



Beacon v2 - Towards Biomedical Beacons

Developments Beyond Basic Variant Discovery



Global Alliance
for Genomics & Health

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 the 'Driver Projects' section of the GA4GH website. It features a red circular icon with a white rocket ship. Below it, the text 'Driver Projects' is displayed. To the right, there is a detailed entry for the 'ELIXIR Beacon' project. The entry includes the ELIXIR logo, a brief description of the project's role in real-world genomic data initiatives, and contact information for the 'Champions': Serena Scollen, Ilkka Lappalainen, and Michael Baudis. The URL www.elixir-europe.org is also provided.

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**
 - **filters** for phenotypic & technical metadata
 - Extended quantitative responses
 - Ubiquitous **deployment** (e.g. throughout ELIXIR network)

Beacon Project - Successes, Challenges, Changes

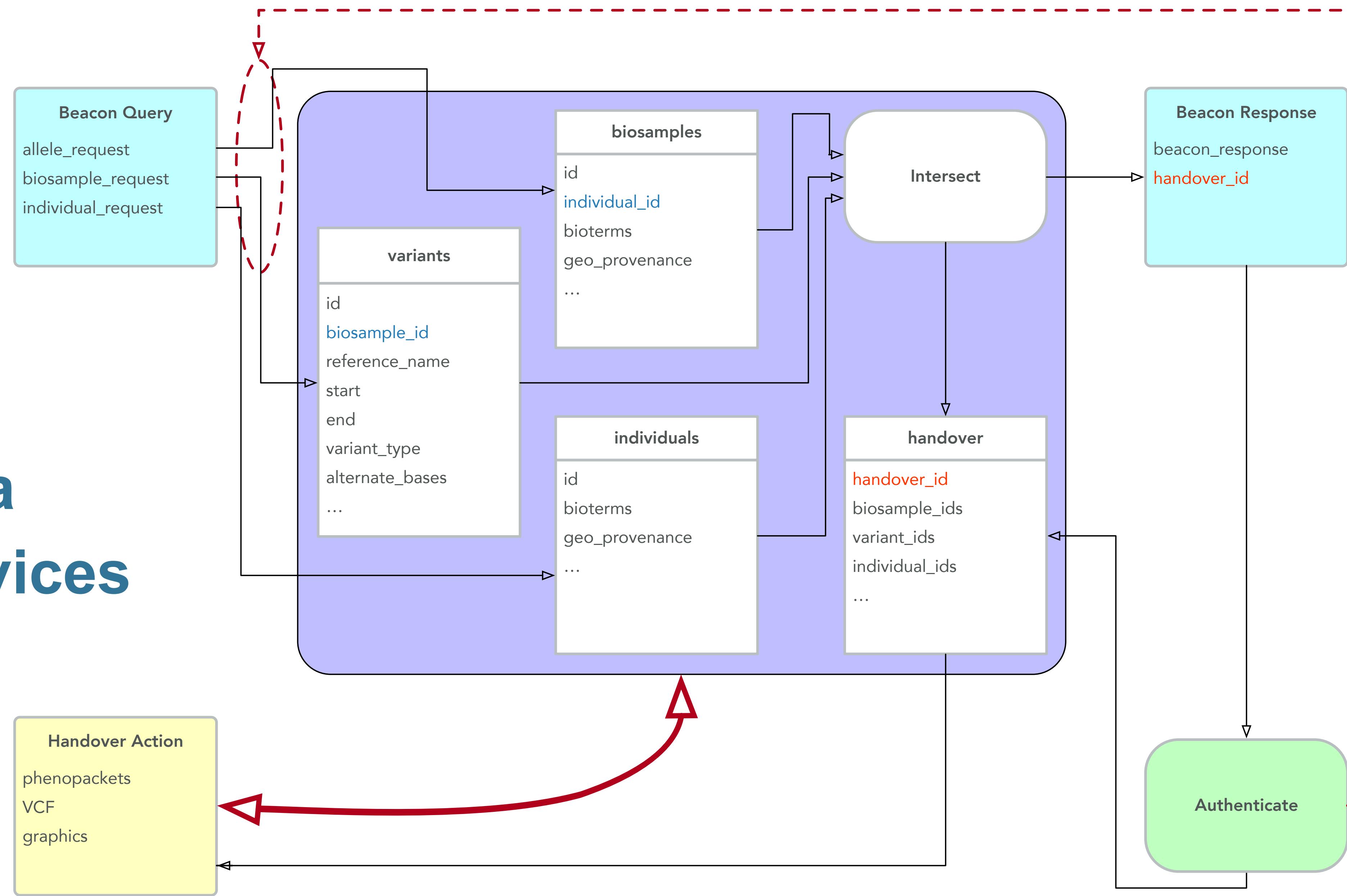
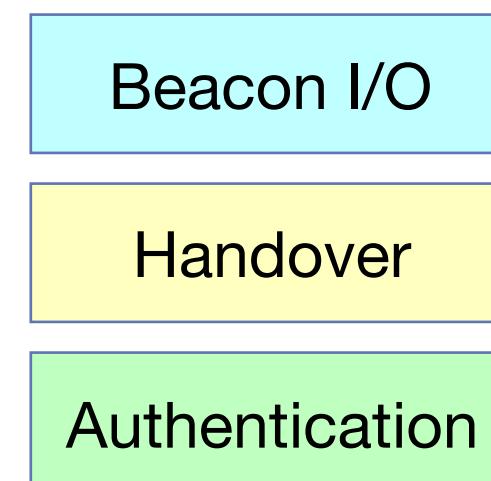


