ELIXIR Beacon Project

Concepts | Status | History | Outlook

Michael Baudis :: ELIXIR-CH:: ELIXIR All Hands :: 2020-06-10

Global Alliance for Genomics & Health

SIB

University of Zurich UZH

Genome screening at the core of “Personalised Health”

- **Genome analyses** (including transcriptome, metagenomics) are core technologies for Personalised Health™ applications
- The unexpectedly large amount of **sequence variants** in human genomes - germline and somatic/cancer - requires huge analysis efforts and creation of **reference repositories**
- **Standardized data formats** and **exchange protocols** are needed to connect these resources throughout the world, for reciprocal, international **data sharing** and **biocuration** efforts
- Our work @ UZH:
  - ***cancer*** genome repositories
  - biocuration
  - protocols & formats

arrayMap

progenetix

elixir

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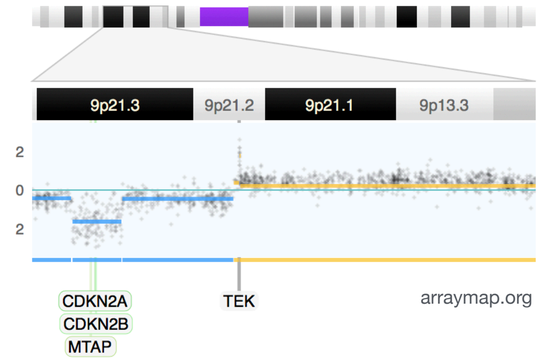
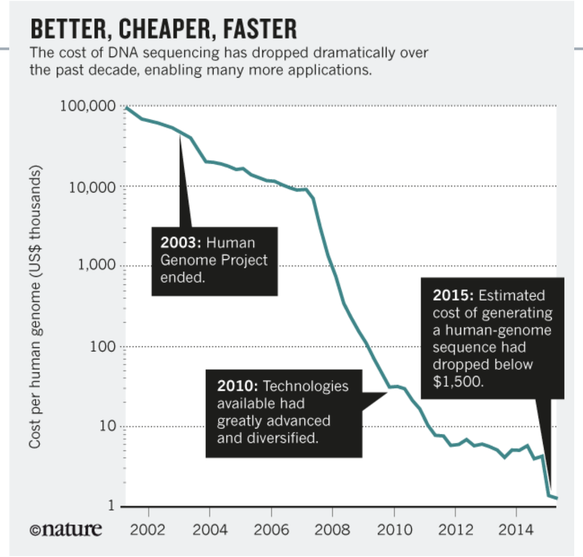
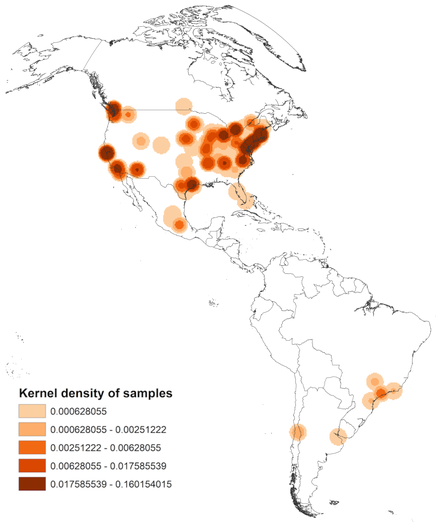
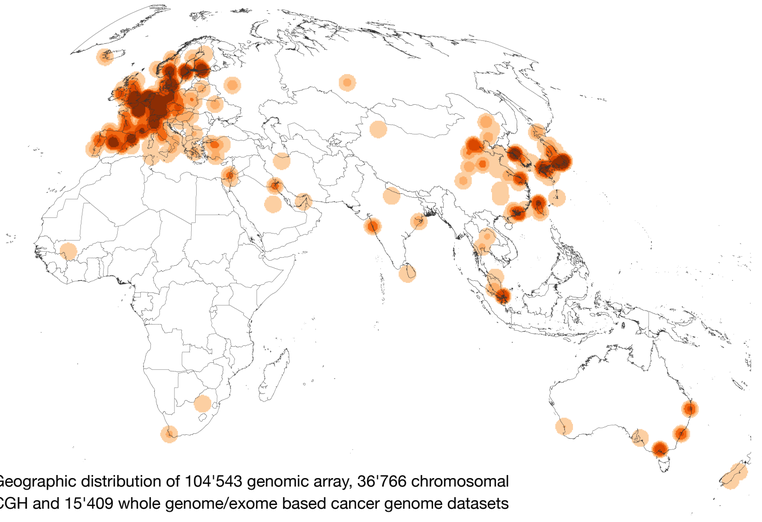




The future of DNA sequencing. Eric D. Green, Edward M. Rubin & Maynard V. Olson. Nature; 11 October 2017 (News & Views)





Geographic distribution of 104'543 genomic array, 36'766 chromosomal CGH and 15'409 whole genome/exome based cancer genome datasets

The vision: Federation of data

GENOMICS

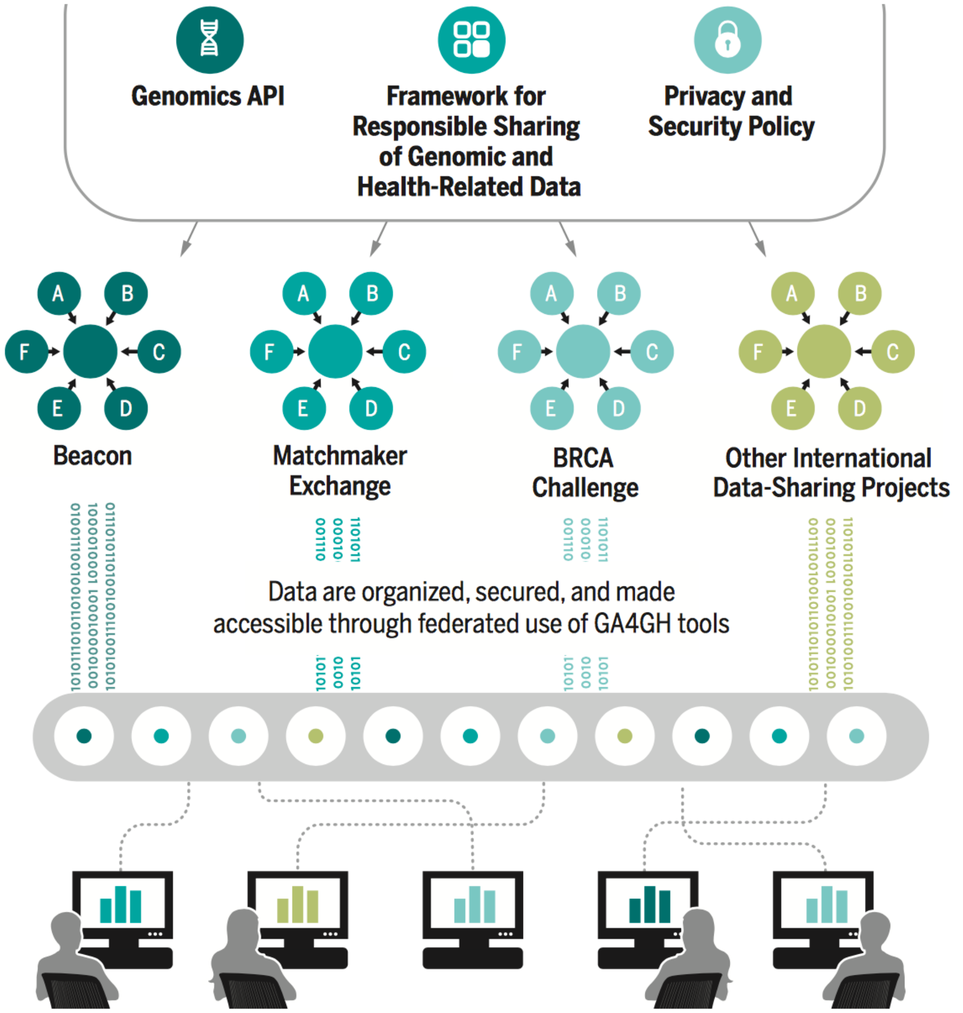
A federated ecosystem for sharing genomic, clinical data

