



# ELIXIR Beacon Project

*Networking Resources Across and Beyond ELIXIR Human Data Communities*

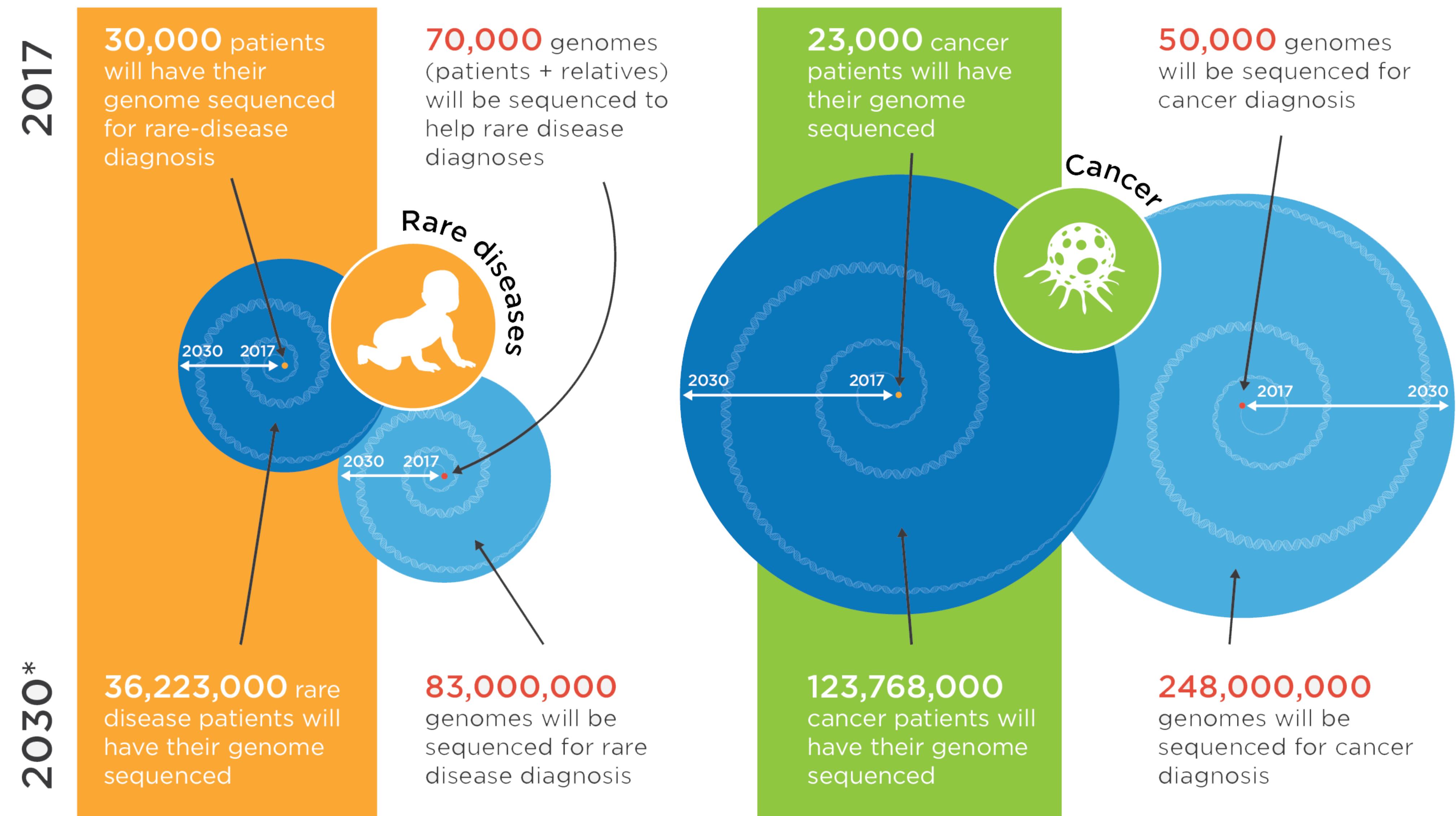
Michael Baudis :: ELIXIR-CH:: ELIXIR Day :: 2020-02-06



Global Alliance  
for Genomics & Health



University of  
Zurich UZH



\* Projected figures, based on current data and known status of genomics initiatives worldwide.

# The vision: Federation of data



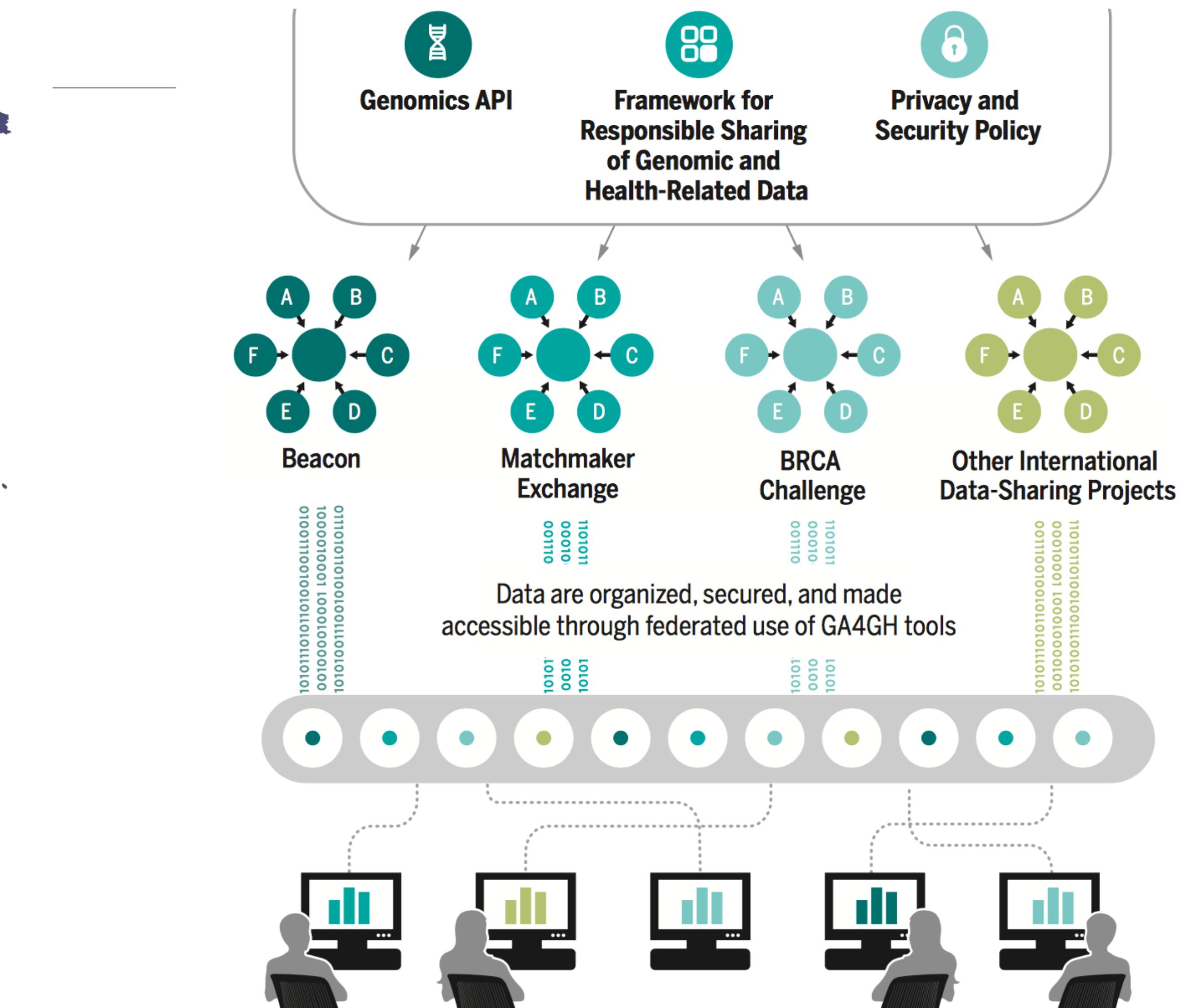


## GENOMICS

# *A federated ecosystem for sharing genomic, clinical data*

Silos of genome data collection are being transformed into seamlessly connected, independent systems

**A federated data ecosystem.** To share genomic data globally, this approach furthers medical research without requiring compatible data sets or compromising patient identity.



