) for the product(s) to which they have contributed.

Assigned Experts

GA4GH core funding is provided by public and private agencies in the US, the UK, and Canada, which is channelled to the four GA4GH Host Institutions, themselves located in those same three countries. In an effort to provide (i) opportunities for more countries to engage with GA4GH at the core level and (ii) a mechanism for underrepresented populations to meaningfully engage with GA4GH, we have developed the Assigned Expert model. This allows national agencies that cannot or do not wish to direct funds outside of their country to have a "dotted line" into the GA4GH staff technical and policy teams.

Assigned Experts are individuals assigned by a national funder (or representative local institution) to commit at least 30% of their time (four calendar months/year) to GA4GH standards development as an official part of their job function while also advocating and advancing local priorities within GA4GH.

Assigned Experts are expected to:

1. focus a documentable portion (30% or more) of their official efforts on GA4GH institutional priorities around standards and policy development.
2. work on behalf of a national or regional genomics landscape, providing an otherwise unmet strategic connection for GA4GH.
3. directly and regularly engage with the GA4GH staff policy and technical teams.

Process for Establishing an Assigned Expert

To become an Assigned Expert, an individual's employer must work with GA4GH leadership to establish the need and validate the appointment. Assigned Experts that are not first discussed with GA4GH leadership may not be recognized by GA4GH as such. Assigned Expert employers are expected to find funding for the appointment, or, conversely, funders of Assigned Experts are expected to secure a local employer to house the expert — these are not the responsibility of GA4GH staff or leadership.

To validate one's contribution as an Assigned Expert, an individual or their employer or funder must be able to provide evidence (on an annual basis) of the individual's commitment to GA4GH standards and policy development and will work with GA4GH staff to ensure that their efforts are aligned with institutional development priorities. An evaluation tool will be provided in early 2023 to validate Assigned Experts and ingest supporting documentation.

Part IV: Strategies for achieving our vision

GA4GH Secretariat

Work Stream Management

Work Stream Managers (WSMs) play a critical role in setting the goals and priorities of the Work Streams, as well as the GA4GH Executive and Steering Committee. Responsibilities include community management, strategy development and implementation, and programme management of several international task-teams and knowledge exchange projects from concept development through to external communications and continued development.

In addition to supporting the assigned Work Streams, WSMs are expected to represent GA4GH at the national and international level, and represent GA4GH projects, teams, and initiatives in international collaborations and national genomics programmes. WSMs also help promote adoption and implementation of GA4GH products, recruit industry and academic partners, and act as a communication channel for the genomics community's technical and policy needs into the Work Streams.

Technical Team

The GA4GH Technical Team (Tech Team) consists of software developers and technical writers/documentation managers who help support the widespread development, maintenance, uptake, and longevity of GA4GH technical standards. Led by a Chief Standards Officer (CSO), the Tech Team develops tools and services that enhance the GA4GH standards ecosystem, increasing visibility and reducing the barriers for newcomers to understand and adopt GA4GH products. Such tools include reference implementations (see starter kit below), testbeds, registries, libraries, and technical training modules. Developers leverage key relationships with other organisations to integrate GA4GH standards in widely used, open source bioinformatics toolkits and libraries. All software supported by the Tech Team is open-source and freely accessible to the research community, aside from small, private tools that accelerate the team's internal development processes. Developers do not write proprietary software for GA4GH stakeholders, but may write open software with the intent that it is usable by a number of implementers.

It is our goal to add significant capacity to the tech team in 2023, in particular in the area of technical writing and documentation management. This area of the team's work will include ensuring uniform documentation quality across all GA4GH technical standards, maintaining documentation on work stream roadmaps, community-driven use cases, and implementation guides.

The Tech Team works on projects that align with the plans outlined in our grant applications and works closely with the GA4GH Work Streams and expert contributor network to ensure that their

projects are tightly aligned with community needs and the overall GA4GH roadmap. The team also helps to identify interoperability problems between new and existing standards, and help to correct these issues to promote comprehensive technical harmonisation across all GA4GH products. Also, as GA4GH-compliant services can be viewed as a large, federated network, the technical team will maintain awareness of the network and ensure services and resources are findable for the wider genomics community. More details on the GA4GH Technical Team can be found in the GA4GH Tech Team Charter document.

