Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.

Genomics Data Federation through Global Alliance for Genomics and Health Standards

Development and Implementation of the GA4GH Beacon Protocol

Michael Baudis

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

1992	Heidelberg	Student of medicine | doctoral thesis in molecular cytogenetics @ DKFZ (Peter Lichter) | resident in clinical hematology/oncology | data, clinical studies & cancer systematics
2001	Stanford	Post-doc in hemato-pathology (Michael Cleary) | molecular mechanisms of leukemogenesis | transgenic models | expression arrays | systematic cancer genome data collection | *Progenetix* website
2003	Gainesville	Assistant professor in paediatric haematology | molecular mechanisms of leukemogenesis | focus on bioinformatics for cancer genome data analysis
2006	Aachen	Research group leader in genetics | genomic array analysis for germline alterations | descriptive analysis of copy number aberration patterns in cancer entities
2007	Zürich	Professor of bioinformatics @ DMLS (2015) | systematic assembly of oncogenomic data | databases and software tools | patterns in cancer genomes | *Progenetix* & *arrayMap* resources | GA4GH | SPHN | ELIXIR

Genomics has seen massive and ongoing changes in technology

200+ Genomic Data Initiatives Globally

Clinical/Genomic Medicine

Research

National

Cohorts

How Many Genomes?

RESEARCH

HEALTHCARE

60M individuals
132.5M sequences

CLINICAL TRIALS

2.7-3M individuals

COHORTS

140M individuals

Global Genomic Data Sharing Can...

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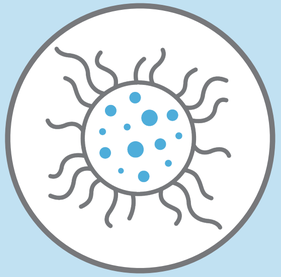


Demonstrate patterns in health & disease

Increase statistical significance of analyses

Lead to “stronger” variant interpretations

Increase accurate diagnosis

Advance precision medicine

Limited Population Diversity in Cancer Studies

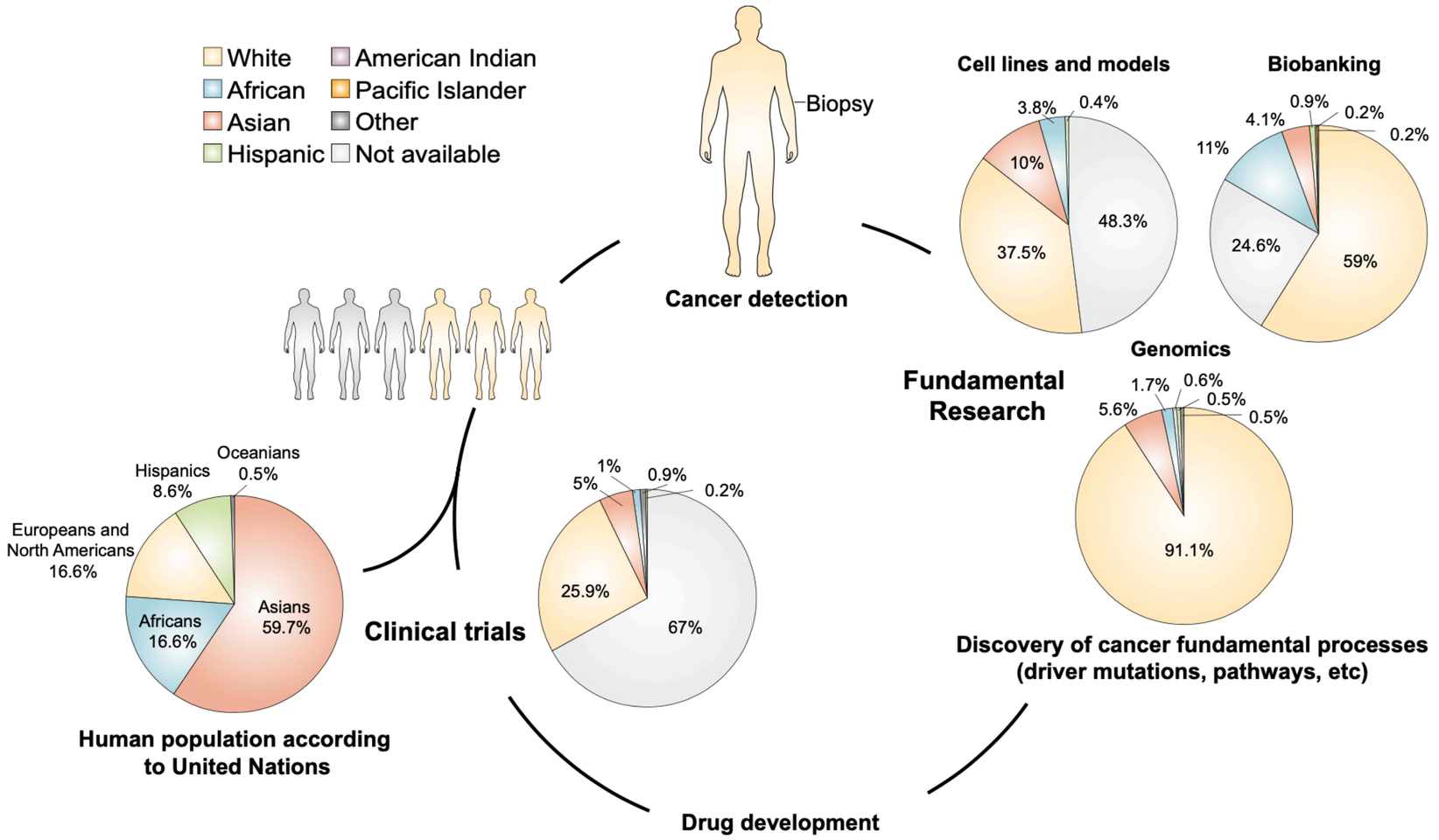
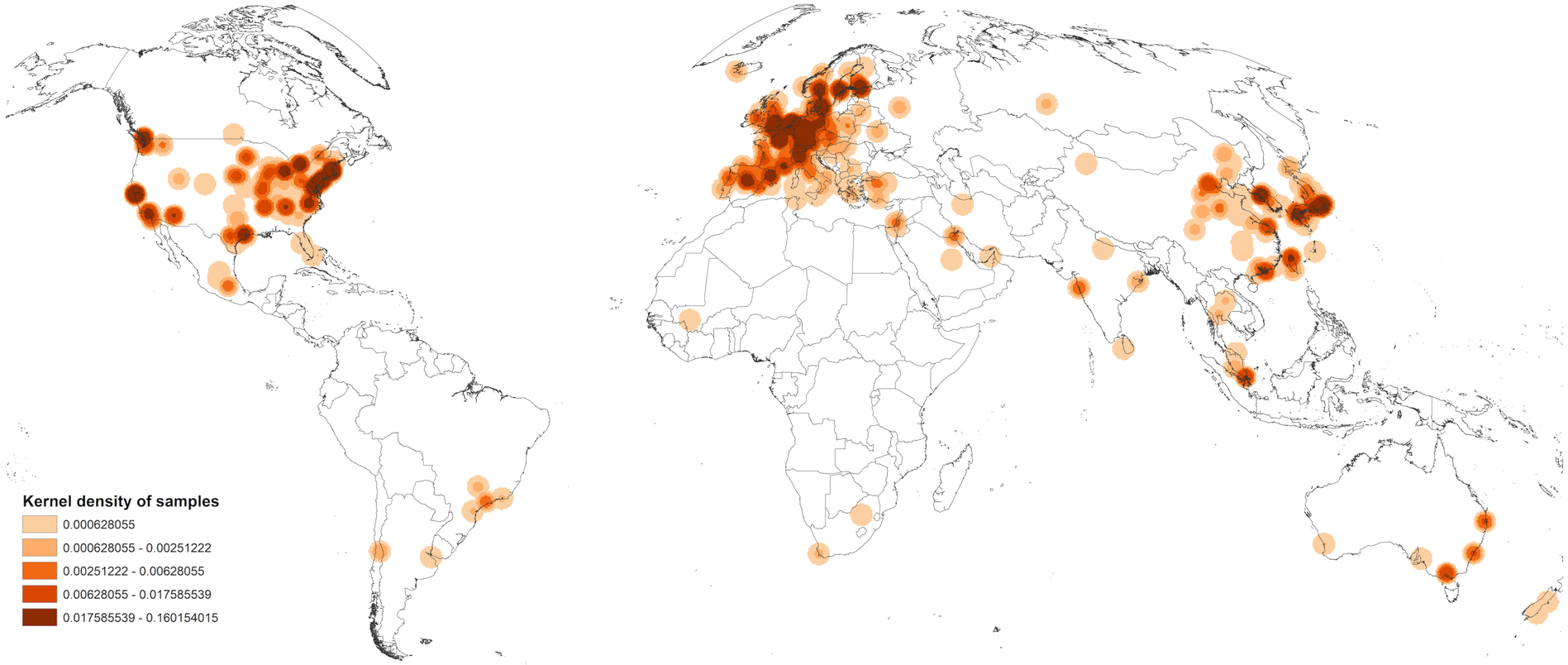


Figure 1. Racial/Ethnic disparities in cancer research. Racial/ethnic inclusion was studied in several aspects of oncological research, from cell lines and patient-derived xenografts to biobanking, genomics and clinical trials.

Guerrero S, López-Cortés A, Indacochea A, et al. Analysis of Racial/Ethnic Representation in Select Basic and Applied Cancer Research Studies. *Sci Rep*. 2018;8(1):13978.

Where does Genomic Data Come From?

Geographic bias in published cancer genome profiling studies



progenetix

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.

Since data is distributed globally, we need interoperable standards to answer research questions

GENOMICS

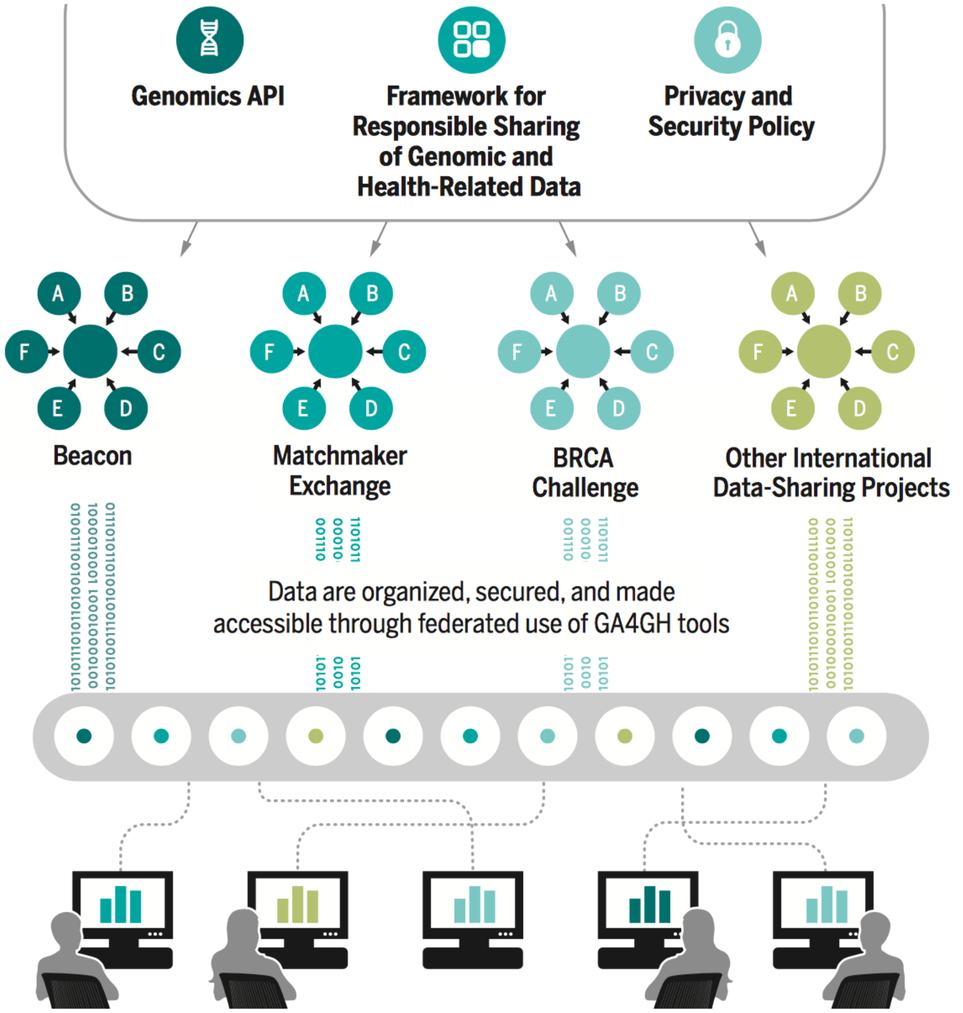
A federated ecosystem for sharing genomic, clinical data

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

GA4GH Timeline

Global Alliance for Genomics & Health

2012 2014 2016 2018 2020

Pre-Launch

GA4GH conceived at first meeting in New York

White Paper published on the need for genomic data sharing

Building Momentum

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

Launch of "GA4GH Connect" and Strategic Roadmap

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

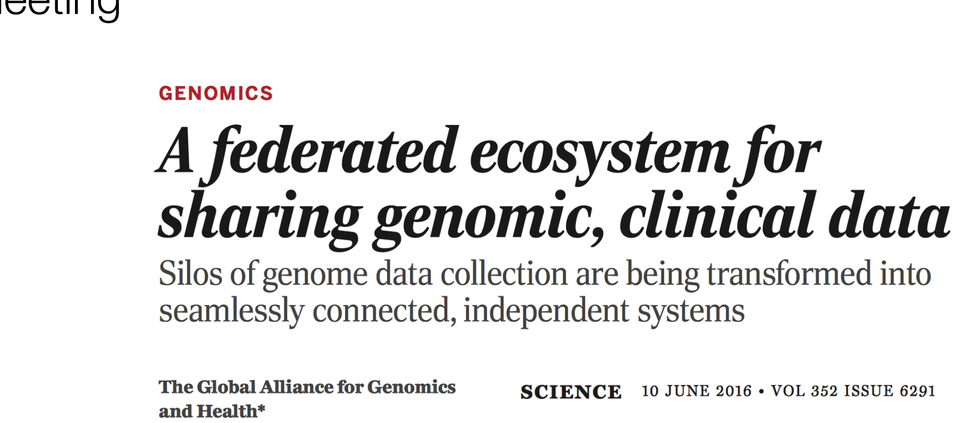
Approved over twenty standards and policies

Over fifty organizations implementing GA4GH standards

The Global Alliance for Genomics and Health

Making genomic data accessible for research and health

- January 2013 - 50 participants from eight countries
- June 2013 - White Paper, over next year signed by 70 “founding” member institutions (e.g. SIB, UZH)
- March 2014 - Working group meeting in Hinxton & 1st plenary in London
- October 2014 - Plenary meeting, San Diego; interaction with ASHG meeting
- June 2015 - 3rd Plenary meeting, Leiden
- September 2015 - GA4GH at ASHG, Baltimore
- October 2015 - DWG / New York Genome Centre
- April 2016 - Global Workshop @ ICHG 2016, Kyoto
- October 2016 - 4th Plenary Meeting, Vancouver
- May 2017 - Strategy retreat, Hinxton
- October 2017 - 5th plenary, Orlando
- May 2018 - Vancouver
- October 2018 - 6th plenary, Basel
- May 2019 - GA4GH Connect, Hinxton
- October 2019 - 7th Plenary, Boston
- October 2020 - Virtual Plenary, June 2021 - Virtual Connect ...
- October 2021 - Virtual Plenary ...
- September 2022 - 10th Plenary, Barcelona
- ...



