Global Alliance for Genomics & Health

GA4GH Connect

2023 Strategic Refresh

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Executive summary

In 2022, through a written survey and a series of open-brainstorming sessions chaired by members of the GA4GH Executive Committee, GA4GH received more than 300 pieces of feedback from more than 120 individual stakeholders commenting on our current and future ability to achieve the three Community Imperatives identified during the 2020 Gap Analysis: (i) improve interoperability and alignment with external standards and between GA4GH products; (ii)

improve implementation support for technical standards; and (iii) align more closely with healthcare and clinical standards.

The most recurrent themes in the Strategic Refresh feedback were recommendations to (i) create a forum for sharing experiences implementing GA4GH products, and mechanisms for implementers to demonstrate usage of GA4GH products; (ii) work with collaborative projects to identify and advance use cases and (iii) ensure clinical relevance of the GA4GH toolkit before promoting it to the clinical community.

In order to respond to these recommendations, GA4GH staff 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.

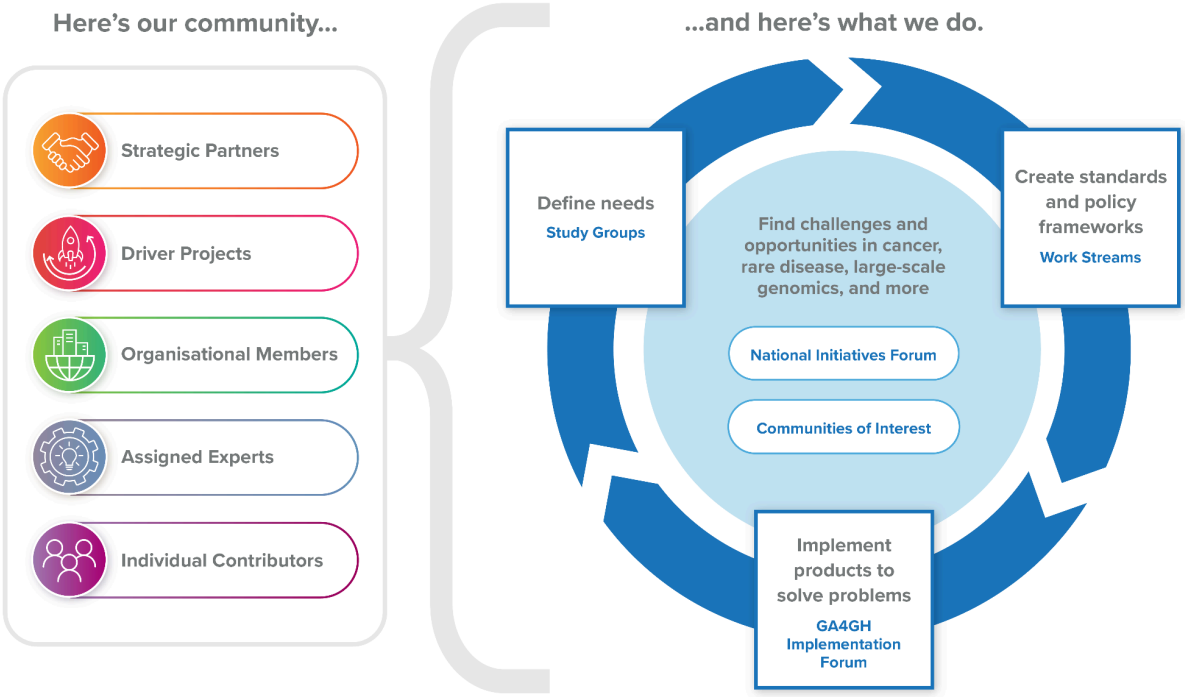
For example, GA4GH is no longer a simple matrix of Driver Projects and Work Streams, as described in the 2018 Strategic Plan. While this matrix remains the foundational strategy underlying all of our work, we also support activities upstream and downstream from the development phase and rely on contributions from a much broader community than those reflected solely in the Driver Project cohort.

In this document we present an updated matrix that better reflects how we now work, and introduce a series of new strategies including (i) a regular application cycle to bring in new Driver Projects (DPs) and introduction of DP term limits; (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). 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 organisations (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 to develop robust implementation support resources. Finally, in an appendix, we describe how we believe these updates will address 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 accelerate progress in genomic research and human health by cultivating a common framework of standards and harmonised approaches for effective and responsible sharing of genomic and related health data. In 2017, the GA4GH Connect five-year strategic plan refocused the organisation on development of technical standards and policy frameworks, introducing the concepts of Work Streams and Driver Projects. 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: (i) improve interoperability and alignment with external standards and between GA4GH products; (ii) improve implementation support for technical standards; and (iii) engage more closely with healthcare and clinical standards. In 2022, GA4GH staff oversaw a "Strategic Refresh" to reflect on our progress against the previous roadmaps and plans.

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 for each of the three Community Imperatives and summarise the feedback we received in 2022 on our current and future ability to achieve them.

