Data Resources, Sharing, Discovery in Biomedical Genetics and Cancer Genomics

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Professor of Bioinformatics
University of Zürich
Swiss Institute of Bioinformatics **SIB**
GA4GH Workstream Co-lead *DISCOVERY*
Co-lead ELIXIR Beacon API Development
Co-lead ELIXIR hCNV Community

Universität Zürich[UZH]

SIB Swiss Institute of Bioinformatics

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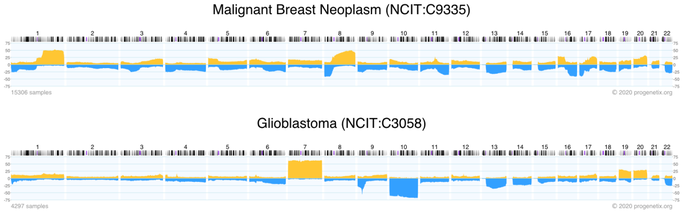
elixir

Theoretical Cytogenetics and Oncogenomics

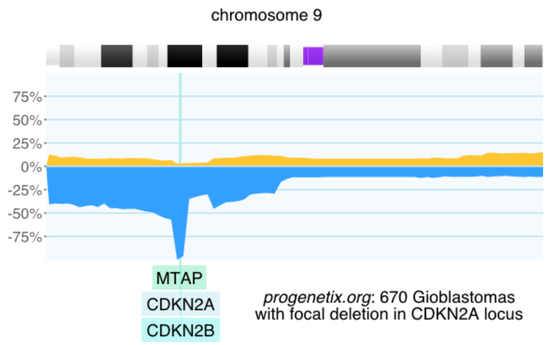
... but what does this entail @baudisgroup?

- patterns & markers in cancer genomics, especially somatic structural genome variants



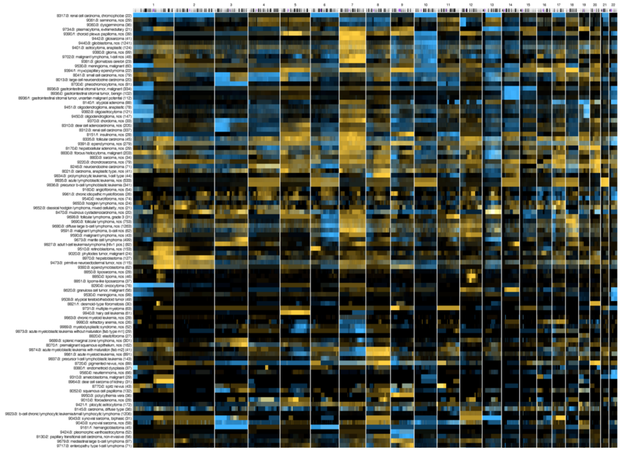
- bioinformatics support in collaborative studies
- reference resources for curated cancer genome variations
- bioinformatics tools & methods
- standards and reference implementations for data sharing in genomics and personalized health
- open research data "ambassadoring"

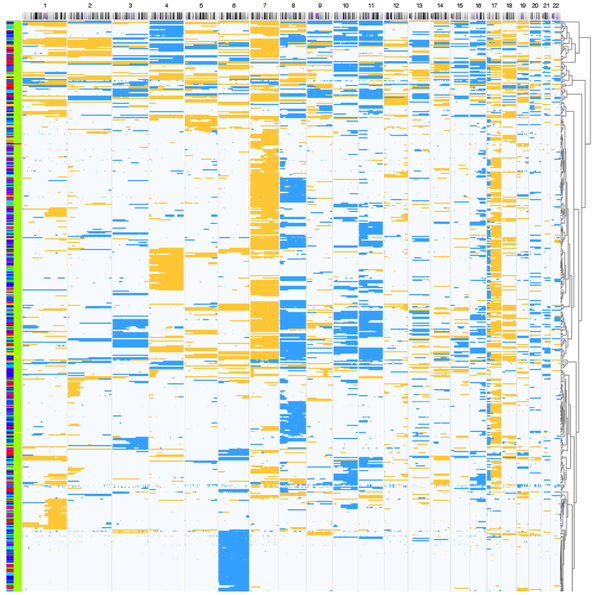
Theoretical Cytogenetics and Oncogenomics Research | Methods | Standards

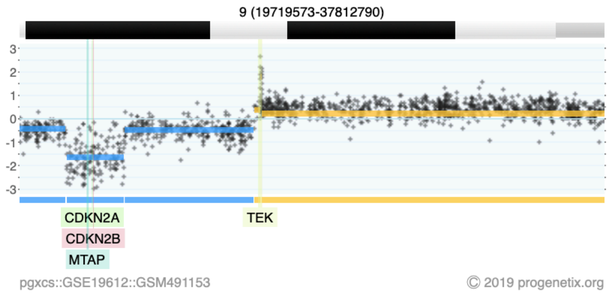


Genomic Imbalances in Cancer - Copy Number Variations (CNV)

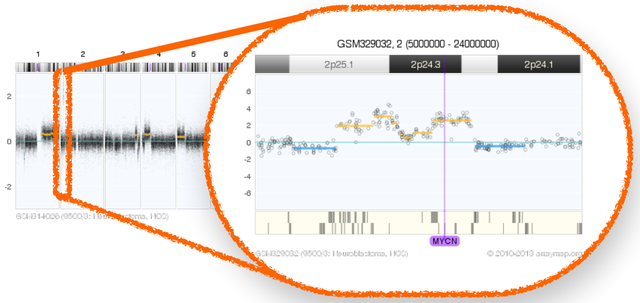
- Point mutations (insertions, deletions, substitutions)
- Chromosomal rearrangements
- **Regional Copy Number Alterations** (losses, gains)
- Epigenetic changes (e.g. DNA methylation abnormalities)







2-event, homozygous deletion in a Glioblastoma



MYCN amplification in neuroblastoma
(GSM314026, SJNB8_N cell line)

progenetix.org

Cancer Genomics Reference Resource

- ***open*** resource for oncogenomic profiles
- over **116'000 cancer CNV profiles**
- more than **800 diagnostic types**
- inclusion of reference datasets (e.g. TCGA)
- standardized encodings (e.g. NCIt, ICD-O 3)
- identifier mapping for PMID, GEO, Cellosaurus, TCGA, cBioPortal where appropriate
- core clinical data (TNM, sex, survival ...)
- data mapping services
- recent addition of SNV data for some series

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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+

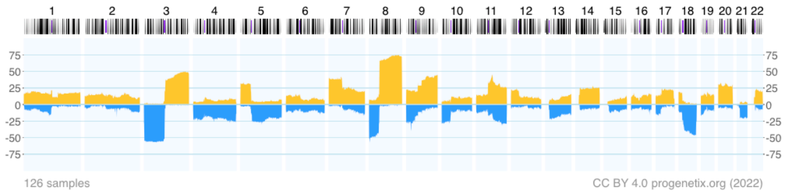
Documentation
- News
- Downloads & Use Cases
- Sevices & API

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.

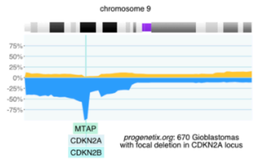
Floor of the Mouth Neoplasm (NCIT:C4401)



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.

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.

progenetix.org

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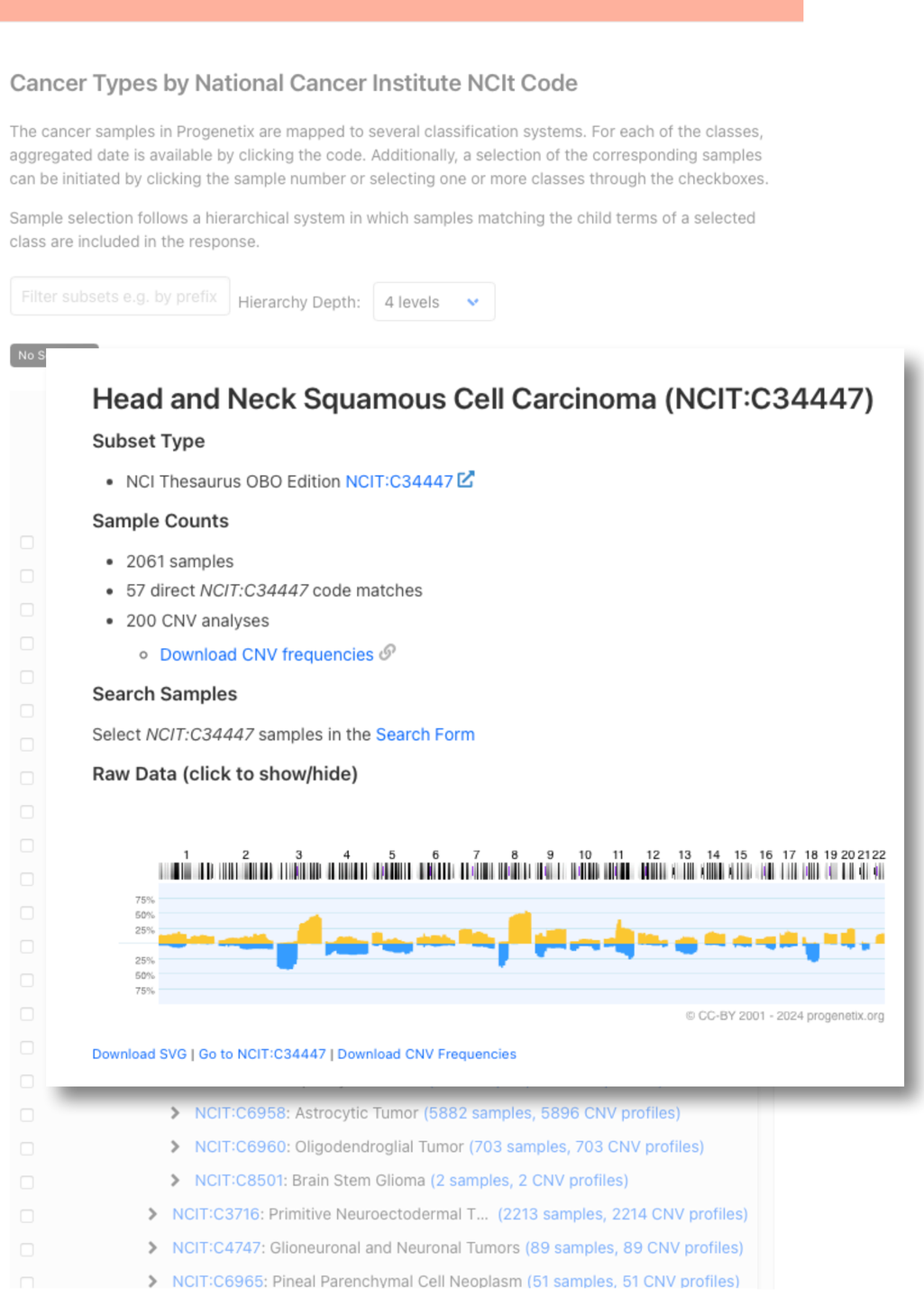

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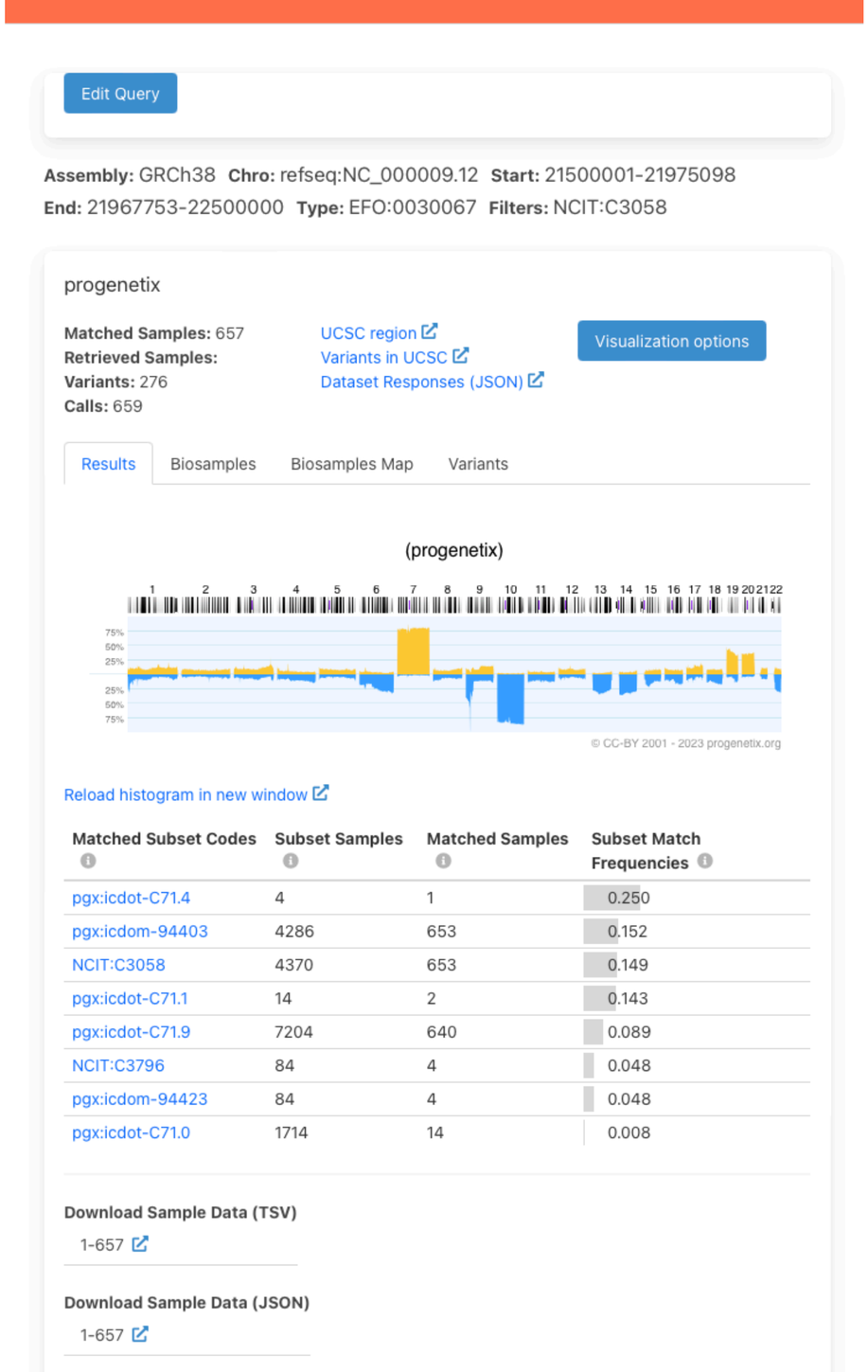