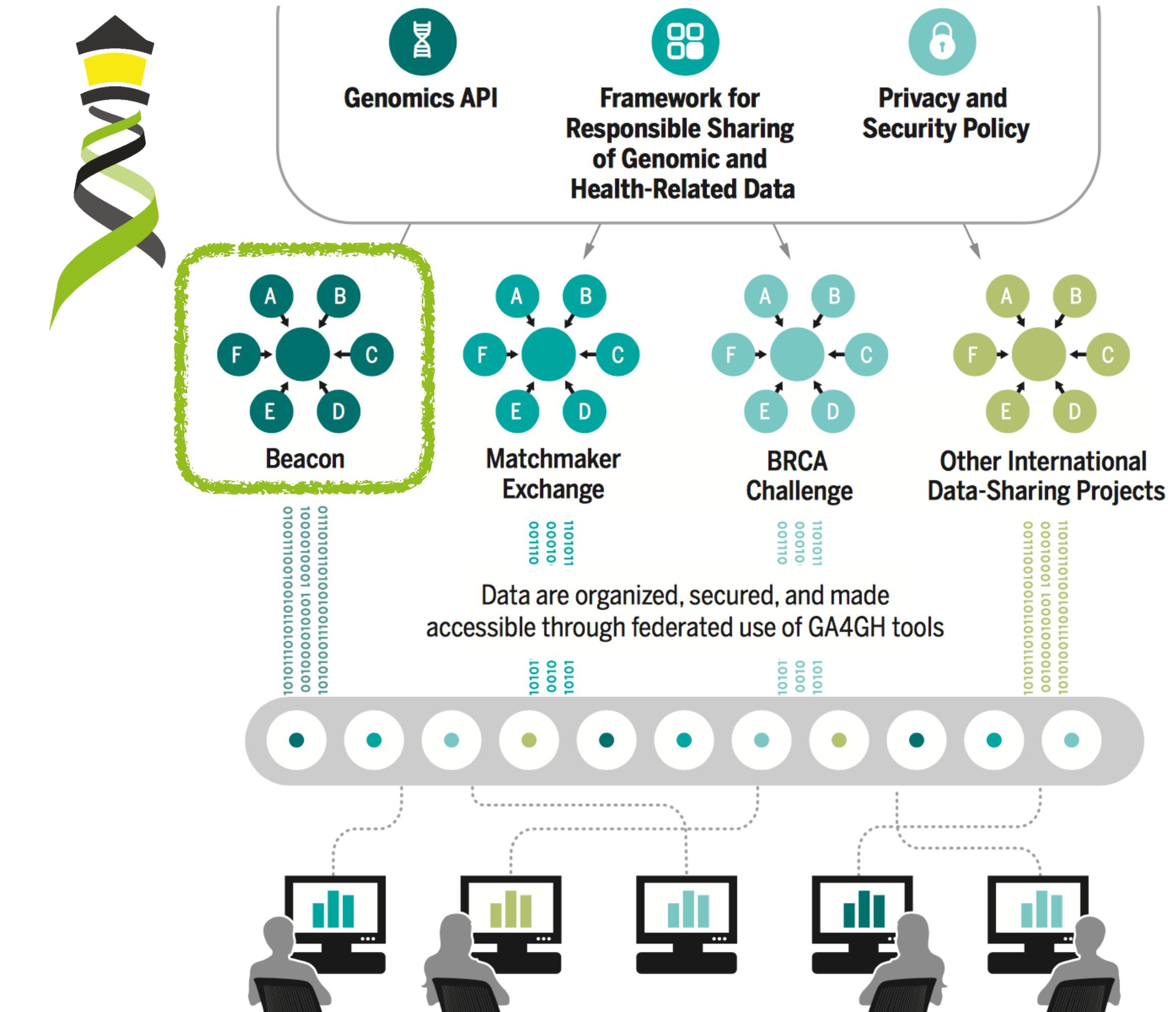


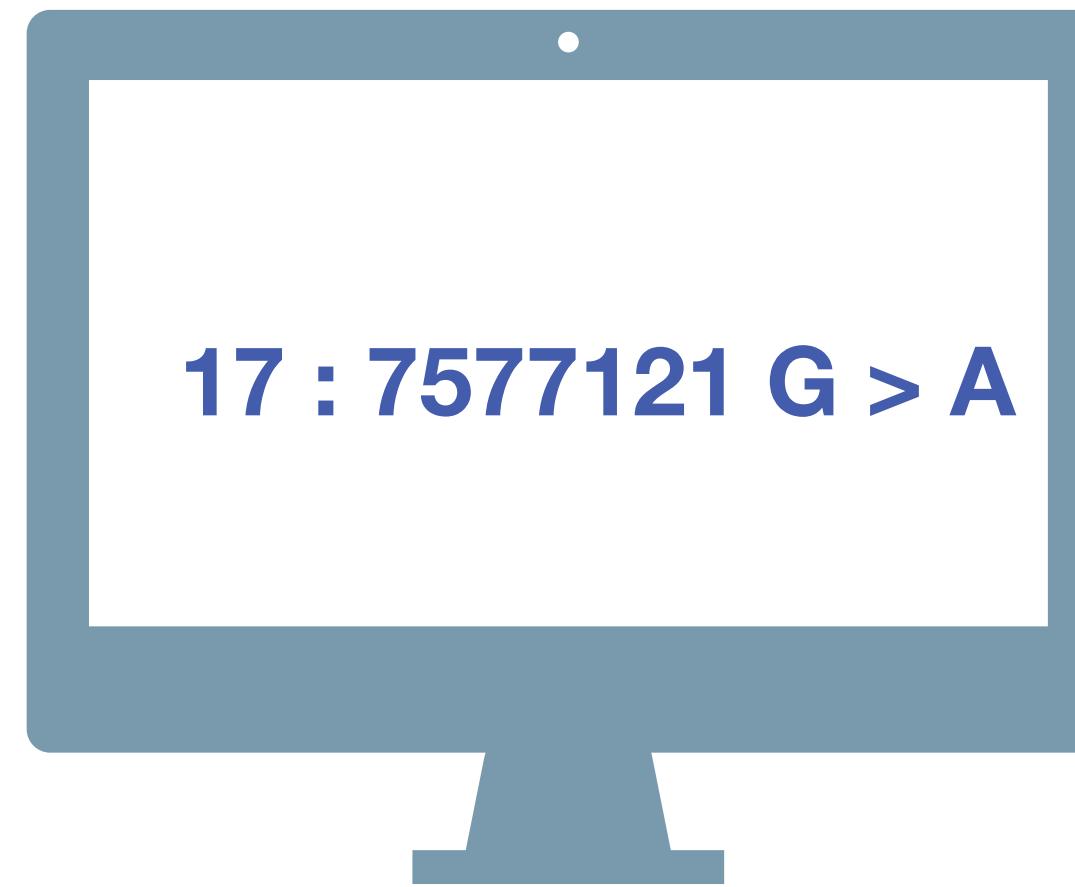
GENOMICS

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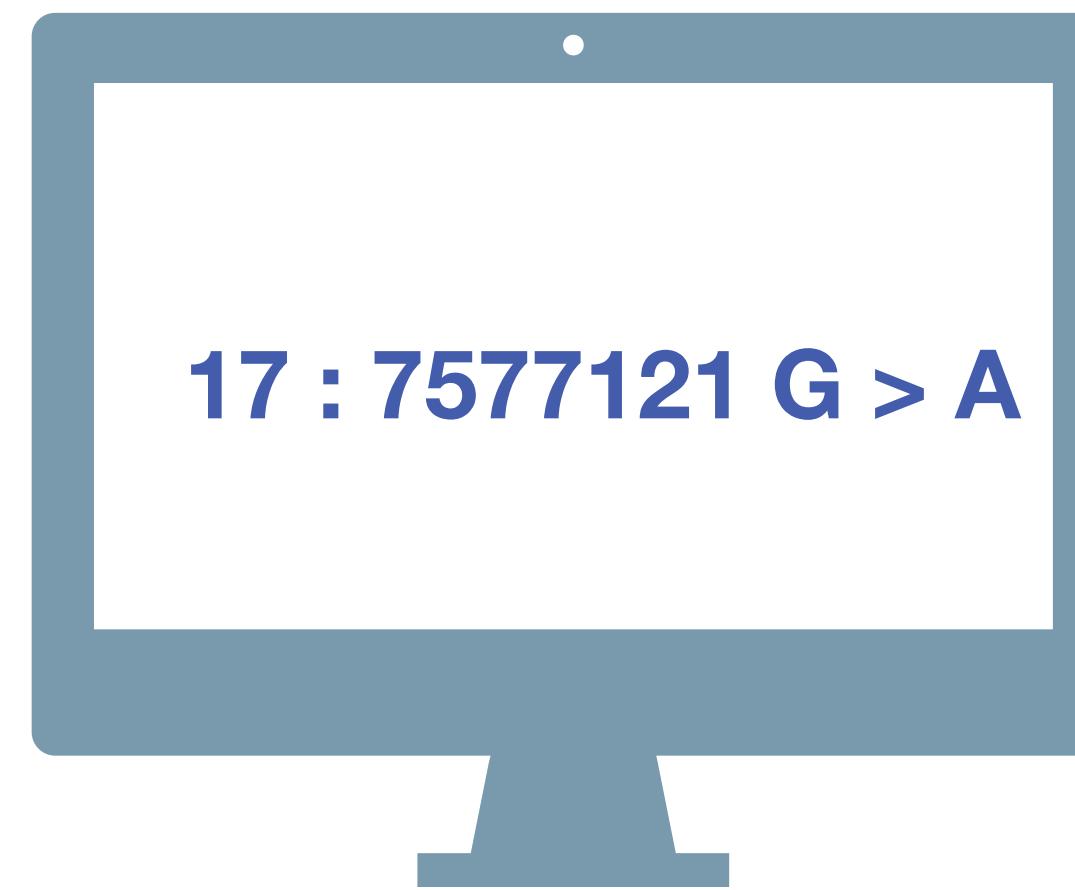