Starter Kit

The GA4GH Starter Kit is a suite of open-source microservice applications developed by the GA4GH Tech Team, each acting as a server reference implementation of an approved GA4GH API Specification. Currently, there are four completed Starter Kits: **DRS Starter Kit**, which provides an indirection layer to datasets stored in the cloud and serves data according to the GA4GH Data Repository Service (DRS); **WES Starter Kit**, which enables the remote submission, monitoring, and cancelling of computational workflows and is based on the GA4GH Workflow Execution Service (WES); **Data Connect Starter Kit**, which enables discovery of datasets and search of data by allowing the researchers to run SQL queries; and **Passport Starter Kit**, both UI and broker, which enables researchers to request required visas and provide a passport token minted with the selected visas. In addition to these core services, a series of Starter Kit support applications provides additional utility in terms of interactivity and maintainability. These resources include a **GA4GH Starter Kit User Interface** for viewing, creating, editing, and deleting GA4GH models served by starter kit web services; **GA4GH Starter Kit Utils**, a command-line tool for performing setup and maintenance operations on starter kit services and databases; **GA4GH Starter Kit Common**, a utils library imported by all starter kit core apps; and **GA4GH Starter Kit Docs**, a static website where users can find a quick start tutorial and a series of concepts and guides for engaging with the starter kit resources in addition to the official documentation for each of the completed Starter Kits.

GA4GH Scripts

GA4GH Scripts are snippets of end-to-end client code written by the GA4GH community to make API calls to real-world data platforms that have implemented GA4GH API specifications. These scripts give users an out-of-the-box resource for accessing, discovering, and analysing controlled data in a standardised, federated pattern using GA4GH standards. They may be applied directly, or adapted to the specific data and analysis the user wants to perform. Users can leverage GA4GH scripts to run workflows locally on data accessed via GA4GH APIs or remotely at the locations where GA4GH APIs reside, and aggregate results from multiple locations. We also encourage users to share their adapted scripts with the broader community via GitHub.

GA4GH scripts also serve to validate that GA4GH APIs can be strung together to accomplish end user tasks before platform implementers embark on their more costly task: extending the ability to orchestrate federated data and services to scientists who do not write scripts.

GA4GH API Tutorials

GA4GH API Tutorials aim to promote awareness and uptake of the GA4GH standards that enable federated data sharing. Tutorials enable (i) researchers to learn how to seamlessly access data from multiple sources in an international network using common tools and protocols, and (ii) data providers to securely share data with trusted researchers while still maintaining ownership and control over their data. Run by the Tech Team and GA4GH Work Stream Contributors, these tutorials allow attendees to explore GA4GH standardised API interfaces to do such things as access information about files that can be used as input to analytical workflows; remotely run workflows defined in Common Workflow Language (CWL), Workflow Description Language (WDL) or Nextflow on an input dataset of interest; or search and filter biomedical datasets based on criteria of interest. In the first section of a GA4GH API tutorial, GA4GH Scripts are used to search and obtain access to controlled data that reside in real-world data platforms that have adopted API standards (e.g., NIH, Seven Bridges, ELIXIR, DNAstack). In the second section, attendees deploy a server implementation network using the GA4GH Starter Kit and use it to run the same search, access, and analyse protocol practised in section one.

Policy Team

The GA4GH Policy Team consists of dedicated policy analysts and research assistants who support the ongoing development of GA4GH regulatory and ethics deliverables, as well as providing guidance and expertise to the community in a number of key areas. The team provides GA4GH leadership with advice and expertise on where and how GA4GH can influence national and international discussions on policy and, where appropriate, they lead or collaborate on the development of position statements and consultation responses. The team also develops stakeholder/network relations, particularly within the genomics and clinical communities and those making policy for those groups, to ensure early sight of emerging policy issues relevant to the work of GA4GH. They also support the GA4GH Product Approval Process (by both completing the regulatory and ethics review of new products and developing process resources), the development of internal GA4GH policies (eg. Code of Conduct, Patent Policy), and the development of regulatory and ethics implementation resources for other Work Stream deliverables, as appropriate.