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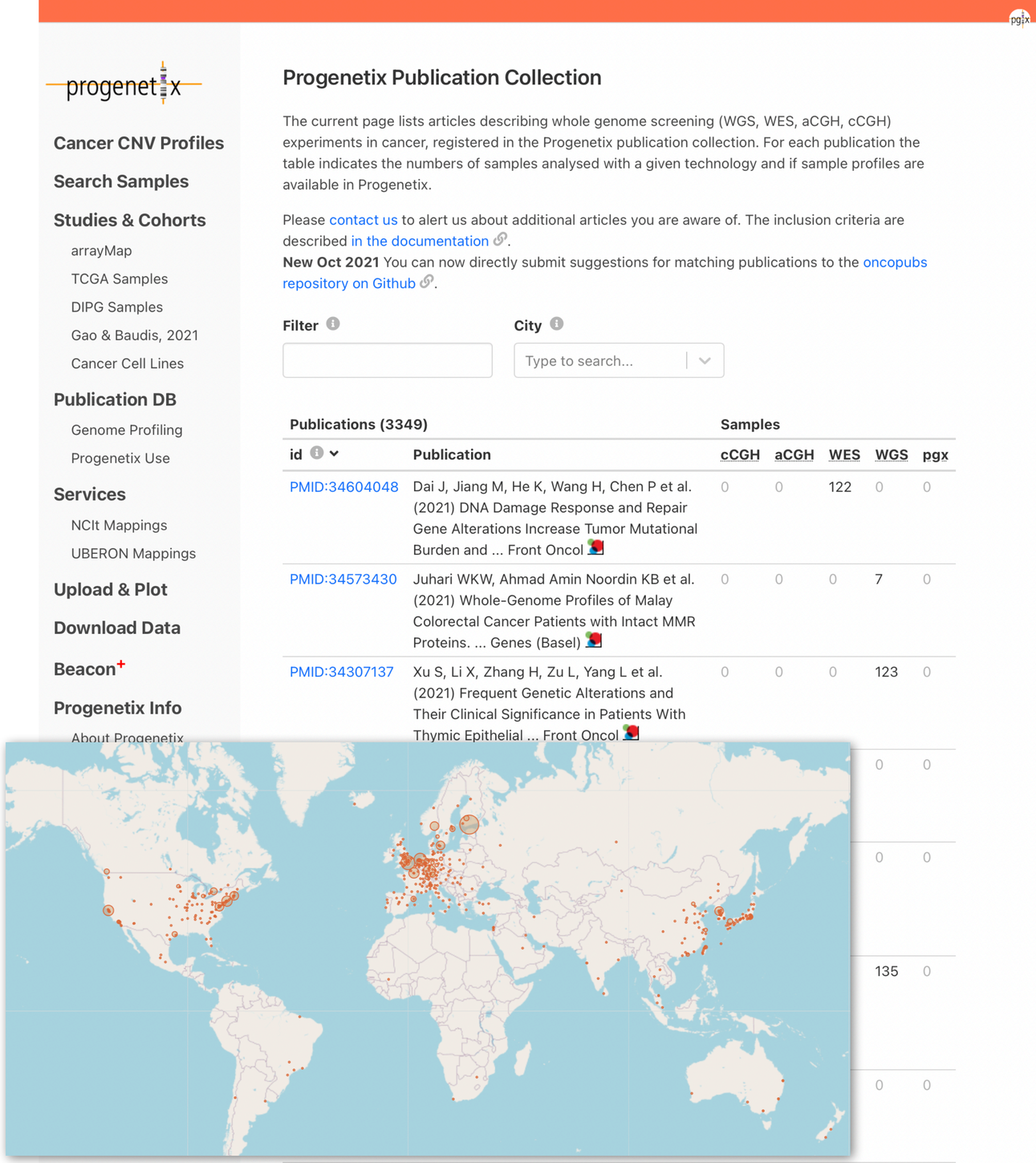

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Cancer Cell Lines

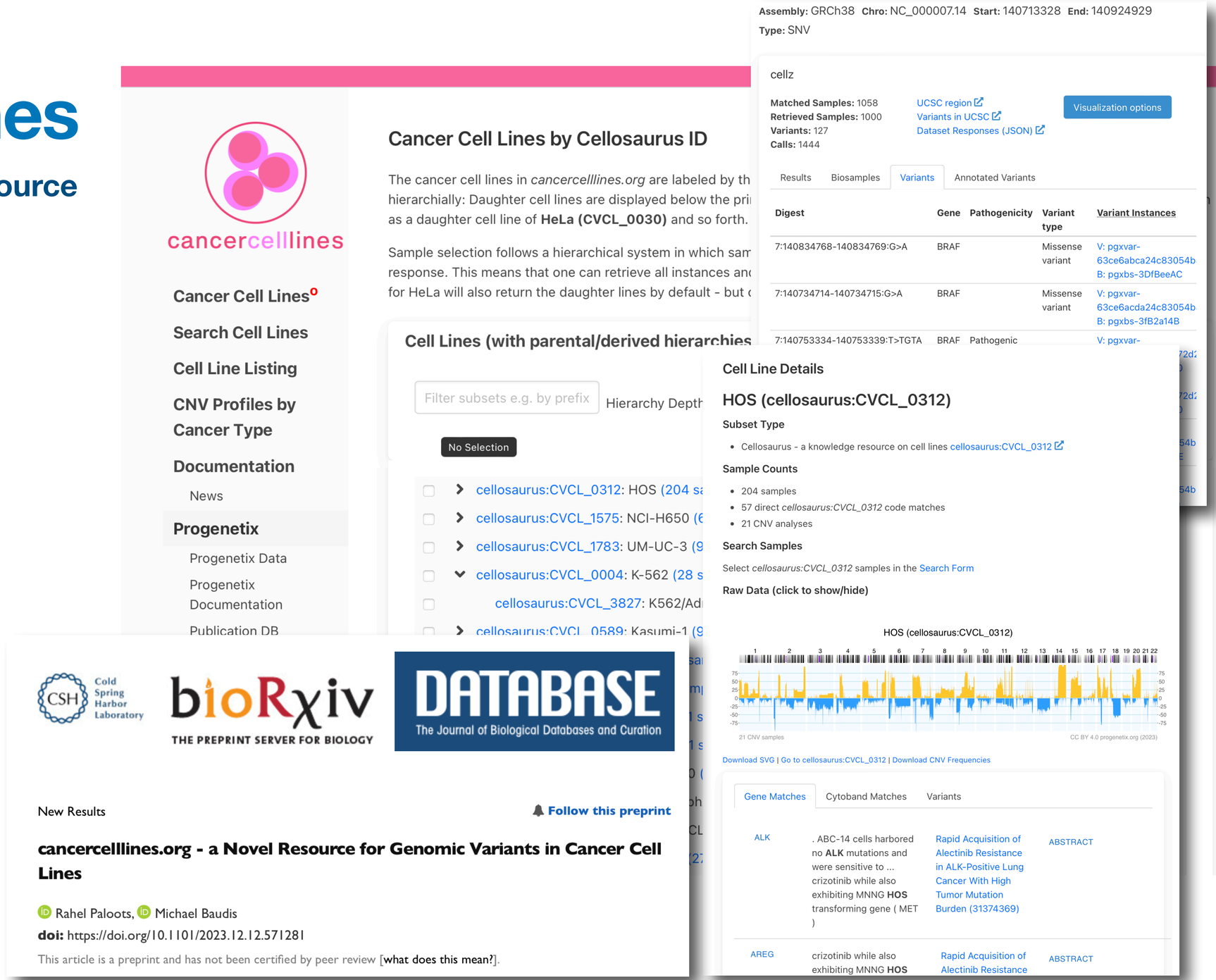
Cancer Genomics Reference Resource

- starting from >5000 cell line CNV profiles
  - 5754 samples | 2163 cell lines
  - 256 different NCIT codes
- genomic mapping of annotated variants and additional data from several resources (ClinVar, CCLE, Cellosaurus...)
  - 16178 cell lines
  - 400 different NCIT codes
- query and data delivery through Beacon v2 API

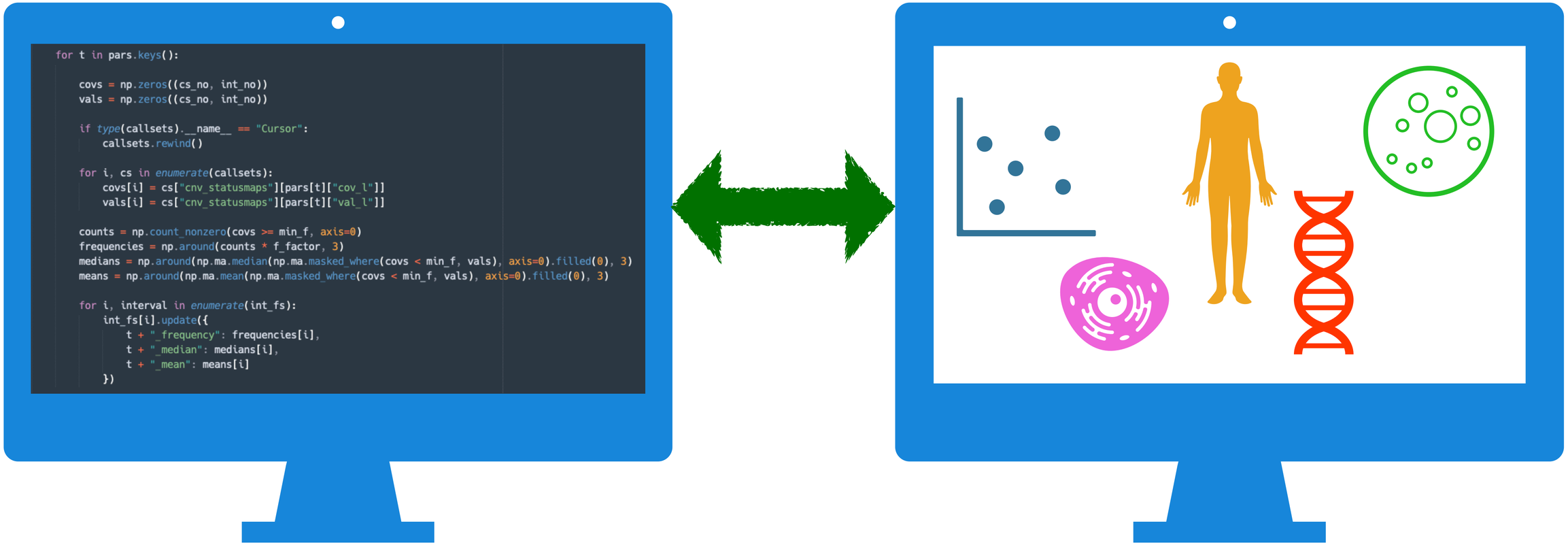
➡ integration in data federation approaches

cancercelllines.org

Lead: Rahel Paloots



{BioInformaticsScience}

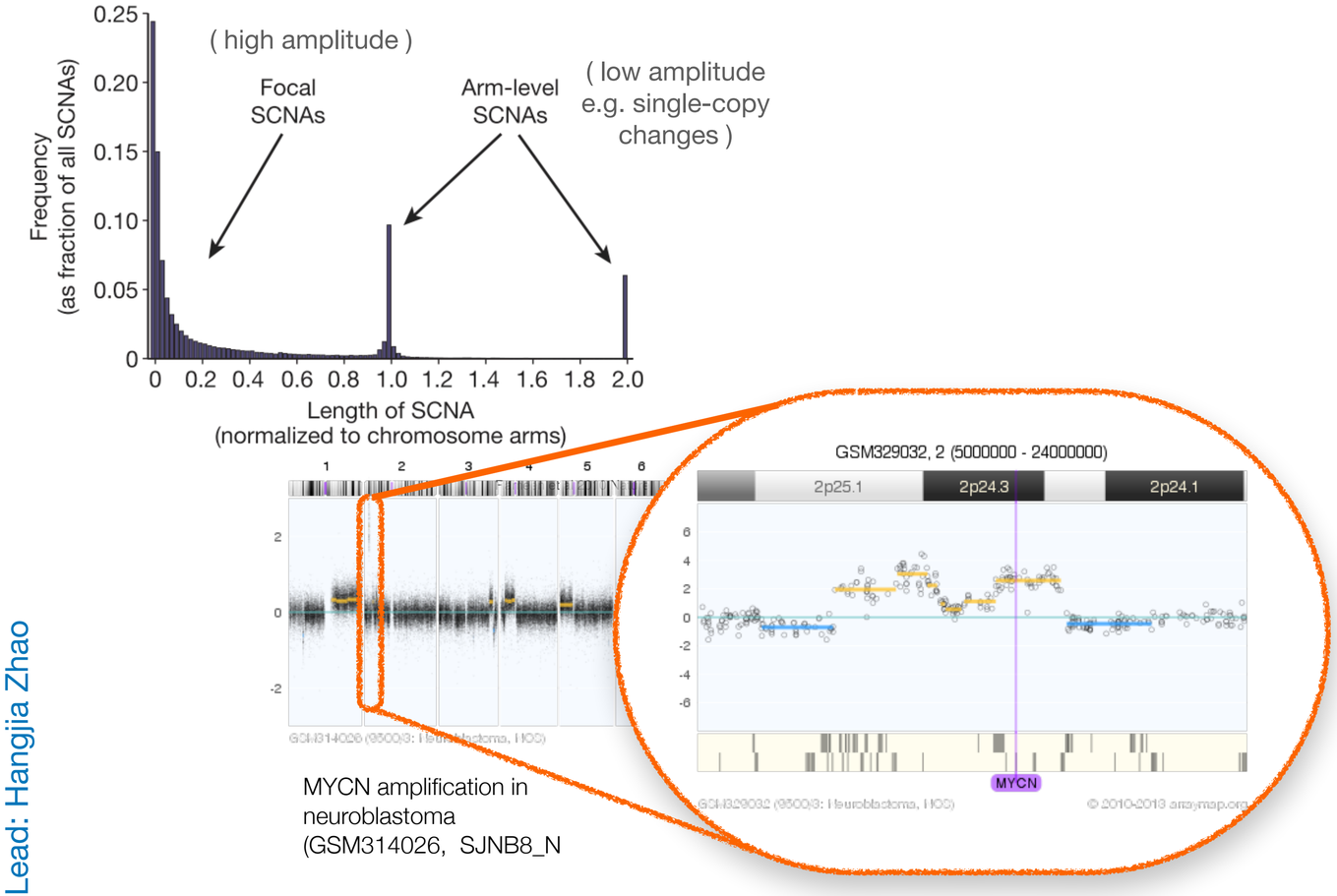


OXFORD

Briefings in Bioinformatics, 2024, **25(2)**, 1–12
https://doi.org/10.1093/bib/bbad541
Problem Solving Protocol

labelSeg: segment annotation for tumor copy number alteration profiles

Hangjia Zhao and Michael Baudis
Corresponding author: Michael Baudis, Department of Molecular Life Sciences, University of Zurich, Winterthurerstrasse 190, CH-8057 Zurich, Switzerland.
Tel.: (+41) 44 635 34 86; E-mail: michael.baudis@mls.uzh.ch



Lead: Hangjia Zhao

GA4GH Variation Representation Specification

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CopyNumberChange

Copy Number Change captures a categorization of copies of a molecule within a system, relative to a baseline. These types of Variation are common outputs from CNV callers, particularly in the somatic domain where integral CopyNumberCount are difficult to estimate and less useful in practice than relative statements. Somatic CNV callers typically express changes as relative statements, and many HGVS expressions submitted to express copy number variation are interpreted to be relative copy changes.

Computational Definition

An assessment of the copy number of a Location or a Feature within a system (e.g. genome, cell, etc.) relative to a baseline ploidy.

Information Model

Some CopyNumberChange attributes are inherited from Variation.

Field	Type	Limits	Description
_id	CURIE	0..1	Variation Id. MUST be unique within document.
type	string	1..1	MUST be “CopyNumberChange”
subject	Location | CURIE | Feature	1..1	A location for which the number of systemic copies is described.
copy_change	string	1..1	MUST be one of “efo:0030069” (complete genomic loss), “efo:0020073” (high-level loss), “efo:0030068” (low-level loss), “efo:0030067” (loss), “efo:0030064” (regional base ploidy), “efo:0030070” (gain), “efo:0030071” (low-level gain), “efo:0030072” (high-level gain).