22 SEPTEMBER 2022 | BARCELONA, SPAIN

GA4GH 10th Plenary

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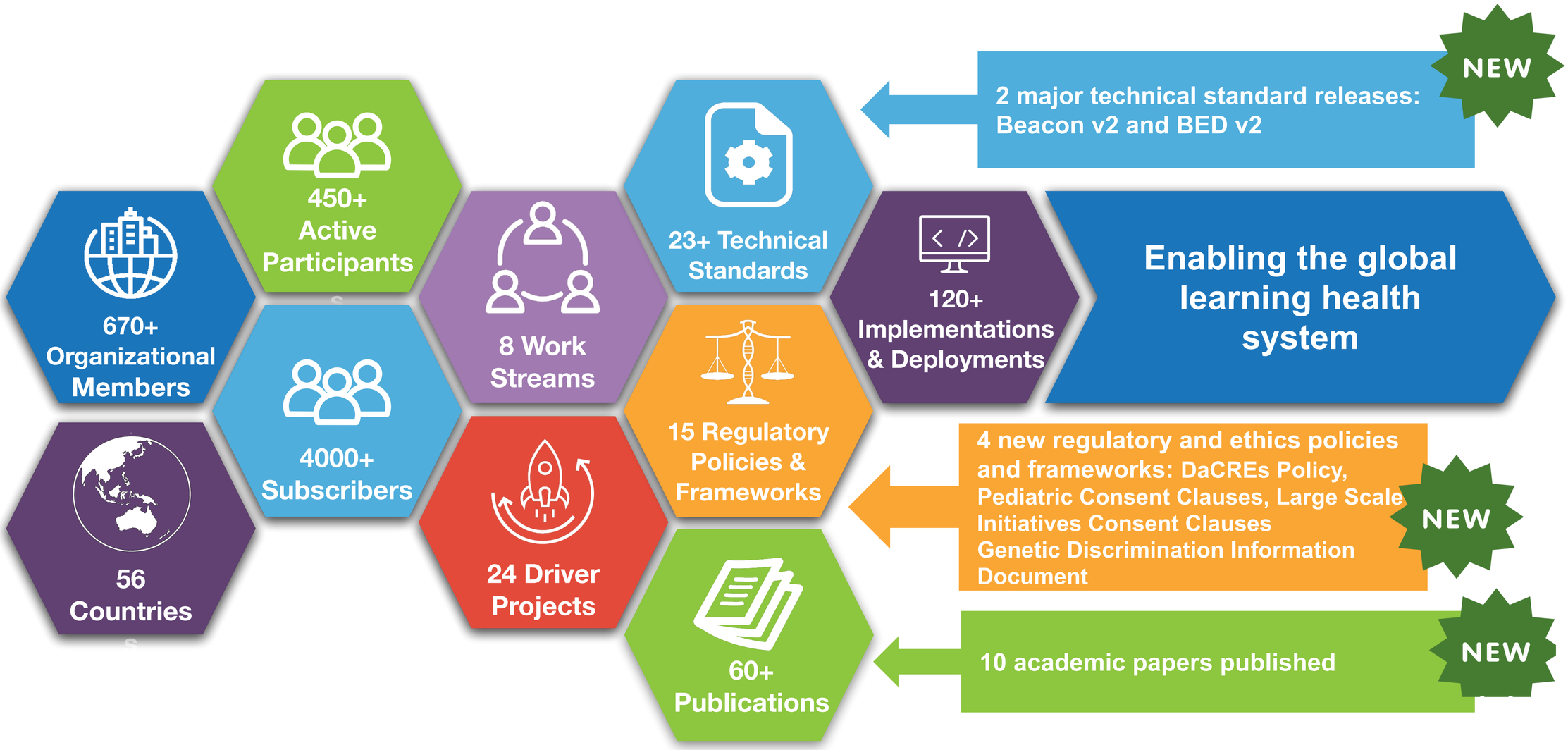
The GA4GH ecosystem and outputs

Global Alliance for Genomics & Health



Federation

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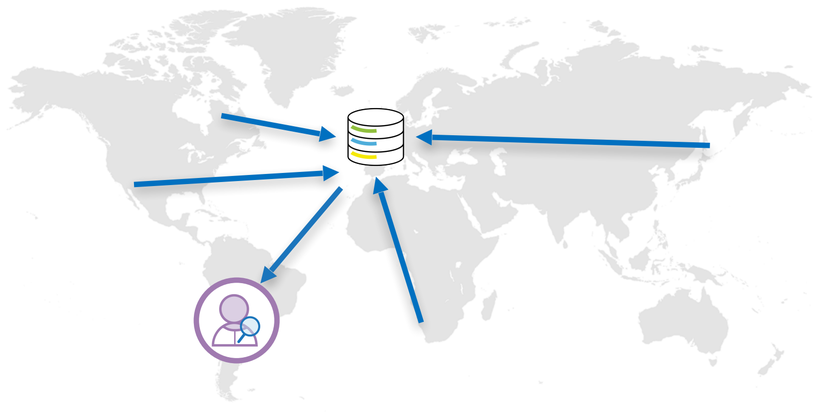
Central Database

Basic research consented for data sharing

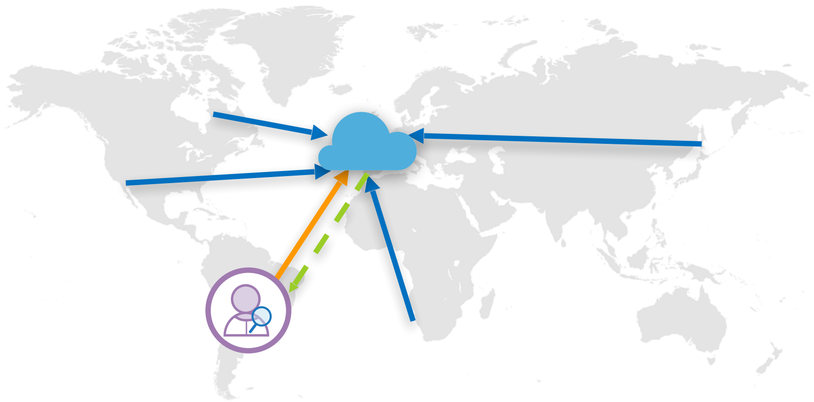


Aggregate data globally

Download, analyze locally

Secure Cloud

Large scale research datasets

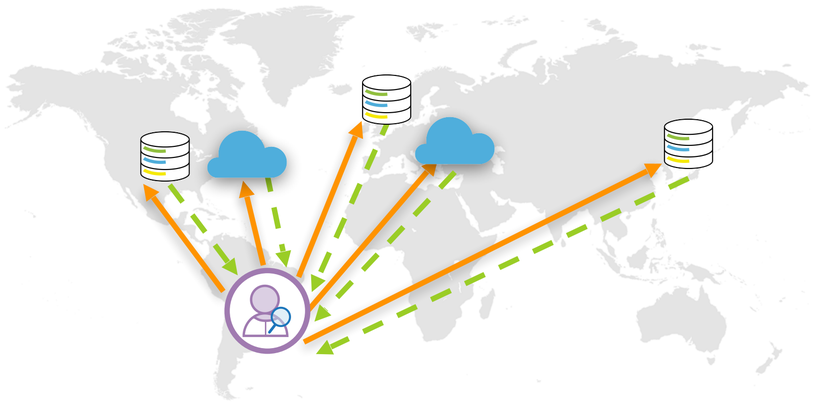


Aggregate data globally

Analyze centrally in secure cloud

Federated Approach

Connecting national genomics initiatives



Host data locally

Analyze data remotely and collate results

User — Data transmission — Data Visiting - - - Results sent back to user

Federation: a solution for data analysis

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No data copying or transfer

Data can remain in original jurisdiction

Ownership and access control retained

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Collaborate. Innovate. Accelerate.

GA4GH Standards Development

GA4GH Roadmap Development Process

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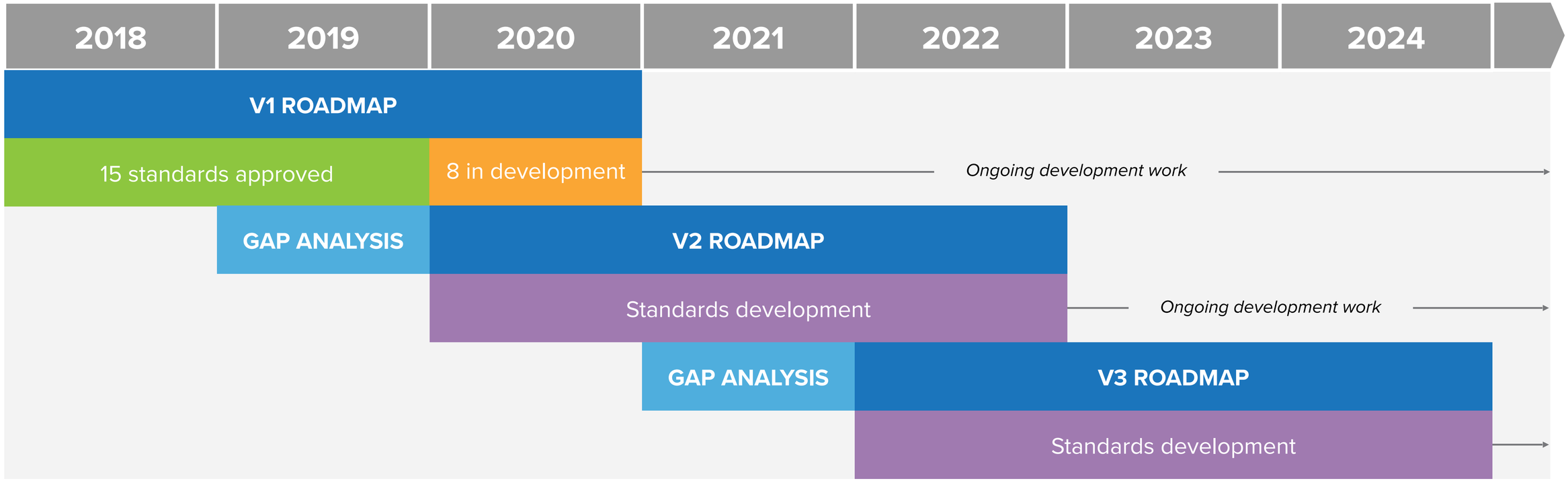

Strategic Roadmap Alignment to F.A.I.R. Principles

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Findable

- Beacon API
- Data Use Ontology
- refget API
- Search API
- Service Registry Prototype
- Tool Registry Service (TRS) API

Accessible

- Authentication and Authorization Infrastructure
- Data Repository Service (DRS) API
- Data Use Ontology
- GA4GH Passports

Interoperable

- Phenopackets/FIHR
- Pedigree Representation
- Genetic variant file formats
- Read file formats
- RNAget API
- Crypt4GH
- Variant Annotation
- Variant Representation
- Task Execution Service (TES) API
- Testbed interoperability demonstration
- Tool Registry Service (TRS) API
- Workflow Execution Service (WES) API

Reusable

- htsget streaming API
- refget API
- Variant Annotation
- Workflow Execution Service (WES) API
- Testbed interoperability demonstration

Alignment with Other Standards Organizations

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Research | Clinical Research | Clinical Care

HL7 FHIR

SNOMED

cdisc

ISO TC276

ISO TC215/ SC1

GA4GH Work Streams

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Clinical and Phenotypic Data Capture

Cloud

Data Use and Researcher Identities

Discovery

Genomic Knowledge Standards

Large Scale Genomics

Regulatory and Ethics

Data Security

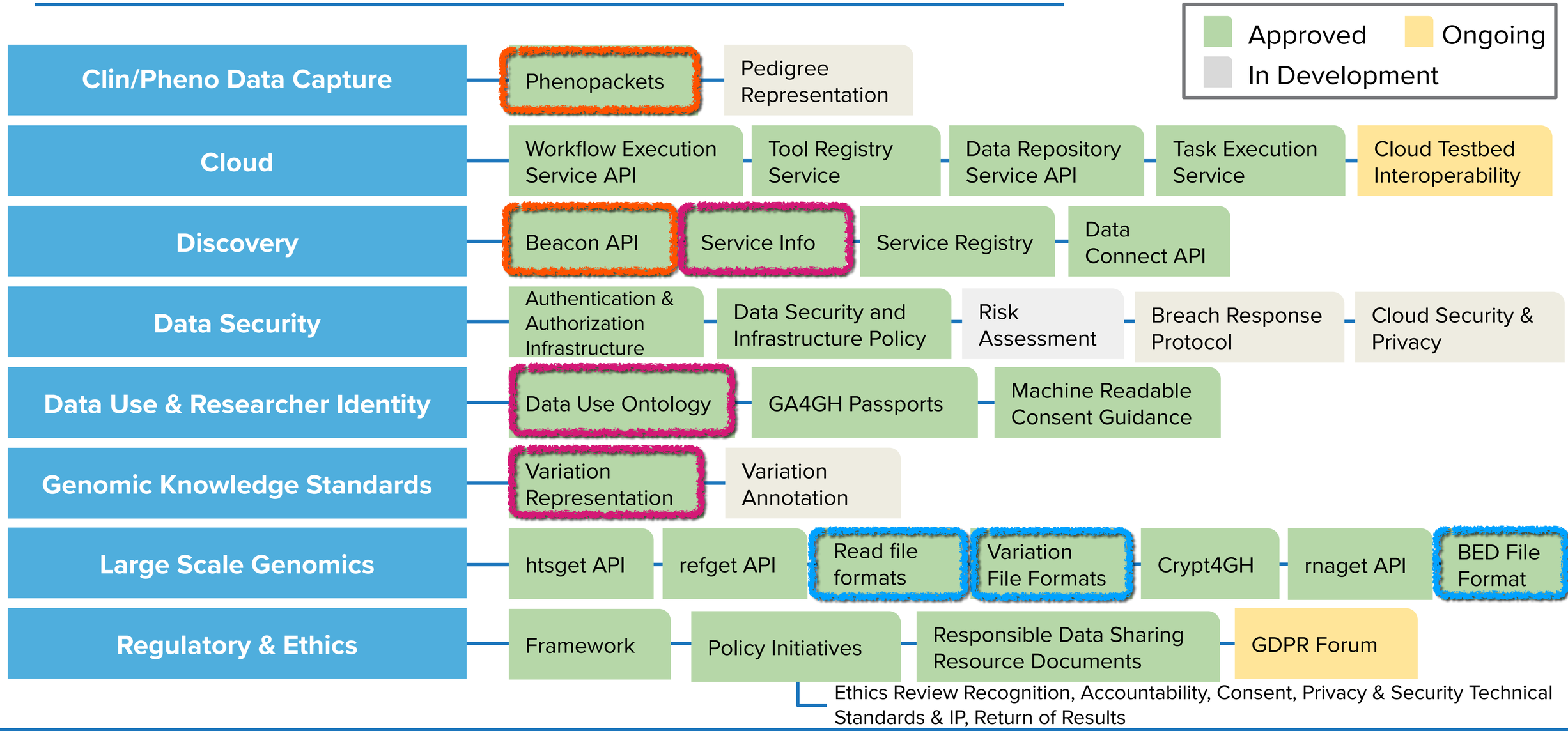
GA4GH 2020-2022 Strategic Roadmap

Global Alliance for Genomics & Health

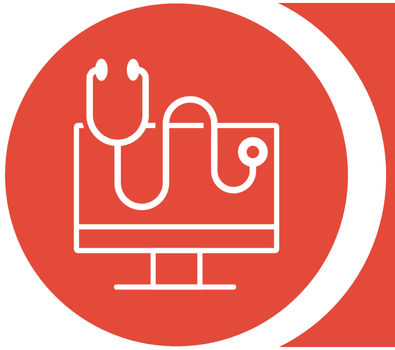
- **Clin/Pheno Data Capture**
  - Phenopackets
  - Pedigree Representation
- **Cloud**
  - Workflow Execution Service API
  - Tool Registry Service
  - Data Repository Service API
  - Task Execution Service
  - Cloud Testbed Interoperability
- **Discovery**
  - Beacon API
  - Service Info
  - Service Registry
  - Data Connect API
- **Data Security**
  - Authentication & Authorization Infrastructure
  - Data Security and Infrastructure Policy
  - Risk Assessment
  - Breach Response Protocol
  - Cloud Security & Privacy
- **Data Use & Researcher Identity**
  - Data Use Ontology
  - GA4GH Passports
  - Machine Readable Consent Guidance
- **Genomic Knowledge Standards**
  - Variation Representation
  - Variation Annotation
- **Large Scale Genomics**
  - htsget API
  - refget API
  - Read file formats
  - Variation File Formats
  - Crypt4GH
  - rnaget API
  - BED File Format
- **Regulatory & Ethics**
  - Framework
  - Policy Initiatives
    - Ethics Review Recognition, Accountability, Consent, Privacy & Security Technical Standards & IP, Return of Results
  - Responsible Data Sharing Resource Documents
  - GDPR Forum

GA4GH 2020-2022 Strategic Roadmap

Global Alliance for Genomics & Health



Global Alliance for Genomics & Health



Support clinical adoption of genomics through information models and standards for describing and exchanging clinical phenotypes.

