



# 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



2017

**30,000** patients will have their genome sequenced for rare-disease diagnosis

**70,000** genomes (patients + relatives) will be sequenced to help rare disease diagnoses

Rare diseases

2030 2017

2030 2017

2030\*

**36,223,000** rare disease patients will have their genome sequenced

**83,000,000** genomes will be sequenced for rare disease diagnosis

**23,000** cancer patients will have their genome sequenced

**50,000** genomes will be sequenced for cancer diagnosis

Cancer

2030 2017

2030 2017

2030 2017

2030 2017

**123,768,000** cancer patients will have their genome sequenced

**248,000,000** genomes will be sequenced for cancer diagnosis

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



# The vision: Federation of data





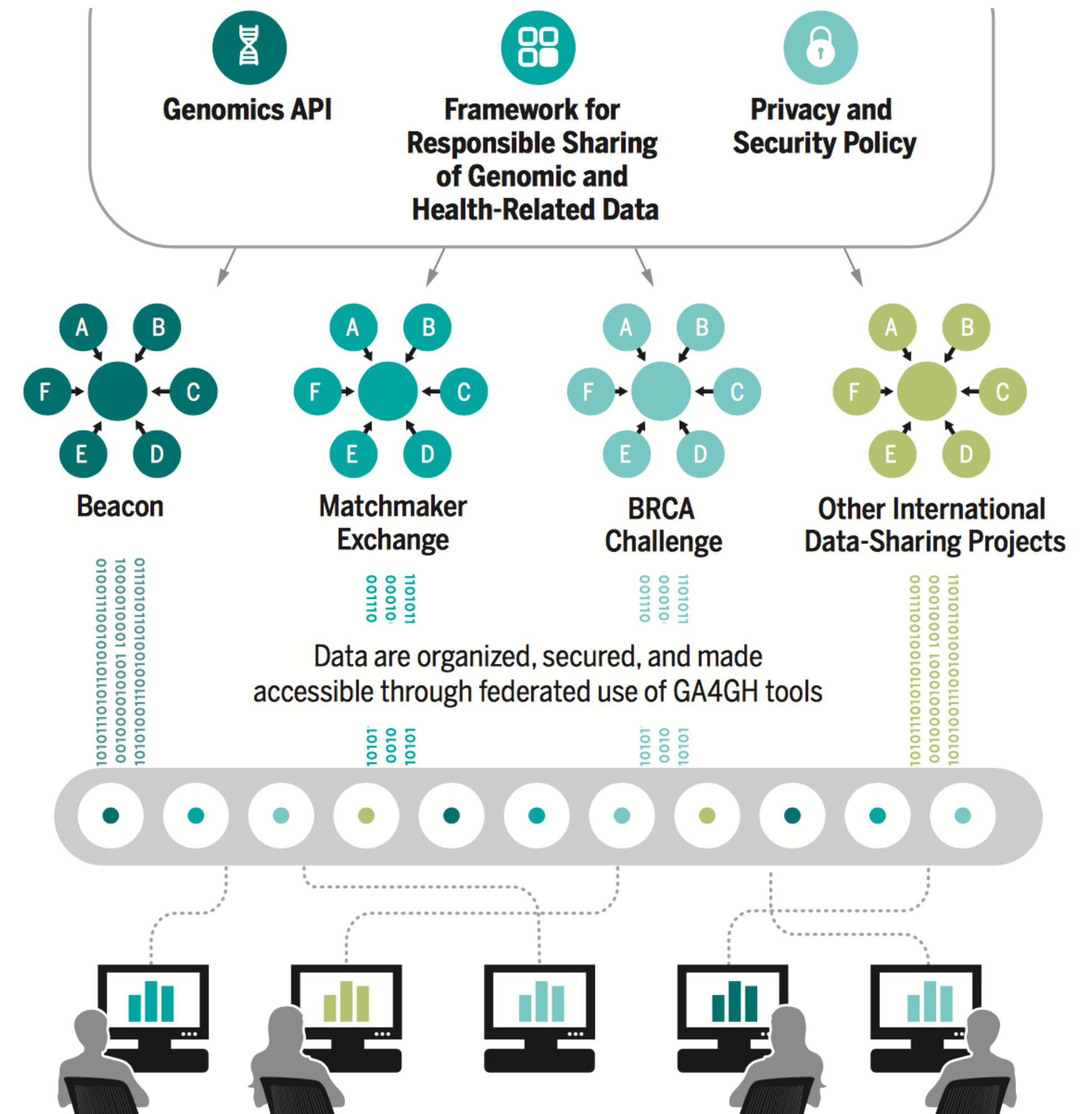


## GENOMICS

# *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.





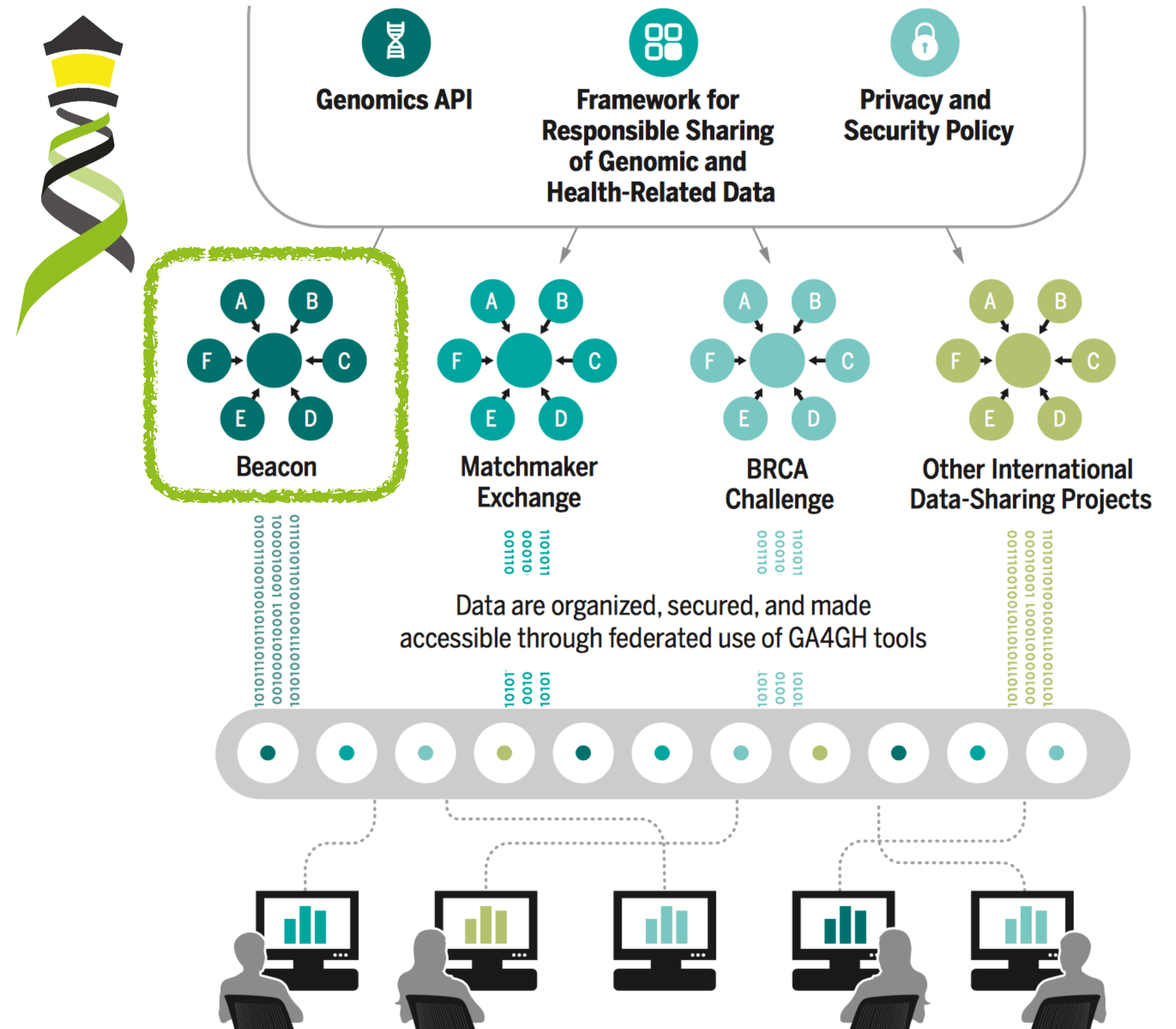


## GENOMICS

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DNASTACK



Global Alliance  
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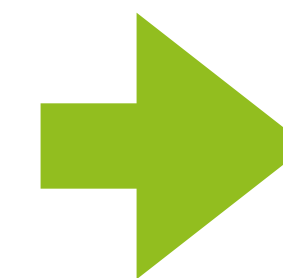
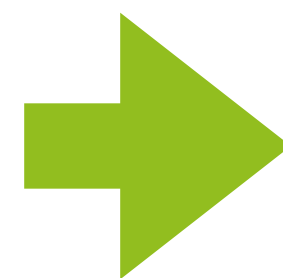




A **Beacon** answers a query for a specific genome variant against individual or aggregate genome collections

**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  
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Here: The variant has  
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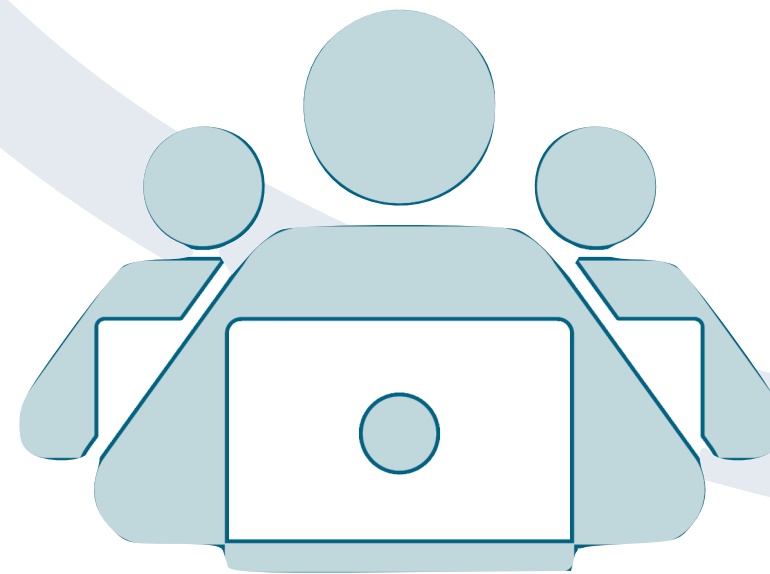
# GA4GH & ELIXIR - Partnering for Standards and Tools in Genomics and Health