- Beacon has been recognized as the *de facto* single forward looking **GA4GH genomics API** (GA4GH plenary, Boston Oct 2019)
- Beacon adoption and recognition are "ubiquitous", but uneven
 - ▶ the majority of Beacon implementations are on old, pre-v1 versions (i.e. only supporting basic SNV queries)
 - ▶ many of the powerful features of v1.n are not widely known/understood (e.g. clinical or variant data delivery through **handover** protocols)
 - ▶ network integration is difficult due to insufficient data about endpoint capabilities
 - ▶ problematic interpretation of variant annotations (VCF ..)
- Many requests for Beacons supporting "evidence" query and delivery



Beacon & Handover

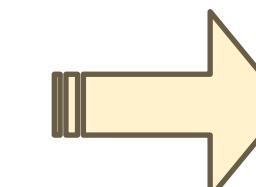
Beacons v1.1
supports data
delivery services



Beacon v2.n - Areas of Change

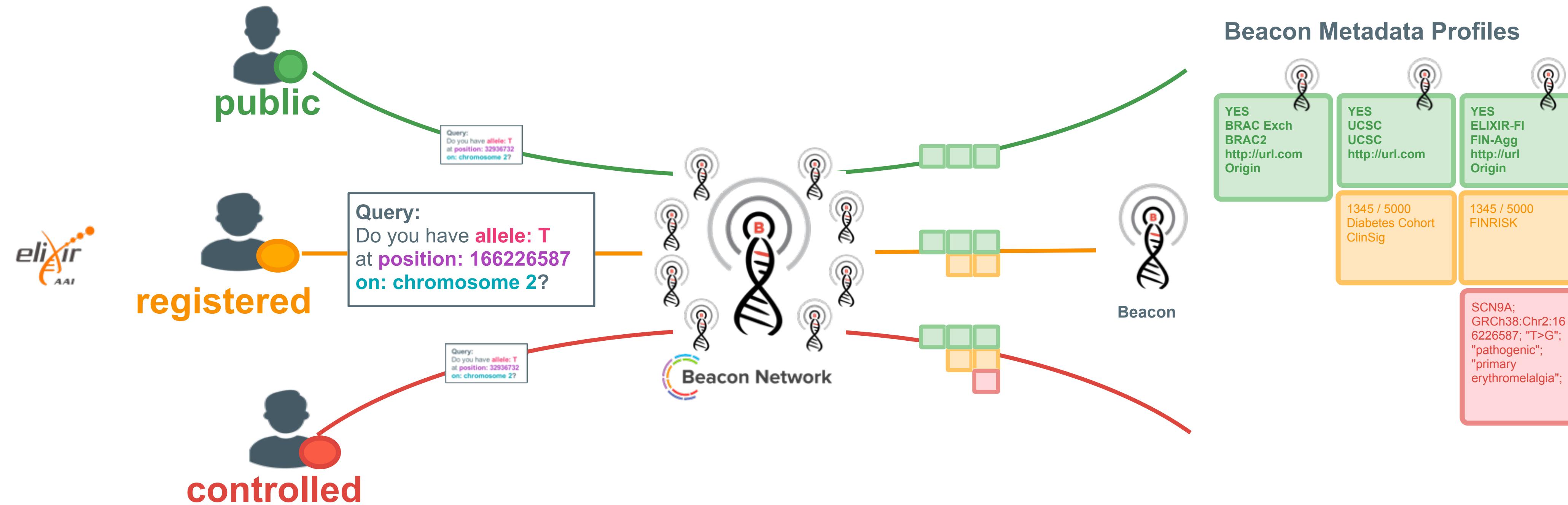
- Separate query types for different genomic variants
 - SNPs **BeaconSnpRequest**
 - Structural Variants **BeaconCnvRequest**
 - Region **BeaconRangeRequest**
 - ...
- Access levels
- Filters
 - Simple general filter schema w/ scoping through prefixes (CURIEs, private implementations)
- New types of queries:
 - By sample, patient, variant effect/evidence
 - Complex queries? (stakeholder driven; e.g. EJP-RD, GEL; Tony Brooks leading effort)
- Schema versions & Service Info
 - Negotiated queries based on individual Beacon capabilities

v2.0
Tested and already implemented by real Beacons



Ongoing → v2.n