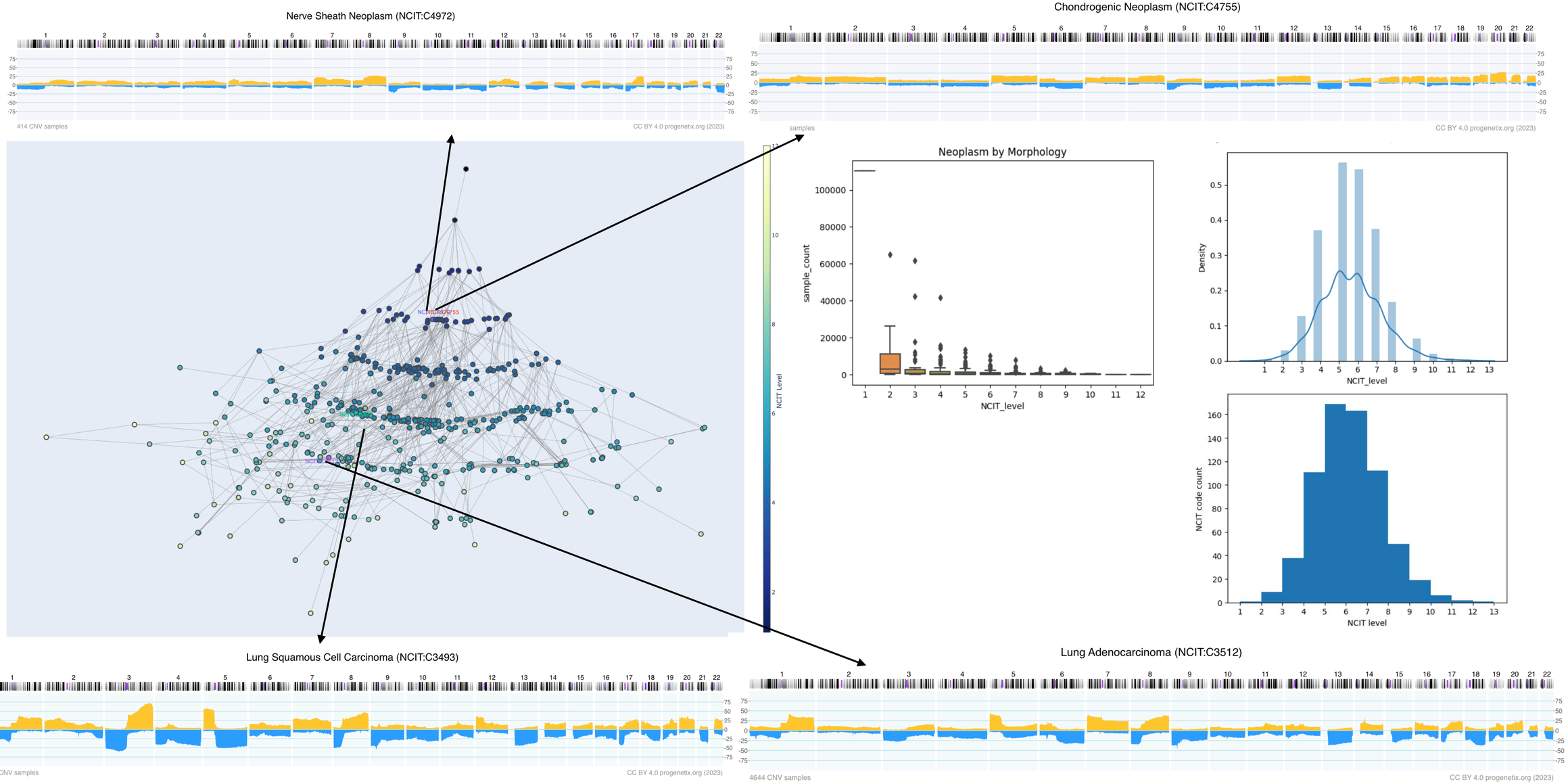
Tumor subpopulations can be matched with highly similar cell lines?!

Lead: Rahel Paloots

CNV profiles heterogeneity vs cancer classification

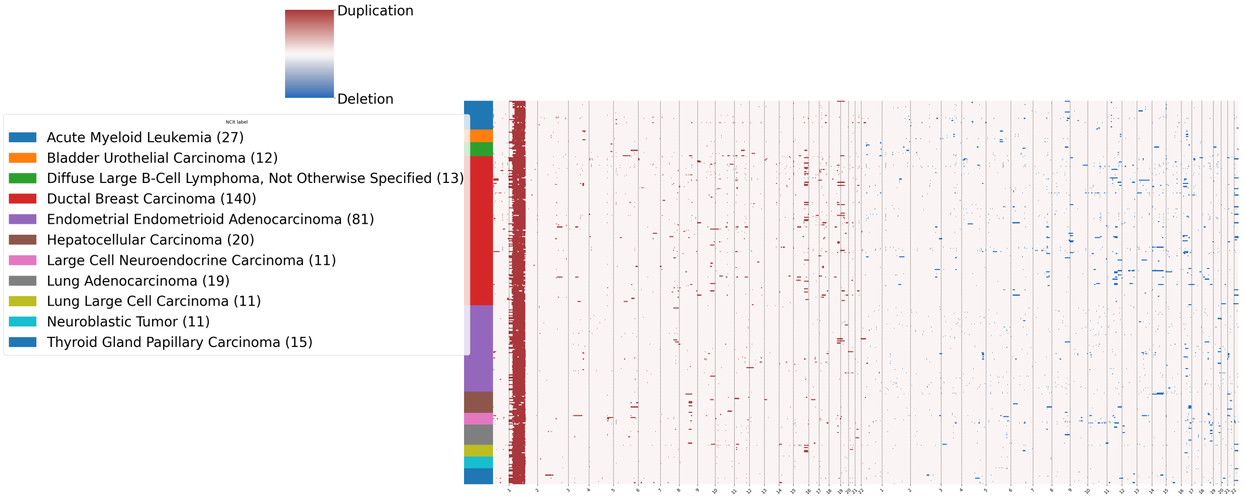
Correspondance of genomic profiles to NCIT cancer hierarchy

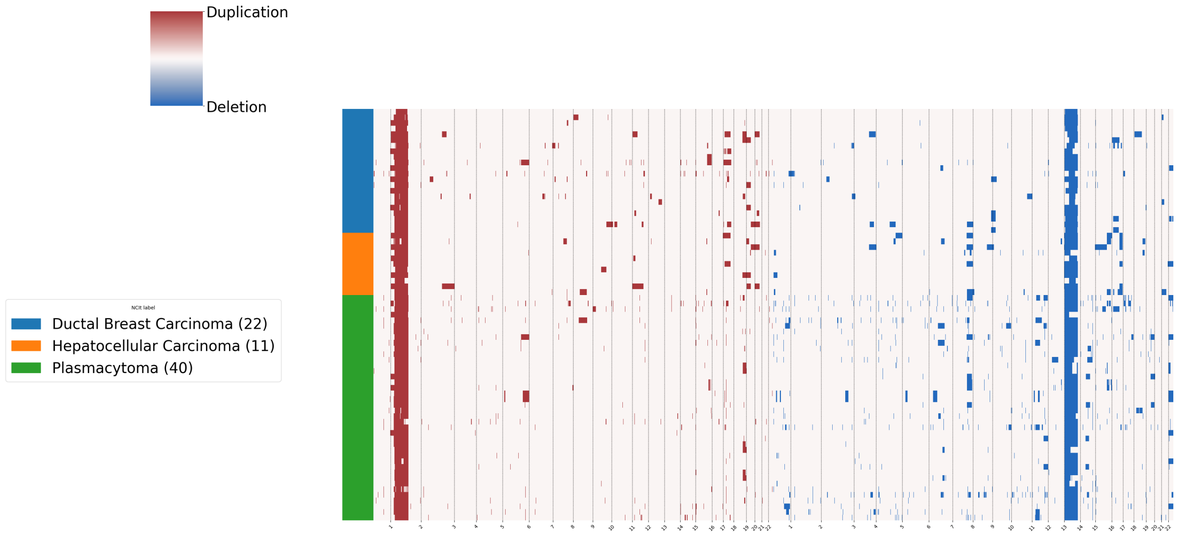
Lead: Ziying Yang

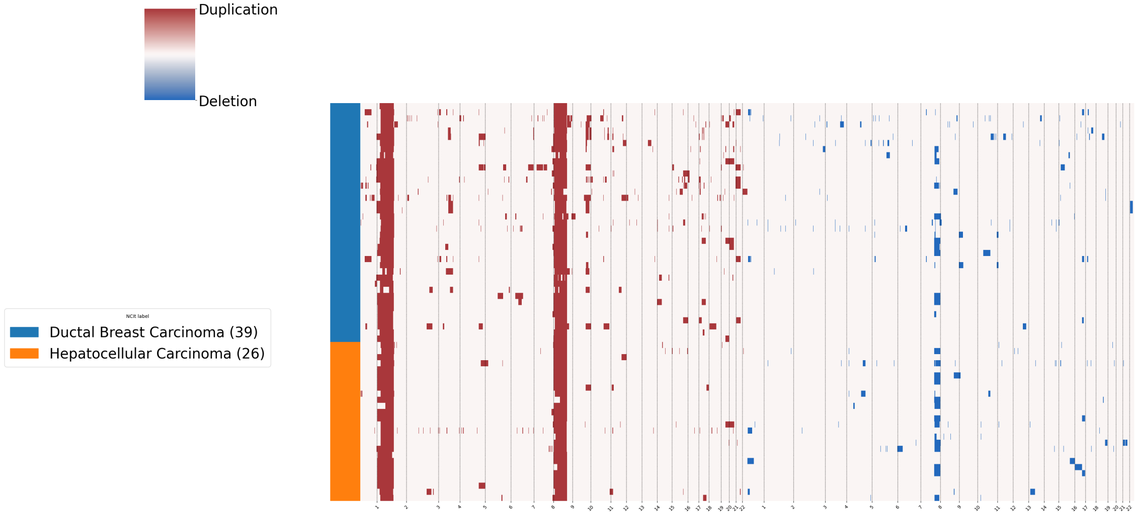


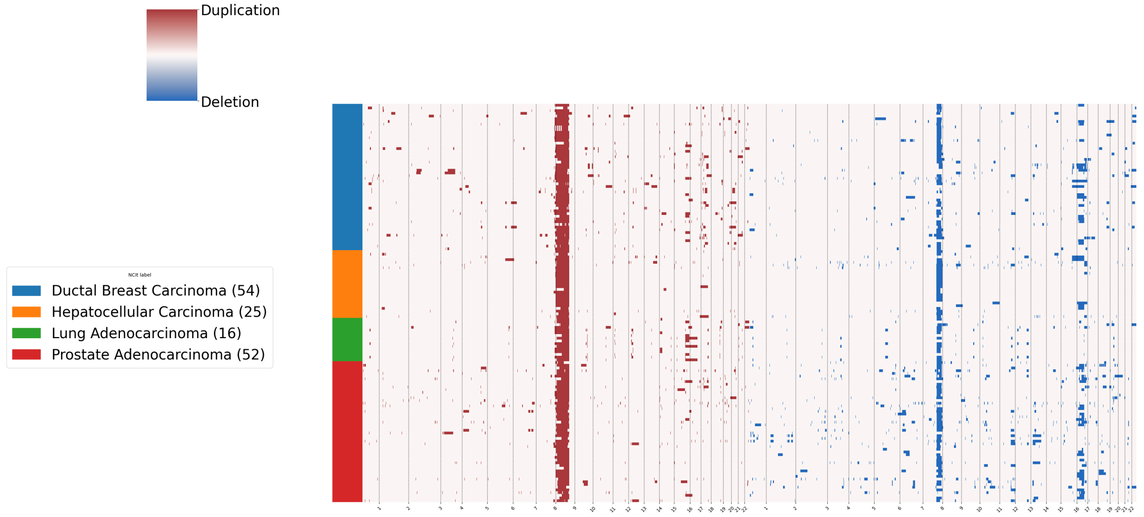
Example Use of Progenetix Data

Inter-tumoral CNV pattern similarity





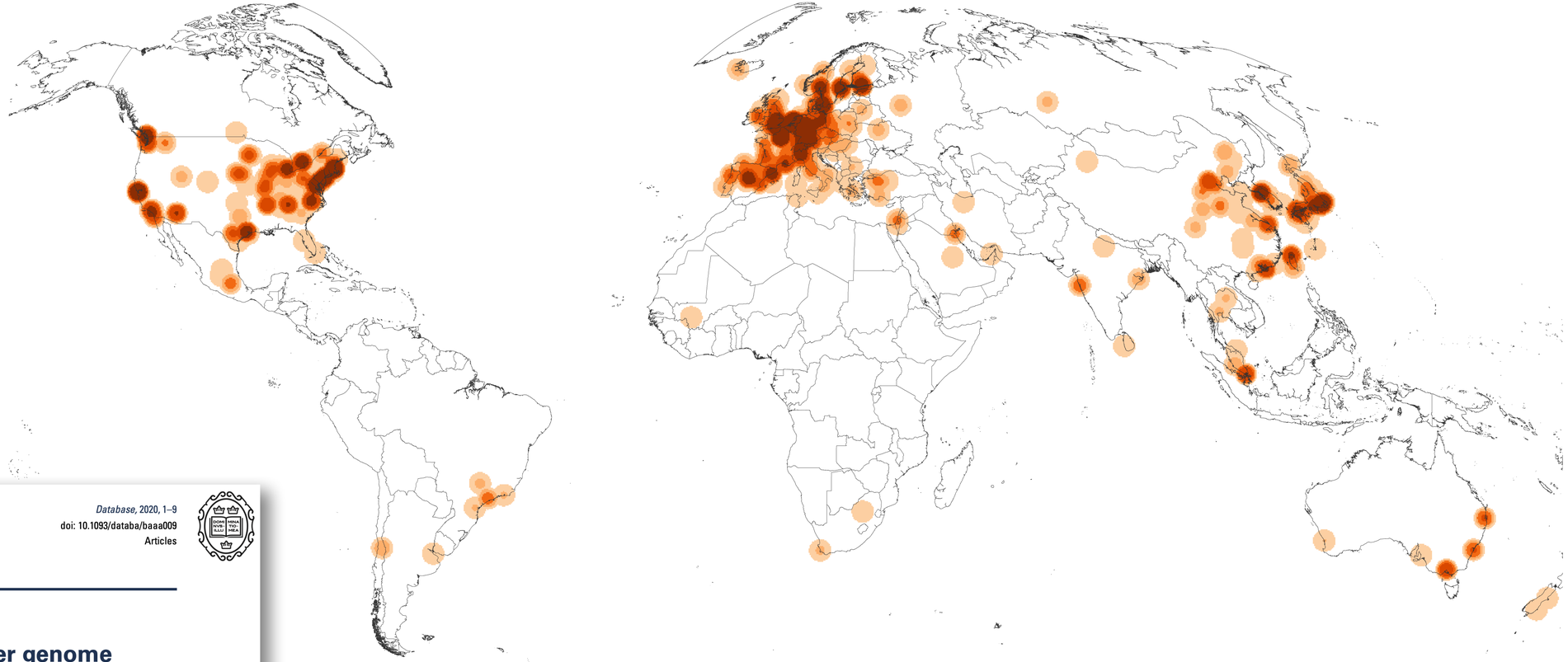


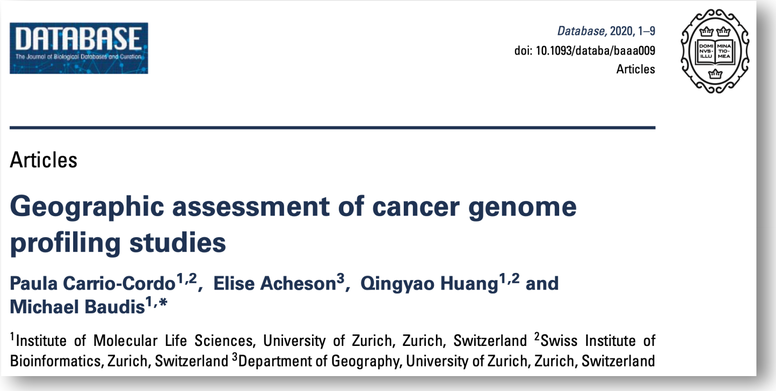


Mostly Carcinoma and Adenocarcinoma in different organs

Where Does Cancer Genomic Data Come From?

Geographic bias in published cancer genome profiling studies





DATABASE
The Journal of Biological Databases and Curation

Database, 2020, 1–9
doi: 10.1093/databa/baaa009
Articles

Articles

Geographic assessment of cancer genome profiling studies

Paula Carrio-Cordo[1,2], Elise Acheson[3], Qingyao Huang[1,2] and Michael Baudis[1,*]

[1]Institute of Molecular Life Sciences, University of Zurich, Zurich, Switzerland [2]Swiss Institute of Bioinformatics, Zurich, Switzerland [3]Department of Geography, University of Zurich, Zurich, Switzerland

Map of the geographic distribution (by first author affiliation) of the 104'543 genomic array, 36'766 chromosomal CGH and 15'409 whole genome/exome based cancer genome datasets. The numbers are derived from the 3'240 publications registered in the Progenetix database.

