ELIXIR::GA4GH Strategic Partnership

Overview

This document outlines the rationale and implementation of the Strategic Partnership between ELIXIR, the European bioinformatics life sciences Research Infrastructure, and the Global Alliance for Genomics and Health (GA4GH), the international standards organisation for genomics. This Strategic Partnership extends from the Collaboration Agreement that has been in place since November 2017 and describes the current and future complementary activities carried out by both organisations in order to establish and implement standards and design tools to overcome technical and regulatory hurdles that aid genomic data-sharing (GA4GH on an international scale, ELIXIR predominantly on an European scale).

Scientific Need

Large cohorts, with potentially millions of participants, are needed to understand the genetic and molecular signatures of diseases, and they provide a cornerstone for the creation of personalised treatments. To enable such datasets, over 200 genomic data initiatives are underway worldwide. However, national efforts will not be sufficient, and in recognition of this 20 European countries recently signed a Declaration to sequence and provide transnational access to at least 1M human genomes by 2022. Collectively, the European Declaration together with similar initiatives around the globe, will generate exabytes of data from millions of participants.

The ELIXIR::GA4GH Strategic Partnership will facilitate the responsible sharing of these sensitive data across Europe in order to help create virtual cohorts with tens of millions of participants.

Securely accessing personal genomic data at a population scale, and across national borders, is an enormous challenge that will require significant investments in national and international infrastructure. The scope and complexity of this infrastructure is beyond the capacity — and jurisdiction — of any single organisation; international collaboration is needed to provide recognised, secure, standardised, documented, and interoperable services. Building on the current Collaboration Agreement between ELIXIR and GA4GH, the ELIXIR::GA4GH Strategic Partnership will coordinate the generation and implementation of the GA4GH suite of interoperable standards and policy frameworks to overcome the technical and regulatory hurdles to genomic data sharing with a focus on Europe. In particular, when it comes to regulatory hurdles, the adoption of the European Regulation on Data Protection (GDPR) in 2016 has led to a new and more challenging situation concerning data protection, which does not only affect the EU but also international partners. The situation is further complicated by an opening clause that allows the countries operating under the GDPR to have different national

provisions for processing health and genetic data. ELIXIR with its broad representation of countries from the European Economic Area (EEA) can therefore bring in the European perspective and the respective nation legislations. These include among others provisions for organisational and technical safeguards to process genetic data, the legal basis for the processing and the possibilities to publish data in open access repositories. The ELIXIR Nodes can further link back to their data protection authorities for the needs of and the interpretation of the provisions for research.

The coordinated development and deployment of the ELIXIR Beacon and ELIXIR Authentication and Authorisation Infrastructure (AAI) standards, along with the suite of standards from the GA4GH Cloud and Large Scale Genomics Work Streams will help translate this vision into reality. GA4GH's interoperable APIs solve a number of technical challenges. For instance, the Data Repository Service (DRS), Authentication and Authorization Infrastructure, htsget, refget, and Data User and Researcher Identity standards (e.g. Data Use Ontology) all aim to enable researchers to discover and obtain secured access to genomic data if the data can be moved outside of national jurisdictions. The ELIXIR Rare Diseases Community has recently implemented a number of the GA4GH standards and the ELIXIR 2019-23 programme demonstrates continued support to implement and deploy open standards working alongside GA4GH.

Many countries in Europe now have nascent personalised medicine programmes meaning that human genomics is undergoing a step change from being a predominantly research-driven activity to one funded through healthcare. However genetic data generated through healthcare is not likely to be shared as widely as research data. Healthcare is subject to national laws, and it is often unacceptable for health data from one country to be exported outside regional or national jurisdictions. When data cannot be moved outside jurisdictions, the GA4GH Cloud standards (Workflow Execution Service (WES), Task Execution Service (TES), and Tools Registry Service (TRS)) allow users to analyse sensitive data in a virtual environment. The ELIXIR::GA4GH Strategic Partnership aims to provide technical solutions based on the constraints of the data providers and the requirements of the researchers while operating in a federated ecosystem of implementers, therefore becoming a solution for implementation as part of the EU 1 Million Genomes Declaration.