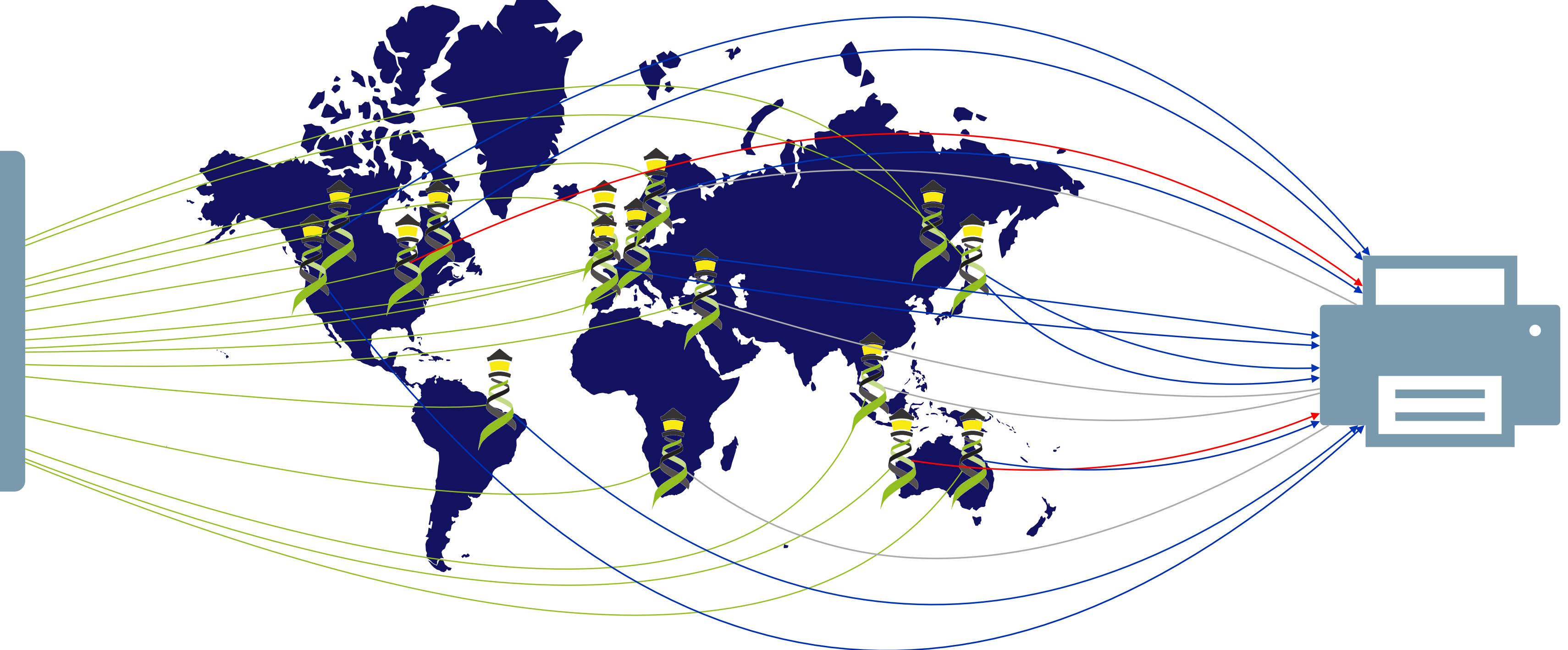
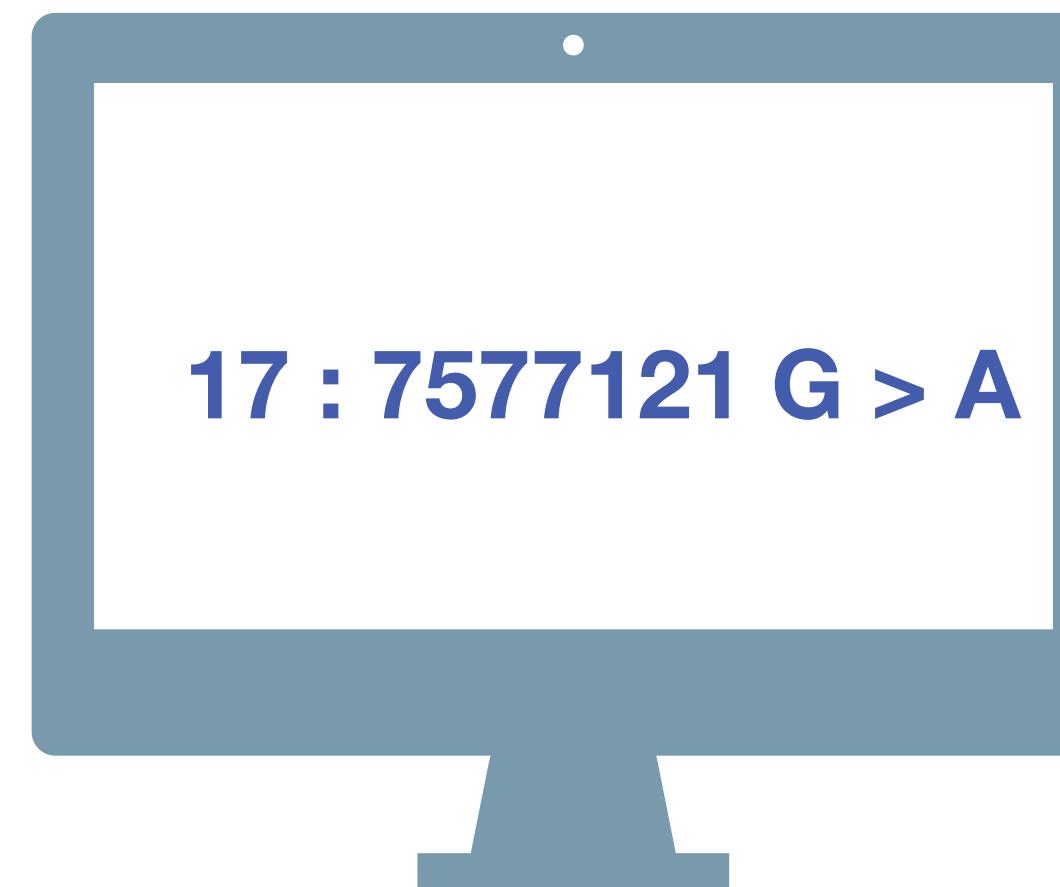
# Beacon

A **Beacon** answers a query for a specific genome variant against individual or aggregate genome collections

**YES | NO | \0**



A Beacon network federates  
*genome variant queries*  
across databases that  
support the ***Beacon API***



Have you seen this variant?  
It came up in my patient  
and we don't know if this is  
a common SNP or worth  
following up.

A Beacon network federates  
genome variant queries  
across databases that  
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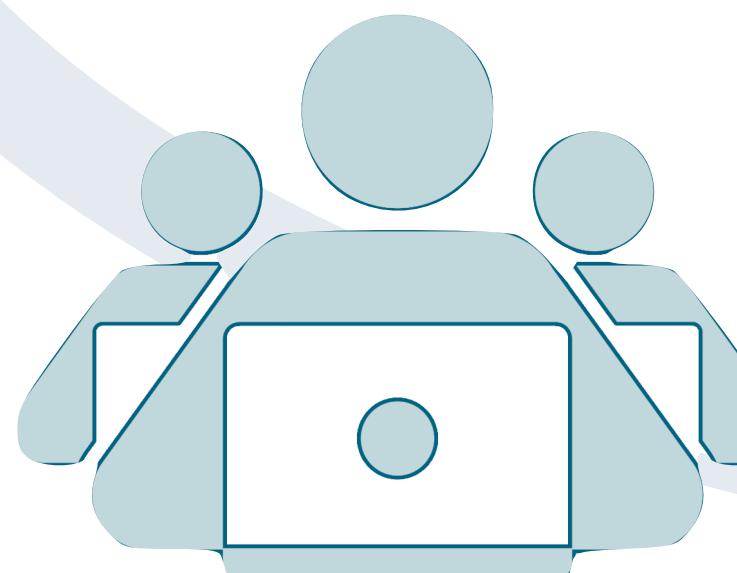
Here: The variant has  
been found in **few**  
resources, and those  
are from **disease**  
specific **collections**.

# GA4GH & ELIXIR - Partnering for Standards and Tools in Genomics and Health



**Global Alliance**  
for Genomics & Health  
Collaborate. Innovate. Accelerate.

Simplify the way people search for and request access to potentially identifiable data in international and national genomic data resources



# ELIXIR Beacon Project

- Driver project on GA4GH roadmap
- aligns with Discovery Work Stream
- strong impact on GA4GH developments as a concrete, funded project

The screenshot shows two cards. The left card is titled 'Driver Projects' and contains text about real-world genomic data initiatives. The right card is titled 'ELIXIR Beacon' and provides links to its implementation studies and champions.

**Driver Projects**  
GA4GH Driver Projects are real-world genomic data initiatives that help guide our development efforts and pilot our tools. Stakeholders around the globe advocate, mandate, implement, and use our frameworks and standards in their local contexts.