Different Approaches to Data Sharing

progenetix

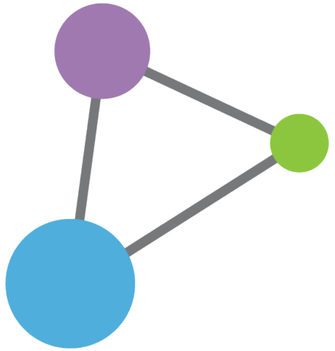
Centralized Genomic Knowledge Bases

European Genome-Phenome Archive

Data Commons
Trusted, controlled repository of multiple datasets

SPHN Swiss Personalized Health Network

Hub and Spoke
Common data elements, access, and usage rules



Linkage of distributed and disparate datasets

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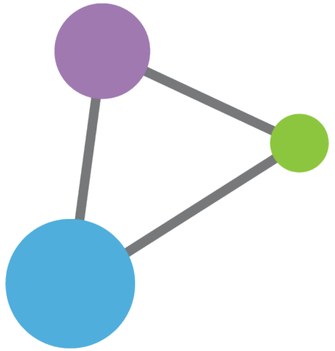


Linkage of distributed and disparate datasets

The EGA

European Genome-Phenome Archive

Long term secure archive for human biomedical research sensitive data, with focus on reuse of the data for further research (or "*broad and responsible use of genomic data*")

EUROPEAN GENOME-PHENOME ARCHIVE

CRG Centre for Genomic Regulation

EMBL-EBI

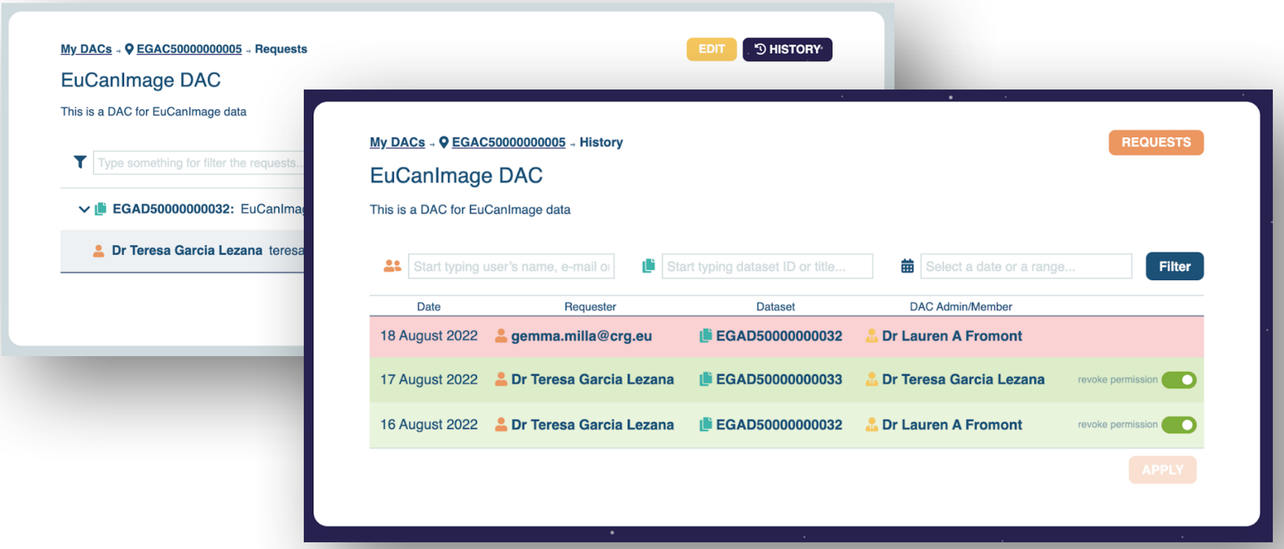
Global Alliance for Genomics & Health
DRIVER PROJECT

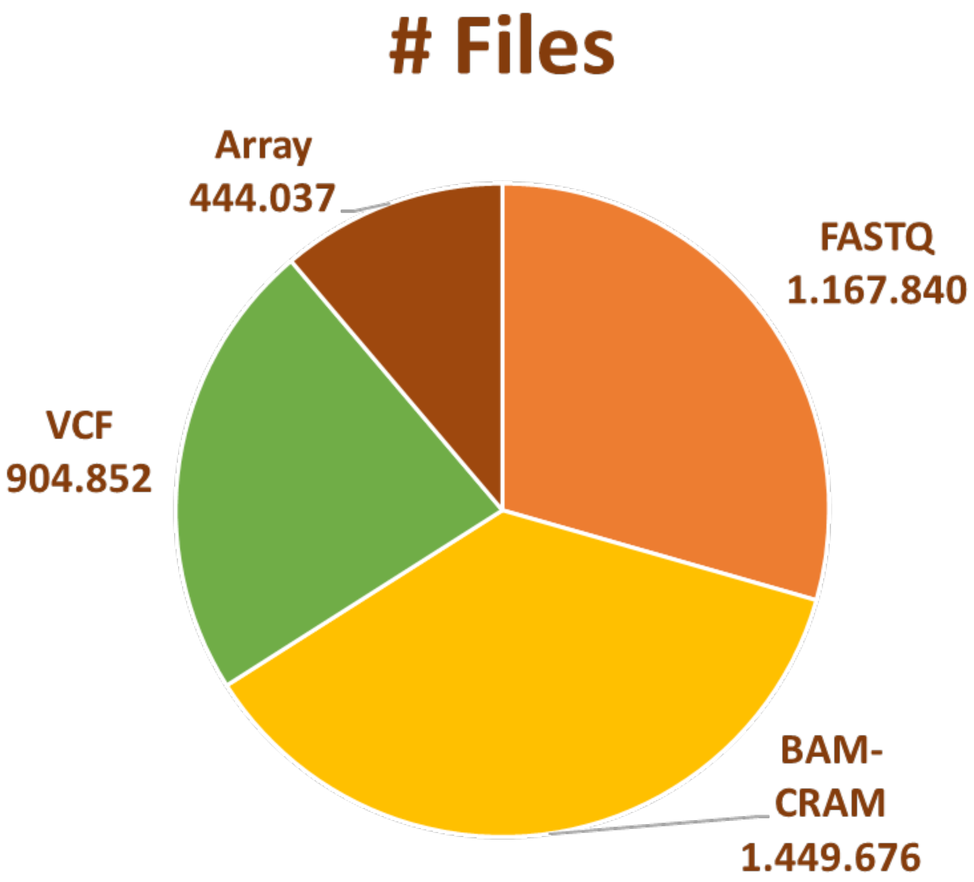
Slide: adapted from Jordi Rambla@ GA4GH 2023

The EGA

European Genome-Phenome Archive

- EGA “owns” nothing; data controllers tell who is authorized to access ***their*** datasets
- EGA admins provide smooth “all or nothing" data sharing process





4,328 Studies released
10,470 Datasets
2,309 Data Access Committees

Slide: adapted from Jordi Rambla@ GA4GH 2023

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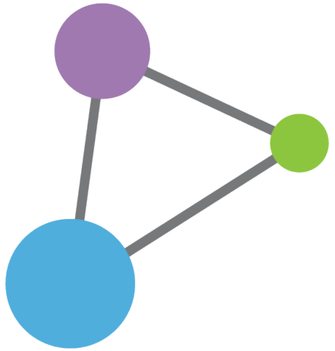

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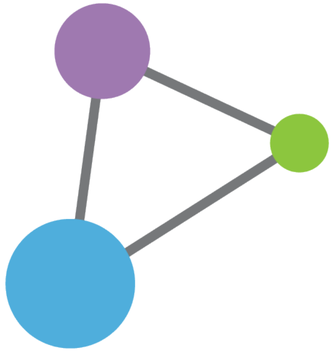

Linkage of distributed and disparate datasets

Federation

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for Genomics & Health

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GENOMICS

A federated ecosystem for sharing genomic, clinical data

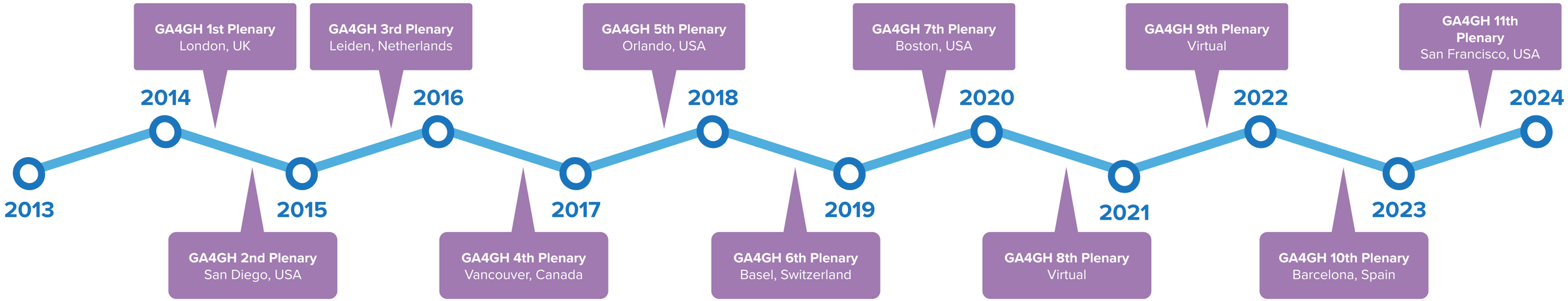
Silos of genome data collection are being transformed into seamlessly connected, independent systems

The Global Alliance for Genomics and Health*

SCIENCE 10 JUNE 2016 • VOL 352 ISSUE 6291

GA4GH timeline

Global Alliance for Genomics & Health



Pre-launch

73 partners sign a letter of intent to form an alliance

Building momentum

Global Alliance for Genomics & Health

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Formal launch of GA4GH

Published *Framework for Responsible Sharing of Genomic and Health-Related Data*

Formed four working groups

Developed three demonstration projects

GA4GH Connect

GA4GH CONNECT

Launch of **GA4GH Connect** and Strategic Roadmap

Formation of new organizational structure consisting of eight Work Streams and over twenty Driver Projects

Gap analysis

Gap analysis identifies three community imperatives

- Interoperability and alignment
- Implementation support
- Engaging with healthcar and clinical standards

Strategic Refresh

Strategic refresh introduces updates to GA4GH to better meet the three community imperatives

Our funders, partners, and Driver Projects

Global Alliance for Genomics & Health

Core Funders

CIHR IRSC Canadian Institutes of Health Research / Instituts de recherche en santé du Canada

UKRI Medical Research Council

GenomeCanada

NIH National Human Genome Research Institute

NIHR | National Institute for Health Research

wellcome

Host Institutions

OICR Ontario Institute for Cancer Research

McGill University — Victor Phillip Dahdaleh Institute of Genomic Medicine at McGill University

Broad Institute

wellcome sanger institute

EMBL-EBI

Supporting Funders

abbvie

aws

DNASTACK

illumina

Ontario Brain Institute / Institut ontarien du cerveau

Roche

Assigned Expert Funders/Employers

NIH National Human Genome Research Institute

University of Cape Town — iYunivesithi yaseKapa · Universiteit van Kaapstad

Strategic Partner

elixir

Driver Projects

All of Us Research Program

Autism Speaks

ClinGen Clinical Genome Resource

Matchmaker Exchange

epi share

Genomics England

VICC

elixir beacon network

NIH National Cancer Institute

monarch initiative

NHLBI BioData Catalyst®

Human Pangenome Project

RIFGC

NCPI

eosc cancer

GDI is funded by the European Commission under the Digital Europe Programme under grant agreement number 101081813 and through co-funding from participating Member States.

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INFORMATICS

Beacon v2 and Beacon networks: … federated data discovery in biome…

Jordi Rambla[1,2] | Michael Baudis[3] | Roberto Ariosa[1] | Tim Beck[4] | Lauren A. Fromont[1] | Arcadi Navarro[1,5,6,7] | Rahel Paloots[3] | Manuel Rueda[1] | Gary Saunders[8] | Babita Singh[1] | John D. Spalding[9] | Juha Törnroos[9] | Claudia Vasallo[1] | Colin D. Veal[4] | Anthony J. Brookes[4]

Cell Genomics

CellPress OPEN ACCESS

Commentary

International federation of genomic medicine databases using GA4GH standards

Adrian Thorogood,[1,2,*] Heidi L. Rehm,[3,4] Peter Goodhand,[5,6] Angela J.H. Page,[4,5] Yann Joly,[2] Michael Baudis,[7] Jordi Rambla,[8,9] Arcadi Navarro,[8,10,11,12] Tommi H. Nyronen,[13,14] Mikael Linden,[13,14] Edward S. Dove,[15] Marc Fiume,[16] Michael Brudno,[17] Melissa S. Cline,[18] and Ewan Birney[19]

Cell Genomics

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Perspective

GA4GH: International policies and standards for data sharing across genomic research and healthcare

Heidi L. Rehm,[1,2,47] Angela J.H. Page,[1,3,*] Lindsay Smith,[3,4] Jeremy B. Adams,[3,4] Gil Alterovitz,[5,47] Lawrence J. Babb,[1] Maxmillian P. Barkley,[6] Michael Baudis,[7,8] Michael J.S. Beauvais,[3,9] Tim Beck,[10] Jacques S. Beckmann,[11] Sergi Beltran,[12,13,14] David Bernick,[1] Alexander Bernier,[9] James K. Bonfield,[15] Tiffany F. Boughtwood,[16,17] Guillaume Bourque,[9,18] Sarion R. Bowers,[15] Anthony J. Brookes,[10] Michael Brudno,[18,19,20,21,38] Matthew H. Brush,[22] David Bujold,[9,18,38] Tony Burdett,[23] Orion J. Buske,[24] Moran N. Cabili,[1] Daniel L. Cameron,[25,26] Robert J. Carroll,[27] Esmeralda Casas-Silva,[123] Debyani Chakravarty,[29] Bimal P. Chaudhari,[30,31] Shu Hui Chen,[32] J. Michael Cherry,[33] Justina Chung,[3,4] Melissa Cline,[34] Hayley L. Clissold,[15] Robert M. Cook-Deegan,[35] Mélanie Courtot,[23] Fiona Cunningham,[23] Miro Cupak,[6] Robert M. Davies,[15] Danielle Denisko,[19] Megan J. Doerr,[36] Lena I. Dolman,[19]

(Author list continued on next page)

Cell Genomics

Technology

The GA4GH Variation Representation Specification: A computational framework for variation representation and federated identification

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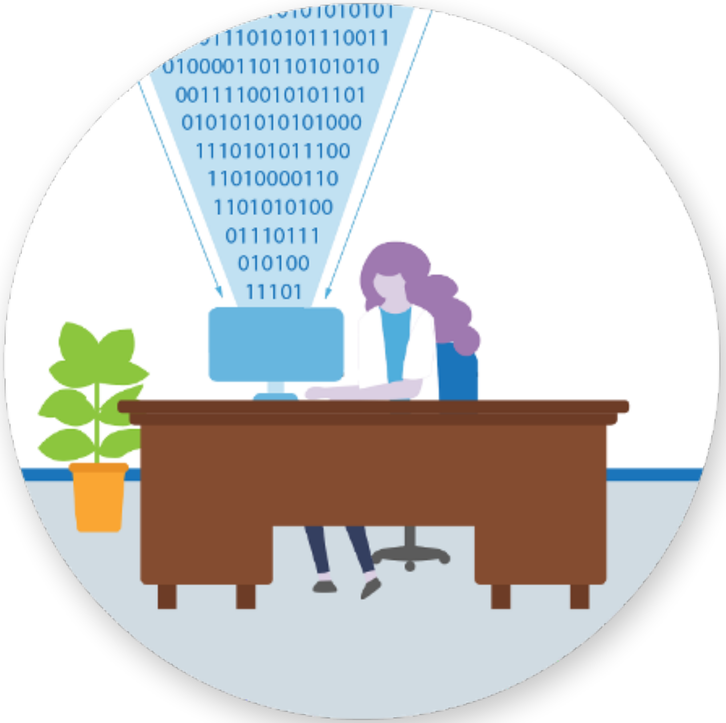
A New Paradigm for Data Sharing