In May 2018, the European General Data Protection Regulation (GDPR) came into force. For example, the GA4GH DURI workstream handles researchers own information that is subject to GDPR. Ethical, Legal and Social Implications (ELSI) concerns for all infrastructures dealing with sensitive human health data are very similar with respect to privacy, consent, protection of personal data, differences in national legislation, and their implementation(s). ELIXIR has held two GDPR focussed workshops, where the approved GA4GH specification (and Driver Project) Beacon has been presented and discussed as a Use Case in order to better understand the impact and implementation of the regulation across the ELIXIR Nodes. Additionally, the ELIXIR Italy Node has held a specific 'Beacon and the GDPR' focussed workshop. The ELIXIR::GA4GH Strategic

Partnership provides a strong opportunity to link work ongoing within ELIXIR in relation to GDPR and ELISI to the foundational GA4GH Regulatory and Ethics Work Stream.

Operational Need

Launched in 2017, the ELIXIR::GA4GH Collaboration Agreement (CA) aimed to affirm ELIXIR's influence on the standards and technology being established for human genomic data-sharing by GA4GH and expand European participation within GA4GH. Through the CA, the two parties committed to supporting ELIXIR's participation in the development of GA4GH standards, and its dissemination of those standards across the community, predominantly across Europe but also on an international scale via global collaborations and initiatives.

Since 2017 the membership of ELIXIR and GA4GH (both in terms of global coverage and individual institutes) has grown significantly; and has resulted in more interactions between the organisations, at project and Work Stream levels (Figure 1). As of January 2019 more than 60% of ELIXIR Nodes have contributed to at least one GA4GH Work Stream and individual ELIXIR members currently hold co-leadership positions in four of the six GA4GH Technical Work Streams (Table 1) or co-lead individual Working Groups within those Work Streams.

The objectives of the Collaboration Agreement have now been achieved and activities have laid a foundation for stronger collaboration between the two organisations going forward.

GA4GH recognizes that it has limited capacity to solicit community involvement through Driver Project relationships alone. Having recently added seven new Driver Projects, it is now managing collaborations with 22 disparate genomic data projects with varying levels of complexity. As GA4GH continues to mature as the global standards organization for genomics, it will need to develop a mechanism to engage the community at a higher level. A Strategic Partnership with ELIXIR will help achieve this by connecting GA4GH to the vast network of European life science initiatives through a single touch point.

Additionally, the partnership will strengthen ELIXIR's influence on GA4GH standards development while still encouraging engagement, participation, and collaboration across the global community. Indeed, GA4GH envisions future Strategic Partnerships with other jurisdictions, which are likely to be modeled on the ELIXIR Partnership. The expectation from GA4GH is that all Driver Projects will over time graduate or be taken into a Strategic Partnership.

Figure 1: ELIXIR Platform and Community activities (contributing, adopting, implementing) across the GA4GH Work Streams.

