



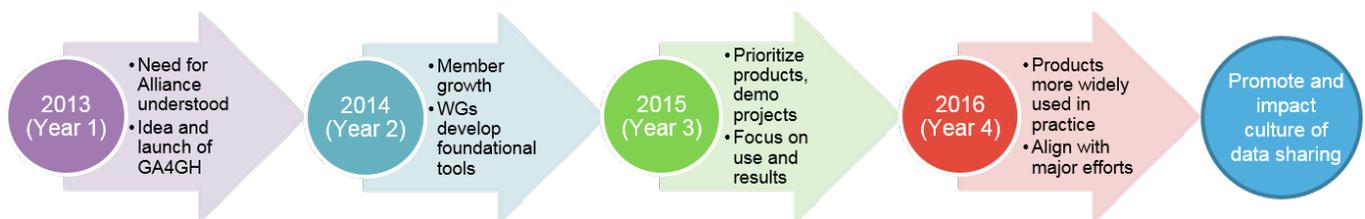
## Global Alliance for Genomics and Health Strategic Road Map (2015/2016)

### GA4GH Mission

The mission of the Global Alliance for Genomics and Health (GA4GH) is to accelerate progress in human health by helping to enable effective and responsible sharing of genomic and clinical data. The world is at an inflection point for the scalability and use of genomic data in medicine, which is why the original GA4GH mission remains valid and of vital importance.

In 2015 and 2016, GA4GH will prioritize development and use of products, as well as the success of demonstration projects, leading to increased genomic data sharing and systemic change. GA4GH will be focused on high-impact activities and tangible results. Longer term, GA4GH will empower and promote a culture of widespread data sharing and help foster the connection of genomic medicine to clinical care.

### GA4GH Goal Progression



### 2015/2016 Objectives

GA4GH will focus the scope of 2015/2016 activities on priority needs to enable data sharing in areas that provide value to members and the field, so that the effort ensures: **results**, **relevance**, and **sustainability**. The objectives and activity directives in the Road Map provide GA4GH Members with the prioritization and framing needed to develop systems, drive concrete actions, and create targeted progress in the key areas.

#### Results

- Objective 1: Develop priority products
- Objective 2: Deliver packaged, working solutions
- Objective 3: Support demonstration projects

#### Relevance

- Objective 4: Align with major data collection and sharing efforts
- Objective 5: Communicate strategically with key audiences
- Objective 6: Establish GA4GH as a thought leader

#### Sustainability

- Objective 7: Build leadership and participation
- Objective 8: Expand organizational capacity and funding streams



## Results

Develop and deliver integrated solutions to significantly advance use of GA4GH products, show proof of concept, and drive towards breakthrough accomplishments that enable data sharing.

### Objective 1: Develop priority GA4GH products that address pressing needs

Working Groups will set priorities and benchmarks for product development, in order to best assign resources, finalize outputs, and promote use. Priority products will be tools, methods, and approaches that are most needed to help remove existing barriers and challenges, and to better enable the sharing of genomic and health-related data.

- Carefully choose products and develop tools and approaches in systematic process across the current Working Group structure.
  - Develop benchmarks with 6-month intervals for Working Group product output for more coordinated and systematic planning.
  - Prioritization and expected timelines for 2015/2016 products to be set by May 2015. The list of all current Working Group products is included in this Road Map as an appendix.
- All new products will target how GA4GH can add value and have a strong focus on accomplishing tangible near-term results.
  - Set criteria and establish process for taking on new products (showing how each will advance two-year GA4GH objectives).
  - Ensure that high priority products are packaged for broad uptake; Guard against risk of groups working on dozens of products without prioritization at the outset.
  - Recognize and focus on the pressing need for a few breakthrough accomplishments for GA4GH.
- Increase uptake of GA4GH products by leveraging driver institutions and Membership, in order to increase responsible sharing.
  - Partner with and encourage funders of research and other key institutions and stakeholders to incentivize use and sharing.
  - Better leverage and communicate to GA4GH members on product development and how products can be utilized.