Interoperability and 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 two aims 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 GA4GH products to share data, particularly sensitive human data. Others urged the GA4GH Work Streams to place a greater emphasis on ingesting external use cases, add formal alignment requirements to the product development and approval process, and create an issue

board for GA4GH-wide, cross-Work-Stream concerns. There was also a suggestion to regularly convene Work Stream leadership in order to identify integration and alignment challenges and opportunities.

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 products. 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 programme." We also learned of several improvements we can make to our working practices, including more comprehensive documentation (which should be consistently translated into multiple spoken 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 and the standards organisations that support it. While our connection to the research side of the "learning health system"[1] was strong, we acknowledged our limited interface with the healthcare side of that cycle, in part due to the diversity of the global clinical community. 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 and the healthcare industry versus practising clinicians).

In 2022, we are still facing the same challenges. Strategic Refresh participants told us that the ultimate targets of implementation — people with clinical backgrounds and clinical genomic experience in the areas of patient care, clinical laboratory roles, and clinical research — are still underrepresented within the GA4GH community. They urged us to introduce more clinically-focused Driver Projects, align with other clinical standards development organisations (SDOs) and professional organisations (such as ACMG, NSGC, ASHG, and Medical Genome

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

Initiative), and 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 electronic health record (EHR) systems to encourage adoption of GA4GH products (such as VRS and VCF). 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 address 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.

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

1. create a forum for sharing experiences implementing GA4GH products;
2. create mechanisms for implementers to demonstrate usage of GA4GH products;
3. work with collaborative projects to identify and advance use cases;
4. ensure clinical relevance of GA4GH toolkit before promoting to the clinical community.

Through these updates, we hope that several other recommendations received from the community will also be in a better position to succeed. We highlight how each proposal relates to specific pieces of feedback in Appendix II: how this proposal will impact the Community Imperatives.

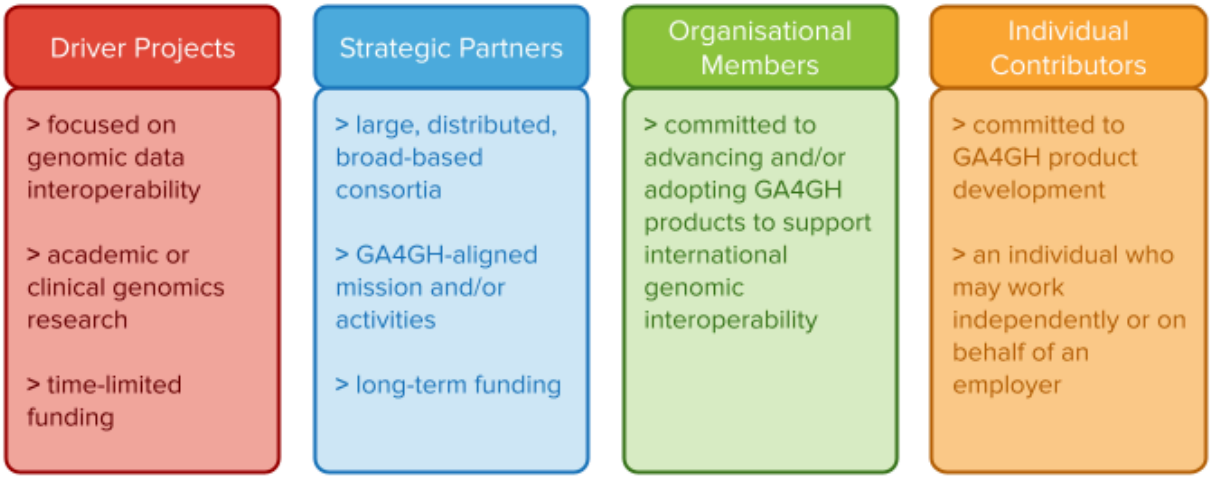
Part II: who we are

Updates to our community

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 where GA4GH activities would 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 GA4GH community has expanded significantly. 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 more scalable mechanisms, GA4GH now seeks to expand the Strategic Partner model, launched in 2017 with the formation of the ELIXIR::GA4GH Strategic Partnership. These two models (Driver Projects and Strategic Partners) draw from our broader, extended community of Individual Contributors and Organisational Members, whose great value we wish to better and more formally recognise 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 who 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 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 product 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 prioritised role within GA4GH Work Streams. With this benefit comes the responsibility of accurately representing the needs of the broader genomics community.

In becoming a Driver Project, initiatives make six formal commitments to GA4GH[2]:

1. contribute to the development of at least three technical products;
2. implement at least three technical products
3. participate in foundational Work Streams (Regulatory & Ethics, Data Security);
4. uphold the the *Framework for responsible sharing of genomic and health-related data* as well as other regulatory and ethics frameworks as appropriate;
5. commit a total dedicated personnel effort of at least two full-time-employee (FTE) equivalents;
6. adhere to GA4GH institutional policies, including the Standards for Professional Conduct, Copyright Policy, and forthcoming Patent Policy.

