

Beacon v2 and Beyond

The Standard for Data *Discovery* and Data *Sharing* in Biomedical Genomics



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Co-lead ELIXIR Beacon API Development

Co-lead ELIXIR hCNV Community



Universität
Zürich^{UZH}



SIB
Swiss Institute of
Bioinformatics



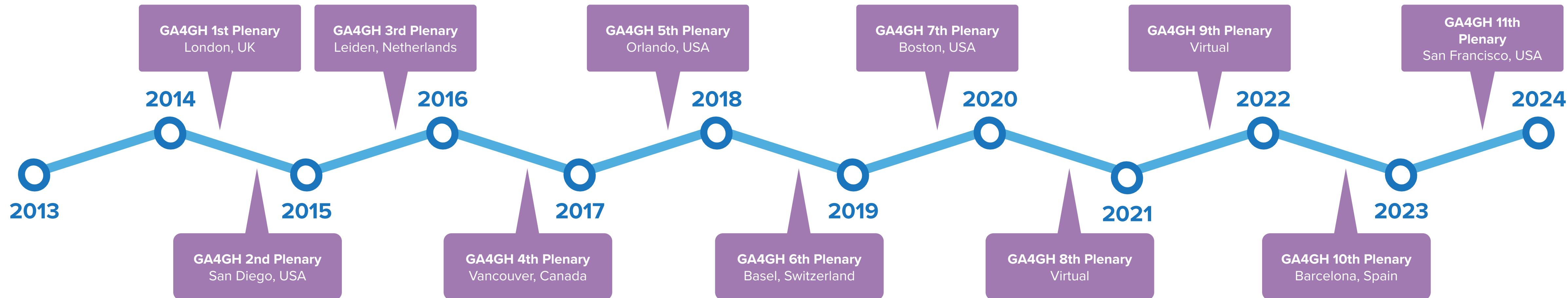
Global Alliance
for Genomics & Health
Collaborate. Innovate. Accelerate.



GA4GH timeline



Global Alliance
for Genomics & Health



Pre-launch	Building momentum	GA4GH Connect	Gap analysis	Strategic Refresh
 <p>73 partners sign a letter of intent to form an alliance</p>	 <p>Global Alliance for Genomics & Health Collaborate. Innovate. Accelerate.</p> <p>Formal launch of GA4GH</p> <p>Published <i>Framework for Responsible Sharing of Genomic and Health-Related Data</i></p> <p>Formed four working groups</p> <p>Developed three demonstration projects</p>	 <p>Launch of GA4GH Connect and Strategic Roadmap</p> <p>Formation of new organizational structure consisting of eight Work Streams and over twenty Driver Projects</p>	<p>Gap analysis identifies three community imperatives</p> <ul style="list-style-type: none"> Interoperability and alignment Implementation support Engaging with healthcare and clinical standards	 <p>Strategic refresh introduces updates to GA4GH to better meet the three community imperatives</p>

Beacon v1 Development

2014

GA4GH founding event; Jim Ostell proposes Beacon concept including "more features ... version 2"

2015

- beacon-network.org aggregator created by DNAstack

2016

- Beacon v0.3 release
- work on queries for structural variants (brackets for fuzzy start and end parameters...)

2017

- OpenAPI implementation
- integrating **CNV parameters** (e.g. "startMin, statMax")
- Beacon v0.4 release in January; feature release for GA4GH approval process
- **GA4GH Beacon v1 approved** at Oct plenary

2019

- ELIXIR Beacon Network

2020



2021

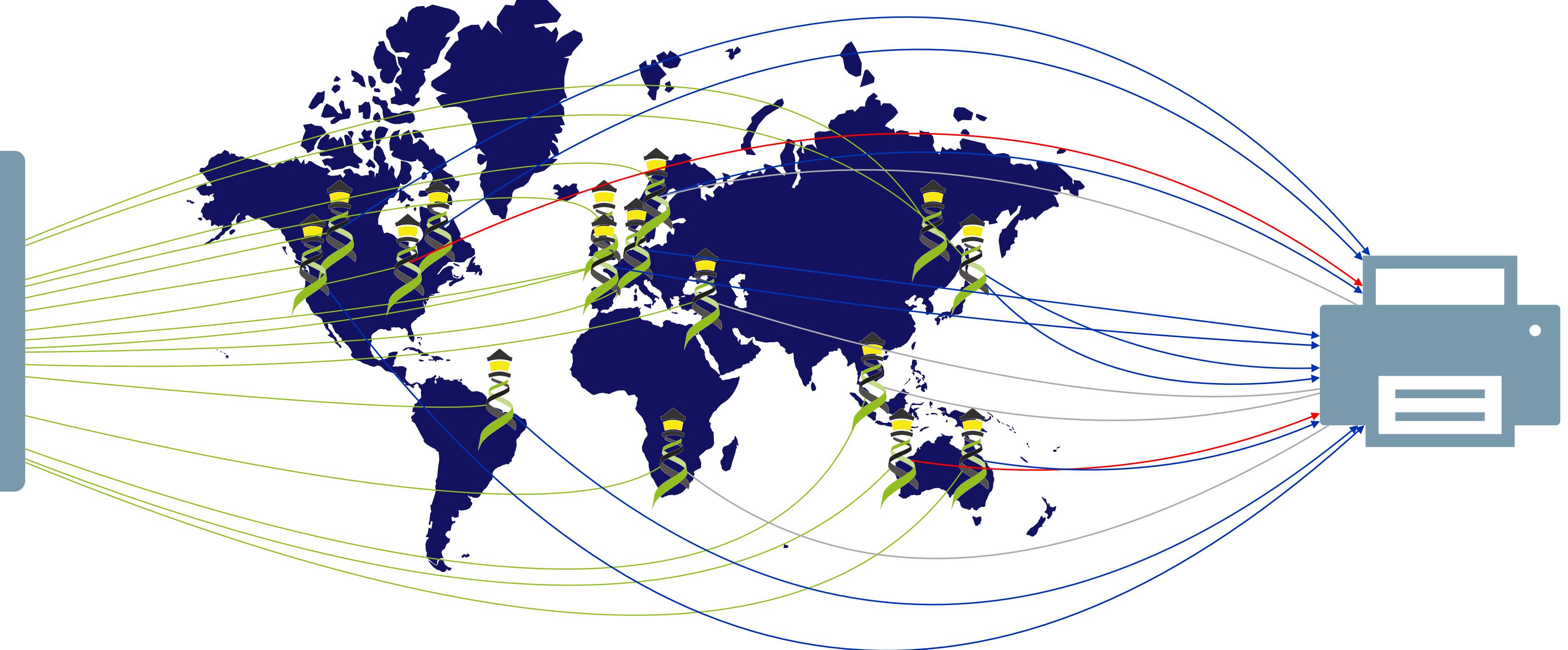
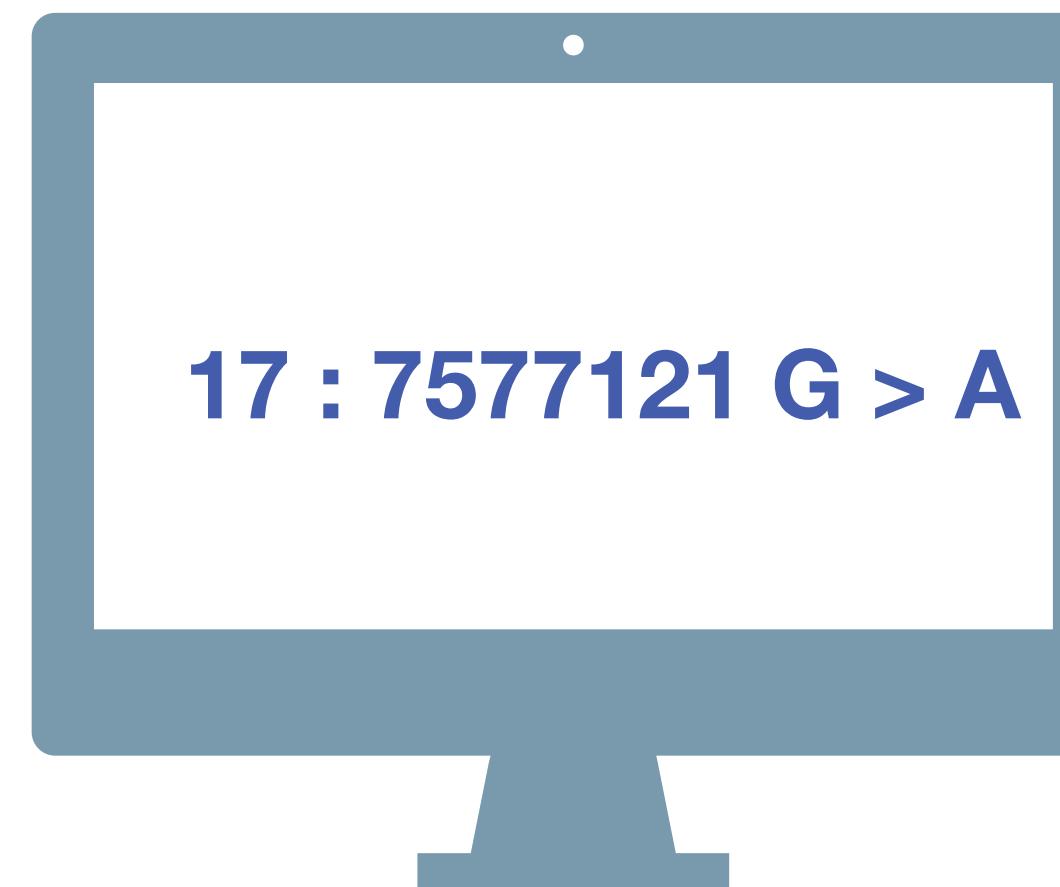
Beacon v2 Development

2022

- Beacon+ concept implemented @ [progenetix.org](#)
- concepts from GA4GH Metadata (ontologies...)
- entity-scoped query parameters ("individual.age")
- Beacon+ demos "handover" concept
- Beacon hackathon Stockholm; settling on **filters**
- Barcelona goes Zurich developers meeting
- Beacon API v2 Kick off
- adopting "handover" concept
- "Scouts" teams working on different aspects - filters, genomic variants, compliance ...
- **framework + models** concept implemented
- range and bracket queries, variant length
- starting of GA4GH review process
- changes in default model, aligning with Phenopackets and VRS
- unified beacon-v2 code & docs repository
- **Beacon v2 approved** at April GA4GH Connect

Related ...