### Objective 2: Deliver packaged, working solutions to promote product use

To help implement GA4GH products in the field and have them more widely applied in practice, GA4GH will not only make tools available, but will take steps to drive dynamic use. These will include combining methods and tools into data sharing “packages” that are relevant for large-scale initiatives and more direct engagement with select groups undertaking data sharing efforts.

- Produce combined toolkits to guide and promote use of products based on need:
  - Start-up kits for responsible data sharing
  - Central portal with compatible tools and methods
  - Update GA4GH website with easier access to and better support for products
  - Increased Working Group alignment and cross-Working Group collaboration
- Provide support to select efforts to facilitate product use.
  - Provide individualized technical assistance where appropriate and with varying level of support to existing/emerging projects and initiatives to facilitate the use of GA4GH tools.
  - Survey membership and ensure transparency in selecting efforts with which to engage.



- Hear from key data sharing efforts to understand obstacles to data sharing in order to drive solutions that are responsive to needs.
- Strategically deploy efforts and guidance, rather than offering a broad brushstroke of technical assistance or large-scale training efforts throughout the ecosystem.

### **Objective 3: Support current and new demonstration projects to show value of responsible data sharing**

Demonstration projects are a key marker of progress now and over the next two years. GA4GH will focus on accelerating a select number of demonstration projects, driving them to the results phase. Demonstration projects will be used as examples for the field and could have major impact once completed, more publicly proving the transformational possibilities of genomic data sharing.

- Have a steady and small number of demonstration projects, cycling new projects in and out as needed, rather than continually increasing GA4GH portfolio.
  - Projects should become fully independent within 2-3 years. In some select cases, projects may be incubated longer if necessary.
- Continue providing needed support to current demonstration projects to produce results and drive towards independence (BRCA Challenge, Matchmaker Exchange, and Beacon).
- Identify gaps in current project portfolio and select 1-2 new demonstration projects:
  - Proposed project focus: Clinical cancer genome project
  - Proposed project focus: Infectious disease/Pathogen genome data sharing
  - Proposed project focus: eHealth
- Consider engaging additional emerging projects.
  - Only add demonstration projects with a high impact for GA4GH mission.
  - Emphasize key demonstration projects rather than accelerating dozens of new initiatives.

#### **Relevance**

As public awareness about the use of genomic data in medicine rapidly increases, the GA4GH must also increase targeted awareness and ensure relevance through alignment with current major efforts and leadership in key issues and debates.

### **Objective 4: Align with major efforts around the world to collect and share data in order to leverage GA4GH impact**

Significant initiatives in precision medicine and genomic data collection have been announced, and additional major national and institutional efforts are likely. GA4GH will align with and directly engage such efforts to increase product use and secure relevance.

- Directly engage major efforts to ensure relevance and use of GA4GH solutions.
  - Partner and interact in ways that are advantageous for efforts, adding value and expertise.
  - Approach efforts through funding agencies and/or with allied stakeholders.
- Create processes to leverage opportunities to align GA4GH approaches and methods with major efforts in field (national and institutional).
  - Examples of efforts: Genomics England, US Precision Medicine Initiative.



### **Objective 5: Communicate in targeted, deliberate way to key audiences**

Selective outreach to promote product use and data sharing – and to make GA4GH a trusted authority in the field – will be the communication focus of 2015/2016.

- Communicate primarily to highlight GA4GH products and projects.
  - GA4GH outreach to include conferences, papers, and other public communications relevant to product development and application.
  - Deploy enhanced web and technology tools to package products and facilitate external communication and member engagement.
  - Tell the story of current successes and future possibilities for genomic data sharing using the development and results of projects and products.
- Build support and trust in GA4GH through external communications to scientific, health, advocacy, and other biomedical stakeholder audiences.
  - Produce major marker paper illustrating GA4GH approach.
- Communicate with members to understand barriers and needs, and engage to ensure product dissemination.
  - GA4GH will use regular webinars, newsletters, panels to introduce products/projects, plenary meetings, and member surveys to routinely inform membership.
- GA4GH will not undertake a large-scale public communication campaign in 2015/2016, given near-term resources and focus.