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;

[2] Approved Driver Projects will be expected to sign a formal, non-binding Driver Project Agreement.

3. participate in quarterly Standards Steering Committee (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) represent a particular DP on various GA4GH activities, such as Work Streams and Study Groups. DP contributors have deep technical, ethics, 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 that are under-resourced for genomics and in low and middle-income countries (LMICs).

Strategic Partners

Launched in 2017, the GA4GH Strategic Partner (SP) model was launched through the creation of the ELIXIR::GA4GH Strategic Partnership. Through this partnership, the European Life- Science Infrastructure (ELIXIR) has committed to support the use of GA4GH products across Europe in order to facilitate the responsible sharing of large-scale, sensitive human genomic data. Likewise, GA4GH has committed to prioritising the needs of ELIXIR in its work.

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

In 2023, we aim to add more SPs as necessary on a rolling basis. GA4GH SPs should be sustainably-funded, broad-based consortia (some will encompass multiple DPs) with an aligned mission to that of GA4GH. They are distinct from DPs 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 GA4GH products are fit for use within that network.

Strategic Partners must:

1. have secure, long-term funding and/or a robust sustainability model;
2. provide services to an international genomics and health community;
3. align philosophically with GA4GH's mission and vision;
4. participate in GA4GH activities (Study Groups, Work Streams, GA4GH Implementation Forum).

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

To initiate a Strategic Partnership, the leadership of the proposed 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 requires only 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:

1. have a mission, operations, and public statements that are consistent with the GA4GH Constitution and Member Agreement;
2. be an established entity (e.g. corporation, not-for-profit organisation, partnership);
3. have an open and professional presence;
4. be active in the field of genomic research/medicine or a related activity;
5. have ownership, governance, funding, and leadership that are clearly defined and accessible.

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 that wish to adopt GA4GH products but are not able to participate in product development. Contributor-implementers both participate in the development of standards and policy frameworks, as well as implement them in real-world practice. Observers support the mission and vision of GA4GH but are not in a position to either

contribute to or implement GA4GH deliverables. ***In order to encourage broader participation among our Organisational Members, we will aim in 2023 to publicly recognise those that are actively contributing to or implementing GA4GH products.***

Individual Contributors

The GA4GH contributor community currently consists of around 1,400 individuals, who can be characterised as observers, contributors, funded contributors, and Assigned Experts, as described below.

Observers

Individual observers have expressed interest in GA4GH development activities and have been added to a GA4GH Google Group for email updates.

Recognition: Observers are not recognised on the GA4GH website.

Contributors

Contributors are individuals who have made meaningful contributions to GA4GH products or work, for example by joining Work Stream calls, reviewing or developing documents, or developing code.

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

Funded contributors

Funded contributors are contributors who have received funding to support their work on GA4GH standards and policy development.

Recognition: Funded contributors are listed as "contributors" on the webpage(s) for the product(s) to which they have contributed. The specific funder is not indicated.

Assigned Experts

Assigned Experts are funded contributors who have been specifically assigned by their employer — a national funder or representative local institution — to commit at least 30% of their time (four calendar months per year) to GA4GH product 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. represent a national or regional genomics landscape, often providing an otherwise unmet strategic connection for GA4GH;
3. directly and regularly engage with the GA4GH policy and technical staff teams.

Process for Establishing an Assigned Expert

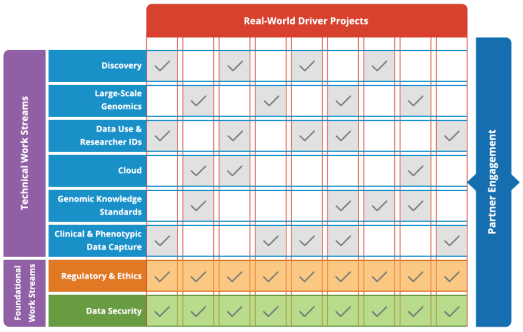
To become an Assigned Expert, an individual's employer must work with GA4GH leadership to establish the need and validate the appointment. Assigned Experts who are not first discussed with GA4GH leadership may or may not be recognised by GA4GH as such. Assigned Expert employers are expected to find funding for the appointment, or, conversely, funders of Assigned Experts are expected to secure a local employer to house the expert. Arranging funding and employers is 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 work with GA4GH staff to ensure that their efforts are aligned with institutional development priorities.

Recognition: Assigned Expert funders are listed on the GA4GH website, and individual Assigned Experts are recognised on the webpage(s) for the product(s) to which they have contributed. Both are also recognised on a webpage dedicated to the Assigned Experts mechanism.