Beacon v1 => v2: Integrating permissions and discovery



<https://www.youtube.com/watch?v=LyfmvAs7LtQ&feature=youtu.be>



Beacon Project - Partner Engagement & Next Steps



- Working with partner communities & projects on deploying Beacons
 - ▶ ELIXIR Human Copy Number Variation Community
 - ▶ clinical groups (e.g. Andalucia, Cancer Core Europe)
 - ▶ variant annotation resources, with possible clinical components (e.g. SVIP-O)
- Improving reference implementation and standards / compliance testing
- Beacon v2 code testing
- aligning w/ GA4GH standards, through "request & adopt"



ELIXIR h-CNV Community

- 1st community meeting September 2018 - Hinxton
- h-CNV community approved in February 2019
- Implementation study accepted: start 2019-06-01
- community workshop ELIXIR All-Hands Lisbon
- chaired by Christophe Béroud and David Salgado (INSERM Marseille)

Node	Name of PI
ELIXIR-FR	Christophe Béroud, David Salgado, Marc Hanauer, Victoria Dominguez
ELIXIR-CH	Michael Baudis
ELIXIR-DE	Jan Korbel
EMBL-EBI	Thomas Keane, Fiona Cunningham
ELIXIR-ES	Joaquin Dopazo, Alfonso Valencia, Salvador Capella, Sergi Beltran, Steven Laurie, Gemma Bullich, Laura I. Furlong, Janet Piñero
ELIXIR Hub	John Hancock, Gary Saunders, Kathi Lauer, Leyla Garcia
ELIXIR-NL	Bauke Ylstra, Daoud Sie, Leon Mei, Morris Swertz (UMCG), Lennart Johansson
ELIXIR-NO	Eivind Hovig, Pubudu Samarakoon
ELIXIR-HU	Attila Gyenessei ,Katalin Monostory
ELIXIR-SI	Brane Leskošek, Polonca Ferk, Marko Vidak
ELIXIR-UK	Krzysztof Poterlowicz