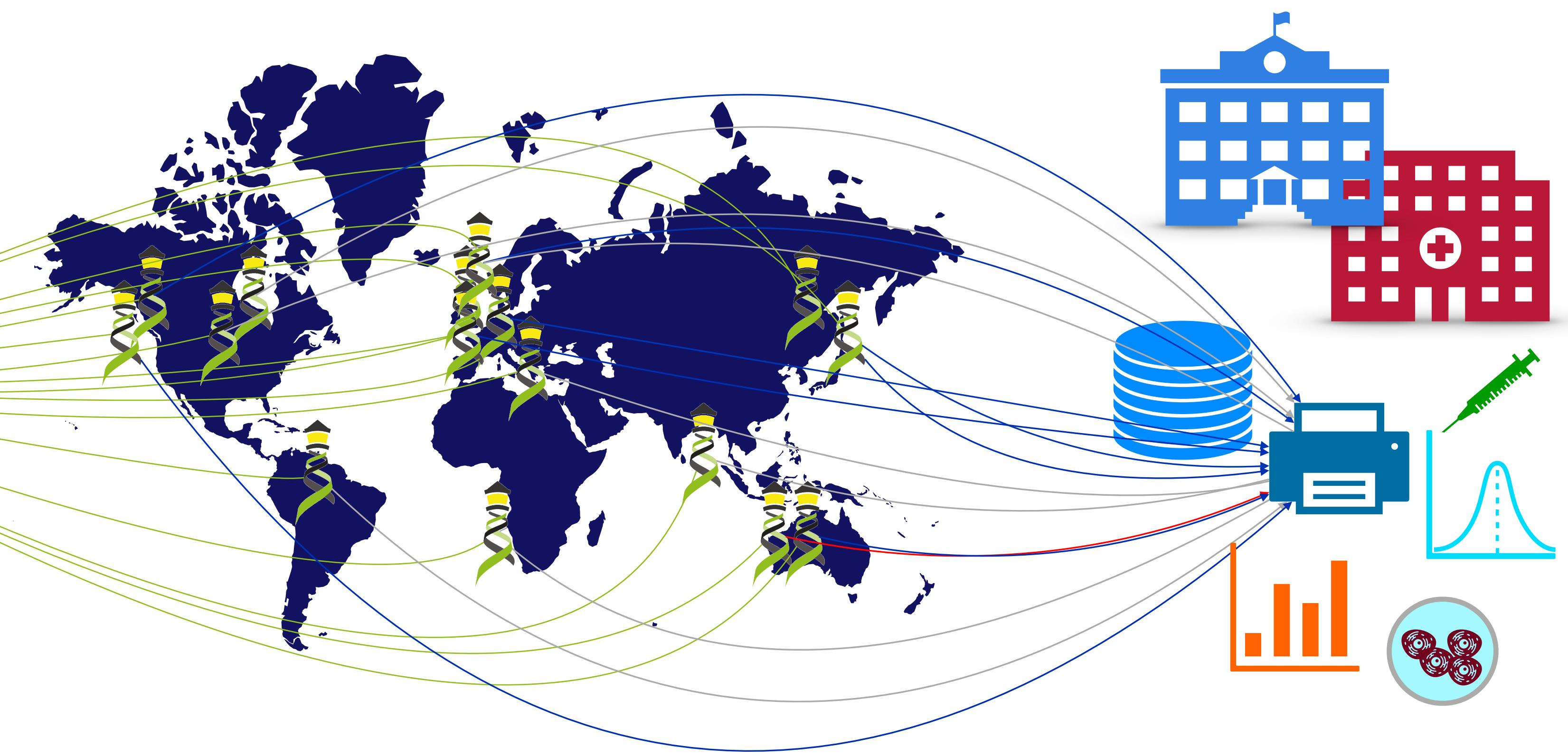
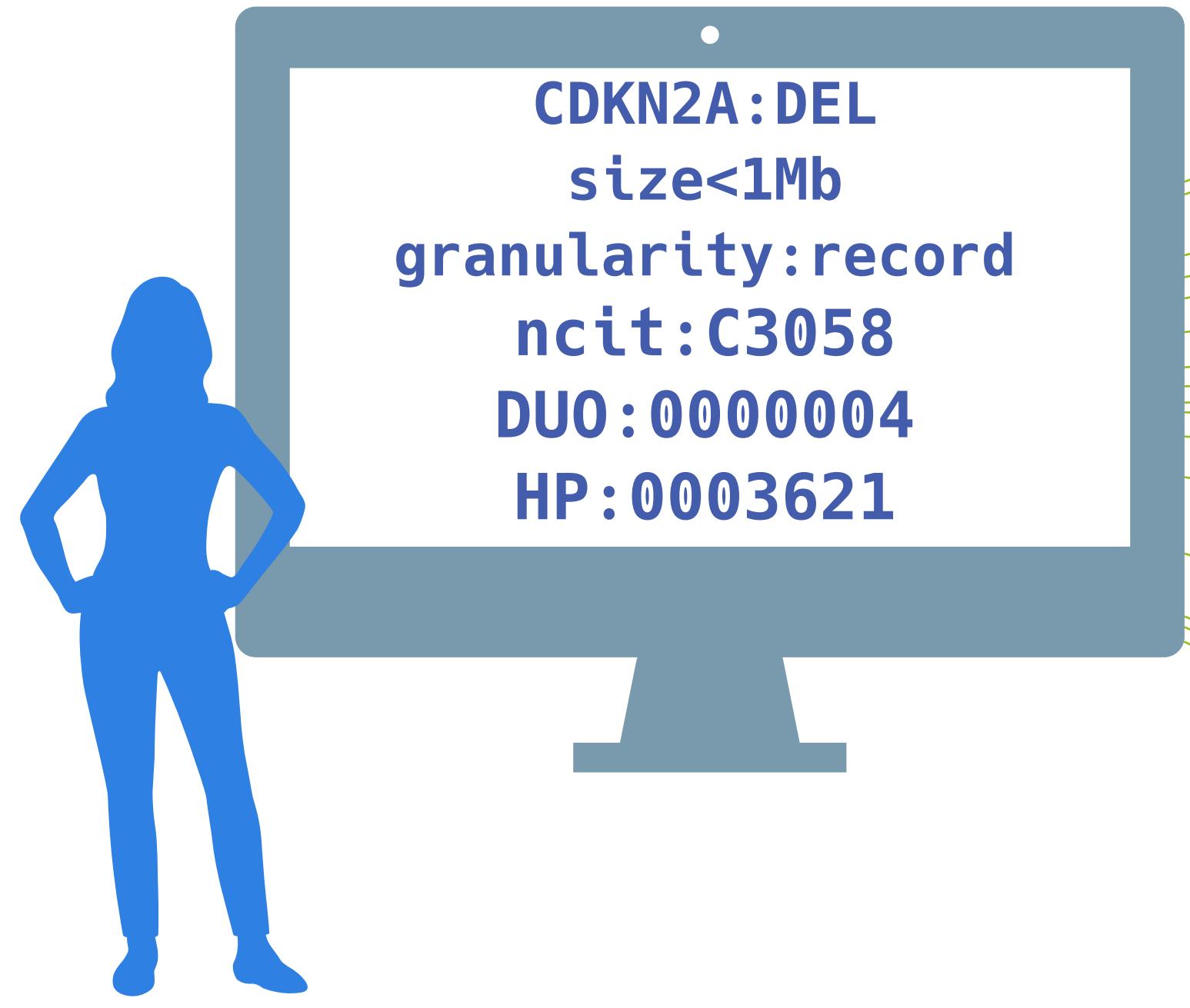
- ELIXIR starts Beacon project support
- GA4GH re-structuring (workstreams...)
- Beacon part of Discovery WS
- new Beacon website (March)
- Beacon publication at Nature Biotechnology
- Phenopackets v2 approved
- [docs.genomebeacons.org](#)



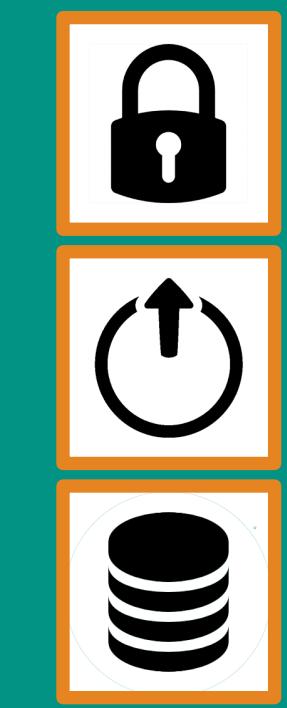
Have you seen this variant?
It came up in my patient
and we don't know if this is
a common SNP or worth
following up.

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**.



Can you provide data about focal deletions in CDKN2A in Glioblastomas from juvenile patients with unrestricted access?

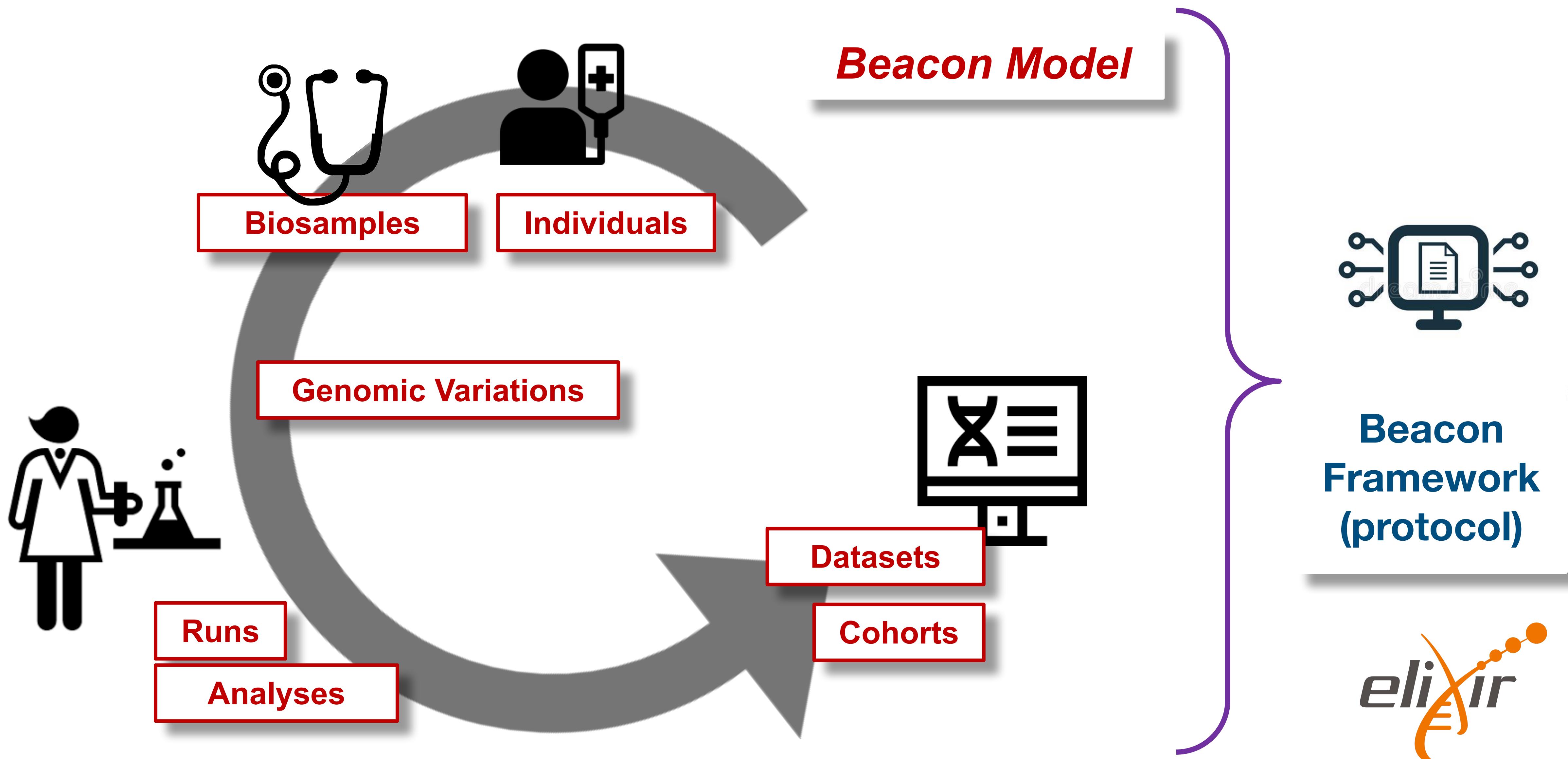


Beacon **v2** API

The Beacon API v2 represents a simple but powerful **genomics API** for **federated** data discovery and retrieval