Representation and Inclusion

The EDI Advisory Group was launched in May 2020 with the goal of recognizing and responding to EDI issues raised within the GA4GH community. This group supports GA4GH by finding equitable and inclusive ways to bring diverse ideas into the standards creation process. Pathways to EDI that the group explores include:

1. Identifying opportunities to attract diverse talent to the GA4GH community;
2. Helping new and existing contributors from all backgrounds feel welcomed and valued by the community; and
3. Ensuring equitable access to leadership and speaking opportunities within GA4GH.

The EDI Advisory group follows a framework that we call “reflected in our teams, reflected in our standards”. This framework is based on the idea that if EDI concepts are incorporated successfully into our teams, then the interests of the broader community are more likely to be reflected in the standards development process and in the standards themselves. Below we outline current activities of the EDI Advisory Group. In 2023, we aim to drive greater participation in this group from the active GA4GH contributor community, as the primary group impacted by this work.

Onboarding

The GA4GH Equity, Diversity, and Inclusion (EDI) Advisory Group aims to ensure our products are developed by and fit the needs of the broadest possible stakeholder community. The GA4GH Onboarding Programme aims to help newcomers feel welcomed and have the knowledge needed to effectively participate in GA4GH. The program will pair newcomers to a specific Work Stream with an Onboarding Guide who will help them become oriented to the Work Stream. Onboarding Guides help GA4GH newcomers to better understand Work Stream-specific content, providing networking support and background information relevant to the working discussions.

During an initial meeting with an Onboarding Guide, a newcomer is able to share their expertise and goals for participating in GA4GH, learn more about the Work Stream’s priorities, milestones, and decisions, as well as details on how the group functions. At the newcomer’s first Work Stream meeting, the Onboarding Guide will ensure that the newcomer is introduced to the rest of the group, help amplify the newcomer’s voice by helping to put comments into context and/or support/reinforce comments made, help the newcomer find points of connection between the current discussion and others within the community, and seek out the newcomer’s views on specific topics, to facilitate their engagement/participation. Over the longer term, the Onboarding Guide is available for the newcomer to reach out to as needed with additional questions or introductions to help deepen their engagement with others in the community.

Our goal with the Onboarding Programme is to provide a safe and comfortable mechanism for GA4GH newcomers to optimally orient themselves into our often complex environment.

Best Practices for Working Effectively

The Equity, Diversity and Inclusion Advisory Group began the Work Stream Best Practices project in 2021 with a workshop aimed at engaging all Work Stream and Subgroup leads. The purpose of this kick off workshop was to collect current Work Stream Best Practices for creating an inclusive working environment that facilitates diverse participation. Best Practices were collected from the

community over a period of time and later categorised by members of GA4GH secretariat. The following seven topic areas emerged from this effort; (i) identifying missing perspectives, (ii) inviting people to participate, (iii) welcoming and onboarding newcomers, (iv) chairing inclusive meetings, (v) ensuring opportunities for engagement, (vi) recognizing and defining contributions, (vii) keeping track of progress, and (viii) communicating decisions/discussions.

In addition to continuing to collect best practices, the next step for the group is to assist members of the GA4GH community who may need extra support in either developing or implementing existing best practices. To support this process, an interactive platform is being developed to host all GA4GH WS Best Practices. This platform will allow users to both submit and request best practices, provide a visual breakdown of current best practices, and provide users with the option to filter best practices based on topic areas. At the end of the project, all GA4GH WS Best Practices will be hosted on ga4gh.org.