Proposed Solution

Standardize exchange formats for representing clinical data and describing clinical phenotypes.

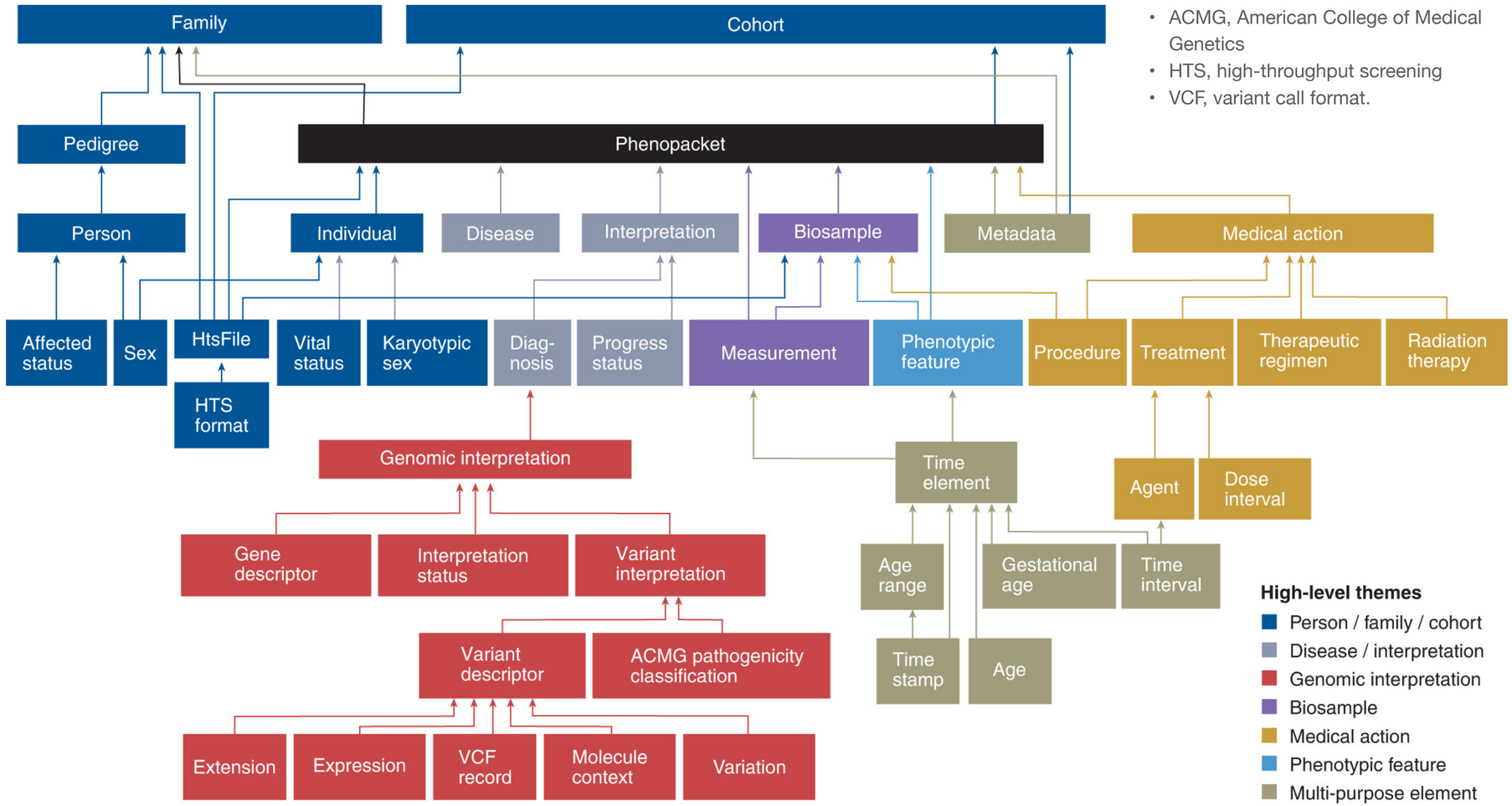
GA4GH Deliverables

- Phenopackets
- Pedigree
- New project: Cohort Representation

The GA4GH Phenopackets v2 Standard
A Computable Representation of Clinical Data

The GA4GH Phenopacket schema consists of several optional elements, each containing information about a certain topic, such as phenotype, variant or pedigree. An element can contain other elements, which allows a hierarchical representation of data.

For instance, Phenopacket contains elements of type *Individual*, *PhenotypicFeature*, *Biosample* and so on. Individual elements can therefore be regarded as **building blocks** of larger structures.



Jacobsen JOB, Baudis M, Baynam GS, Beckmann JS, Beltran S, Buske OJ, Callahan TJ, *et al.* 2022.
The GA4GH Phenopacket Schema Defines a Computable Representation of Clinical Data.
Nature Biotechnology 40 (6): 817–20.

Phenopackets Available via GA4GH and ISO

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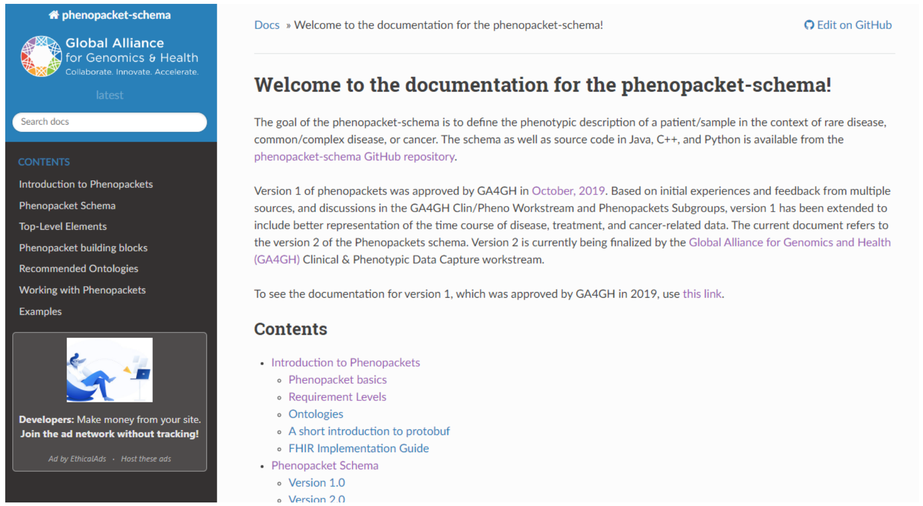
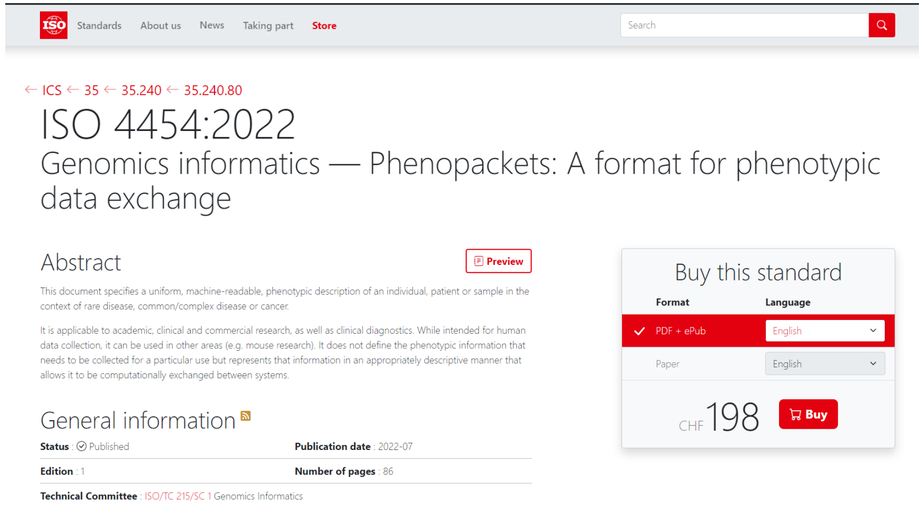
GA4GH



https://bit.ly/PhenopacketsDocs

ISO



https://www.iso.org/standard/79991.html

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Facilitate the discovery and utilization of data sources and services

Proposed Solution

Establish a unified interface for aggregating data sources and services that can be crawled and indexed

GA4GH Deliverables

- Beacon API
- Data Connect API
- Service Info & Service Registry
- SchemaBlocks {S}[B]

GA4GH {S}[B]

SchemaBlocks

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

schemablocks.org

Service Info and Service Registry APIs

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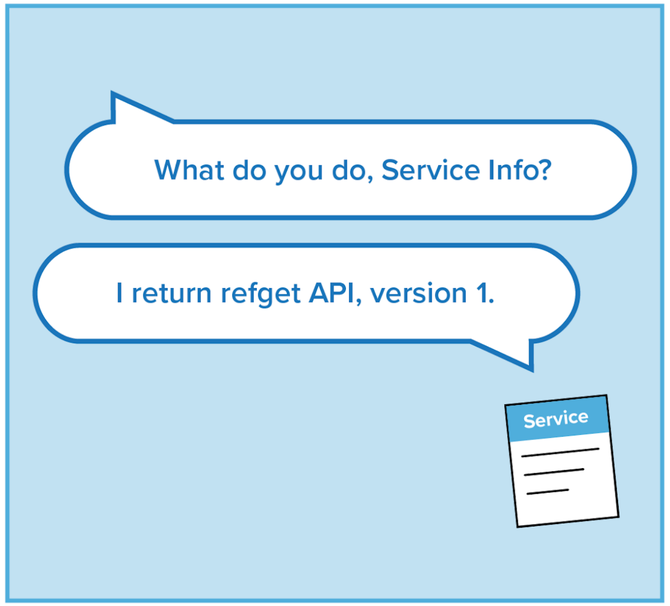
The Service Info and Registry APIs are minimalistic, light-weight APIs that provide a standard format for describing and listing genomics web services along with their metadata.

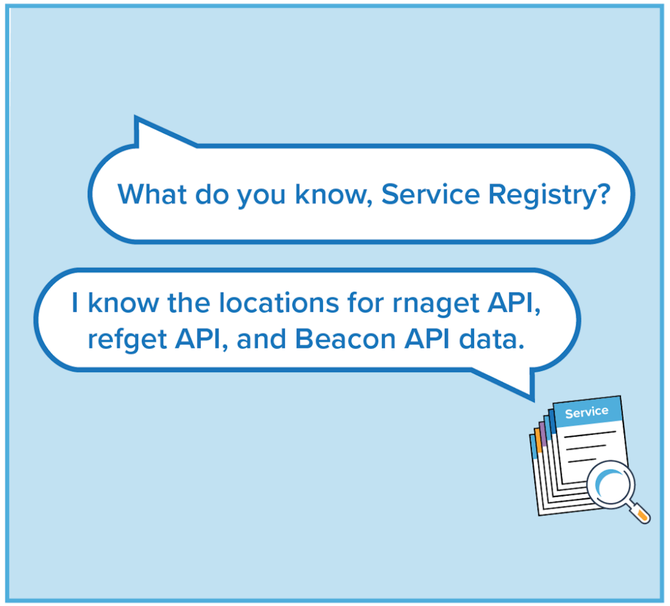
Approved: January 22, 2020

Example Users

elixir

AUTISM SHARING INITIATIVE



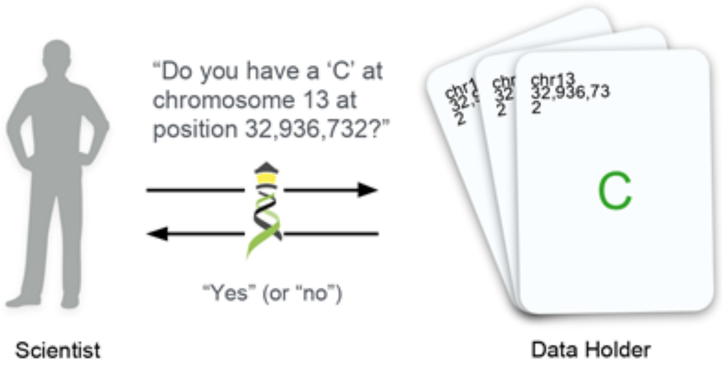


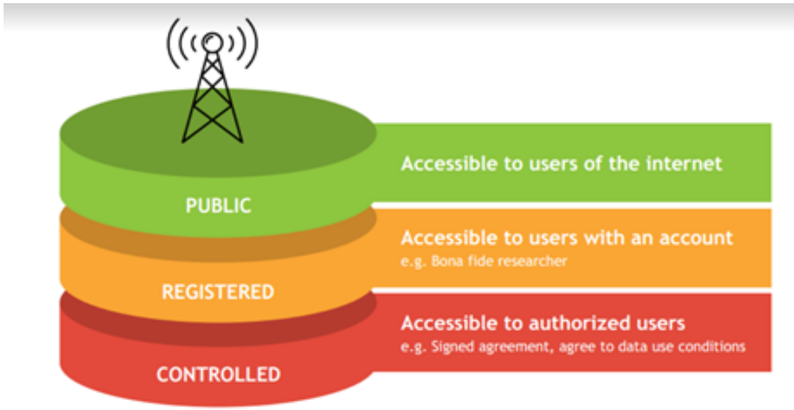
The GA4GH Beacon Protocol

A GA4GH standard for genomics data discovery (and exchange)

The Beacon API can be implemented as a web-accessible service that users may query for information about a specific allele.