Beacon v2

docs.genomebeacons.org



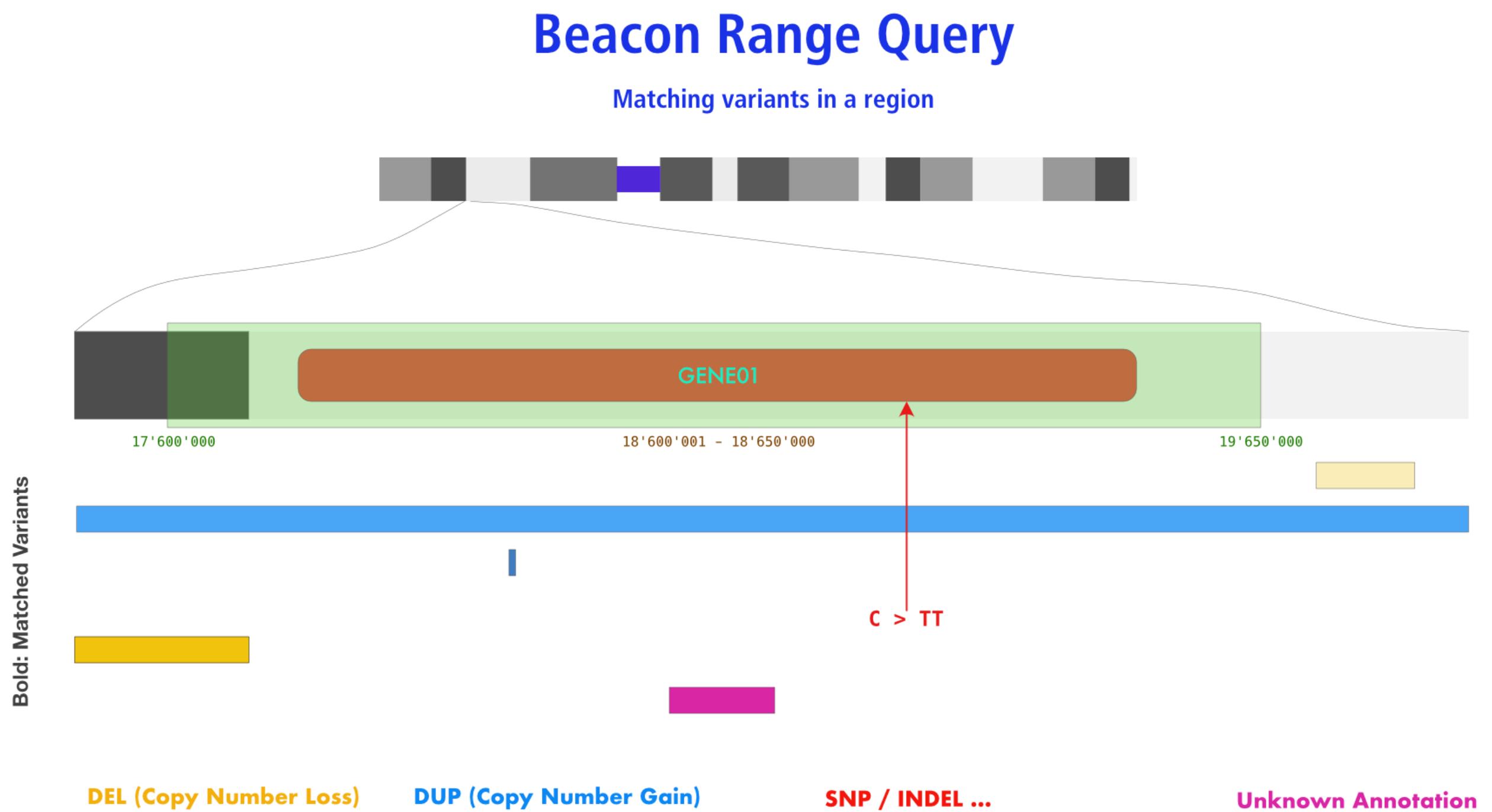
**Beacon
Framework
(protocol)**



Variation Queries

Range ("anything goes") Request

- defined through the use of 1 start, 1 end
- any variant... but can be limited by type etc.



Beacon Query Types

Sequence / Allele CNV (Bracket) **Genomic Range** Aminoacid Gene ID HGVS Sam

Dataset: Test Database - examplez

Chromosome: 17 (NC_000017.11)

Variant Type: SO:0001059 (any sequence alteration - S...)

Start or Position: 7572826

End (Range or Structural Var.): 7579005

Reference Base(s): N

Alternate Base(s): A

Select Filters: Chromosome 17

Query Database

Form Utilities: Gene Spans, Cytoband(s)

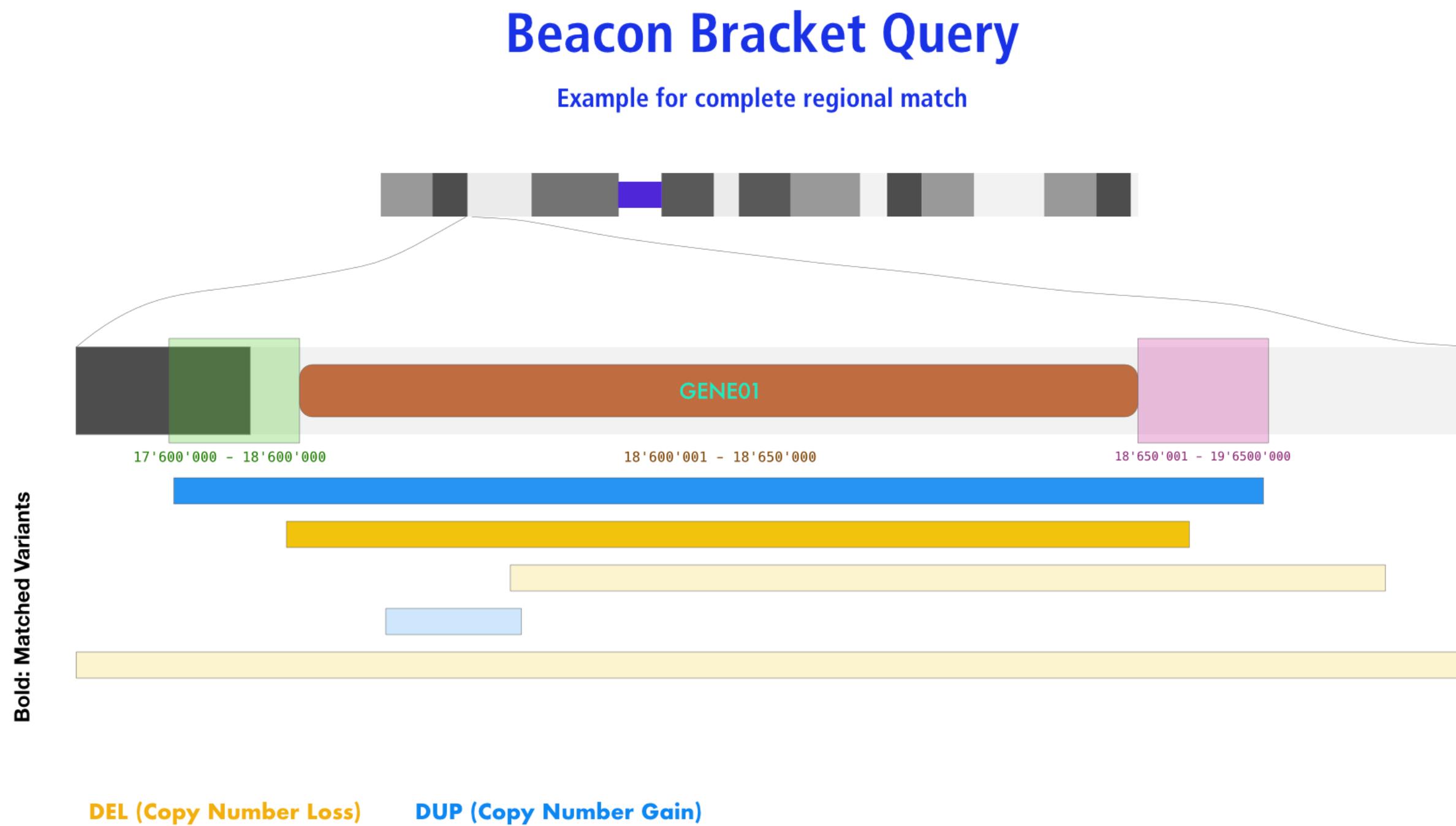
Query Examples: CNV Example, SNV Example, Range Example, Gene Match, Aminoacid Example, Identifier - HeLa

As in the standard SNV query, this example shows a Beacon query against mutations in the EIF4A1 gene in the DIPG childhood brain tumor dataset. However, this range + wildcard query will return any variant with alternate bases (indicated through "N"). Since parameters will be interpreted using an "AND" paradigm, either Alternate Bases OR Variant Type should be specified. The exact variants which were being found can be retrieved through the variant handover [H->O] link.

Variation Queries

Bracket ("CNV") Query

- defined through the use of 2 start, 2 end
- any contiguous variant...



Beacon Query Types

Sequence / Allele **CNV (Bracket)** Genomic Range Aminoacid Gene ID HGVS Sam

Dataset

Test Database - examplez X | ▼

Chromosome i

9 (NC_000009.12) | ▼

Variant Type i

EFO:0030067 (copy number deletion) | ▼

Start or Position i

21000001-21975098

End (Range or Structural Var.) i

21967753-23000000

Select Filters i

NCIT:C3058: Glioblastoma (100) X | ▼

Chromosome 9 i

21000001-21975098



Query Database

Form Utilities

⚙️ Gene Spans

⚙️ Cytoband(s)

Query Examples

[CNV Example](#)

[SNV Example](#)

[Range Example](#)

[Gene Match](#)

[Aminoacid Example](#)

[Identifier - HeLa](#)

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. $\leq \sim 2\text{Mbp}$ in size). The query is against the examplez collection and can be modified e.g. through changing the position parameters or data source.

Standards Development & Implementation: CNV Terms

in computational (file/schema) formats

- EFO:0030064
- EFO:0030067
 - | - EFO:0030068
 - \ - EFO:0020073
 - \ - EFO:0030069
- EFO:0030070
 - | - EFO:0030071
 - \ - EFO:0030072

GA4GH VRS1.3+	Beacon v2	VCF v4.4	SO
EFO:0030070 gain	DUP or EFO:0030070	DUP SVCLAIM=D	SO:0001742 copy_number_gain
EFO:0030071 low-level gain	DUP or EFO:0030071	DUP SVCLAIM=D	SO:0001742 copy_number_gain
EFO:0030072 high-level gain	DUP or EFO:0030072	DUP SVCLAIM=D	SO:0001742 copy_number_gain
EFO:0030072 high-level gain	DUP or EFO:0030073	DUP SVCLAIM=D	SO:0001742 copy_number_gain
EFO:0030067 loss	DEL or EFO:0030067	DEL SVCLAIM=D	SO:0001743 copy_number_loss
EFO:0030068 low-level loss	DEL or EFO:0030068	DEL SVCLAIM=D	SO:0001743 copy_number_loss
EFO:0020073 high-level loss	DEL or EFO:0020073	DEL SVCLAIM=D	SO:0001743 copy_number_loss
EFO:0030069 complete genomic loss	DEL or EFO:0030069	DEL SVCLAIM=D	SO:0001743 copy_number_loss

Beacon v2 Filters

Example: Use of hierarchical classification systems (here NCI neoplasm core)

- Beacon v2 relies heavily on "filters"
 - ontology term / CURIE
 - alphanumeric
 - custom
 - Beacon v2 "filters" assumes inclusion of child terms when using hierarchical classifications
 - implicit *OR* with otherwise assumed *AND*
 - implementation of hierarchical annotations overcomes some limitations of "fuzzy" disease annotations