**ELIXIR Beacon**  
<https://www.elixir-europe.org/about/implementation-studies/beacons>

Europe  
**Champions:** Jordi Rambla, Juha Tornroos, Gary Saunders

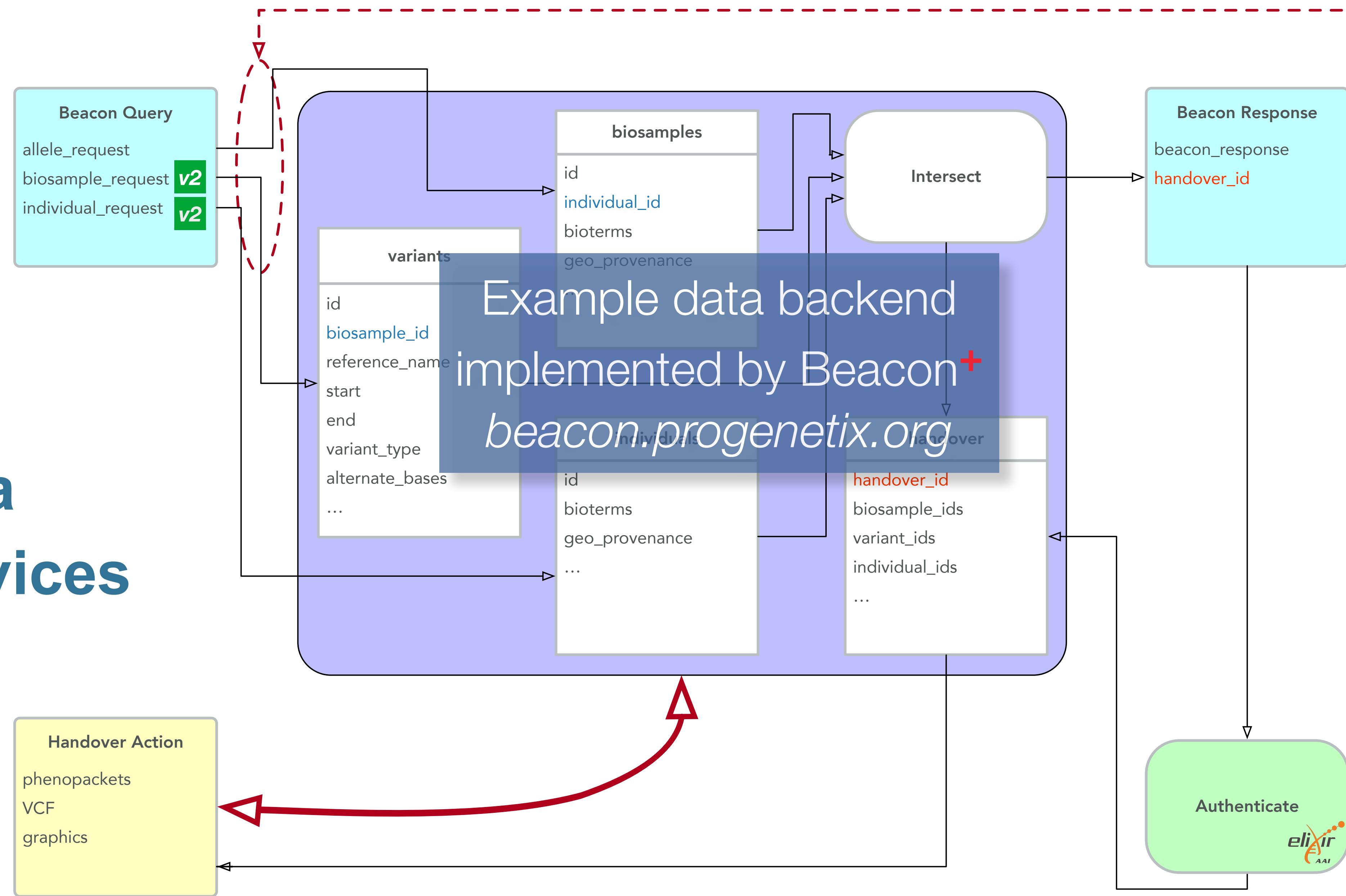
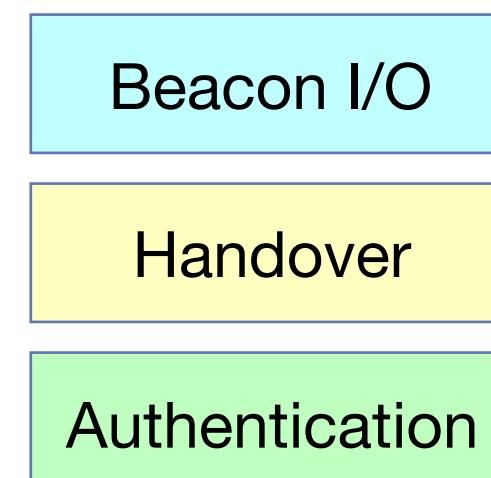
## v1.1 and roadmap

- structural variations** (DUP, DEL) in addition to SNV
- ... more structural queries (translocations/fusions...)
- Beacon queries as entry for **data handover** (outside Beacon protocol)
- layered authentication system using **ELIXIR AAI**
- v2** **filters** for phenotypic & technical metadata
- v2** Extended quantitative responses
- Ubiquitous **deployment** (e.g. throughout ELIXIR network)