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

The Global Alliance for Genomics and Health*

SCIENCE 10 JUNE 2016 • VOL 352 ISSUE 6291

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



Global Alliance for Genomics & Health

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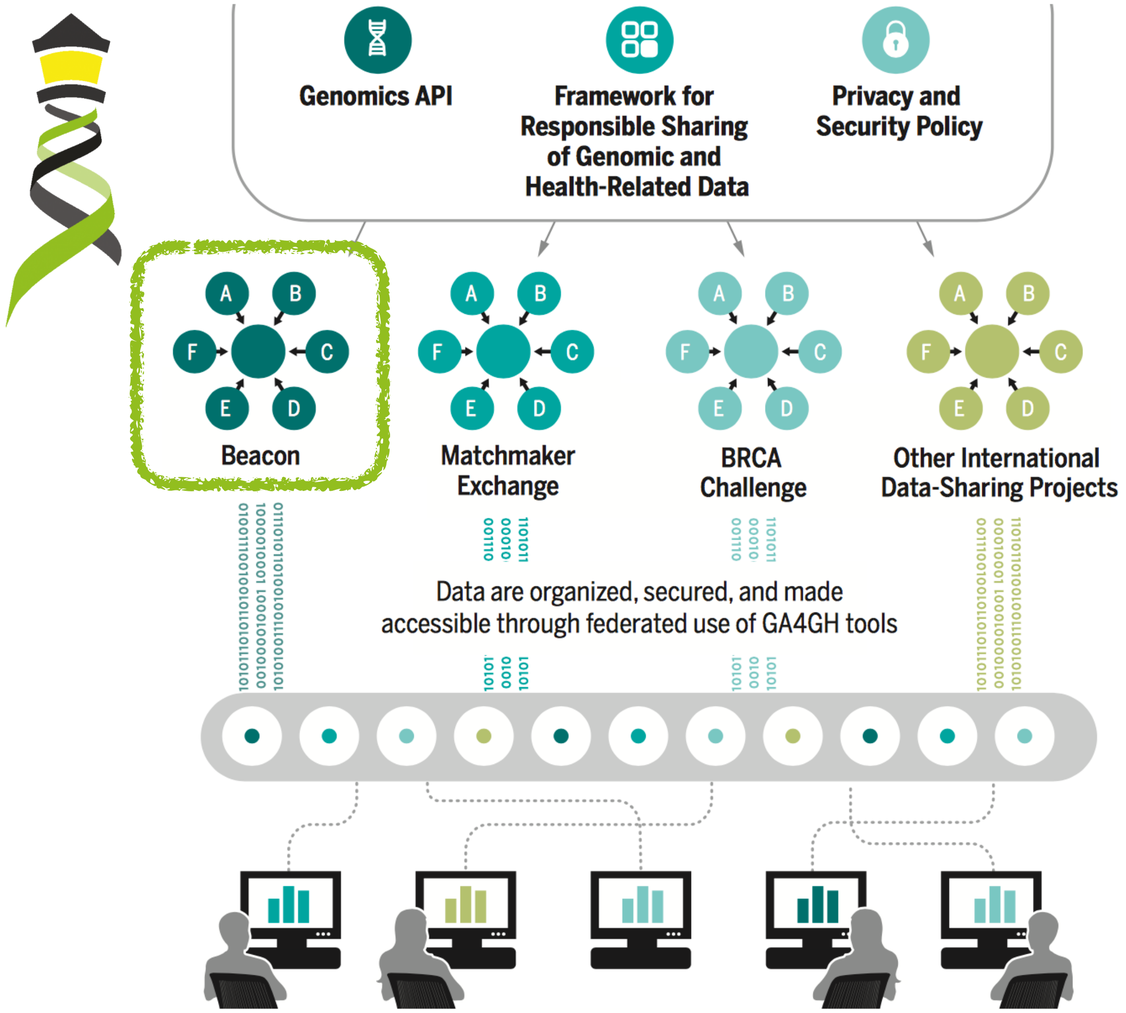


DNASTACK

Global Alliance for Genomics & Health

Global Alliance “Beacon” - Jim Ostell, NCBI, March 7, 2014

Global Alliance for Genomics & Health

Introduction

… I proposed a challenge application for all those wishing to seriously engage in ***international*** data sharing for human genomics. …

1. Provide a public web service
2. Which accepts a query of the form “Do you have any genomes with an “A” at position 100,735 on chromosome 3?”
3. And responds with one of “Yes” or “No” …

“Beacon” because … people have been scanning the universe of human research for ***signs of willing participants in far reaching data sharing***, but … it has remained a dark and quiet place. The hope of this challenge is to 1) ***trigger the issues*** blocking groups … in way that isn’t masked by the … complexities of the science, fully functional interfaces, and real issues of privacy, and to 2) in ***short order*** … see ***real beacons of measurable signal*** … from ***at least some sites*** … Once your “GABeacon” is shining, you can start to take the ***next steps to add functionality*** to it, and ***finding the other groups*** … following their GABeacons.

Utility

Some have argued that this simple example is not “useful” so nobody would build it. Of course it is not the first priority for this application to be scientifically useful. …intended to provide a ***low bar for the first step of real*** … ***engagement***. … there is some utility in …locating a rare allele in your data, … not zero.

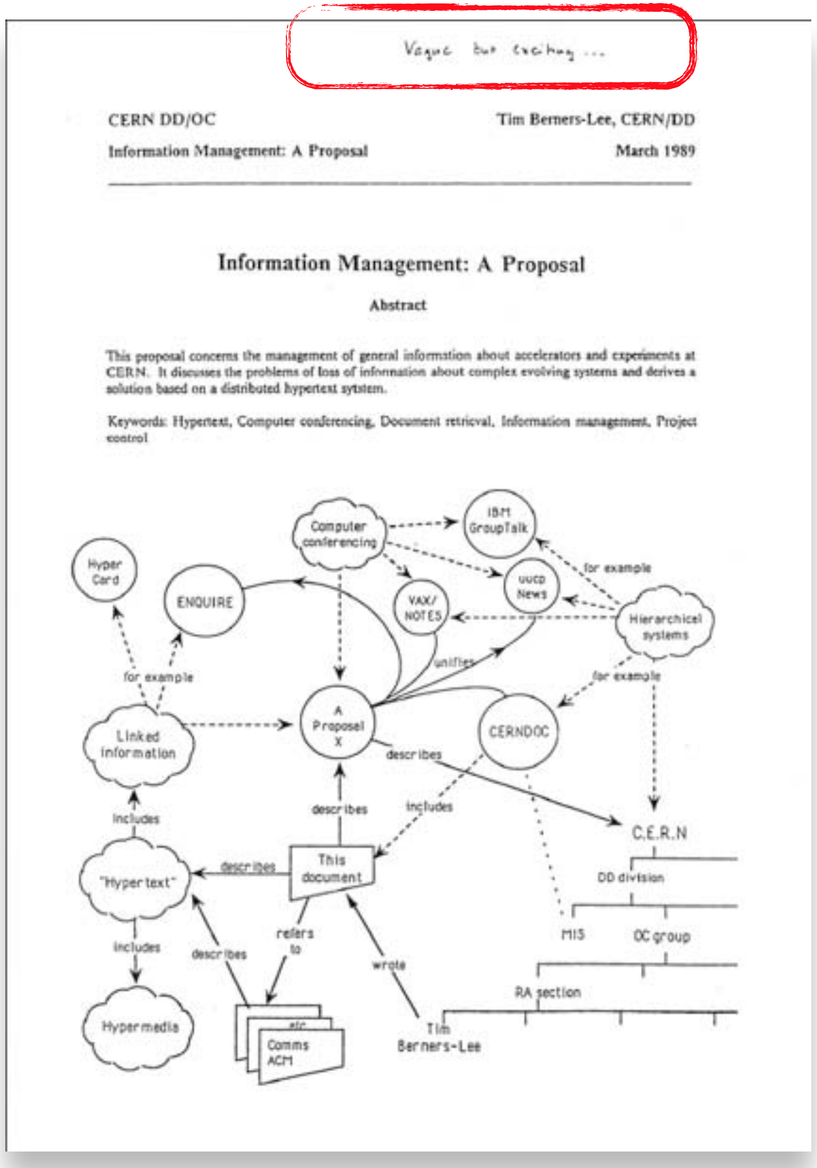
A number of more useful first versions have been suggested.

1. Provide ***frequencies of all alleles*** at that point
2. Ask for all alleles seen in a gene ***region*** (and more elaborate versions of this)
3. Other more complicated queries

"I would personally recommend all those be held for version 2, when the beacon becomes a service."
Jim Ostell, 2014

Implementation

1. Specifying the chromosome … The interface needs to specify the ***accession.version*** of a chromosome, or ***build number***…
2. Return values … right to ***refuse*** to answer without it being an error … DOS ***attack*** … or because …especially ***sensitive***…
3. Real time response … Some sites suggest that it would be necessary to have a ***“phone home” response*** …



Vague but exciting ...

CERN DD/OC — Tim Berners-Lee, CERN/DD

Information Management: A Proposal — March 1989

Information Management: A Proposal

Abstract

This proposal concerns the management of general information about accelerators and experiments at CERN. It discusses the problems of loss of information about complex evolving systems and derives a solution based on a distributed hypertext sytstem.

Keywords: Hypertext, Computer conferencing, Document retrieval, Information management, Project control

Tim Berners-Lee: Information Management: A Proposal (CERN 1989) & WWW: First Page (1990)

World Wide Web

The WorldWideWeb (W3) is a wide-area hypermedia information retrieval initiative aiming to give universal access to a large universe of documents.

Everything there is online about W3 is linked directly or indirectly to this document, including an executive summary of the project, Mailing lists , Policy , November's W3 news , Frequently Asked Questions .

What's out there?
: Pointers to the world's online information, subjects , W3 servers, etc.

Help
: on the browser you are using

Software Products
: A list of W3 project components and their current state. (e.g. Line Mode ,X11 Viola , NeXTStep , Servers , Tools , Mail robot , Library)

Technical
: Details of protocols, formats, program internals etc

Bibliography
: Paper documentation on W3 and references.