### **Objective 6: Establish GA4GH as a thought leader in the sharing of genomic data**

By acting as a "think tank," GA4GH will become a trusted voice in key debates and discussions, mainly through the production of information, prioritizing a few key areas.

- Address the most pressing longer-term challenges and opportunities to advancing genomics and health through responsible data sharing.
  - Proposed topic: Benefits and evolution of federated versus centralized data repositories
  - Proposed topic: Million whole genome global opportunity
- Produce a small number of new ideas and think pieces annually, in the form of reports, white papers, and other public information. Draw on the cross-sector expertise and cross-disciplines of GA4GH membership for content.

#### **Sustainability**

Build stronger participation, target new resources, and increase organizational capacity to ensure a stable and continuing GA4GH.

### **Objective 7: Build stable leadership and participation**

GA4GH will seek active participation from new and untapped individuals (for leadership positions and contributing Working Groups members) in order to expand current circle of active engagement, prevent burn out, and diversify views and work streams.



- Identify and engage new leaders to expand the committed set of members that participate actively in GA4GH work.
  - Focus primarily on cultivating members currently demonstrating ability and commitment, or with interest areas aligned with GA4GH priorities.
- Better leverage current members and engage new stakeholders most closely aligned with GA4GH projects and products.
  - Incentivize and encourage participation and work (examples: target interest areas, professional recognition and credit).
  - Use staff resources to reach out to identified individuals to recruit for deeper engagement.
- Establish Steering Committee and Working Group Chair rotation and nomination process.
- Engagement will focus on increasing active participation in GA4GH, not on building a significantly larger membership base.

**Objective 8: Increase organizational capacity in key areas and expand funding streams to support progress**

GA4GH will strategically increase full-time staff and resources will be more targeted in order to ensure a sustainable and forward-focused GA4GH system of support. GA4GH will also drive towards a more distributed and sustainable funding model.

- Increase GA4GH staffing to support GA4GH progress:
  - Working Group coordination and product development
  - Demonstration projects coordination and driving
  - External engagement
  - Administrative and fundraising
- Develop criteria and mechanism for identifying needed staff and organization to support products and projects.
  - Large focus of Working Group staff will be moving GA4GH products toward completion and use.
  - Staffing structure will remain lean and distributed. Resources will be focused on key areas and based on need with leadership and Member feedback.
- Improve processes for more systematic lines of communications internally, with leadership, and between Working Groups. Ensure consistency in GA4GH products, outreach, and activities.
- GA4GH will transition from current funding model (three host sites) to more diverse and distributed sources of funding.
  - Combine host support with leading national funders to increase resource base in 2015.
  - Drive towards a more distributed model, including philanthropic and institutional funding, by 2016.



## Markers of Progress

GA4GH will not succeed with a focus on “winning the battle, but not the war,” i.e. completing many activities, but failing to achieve the broader GA4GH mission. GA4GH activities and commitments will remain nimble and will consistently focus on how we facilitate a large-scale evolution and advancement of a data sharing culture.

As next steps for objectives and activity directives in the 2015/2016 Road Map, GA4GH will develop more defined systems for implementation, as well as timelines and markers of progress or metrics, measuring GA4GH actions and resources used against the stated objectives and our overall mission.