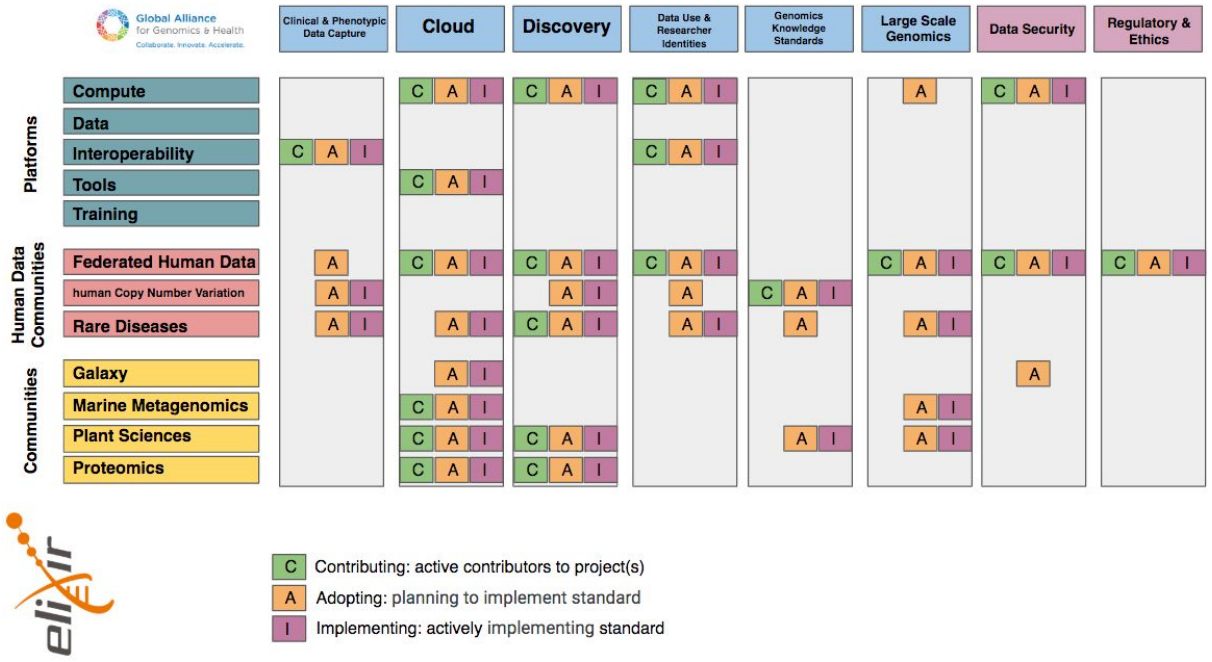


Table 1: Leadership structure of the six GA4GH Technical Work Streams. (* denotes ELIXIR member)

GA4GH Work Stream	Co-leads
Clinical and Phenotypic Data Capture	Melissa Haendel (Oregon State University) David Hansen (Australian e-Health Research Centre)
Cloud	David Glazer (Verily) Brian O'Connor (UCSC)
Data Use & Researcher Identities	Moran Cabili (Foundation Medicine) Tommi Nyrönen* (CSC)
Discovery	Marc Fiume (DNAStack) Michael Baudis* (University of Zurich)
Genomics Knowledge Standards	Bob Freimuth (Mayo Clinic) Andy Yates* (EMBL-EBI)
Large Scale Genomics	Oliver Hofmann (University of Melbourne) Thomas Keane* (EMBL-EBI)

The added value of the ELIXIR::GA4GH Strategic Partnership

ELIXIR is an active contributing member of GA4GH, and has provided funding towards both standards generation and implementation since 2015. As we move into the next phase of this collaboration, the two organisations will form a Strategic Partnership to ensure that:

1) A collaborative arrangement is formed that can influence standards and technology being established for human genomic data-sharing in the ELIXIR and GA4GH organisations through a role in the GA4GH Technical Work Streams that is stronger than any individual Driver Project, or suite of Driver Projects - ensuring standards are fit for purpose across multiple European countries/ELIXIR Nodes and rolled out across Europe
2) The Strategic Partnership model presents a scalable solution to the growth of both ELIXIR and GA4GH, and becomes a model for other organisations that are invited to form this level of relationship
3) ELIXIR::GA4GH Strategic Partnership roles provide (a) clear coordination and structure for the ELIXIR::GA4GH touchpoints and (b) an effective, efficient mechanism to raise issues within the ELIXIR governance structure that can then be delivered to GA4GH
4) ELIXIR will seek to sponsor GA4GH Plenary Meetings and both ELIXIR and GA4GH will aim to co-host workshops and meetings where relevant