People
: A list of some people involved in the project.

History
: A summary of the history of the project.

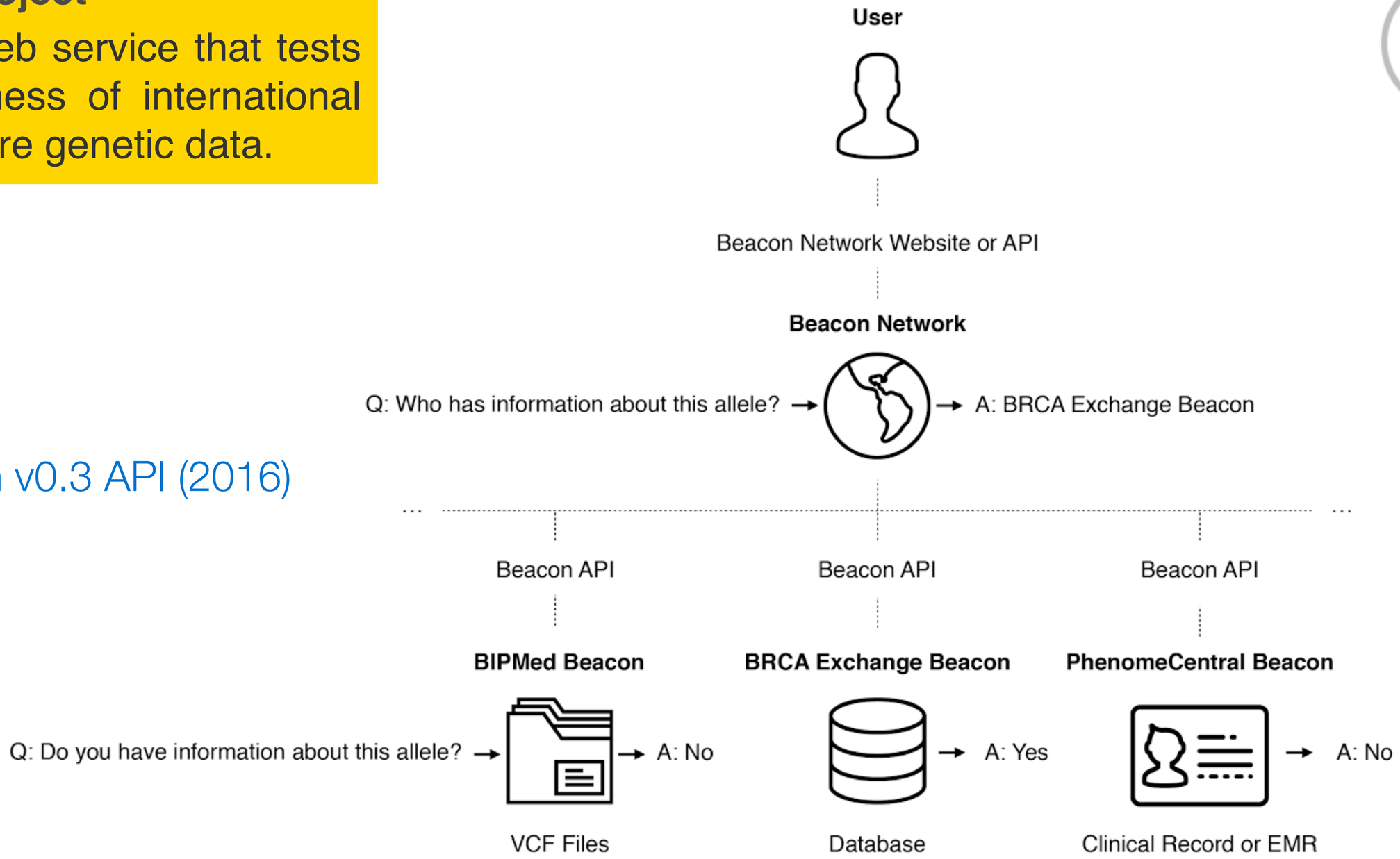
How can I help ?
: If you would like to support the web..

Getting code
: Getting the code by anonymous FTP , etc.

Beacon Project

An open web service that tests the willingness of international sites to share genetic data.

Beacon v0.3 API (2016)



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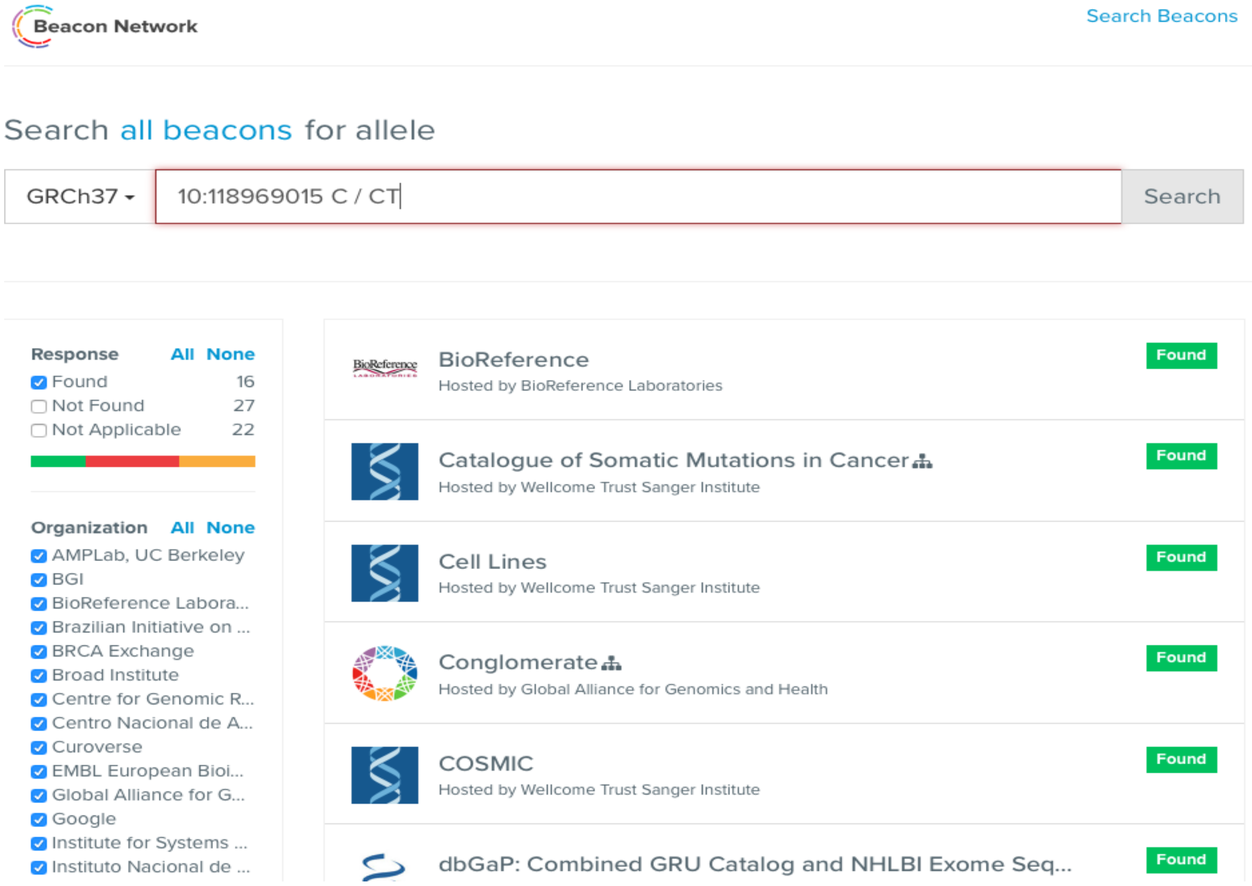