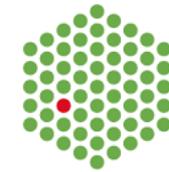
# Beacon & Handover

Beacons v1.1  
supports data  
delivery services



# ELIXIR Beacons

EMBL-EBI



elixir  
FINLAND

elixir  
SWEDEN

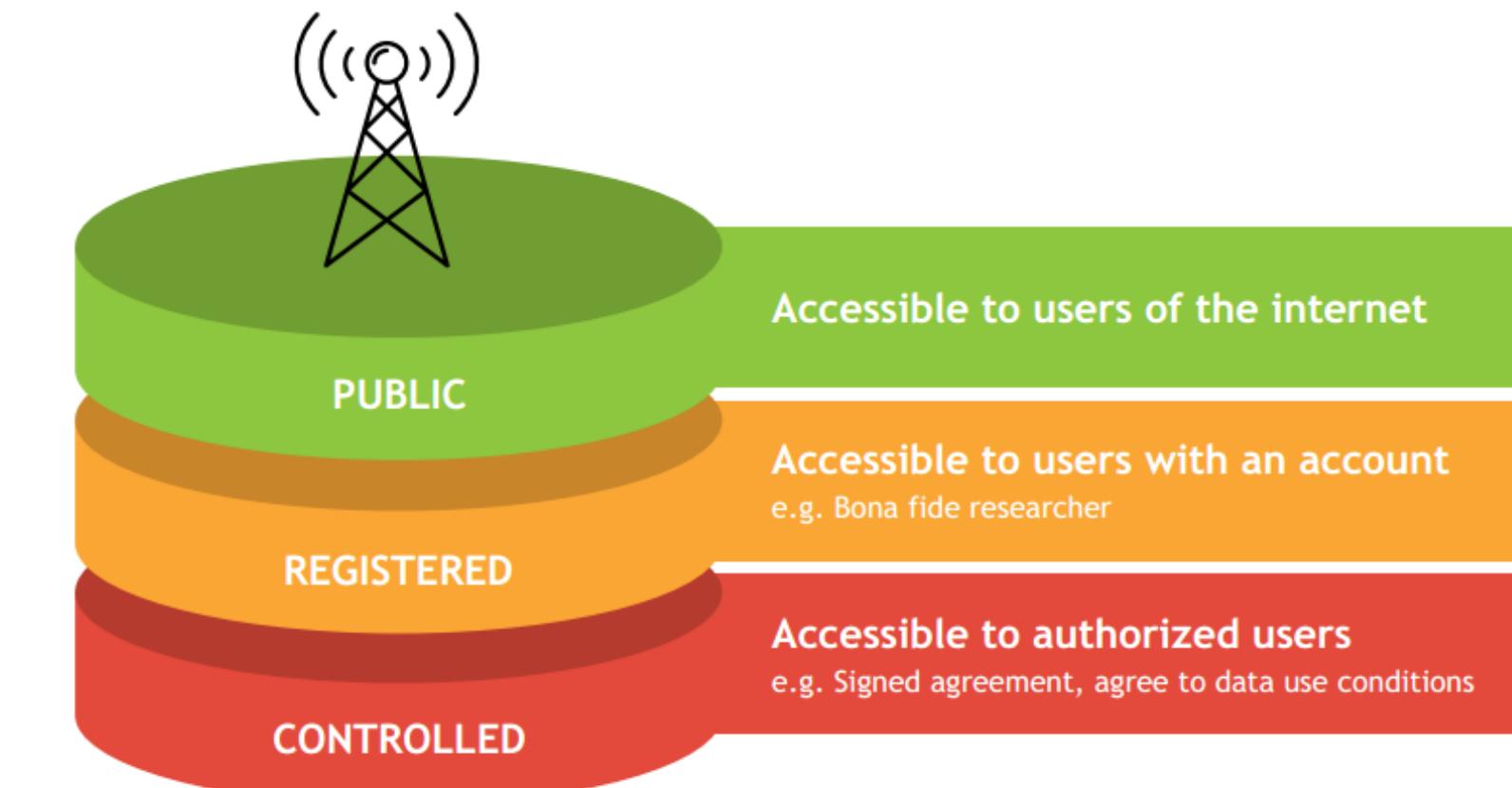
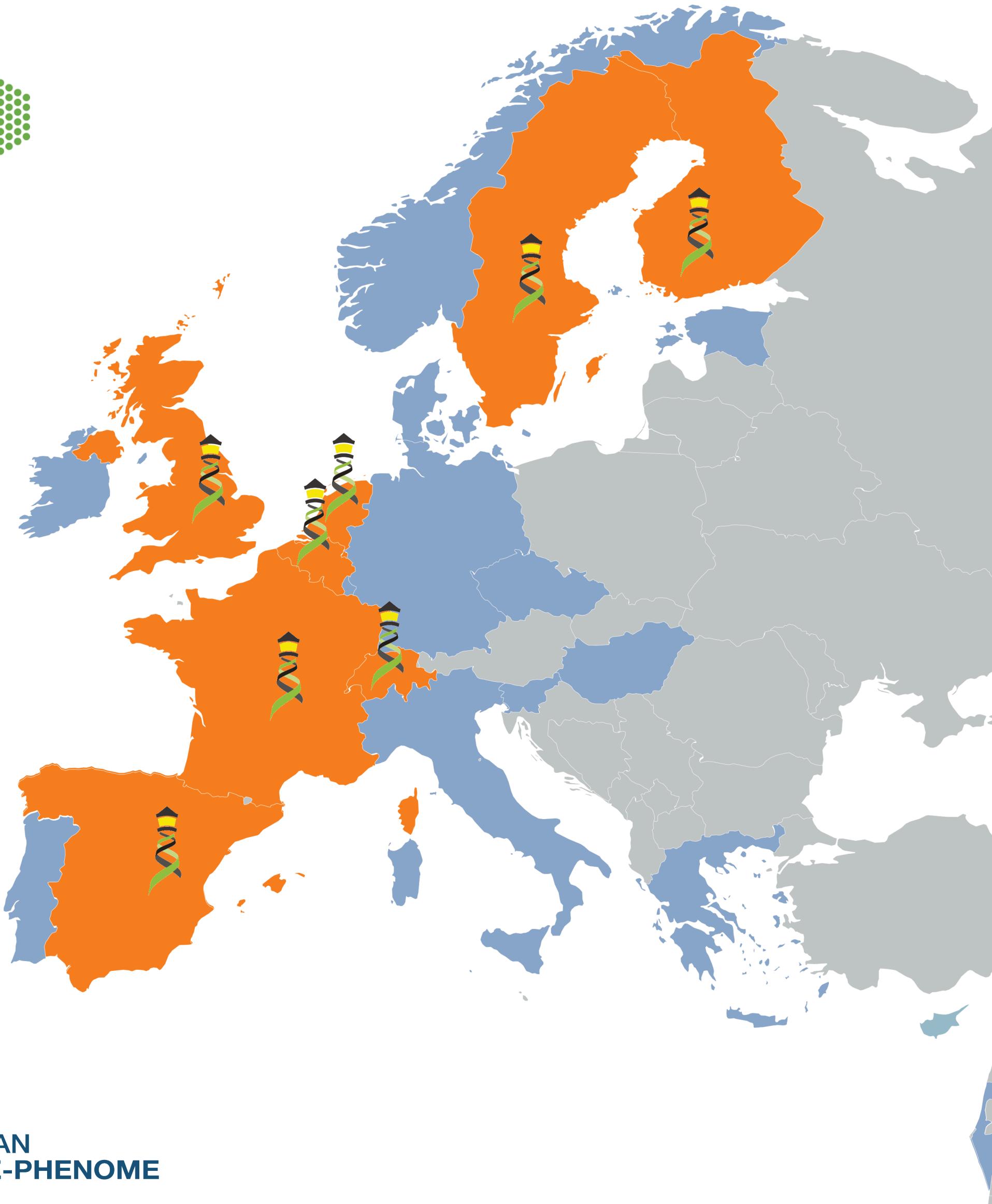
elixir  
BELGIUM

elixir  
NETHERLANDS

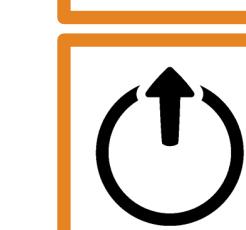
elixir  
SWITZERLAND

elixir  
SPAIN

EUROPEAN  
GENOME-PHENOME  
ARCHIVE



elixir  
AAI



Driving implementation of Beacon technology in ELIXIR Nodes

→ 9 National Nodes have lit Beacons

ELIXIR Authentication and Authorization Infrastructure (AAI)

Beacon



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elixir

# ELIXIR Beacon Network

- developed under lead from ELIXIR Finland
- **authenticated access w/ ELIXIR AAI**
- **incremental extension**, starting with ELIXIR Beacon resources adhering to the **latest specification** (contrast to legacy networks)
- service details provided by individual Beacons, using **GA4GH service-info**
- **registration service**
  - **integrator throughout ELIXIR Human Data**
  - **starting point for "beyond ELIXIR" feature rich federated Beacon services**



GRCh38 ▾ 17 : 7577121 G > A Search

[Example variant query](#) [Advanced Search](#)

**baudisgroup at UZH and SIB**  
Progenetix Cancer Genomics Beacon+

Beacon+ provides a forward looking implementation of the Beacon API, with focus on structural variants and metadata based on the cancer and reference genome profiling data represented in the Progenetix oncogenomic data resource (<https://progenetix.org>).

[Visit Us](#) · [Beacon API](#) · [Contact Us](#)

**CSC - IT Center for Science**  
Development Beacon

Beacon API Web Server based on the GA4GH Beacon API

[Visit Us](#) · [Beacon API](#) · [Contact Us](#)

**Research Programme on Biomedical Informatics**  
DisGeNET Beacon

Variant-Disease associations collected from curated resources and the literature

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**University of Tartu Institute of Genomics, Estonia**  
Beacon at the University of Tartu, Estonia

Beacon API Web Server based on the GA4GH Beacon API

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**National Bioinformatics Infrastructure Sweden**  
SweFreq Beacon

Beacon API Web Server based on the GA4GH Beacon API

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**LCSB at University of Luxembourg**  
ELIXIR.LU Beacon

ELIXIR.LU Beacon

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**European Genome-Phenome Archive (EGA)**  
EGA Beacon

This [Beacon](https://beacon-project.io/) is based on the GA4GH Beacon [v1.1.0](https://github.com/ga4gh/beacon/specification/blob/develop/beacon.yaml)

[Visit Us](#) · [Beacon API](#) · [Contact Us](#)