Part III: what we do

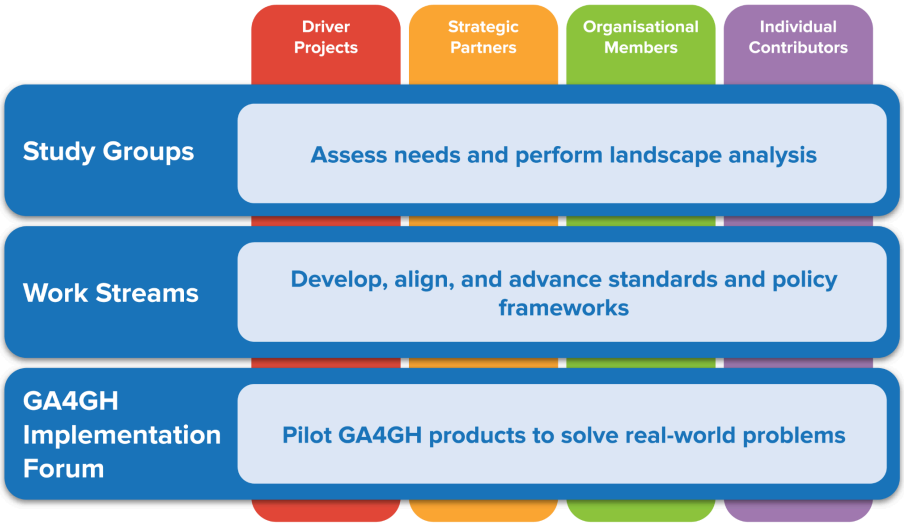
Updates to our activities



Just as the GA4GH community has expanded since 2017, so has the repertoire of activities in which GA4GH stakeholders engage. Our work now includes activities that come before product development commences and after initial product approval. In 2023, we aim to better communicate how we work by formally adding two more horizontal categories to the official 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. We must also emphasise that these three activities feed into one another in a continuous feedback cycle.

Study Groups

As described in the update to the GA4GH Product Development and Approval Process (still in draft form), 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 Data 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 recognise 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 (e.g. researchers, bioinformaticians, clinical stakeholders). Many

members of a Study Group will often go on to form the seed of a Work Stream subgroup to develop a new product.

Work Streams

GA4GH Work Streams develop technical standards and policy frameworks and tools 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 and Driver Projects in the areas of regulation, ethics, and data security in genomics. The Regulatory & Ethics and Data Security Work Streams develop products 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. Currently, there are six technical Work Streams, each focused on a different aspect of genomic interoperability and data sharing: Cloud, Clinical & Phenotypic Data Exchange (Clin/Pheno), Data Use and Researcher Identities (DURI), Discovery, Genomic Knowledge Standards, and Large Scale Genomics.

Work Stream activities are organised and advanced under the direction of Work Stream Leads. While the working practices of each group may differ, GA4GH will provide guidance on best practices for effectively running the Work Stream and advancing its goals. This resource is currently in development.

Technical Alignment Subcommittee

Established in 2019, the Technical Alignment Subcommittee (TASC) aims to provide a mechanism for generating internal consistency across Work Streams and products. 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 issues related to GitHub structure and Docker.

Work Stream contributors

Anyone in the broad genomics community — whether or not they are affiliated with a GA4GH stakeholder organisation — who has an interest in using or helping to develop GA4GH standards or policy frameworks and tools is invited to participate in the GA4GH Work Streams. Participation

includes attendance at meetings and discussion on the associated Slack channels and/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 products. All Work Streams must have representation from at least two GA4GH stakeholder groups at all times, including at least one Driver Project.

There are a number of different contributor roles within a Work Stream:

- **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. WSLs are nominated by the community and appointed by the Executive Committee.

- **Product Leads**
  Each product developed by a Work Stream will have at least one Product Lead (PL). These are individuals with both technical expertise and bandwidth to ensure timely delivery of product-level milestones. PLs are expected to attend both Work Stream meetings and any relevant product-specific subgroup meetings, which may occur more frequently than Work Stream meetings. PLs are appointed by WSLs and 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 and resources to actively contribute to Work Streams and subgroups in the development of GA4GH products. WSCs may participate on behalf of any GA4GH stakeholder organisation or as an Individual Contributor unaffiliated with a member organisation.

Advancing products within a Work Stream

The update to the GA4GH Product Development and Approval Process (still in draft form) outlines a complete product development process, beginning with a formal proposal and ending with review and approval by the GA4GH Standards Steering Committee (SSC), the Regulatory & Ethics and Data Security Work Streams, and a specially convened Product Review Committee specific to the product in question. The process gives clear direction on 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 SSC to ensure that the development and approval 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 Work Stream. 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 Work Streams should persist in perpetuity. If the group decides that it makes more sense to evolve or dissolve, GA4GH leadership 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 and tools, but feels more work is needed to promote uptake or perform necessary testing, and the current Work Stream contributors and processes do not support these new needs, then GA4GH staff (particularly the Work Stream Managers) will collaborate with existing leadership to develop a transition plan for the Work Stream.

Cross-Work Stream communication

A major point of discussion during the Strategic Refresh consultation 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. TASC is well positioned to address the final of these requirements; however, additional opportunities are needed for the first two.