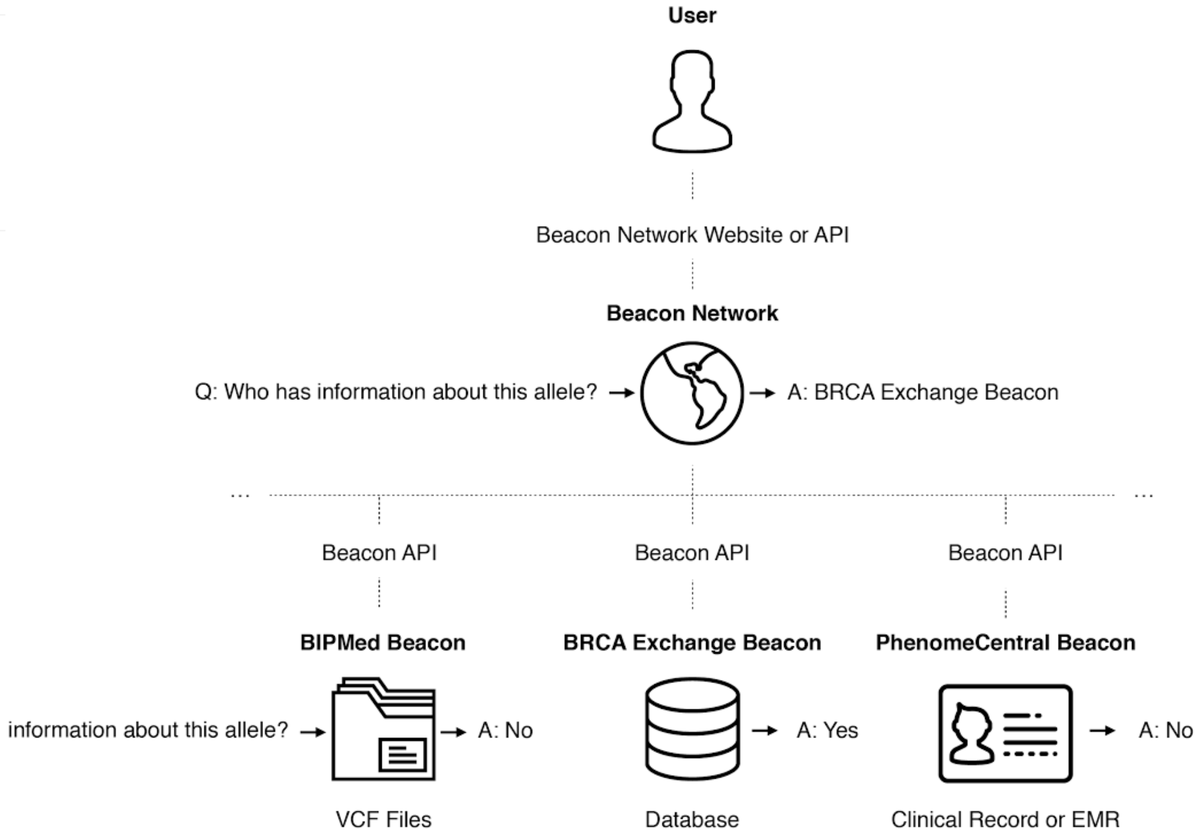

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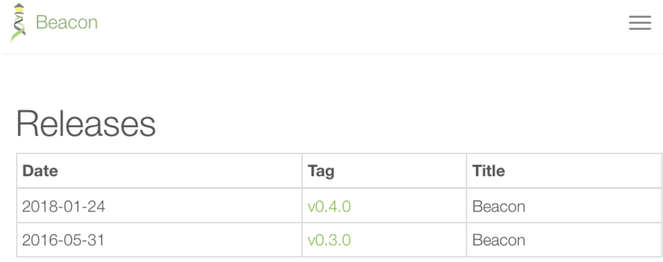
35+ Organizations

90+ Beacons

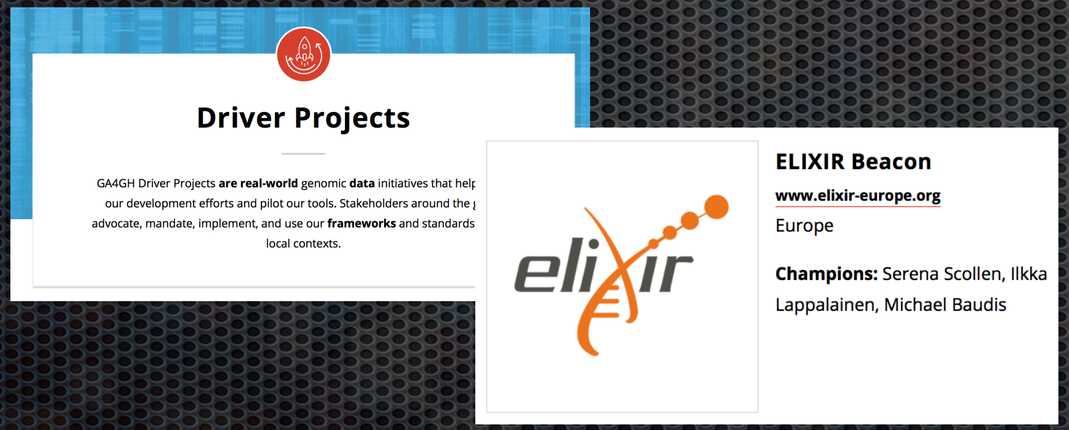
200+ Datasets

100K+ Individuals



2016++: **ELIXIR** Beacon Project

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



Beacon v0.4 forward

- **structural variations** (DUP, DEL) in addition to SNV
- **metadata** queries
- layered **authentication** system using **ELIXIR** AAI
- **quantitative** responses
- Beacon queries as entry for **data delivery** (outside Beacon protocol)
- Ubiquitous **deployment** (e.g. throughout **ELIXIR** network)

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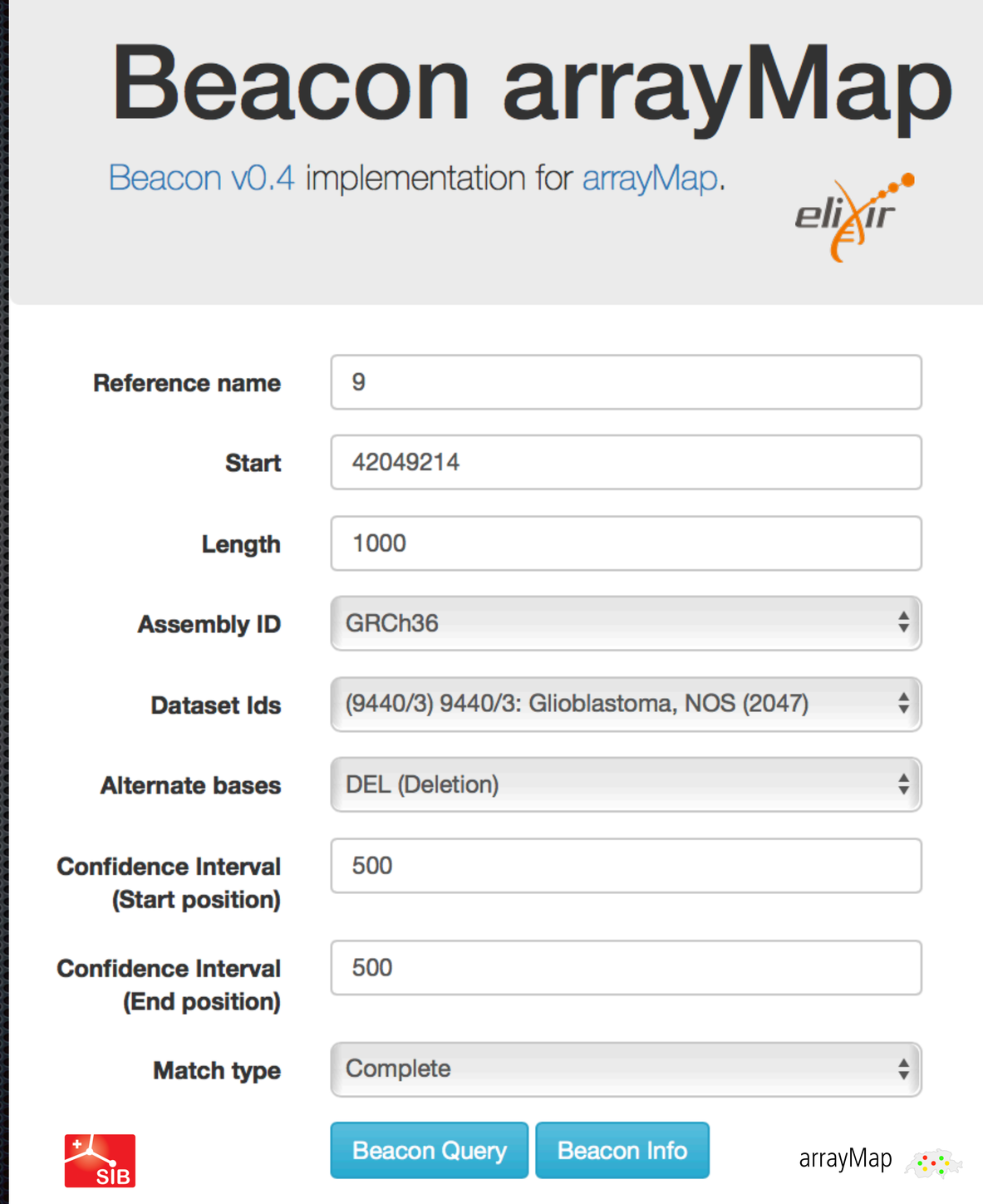
for Genomics & Health