Community Alignment

In addition to the three primary activities of the GA4GH matrix — study groups, work streams, and implementation forum — GA4GH serves as an important convening body in the field of genomics and data interoperability. Below we outline the various efforts currently underway to ensure we are effectively meeting the needs of the broader genomics community and driving uptake of GA4GH standards across the international genomics landscape.

National Initiatives Forum

The GA4GH National Initiatives Forum aims to support more accurate data interpretation, diagnosis, and innovative solutions across healthcare through global cooperation in data sharing and clinical implementation of genomics. The forum helps initiatives reduce both duplication of effort and promote systems integration and the collection of compatible clinical genomic data and health information to enable global data sharing.

The NI forum is primarily focused on supporting uptake of GA4GH standards within large-scale genomics initiatives and providing real-world insights to the GA4GH Work Streams as they develop new standards (and improve existing standards) to support data sharing and interoperability. The group also enables collective learning by sharing best practices, challenges, and opportunities with a technical, regulatory, clinical, or educational focus.

The NI Forum has three primary goals:

- Identify areas of collaboration and resource/expertise sharing
- Identify real-world implementation use cases for global data sharing
- Identify common needs for standards across initiatives

GA4GH National Initiative Forum members have the following characteristics:

- Has secured long-term funding

- Focused on advancing a large-scale genomics strategy across a health system
- Actively working to adopt GA4GH standards

National initiatives must apply and be approved to become a member of the GA4GH National Initiatives Forum and will be expected to dedicate a to-be-determined minimum amount of personnel effort to GA4GH activities.

NI Forum members are invited to two in-person meetings per year, held in conjunction with GA4GH Plenary and GA4GH Connect meetings. This is a time for Forum members to share experiences of implementing genomics at a national scale and to learn from others who are doing the same. The NI Forum will also help steer the course of GA4GH standards development and optimization through their participation in the GA4GH Implementation Forum, Study Groups, and Work Streams.

Communities of Interest

Communities of Interest are groups of like-minded initiatives and stakeholders from across the globe who are interested in using GA4GH standards to advance real-world scientific or clinical use cases in a specific disease area. Communities of Interest represent a deliberate effort to identify standards gaps and implementation opportunities. GA4GH will hold space at all hybrid and virtual meetings for Communities of Interest to convene, should they wish to do so. Communities are also encouraged to meet virtually throughout the year according to their members' needs and expectations.

The current Communities of Interest are:

Rare Disease Community

Launched at GA4GH 10th Plenary in 2022, the GA4GH Rare Disease Community is initially focused on developing use cases in the areas of standardised phenotype collection, variant matching implementation, and building knowledge for the phenotypic spectrum of a gene in order to advance rare disease diagnosis and treatment.

Cancer Community

Founded in 2020, the GA4GH Cancer Community aims to identify and develop real-world use cases for using and advancing GA4GH standards in support of oncology research and clinical care. The group is currently focused on developing use cases in the areas of cancer variant representation, variants of uncertain significance (VUS) interpretation, and methods for acquiring data on rare tumours such as paediatric intracranial germ cell tumours.

Infectious Disease Community

The GA4GH Infectious Disease Community convenes international groups focused on advancing human genomics for the purpose of treating and diagnosing infectious disease. It interfaces and collaborates with the pathogen genomics community, specifically through a partnership with the

Public Health Alliance for Genomic Epidemiology (PHA4GE). The community was founded in 2022 and is currently in the process of collecting focus areas for advancing use cases. A current topic of interest is exploring the potential for standardisation amongst experimental metadata.

Alignment with Other Standards Development Organisations

In addition to GA4GH, there are a number of standards development organisations (SDOs) working to set standards for human genomics, such as the International Organisation for Standardisation (ISO) Health Informatics Sub-Committee for Genomics Informatics (TC215/SC1), Health Level Seven (HL7) and the Clinical Data Interchange Standards Consortium (CDISC). As an early work product of the Sub-Committee, ISO TC215/SC1 launched a task force to map the genomics standards work items underway in each of the above-mentioned SDOs. This mapping will inform a cross-SDO roadmap for genomics, identifying alignment of current activities and gaps, as well as opportunities for collaborative standards development. Participating in these alignment activities will reduce duplication of efforts and amplify the impact of standards, promoting a truly global infrastructure that benefits research and healthcare outcomes.