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**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

## v1.1 and roadmap

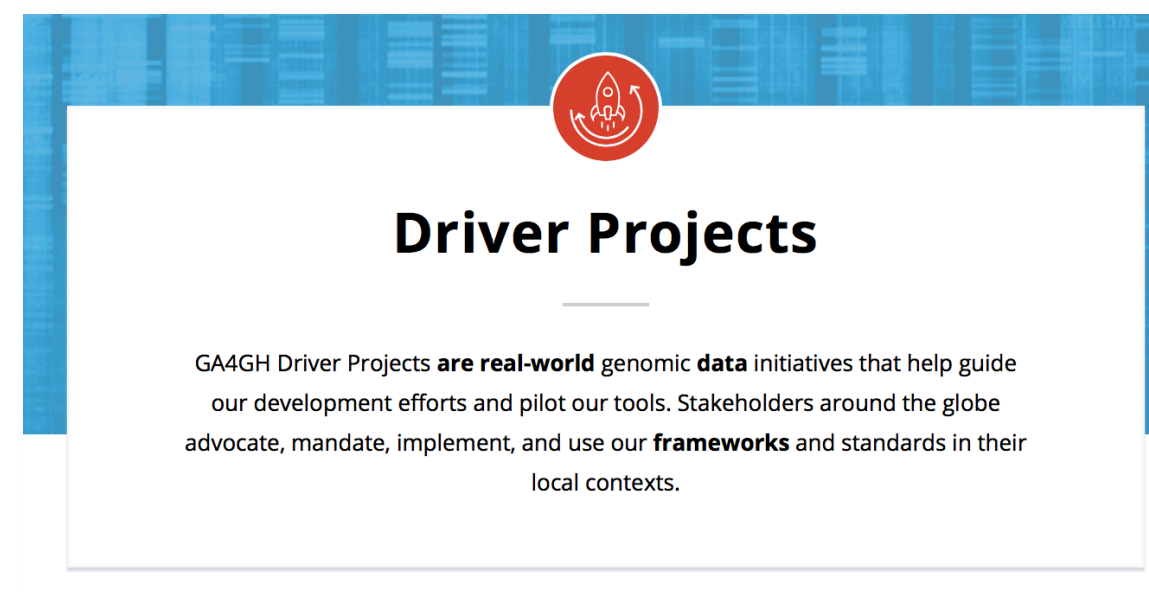


- ☒ **structural variations** (DUP, DEL) in addition to SNV
- ☒ ... more structural queries (translocations/fusions...)
- ☒ Beacon queries as entry for **data handover** (outside Beacon protocol)
- ☒ layered authentication system using **ELIXIR AAI**

**v2** **filters** for phenotypic & technical metadata

**v2** Extended quantitative responses

- ☐ Ubiquitous **deployment** (e.g. throughout ELIXIR network)