Beacon+ Concept

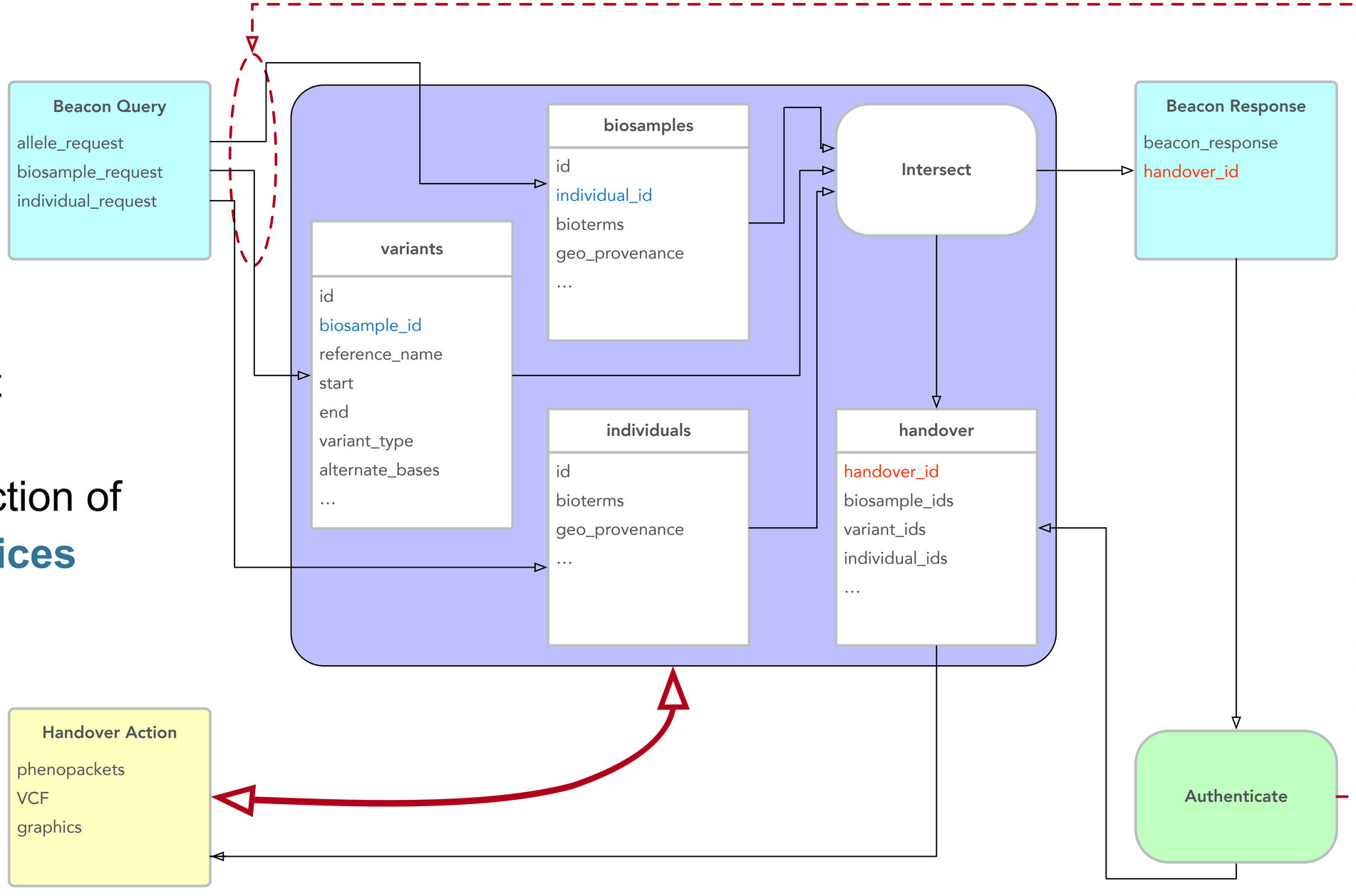
- Implementation of cancer beacon prototype, backed by arrayMap data
- structural variations
- quantitative queries
- metadata
- current version uses GA4GH schema compatible, non-SQL database backend (MongoDB)

Heinz Stockinger, Séverine Duvaud & SIB Technology Group



Beacon & Handover

Beacons to support advanced types of queries and connection of **data delivery services**



Beacon I/O

Handover

Authentication

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Beacon Handover

- only exposure of access handle to data stored in secure system
- one-step authentication and selection of *handover* action; other scenarios possible / likely
- *handover* response **outside of Beacon protocol / system**


```
{
"alleleRequest": {
"endMax": "26000000",
"referenceName": "9",
"startMax": "21975098",
"endMin": "21967753",
"startMin": "18000000",
"alternateBases": "N",
"variantType": "DEL",
"referenceBases": "*"
},
"url": "https://beacon.progenetix.org/beacon/info/",
"beaconId": "progenetix-beacon",
"datasetAlleleResponses": [
{
"externalUrl": "https://beacon.progenetix.org/beacon/info/",
"datasetId": "arraymap",
"variantCount": 588,
"info": {
"distinctVarCount": 551,
"description": "The query was against database \"arraymap\", variant collection \"variants\". 588 matched callsets for 588 distinct variants."},
"error": null,
"exists": true,
"datasetHandover": [
  {
    "url": "https://beacon.progenetix.test/beaconplus-server/beacondeliver.cgi?do=biosamplesdata&accessid=5d76f88d-4012-11e9-a0b4-d9893b611ec4",
    "handoverType": { "label": "Biosamples", "id": "pgx:handover:biosamplesdata" },
    "description": "retrieve data of the biosamples matched by the query"
  },
  {
    "url": "https://beacon.progenetix.test/beaconplus-server/beacondeliver.cgi?do=callsetsvariants&accessid=5d77fb88-4012-11e9-a0b4-bb5a9c8cf98a",
    "description": "export all variants of matched callsets - potentially huge dataset...",
    "handoverType": { "label": "Callsets Variants", "id": "pgx:handover:callsetsvariants" }
  },
  {
    "handoverType": {"id": "pgx:handover:cnvhistogram","label": "CNV Histogram"},
    "description": "create a CNV histogram from matched callsets",
    "url": "https://beacon.progenetix.test/beaconplus-server/beacondeliver.cgi?do=cnvhistogram&accessid=5d77fb88-4012-11e9-a0b4-bb5a9c8cf98a"
  },
  {
    "handoverType": { "label": "Variants", "id": "pgx:handover:variantsdata" },
    "description": "retrieve data of the variants matched by the query",
    "url": "https://beacon.progenetix.test/beaconplus-server/beacondeliver.cgi?do=variantsdata&accessid=5d6e982b-4012-11e9-a0b4-c5ce5cc21906"
  }
],
"callCount": 588,
"varResponses": [
"9:21773941-21968713:DEL",
"9:21732467-23813102:DEL",
"9:21785019-21968713:DEL",
"9:21968713-22031006:DEL",
```


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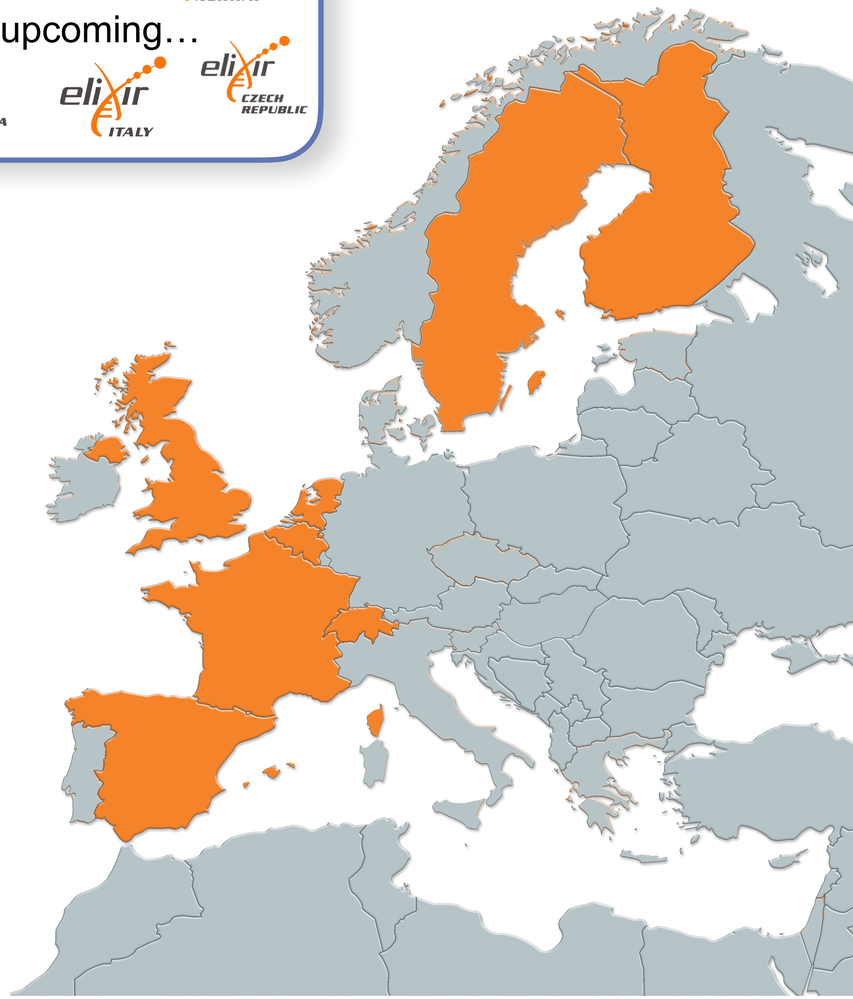
2018 Progress