Approved: October 3, 2018





Example Users

EMBL-EBI

Australian Genomics

SciLifeLab

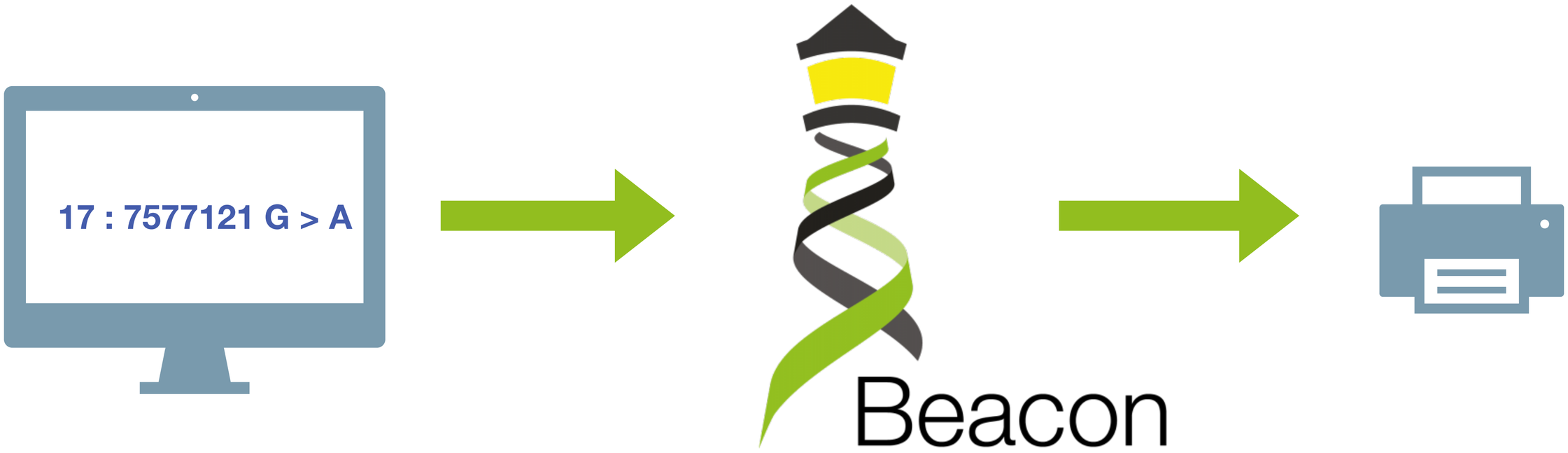
elixir

UNIVERSITY OF CALIFORNIA SANTA CRUZ

BROAD INSTITUTE

EUROPEAN GENOME-PHENOME ARCHIVE

International Cancer Genome Consortium



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**.

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*** …

ELIXIR - Making Beacons Biomedical

- Authentication to enable non-aggregate, patient derived datasets
  - ELIXIR AAI with compatibility to other providers (OAuth...)
- Scoping queries through "biodata" parameters
- Extending the queries towards clinically ubiquitous variant formats
  - cytogenetic annotations, named variants, variant effects
- Beacons as part of local, secure environments
  - local EGA ...
- Beacon queries as entry for **data delivery**
  - handover to stream and download using htsget, VCF, EHRs
- Interacting with EHR standards
  - FHIR translations for queries and handover ...

2016 and beyond ...

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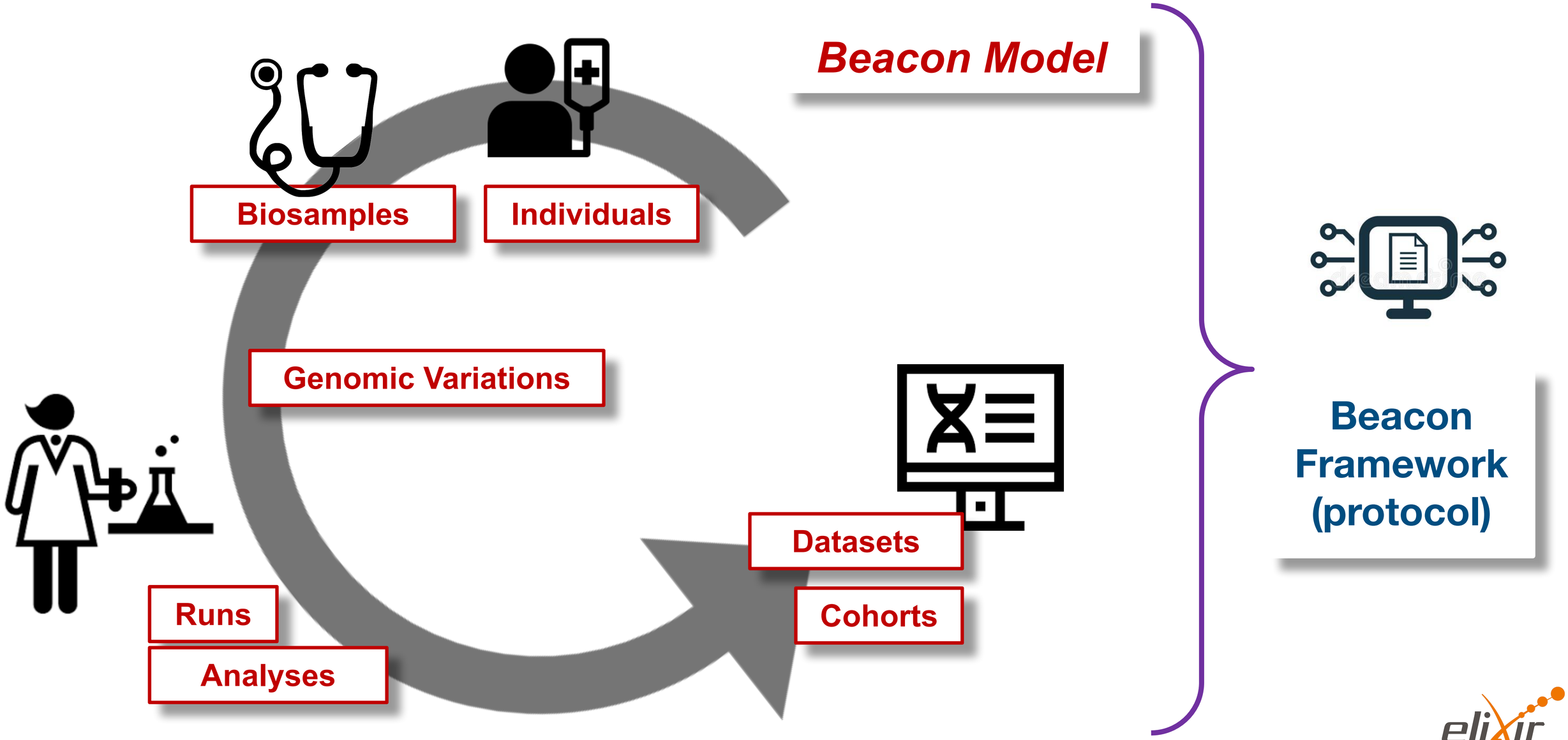
elixir

	Beacon v1 Development	Beacon v2 Development	Related ...
2014	**GA4GH founding event; Jim Ostell proposes Beacon concept with "more features… version 2"**		
2015	• beacon-network.org aggregator created by DNAstack • Beacon v0.3 release		• ELIXIR starts Beacon project support
2016	• 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	• Beacon hackathon Stockholm; settling on "filters" • Barcelona goes Zurich developers meeting • Beacon API v2 Kick off • adopting "handover" concept	• new Beacon website (March)
2019	• ELIXIR Beacon Network	• "Scouts" teams working on different aspects - filters, genomic variants, compliance ... • discussions w/ clinical stakeholders	• Beacon publication at Nature Biotechnology
2020		• framework + models concept implemented • range and bracket queries, variant length • starting of GA4GH review process	
2021		• further changes esp. in default model, aligning with Phenopackets and VRS • unified beacon-v2 code & docs repository • **Beacon v2 approved at Apr GA4GH Connect**	
2022			• *docs.genomebeacons.org*

Beacon v2

docs.genomebeacons.org

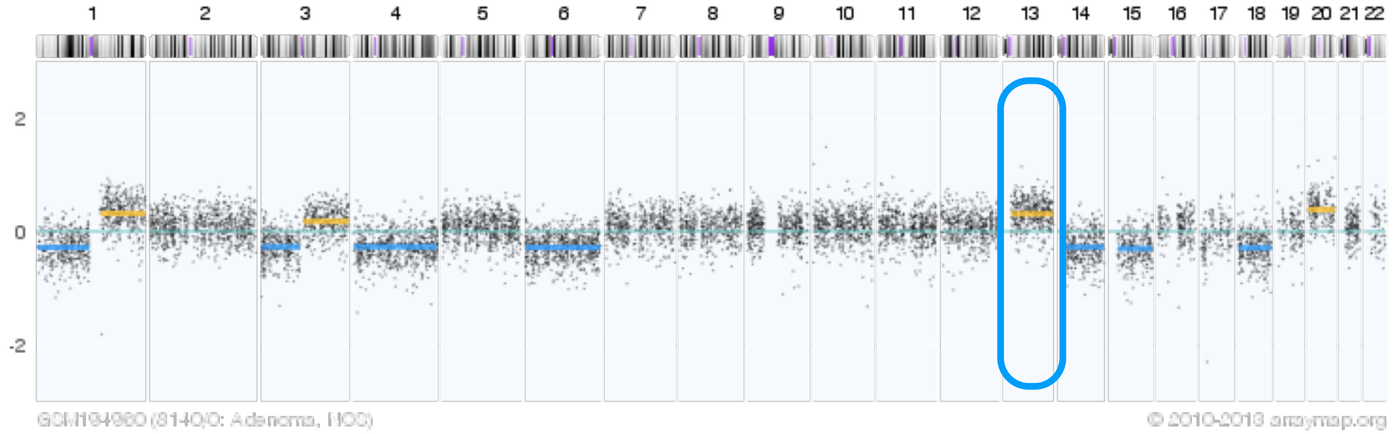


Progenetix and GA4GH Beacon

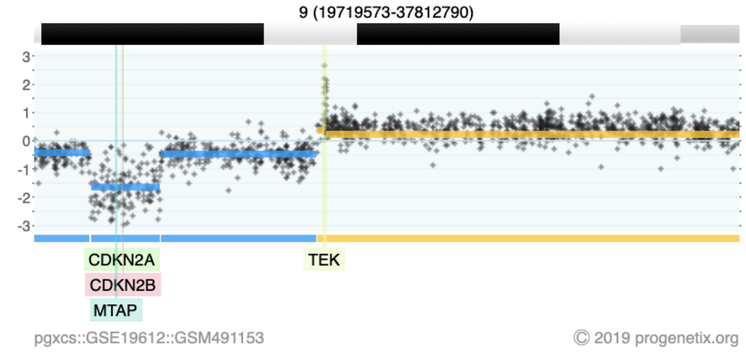
Implementation driven development of a GA4GH standard

progenetix

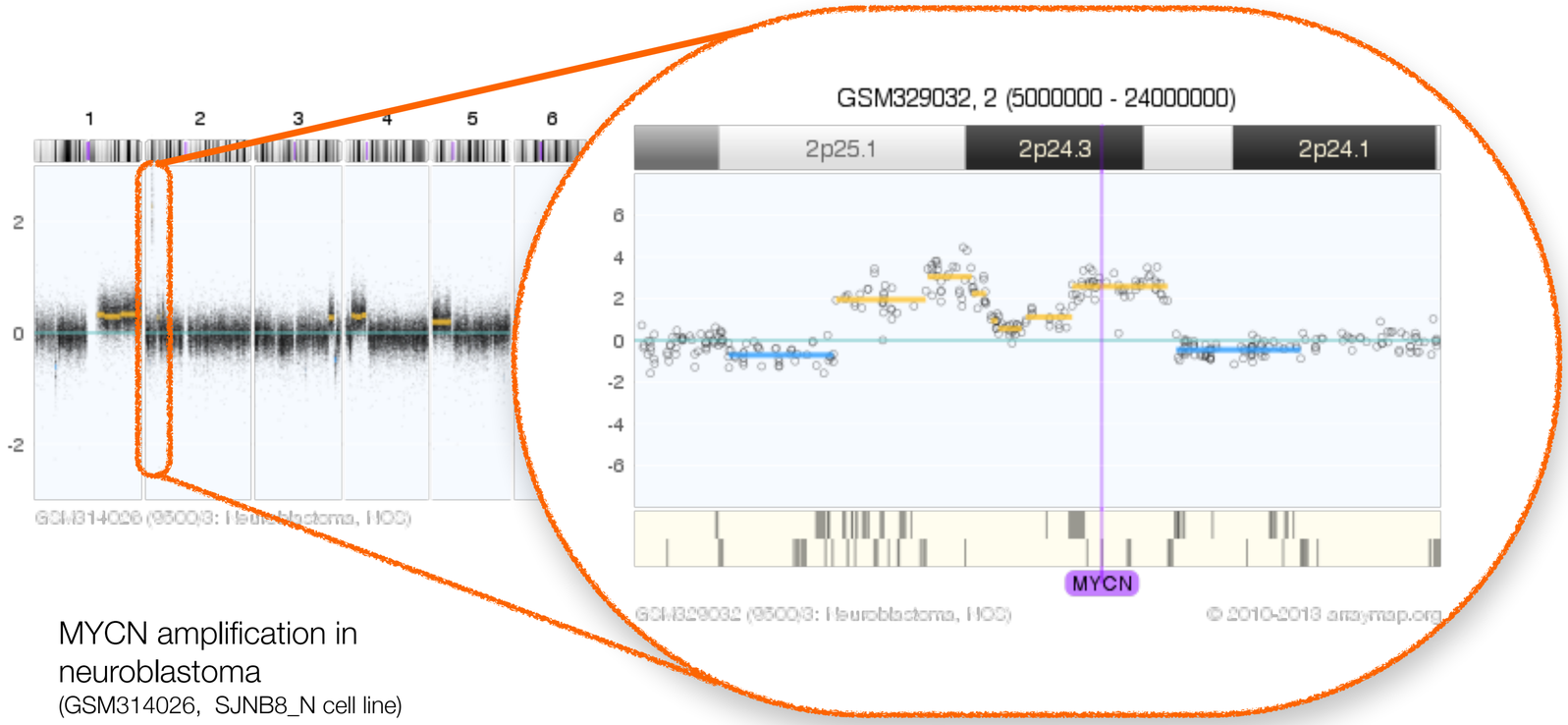
Somatic Copy Number Variations



Gain of chromosome arm 13q in colorectal carcinoma



2-event, homozygous deletion in a Glioblastoma



MYCN amplification in neuroblastoma
(GSM314026, SJNB8_N cell line)

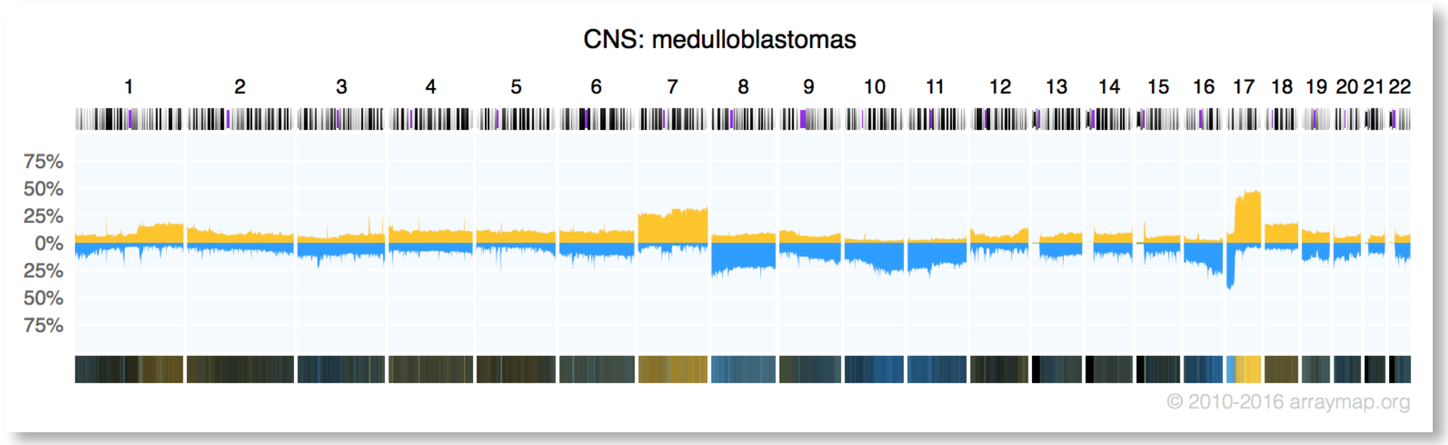
low level/**high level** copy number alterations (CNAs)