In 2023, GA4GH staff will aim 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-Work-Stream 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, led by the Equity, Diversity, and Inclusion Advisory Group, aims to identify opportunities for GA4GH contributors to engage more fully across Work Streams.

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. Additionally, 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 authorisation and authentication; and Beacon for discovering datasets of interest. While more ambitious goals are on the horizon (e.g. 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 (which replaces both FASP[3] and the Genomics in Health Implementation Forum[4]), we aim to identify a series of multi-stakeholder use cases focused on implementing production-ready standards to advance a real-world scientific or clinical problem. GA4GH will support three to four flagship use cases at a time, all of which:

1. **consist of a set of actions** performed to achieve a stated goal through either multiple GA4GH products working together, or one standard being implemented across multiple projects to demonstrate interoperability;
2. **produce observable results** to answer a scientific or clinical research question using real-world data;
3. **are of value to users** (at least three distinct global initiatives) in the area of genomic data interoperability;
4. **have dedicated resources** to advance the use case.

These GIF Projects — focused on such things as federated imputation or federated variant analysis — will be identified in multiple venues within the GA4GH community, such as the National Initiatives Forum, Work Streams, GA4GH meetings, and Communities of Interest. Once a use case has been identified, the larger community will be notified and invited to participate.

GIF Projects will be advanced using the FASP framework, which focused on enabling "third parties to perform real-world, scientific use cases by connecting GA4GH standards." This framework leads to three key outputs for GIF Project:

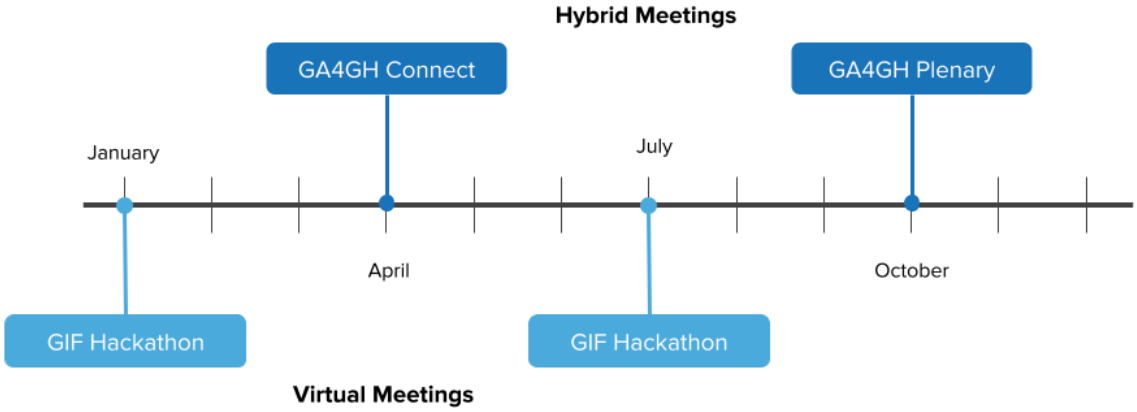
1. resources to enable the broader genomics community to solve real-world challenges using GA4GH products;
2. demonstration of GA4GH-supported interoperability;
3. insights about using the products in practice, which can be fed back to those developing and/or maintaining them (e.g. the GA4GH Work Streams).

GIF meetings

Two hybrid GIF meetings will be held each year, in conjunction with GA4GH Plenary and with GA4GH Connect, our annual hybrid working meeting focused on cross-Work-Stream development. A biannual GIF Hackathon will allow engineers to advance use cases in a virtual setting in the time between annual meetings. Individual GIF Project teams will define the frequency and regularity with which they meet. GIF Projects will also provide valuable content for

[3] Previous FASP activities, such as developing implementation support resources, that are not covered by GIF will become the remit of the GA4GH technical staff team, Assigned Experts, or TASC.
[4] The initiatives that previously comprised the Genomics in Health Implementation Forum will continue to convene as the National Initiatives Forum.

the annual GA4GH Plenary Meeting, namely evidence of GA4GH standards being used in real-world settings. GIF milestones will align with these annual meetings in order to drive activity.



GIF leadership

Each GIF Project will be led by two or more project champions. Champions will work with GA4GH staff to ensure that all required documentation is complete and up-to-date, the project progresses and has clear milestones/goals, and that a presenter is available to provide updates at GIF meetings. The full group of GIF champions will meet quarterly to ensure alignment across projects and with GA4GH. For strategy development and coordination purposes, the group of champions will annually elect one of their number to serve as chair.

More details on the operational aspects of GIF can be found in the GIF Working Practices document.

Partner engagement

In addition to the three primary activities of the GA4GH matrix — Study Groups, Work Streams, and the 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 effectively meet the needs of the broader genomics community and drive uptake of GA4GH standards across the international genomics landscape.