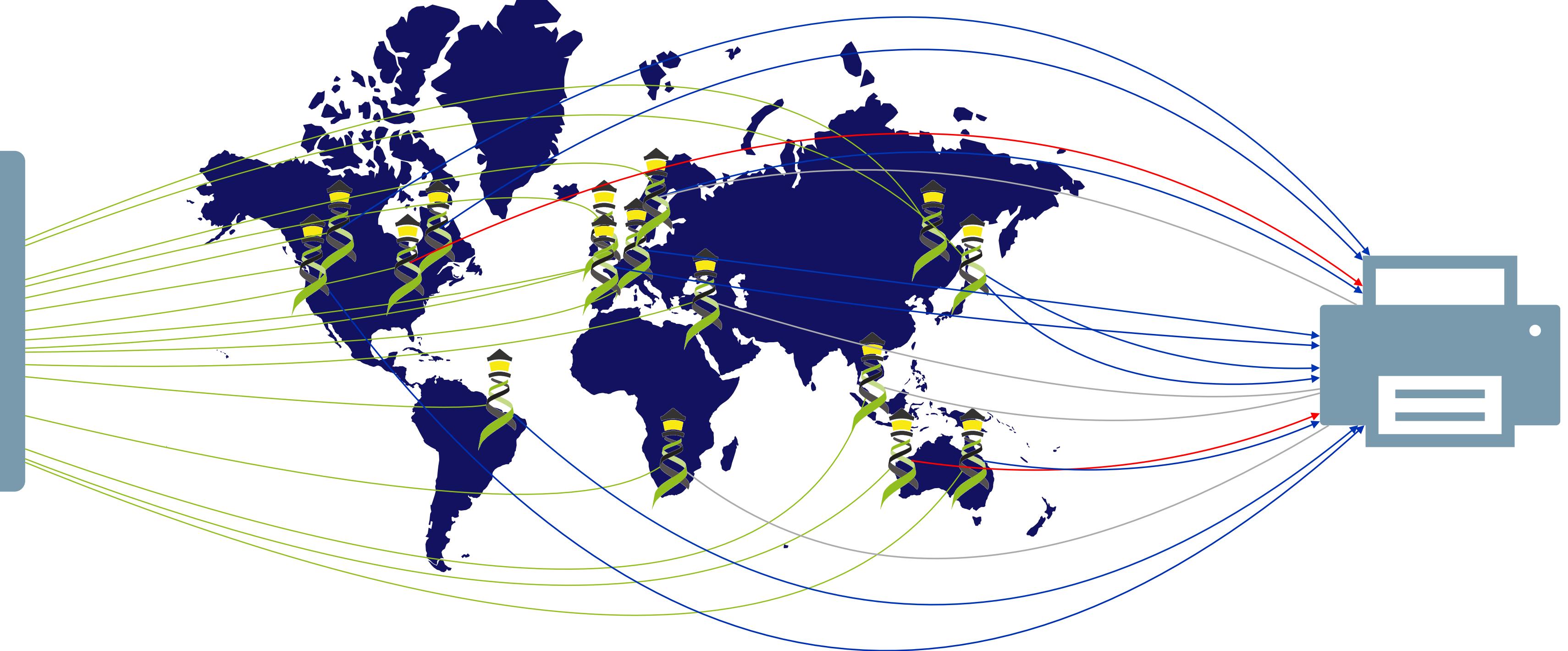
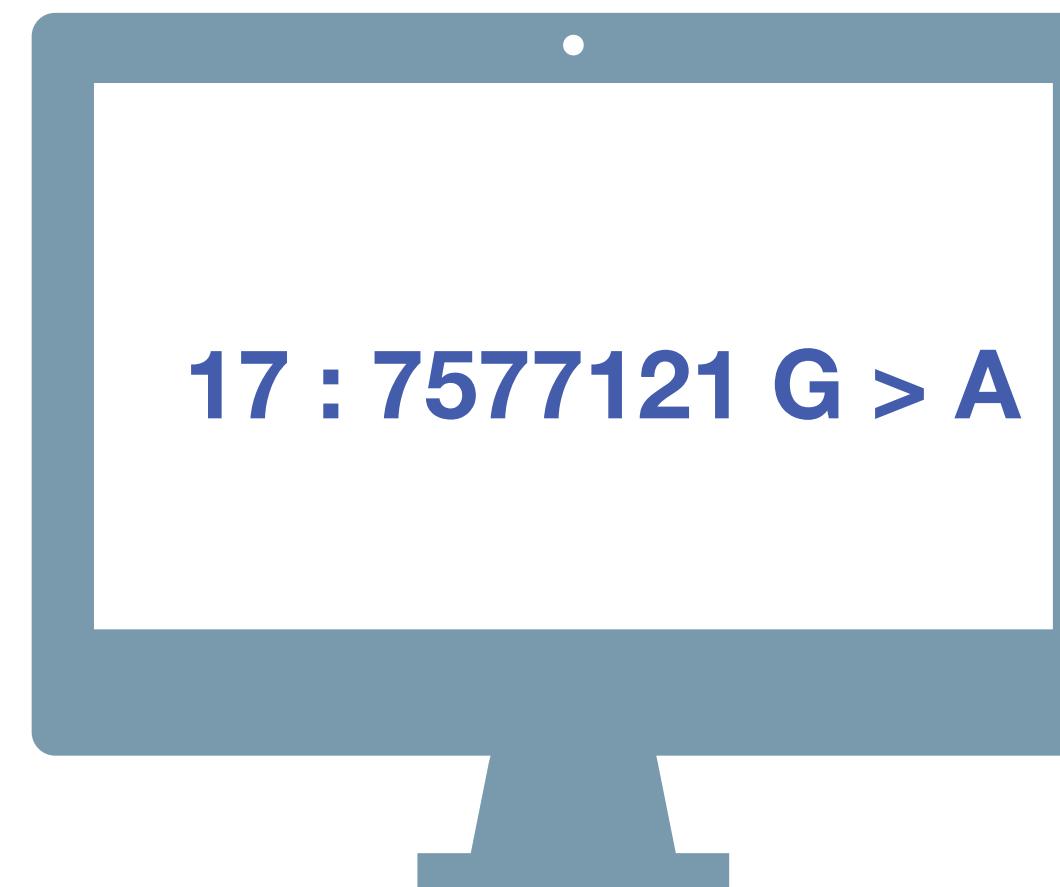
**CSC - IT Center for Science**  
Production Beacon

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# Beacon v2 - Areas of Change

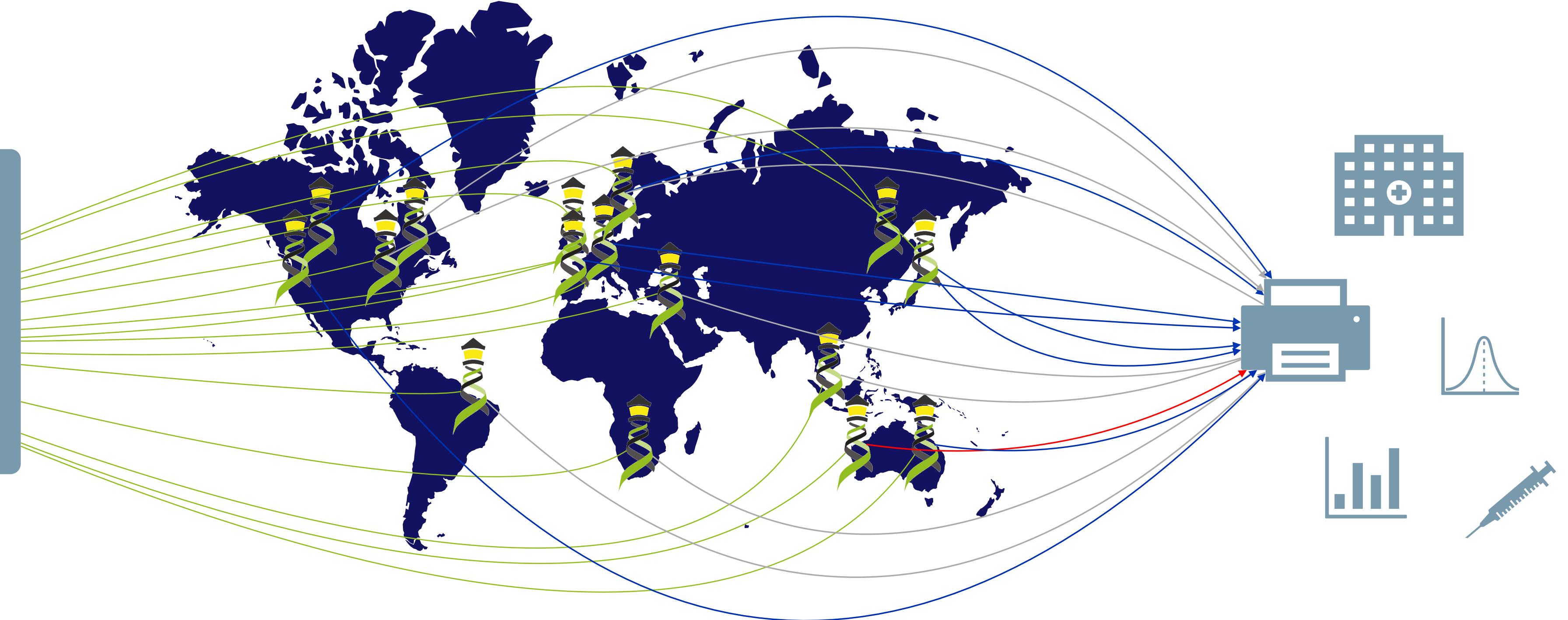
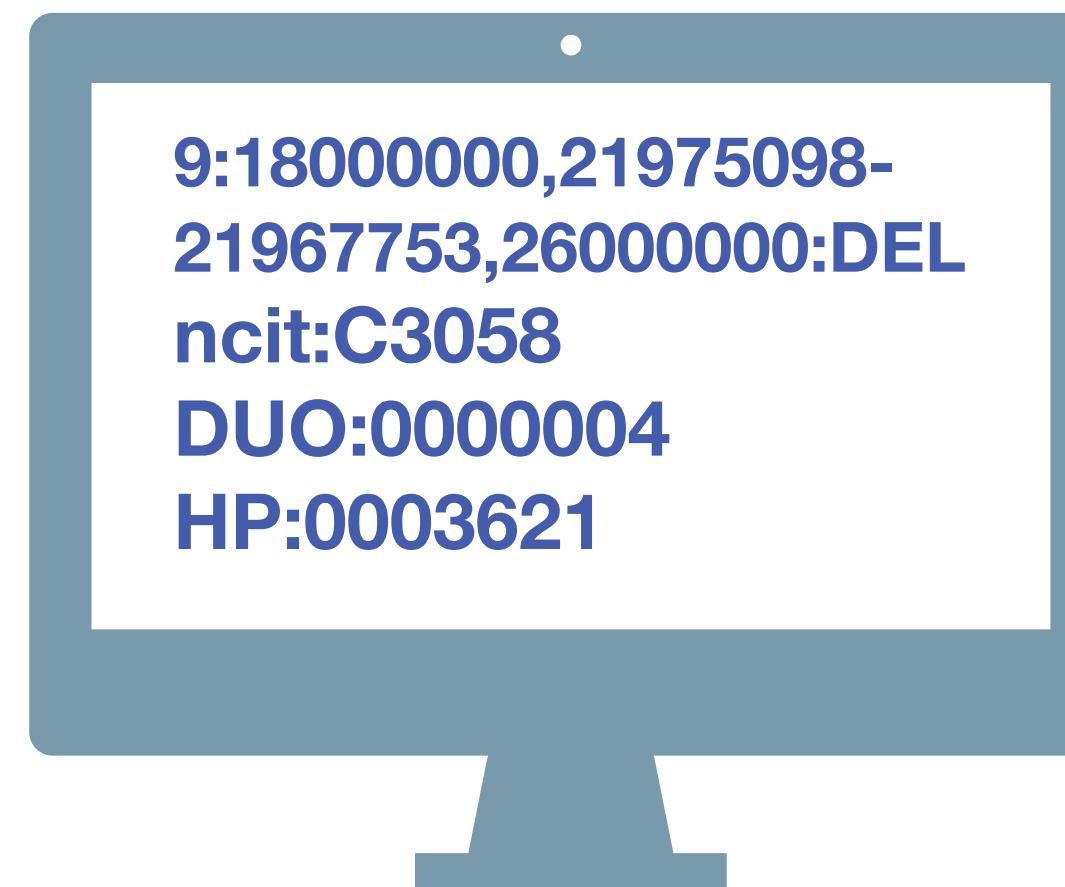
- Separate query types for different genomic variants
    - SNPs **BeaconSnpRequest**
    - Structural Variants **BeaconCnvRequest**
    - Region **BeaconRangeRequest**
    - ...
  - Access levels
  - Filters
    - Simple general filter schema w/ **scoping through prefixes** (CURIEs, private implementations)
  - New types of queries:
    - By sample, patient, variant effect/evidence
    - Complex queries? (stakeholder driven; e.g. EJP-RD, GEL...)
  - Schema versions & Service Info
    - Negotiated queries based on individual Beacon capabilities
- 
- The diagram uses yellow curly braces to group items. The first group, containing the first four bullet points, is associated with 'v2.0' and a green arrow pointing right, indicating GA4GH approval process ("major product update"). The second group, containing the last three bullet points, is associated with 'v2.n' and a yellow arrow pointing right, indicating incremental rollout after v2.0.
- Tested and already implemented by Beacons      **v2.0**
- Ongoing      **v2.n**



