GA4GH Beacon v2 - Evolving Reference Standard for Genomic Data Exchange

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For the ELIXIR Beacon project, GA4GH Discovery work stream and the international network of Beacon API developers

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For more information please visit beacon-project.io and the code repositories at github.com/ga4gh-beacon

The Beacon Protocol - From the first approved GA4GH standard towards supporting an "Internet of Genomics"

A ***Beacon*** answers a query for a specific genome variant against individual or aggregate genome collections
YES | **NO** | \0

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 support the ***Beacon API***

Here: The variant has been found in **few** resources, and those are from **disease** specific **collections**.

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

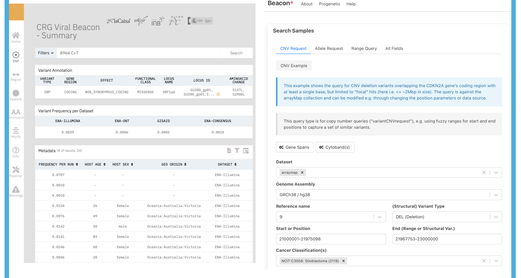
The Beacon API v2 proposal opens the way for the design of a simple but powerful "**genomics API**".

Beacon v2 - Areas of Change and Future Developments

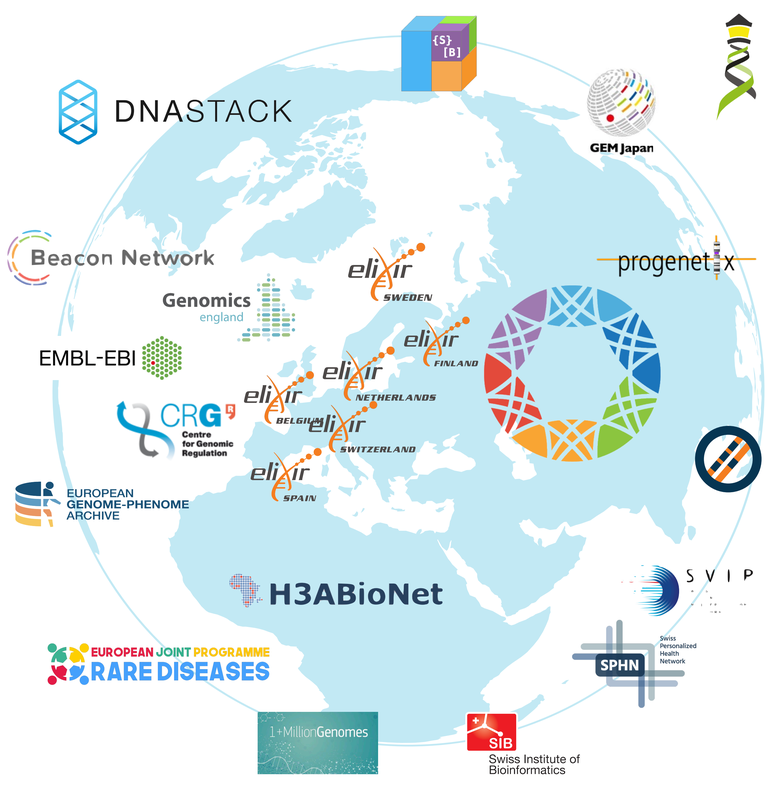
The Beacon driver project was one of the earliest initiatives of the Global Alliance for Genomics and Health with the Beacon v1.0 API as first approved GA4GH standard.

Version 2 of the protocol is slated to provide fundamental changes, towards a *Internet of Genomics* foundational standard:

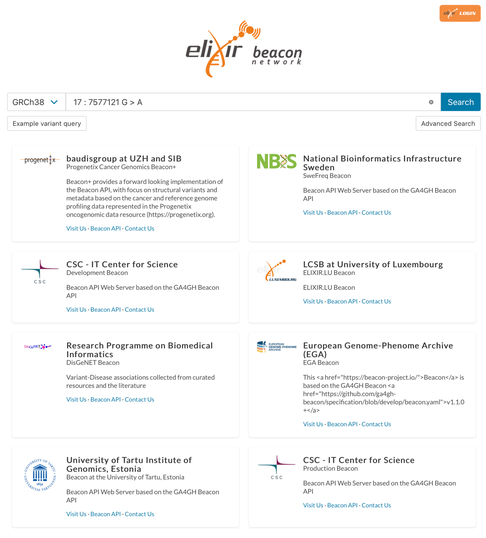
- requests beyond genomic variants ("filters")
- payload responses, secured through open AAI
- aligning w/ GA4GH standards (Phenopackets, VRS, DUO...) through SchemaBlocks {S}[B]
- Working with international partners on deployment of advanced implementations



*Examples of Beacon v2 test implementations: The CRG Viral Beacon provides extensive information about COVID19 variants (left). The Beacon+ server allows query on cancer genomes using disease code **filters** (right).*



Beacon Networks with AAI Support

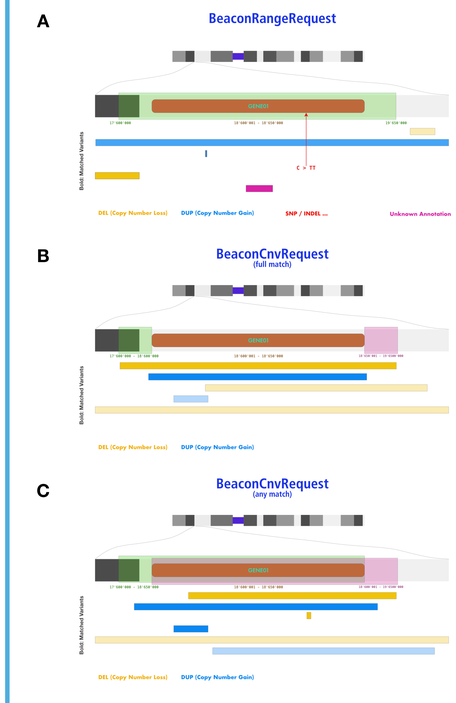


The Beacon protocol draws its power from networking individual resources with federated queries and response aggregation. Current implementation of Beacon networks such as developed by ELIXIR enable the latest versions of Beacon query types, and support open authentication and authorization protocols (AAI), for a layered access control in data sensitive environments.

Service Registry API VALID {-} build passing license Apache 2
GA4GH service-info specification VALID {-} build passing license Apache 2

Networking Beacon resources is supported through GA4GH standards such as the Service Registry and Service Info specifications.

Beacon v2 Typed Variant Requests



The Beacon v2 protocol introduces a growing number of typed queries, to facilitate the detection of diverse or incompletely defined genome variants. Scenarios can include the scanning for any variant in a genomic range (A); the retrieval of copy number variants duplicating a gene's CDR (B) or deletion CNVs affecting a gene in any extent, but limiting this to "focal" events (C).

Michael Baudis 2020-09-28