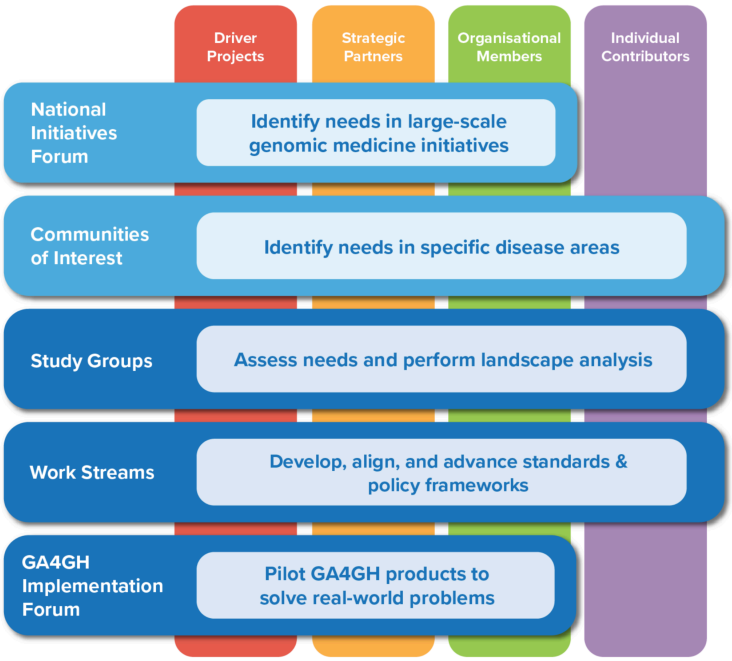
National Initiatives Forum

In response to the strong feedback received around the need to better engage the clinical community, GA4GH will put more emphasis and resources on its work with large-scale clinical genomic data initiatives, such as Genomics England and Australian Genomics.

Launched in 2016, the GA4GH National Initiatives Forum (previously called the Genomics in Health Implementation Forum, or GHIF) aims to support more accurate data interpretation and diagnosis and the development of innovative solutions across healthcare through global cooperation and data sharing. The forum helps initiatives reduce duplication of effort and

promote systems integration and the collection of compatible clinical genomic data and health information to enable global data sharing.

The forum is primarily focused on supporting uptake of GA4GH standards within large-scale clinical genomics initiatives and providing real-world insights to the GA4GH Work Streams as they develop new standards (and improve existing standards) that 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 forum's primary goals are to:

1. identify areas for collaboration and the sharing of resources and expertise;
2. identify real-world use cases for global genomic data sharing;
3. identify common needs for standards across initiatives.

All GA4GH National Initiatives Forum members:

1. have secured long-term funding;
2. are focused on advancing a large-scale genomics strategy across a health system;
3. are actively working to adopt GA4GH standards.

Unless they are already a GA4GH Driver Project, national initiatives interested in participating in the forum must apply and be approved to become a member. They will be expected to dedicate a to-be-determined minimum amount of personnel effort to GA4GH activities.

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 forum will also help steer the course of GA4GH standards development and optimization through members' 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 products 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 GA4GH standards that support oncology research and clinical care. The group is currently focused on developing use cases in the areas of cancer variant representation, federated sharing of data for variant 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 of 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). Early in its history, ISO TC215/SC1 launched a task force to map the genomics standards work items underway in each of the above-mentioned SDOs. This mapping aims to inform a cross-SDO roadmap for genomics, identifying alignment of current activities, gaps, and opportunities for collaborative standards development. By participating in these alignment activities, GA4GH aims to help reduce duplicative efforts and amplify the impact of our standards, promoting a truly global infrastructure that benefits research and healthcare outcomes.

Part IV: Strategies for achieving our vision

GA4GH Funders Forum

The GA4GH Funders Forum consists of Host Institutions, Core Funders, Supporting Funders, and Assigned Expert Funders. The Funders Forum provides financial support and meets annually to provide additional strategic guidance.

Host Institutions

Host Institutions provide services and administrative support — such as space, grants management, and human resources — on behalf of GA4GH. All core GA4GH staff are employed at a GA4GH Host Institution. The current Host Institutions are the Broad Institute of MIT and Harvard in the US, the Ontario Institute for Cancer Research in Canada, the Wellcome Sanger Institute in the UK, and EMBL's European Bioinformatics Institute in the UK.

Core Funders

Core Funders commit to funding more than 200,000 USD annually for a minimum of three years. They meet as a group once per year to provide high-level consultation on GA4GH functions and direction and have the opportunity to nominate the majority of directors of GA4GH Inc. Public and private agencies in the US, the UK, and Canada currently provide GA4GH core funding, which is channelled to the four GA4GH Host Institutions.

Supporting Funders

Supporting Funders provide financial support to GA4GH at a financial level below the Core Funder threshold.

Assigned Expert Funders

National funding agencies and other institutions have the opportunity to join the GA4GH Funders Forum by assigning a technical or regulatory expert to GA4GH product development activities for at least four calendar months per year.