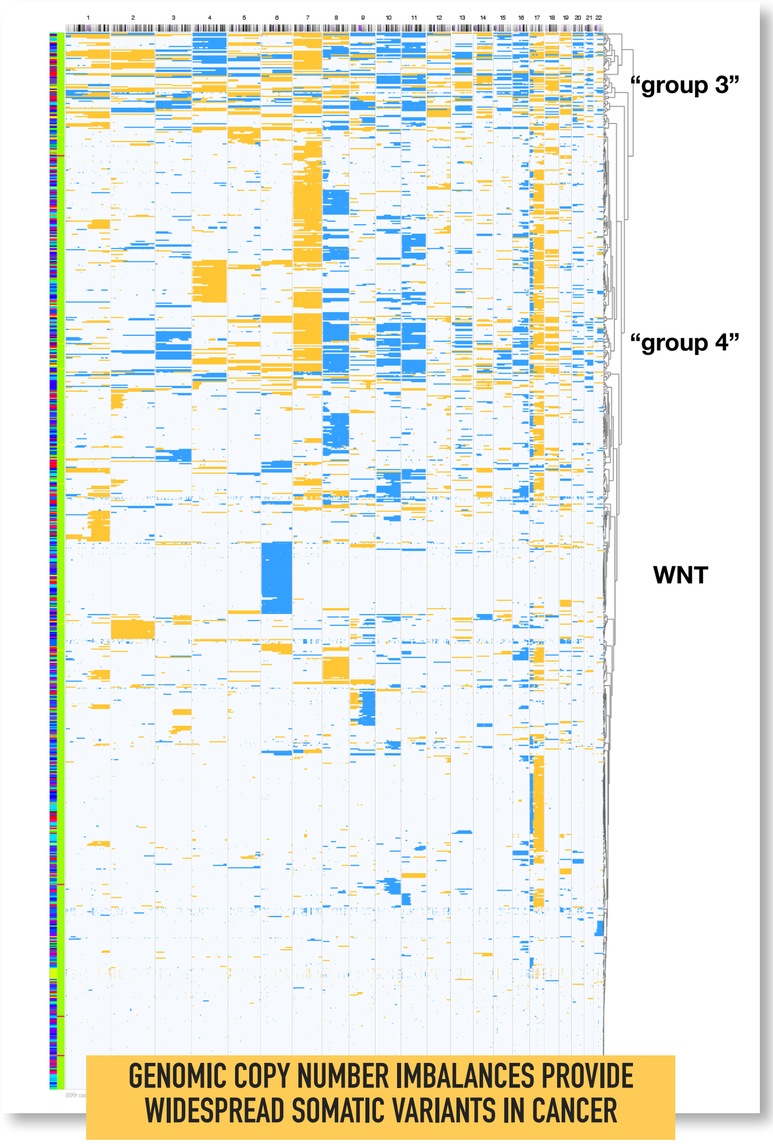
progenetix

Somatic CNVs In Cancer

Recurrent mutation patterns

How can those patterns be used for classification and determination of biological mechanisms?





A genomic copy number histogram for malignant medulloblastomas, the most frequent type of pediatric brain tumors, displaying regions of genomic duplications and deletions. These can be decomposed into individual tumor profiles which segregate into several clusters of related mutation patterns with functional relevance and clinical correlation.

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Progenetix in 2022

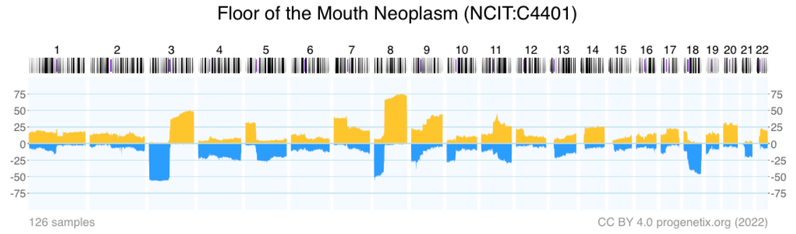
Cancer Genomics Reference Resource

- open resource for curated oncogenomic profiles
- >116'000 cancer CNV profiles, from >800 types
- majority of data from genomic arrays with ~50% overall from SNP platforms with original data re-processing
- standardized encodings (e.g. NCIt, ICD-O 3)
- identifier mapping for PMID, GEO, Cellosaurus, TCGA, cBioPortal where appropriate
- core biosample and technical metadata where accessible (TNM, sex, survival ...)
- publication database and code mapping services

progenetix

Cancer genome data @ progenetix.org

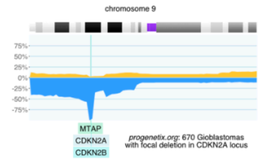
The Progenetix database provides an overview of mutation data in cancer, with a focus on copy number abnormalities (CNV / CNA), for all types of human malignancies. The data is based on *individual sample data* from currently **142063** samples.



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.

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 in 2022

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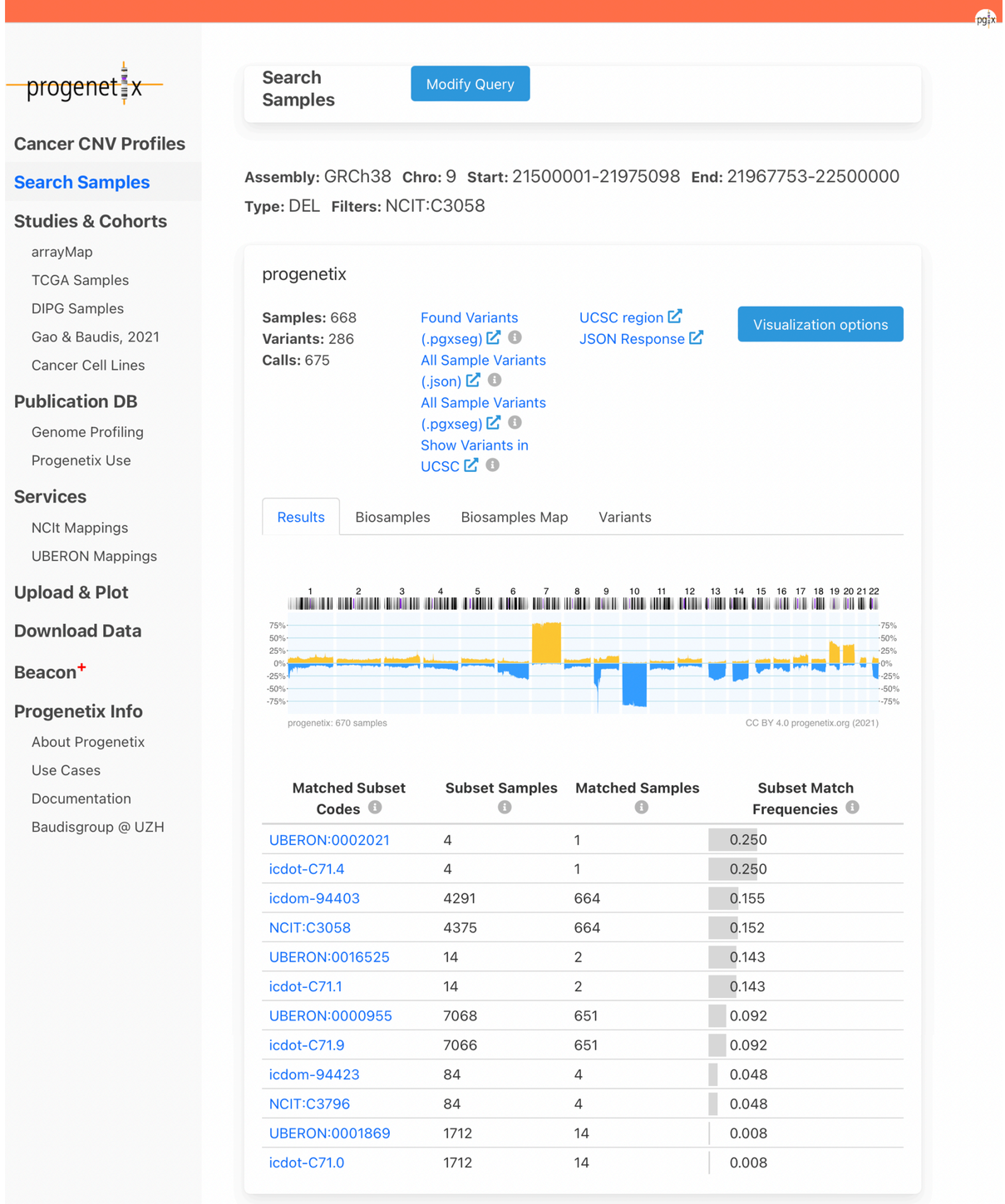

Progenetix in 2022

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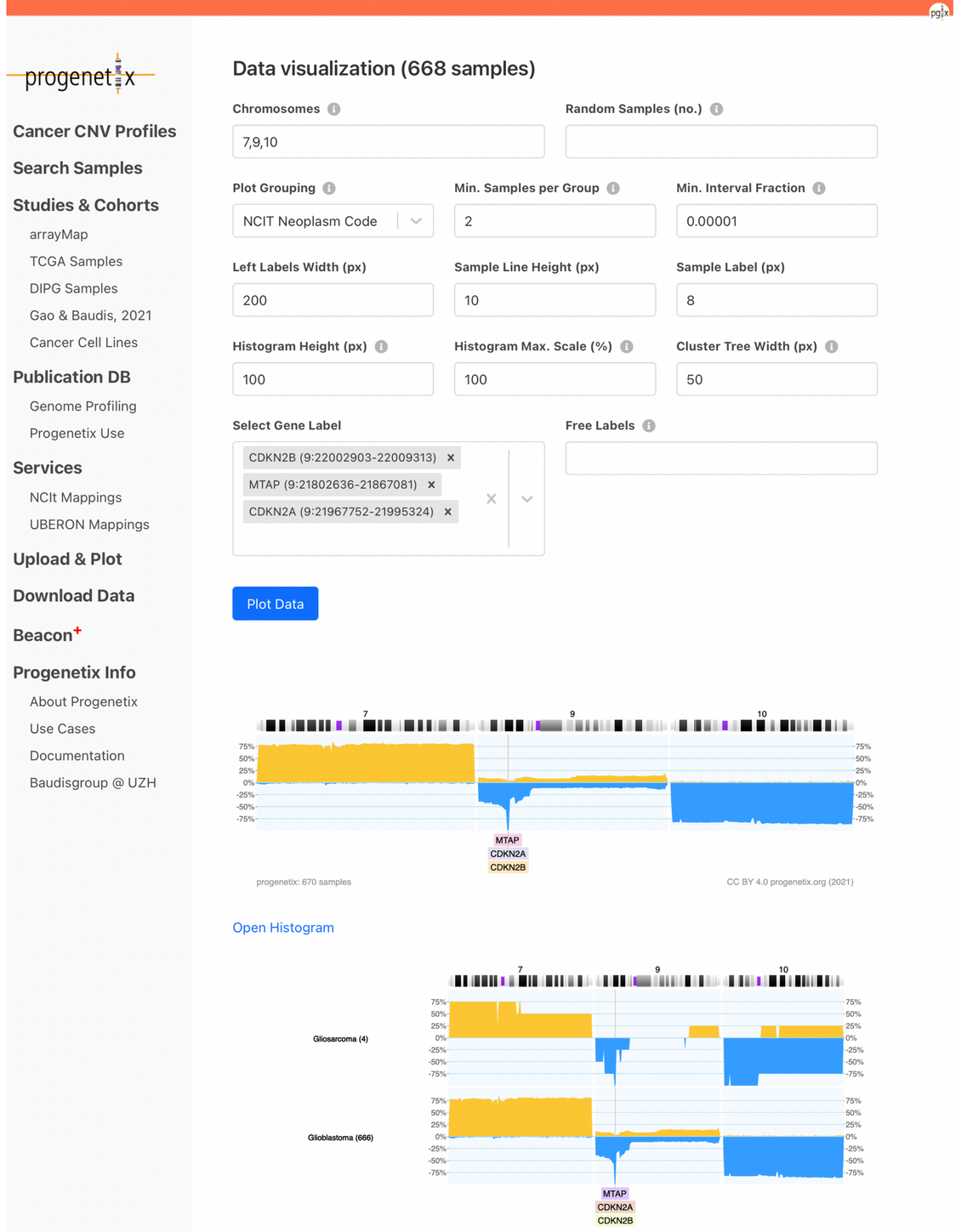

DX Ontologies

Example: Hierarchical NCIt Neoplasm Core replaces heterogeneous primary annotations

- heterogeneous and inconsistent diagnostic annotations are common in clinical reports and research studies ("text", ICD-10, ICD-O 3, OncoTree, domain-specific)
- highly **variable granularity** of annotations as major road block for large scale data integration
  - "Colorectal Cancer" or "Rectal Mucinous Adenoca."
- initiatives and services such as **Phenopackets**, MONDO, OXO ... rely on and/or provide mappings to hierarchical ontologies
- consistent CURIEs were instrumental in the development & testing of the Beacon v2 "Filters" paradigm
  - final "Filtering Term" object dev. by Tim Beck, U. of Leicester

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NCIt Neoplasm Core coded display (excerpt) for samples in the Progenetix cancer genome data resource allows sample selection on multiple hierarchy levels →

Collapse all | Expand | All

Subsets	Samples
NCIT:C3262: Neoplasm	88844
NCIT:C3263: Neoplasm by Site	84747
NCIT:C156482: Genitourinary System Neoplasm	11616
NCIT:C156483: Benign Genitourinary System Neoplasm	219
NCIT:C4893: Benign Urinary System Neoplasm	90
NCIT:C4778: Benign Kidney Neoplasm	90
NCIT:C159209: Kidney Leiomyoma	1
NCIT:C4526: Kidney Oncocytoma	82
NCIT:C8383: Kidney Adenoma	7
NCIT:C7617: Benign Reproductive System Neoplasm	129
NCIT:C4934: Benign Female Reproductive System Neoplasm	129
NCIT:C2895: Benign Ovarian Neoplasm	58
NCIT:C4510: Benign Ovarian Epithelial Tumor	58
NCIT:C40039: Benign Ovarian Mucinous Tumor	58
NCIT:C4512: Ovarian Mucinous Cystadenoma	58
NCIT:C4060: Ovarian Cystadenoma	58
NCIT:C4512: Ovarian Mucinous Cystadenoma	58
NCIT:C3609: Benign Uterine Neoplasm	71
NCIT:C3608: Benign Uterine Corpus Neoplasm	71
NCIT:C3434: Uterine Corpus Leiomyoma	71
NCIT:C156484: Malignant Genitourinary System Neoplasm	11171
NCIT:C157774: Metastatic Malignant Genitourinary System Neoplasm	2
NCIT:C146893: Metastatic Genitourinary System Carcinoma	2
NCIT:C8946: Metastatic Prostate Carcinoma	2
NCIT:C164141: Genitourinary System Carcinoma	10561
NCIT:C146893: Metastatic Genitourinary System Carcinoma	2
NCIT:C8946: Metastatic Prostate Carcinoma	2
NCIT:C3867: Fallopian Tube Carcinoma	19

Ontologies and Classifications

Services: Ontologymaps (NCIt)

The **ontologymaps** service provides equivalency mapping between ICD-O and other classification systems, notably NCIt and UBERON. It makes use of the sample-level mappings for NCIT and ICD-O 3 codes developed for the individual samples in the Progenetix collection.