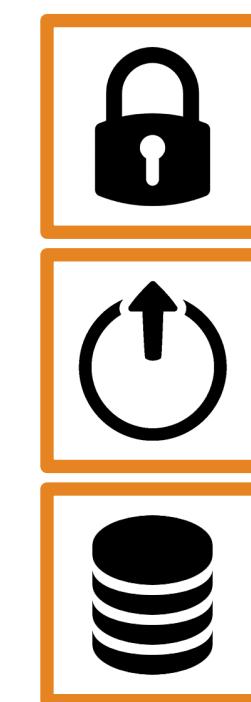
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Have you seen deletions in this region on chromosome 9 in Glioblastomas from a juvenile patient, in a dataset with unrestricted access?



## Beacon v2 API

The Beacon API v2 proposal opens the way for the design of a simple but powerful "genomics API".

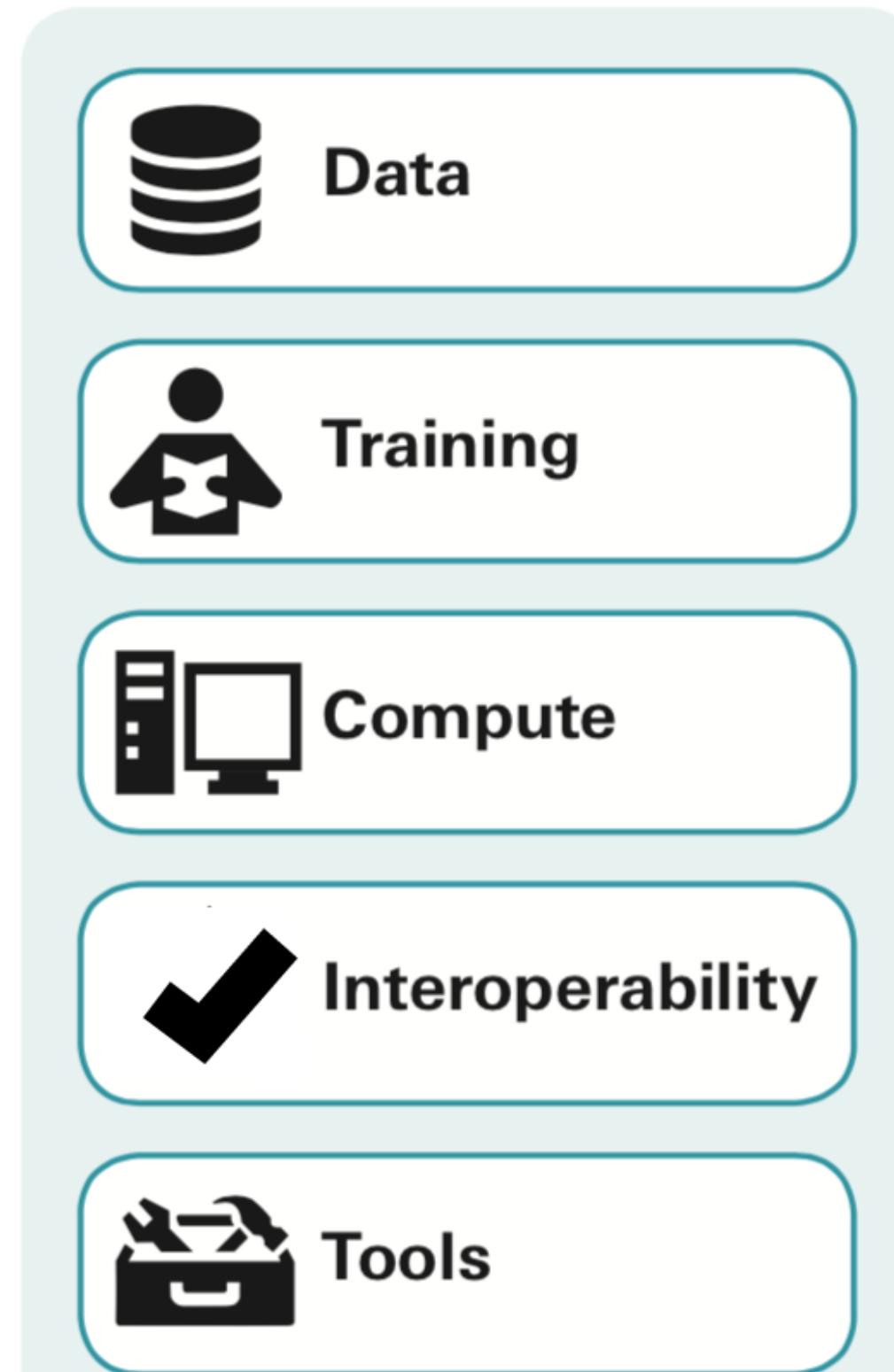


# Beacon Project - Partner Engagement & Next Steps

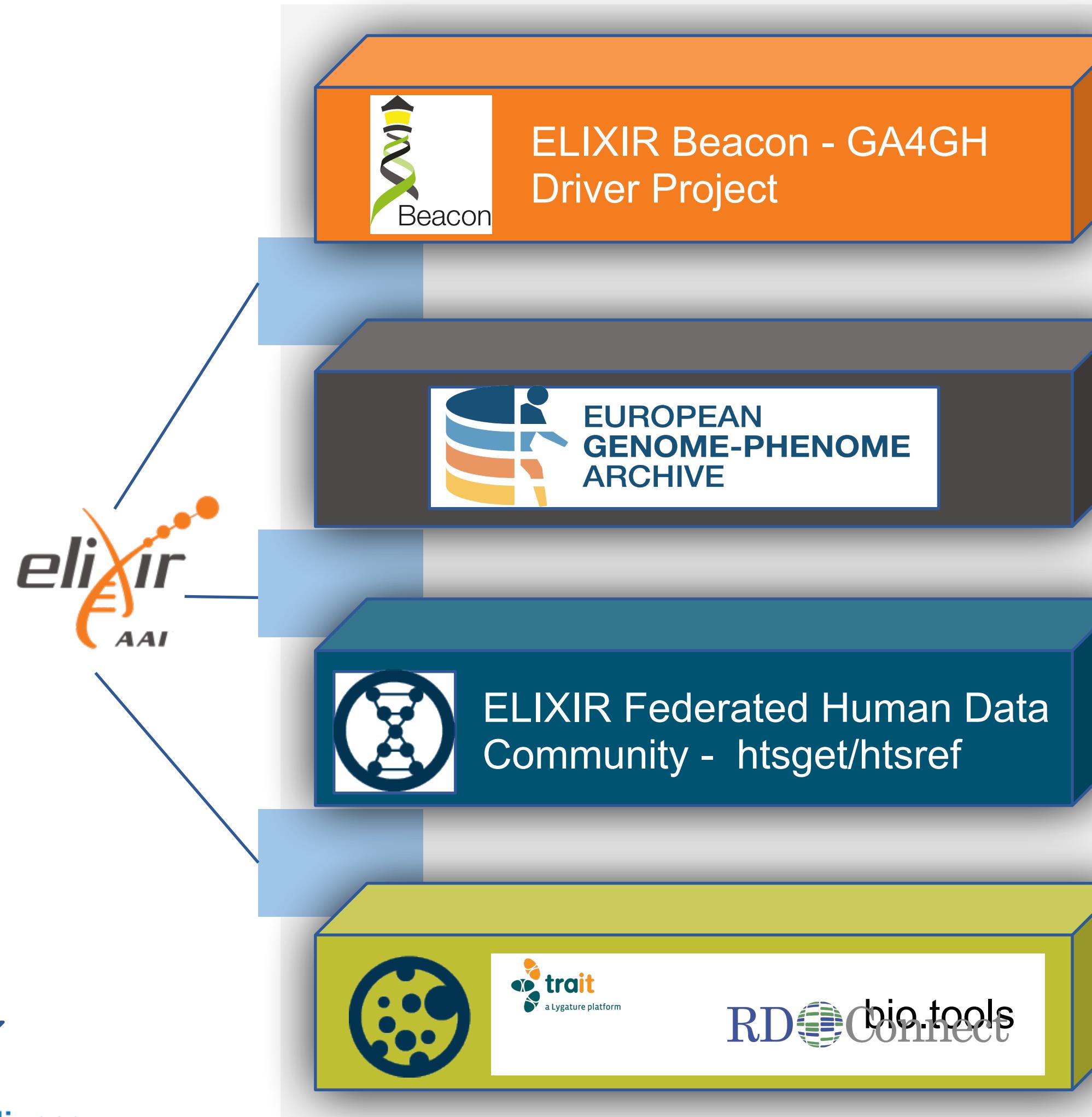
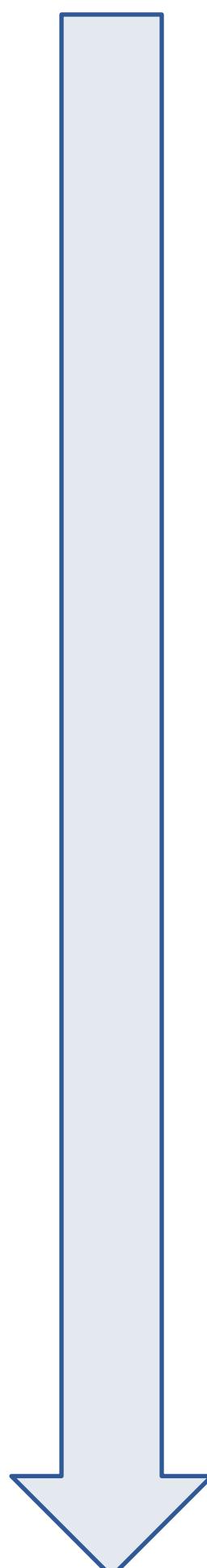
- Working with **partner communities & projects** on *deploying Beacons*
  - ELIXIR hCNV Community
  - European Joint Program on Rare Diseases
  - clinical groups & data initiatives (e.g. Andalucia, Cancer Core Europe, SPHN)
  - variant annotation resources, with optional clinical components (e.g. SVIP-O)
- Improving reference implementation and standards / **compliance** testing
- Beacon **v2** "fast forward" development
- aligning w/ GA4GH standards, through "request & adopt" => SchemaBlocks **{S}[B]**
- networks **throughout & beyond ELIXIR**



# Beacon as Part of ELIXIR Human Genomics & Translational Data



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## Data Discoverability

Federating lightweight discoverability of data, and datasets across ELIXIR

## Data Archival

Utilising the ELIXIR Deposition Databases to ensure secure, long-term, efficient archival of data

## Federated Data Access

Coordinating a collection of interoperable EGA-like resources to ensure secure management of sensitive data across the ELIXIR Nodes

## Data Analysis

Bringing ‘analysis to data’ via common workflow languages, workflows, containers, and tools



The ***ELIXIR Beacon Project*** has a central position in the alignment of **ELIXIR human data communities** and other large European data initiatives.

Through its partnership with **GA4GH**, Beacon drives the **world-wide** development, coordination and uptake of genomics standards.

The **Beacon Project** mirrors the **core mission of GA4GH**, driven forward through ELIXIR.





**ELIXIR Genome**

**Beacons**

A Driver Project of the Global  
Alliance for Genomics and  
Health

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**Examples, Guides & FAQ**

**Specification**

**Roadmap**

**Beacon Networks**

**Meeting Minutes**

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Beacon @ ELIXIR

GA4GH

Beacon+

beacon-network.org

GA4GH::SchemaBlocks

GA4GH::Discovery

GA4GH::CLP

GA4GH::GKS

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## Roadmap