Beacon+ specific: Multiple term selection with OR logic

<input checked="" type="checkbox"/>	> NCIT:C4914: Skin Carcinoma	213
<input type="checkbox"/>	> NCIT:C4475: Dermal Neoplasm	109
<input checked="" type="checkbox"/>	> NCIT:C45240: Cutaneous Hematopoietic and Lymphoid Cell Neoplasm	310

Filters: NCIT:C4914, NCIT:C4819, NCIT:C9231, NCIT:C2921, NCIT:C45240, NCIT:C6858, NCIT:C3467, NCIT:C45340, NCIT:C7195, NCIT:C3246, NCIT:C7217

progenetix							
Variants: 0	$f_{alleles}$: 0	Callsets	Variants	UCSC region	Legacy Interface	 Show JSON Response	
Calls: 0							
Samples: 523							
Results	Biosamples						
Id	Description	Classifications		Identifiers	DEL	DUP	CNV
PGX_AM_BS_MCC01	Merkel cell carcinoma	icdot-C44.9 Skin, NOS icdom-82473 Merkel cell carcinoma NCIT:C9231 Merkel Cell Carcinoma		PMID:9537255	0.116	0.104	0.22
PGX_AM_BS_MCC02	Merkel cell carcinoma	icdot-C44.9 Skin, NOS icdom-82473 Merkel cell carcinoma NCIT:C9231 Merkel Cell Carcinoma		PMID:9537255	0.154	0.056	0.21
PGX_AM_BS_MCC03	Merkel cell carcinoma	icdot-C44.9 Skin, NOS icdom-82473 Merkel cell carcinoma NCIT:C9231 Merkel Cell Carcinoma		PMID:9537255	0.137	0.21	0.347
PGX_AM_BS_MCC04	Merkel cell carcinoma	icdot-C44.9 Skin, NOS icdom-82473 Merkel cell carcinoma NCIT:C9231 Merkel Cell Carcinoma		PMID:9537255	0.158	0.056	0.214
PGX_AM_BS_MCC05	Merkel cell carcinoma	icdot-C44.9 Skin, NOS icdom-82473 Merkel cell carcinoma NCIT:C9231 Merkel Cell Carcinoma		PMID:9537255	0.107	0.327	0.434
				Page 1 of 105			

Begriffsbestimmung

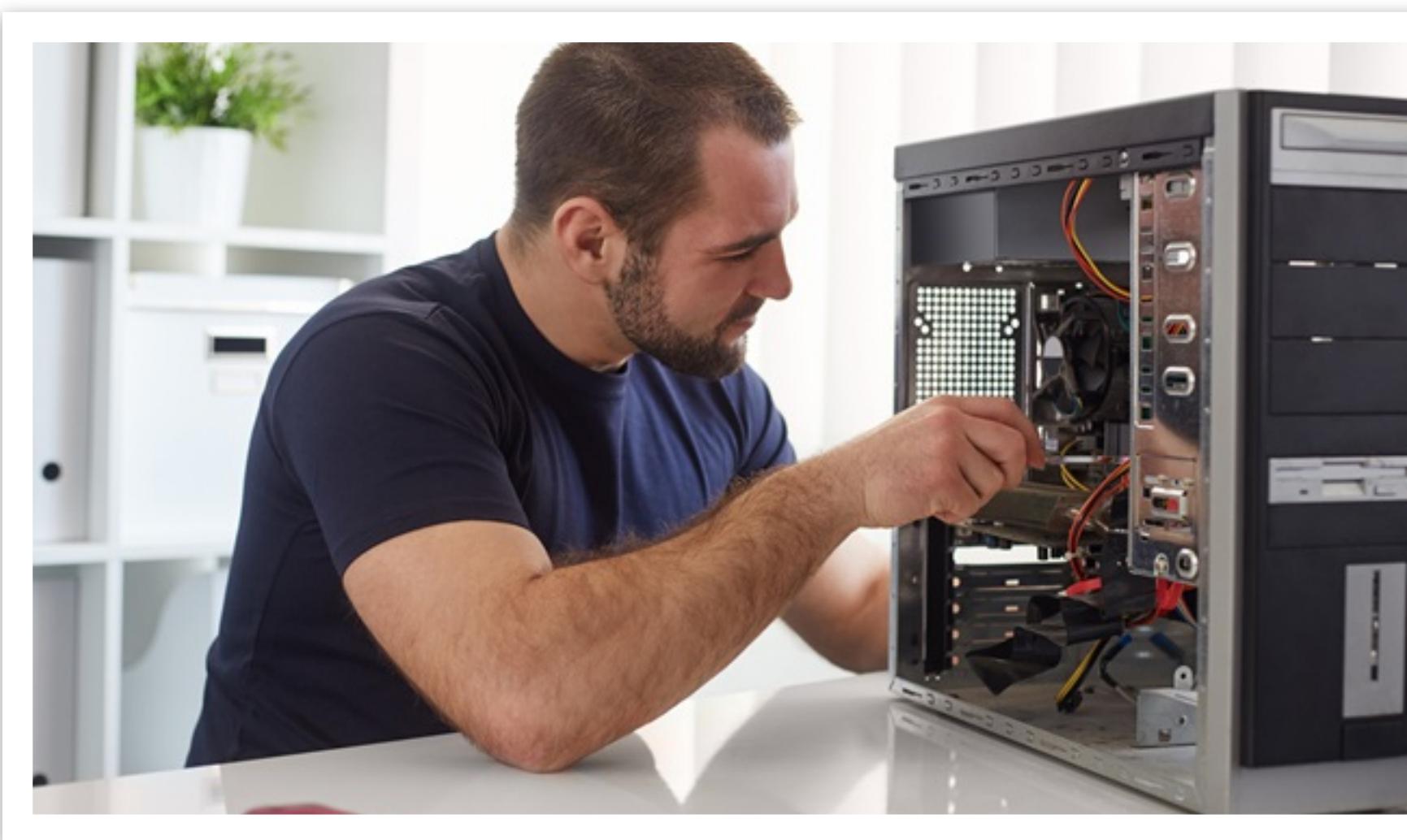
The right expressions help to conceptualize...

- **Beacon:** The protocol/API, with framework and default model
- **beacon:** Implementation of Beacon
 - using the Beacon v2 framework & supporting at minimum boolean responses
 - suggested support of Beacon v2 default model but can choose other
- Beacon **Aggregator:** service distributes queries to beacons and aggregates responses into a single Beacon response
 - potential to liftover genomes, remap filtering terms, translate between protocol versions...
 - entry point to or potentially itself node in a ...
- Beacon **Network:** Set of beacons with shared entry point for distributed queries and aggregated response delivery
 - "true" beacon networks should have managed aspects - scope, term use...
 - networks may combine mixes of internal (protected, rich data, additional extensions...) and external interfaces



Beacon v2 deployment

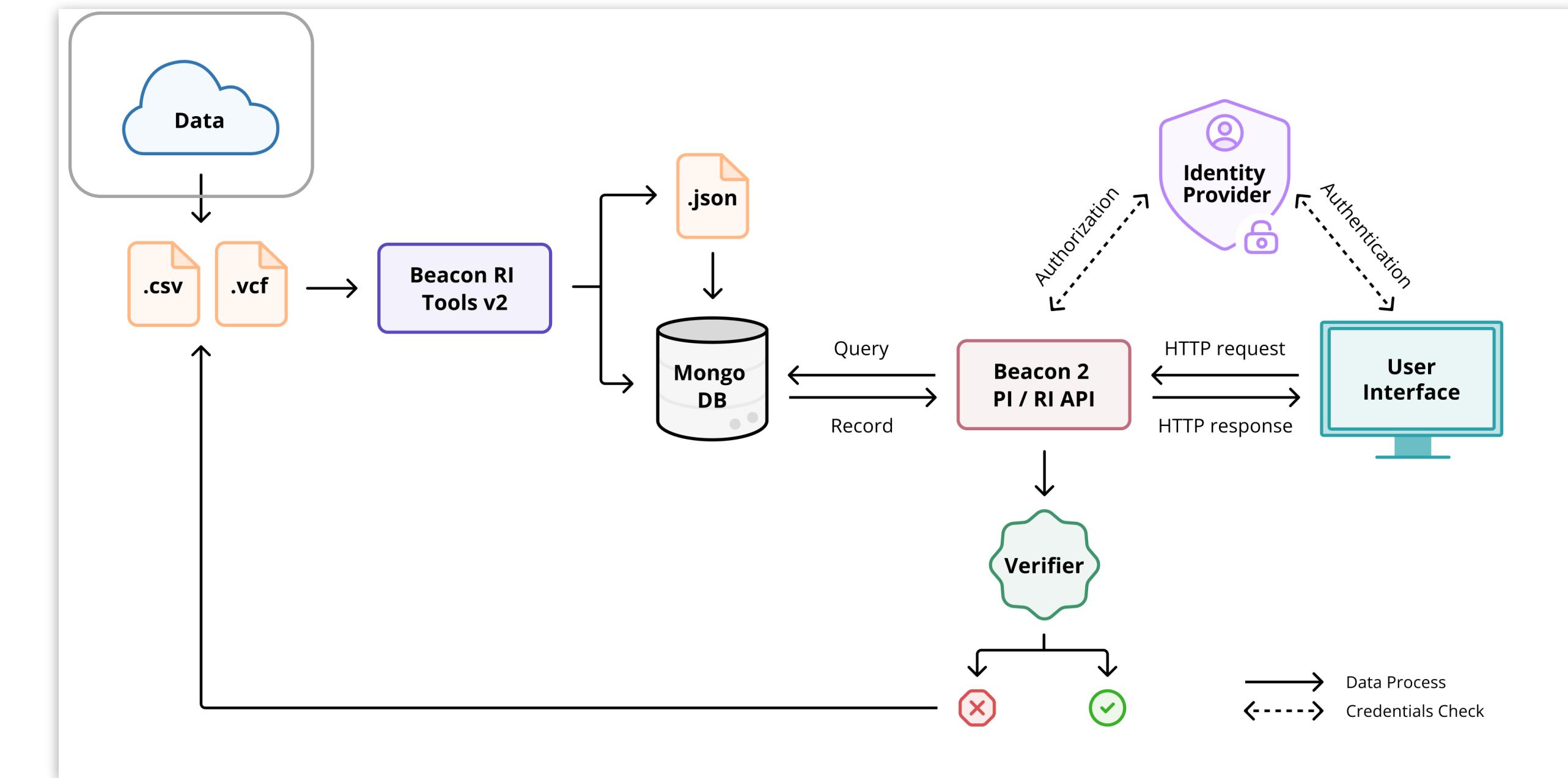
Build it yourself



Beacon v2 API

<https://github.com/ga4gh-beacon/beacon-v2>

Toolkit for production environments



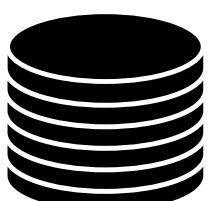
Beacon v2 Production Implementation (released Oct 2024)

<https://github.com/ga4gh-beacon/beacon-v2>

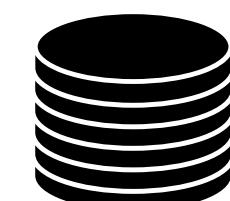
bycon based Progenetix Stack



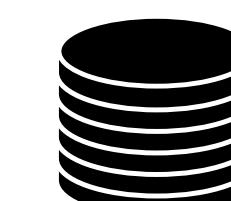
- JavaScript front-end is populated for query results using asynchronous access to multiple handover objects
 - ▶ biosamples and variants tables, CNV histogram, UCSC .bed loader, .pgxseg variant downloads...
- the complete middleware / CGI stack is provided through the *bycon* package
 - ▶ schemas, query stack, data transformation (Phenopackets generation)...
- data collections mostly correspond to the main Beacon default model entities
 - ▶ no separate *runs* collection; integrated w/ analyses
 - ▶ *variants* are stored per observation instance