Global Policy Forum

There are many international health research consortia that generate or use genomic and clinical samples and data, and have established working groups to address ethical, legal, and social implications (ELSI) arising during their work.

GA4GH has begun to align with these ELSI working groups in effort to to work synergistically to share resources, learn from each other's work, and collaboratively identify and address outstanding challenges.

Together with ELSI working groups from the Public Health Alliance for Genomic Epidemiology (PHA4GE), the International Common Disease Alliance (ICDA), the Global Genomic Medicine Collaborative (G2MC), and the International HundredK+ Cohorts Consortium (IHCC), and the Human Genome Organisation (HUGO), GA4GH participates in this inclusive environment that is open to all ELSI working groups from relevant networks and consortia.

The groups held their first online meeting on 1 June 2021 and its first in-person meeting on 21 September 2022, where participating working groups provided an overview of their ongoing programmes and began collating sharable resources to reduce duplication of efforts. An agreement was reached to continue to work together through the forum, with a proposal to continue with an informal online meeting of the community one to two times per year. The first collaborative effort will be a publication and/or report that will be of use to funders and policy makers to assist with understanding the landscape of work ongoing in ELSI working groups related to genomics.

Annual Meetings

GA4GH Plenary

Since 2014, annual GA4GH Plenary Meetings have convened a diverse set of stakeholders to address the complexities of sharing genomic data across traditional bounds. Attendance at these meetings has steadily grown, with the virtual 9th Plenary Meeting in September 2021 welcoming over 500 participants. Following the SARS-cov-2 global pandemic, all GA4GH Plenary Meetings are now held as hybrid events, with all sessions being live streamed. Associated working meetings welcome engaged participation from the Work Stream community via Zoom.

GA4GH Connect

First launched in 2017 as part of the strategic planning effort, GA4GH Connect Meetings bring together GA4GH's most active contributors to advance technical standards and policy frameworks; identify emerging needs for genomic and health data sharing; and address any challenges in implementing GA4GH products. Connect Meetings are also the primary opportunity for advancing cross-interaction between Work Streams. Going forward, Connect Meetings will also reserve space for the National Initiatives Forum and the Communities of Interest to convene and identify new opportunities and goals.

GIF Hackathons

As described above (see GIF Meetings), GIF hackathons are 7-14 day events that focus on improving GA4GH APIs and other standards, and implementing GA4GH specifications in new environments. Hackathons are composed of multiple topics, which are put forth by GA4GH contributors. Such topics aim to either clarify and/or formalise a new feature in one or more existing GA4GH specifications that will enable more research use cases and/or develop/deploy implementations of GA4GH standards to facilitate a novel, federated use case. GIF Hackathons will welcome the entire GA4GH community, but secretariat support will be focused on the flagship GIF use cases described above.

Appendix I: Summary of Past Strategic Planning

GA4GH Connect (2017)

GA4GH Connect laid out a strategic approach focused on connecting standards and policy developers with real-world users to ensure fit-for-purpose utility and broad uptake. In particular, it focused on adoption and implementation by organisations around the globe and projects with real-world genomic data to share. GA4GH strategically partnered with active genomic medicine and research initiatives ("Driver Projects") to create the policies and standards that would meet their data sharing needs, as exemplars of the broader global genomics community. The Driver Projects would come to play a crucial role in determining the policies, standards, and tools on which GA4GH would focus its development efforts over the coming years. The plan envisaged that embedding Driver Projects within Work Streams would increase the likelihood that completed policies and standards would meet the varying needs of the broader genomics community. These stakeholders worked together to develop an accompanying product roadmap, released in 2018. The plan also pointed to a need for proactive engagement at national and organisational levels to ensure that GA4GH standards are easily accessible and ready for use for the broader community.