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

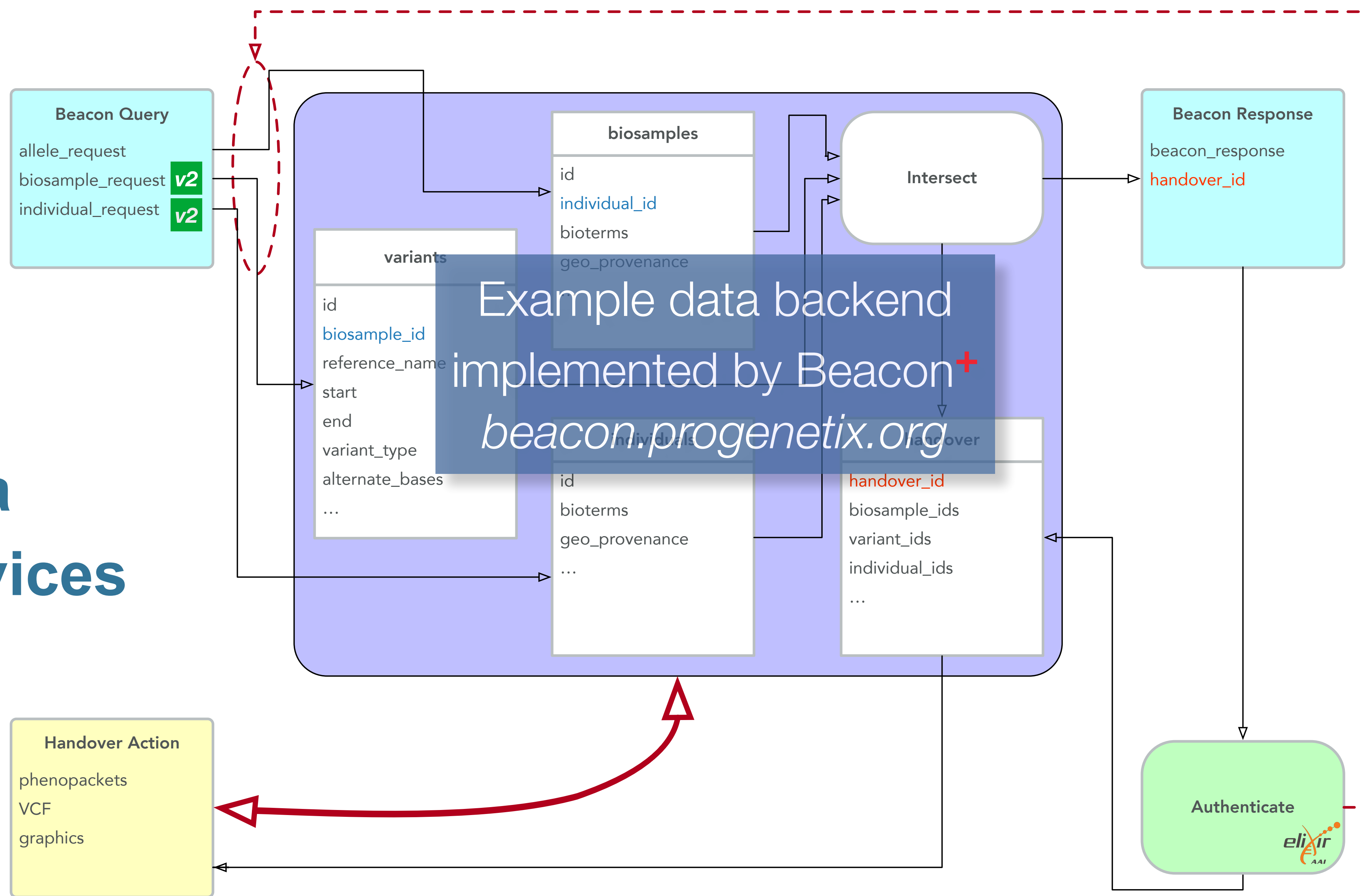
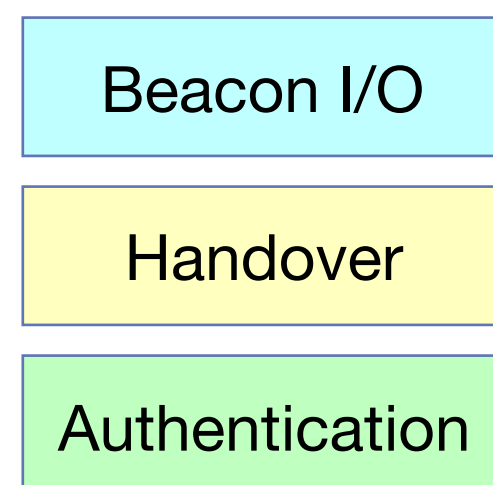
Europe