FROM

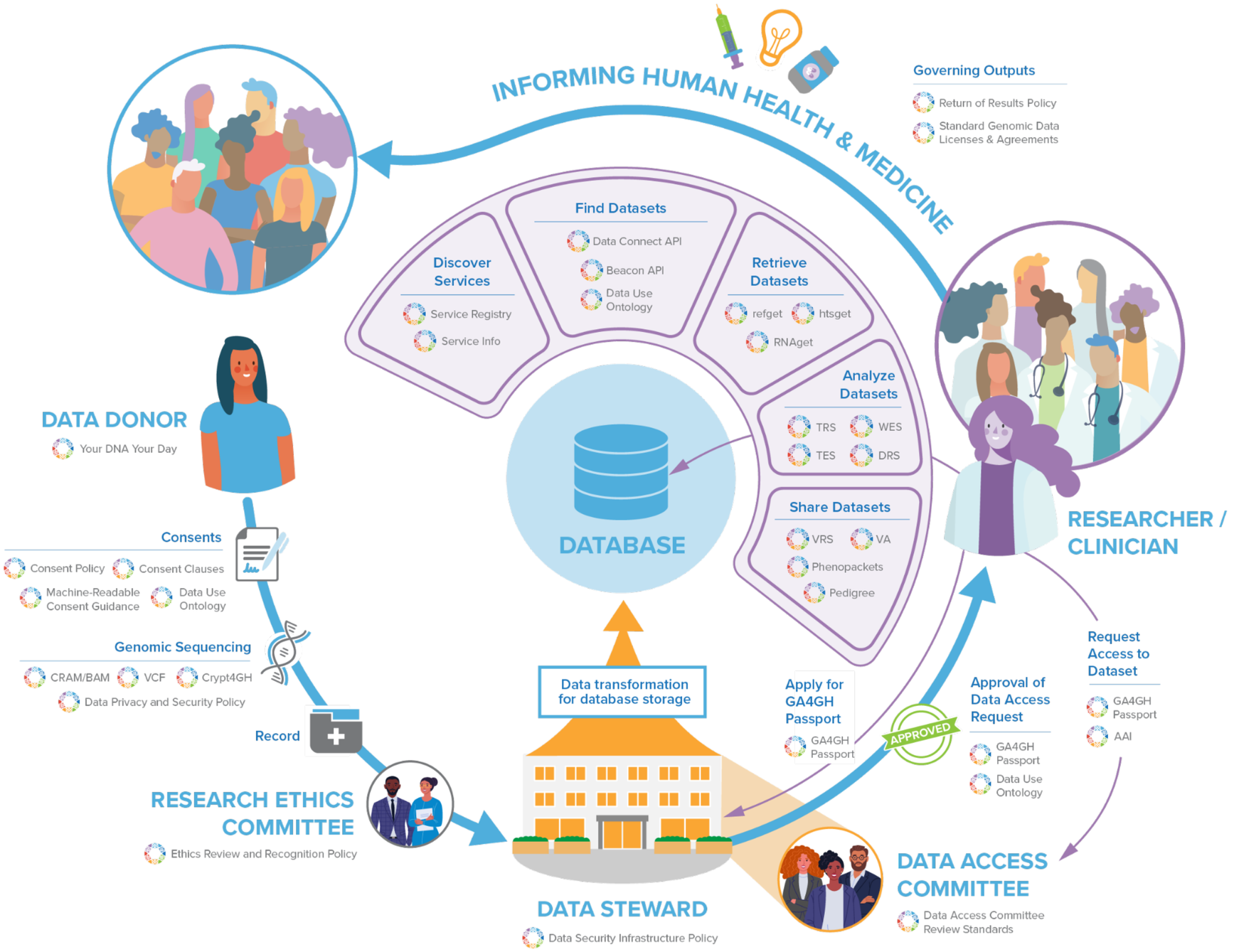


Data Copying

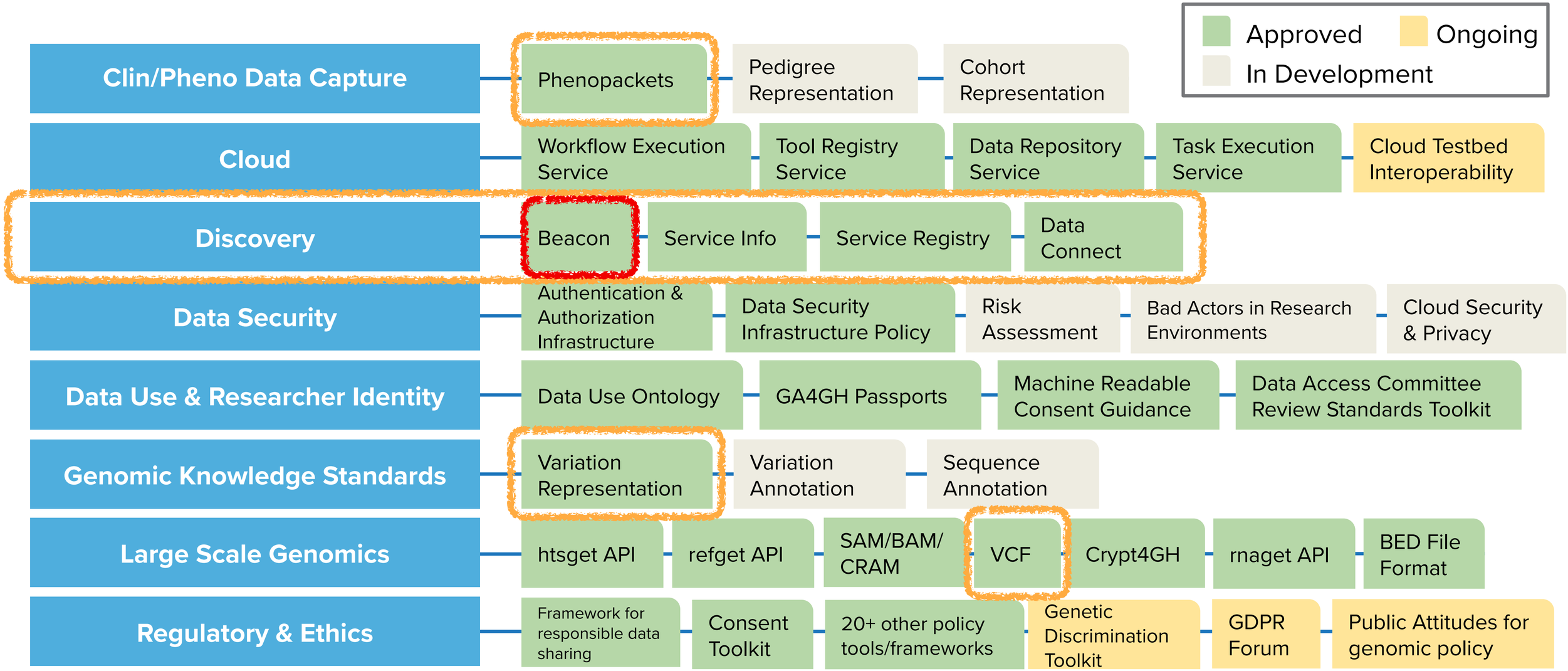
STANDARDS

TO

Data Visiting


INFORMING HUMAN HEALTH & MEDICINE
Governing Outputs
Return of Results Policy
Standard Genomic Data Licenses & Agreements
Discover Services
Service Registry
Service Info
Find Datasets
Data Connect API
Beacon API
Data Use Ontology
Retrieve Datasets
refget
htsget
RNAget
Analyze Datasets
TRS
WES
TES
DRS
Share Datasets
VRS
VA
Phenopackets
Pedigree
DATABASE
DATA DONOR
Your DNA Your Day
Consents
Consent Policy
Consent Clauses
Machine-Readable Consent Guidance
Data Use Ontology
Genomic Sequencing
CRAM/BAM
VCF
Crypt4GH
Data Privacy and Security Policy
Record
RESEARCH ETHICS COMMITTEE
Ethics Review and Recognition Policy
Data transformation for database storage
DATA STEWARD
Data Security Infrastructure Policy
Apply for GA4GH Passport
GA4GH Passport
APPROVED
Approval of Data Access Request
GA4GH Passport
Data Use Ontology
Request Access to Dataset
GA4GH Passport
AAI
DATA ACCESS COMMITTEE
Data Access Committee Review Standards
RESEARCHER / CLINICIAN

Overview of GA4GH standards and frameworks



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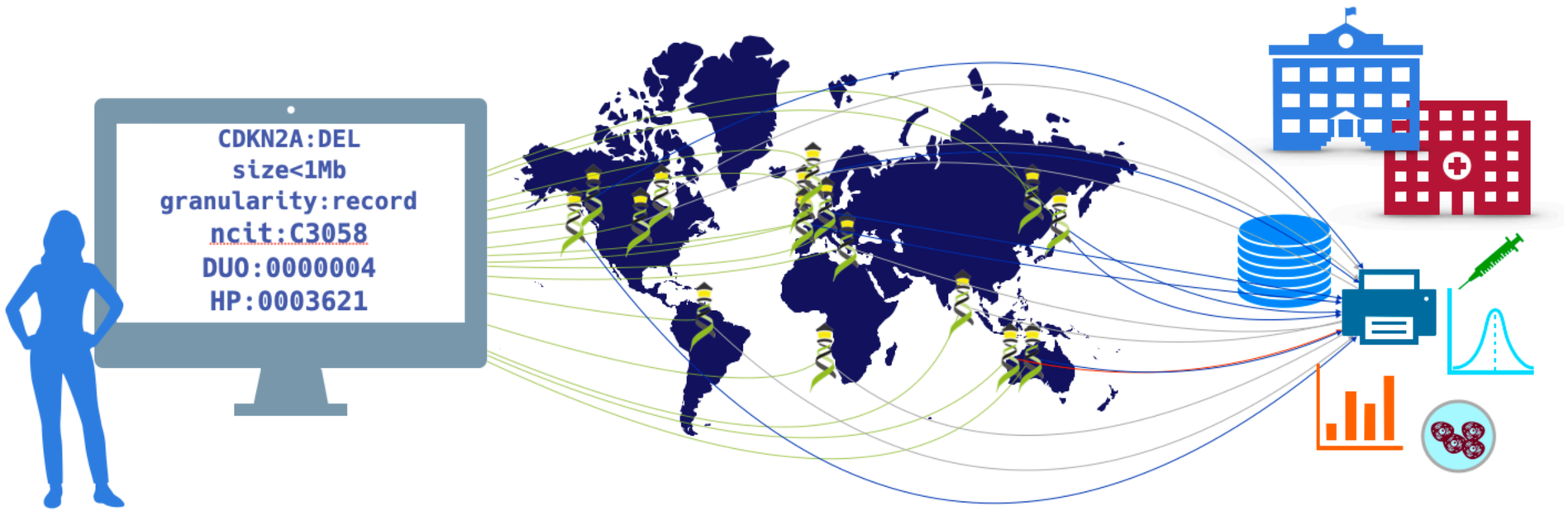
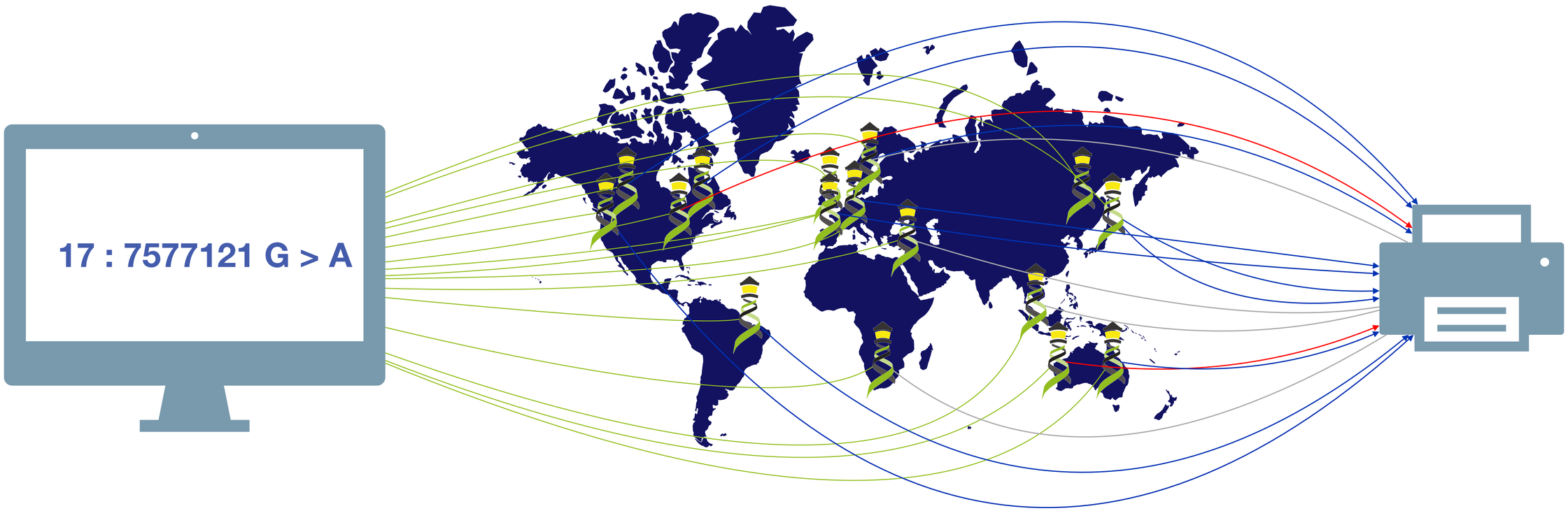


The GA4GH Beacon Protocol

Federating Genomic Discoveries

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

YES | **NO** | **\0**



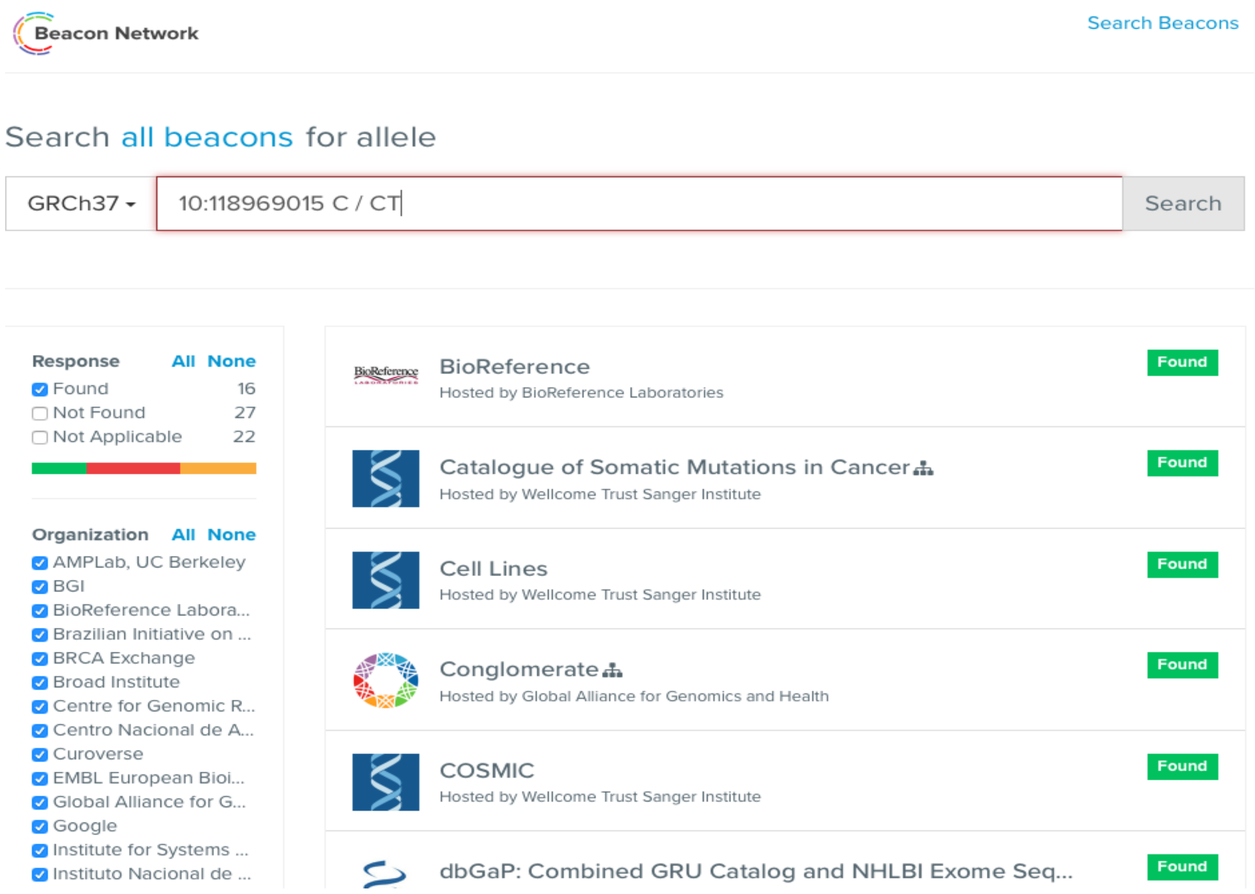
Have you seen this variant? It came up in my patient and we don't know if this is a common SNP or worth following up.