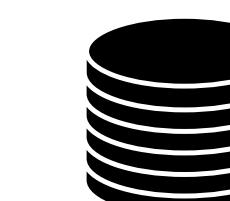
variants



analyses



biosamples



individuals



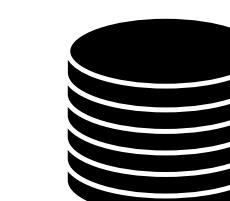
github.com/progenetix/bycon



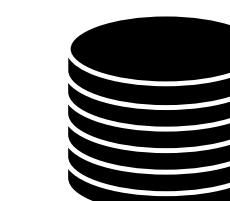
Entity collections



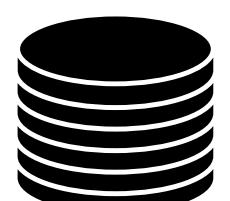
Utility collections



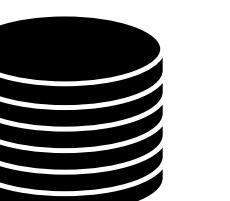
collations



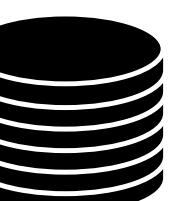
geolocs



genespans publications



publications



qBuffer

- *collations* contain pre-computed data (e.g. CNV frequencies, statistics) and information for all grouping entity instances and correspond to **filter values**
 - ▶ PMID:10027410, NCIT:C3222, pgx:cohort-TCGA, pgx:icdom-94703...
- *querybuffer* stores id values of all entities matched by a query and provides the corresponding access handle for **handover** generation

```
_id: ObjectId("6249bb654f8f8d67eb94953b"),
id: '0765ee26-5029-4f28-b01d-9759abf5bf14',
source_collection: 'variants',
source_db: 'progenetix',
source_key: '_id',
target_collection: 'variants',
target_count: 667,
target_key: '_id',
target_values: [
  ObjectId("5bab578b727983b2e00ca99e"),
  ObjectId("5bab578d727983b2e00cb505")]
```

Cancer Genomics Reference Resource

- **open** resource for oncogenomic profiles
- over **140'000 cancer CNV profiles**
- SNV data for some series (e.g. TCGA)
- more than **900 diagnostic types**
- inclusion of reference datasets (e.g. TCGA)
- standardized encodings (e.g. NCIIt, ICD-O 3)
- identifier mapping for PMID, GEO, Cellosaurus, TCGA, cBioPortal where appropriate
- core clinical data (TNM, sex, survival ...)
- data mapping services



Cancer CNV Profiles

ICD-O Morphologies
ICD-O Organ Sites
Cancer Cell Lines
Clinical Categories

Search Samples

arrayMap
TCGA Samples
1000 Genomes
Reference Samples
DIPG Samples
cBioPortal Studies
Gao & Baudis, 2021

Publication DB

Genome Profiling
Progenetix Use

Services

NCIt Mappings
UBERON Mappings

Upload & Plot

Beacon⁺

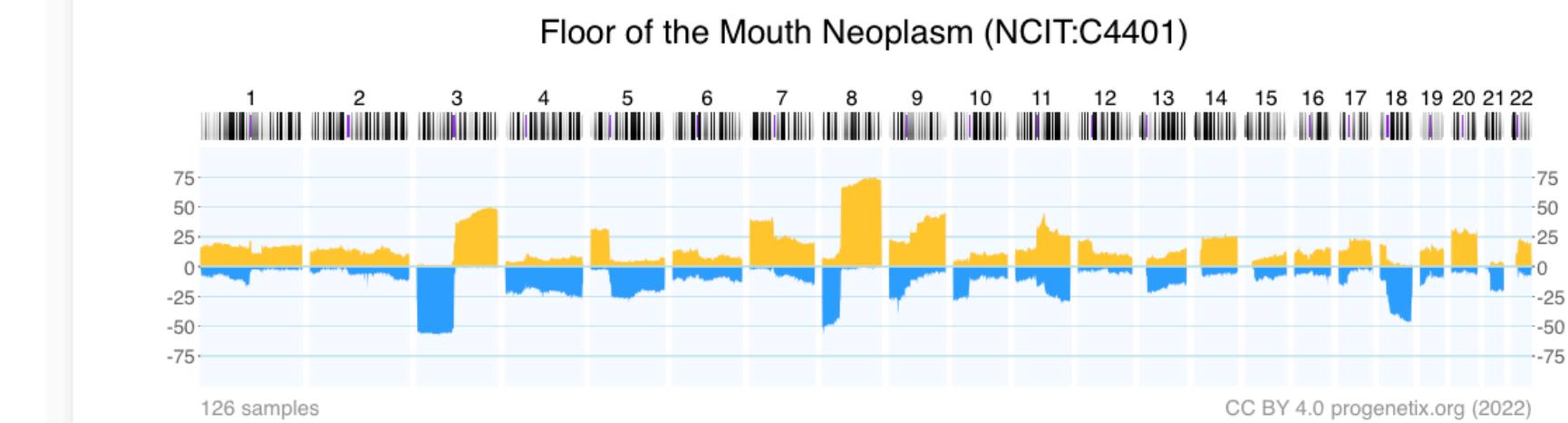
Documentation

News
Downloads & Use
Cases
Sevices & API

Baudisgroup @ UZH

Cancer genome data @ progenetix.org

The Progenetix database provides an overview of mutation data in cancer, with a focus on copy number abnormalities (CNV / CNA), for all types of human malignancies. The data is based on *individual sample data* from currently **142063** samples.



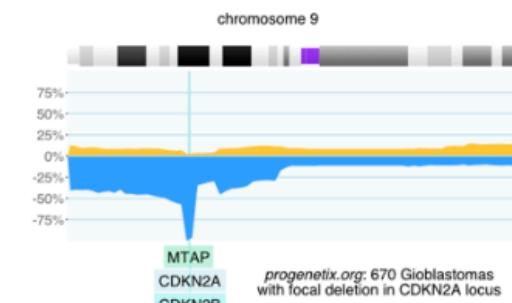
[Download SVG](#) | [Go to NCIT:C4401](#) | [Download CNV Frequencies](#)

Example for aggregated CNV data in 126 samples in Floor of the Mouth Neoplasm.
Here the frequency of regional **copy number gains** and **losses** are displayed for all 22 autosomes.

Progenetix Use Cases

Local CNV Frequencies

A typical use case on Progenetix is the search for local copy number aberrations - e.g. involving a gene - and the exploration of cancer types with these CNVs. The [\[Search Page \]](#) provides example use cases for designing queries. Results contain basic statistics as well as visualization and download options.



progenetix.org: 670 Glioblastomas with local deletion in CDKN2A locus

Cancer CNV Profiles

The progenetix resource contains data of **834** different cancer types (NCIt neoplasm classification), mapped to a variety of biological and technical categories. Frequency profiles of regional genomic gains and losses for all categories (diagnostic entity, publication, cohort ...) can be accessed through the [\[Cancer Types \]](#) page with direct visualization and options for sample retrieval and plotting options.

Cancer Genomics Publications

Through the [\[Publications \]](#) page Progenetix provides **4164** annotated references to research articles from cancer genome screening experiments (WGS, WES, aCGH, cCGH). The numbers of analyzed samples and possible availability in the Progenetix sample collection are indicated.

pgxRpi: an R/Bioconductor package

Client for Accessing Beaconized Data

- **Query and export variants**

https://progenetix.org/beacon/biosamples/pgxbs-kftvh94d/g_variants

```
> variants <- pgxLoader(type="variant",biosample_id="pgxbs-kftvh94d")
```

- **Query metadata of biosamples and individuals by filters (e.g. NCIt, PMID)**

<http://progenetix.org/services/sampletable/?filters=NCIT:C3697>

```
> biosamples <- pgxLoader(type="biosample",filters="NCIT:C3697")
```

- **Query and visualize CNV frequency by filters**

<http://www.progenetix.org/services/intervalFrequencies/?filters=NCIT:C3512>

```
> freq <- pgxLoader(type="frequency",output="pgxfreq",filter  
> pgxFreqplot(freq)
```

- **Process local .pgxseg files**

```
> info <- pgxSegprocess(file=file, show_KM_plot = T,  
return_seg = T, return_metadata = T, return_frequency = T)
```

pgxRpi

This is the **development** version of pgxRpi; for the stable release version, see [pgxRpi](#).

R wrapper for Progenetix

platforms all rank 2178 / 2266 support 0 / 0 in Bioc < 6 months build unknown updated < 1 month dependencies 137

DOI: [10.18129/B9.bioc.pgxRpi](https://doi.org/10.18129/B9.bioc.pgxRpi)

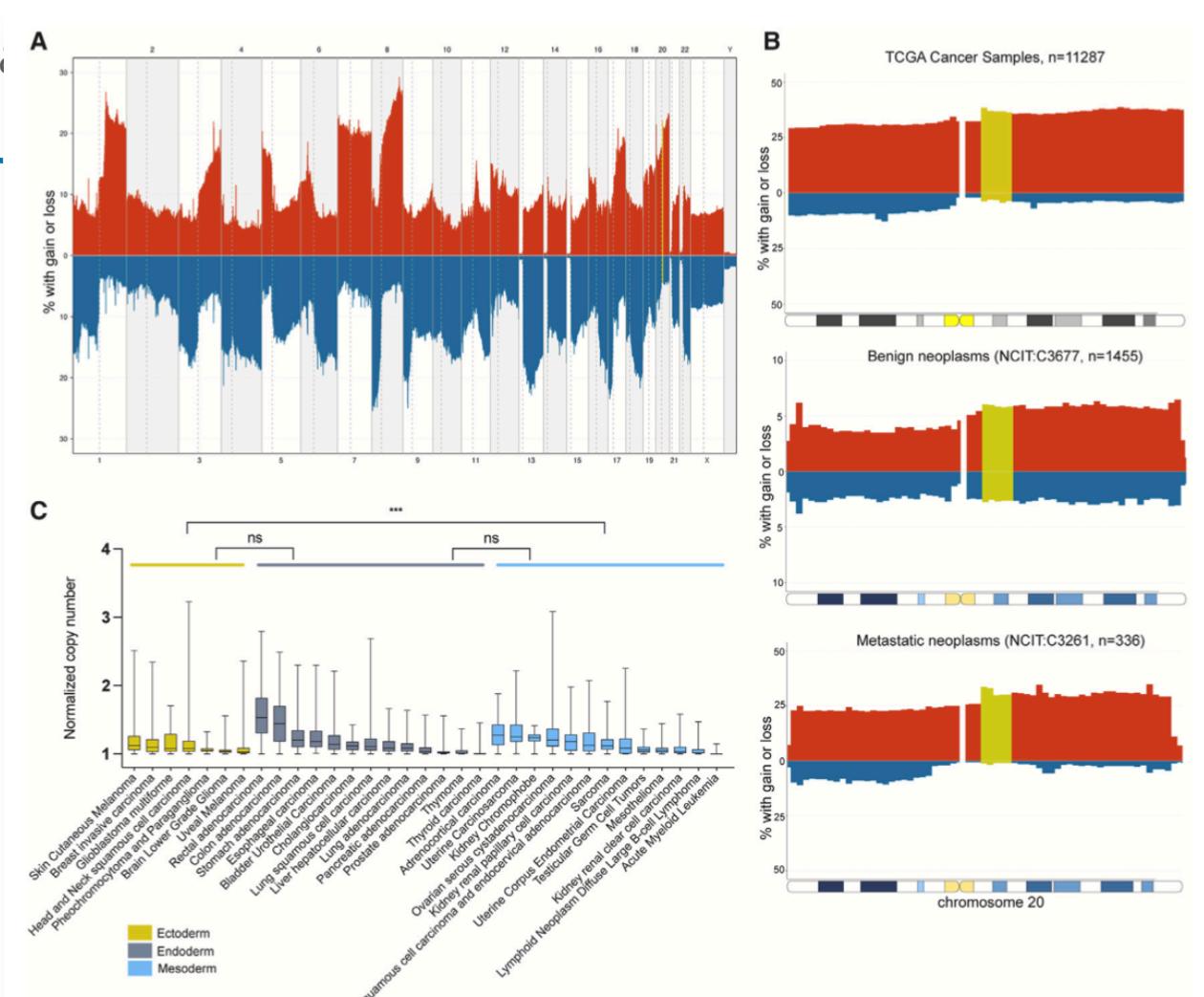
Bioconductor version: Development (3.20)

The package is an R wrapper for Progenetix REST API built upon the Beacon v2 protocol. Its purpose is to provide a seamless way for retrieving genomic data from Progenetix database—an open resource dedicated to curated oncogenomic profiles. Empowered by this package, users can effortlessly access and visualize data from Progenetix.