NCIT and ICD-O 3

While NCIT treats diseases as **histologic** and **topographic** described entities (e.g. **NCIT:C7700**: **Ovarian adenocarcinoma**), these two components are represented separately in ICD-O, through the **Morphology** and **Topography** coding arms (e.g. here **8140/3** + **C56.9**).

More documentation with focus on the API functionality can be found on the documentation pages.

The data of all mappings can be retrieved trough this API call: {JSON↗}

Code Selection

NCIT:C4337: Mantle Cell Lymphoma

Optional: Limit with second selection

Matching Code Mappings {JSON↗}

NCIT:C4337: Mantle Cell Lymphoma	pgx:icdom-96733: Mantle cell lymphoma	pgx:icdot-C77.9: Lymph nodes, NOS
NCIT:C4337: Mantle Cell Lymphoma	pgx:icdom-96733: Mantle cell lymphoma	pgx:icdot-C18.9: large intestine, excl. rectum and rectosigmoid junction
NCIT:C4337: Mantle Cell Lymphoma	pgx:icdom-96733: Mantle cell lymphoma	pgx:icdot-C42.2: Spleen

More than one code groups means that either mappings need refinements (e.g. additional specific NCIT classes for ICD-O T topographies) or you started out with an unspecific ICD-O M class and need to add a second selection.

In Progenetix all cancer diagnoses are coded to both NCIt neoplasm codes and ICD-O 3 Morphology + Topography combinations. The matched mappings are provided as lookup-service since neither an official ICD-O ontology nor such a "disease defined by ICD-O M+T" concept is codified anywhere.

List of filters recognized by different query endpoints

Public Ontologies with CURIE-based syntax

CURIE prefix	Code/Ontology	Examples
NCIT	NCIt Neoplasm[1]	NCIT:C27676
HP	HPO[2]	HP:0012209
PMID	NCBI Pubmed ID	PMID:18810378
geo	NCBI Gene Expression Omnibus[3]	geo:GPL6801, geo:GSE19399, geo:GSM491153
arrayexpress	EBI ArrayExpress[4]	arrayexpress:E-MEXP-1008
cellosaurus	Cellosaurus - a knowledge resource on cell lines [5]	cellosaurus:CVCL_1650
UBERON	Uberon Anatomical Ontology[6]	UBERON:0000992
cbioportal	cBioPortal[9]	cbioportal:msk_impact_2017

Private filters

Since some classifications cannot directly be referenced, and in accordance with the upcoming Beacon v2 concept of "private filters", Progenetix uses additionally a set of structured non-CURIE identifiers.

For terms with a `pgx` prefix, the identifiers.org resolver will

Filter prefix / local part	Code/Ontology	Example
pgx:icdom-...	ICD-O 3[7] Morphologies (Progenetix)	pgx:icdom-81703
pgx:icdot...	ICD-O 3[7] Topographies(Progenetix)	pgx:icdot-C04.9
TCGA	The Cancer Genome Atlas (Progenetix)[8]	TCGA-000002fc-53a0-420e-b2aa-a40a358bba37
pgx:pgxcohort-...	Progenetix cohorts [10]	pgx:pgxcohort-arraymap

Progenetix in 2022

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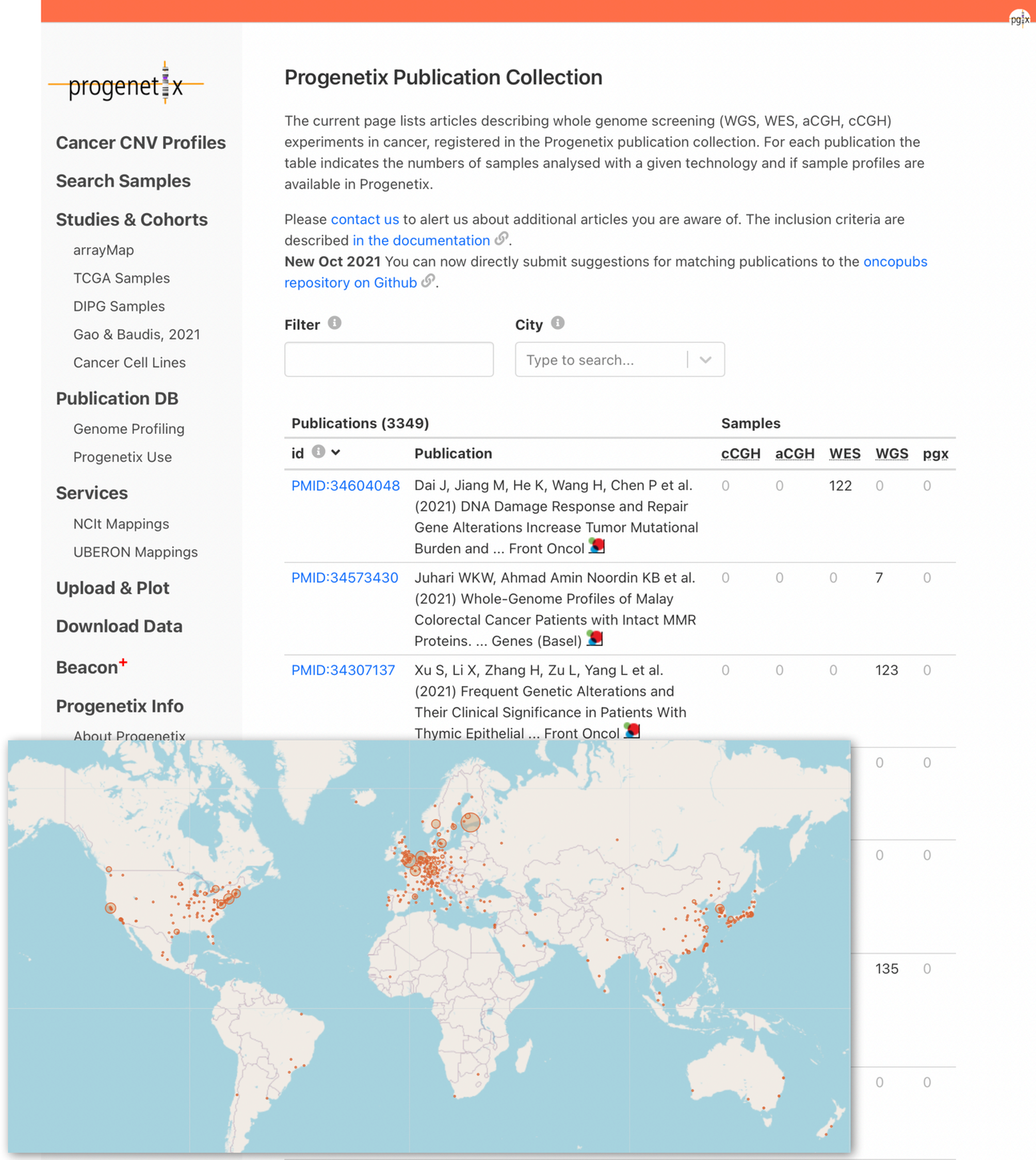
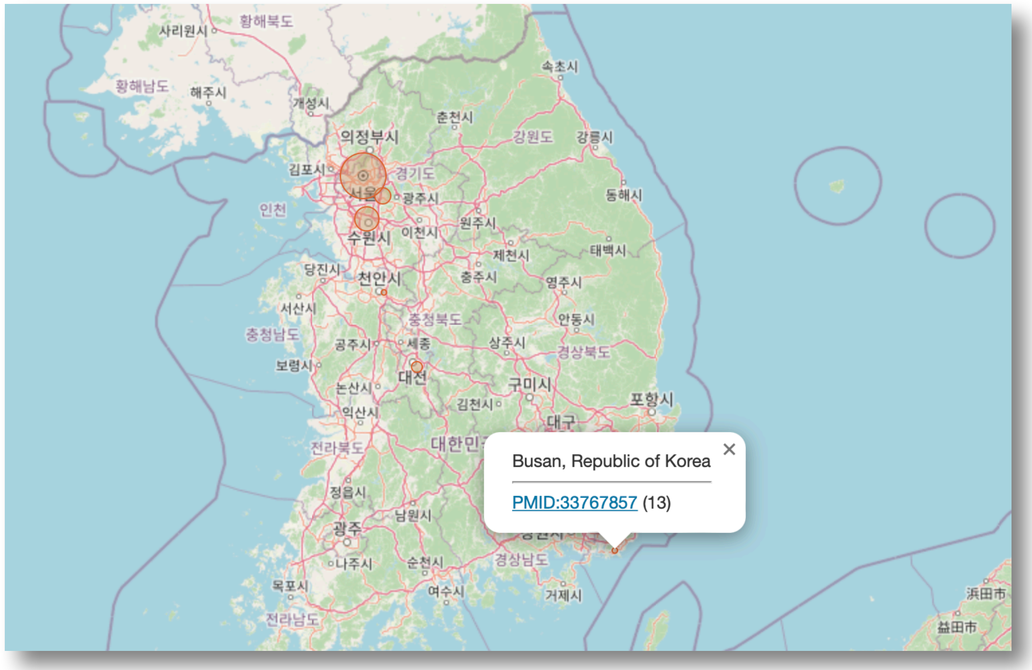


Service: Publications

Location Mapping for Statistics and Discovery...

- all publications are tagged for "best fit" geographic origin in order
    1. specific sample origin
    2. processing laboratory
    3. corresponding author
- enables searches for e.g. "all publications or samples in HCC from 2000km around Taipeh"
- handy utility for discovering locally performed research, partners...



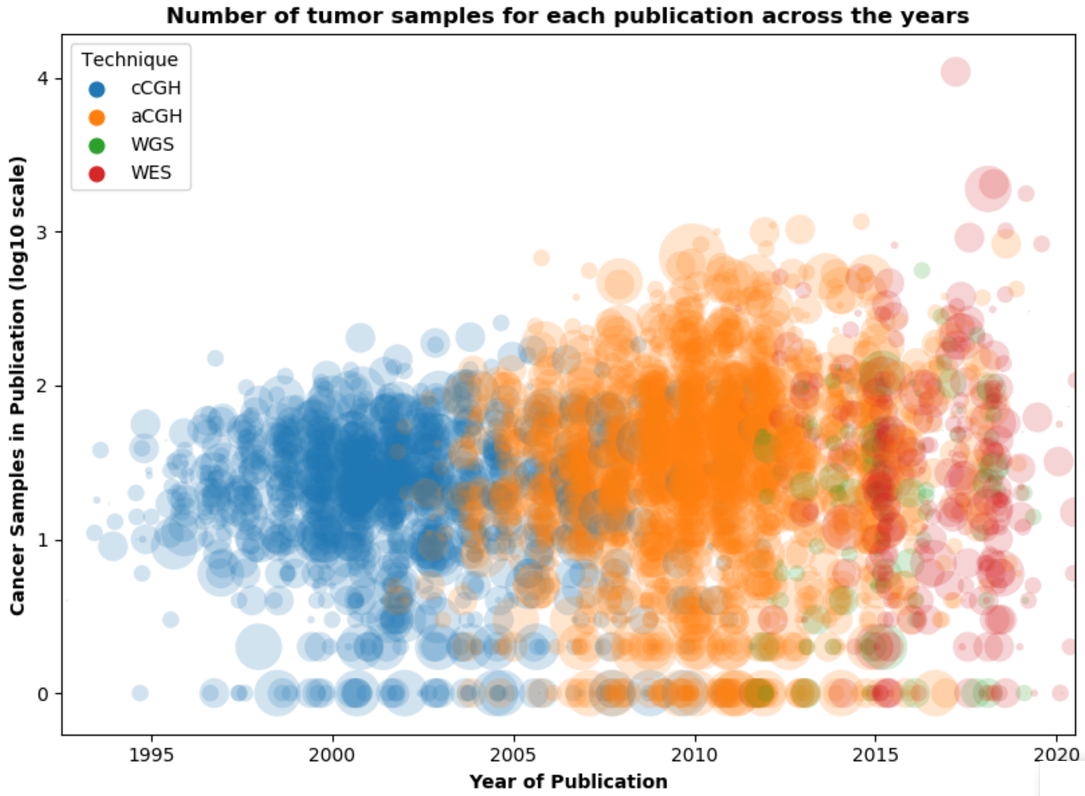
PMID:33767857

Methylation and molecular profiles of ependymoma: Influence of patient age and tumor anatomic location.

Cho HJ, Park HY, Kim K, Chae H, Paek SH, Kim SK, Park CK, Choi SH, Park SH.

Mol Clin Oncol PMID:33767857

http://progenetix.org/services/publications/?filters=genomes:>0&ISO3166alpha2=kr&output=map



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.

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- Cancer CNV Profiles
- Search Samples
- Studies & Cohorts
  - arrayMap
  - TCGA Samples
  - DIPG Samples
  - Gao & Baudis, 2021
  - Cancer Cell Lines
- Publication DB
- Services
  - NCIt Mappings
  - UBERON Mappings
- Upload & Plot
- Download Data

Progenetix Publication Collection

The current page lists articles describing whole genome screening (WGS, WES, aCGH, cCGH) experiments in cancer, registered in the Progenetix publication collection. For each publication the table indicates the numbers of samples analysed with a given technology and if sample profiles are available in Progenetix.

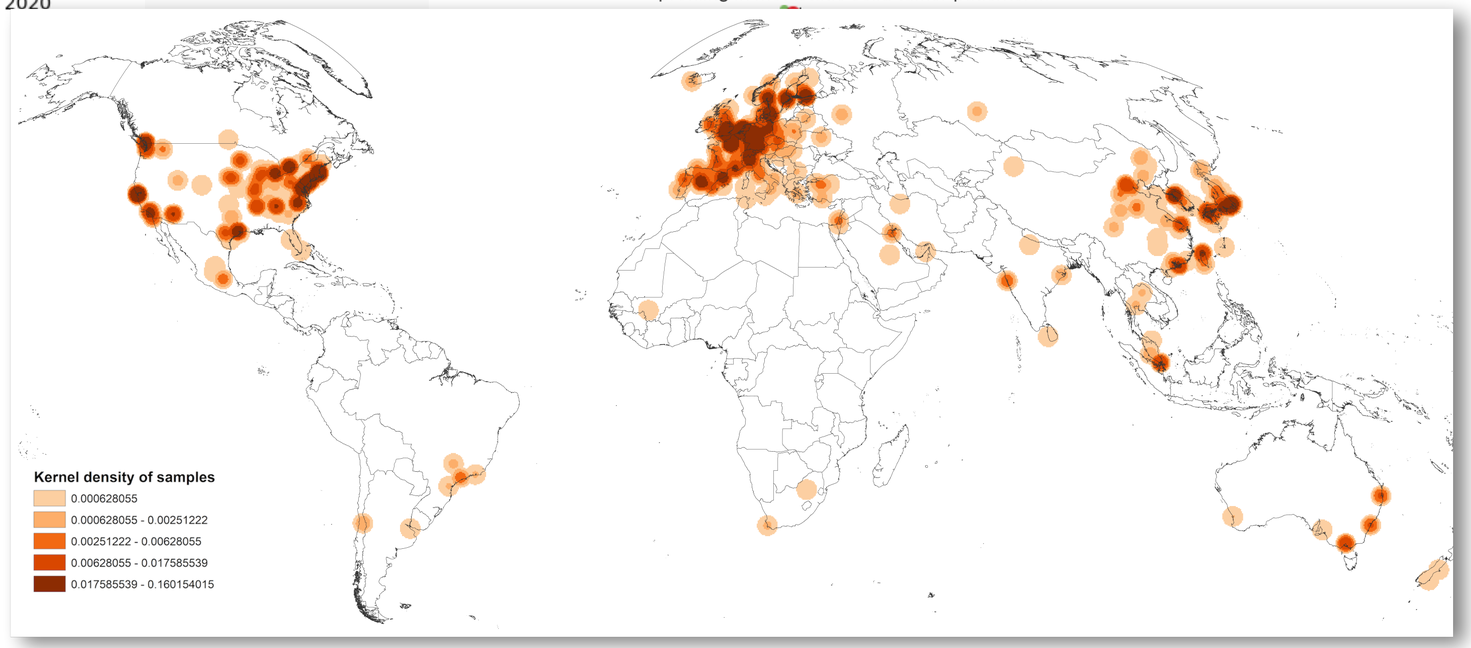
Please contact us to alert us about additional articles you are aware of. The inclusion criteria are described in the documentation.