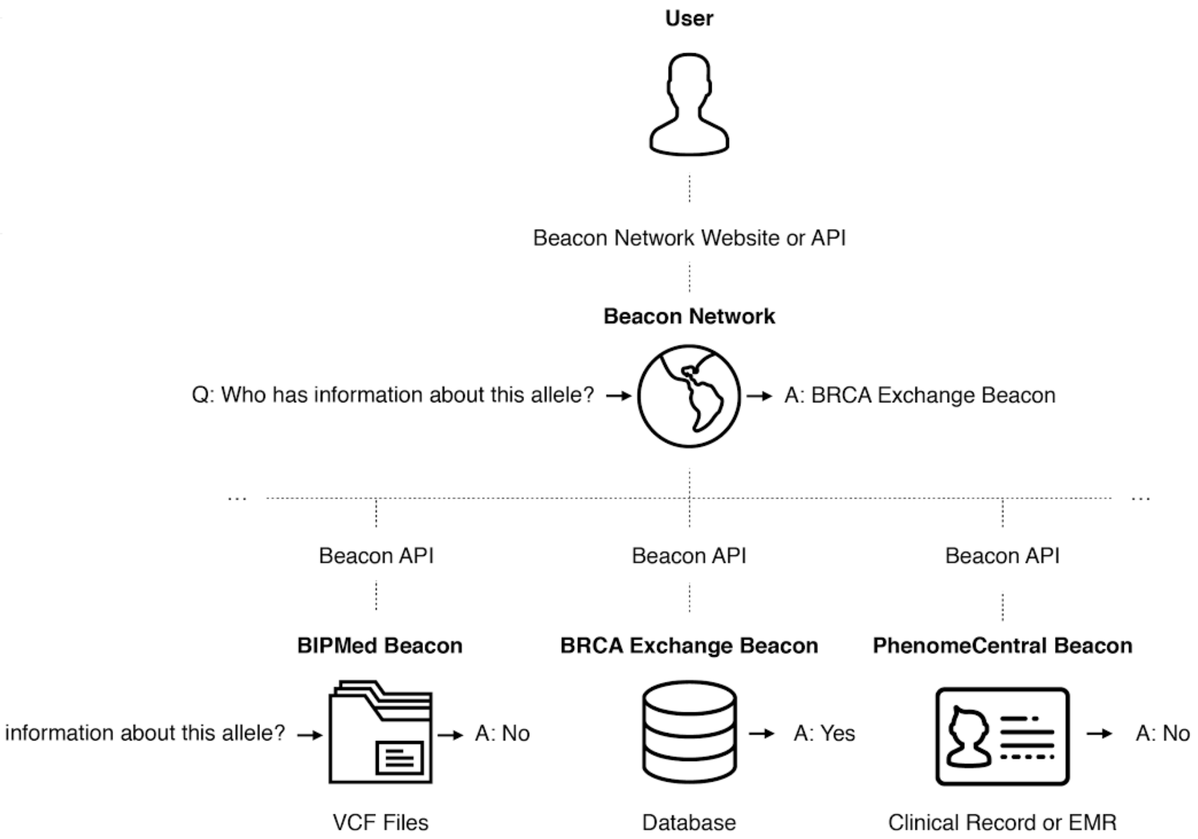
A Beacon network federates *genome variant queries* across databases that support the ***Beacon API***

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

Beacon Project in 2016

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





Beacon

Releases

Date	Tag	Title
2018-01-24	v0.4.0	Beacon
2016-05-31	v0.3.0	Beacon

35+ Organizations

90+ Beacons

200+ Datasets

100K+

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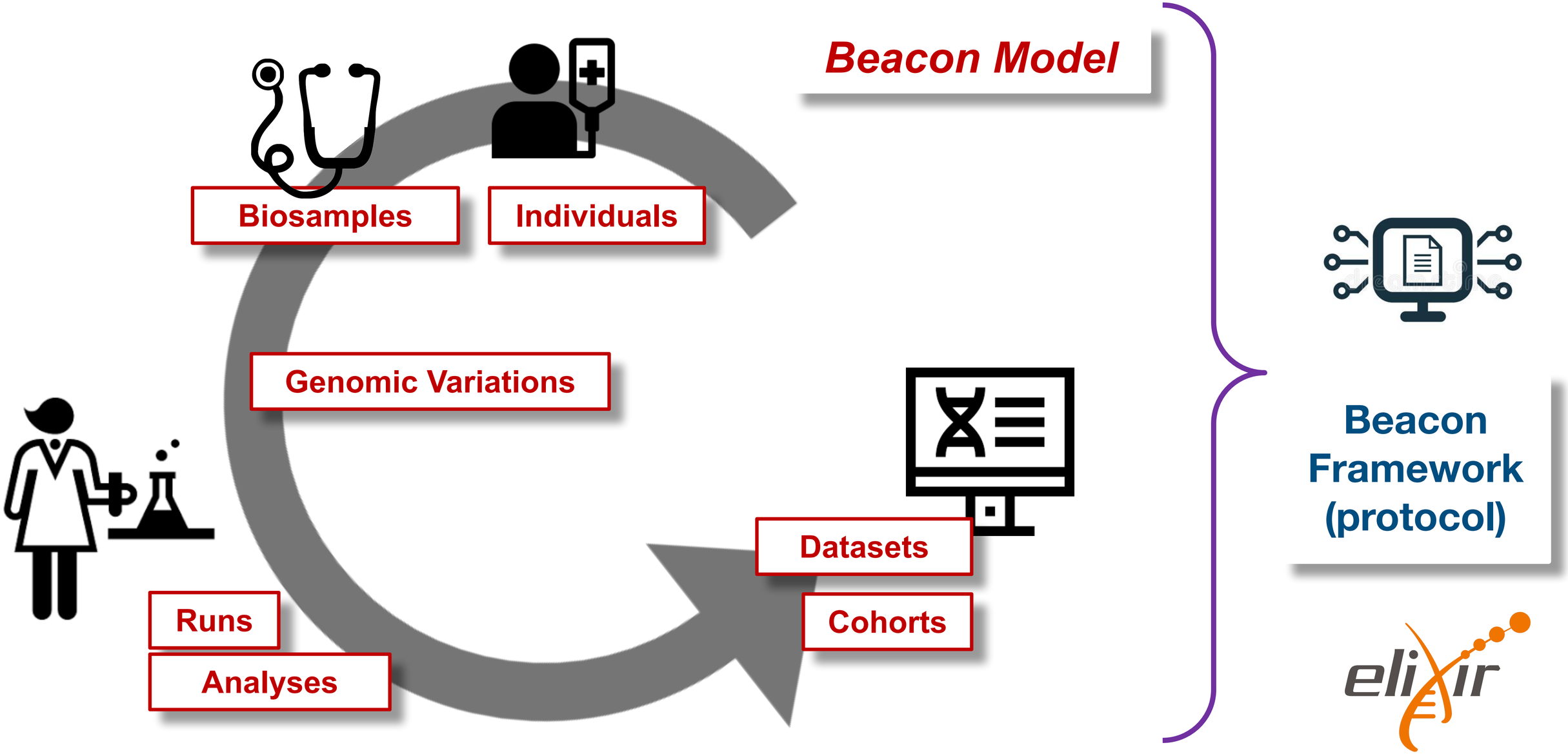
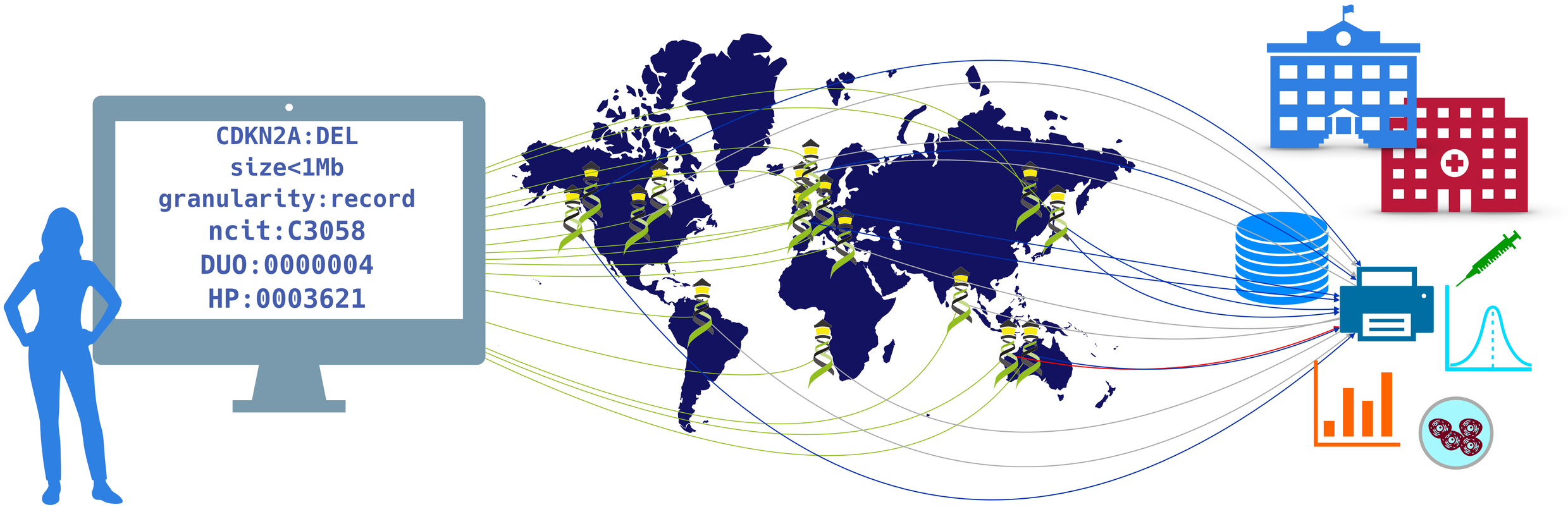
	Beacon v1 Development	Beacon v2 Development	Related ...
2014	GA4GH founding event; Jim Ostell proposes Beacon concept including "more features ... version 2"		
2015	• beacon-network.org aggregator created by DNAstack		• ELIXIR starts Beacon project support
2016	• Beacon v0.3 release • work on queries for structural variants (brackets for fuzzy start and end parameters...)	• Beacon+ concept implemented on progenetix.org • concepts from GA4GH Metadata (ontologies…) • entity-scoped query parameters ("individual.age")	
2017	• OpenAPI implementation • integrating CNV parameters (e.g. "startMin, statMax")	• Beacon+ demos "handover" concept	• GA4GH re-structuring (workstreams...) • Beacon part of Discovery WS
2018	• Beacon v0.4 release in January; feature release for GA4GH approval process • GA4GH Beacon v1 approved at Oct plenary		• new Beacon website (March)
2019	• ELIXIR Beacon Network	• Beacon hackathon Stockholm; settling on "filters" • Barcelona goes Zurich developers meeting • Beacon API v2 Kick off • adopting "handover" concept	• Beacon publication at Nature Biotechnology
2020		• "Scouts" teams working on different aspects - filters, genomic variants, compliance ... • discussions w/ clinical stakeholders	
2021		• framework + models concept implemented • range and bracket queries, variant length parameters • starting of GA4GH review process	• Phenopackets v2 approved
2022		• further changes esp. in default model, aligning with Phenopackets and VRS • unified beacon-v2 code & docs repository • Beacon v2 approved at Apr GA4GH Connect	• *docs.genomebeacons.org*