Author: Hangjia Zhao [aut, cre]  Michael Baudis [aut] 

Maintainer: Hangjia Zhao <hangjia.zhao at uzh.ch>

Use case: 2024 article using Progenetix' *pgxRpi* to retrieve & visualize 117'587 cancer CNV profiles for a study into pluripotent stem cells' genomics



Stem Cell Reports Review



OPEN ACCESS

Gains of 20q11.21 in human pluripotent stem cells: Insights from cancer research

Nuša Krivec,^{1,2} Manjusha S. Ghosh,^{1,2} and Claudia Spits^{1,2,*}

¹Research Group Reproduction and Genetics, Faculty of Medicine and Pharmacy, Vrije Universiteit Brussel, Brussels, Laarbeeklaan 103, 1090 Brussels, Belgium

²These authors contributed equally

*Correspondence: claudia.spits@vub.be

<https://doi.org/10.1016/j.stemcr.2023.11.013>

Figure 2. Copy-number alterations of human chromosome 20q11.21 in cancers

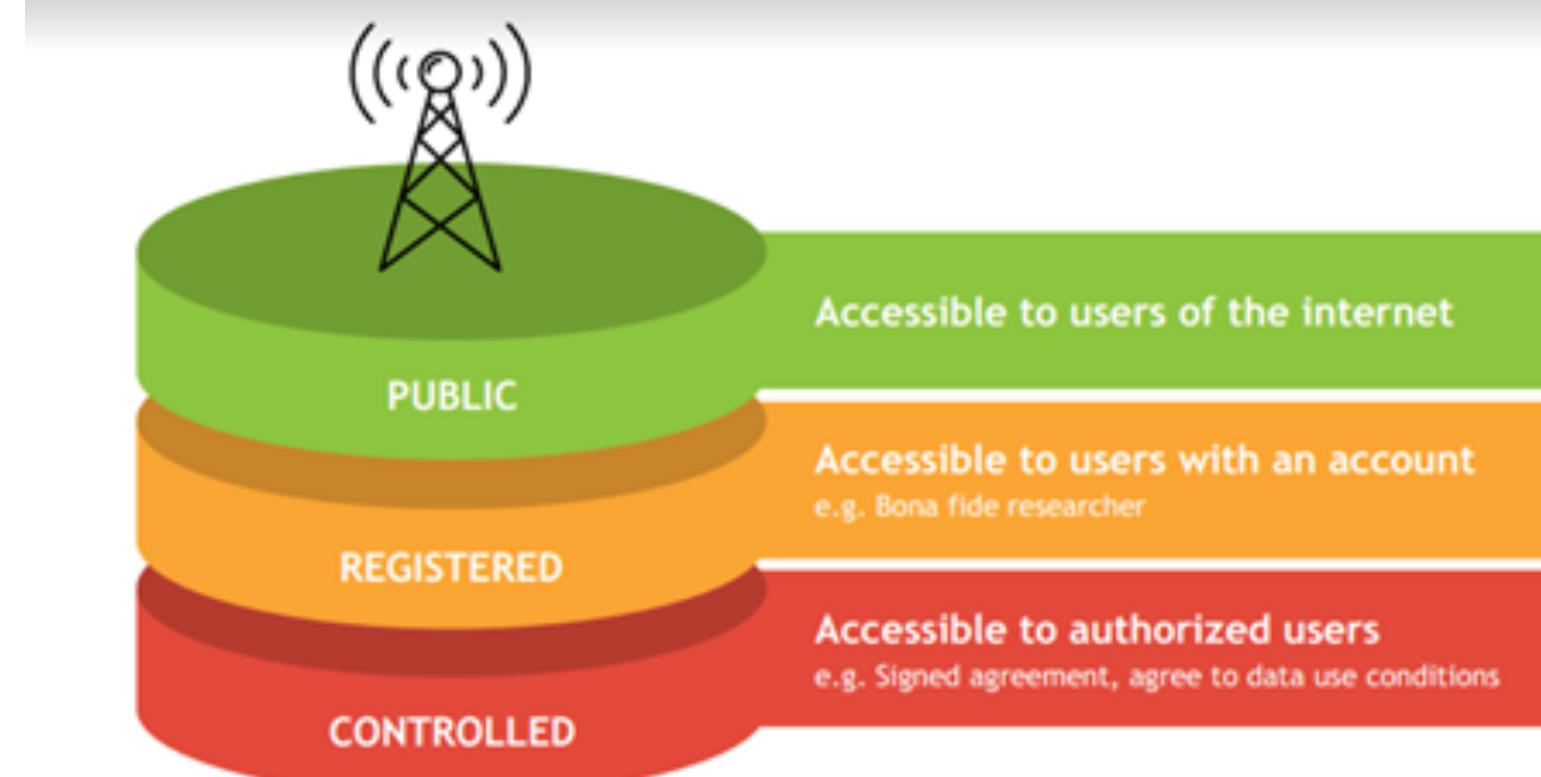
(A) Aggregated copy-number variation (CNV) data of 117,587 neoplasms (NCIT: C3262) from the Progenetix database (Huang et al., 2021) were plotted using R library pgxRpi. The percentage of samples with aberrations (red, gain; blue, loss) for the whole chromosome are indicated on the y axis. Chromosomal regions are depicted on the x axis; the minimal region of interest at chr20:31216079–35871578 is marked in moss green. NCIT, National Cancer Institute Thesaurus.

(B) Top to bottom: Aggregated CNV data of 11,287 TCGA cancer samples, 336 metastatic neoplasms (NCIT: C3261), and 1,455 benign neoplasms (NCIT: C3677) from the Progenetix database (Huang et al., 2021), respectively, were plotted using R library pgxRpi. The percentage of samples with aberrations (red, gain; blue, loss) for the whole chromosome are indicated on the y axis. Chromosomal regions are depicted on the x axis; the minimal region of interest at chr20:31216079–35871578 is marked in moss green.

Beacon Security

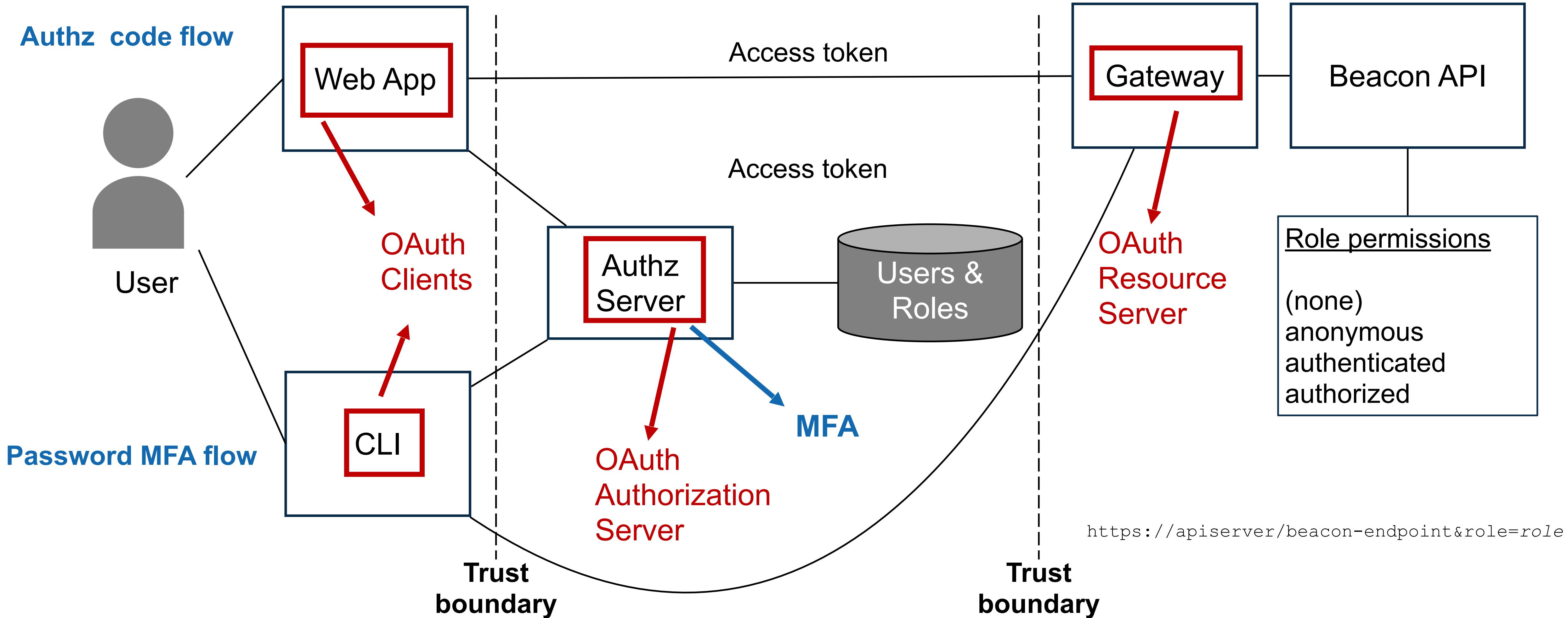
Security by Design ... if Implemented in the Environment

- the beacon API specification does not implement explicit security (e.g. checking user authentication and authorization)
- the framework implements different levels of response granularity which can be mapped to authorization levels (**boolean** / **count** / **record** level responses)
- implementations can have beacons running in secure environments with a **gatekeeper** service managing authentication and authorization levels, and potentially can filter responses for escalated levels
- the backend can implement additional access reduction, on a user <-> dataset level if needed



Architecture

Running the *bycon* stack in a secure environment

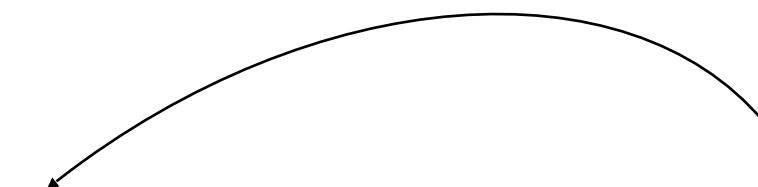




Beacon as a global standard



Beacon Scouts



Real-world needs

Cancer

Common diseases

Rare Diseases

...

- **Beacon Filters** – improve current filter solutions
- **Beacon Cohorts** – develop aggregated request and response (e.g. counts by sex and age)
- **Beacon Variants** – expand specification to cover new use cases and typed queries
- **Beacon Dev** – improve API (cleaning code, GitHub issues)
- **Beacon Matchmaking** – implementation in matchmaking use cases

Beacon Scouts

Finding the Paths to Beacon's Future

● Genomic Variation Scouts

- ➡ extension to the query model based on assessed needs
 - ▶ fusions/breakpoints, cytogenetic annotations, repeats, categorical variants...
- ➡ adoption of evolving VRS... standards for variant representation
 - ▶ adjacency, repeats...