Filter

City

Type to search...

Publications (3324)		Samples				
id	Publication	cCGH	aCGH	WES	WGS	pgx
PMID:34103027	Peng G, Chai H, Ji W, Lu Y, Wu S et al. (2021) Correlating genomic copy number alterations with clinicopathologic findings in 75 cases of ... BMC Med Genomics	0	79	0	0	0
PMID:34059130	Tsui DWY, Cheng ML, Shady M, Yang JL et al. (2021) Tumor fraction-guided cell-free DNA profiling in metastatic solid tumor patients. ...	0	0	5	113	0



Progenetix Stack

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- 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

React

APACHE HTTP SERVER PROJECT

python™

mongoDB

- *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"),
```

variants analyses biosamples individuals

Entity collections

collations geolocs genespans publications qBuffer

Utility collections

Documentation Home

Search

progenetix-web
☆3 1

Progenetix Documentation

- Documentation Home
  - Progenetix Source Code
    - bycon
    - progenetix-web
    - PGX
    - Additional Projects
- News & Changes
- Pages & Forms
- Services & API
- Use Case Examples
- Classifications, Ontologies & Standards
- Publication Collection
- Data Review
- Beacon+ & bycon
- Technical Notes
- Progenetix Data
- Baudisgroup @ UZH

Progenetix Source Code ¶

With exception of some utility scripts and external dependencies (e.g. MongoDB the software (from database interaction to website) behind Progenetix and Beac

`bycon`

- Python based service based on the GA4GH Beacon protocol
- software powering the Progenetix resource
- Beacon+ implementation(s) use the same code base

`progenetix-web`

- website for Progenetix and its Beacon+ implementations
- provides Beacon interfaces for the `bycon` server, as well as other Progenetix sevices (e.g. the publicat
- implemented as React / Next.js project
- contains this documentation tree here as `mkdocs` project, with files in the `docs` directory

Org.progenetix

Search

beacon-v2
☆2 8

Base `/biosamples`

`/BIOSAMPLES/` + QUERY

- /biosamples?filters=cellosaurus:CVCL_0004
- this example retrieves all biosamples having an annotation for the Cellosaurus *CVCL_0004* identifier (K562)

es/pgxbs-kftva5c9

a single biosample

MODE=TRUE

es?testMode=true

some random samples

- for testing API responses

`/BIOSAMPLES/{ID}/G_VARIANTS`

- /biosamples/pgxbs-kftva5c9/g_variants/
- retrieval of all variants from a single biosample

Base `/individuals`

`/INDIVIDUALS` + QUERY ¶

/individuals?filters=NCIT:C7541

Rapidly evolving documentation of both the Beacon API itself and its use and technical implementation on
docs.genomebeacons.org
docs.progenetix.org

Shoutout to Laure(e)n Fromont & Manuel Rueda for being instrumental in the Beacon v2 documentation!

Beacon API

Beacon-style JSON responses

The Progenetix resource's API utilizes the `bycon` framework for data query and delivery and represents a custom implementation of the Beacon *v2* API.

The standard format for JSON responses corresponds to a generic Beacon v2 response, with the `meta` and `response` root elements. Depending on the endpoint, the main data will be a list of objects either inside `response.results` or (mostly) in `response.resultSets.results`. Additionally, most API responses (e.g. for biosamples or variants) provide access to data using *handover* objects.

Beacon v2 Documentation

Search

beacon-v2
☆2 8

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Progenetix & Beacon+

The Beacon+ implementation - developed in the Python & MongoDB based `bycon` project - implements an expanding set of Beacon v2 paths for the Progenetix resource.

Scoped responses from query object

In queries with a complete `beaconRequestBody` the type of the delivered data is independent of the path and determined in the `requestedSchemas`. So far, Beacon+ will compare the first of those to its supported responses and provide the results accordingly; it doesn't matter if the endpoint was `/beacon/biosamples/` or `/beacon/variants/` etc.

Below is an example for the standard test "small deletion CNVs in the CDKN2A locus, in gliomas" Progenetix test query, here responding with the matched variants. Exchanging the `entityType` entry to

- `{ "entityType": "biosample", "schema:": "https://progenetix.org/services/schemas/Biosample/"}`

would change this to a biosample response. The example ccan be tested by POSTing this as `application/json` to `http://progenetix.org/beacon/variants/` or `http://progenetix.org/beacon/biosamples/`.

```
{
    "$schema":"beaconRequestBody.json",
    "meta": {
        "apiVersion": "2.0",
        "requestedSchemas": [
            {
                "entityType": "genomicVariant",
                "schema:": "https://progenetix.org/services/schemas/genomicVariant"
            }
        ]
    },
    "query": {
        "requestParameters": {
```

Onboarding

Demonstrating Compliance

- Progenetix Beacon+ has served as implementation driver since 2016
- Beacon v2 as service with protocol-driven registries for federation
- GA4GH approved Beacon v2 in April 2022

Global Alliance for Genomics & Health

	Beacon v1 Development	Beacon v2 Development	Related ...
2014	**GA4GH founding event; Jim Ostell proposes Beacon concept with "more features… version 2"**		
2015	• beacon-network.org aggregator created by DNAstack • Beacon v0.3 release	• Beacon+ concept implemented on progenetix.org	• ELIXIR starts Beacon project support
2016	• work on queries for structural variants (brackets for fuzzy start and end parameters...)	• 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	• 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 ... • discussions w/ clinical stakeholders	• new Beacon website (March)
2019	• ELIXIR Beacon Network		• Beacon publication at Nature Biotechnology
2020		• framework + models concept implemented • range and bracket queries, variant length • starting of GA4GH review process	
2021		• further changes esp. in default model, aligning with Phenopackets and VRS • unified beacon-v2 code & docs repository • **Beacon v2 approved at Apr GA4GH Connect**	
2022			• *docs.genomebeacons.org*

beacon.progenetix.org

Beacon Implementations

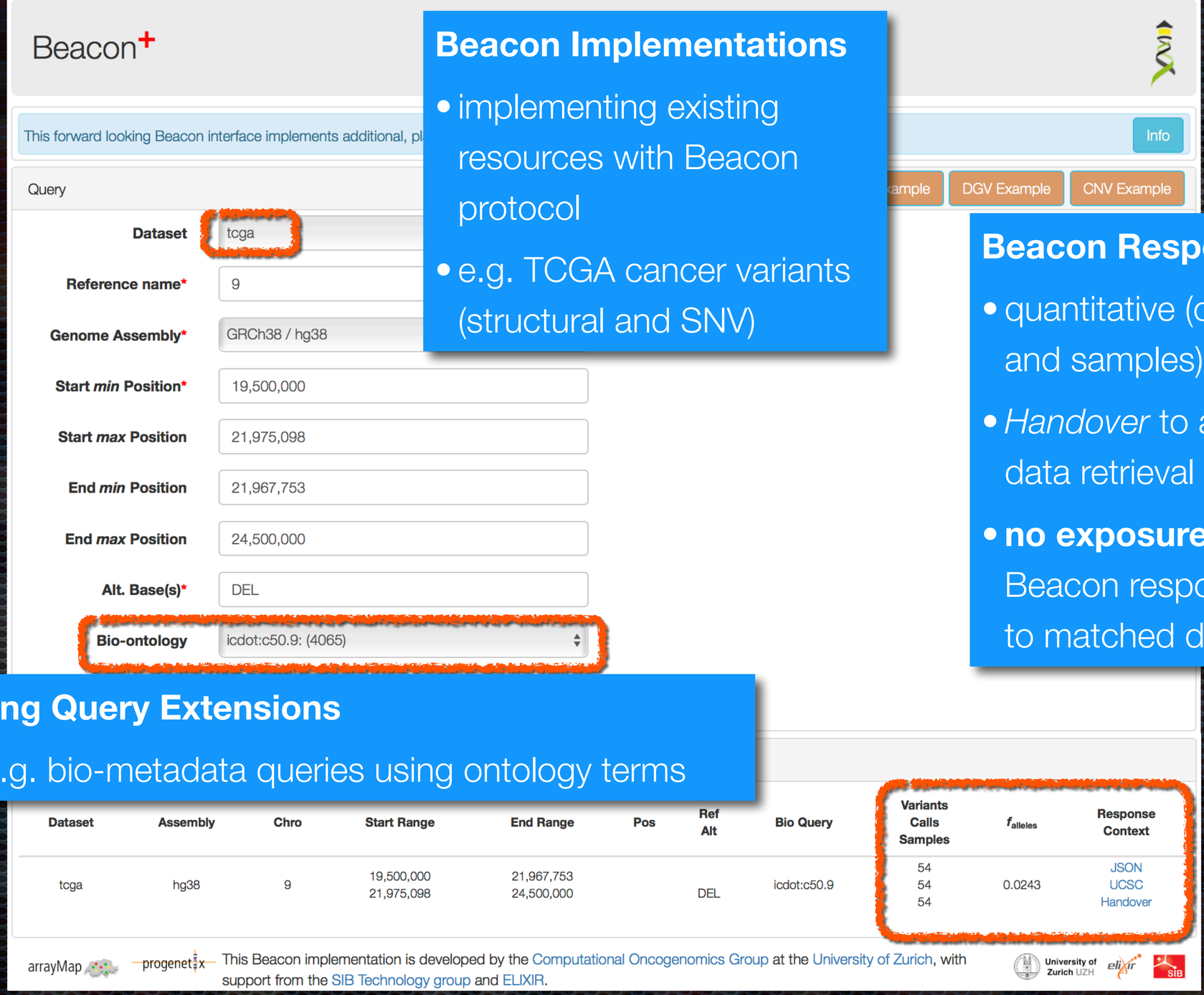
- implementing existing resources with Beacon protocol
- e.g. TCGA cancer variants (structural and SNV)

Beacon Response

- quantitative (counts for variants, callsets and samples)
- *Handover* to authentication system for data retrieval
- **no exposure** of data beyond standard Beacon response and additional pointer to matched data

Prototyping Query Extensions

- testing e.g. bio-metadata queries using ontology terms

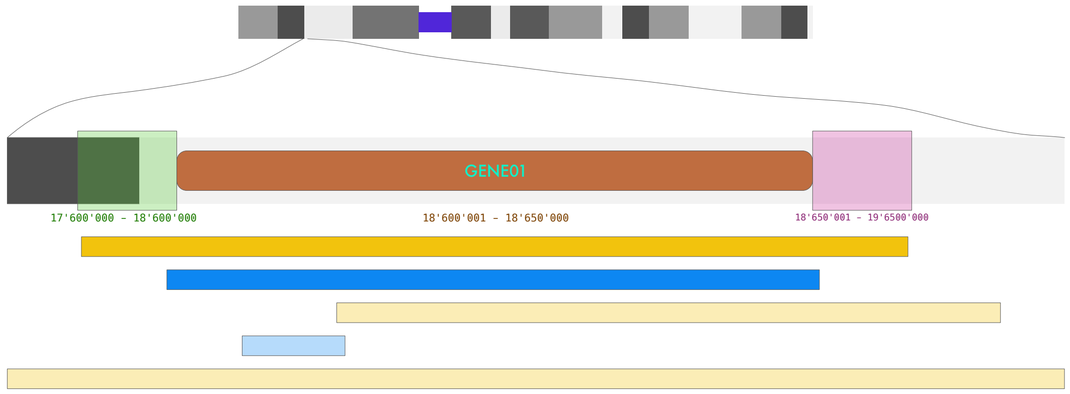


Progenetix in 2022

Variant and Metadata for Sample Discovery

- positional queries for genomic variants using the **GA4GH Beacon protocol**
- metadata queries (diagnoses, identifiers, clinical classes ...) using **Beacon "filters"**

Genome Bracket Query
(full match)



Bold: Matched Variants

Shaded: Unmatched

DEL (Copy Number Loss) DUP (Copy Number Gain)

progenetix

- Cancer CNV Profiles
- Search Samples
- Studies & Cohorts
  - arrayMap
  - TCGA Samples
  - DIPG Samples
  - Gao & Baudis, 2021
  - Cancer Cell Lines
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- Services
  - NCIt Mappings
  - UBERON Mappings
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- Progenetix Info
  - About Progenetix
  - Use Cases
  - Documentation
  - Baudisgroup @ UZH

Search Samples

CDKN2A Deletion Example | MYC Duplication | TP53 Del. in Cell Lines | K-562 Cell Line | Gene Spans | Cytoband(s)

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 "highly focal" hits (here i.e. <= ~1Mbp in size). The query can be modified e.g. through changing the position parameters or diagnosis.

Gene Symbol: Select...

Chromosome: 9

(Structural) Variant Type: DEL (Deletion)

Start or Position: 21500001-21975098

End (Range or Structural Var.): 21967753-22500000

Minimum Variant Length

Maximal Variant Length

Reference ID(s): Select...

Cancer Classification(s): NCIT:C3058: Glioblastoma (4375)

Clinical Classes: Select...

Genotypic Sex: Select...

Biosample Type: Select...

Filters

Filter Logic: AND

Filter Precision: exact

City: Select...

Chromosome 9

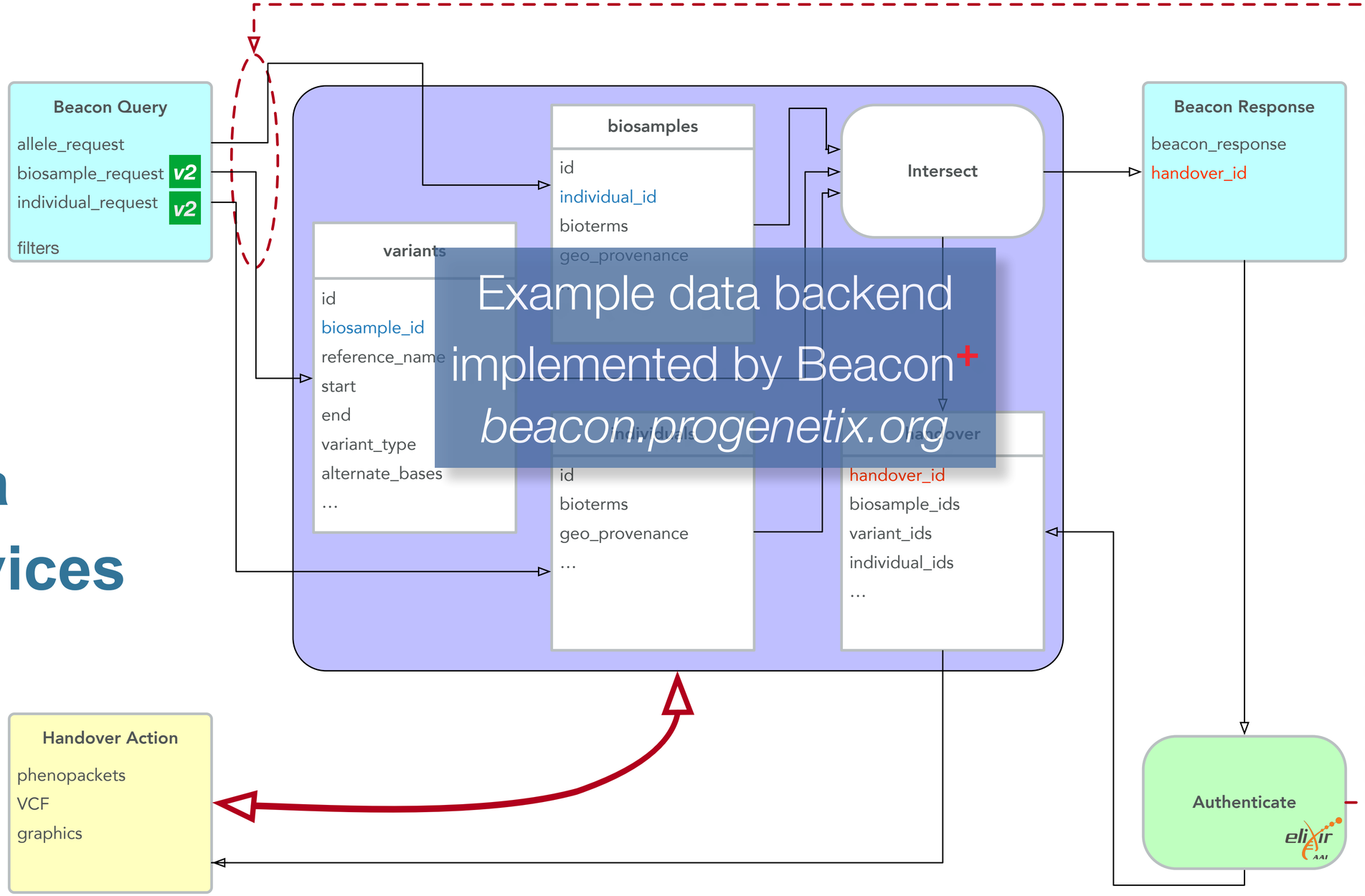
Query Database

Filter types

- Ontology Filters
  - **Hierarchical ontology query** is assumed by default, where the requested ontology filter(s) and all descendent terms are queried
  - **Exact term match** requests, where descendent terms are excluded, are supported
  - **Semantic similarity queries** for entities that are associated with terms that are similar to the requested filters are supported by Beacon 2.0
    - Agnostic to the semantic similarity model used by a Beacon
    - Relative similarity thresholds of *high*, *medium* and *low*
- Numeric Filters … (using equality and relational operators)
- Alphanumeric Filters … (e.g. string matches)

Beacon & Handover

Beacons v1.1 supports **data delivery services**



Beacon I/O

Handover

Authentication

Michael Baudis

Global Alliance for Genomics & Health

SIB

University of Zurich UZH

elixir

Beacon v2 Conformity and Extensions in Progenetix

Putting the + into Beacon ...

- support & use of standard Beacon v2 PUT & GET variant queries, filters and meta parameters
  - ➡ variant parameters, geneId, lengths, EFO & VCF CNV types, pagination
  - ➡ widespread, self-scoping filter use for bio-, technical- and and id parameters with switch for descending terms use (globally or per term if using POST)
- extensive use of handovers
  - ➡ asynchronous delivery of e.g. variant and sample data, data plots
- + extensions of query logic
  - ➡ optional use of OR logic for filter combinations (global)
- + extension of query parameters
  - ➡ geographic queries incl. $geonear and use of GeoJSON in schemas
- ┐(ツ ▽ ツ)┌ no implementation of authentication on this open dataset

Progenetix provides a number of additional services and output formats which are initiated over the / services path or provided as request parameters and are not considered Beacon extensions (though they follow the syntax where possible).

progenetix

pgxRpi

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

Beacon Path: Retrieve variants by biosample id(s)