GA4GH Standards Steering Committee

The GA4GH Standards Steering Committee (SSC) consists of Work Stream Leads, Driver Project (DP) Champions, and Strategic Partner (SP) representatives. All leaders in their fields, SSC members are either appointed through an open nomination process (i.e. Work Stream Leads) or by the project with which they are affiliated (i.e. DP Champions and SP representatives). As a group, SSC members reflect the diverse perspectives, backgrounds, and geography of the GA4GH community. While multiple DP Champions and SP representatives may join SSC meetings, DPs and SPs are allotted one vote each. If an SP is associated with one or more GA4GH Driver Projects (DPs), the SP vote is additional to the DP vote(s).

The SSC is primarily responsible for ensuring the product development and approval process is followed appropriately. The group takes key votes during a product's life cycle: at the proposal phase, when the SSC approves a standard for development within a Work Stream, and at the product approval phase, when the SSC confirms that the product has been developed according to the agreed process. In both cases, a simple majority vote is sufficient for approval. However, GA4GH strives to ensure unanimous approval through steps taken prior to the vote.

While Organisational Members are not represented on the SSC, all products are made available for open comment for one month prior to the voting period and all GA4GH stakeholders — as well as the general public — are encouraged to share their feedback during that time. Additionally, Driver Projects are intended to serve as proxies for this broader community, ensuring its needs are at least indirectly represented

GA4GH staff

Work Stream management

Work Stream Managers (WSMs) play a critical role in setting the goals and priorities of the Work Streams and related activities. Their responsibilities include community management, strategy development and implementation, and programme management of several international task-teams and knowledge exchange projects from concept development through 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, as well as 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 to bring 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 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 of standards and reducing the barriers for newcomers to understand and adopt GA4GH products. Tools developed by the tech team include reference implementations, 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.

GA4GH aims 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, community-driven use cases, and implementation guides.

The tech team works on projects that align with plans outlined in our grant applications and collaborates 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 tech team also helps to identify interoperability problems between new and existing standards and correct these issues, promoting comprehensive technical harmonisation across all GA4GH products. As GA4GH-compliant services can be viewed as a large, federated network, the tech team will also 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. Users are encouraged 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, giving platform implementers confidence before they embark on their more costly task: extending the ability to orchestrate federated data and services to scientists who do not write scripts.

GA4GH Tutorials

GA4GH Tutorials aim to promote awareness and uptake of the GA4GH products that enable federated data sharing. Tutorials enable researchers to learn how to seamlessly access data from multiple sources in an international network using common tools and protocols. They also enable 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 participants to learn more about GA4GH products, and in some cases, get hands-on experience with tasks such as accessing information, running workflows, and searching and filtering biomedical datasets based on criteria of interest.

Policy team

The GA4GH policy team consists of dedicated policy analysts and research assistants who support the ongoing development of GA4GH regulatory and ethics products, as well as provide guidance and expertise to the community in a number of key areas. The team offers GA4GH leadership advice and expertise on where and how GA4GH can influence national and international discussions on policy. The team also leads and/or collaborates on the development of position statements and consultation responses where appropriate. Team members develop relationships with stakeholders and networks — particularly within the genomics and clinical communities and with policymakers for those communities — to ensure early awareness of emerging policy issues relevant to the work of GA4GH. The team also supports 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 (e.g. Code of Conduct, Patent Policy); and the development of regulatory and ethics implementation resources for other Work Stream products, as appropriate.

Representation and inclusion

The GA4GH Equity, Diversity, and Inclusion (EDI) Advisory Group was launched in May 2020 with the goal of recognising 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 product 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;
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 the resulting products. Below we outline current activities of the EDI Advisory Group.

Onboarding

The 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 helps newcomers feel welcomed and have the knowledge needed to effectively participate in GA4GH. The programme pairs newcomers to a specific Work Stream with an Onboarding Guide, who helps them become oriented to the Work Stream. Onboarding Guides aid GA4GH newcomers to better understand Work-Stream-specific content by providing networking support and relevant background information.

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, and gain knowledge about 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, amplify the newcomer’s voice by helping to put comments into context and/or support and reinforce comments made; helping the newcomer find points of connection between the current discussion and others within the community; and seeking out the newcomer’s views on specific topics to facilitate their engagement and participation. Over the longer term, the Onboarding Guide is available to the newcomer as needed with additional questions or introductions to 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 EDI Advisory Group began the Best Practices for Working Effectively project in 2021 with a workshop aimed at engaging all Work Stream and product leads. Following the workshop, the team worked to collect Work Stream best practices for creating an inclusive working environment that facilitates diverse participation. These were later categorised by members of GA4GH staff. The following seven practice areas emerged from this effort: identifying missing perspectives,

inviting people to participate, welcoming and onboarding newcomers, chairing inclusive meetings, ensuring opportunities for engagement, recognising and defining contributions, keeping track of progress, and communicating decisions and discussions.