## Appendix: Current Working Group Products

<p><b>Clinical</b></p> <p>Clinical Cancer Genome</p> <ul style="list-style-type: none"> <li>• White Paper</li> <li>• Resource (cancer data sharing initiatives)</li> </ul> <p>eHealth</p> <ul style="list-style-type: none"> <li>• Resource (eHealth use cases)</li> <li>• Resource (analyzing genomic data)</li> <li>• Resource (pedigree consent and family history)</li> <li>• White Paper (federated vs. centralized data)</li> <li>• Catalogue</li> </ul> <p>Phenotype Ontologies</p> <ul style="list-style-type: none"> <li>• Resource (cancer use cases)</li> <li>• Standard (rare disease requirements for ontology)</li> </ul>	<p><b>Data</b></p> <p>Variant Annotation</p> <ul style="list-style-type: none"> <li>• Standard (data annotation)</li> </ul> <p>Containers and Workflows</p> <ul style="list-style-type: none"> <li>• Standard (interoperable GA4GH APIs with open source)</li> </ul> <p>File Formats</p> <ul style="list-style-type: none"> <li>• Standard (file formats)</li> </ul> <p>Genotype2Phenotype Association</p> <ul style="list-style-type: none"> <li>• Standard</li> </ul> <p>Metadata</p> <ul style="list-style-type: none"> <li>• Standard (metadata schema)</li> </ul> <p>Reference Implementation</p> <ul style="list-style-type: none"> <li>• Standard (GA4GH API)</li> </ul> <p>Reference Variation</p> <ul style="list-style-type: none"> <li>• Standard (graph-based reference)</li> <li>• Implementation</li> </ul> <p>RNA and Gene Expression</p> <ul style="list-style-type: none"> <li>• Standard (RNA sequence reads)</li> </ul> <p>Variance Annotation</p> <ul style="list-style-type: none"> <li>• Standard (data representation)</li> </ul>	<p><b>Regulatory and Ethics</b></p> <p>Accountability</p> <ul style="list-style-type: none"> <li>• Policy (accountability)</li> </ul> <p>Consent</p> <ul style="list-style-type: none"> <li>• Policy (consent)</li> <li>• Tools (consent tools)</li> </ul> <p>Data Protection Regulation</p> <ul style="list-style-type: none"> <li>• Resource (communications on data protection regulations)</li> </ul> <p>Ethics Review Equivalency</p> <ul style="list-style-type: none"> <li>• Resource (models that allow for mutual recognition of ethics review)</li> </ul> <p>Machine-Readable Consent</p> <ul style="list-style-type: none"> <li>• Standard (data use types in consent forms)</li> </ul> <p>Paediatric</p> <ul style="list-style-type: none"> <li>• Policy (retention/release/future use of DNA obtained for newborn screening)</li> <li>• Standard (databases)</li> <li>• Resource (priority list of gene candidates)</li> </ul> <p>Privacy and Security</p> <ul style="list-style-type: none"> <li>• Policy (privacy and security)</li> </ul>	<p><b>Security</b></p> <p>Security Technology Infrastructure</p> <ul style="list-style-type: none"> <li>• Security Infrastructure specification</li> <li>• Standards and guidelines</li> </ul> <p>Software Security</p> <ul style="list-style-type: none"> <li>• Guidance</li> <li>• Reference implementation</li> </ul> <p>Cloud Security</p> <ul style="list-style-type: none"> <li>• Guidance for cloud security providers</li> <li>• Guidance for purchasers of cloud security services</li> </ul> <p>Incident Response (pending)</p> <ul style="list-style-type: none"> <li>• Incident oversight and response process</li> <li>• Process implementation</li> </ul> <p>Genomic Privacy Technology (pending)</p> <ul style="list-style-type: none"> <li>• Guidance</li> <li>• Reference implementation</li> </ul>
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### Appendix Key

*Resource:* documentation or reference material

*Tool:* a dynamic resource with a specific purpose or function

*Catalogue:* a dynamic, searchable tool that maps activity in a specific area

*Standard:* a benchmark or reference that may be used/applied broadly

*Policy:* a course or principle of action that guides or informs best practice

*Implementation:* an application of a software standard