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Human Copy Number Variation

Human Copy Number Variation

Most human cells divide by mitosis, where one parent cell divides to produce two daughter cells. Just before a cell divides it makes a copy of its own DNA, but this copy may not have the same gene sequence as the parent DNA. One gene, for example, may be copied twice into the new DNA, or not copied at all. This phenomenon is called **Copy Number Variation (CNV)**.

It is thought that these Copy Number Variations are vital for evolution, but they also play important roles in disease. Despite the fact that Copy Number Variations are the most prevalent genetic mutation type, identifying and interpreting them is still a major challenge. The ELIXIR human Copy Number Variation (hCNV) Community aim to implement processes to make the detection, annotation and interpretation of these variations easier.



COPY NUMBER VARIATION - DATA COLLECTION & ANALYSIS
Göteborg, Sweden
 Friday 14th June | satellite to ESHG





{B}{H}
BIOHACKATHON-EUROPE
PARIS 2019 November 18th - 22th
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GA4GH {S}[B] SchemaBlocks

- “cross-workstreams, cross-drivers” initiative to document GA4GH object standards and prototypes, data formats and semantics
- launched in December 2018
- documentation and implementation examples provided by GA4GH members
- no attempt to develop a rigid, complete data schema
- object vocabulary and semantics for a large range of developments
- currently not “authoritative GA4GH recommendations”



GA4GH :: SchemaBlocks

An Initiative by Members of the Global Alliance for Genomics and Health

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Data Schemas

Examples, Guides & FAQ

Meeting minutes

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[ELIXIR Beacon](#)

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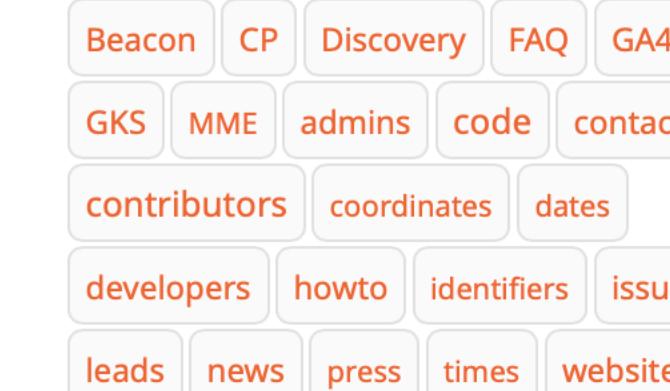
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schemablocks.org

GA4GH Data Model

Recommendation (DRAFT)

The GA4GH data model recommends the use of a default object hierarchy in standard and product design processes. While it reflects concepts from the original [GA4GH schema](#), it provides mostly a structural guideline for API and data store design, but is not thought to provide a set of absolute implementation requirements.