The ELIXIR Beacon Roadmap delineates short-, mid- and long-term objectives, to expand functional scope and reach of Beacon as a protocol and genomic data ecosystem.

### Beacon Flavours

Beacons may be able to increase their functionality through the development of "flavours". These can extend the core Beacon concept for specific use cases.

@mbaudis 2018-10-24: more ...

### Bio-metadata Query Support

Future Beacon API versions will support querying for additional, non-standard genomic data.

@mbaudis 2018-10-18: more ...

### EvidenceBeacon Notes - GA4GHconnect 2018

The topic of "EvidenceBeacon" was discussed with many different attendees during the GA4GHconnect 2018 session and beyond, leading to some clearer picture about the (widely) different approaches.

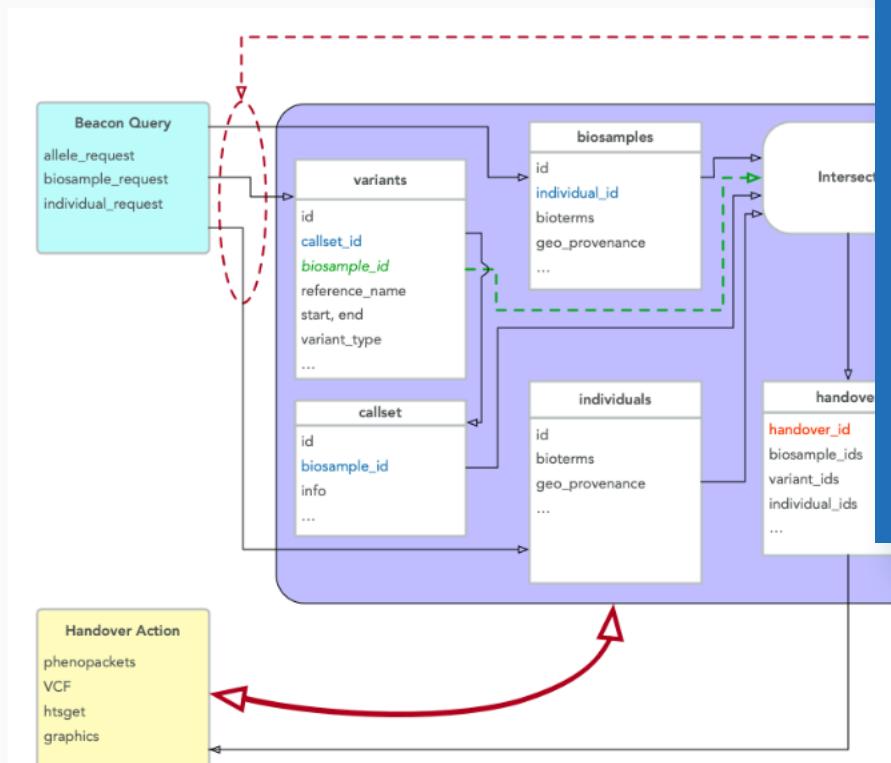
@mbaudis 2019-04-30: more ...

### [H→O] Beacon Handover for Data Delivery

While the Beacon response should be restricted to aggregate data (yes/no), the usage of the protocol could be greatly expanded by providing an access to the underlying data objects matched by a Beacon query.

As part of the mid-term product strategy, the ELIXIR Beacon team is evaluating the possibility of extending the protocol, in which rich data content (e.g. variant data, phenotypic information, linked services, etc.) can be provided from linked services, initiated through a Beacon query. This would involve several steps like protocol selection, authentication...). A discussion of the topic is ongoing in the developer area on Github (issue #114).

As of 2018-11-13, the **handover** concept has become part of the ongoing specification.



beacon-project.io



## Beacon

Beacon Project, Global Alliance for Genomics & Health.

# API Leads

## Jordi Rambla

## Anthony Brooks

## Juha Törnroos

# Discovery WS

## Michael Baudis (Beacon)

## Marc Fiume (Networks)

# ELIXIR

## Gary Saunders

## David Lloyd

## Serena Scollen

openapi beacon ga4gh

Apache-2.0 23 ★ 28 41 7 Updated on May 9

Projects 1 Settings

Customize pinned repositories

specification

GA4GH Beacon specification.

28 23

Type: All ▾ Language: All ▾ New

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- JavaScript
- Java
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Most used topics

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- ga4gh

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Sabela de la Torre Pernas

Authenticate

elair

github.com/ga4gh-beacon/



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