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





# Beacons v1.1 supports **data delivery services**





# ELIXIR Beacons

EMBL-EBI 

*elixir*  
FINLAND

*elixir*  
SWEDEN

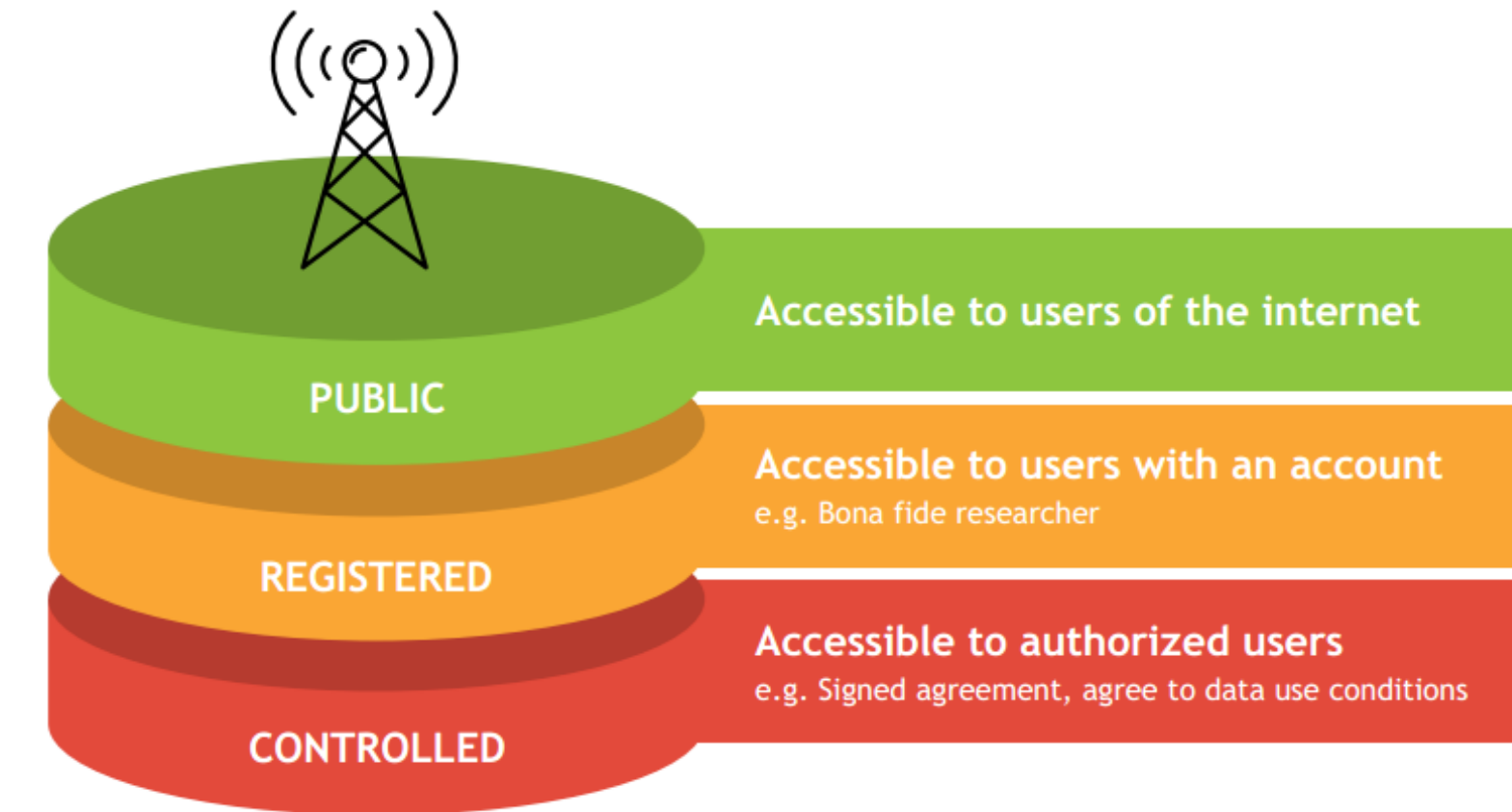
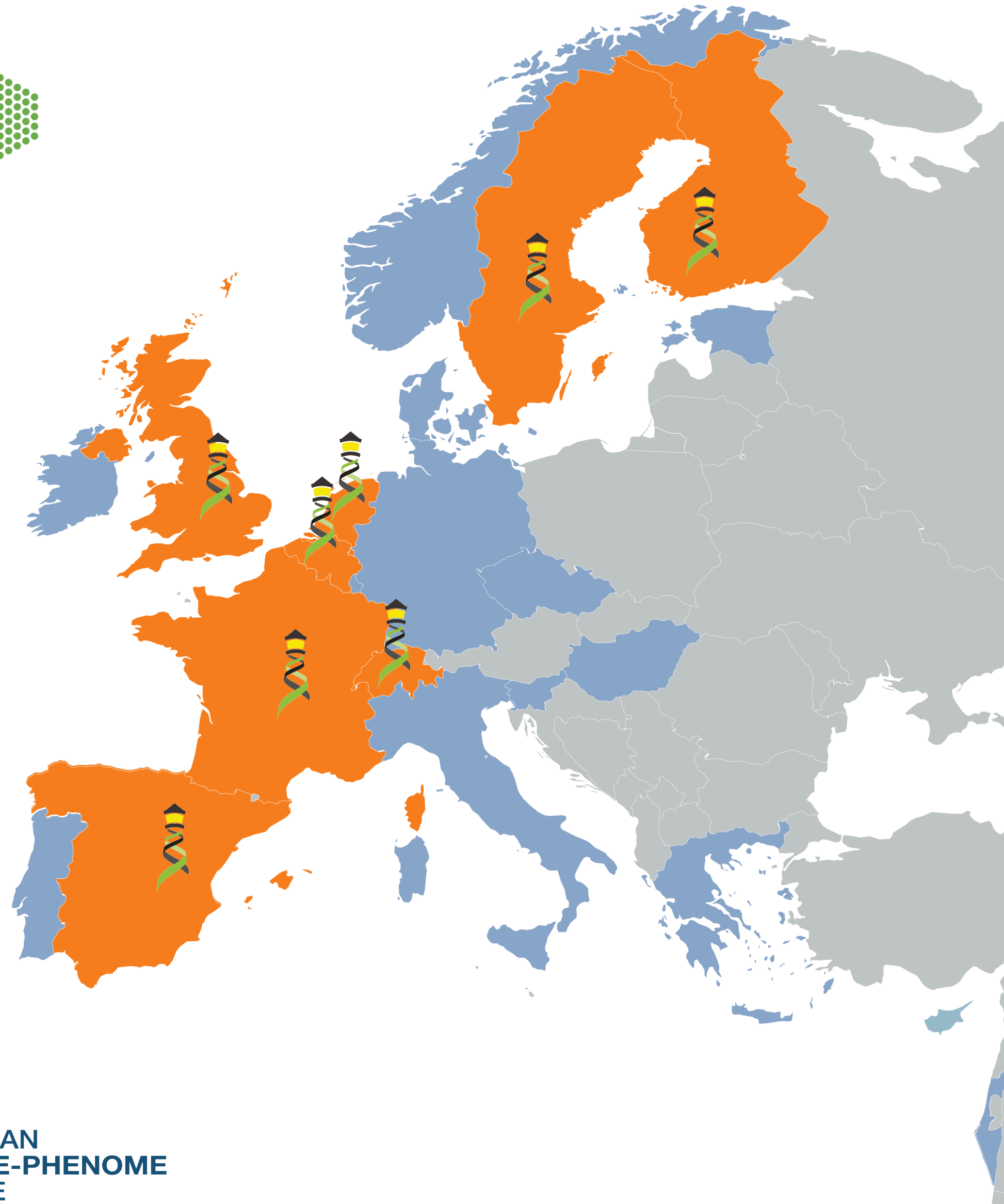
*elixir*  
BELGIUM