Beacon v2

docs.genomebeacons.org





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

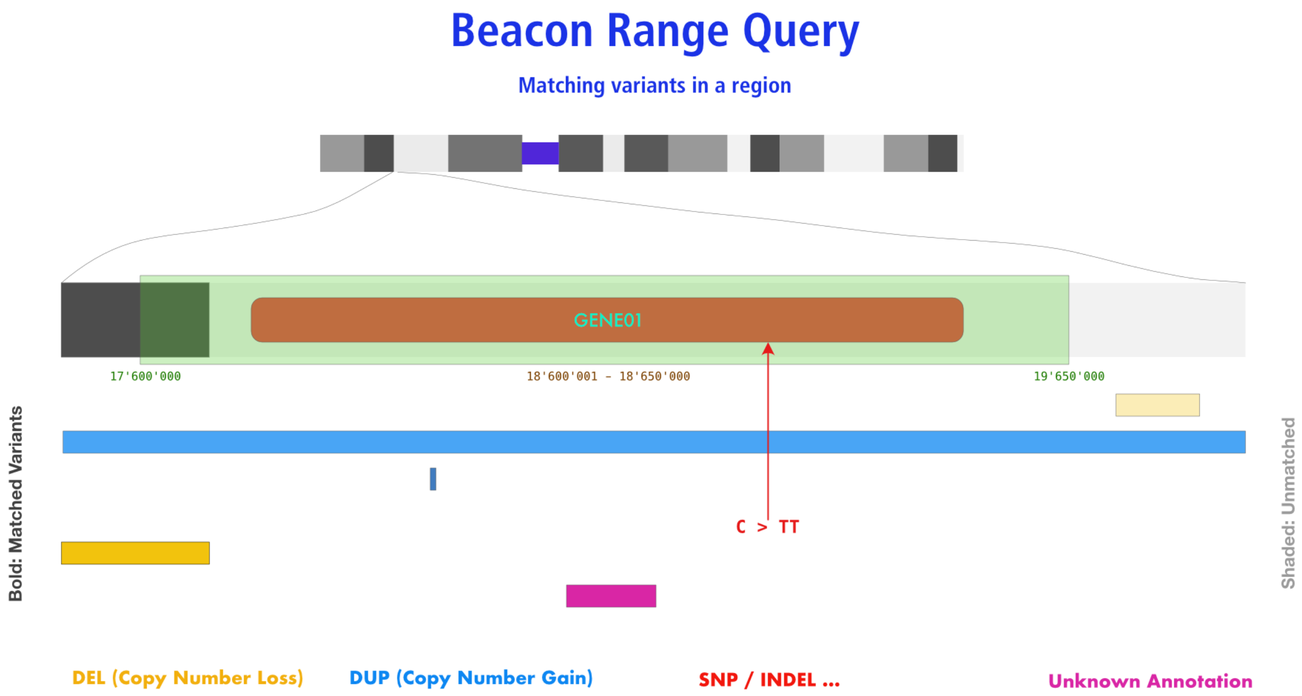
Beacon *v2* API

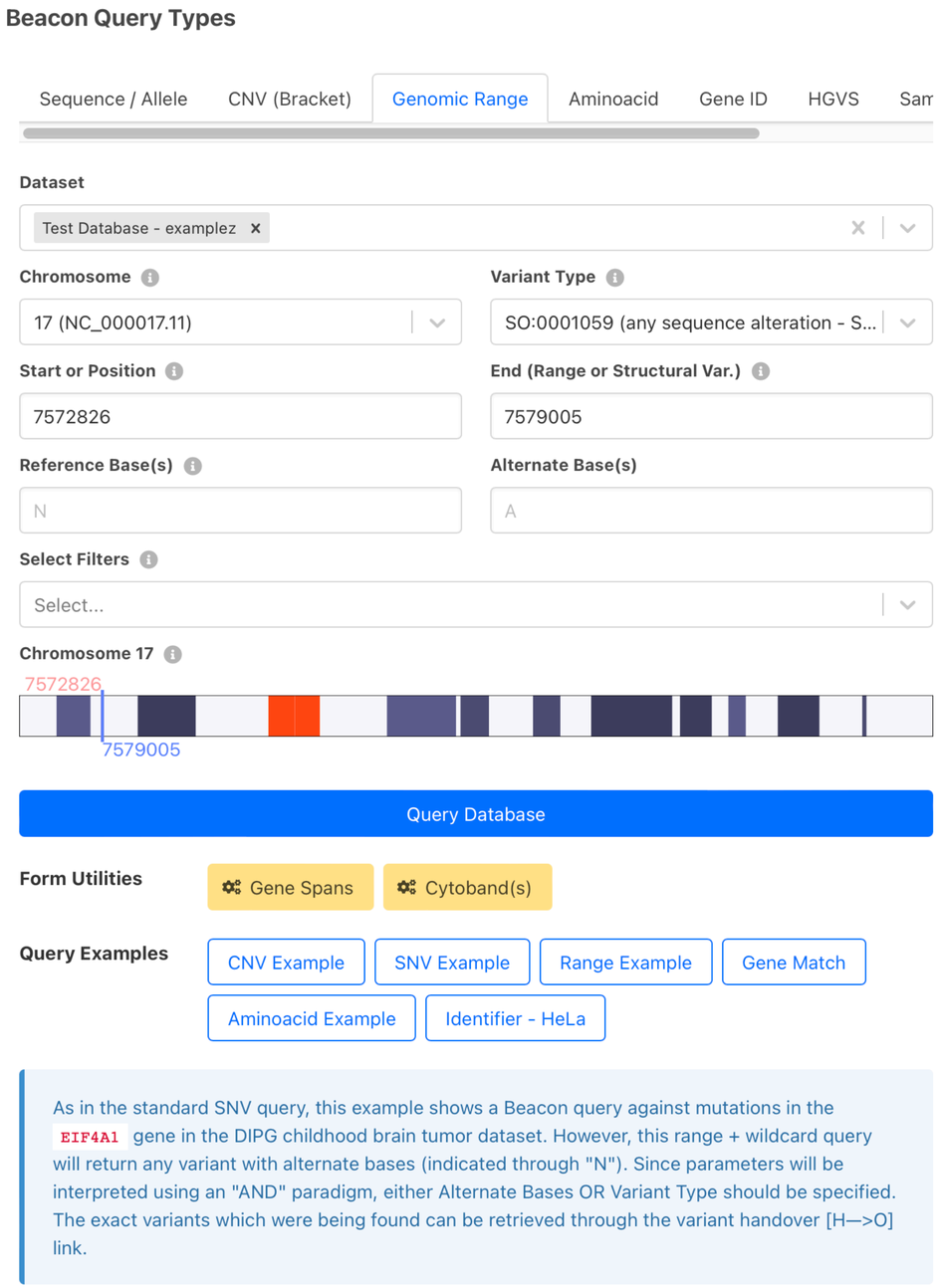
The Beacon API v2 represents a simple but powerful **genomics API** for ***federated*** data discovery and retrieval

Variation Queries

Range ("anything goes") Request

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

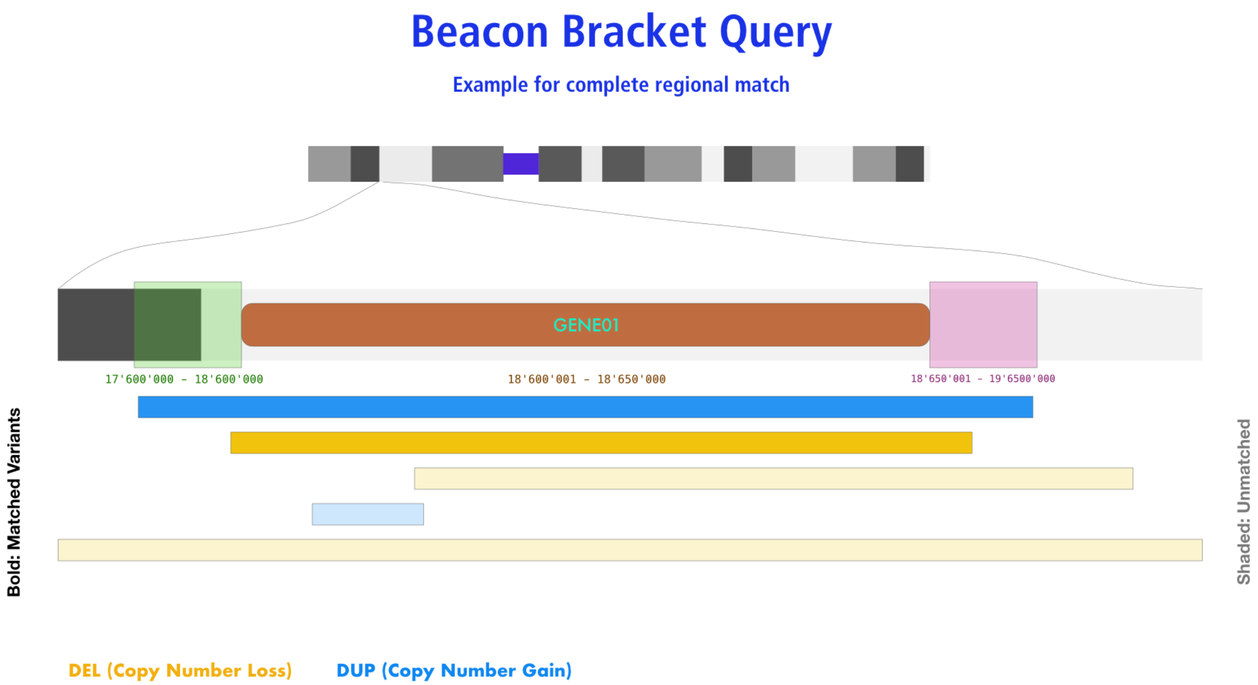


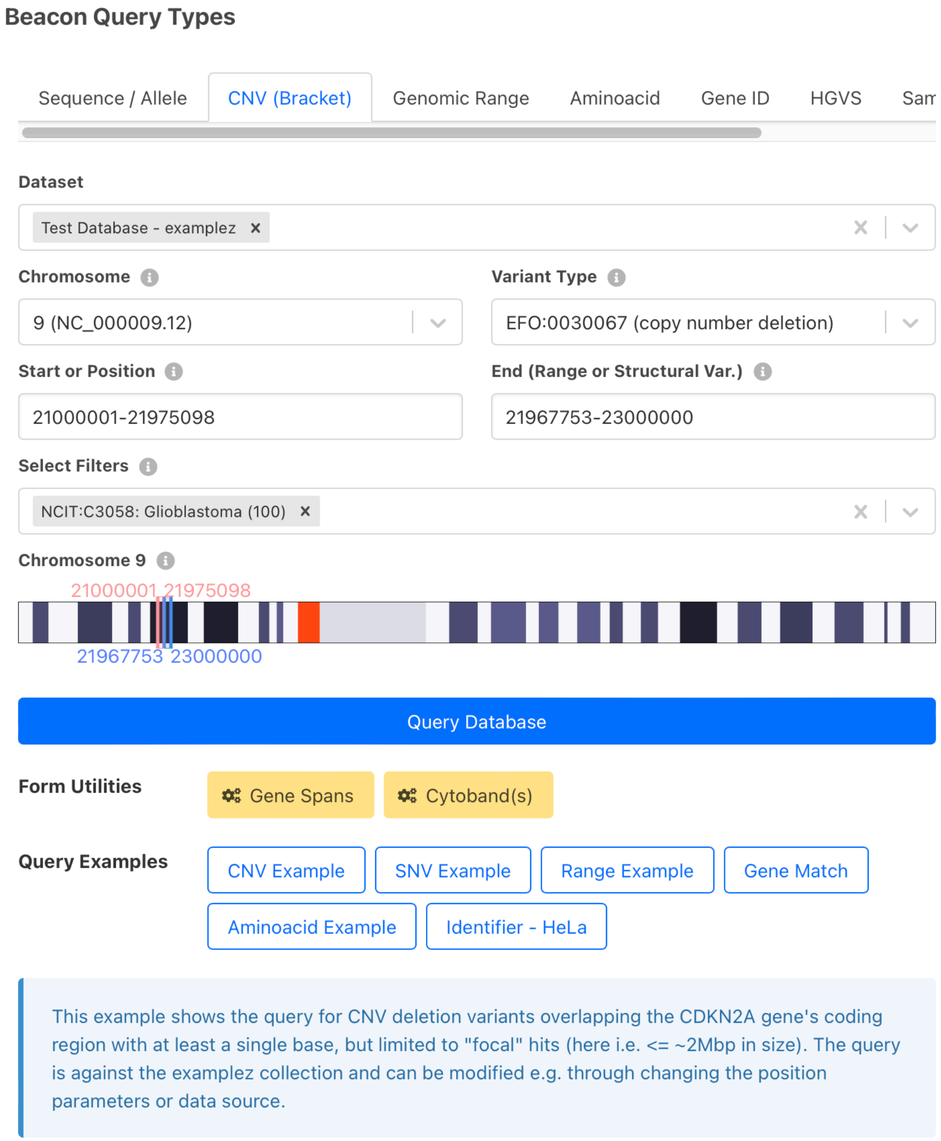


Variation Queries

Bracket ("CNV") Query

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





Beacon v2 Filters

Example: Use of hierarchical classification systems (here NCIt 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

☑	NCIT:C4914: Skin Carcinoma	213
☐	NCIT:C4475: Dermal Neoplasm	109
☑	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
Calls: 0
Samples: 523

$f_{alleles}$: 0

Callsets Variants
Legacy Interface
UCSC region

Show JSON Response

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

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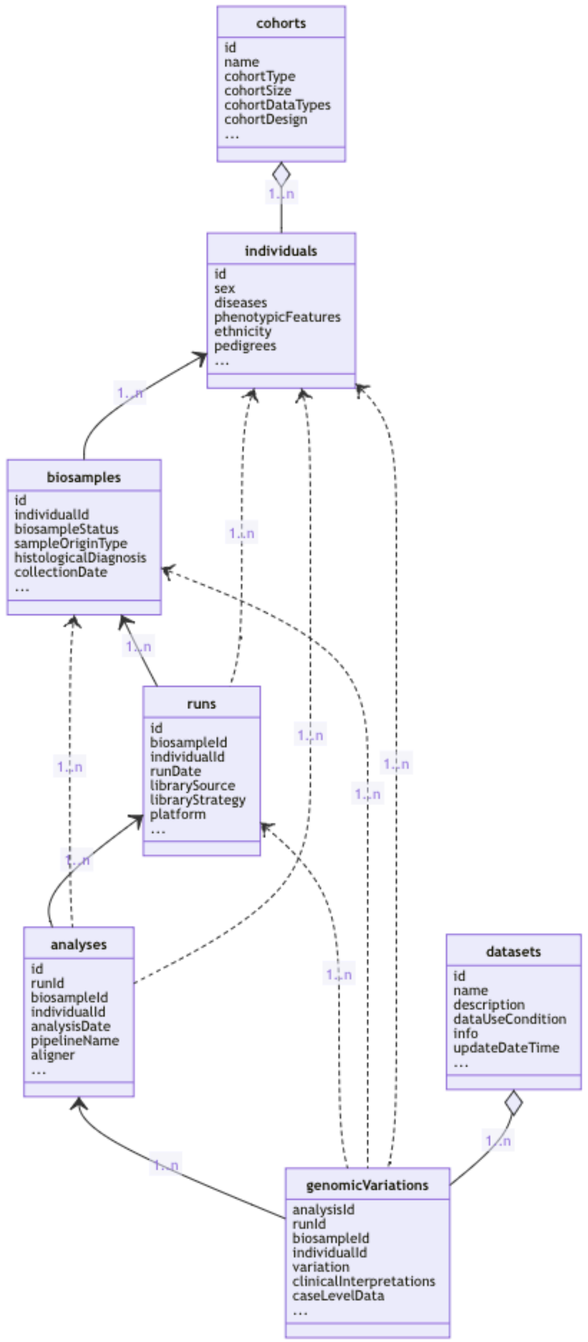
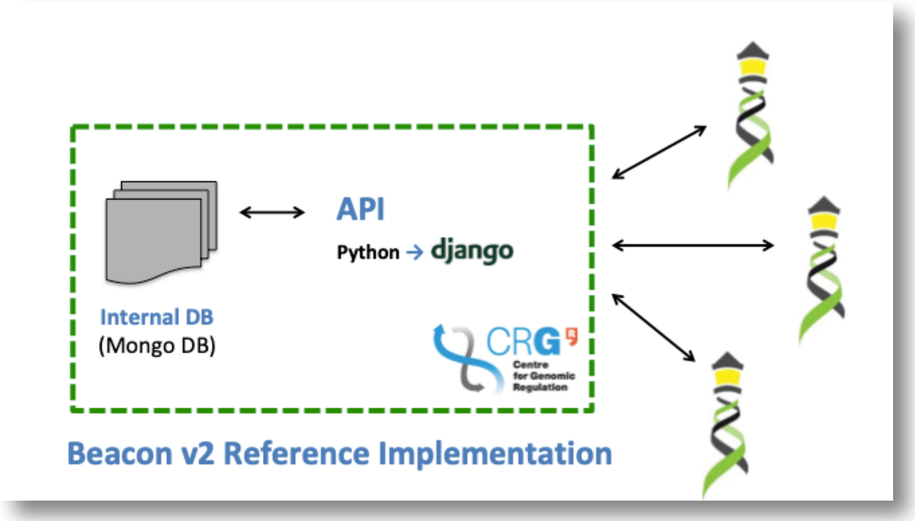
Beacon Default v2 Model