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

GA4GH Beacon Genomic Variation Query Standards

Search GitHub elijah

Beacon VQS Requests

The `VQSRequest` type represents the generic collection of variant parameters supported in Beacon v2+ requests. These include parameters with close alignment to VRS v2 concepts and replacing some Beacon v1/v2 generics with tighter definitions (e.g. `referenceAccession` instead of `referenceName` and `accession` or `copyChange` for a specific subset of former `variantType` values) but also keep some concepts beyond VRS scope or specifically geared towards query applications (`geneId`, `sequenceLength`)

For the parameter definitions please see the [requestParameterComponents page](#).

VQSRequest Parameters

```
requestProfileId: ./requestParameterComponents.yaml#/defs/RequestProfileId
referenceAccession: ./requestParameterComponents.yaml#/defs/RefgetAccession
start: ./requestParameterComponents.yaml#/defs/SequenceStart
end: ./requestParameterComponents.yaml#/defs/SequenceEnd
sequence: ./requestParameterComponents.yaml#/defs/Sequence
copyChange: ./requestParameterComponents.yaml#/defs/CopyChange
adjacencyAccession: ./requestParameterComponents.yaml#/defs/AdjacencyAccession
adjacencyStart: ./requestParameterComponents.yaml#/defs/AdjacencyStart
adjacencyEnd: ./requestParameterComponents.yaml#/defs/AdjacencyEnd
repeatSubunitCount: ./requestParameterComponents.yaml#/defs/RepeatSubunitCount
repeatSubunitLength: ./requestParameterComponents.yaml#/defs/RepeatSubunitLength
geneId: ./requestParameterComponents.yaml#/defs/GeneId
aminoacidChange: ./requestParameterComponents.yaml#/defs/AminoacidChange
genomicAlleleShortForm:
./requestParameterComponents.yaml#/defs/GenomicAlleleShortForm
sequenceLength: ./requestParameterComponents.yaml#/defs/SequenceLength
vrsType: ./requestParameterComponents.yaml#/defs/VRStype
```

<https://genomebeacons.org/variant-query-types/variant-scouts-home/>

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Copy number gains involving the whole locus chr2:54,700,000-63,900,000
Focal high-level deletion involving the CDKN2A locus
Find t(8;14)(q24;q32) translocations
CAG repeat in the first exon of the huntingtin gene (HTT)
CAG repeat in the first exon of the huntingtin gene (HTT)
CGG trinucleotide repeat expansion in the FMR1 gene
Query for a focal deletion involving TP53

Progenetix Cancer Genomics Beacon+

/api

Beacon+ provides a forward looking implementation of the Beacon v2 API, with focus on structural genome variants and metadata based on the cancer and reference genome profiling data in the Progenetix resource (<https://progenetix.org>).

This page presents a prototype for an OpenAPI (Swagger) definition for the [GA4GH Beacon API](#). The definitions are generated from the `entity_defaults` and `argument_definitions` in the [bycon project](#). The complete. Please be aware that the whole capabilities of the project cannot be represented solely through the OpenAPI definitions and also involve features such as filtering terms logic and result aggregation entities. Additionally, the bycon project implements a number of data services beyond Beacon standards which again are only partially covered here.

bycon and Data Aggregation

The Beacon standard implements a REST style syntax - e.g. consistent id-based document retrieval for entities indicated through their framework provide full data aggregation; i.e. queries with parameters against **any** of the main data entities (g_variants, runs, analyses, intersection of the query results at the level of the response entity).

[Beacon v2 API] | [Example: Bv2 CNV / bracket] | [Proposal: VQS CNV / bracket] | [Proposal: VQS CNV by Gene ID] | [Proposal: VQS CNV by Reference Name]

Contact the developer

Servers

<https://progenetix.org>

Beacon

[GET /beacon/info](#) Get info entries

[GET /beacon/datasets](#) Get dataset entries

[GET /beacon/cohorts](#) Get cohort entries

[GET /beacon/g_variants](#) Get genomicVariant entries

[GET /beacon/g_variants/{id}](#) Get genomicVariant entries

[GET /beacon/g_variants/{id}/analyses](#) Get analysis entries

[GET /beacon/g_variants/{id}/biosamples](#) Get biosample entries

[GET /beacon/g_variants/{id}/individuals](#) Get individual entries

[GET /beacon/analyses](#) Get analysis entries

[GET /beacon/analyses/{id}](#) Get analysis entries

[GET /beacon/analyses/{id}/g_variants](#) Get genomicVariant entries

[GET /beacon/analyses/{id}/biosamples](#) Get biosample entries

Bv2minimalAlleleRequest

[GET /beacon/g_variants](#) Get genomicVariant entries

Parameters

Name Description

referenceName string (query)

Examples: Chromosome 17

17

start array<integer> (query)

Examples: Base position on chromosome 17

7577120

Add integer item

alternateBases string (query)

Examples: An 'A' allele at the specified position

A

referenceBases string (query)

Examples: A reference 'G' allele at the specified position

G

skip integer (query)

skip

limit integer (query)

limit

requestedGranularity string (query)

Examples: The minimal boolean response

boolean

Bv2cnvbracketquery

[GET /beacon/g_variants](#) Get genomicVariant entries

Get genomicVariant entries

Parameters

Name Description

filters array<string> (query)

Examples: Glioblastoma

NCIT:C3058

Add string item

referenceName string (query)

Examples: Chromosome 9 (GRCh38)

refseq:NC_000009.12

start array<integer> (query)

Examples: Range for start of CNV involving CDKN2A

21000001

21975098

Add integer item

end array<integer> (query)

Examples: Range for end of CNV involving CDKN2A

21967753

23000000

Add integer item

variantType string (query)

Examples: High-level copy number loss

EFO:0020073

VQSadjacencyRequest

[GET /beacon/g_variants](#) Get genomicVariant entries

[GET /beacon/analyses](#) Get analysis entries

[GET /beacon/biosamples](#) Get biosample entries

Get biosample entries

Parameters

Name Description

filters array<string> (query)

Examples: Malignant lymphoma, NOS (ICD-O 3 code 9680/3)

pgx:icd0-95903

Add string item

referenceAccession string (query)

Examples: RefSeq ID for Chromosome 8 (GRCh38)

refseq:NC_000008.11

breakpointRange array<integer> (query)

Examples: Range for band q24 on chromosome 8

11670000

145138636

Add integer item

adjacencyAccession string (query)

Examples: RefSeq ID for Chromosome 14 (GRCh38)

refseq:NC_000014.9

adjacencyRange array<integer> (query)

Examples: Range for band q32 on chromosome 14

89300000

107043718

Add integer item

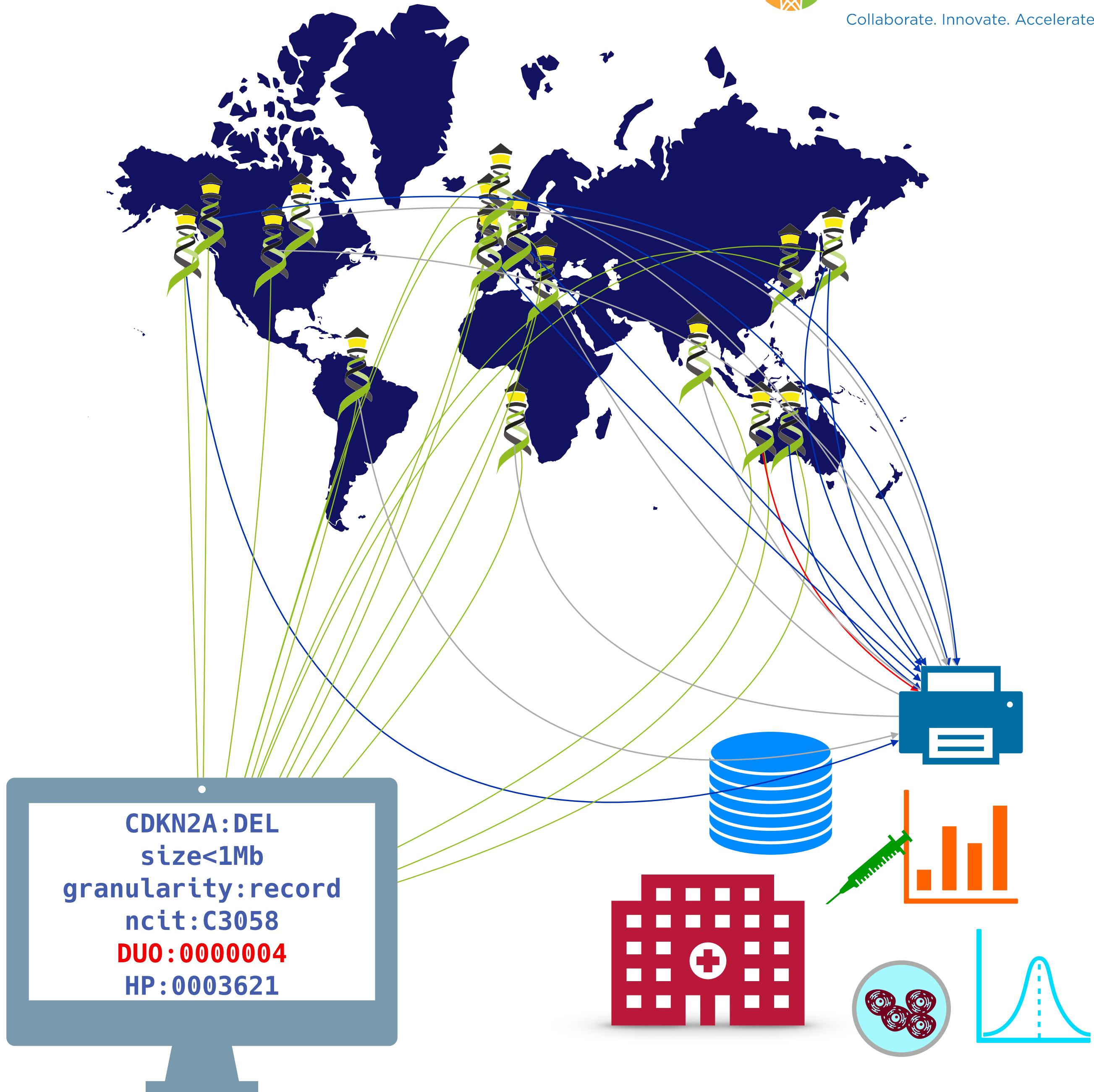
vrsType string (query)