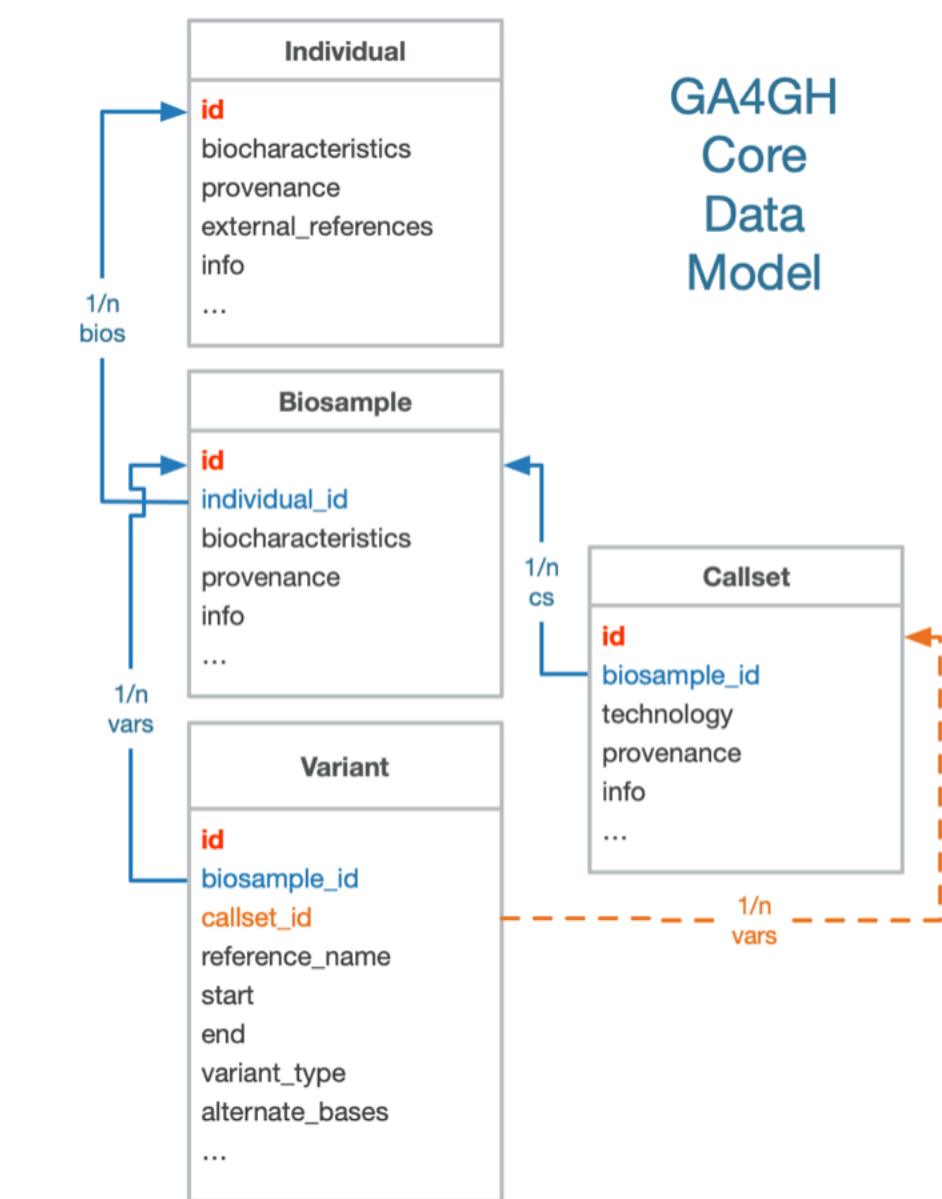
Contributors

- [@mcourtot](#)
- [@mbaudis](#)

Summary

The GA4GH data model for genomics recommends the use of a principle object hierarchy, consisting of

- **variant**
 - a single molecular observation, e.g. a genomic variant observed in the analysis of the DNA from a biosample
- **callset**
 - the entirety of all variants, observed in a single experiment on a single sample
 - a **callset** can be compared to a data column in a **VCF** variant annotation file
 - **callset** has an optional position in the object hierarchy, since **variants** describe biological observations in a biosample
- **biosample**
 - a reference to a physical biological specimen on which analyses are performed
- **individual**
 - in a typical use a human subject from which the biosample(s) was/were extracted



A graph showing recommended basic objects and their relationships. The names and attributes are examples and may diverge in count and specific wording (e.g. "subject" instead of "individual") in specific implementations.

These basic definitions will be detailed further on.

Additional concepts (e.g. *dataset*, *study* ...) may be added in the future.





ELIXIR Genome Beacons

A Driver Project of the Global Alliance for Genomics and Health

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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 distinct **flavours**, which 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 related data types.