Gap Analysis (2020)

The 2022 gap analysis included a bottom-up approach with Work Streams defining roadmap updates to support their respective efforts, as well as a top-down process engaging aligned sets of Driver Projects to gather input on areas of need that were not being adequately addressed by existing or planned GA4GH products. This was followed by re-engagement with Work Streams to incorporate the collected feedback into their plans. The effort culminated in a summary document outlining the aggregated feedback, an updated product roadmap (which now included more than 30 deliverables), and a revised strategic roadmap, which outlined the three community imperatives identified by the GA4GH community (improve interoperability and alignment with external standards and between GA4GH products, improve implementation support for technical standards, and engage more closely with healthcare and clinical standards).

Strategic Refresh (2022)

Many of the activities identified in 2020 to advance the three community imperatives have been well underway for some time, including the Technical Alignment Subcommittee, the Federated Analysis Systems Project, the Genomics in Health Implementation Forum (GHIF), and more. Nevertheless, the GA4GH executive leadership decided to support a strategic "refresh" to ensure our work is meeting the needs of the community and that our existing organisational structures and practices are optimised for the community's current priorities. In January 2022, the Secretariat kicked off a series of open-brainstorming sessions ("Town Hall Meetings) chaired by members of the executive committee across multiple time zones and invited the entire genomics

community to provide feedback on our activities and working practices. These sessions were specifically focused on soliciting suggestions for improving our efforts toward the three community imperatives identified in 2020. Through these discussions, as well as a written survey, we collected over 300 pieces of feedback from more than 120 individual stakeholders.

Appendix II: How this proposal will impact the Community Imperatives

In this section we identify the areas of the 2022 Strategic Plan that aim to address specific pieces of feedback heard during the Strategic Refresh consultation process.

Interoperability & Alignment

- ***Better support and engagement FASP and TASC, the existing centralised forums for discussing ongoing GA4GH technical details.***
    - *While FASP will be retired with the introduction of this strategic plan, its efforts have laid a strong foundation for both the Interoperability Testbed as well as the GA4GH Implementation Forum.*
- ***Bring together groups of disparate Driver Projects to formally work together to implement standards to share data, particularly sensitive human data.***
    - *The GA4GH Implementation Forum will bring together groups of Driver Projects and other GA4GH stakeholders to work together on real world problems.*
- ***Place greater emphasis on ingesting external use cases***
    - *Introduction of Study Groups,* which provide a mechanism for early exploration into overlap and synergy with other standards and work streams.
    - *Introduction of the GA4GH Implementation Forum.*
- ***Add formal alignment requirements to the product approval and development process***
    - *Introduction of Study Groups*
    - *Updates to the Product Development and Approval Process*
- ***Create an issue board for GA4GH-wide, cross-work stream concerns.***
    - *This has not yet been addressed, however we expect additional support to the Technical Team to provide a route for this, or a similar technical solution for considering cross-work stream concern, to emerge.*
- ***Regularly convene Work Stream leadership to identify integration and alignment challenges and opportunities, perhaps through the existing TASC framework.***
    - *Introduction of the Interoperability Testbed*

Implementation Support

- ***Provide more opportunities for individuals with clinical backgrounds and clinical genomic experience to engage with GA4GH***
    - *Updated Driver Project model with regular open calls for new projects beginning in 2023, will include specific outreach to the clinical community to ensure better representation therein.*