*elixir*  
NETHERLANDS

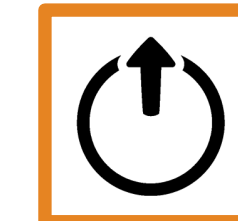
*elixir*  
SWITZERLAND

*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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for Genomics & Health  
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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](#)

**National Bioinformatics Infrastructure Sweden**  
SweFreq Beacon  
Beacon API Web Server based on the GA4GH Beacon API  
[Visit Us](#) · [Beacon API](#) · [Contact Us](#)

**CSC - IT Center for Science**  
Development 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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**Research Programme on Biomedical Informatics**  
DisGeNET Beacon  
Variant-Disease associations collected from curated resources and the literature  
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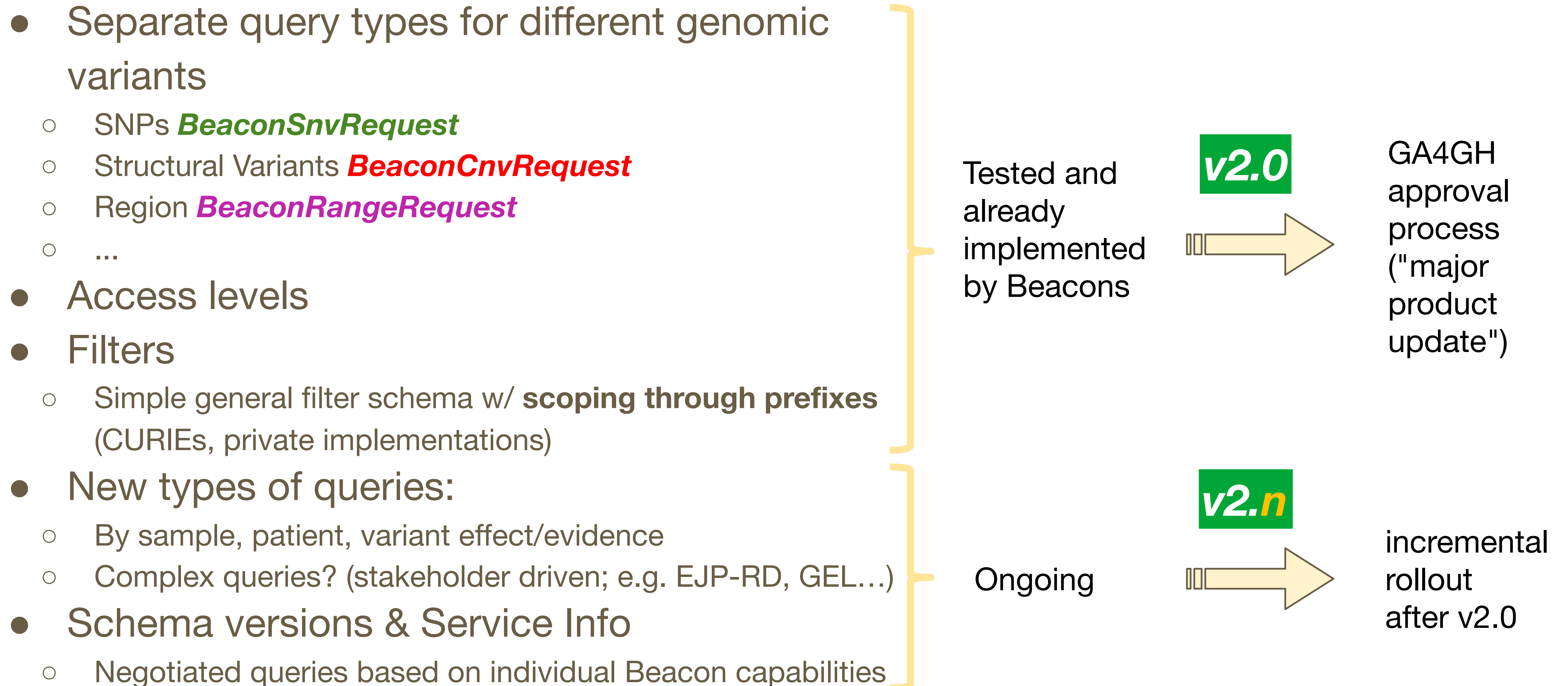
**European Genome-Phenome Archive (EGA)**  
EGA Beacon  
This [Beacon](https://beacon-project.io/) is based on the GA4GH Beacon [v1.1.0](https://github.com/ga4gh-beacon/specification/blob/develop/beacon.yaml)  
[Visit Us](#) · [Beacon API](#) · [Contact Us](#)

**University of Tartu Institute of Genomics, Estonia**  
Beacon at the University of Tartu, Estonia  
Beacon API Web Server based on the GA4GH Beacon API  
[Visit Us](#) · [Beacon API](#) · [Contact Us](#)

**CSC - IT Center for Science**  
Production Beacon  
Beacon API Web Server based on the GA4GH Beacon API  
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# Beacon v2 - Areas of Change







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.