@mbaudis 2018-10-18: [more ...](#)

EvidenceBeacon Notes - GA4GHconnect 2019

The topic of "EvidenceBeacon" was discussed with many different attendants during the speed dating session and beyond, leading to some clearer picture about the (widening) extent & next steps.

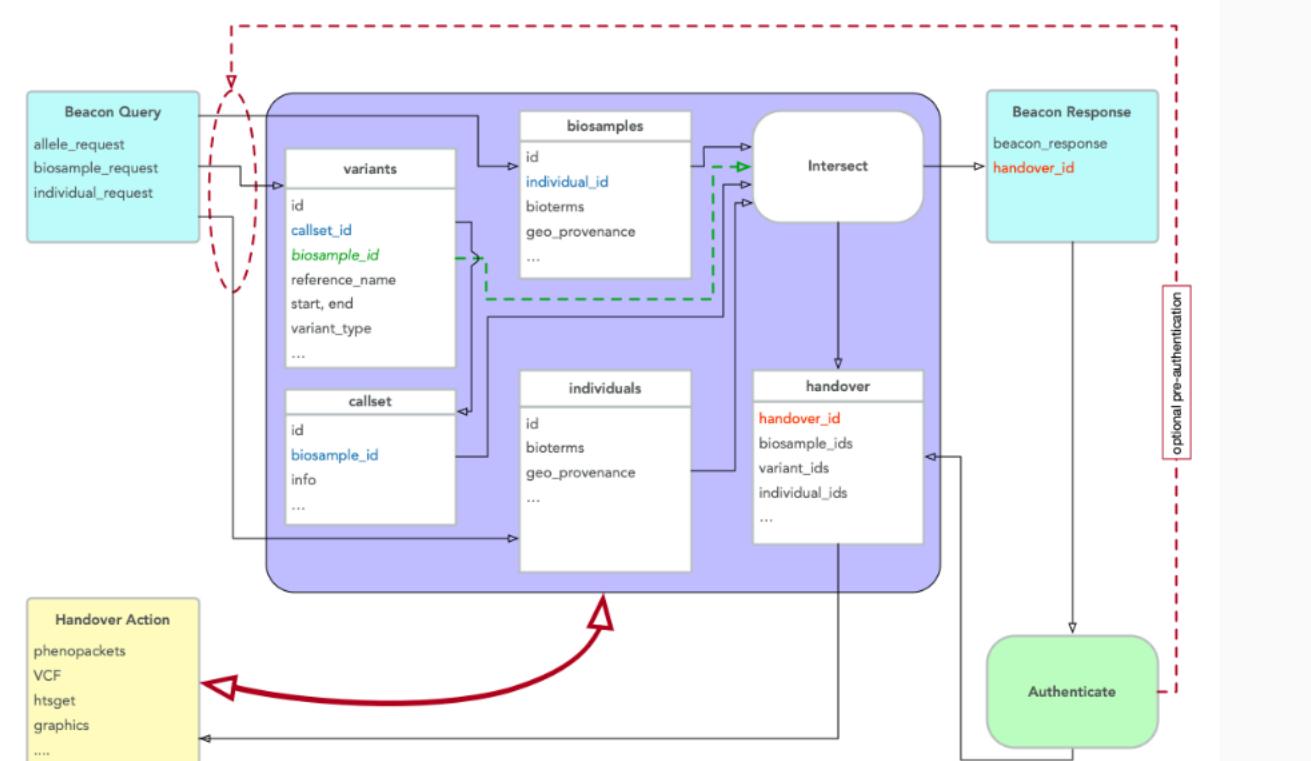
@mbaudis 2019-04-30: [more ...](#)

[H→O] Beacon Handover for Data Delivery

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

As part of the mid-term product strategy, the ELIXIR Beacon team is evaluating the use of a "handover" protocol, in which rich data content (e.g. variant data, phenotypic information, low-level sequencing results) can be provided from linked services, initiated through a Beacon query (and possibly additional steps like protocol selection, authentication...). A discussion of the topic can e.g. be found in the Beacon developer area on Github (issue #114).