- ***Encourage leading stakeholders to set the tone for the community by supporting and promoting GA4GH standards.***
    - *Introduction of Strategic Partner mechanism and targeted outreach in the open call for Driver Projects*
- ***Create a forum for implementers to share their experiences engaging with GA4GH resources, both to learn from one another as well as to drive improvements in standards and policies.***
    - *Introduction of the GA4GH Implementation Forum and Interoperability Testbed*
- ***Encourage journals to include GA4GH in their author guidelines***
    - *Strategic Partner mechanism*
    - *While not discussed above, GA4GH will continue to pursue its role as a thought leader and engage in discussions with journals*
- ***Encourage funders to create specific GA4GH-focused requests for proposals***
    - *While not discussed above, GA4GH will continue to engage with funders to develop opportunities for the community that help promote interoperability and data sharing*
- ***Create a mechanism for users to both test the success of their implementations and to share their uptake through an associated badge program.***
    - *Introduction of the GA4GH Implementation Forum and Interoperability Testbed*
- ***Make documentation more comprehensive and accessible (e.g., by consistently translating into multiple languages)***
    - *Expansion of Tech Team and introduction of technical writing expertise therein*
- ***Create better processes for users to provide ongoing feedback***
    - *Users will have more opportunities to provide feedback to GA4GH through their participation in the GA4GH Implementation Forum.*
- ***Create a mechanism for more rigorous review and testing of deliverables prior to approval***
    - *Updates to the Product Development and Approval process (external link)*
- ***The updated GA4GH Strategic Plan outlines additional mechanisms for improved implementation support:***
    - *Starter Kit resources make it easier for GA4GH standards*
    - *Assigned Experts*
    - *Best practices, onboarding,*
    - *DP spots reserved for LMICs as mechanisms to ensure better support for lower resourced communities*

Engaging Healthcare and Other SDOs

- ***Introduce more clinically-focused Driver Projects***

    - *The open call for new Driver Projects will reserve a minimum number of spots for clinical research projects*
- ***Align with other clinical Standards Development Organisations (SDOs) and professional organisations (such as ACMG, NSGC, ASHG and MGI)***
    - *GA4GH has joined a multi-stakeholder community of standards development organisations working in the area of genomics to ensure alignment between disparately produced standards.*
    - *The Strategic Partners mechanism provides an opportunity for other SDOs as well as professional organisations to formally engage with GA4GH*
- ***Work with stakeholders to identify real-world clinical interoperability use cases.***
    - *The National Initiatives Forum provides a strong intersection with national health care services that are focused on system-wide clinical genomics implementation.*
    - *The GA4GH Implementation Forum will provide an opportunity for groups of multiple GA4GH stakeholders to identify and advance real world use cases, including in the area of clinical interoperability*
- ***Engage with Trusted Research Environments (TREs), which provide a safe haven for research use of clinically generated data***
    - *While not discussed above, members of the GA4GH leadership and broader community have already begun discussions in this area and will continue to explore opportunities.*
- ***Work with key clinical genomics labs to identify appropriate standards and applications and with EHR systems to encourage adoption of GA4GH standards.***
    - *We will encourage Clinical Genomics Labs and EHR systems to join as new Driver Projects and will reserve a minimum number of spots for clinical research projects.*
    - *The Strategic Partners mechanism provides a new, additional opportunity for these groups to engage with GA4GH*
- ***Create implementation guidelines for practical use in clinical environments***
    - *While not described above, this is a common practice among our Clinical and Phenotypic Data Capture Work Stream, which will consider this feedback in its future roadmapping activities.*
- ***Create training materials targeted at clinical stakeholders***
    - *While not described in above, we expect that working with additional clinically-focused projects — through new Driver Project relationships, Strategic Partners, and National Initiatives — will put us in a better position to identify needs in this area and create appropriate materials and training activities. We will also consider how we can adopt the Tech Team Tutorials for clinical audiences*
- ***Leverage and build on ClinGen's database of Genomic Analysis Software Platforms Meeting Minimum Requirements for Data Sharing to Support Quality Assurance.***

- *The GA4GH standard Phenopackets is already included on this list. We will work with ClinGen leadership to consider adding further GA4GH products and will help promote the tool across the community.*

Appendix III: Attribution

This document was produced by the GA4GH Secretariat Team, with considerable efforts from:

- Angela Page, Director of Strategy and Engagement
- Lindsay Smith, Work Stream and Clinical Projects Manager
- Neerjah Skantharajah, Implementation Forum Manager

The GA4GH Executive Team and Standards Steering Committee reviewed pieces of the document throughout its creation.

The contents of the document, including strategic refresh feedback, was gathered from members of the GA4GH Community via a series of town hall forums, surveys, and interviews.