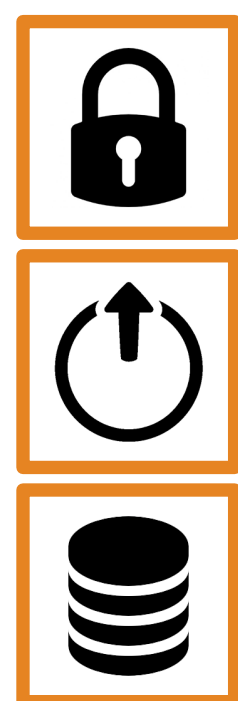
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Have you seen deletions in this region on chromosome 9 in Glioblastomas from a juvenile patient, in a dataset with unrestricted access?



## Beacon v2 API

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





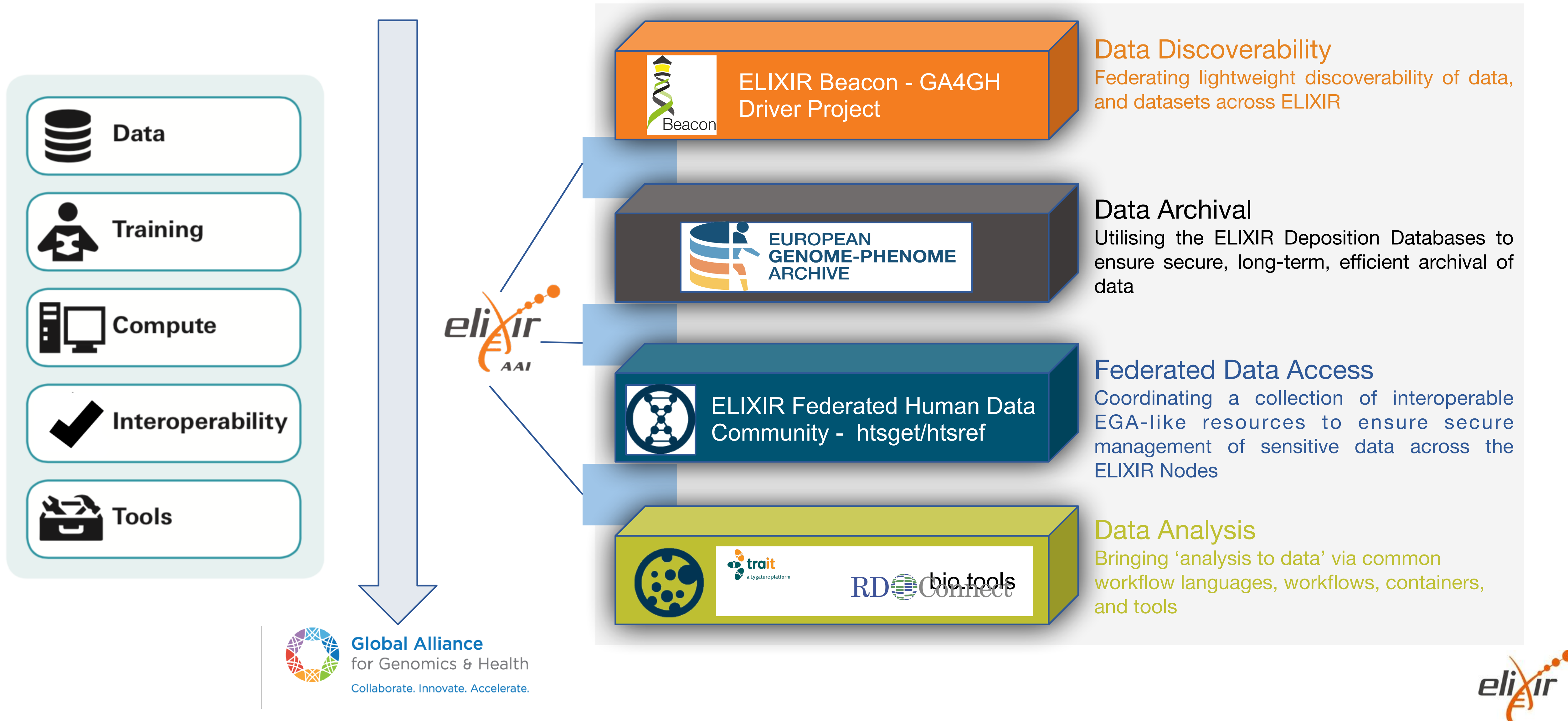
# Beacon Project - Partner Engagement & Next Steps

- Working with **partner communities & projects** on *deploying* Beacons
  - ELIXIR hCNV Community
  - **European Joint Program on Rare Diseases**
  - clinical groups & data initiatives (e.g. Andalusia, 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






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

About...