- Release of v.1.0 specification
  - basis for **GA4GH Beacon v.1.0**
  - GA4GH certification (first product)
- Building flexible **Beacon Network**(s)
- Beacon paper under review
- Post-2018 planning, documented in **5-year plan**
- Widening Node participation & additional use cases:
  - ELIXIR Norway joined Implementation Study, to light Beacon(s) against **Marine data**
  - Widen this out to more use cases? Plants, other…



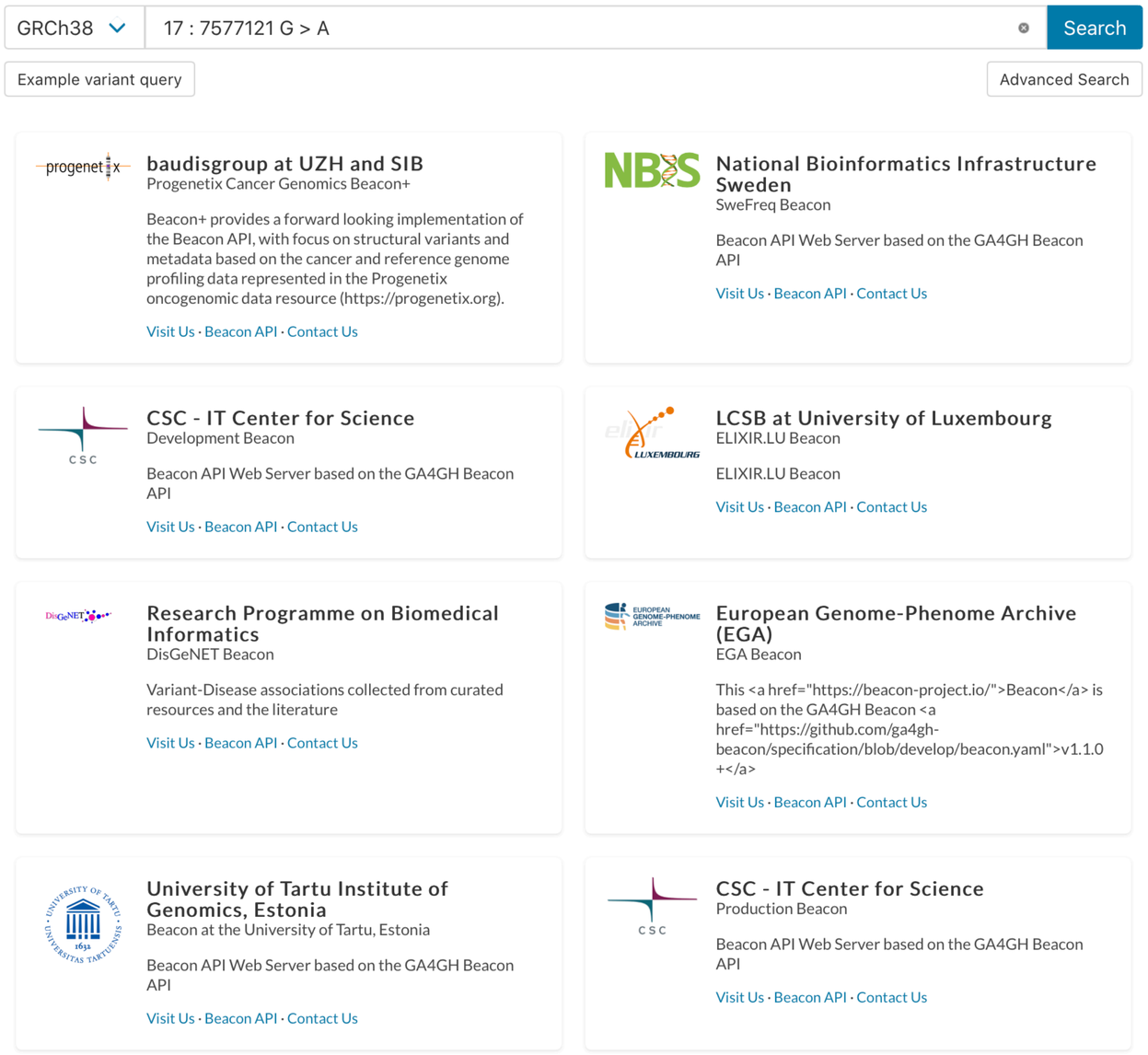
www.elixir-europe.org/beacons

ELIXIR Beacon Project Plan 2019-2021

- Extend the Beacon **protocol** to become the **reference ELIXIR Data Discovery product**, through expanding query options and providing richer responses, with view on biomedical applications, and in alignment with developing GA4GH standards
- Deliver ELIXIR Beacon Network as an established **ELIXIR service**
- Leverage ELIXIR Nodes to increase **data flow** through Beacon services
- Actively support the **integration** of the Beacon API **with human data resources** throughout ELIXIR, with particular view on National Genomics Initiatives, biobanks, and **H**uman **D**ata **C**ommunities (such as Rare Diseases and Human CNV)

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**



FEATURES AND POSSIBILITIES OF THE CURRENT BEACON SPECIFICATION

- precise variant queries (chr17: 7673767 C>T)
- range queries ("any variant from here to there")
- variant frequencies
- structural genome variants, e.g. CNVs ("any deletion overlapping CDKN2A CDR coordinates")
- delivery of any kind of data matching a given query (variants, sample information, patient data ...) utilising "handover" objects (anonymous links to external services with their own security / privacy implementations)
- networking of v1.n Beacons with AAI integration as demonstrated by the ELIXIR Beacon Network

IMPLEMENTATION CHALLENGES

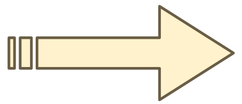
- interpretation and counting issues, especially w/ multi-allelic variants from VCF and "borderline" structural variants (e.g. INDEL vs. DEL, DUP:TANDEM vs. DUP...) - much of this related to using VCF instead of a variant model
- limited recognition & uptake of "recent" API versions
- only recent emergence of modern Beacon network aggregator
- protocol as "moving target" - while v1.n potential for widespread use, the rapid move towards v2 limits dissemination
- lack of well documented, flexible Beacon distributions
  - the "reference implementation" is limited & more for drop-in testing ("get your VCF online") than a scalable solution
  - other implementations (Ensembl, Beacon+ ...) more part of own ecosystem than for easy distribution

Beacon v2 - Areas of Change

- Separate query types for different genomic variants
  - SNPs ***BeaconSnvRequest***
  - 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

Tested and already implemented by Beacons

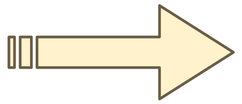
v2.0



GA4GH approval process ("major product update")

Ongoing

v2.n



incremental rollout after v2.0

adapted from Jordi Rambla | Feb 2020

Code leads: Sabela de la Torre Penas & Jordi Rambla

Beacon v2 Filters

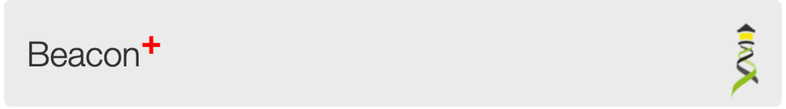
An *v2* extension of the Beacon protocol will allow the query for additional data beyond genome variants, using a proposed `filters` extension. Such filters are thought to be prefixed attributes, where the (public or private) prefix becomes the basis of scoping the value to the correct database value.

Overview of Beacon filters

The Beacon v2 API supports the discovery of genomics and clinical datasets, and includes a powerful feature to enable the "filtering" of beacon responses by biomedical properties (e.g. phenotypes) and procedural metadata.

Filters belong to one of currently three super-classes:

- `Filters` correspond to classes from bio-ontologies for biomedical data or procedural metadata that are contained in public repositories such as the Ontology Lookup Service or NCBO's BioPortal. Filters are identified using the full bio-ontology term/class identifier as CURIE, e.g. "HP:0100526". Examples of bio-ontologies are, among others:
  - Human Phenotype Ontology (HPO)
    - `HP:0100526`: Neoplasm of the lung
  - Phenotypic Qualities Ontology (PATO)
    - `PATO:0020000`: female genetic sex
  - National Cancer Institute Thesaurus (NCIT)
    - `NCIT:C8430`: Ovarian Papillary Tumor
    - `NCIT:C48724`: T2 Stage Finding
  - Experimental Factor Ontology (EFO)
    - `EFO:0009656`: neoplastic sample
- `CustomFilters` are biomedical or metadata terms that are locally defined by a Beacon (e.g. not corresponding to known bio-ontology terms). As with standard `Filters`, the Beacon v2 API is agnostic to how `CustomFilters` are implemented by the Beacon, so this permits maximum flexibility with regards to identifying, labelling and grouping CustomFilters. For example, related phenotype terms or experimental sets could be grouped into local "dictionaries", which could be addressed through a local identifier. The only requirement is that each custom filter term contains a unique identifier that can be used in Beacon requests.
- `FuzzyFilters` are implementations of classifiers which allow for some alternatives in matching and mostly can be drop-ins where ontologies are incomplete. Logically, through the potential matching of multiple values they provide a limited alternative mechanism to allow `OR`-style queries.