Examples: Adjacency

Adjacency

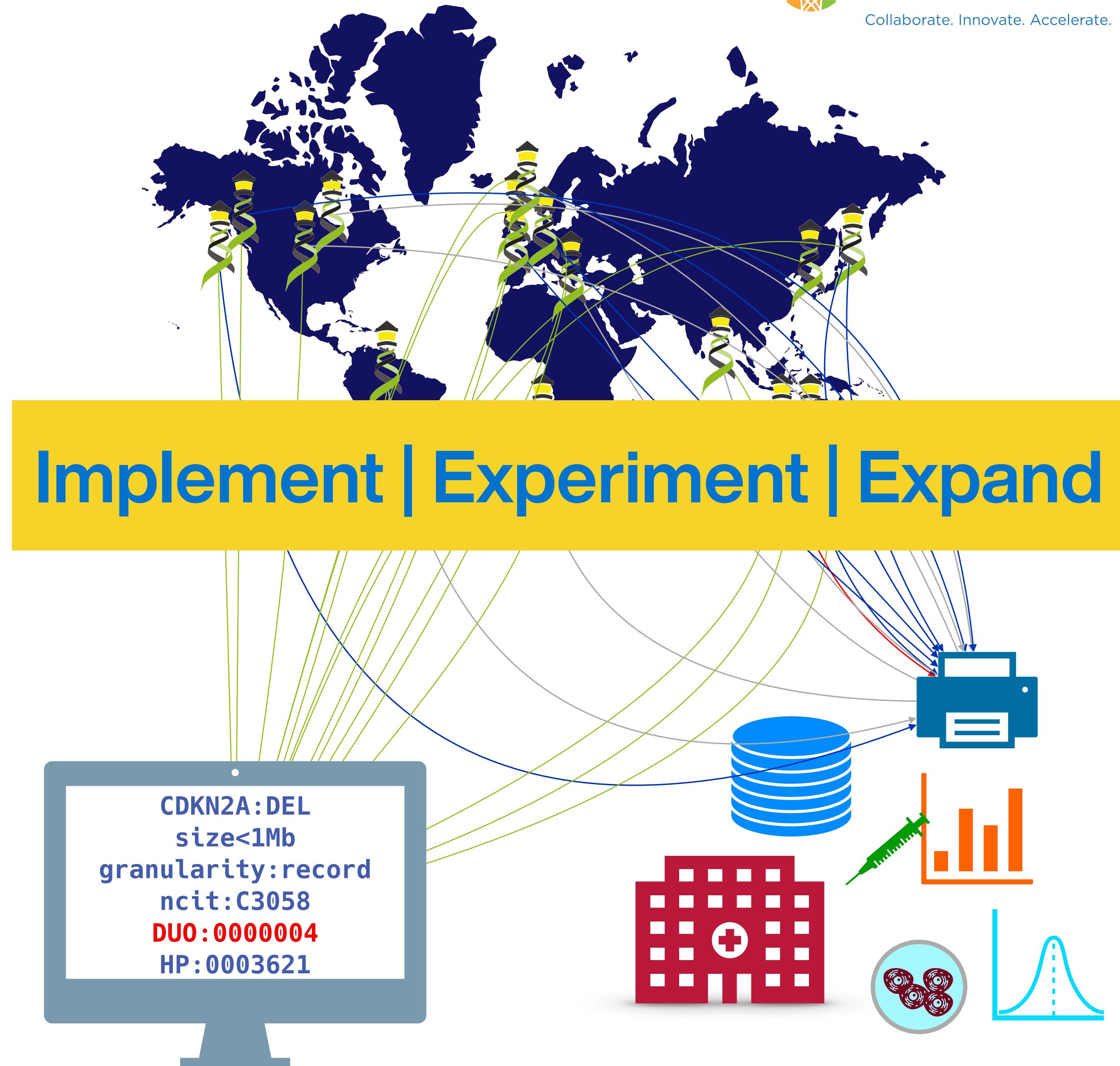
What Can You Do?

- find a way to make your (patients') **data discoverable** - through adding at least the relevant metadata to national or project centric repositories
- use forward looking consent and data protection models (**ORD** principle "as secure as necessary, as open as possible")
- **support** and/or get involved with international **data standards** efforts and projects



Beacon for Genomic Discovery Proxies

- Feature beacons for privacy protecting data discovery
 - privacy protection through aggregated data, cohorts
 - alternative is "**horizontal gatekeeping**": separate Beacons for **discovery** of e.g. genomic and phenotypic data and **data delivery** upon request / authentication
 - We'd love to help launching your beacon (especially as a **bycon**...)



Save the dates!



Global Alliance
for Genomics & Health

April Connect 2025

1 to 4 April 2025

Broad Institute, Cambridge, USA

[Registration Open Now](#)



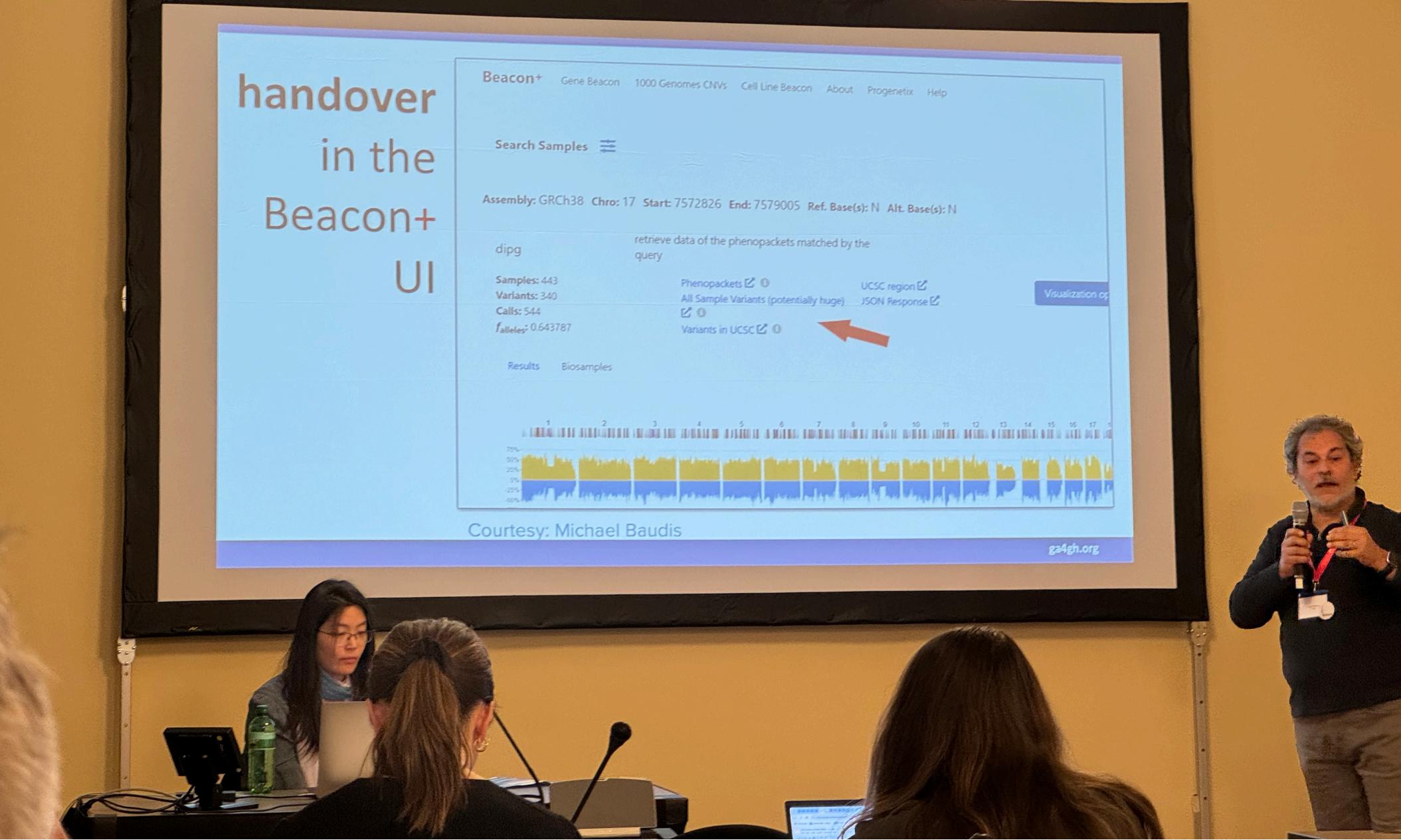
13th Plenary

6 to 10 October

UKK, Uppsala, Sweden

Registration Opening Soon





The Global Alliance for Genomics and Health (GA4GH) gathered for the 2024 [April Connect meeting](#) in Ascona, Switzerland and online from 21 to 24 April. The GA4GH Connect meetings provide an opportunity for contributors to advance the GA4GH Road Map, showcase GA4GH standards and policies in action, and gather feedback on product development and community needs. The meeting brought together 103 in-person attendees and 312 virtual attendees for updates from Work Streams and Driver Projects, breakout sessions, and themed events.





University of
Zurich UZH



Michael Baudis

Hangjia Zhao

Ziying Yang

Ramon Benitez

Brito

Rahel Paloots

Bo Gao

Qingyao Huang



Jordi Rambla

Arcadi Navarro

Roberto Ariosa

Manuel Rueda

Lauren Fromont

Mauricio Moldes

Claudia Vasallo

Babita Singh

Sabela de la Torre

Fred Haziza



Augusto Rendon
Ignacio Medina
Javier Lopez
Jacobo Coll
Antonio Rueda

Swiss Institute of
Bioinformatics

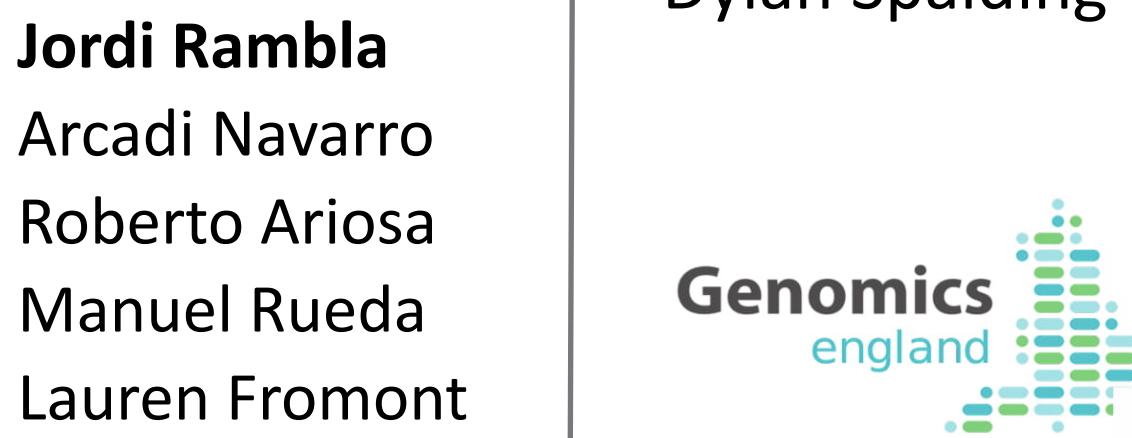
Tony Brookes

Tim Beck
Colin Veal

Tom Shorter



Juha Tornroos
Teemu Kataja
Ilkka Lappalainen
Dylan Spalding



Augusto Rendon
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Giselle Kerry
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H3Africa
Human Heredity & Health in Africa

Nicola Mulder
Mamana
Mbiyavanga
Ziyaad Parker

EU CAN
CAN.
David
Torrents
Dean Hartley

AUTISM SPEAKS



Fundación Progreso y Salud
CONSEJERÍA DE SALUD

Joaquin Dopazo

Javier Pérez
J.L. Fernández
Gema Roldan

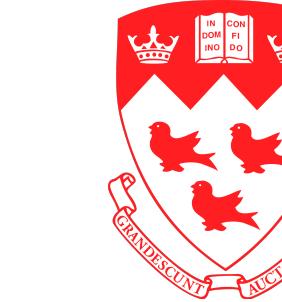
CINECA

Thomas Keane
Melanie Courtot
Jonathan Dursi

Heidi Rehm
Ben Hutton

GEM Japan
Toshiaki
Katayama

Diana Lemos



Stephane Dyke

DNA STACK
Marc Fiume
Miro Cupak



Melissa Cline



EMBL-EBI
Diana Lemos

EUROPEAN JOINT PROGRAMME
RARE DISEASES

VICC Variant Interpretation
for Cancer Consortium

GA4GH Phenopackets

Peter Robinson
Jules Jacobsen

GA4GH VRS
Alex Wagner
Reece Hart

Beacon PRC

Alex Wagner
Jonathan Dursi
Mamana Mbiyavanga
Alice Mann
Neerjah Skantharajah

elixir

The Beacon team through the ages