News & Press

Contributors

Events

Examples, Guides & FAQ

Specification

Roadmap

Beacon Networks

Meeting Minutes

Contacts

Related Sites

Beacon @ ELIXIR

GA4GH

Beacon+

beacon-network.org

GA4GH::SchemaBlocks

GA4GH::Discovery

GA4GH::CLP

GA4GH::GKS

Github Projects

ELIXIR Beacon

SchemaBlocks

Tags

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FAQ

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developers

development

minutes

network

press

proposal




queries

releases

specification

versions

website



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 extensions that 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-sequence data.

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

EvidenceBeacon Notes - GA4GHconnect 2019

The topic of "EvidenceBeacon" was discussed with many different attendees at the GA4GHconnect 2019 session and beyond, leading to some clearer picture about the (widening) scope of the project.

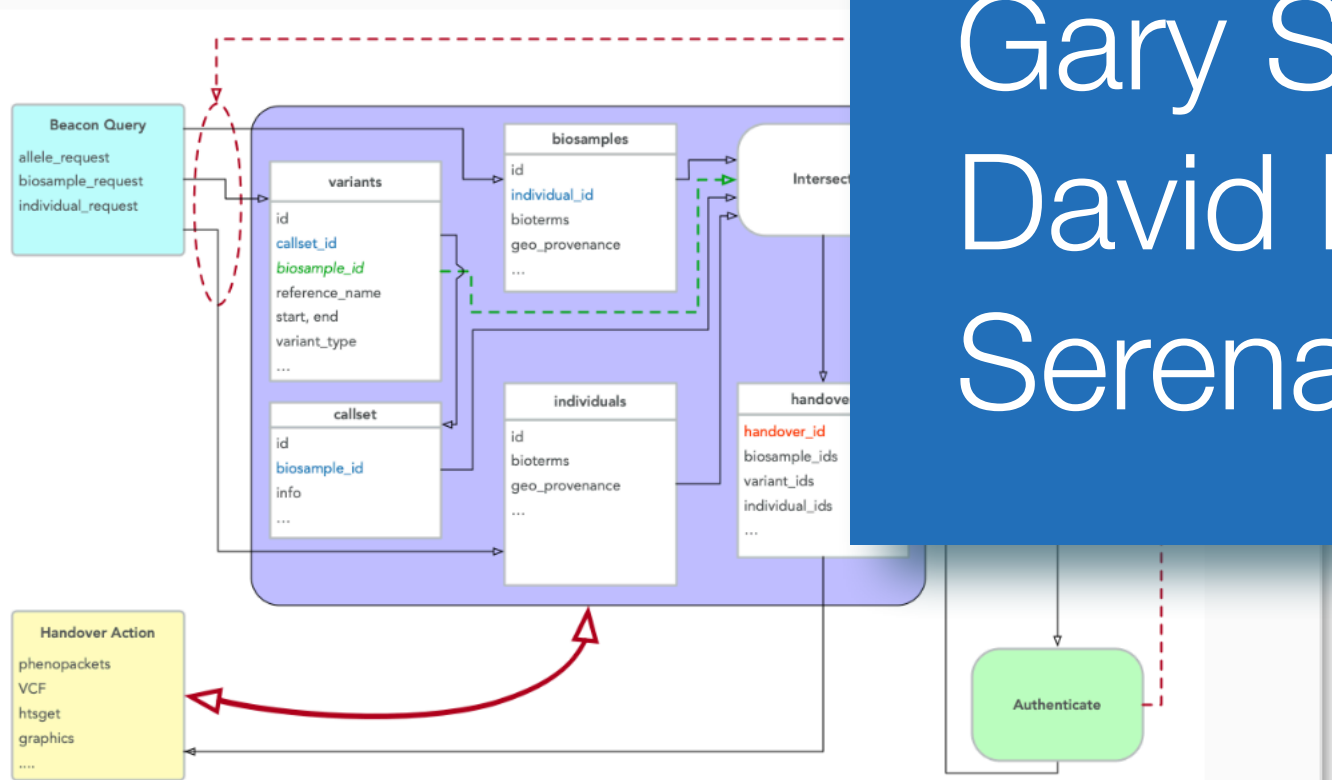
@mbaudis 2019-04-30: more ...


[H→O] Beacon Handover for Data Delivery

While the Beacon response should be restricted to aggregate data (yes, the use of the protocol could be greatly expanded by providing an access to individual data), the use of the protocol could be greatly expanded by providing an access to individual data matched by a Beacon query.

As part of the mid-term product strategy, the ELIXIR Beacon team is evolving the protocol, in which rich data content (e.g. variant data, phenotypic information, etc.) can be provided from linked services, initiated through a Beacon query. 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 work.





Beacon

Beacon Project, Global Alliance for Genomics & Health.

2

Projects 1

Settings

Customize pinned repositories

specification

GA4GH Beacon specification.

28

23

Type: All

Language: All

New

Top languages

JavaScript

Java

HTML

PLpgSQL

Most used topics

beacon

ga4gh

Manage

Updated 9 days ago

openapi

beacon

ga4gh

Apache-2.0

23

28

41

7

Updated on May 9

API Leads  
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Anthony Brooks  
Juha Törnroos  
Discovery WS  
Michael Baudis (Beacon)  
Marc Fiume (Networks)  
ELIXIR  
Gary Saunders  
David Lloyd  
Serena Scollen

Sabela de la Torre Pernas