This example shows a core Beacon query, against a specific mutation in the TP53 gene, in cellosaurus, with ClinVar data.

CNV Example | SNV Range Example | SNV Example | ClinVar Example | Beacon Help

Dataset*

arraymap
progenetix
cellosaurus
dipg
BeaconSpecTest2
BeaconSpecTest

Genome Assembly*

GRCh38 / hg38

Dataset Responses

All Selected Datasets

Reference name*

17

Gene Coordinates

TP53

Cytoband(s)

17p13.1

Start

7673767

Ref. Base(s)

C

Alt. Base(s)

T

Bio-ontology

no selection
NCIT:C102872: Pharyngeal squamous cell carcinoma (2)
NCIT:C103968: Pyruvate dehydrogenase deficiency (1)
NCIT:C105555: High grade ovarian serous adenocarcinoma (75)
NCIT:C105556: Low grade ovarian serous adenocarcinoma (10)
NCIT:C111802: Dyskeratosis congenita (3)

Other Filters

additional comma-separated, prefixed filters

Beacon Query

Beacon+
Flexible Modeling of New Features

Our Beacon platform is being used for the rapid testing of queries and responses - both v1.n and v2.0.a - against a number of partially large-scale genome datasets.

- Progenetix (>100000 cancer CNV profiles)
- DIPG (childhood brain tumor study)
- NEW: Cellosaurus ClinVar annotations for evidence representation
- Brewing: COVID-19

Currently running on a Perl+MongoDB stack, a Python-based OS solution is in early development.


```
[
  {
    "callset_id": "cs-cellosaurus:CVCL_EI02",
    "info": {
      "cellosaurus": {
        "cell_line": "BT474-LAPRa",
        "id": "CVCL_EI02",
        "cellosaurus_variant_name": "TP53 p.Glu285Lys (c.853G&gt;A)"
      },
      "clinvar": {
        "gene_id": "7157",
        "allele_id": "410258",
        "assembly": "GRCh38",
        "cytoband": "17p13.1",
        "variant_type": "single nucleotide variant",
        "origin": "germline;somatic",
        "phenotype": "Hereditary cancer-predisposing syndrome;Li-Fraumeni
syndrome;PARP Inhibitor response;not provided",
        "clinical_significance": "Pathogenic/Likely pathogenic",
        "clinvar_full_name": "NM_001126112.2(TP53):c.853G&gt;A (p.Glu285Lys)"
      }
    },
    "start_min": 7673766,
    "reference_name": "17",
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    ],
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    "start_max": 7673766
  },
  {
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    "start_max": 7673766,
    "callset_id": "cs-cellosaurus:CVCL_AQ07",
    "start_min": 7673766,
    "info": {
      "cellosaurus": {
        "cellosaurus_variant_name": "TP53 p.Glu285Lys (c.853G&gt;A)",
        "cell_line": "BT-474 Clone 5",
        "id": "CVCL_AQ07"
      },
      "clinvar": {
        "assembly": "GRCh38",
        "allele_id": "410258",
        "gene_id": "7157",
        "cytoband": "17p13.1",
        "variant_type": "single nucleotide variant",
        "phenotype": "Hereditary cancer-predisposing syndrome;Li-Fraumeni
syndrome;PARP Inhibitor response;not provided",
        "origin": "germline;somatic",
        "clinvar_full_name": "NM_001126112.2(TP53):c.853G&gt;A (p.Glu285Lys)",
        "clinical_significance": "Pathogenic/Likely pathogenic"
      }
    },
    "end_min": 7673767,
    "biosample_id": "bios-cellosaurus:CVCL_AQ07",
    "reference_name": "17"
  },
  {
    "alternate_bases": [
      "T"
    ],
    "reference_bases": "C",
    "digest": "17_7673767_C_T",
    "end_max": 7673767,
    "variantset_id": "cellosaurus_clinvar_GRCH38",
    "start_max": 7673766,
    "callset_id": "cs-cellosaurus:CVCL_0179",
```
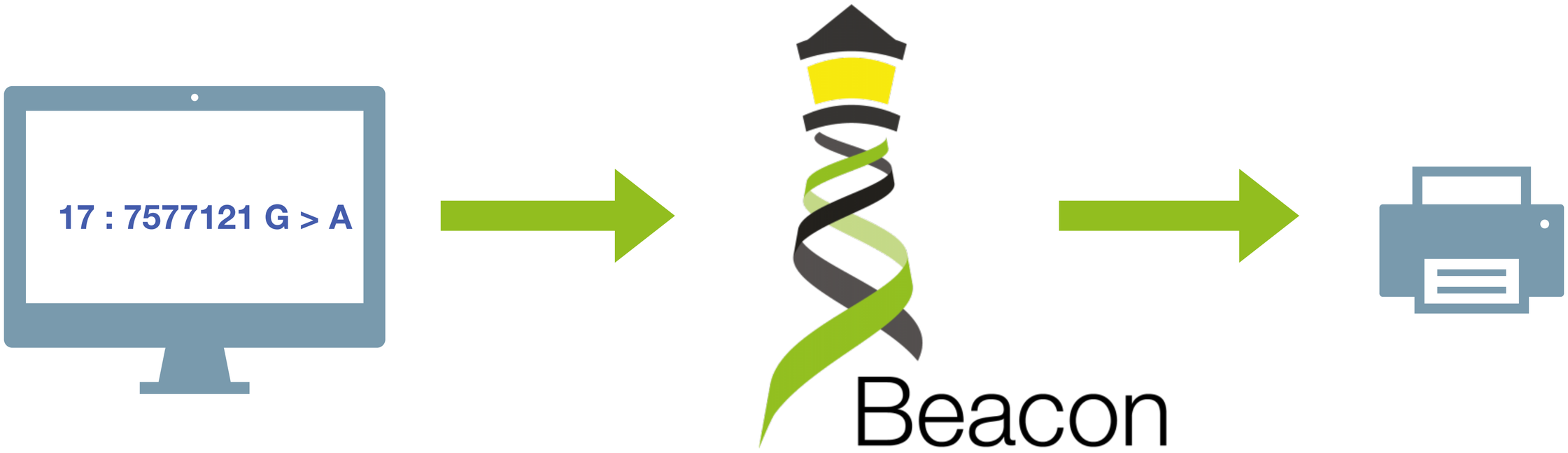



A ***Beacon*** answers a query for a specific genome variant against individual or aggregate genome collections

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.

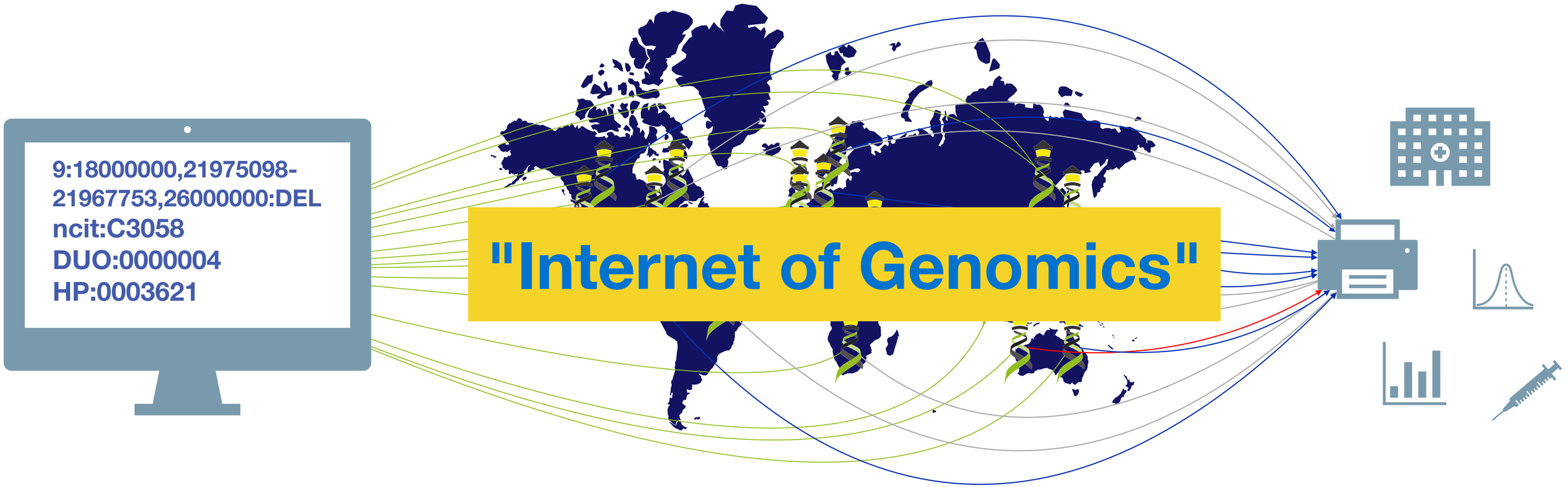
A Beacon network federates *genome variant queries* across databases that support the ***Beacon API***

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**".