As of 2018-11-13, the **handover** concept has become part of the [ongoing code development](#).



beacon-project.io



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Beacon Project, Global Alliance for Genomics & Health.

<http://beacon-project.io>

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GA4GH Beacon specification.

★ 28 ⚡ 23

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Elixir Beacon Reference Implementation

Java ⚡ 4 ★ 9 ⚡ 3 ⚡ 0 Updated 21 hours ago



Top languages

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PLpgSQL

ga4gh-beacon.github.io

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website beacon ga4gh

HTML Apache-2.0 ⚡ 2 ★ 3 ⚡ 15 ⚡ 1 Updated 9 days ago



Most used topics

beacon ga4gh

Manage

specification

GA4GH Beacon specification.

openapi beacon ga4gh

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



People



github.com/ga4gh-beacon/



ELIXIR Beacon Network



Juha Törnroos, ELIXIR-FI

Michael Baudis, ELIXIR-CH

Jordi Rambla, ELIXIR-ES

ELIXIR Programme TC

28/11/2019

www.elixir-europe.org

ELIXIR Beacon Network service

- One of the main objectives of ELIXIR Beacon 2019-2021 implementation study is build ELIXIR Beacon Network and establish a basis to operate that as a service. ELIXIR Beacon is also GA4GH driver project.
- We started the work by creating a specification for ELIXIR Beacon Network and since then this specification has been aligned with GA4GH Discovery work stream. The Network specification was divided into three separate specifications: GA4GH Service Info, GA4GH Service Registry and Beacon Aggregator.
- We have implemented the network components (Registry, Aggregator) and the user interface that can distribute and the query across the network and aggregate the results.



ELIXIR Beacon Network service

- The prototype user interface is developed together by the project partners, operated together by CSC and BSC and it is available at <https://dev-ui-beacon.rahtiapp.fi>
- The prototype supports ELIXIR AAI with three access tiers (public, registered and controlled access) and new GA4GH Passport standard.
- It is possible to query controlled access data from the Beacons in the Network (EGA datasets are being discussed)





GRCh38 ▾

Chromosome : Position ReferenceBase > AlternateBase|VariantType

Search

Quickstart: Example of a mitochondrial variant query

Advanced Search

Connected Beacons

About Beacons

How to use Beacon Network



LCSB at University of Luxembourg

ELIXIR.LU Beacon

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University of Tartu Institute of Genomics

GA4GHBeacon at the University of Tartu

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Beacon for testing GA4GH Passports

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European Genome-Phenome Archive (EGA)

EGA Beacon

This <[a href="https://beacon-project.io/">Beacon> is based on the GA4GH Beacon
<\[a href="https://github.com/ga4gh">GA4GH>\]\(https://github.com/ga4gh\)](https://beacon-project.io/)

Why ELIXIR Beacon Network

- We want to ease data discoverability in Europe and in ELIXIR Core Data Resources
- Provide seamless integration into existing services (e.g. handover to EGA) and leverage ELIXIR Infrastructure services, like ELIXIR AAI
- We are working on two parallel tracks:
- Increasing the size by expanding the Network with European Beacons
- Improving the quality by bringing data from EGA to the Network



Operating as a service

- The long-term goal is to operate ELIXIR Beacon Network as a service.
- Our roadmap includes things like:
 - Documented processes (e.g. how to join to the Network)
 - Defined metadata requirements for individual Beacons (e.g. metadata in info endpoints)
 - Defined security requirements (aligned with GA4GH Security WS)
 - Monitoring and helpdesk operations
 - Metrics and KPIs (defined in previous IS)
 - Estimate for operational costs
- We are also planning to move the prototype to <http://beacon-network.elixir-europe.org> domain name