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 more than 500 participants. The 2022 meeting welcomed 250 in-person attendees, and hundreds more virtually. All GA4GH Plenary meetings are held as hybrid events, with all sessions 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 interaction between Work Streams. Connect meetings 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, GIF hackathons are 7- to 14-day events that focus on improving GA4GH APIs and other standards, as well as implementing GA4GH specifications in new environments. Hackathons are composed of multiple topics put forth by GA4GH contributors. Such topics aim to clarify and/or formalise a new feature in one or more existing GA4GH specifications that will enable more research use cases, and/or will develop or deploy implementations of GA4GH standards to facilitate a novel, federated use case. GIF Hackathons welcome the entire GA4GH community.

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 thatGA4GH would focus its development efforts on 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 in the broader genomics and health communities.

Gap analysis (2020)

The 2020 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 that engaged 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 underway for some time, including the Technical Alignment Subcommittee, the Federated Analysis Systems Project, the Genomics in Health Implementation Forum (GHIF), and more. Nevertheless, 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, GA4GH 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.

Appendix II: how this refresh will impact the Community Imperatives

In this section we identify updates to the GA4GH strategic plan that aim to address specific pieces of feedback heard during the 2022 Strategic Refresh consultation process.

Interoperability and alignment

- ***Better support and engagement on 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 Technical Alignment Sub-Committee and 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.***
    - *The introduction of Study Groups provides a mechanism for early exploration into overlap and synergy with other standards and work streams.*
    - *The introduction of the GA4GH Implementation Forum will refocus efforts on external use cases.*
- ***Add formal alignment requirements to the Product Approval and Development Process.***
    - *The introduction of Study Groups will raise questions of alignment earlier in product development.*
    - *Updates to the Product Development and Approval Process will increase alignment throughout the product life cycle.*
- ***Create an issue board for GA4GH-wide, cross-Work-Stream concerns.***
    - *This is being discussed by the technical team in conjunction with the Technical Alignment Sub-Committee.*
- ***Regularly convene Work Stream leadership to identify integration and alignment challenges and opportunities, perhaps through the existing TASC framework.***
    - *The Technical Alignment Sub-Committee, which consists of WS leads and/or their delegates, will refine its mandate and draw on expanded staff support to more effectively convene Work Stream leadership.*

Implementation support

- ***Provide more opportunities for individuals with clinical backgrounds and clinical genomic experience to engage with GA4GH.***

    - *The 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.*
- ***Encourage leading stakeholders to set the tone for the community by supporting and promoting GA4GH standards.***
    - *The introduction of the Strategic Partner mechanism and targeted outreach in the open call for Driver Projects will recruit more community leaders to support GA4GH standards.*
- ***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.***
    - *The introduction of the GA4GH Implementation Forum and the Technical Alignment Sub-Committee will bring implementers together.*
- ***Encourage journals to include GA4GH in their author guidelines.***
    - *The Strategic Partner mechanism may create opportunities for collaborations with journals.*
    - *While not discussed explicitly in this document, GA4GH will continue to pursue its role as a thought leader and engage in discussions with journals*
- ***Encourage funders to create GA4GH-focused requests for proposals.***
    - *While not discussed explicitly in this document, 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 share their GA4GH product uptake through an associated badge programme.***
    - *The introduction of the GA4GH Implementation Forum and support from the GA4GH technical team will help establish such a mechanism.*
- ***Make documentation more comprehensive and accessible (e.g. by consistently translating into multiple languages).***
    - *The expansion of the tech team and introduction of technical writing expertise therein will improve documentation.*
- ***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 products prior to approval.***
    - *Updates to the Product Development and Approval Process increase rigour throughout the product life cycle.*
- ***Outline additional mechanisms for improved implementation support.***
    - *Starter Kit resources make it easier for organisations to implement GA4GH standards.*

- *Assigned Experts will ideally focus a portion of their efforts on local implementation.*
- *Best practices and onboarding will support more effective working practices in GA4GH groups.*
- *DP spots will be 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 separately-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 explicitly in this document, members of GA4GH leadership and the 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 Strategic Partners, 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 explicitly in this document, creating clinical implementation guidelines is a common practice in our Clinical & 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 explicitly in this document, we expect that working with additional clinically-focused projects — through new Driver Project relationships, Strategic Partners, and the National Initiatives Forum — 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 GA4GH 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 GA4GH staff, with considerable efforts from:

1. Angela Page, Director of Strategy and Engagement;
2. Lindsay Smith, Work Stream and Clinical Projects Manager;
3. Neerjah Skantharajah, Implementation Forum Manager;

and input from the GA4GH Executive Committee:

1. Peter Goodhand, CEO;
2. Ewan Birney, Chair;
3. Heidi Rehm, Vice-Chair;
4. Kathryn North, Vice-Chair.

The Standards Steering Committee reviewed pieces of the document throughout its creation.

The contents of the document, including Strategic Refresh feedback, were gathered from members of the GA4GH Community via a series of town hall meetings, surveys, and interviews.