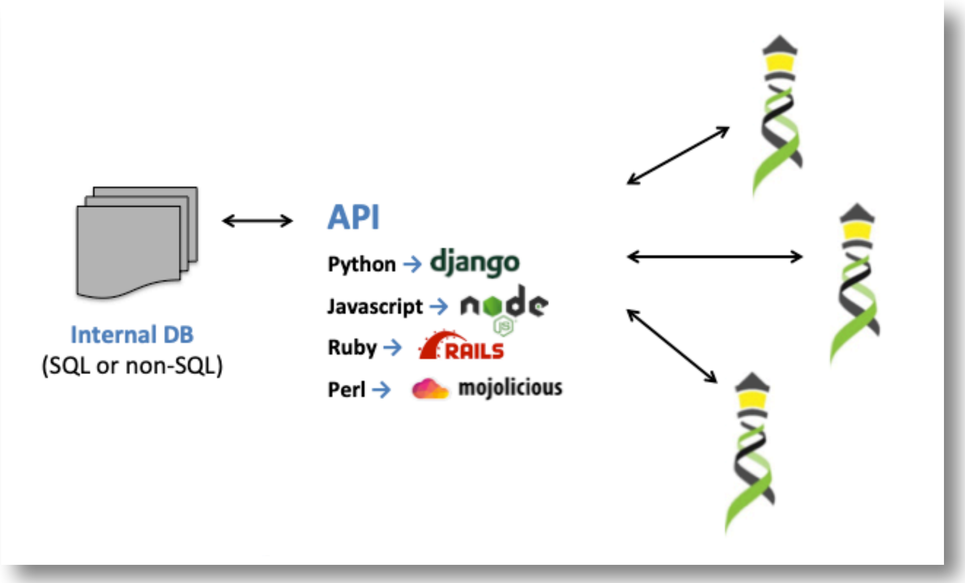
- The Beacon ***framework*** describes the overall structure of the API requests, responses, parameters, the common components, etc.
- Beacon ***models*** describe the set of concepts included in a Beacon, like individual or biosample, and also the relationships between them.
- Besides logical concepts, the Beacon ***models*** represent the schemas for data delivery in “record” granularity
- Beacon explicitly allows the use of *other models* besides its *version specific default*.
- Adherence to a shared ***model*** empowers federation
- Use of the ***framework*** w/ different models extends adoption

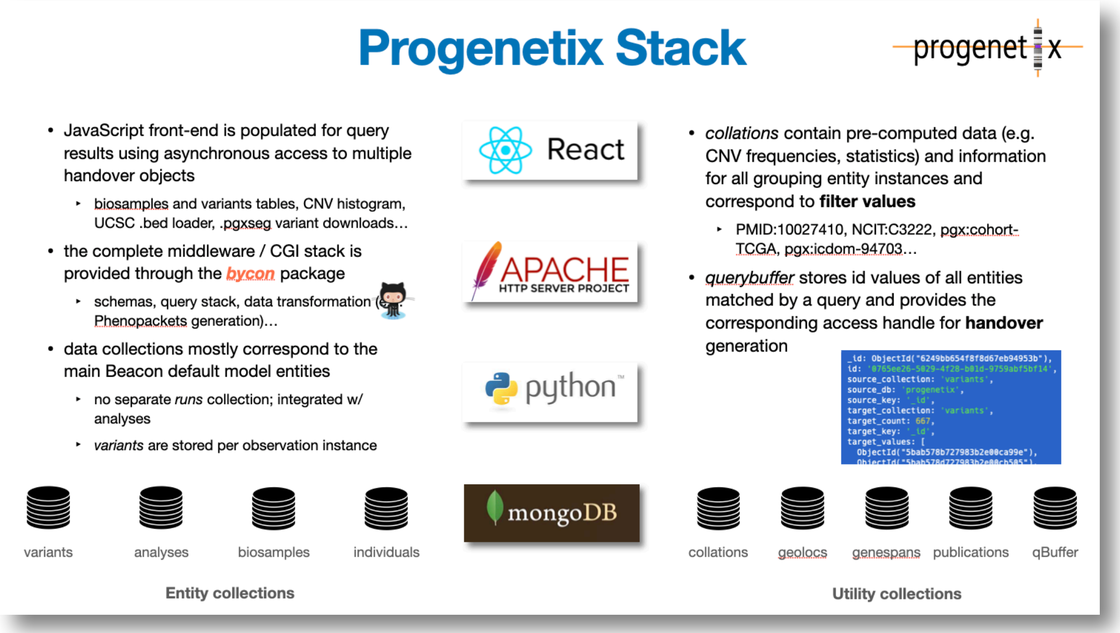


Implementing Beacon v2

... its just code ¯_(ツ)_/¯







Universität Zürich[UZH]

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bycon for GA4GH Beacon

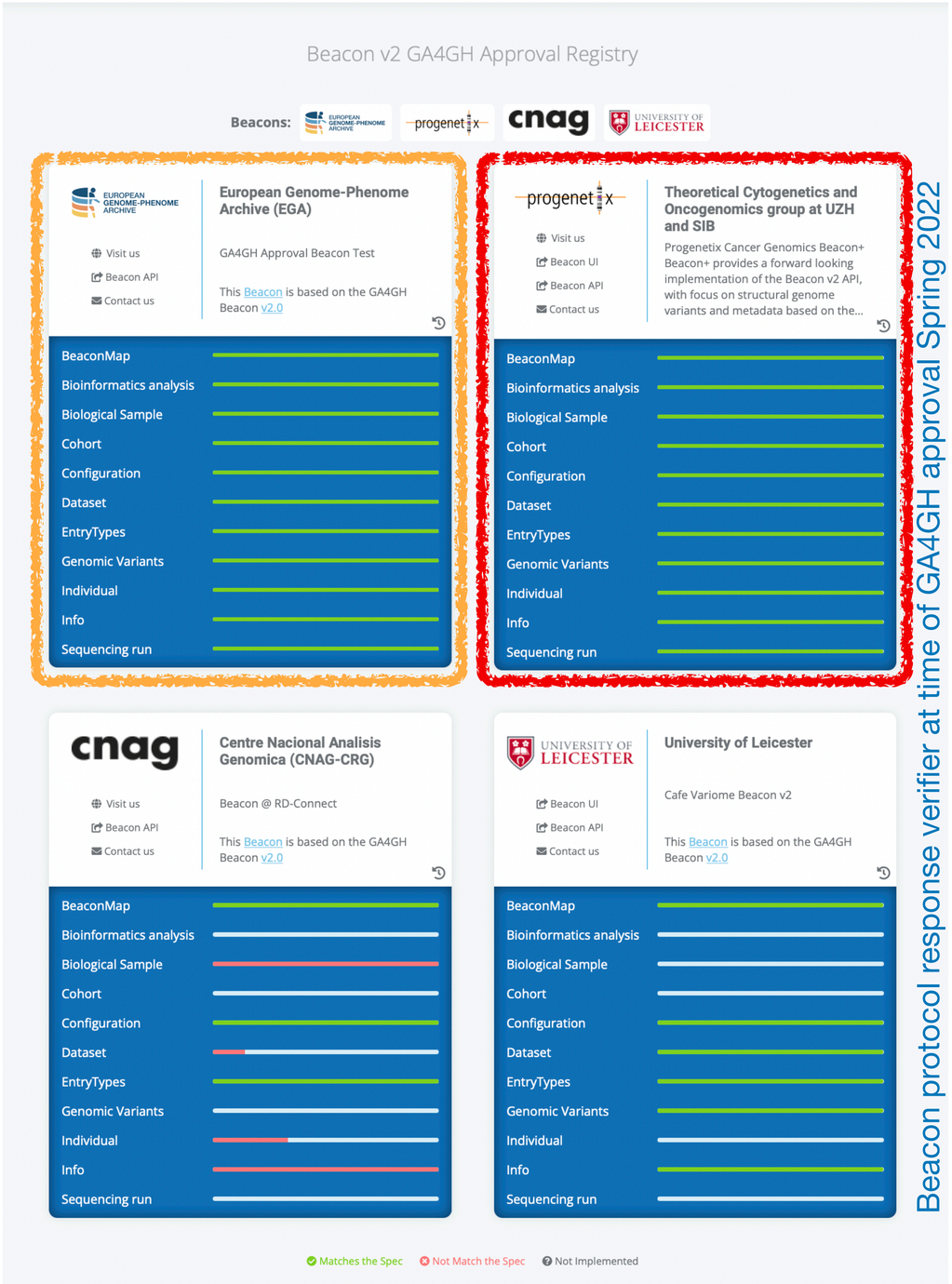
Implementation driven development of a GA4GH standard

progenetix

bycon Beacon

Implementation driven standards development

- Progenetix' Beacon+ has served as implementation driver since 2016
- the *bycon* package is used to prototype advanced Beacon features such as
  - ➡ structural variant queries
  - ➡ data handovers
  - ➡ Phenopackets integration
  - ➡ variant co-occurrences
  - ➡ ...



Beacon protocol response verifier at time of GA4GH approval Spring 2022

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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 (e.g. 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

- *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"),
```

React

APACHE HTTP SERVER PROJECT

python™

mongoDB

variants | analyses | biosamples | individuals

Entity collections

collations | geolocs | genespans | publications | qBuffer

Utility collections

bycon.progenetix.org
github.com/progenetix/bycon/

progenetix / **byconaut**

Type / to search

Code | Issues | Pull requests | Actions | Projects | Wiki | Security | Insights | Settings

byconaut Public

Edit Pins | Unwatch 2 | Fork 1 | Star 0

main | 2 branches

mbaudis get_plot_parameters

bin	
docs	
exports	
imports	
local	
rsrc	
services	
tmp	
.gitignore	
LICENSE	
README.md	
__init__.py	
install.py	
install.yaml	
mkdocs.yaml	

beaconplus.progenetix.org
.../progenetix/beaconplus-web/

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forked from progenetix/progenetix-web

Edit Pins | Watch 0 | Fork 3 | Star 0

main | 1 branch | 0 tags | Go

This branch is 44 commits ahead, 24 commits behind progenetix:main.

mbaudis code cleaning, no feature changes 2a

.github/workflows	cleanup
docs	still first implementation clean-up
extra	documentation
public	graphic refinement
src	code cleaning, no feature changes
.babelrc	Simplify query generation and add
.env.development	first working version
.env.local	first working version
.env.production	env
.env.staging	env
.eslintrc.json	BioSubsetsPage perf optimisations

progenetix / **bycon**

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main | 4 branches | 25 tags | Go to file | Add file | Code

mbaudis 1.3.6 ... be19a12 3 days ago 852 commits

.github/workflows	Create mk-bycon-docs.yaml	8 months ago
bycon	1.3.6	3 days ago
docs	1.3.6	3 days ago
local	1.3.5 preparation	2 weeks ago
.gitignore	Update .gitignore	3 months ago
LICENSE	Create LICENSE	3 years ago
MANIFEST.in	major library & install disentanglement	9 months ago
README.md	#### 2023-07-23 (v1.0.68)	4 months ago
install.py	1.3.6	3 days ago
install.yaml	v1.0.57	5 months ago
mkdocs.yaml	1.1.6	3 months ago
requirements.txt	1.3.6	3 days ago
setup.cfg	...	10 months ago
setup.py	1.3.6	3 days ago
updev.sh	1.3.6	3 days ago

About

Bycon - A Python Based Beacon API (beacon-project.io) implementation leveraging the Progenetix (progenetix.org) data model

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bycon.progenetix.org
github.com/progenetix/bycon/

pgxRpi

An interface API for analyzing Progenetix CNV data in R using the Beacon+ API

GitHub: https://github.com/progenetix/pgxRpi

README.md

pgxRpi

Welcome to our R wrapper package for Progenetix REST API that leverages the capabilities of Beacon v2 specification. Please note that a stable internet connection is required for the query functionality. This package is aimed to simplify the process of accessing oncogenomic data from Progenetix database.

You can install this package from GitHub using:

```
install.packages("devtools")
devtools::install_github("progenetix/pgxRpi")
```

For accessing metadata of biosamples/individuals, or learning more about filters, get started from the vignette Introduction_1_loadmetadata.

For accessing CNV variant data, get started from this vignette Introduction_2_loadvariants.

For accessing CNV frequency data, get started from this vignette Introduction_3_loadfrequency.

For processing local pgxseg files, get started from this vignette Introduction_4_process_pgxseg.

If you encounter problems, try to reinstall the latest version. If reinstallation doesn't help, please contact us.

Bioconductor

pgxRpi

platforms all | rank 2218 / 2221 | support 0 / 0 | in Bioc devel only
build ok | updated < 1 month | dependencies 144

DOI: 10.18129/B9.bioc.pgxRpi
This is the **development** version of pgxRpi; to use it, please install the devel version of Bioconductor.

R wrapper for Progenetix

Bioconductor version: Development (3.19)

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>

Citation (from within R, enter `citation("pgxRpi")`):

Zhao H, Baudis M (2023). *pgxRpi: R wrapper for Progenetix*. doi:10.18129/B9.bioc.pgxRpi, R package version 0.99.9, https://bioconductor.org/packages/pgxRpi.

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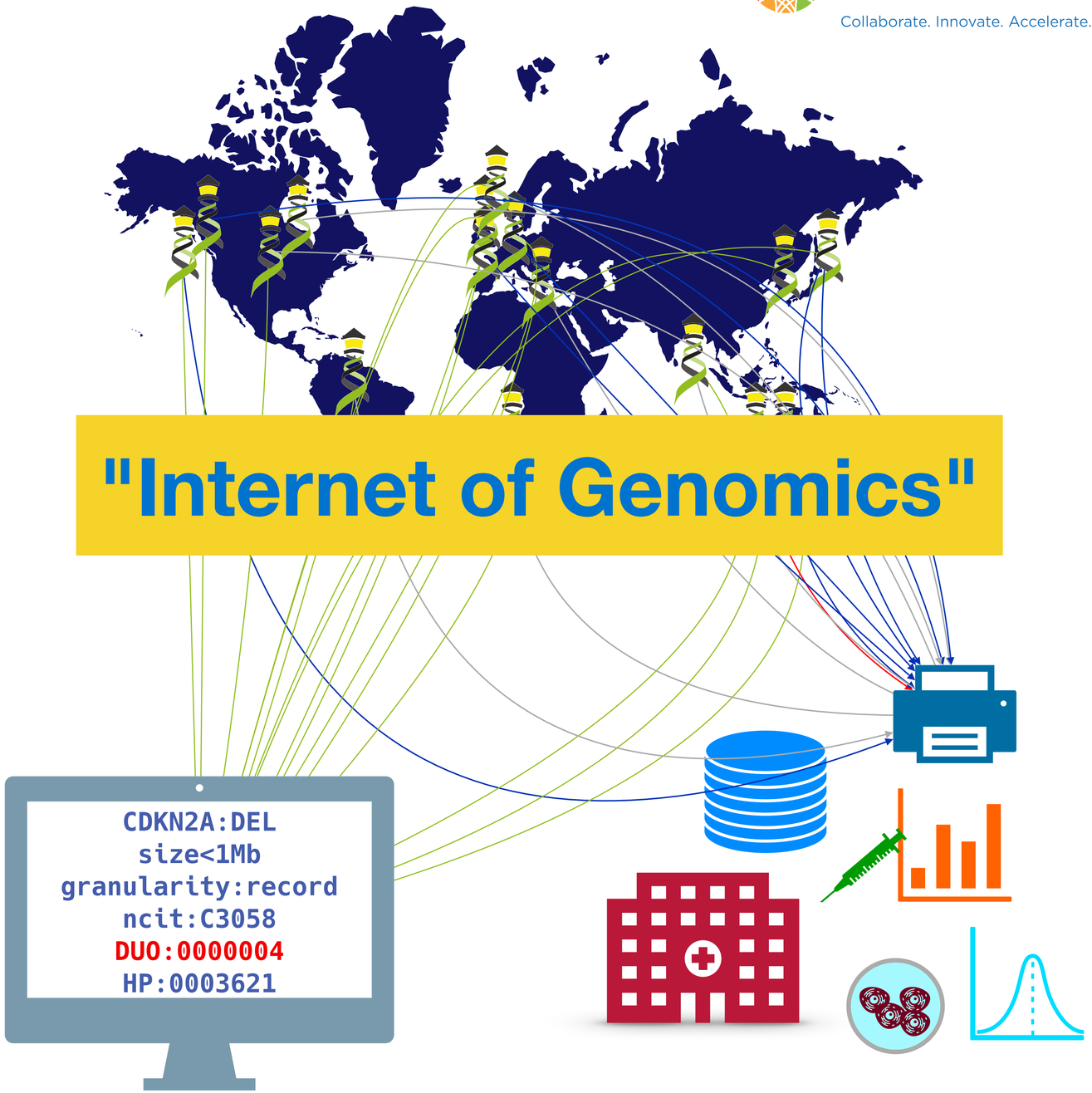
What Can You Do?

- 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 project
- help us with data & code ¯_(ツ)_/¯



The Beacon team through the ages

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