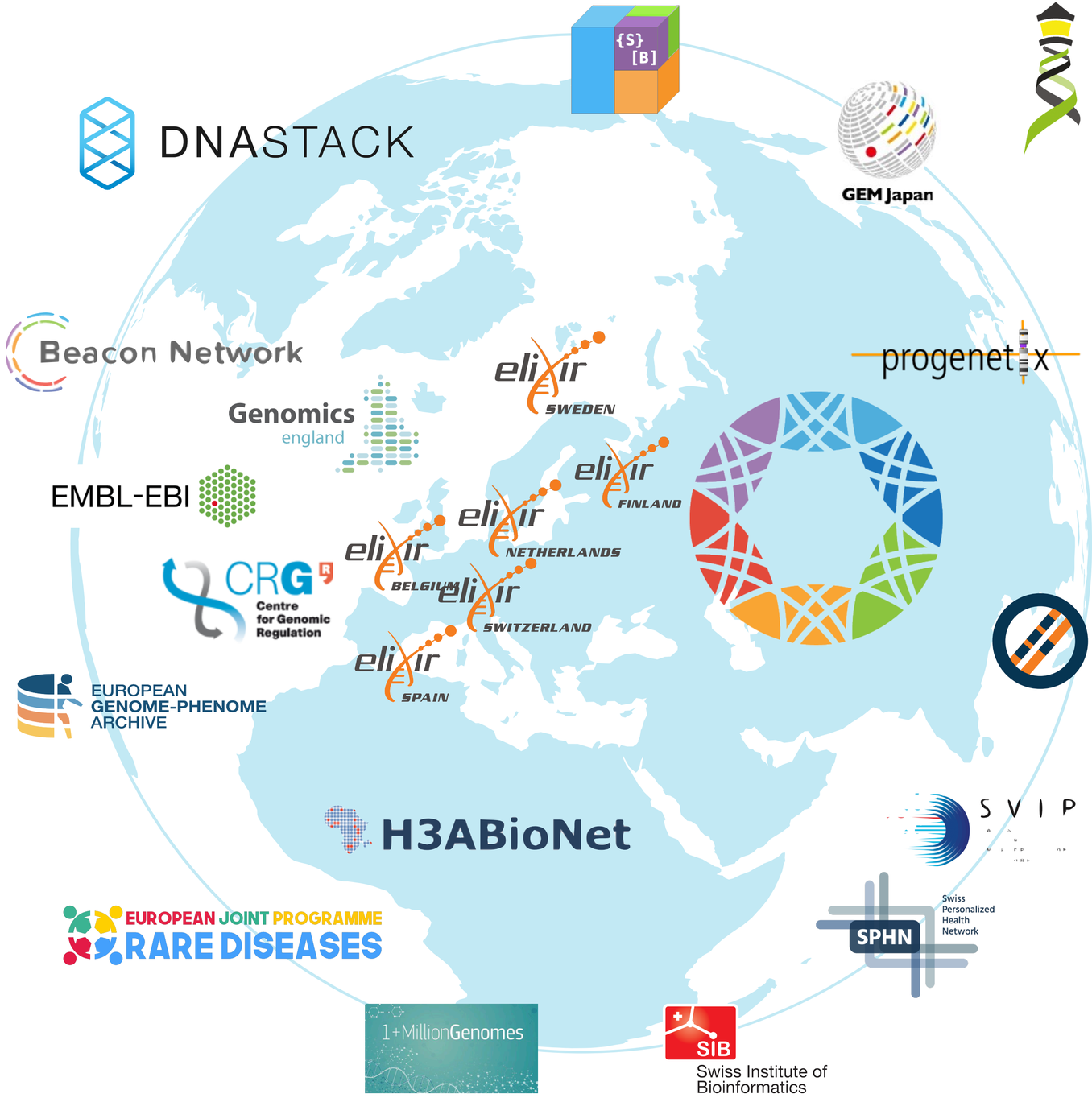
Have you seen deletions in this region on chromosome 9 in Glioblastomas from a juvenile patient, in a dataset with unrestricted access?

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
  - **E**uropean **J**oint **P**rogram on **R**are **D**iseases
  - 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



GA4GH Genome Beacons

A Driver Project of the Global Alliance for Genomics and Health GA4GH and supported through ELIXIR

News
Specification & Roadmap
Beacon Networks
Events
Examples, Guides & FAQ
Contributors & Teams
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Meeting Minutes

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GA4GH::Discovery

Github Projects

Beacon API and Tools
SchemaBlocks

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Beacon Protocol for Genomic Data Sharing

Beacons provide discovery services for genomic data using the Beacon API developed as a key driver project of the **Global Alliance for Genomics and Health** (GA4GH). The *Beacon* protocol itself defines an open standard for genomics data discovery. It provides a framework for public web services responding to queries against genomic data collections, for instance from population based or disease specific genome repositories.

The original Beacon protocol had been designed to be:

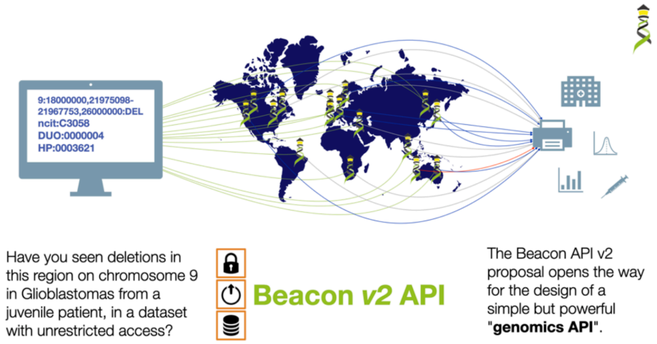
- **Simple:** focus on robustness and easy implementation
- **Federated:** maintained by individual organizations and assembled into a network
- **General-purpose:** used to report on any variant collection
- **Aggregative:** provide a boolean (or quantitative) answer about the observation of a variant
- **Privacy protecting:** queries do not return information about single individuals

Sites offering *beacons* can scale through aggregation Beacon Networks, which distribute single genome queries among a potentially large number of international *beacons* and assemble their responses.

Since 2015 the development of the Beacon protocol has been led by ELIXIR in collaboration with GA4GH and international participants. Recent versions of the *Beacon* protocol have expanded the original concept by e.g.:

- providing a framework for other types of genome variation data (i.e. range queries and structural variants
- allowing for data delivery using ***handover*** protocol, e.g. to link with clinical information in protected environments and allow for data delivery and visualisation services

Beacon v2 - Towards Flexible Use and Clinical Applications



beacon-project.io

Beacon+

This example shows the query for CNV deletion variants overlapping the CDKN2A gene's coding region with at least a single base, but limited to "focal" hits (here i.e. <= ~4Mbp in size). The query is against the arrayMap collection and can be modified e.g. through changing the position parameters or data source.

Query

BND Example | SNV Example | SNV Range Example | CNV Example

Dataset*	tcga
Reference name*	5
Genome Assembly*	GRCh38 / hg38
(structural) variantType	DEL (Deletion)
Gene Coordinates	[PDE4D] 5:58974663-60185597
Start *min* Position*	56974663
Start *max* Position	60185596
End *min* Position	58974664
End *max* Position	62185597
Bio-ontology	no selection icdom:94403: Glioblastoma, NOS (2048) icdom:94423: Gliosarcoma (9) ncit:C102872: Pharyngeal squamous cell... (9) ncit:C105555: High grade ovarian serous... (78) ncit:C114656: Endometrial adenosquamous... (4)

Beacon Query

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Bo Gao
Qingyao Huang
Rahel Paloots
Paula Carrio Cordo
Séverine Duvaud (SIB)

Response

Dataset	Assembly	Chro	Position Start Range End Range	Ref Alt Type	Bio Query	Variants Calls Samples	$f_{alleles}$	Response Context
arraymap	GRCh38	5	56974663 - 60185596 58974664 - 62185597	* N DEL	icdot:C61.9	60 47 47	0.0008	JSON UCSC [H->O] Biosamples [H->O] Callsets Variants [H->O] CNV Histogram [H->O] Individuals [H->O] Variants
tcga	GRCh38	5	56974663 - 60185596 58974664 - 62185597	* N DEL	icdot:C61.9	35 27 27	0.0012	JSON UCSC [H->O] Biosamples [H->O] Callsets Variants [H->O] CNV Histogram [H->O] Individuals [H->O] Variants

beacon.progenetix.org

Session Schedule

15 min	**Beacon - where are we now, and how did we get here?**	*Michael Baudis*
15 min	**ELIXIR Beacon Network: what does it offer to users?**	*Juha Tornroos*
15 min	**Beacon v2 - status, Use Cases, and implementations: examples from ES**	*Jordi Rambla*
10 min	**Beacon is secure: what does this mean, how does this interact with GA4GH, and how will this work with v2**	*Dylan Spalding*
10 min	**The Beacon API Compliance Tester: do the Beacons really speak the same language?**	*Malin Klang*
5 min	**Refreshment break**	
25 min	**Beacon and Covid-19**	*Jordi Rambla*
10 min	**Beacon as a discovery tool for human data outside of genomics: Proteomics**	*Juan Antonio Vizcaino*
15 min	**Beacon in the ELIXIR Programme - Node perspective from FI**	*Juha Törnroos*