```
https://progenetix.org/beacon/g_variants/
?biosampleIds=pgxbs-kftvh94d,pgxbs-kftvh94g,pgxbs-kftvh972
&output=pgxseg
```

Beacon Path: Get biosamples by filter(s)

```
http://progenetix.org/beacon/biosamples/
?filters=NCIT:C3697&output=datatable
```

Service Path: Retrieve CNV frequencies by filter(s)

```
http://www.progenetix.org/services/intervalFrequencies/
?id=NCIT:C4323&output=pgxseg
```

Author: Hangjia Zhao | @hangjiaz

README.md

pgxRpi

This is an API wrapper package to access data from Progenetix database.

You can install this package from GitHub using:

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

If you are interested in accessing CNV variant data, get started from this vignette

If you are interested in accessing CNV frequency data, get started from this vignette

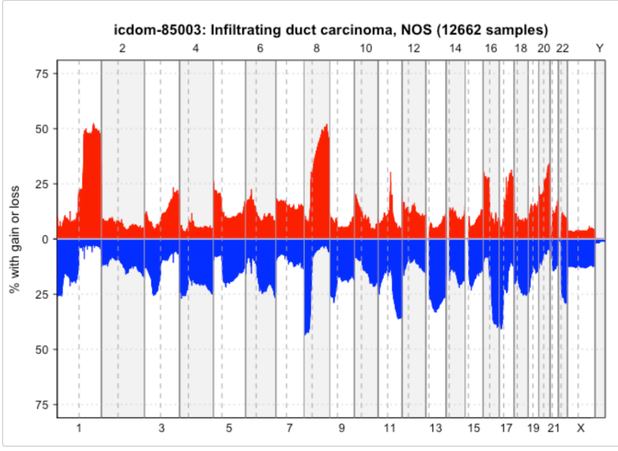
When you face problems, try to reinstall the latest version. If reinstallation doesn't help, please contact us.

```
variant_1 <- pgxLoader(type="variant", biosample_id = biosample_id)
```

```
biosamples <- pgxLoader(type="biosample", filters = "NCIT:C3059",codematches = TRUE,
                        biosample_id = c("pgxbs-kftva5zv","pgxbs-kftva5zw"))
```

```
freq_pgxseg <- pgxLoader(type="frequency", output ='pgxseg',
                         filters=c("NCIT:C4038","pgx:icdom-85003"),
                         codematches = TRUE)
```

```
pgxFreqplot(freq_pgxseg, filters='pgx:icdom-85003')
```



Beacon+: Phenopackets

Testing alternative response schemas...

http://progenetix.org/beacon/biosamples/pgxbs-kftvhyvb/phenopackets

- the v2 default schemas are mostly aligned w/ Phenopackets v2
- creating phenopackets can be done mostly by re-wrapping of Beacon entities (individual, biosample)
- variants can be included through file resource URLs; in Beacon+ this is done through *ad hoc* handover URIs


```
"id": "pgxpxf-kftx3tl5",
"metaData": {
  "phenopacketSchemaVersion": "v2",
  "resources": [
    {
      "id": "NCIT",
      "iriPrefix": "http://purl.obolibrary.org/obo/NCIT_",
      "name": "NCIt Plus Neoplasm Core",
      "namespacePrefix": "NCIT",
      "url": "http://purl.obolibrary.org/obo/ncit/neoplasm-core.c
      "version": "2022-04-01"
    },


"subject": {
  "dataUseConditions": {
    "id": "DUO:0000004",
    "label": "no restriction"
  },
  "diseases": [
    {
      "clinicalTnmFinding": [],
      "diseaseCode": {
        "id": "NCIT:C3099",
        "label": "Hepatocellular Carcinoma"
      },
      "onset": {
        "age": "P48Y9M26D"
      },
      "stage": {
        "id": "NCIT:C27966",
        "label": "Stage I"
      }
    }
  ],
  "id": "pgxind-kftx3tl5",
  "sex": {
    "id": "PATO:0020001",
    "label": "male genotypic sex"
  },
  "updated": "2018-12-04 14:53:11.674000",
  "vitalStatus": {
    "status": "UNKNOWN_STATUS"
  }
}
```

```
"biosamples": [
  {
    "biosampleStatus": {
      "id": "EFO:0009656",
      "label": "neoplastic sample"
    },
    "dataUseConditions": {
      "id": "DUO:0000004",
      "label": "no restriction"
    },
    "description": "Primary Tumor",
    "externalReferences": [
      {
        "id": "pgx:TCGA-0004d251-3f70-4395-b175-c94c2f5b1b81",
        "label": "TCGA case_id"
      },
      {
        "id": "pgx:TCGA-TCGA-DD-AAVP",
        "label": "TCGA submitter_id"
      },
      {
        "id": "pgx:TCGA-9259e9ee-7279-4b62-8512-509cb705029c",
        "label": "TCGA sample_id"
      },
      {
        "id": "pgx:TCGA-LIHC",
        "label": "TCGA LIHC project"
      }
    ],
    "files": [
      {
        "fileAttributes": {
          "fileFormat": "pgxseg",
          "genomeAssembly": "GRCh38"
        },
        "uri": "https://progenetix.org/beacon/biosamples/pgxbs-kftvhyvb/variants/?output=pgxseg"
      }
    ],
    "histologicalDiagnosis": {
      "id": "NCIT:C3099",
      "label": "Hepatocellular Carcinoma"
    },
    "id": "pgxbs-kftvhyvb",
    "individualId": "pgxind-kftx3tl5",
    "pathologicalStage": {
      "id": "NCIT:C27966",
      "label": "Stage I"
    },
    "sampledTissue": {
      "id": "UBERON:0002107",
      "label": "liver"
    },
    "timeOfCollection": {
      "age": "P48Y9M26D"
    },
```

Beacon+: Phenopackets

Testing alternative response schemas...

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Progenetix & Beacon+

A cancer genomics reference resource powered by GA4GH standards

- Copy number variations constitute a complex, exciting and still poorly understood research topic in cancer and rare disease genomics
- Progenetix is the largest public resource for CNV in cancers (and increasingly reference genomes)
- The complexity of inherited and somatic genomic variations requires data access beyond individual resources => **Federated Data Access**
- The Global Alliance for Genomics and Health (GA4GH) is a policy-framing and technical standards-setting organization with focus on genomic data sharing
- Beacon v2 is the main GA4GH data discovery and sharing protocol, developed with support from the European bioinformatics organization ELIXIR
- Progenetix serves as a testbed for the early implementation of GA4GH standards such as Beacon extensions, Phenopackets and VRS

Beacon's v2.n Future?

Some proposals for a stepwise Beacon protocol extension

- Query language expansion, e.g. Boolean options for chaining filters

  ➡use of heterogeneous/alternative annotations within and across resources

- **Phenopackets** support as a (the?) default format for biodata export
- **Phenopackets** as **request** documents
- Focus on service & **resource discovery**
- **ELIXIR Beacon Network**, including translations for federated queries to Beacon and Beacon-like resources

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.

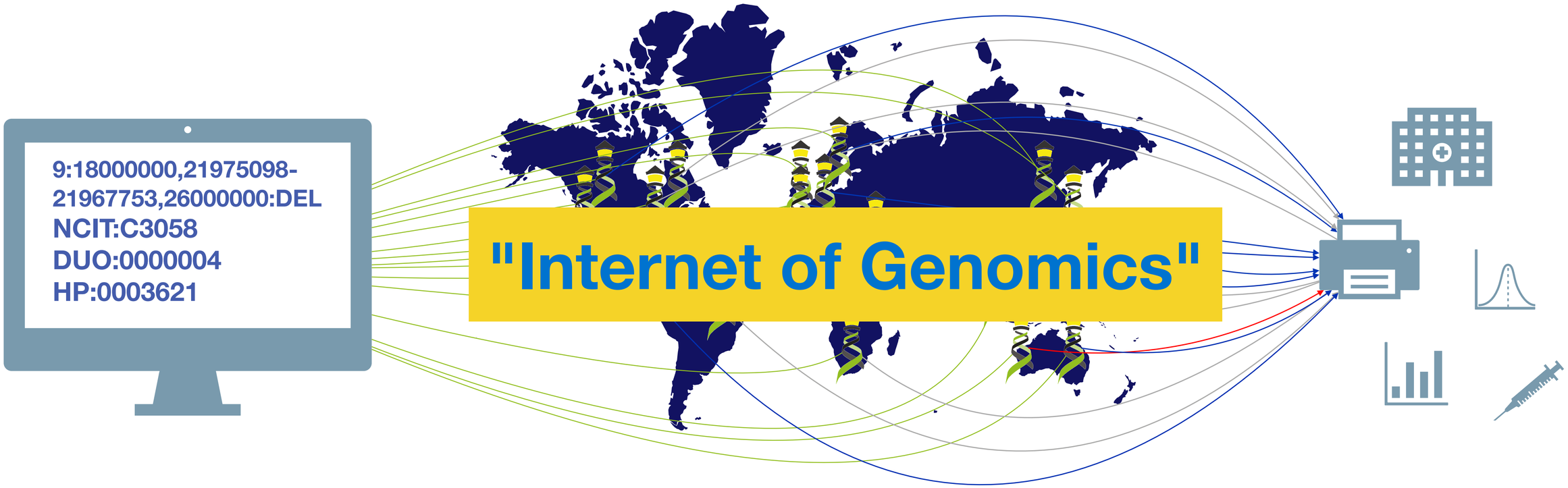
A Beacon network federates *genome variant queries* across databases that support the ***Beacon API***

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**".

The Beacon team

EUROPEAN GENOME-PHENOME ARCHIVE

CRG Centre for Genomic Regulation

Jordi Rambla
Arcadi Navarro
Roberto Ariosa
Manuel Rueda
Lauren Fromont
Mauricio Moldes
Claudia Vasallo
Babita Singh
Sabela de la Torre
Marta Ferri
Fred Haziza

CSC

Juha Törnroos
Teemu Kataja
Ilkka Lappalainen
Dylan Spalding

UNIVERSITY OF LEICESTER

Cafe Variome Central

Tony Brookes
Tim Beck
Colin Veal
Tom Shorter

SPHN Swiss Personalized Health Network

University of Zurich[UZH]

Michael Baudis
Rahel Paloots
Hangjia Zhao
Bo Gao

Genomics england

Augusto Rendon
Ignacio Medina
Javier López
Jacobo Coll
Antonio Rueda

cnag centre nacional d'anàlisi genòmica centro nacional de análisis genómico

Sergi Beltran
Carles Hernandez

Inserm Institut national de la santé et de la recherche médicale

David Salgado

BSC Barcelona Supercomputing Center Centro Nacional de Supercomputación

Salvador Capella
Dmitry Repchevski
JM Fernández

DisGeNET

Laura Furlong
Janet Piñero

elixir

B1MG

Serena Scollen
Gary Saunders
Giselle Kerry
David Lloyd

H3Africa Human Heredity & Health in Africa

Nicola Mulder
Mamana Mbiyavanga
Ziyaad Parker

EU Can CAN.

David Torrents

AUTISM SPEAKS

Dean Hartley

JUNTA DE ANDALUCIA

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

Toshiaki Katayama

GEM Japan

Stephane Dyke

DNASTACK

Marc Fiume
Miro Cupak

BRCA EXCHANGE

Melissa Cline

ENA

EMBL-EBI

Diana Lemos

EUROPEAN JOINT PROGRAMME RARE DISEASES

VICC Variant Interpretation for Cancer Consortium

—-GA4GH teams—-

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

Get Involved! Visit GA4GH.ORG

Global Alliance
for Genomics & Health

Join a Work Stream!

Contact secretariat@ga4gh.org

Become an Organizational Member
ga4gh.org/members

Subscribe to GA4GH Updates
ga4gh.org/subscribe

Contributors: varied backgrounds, many roles

Global Alliance
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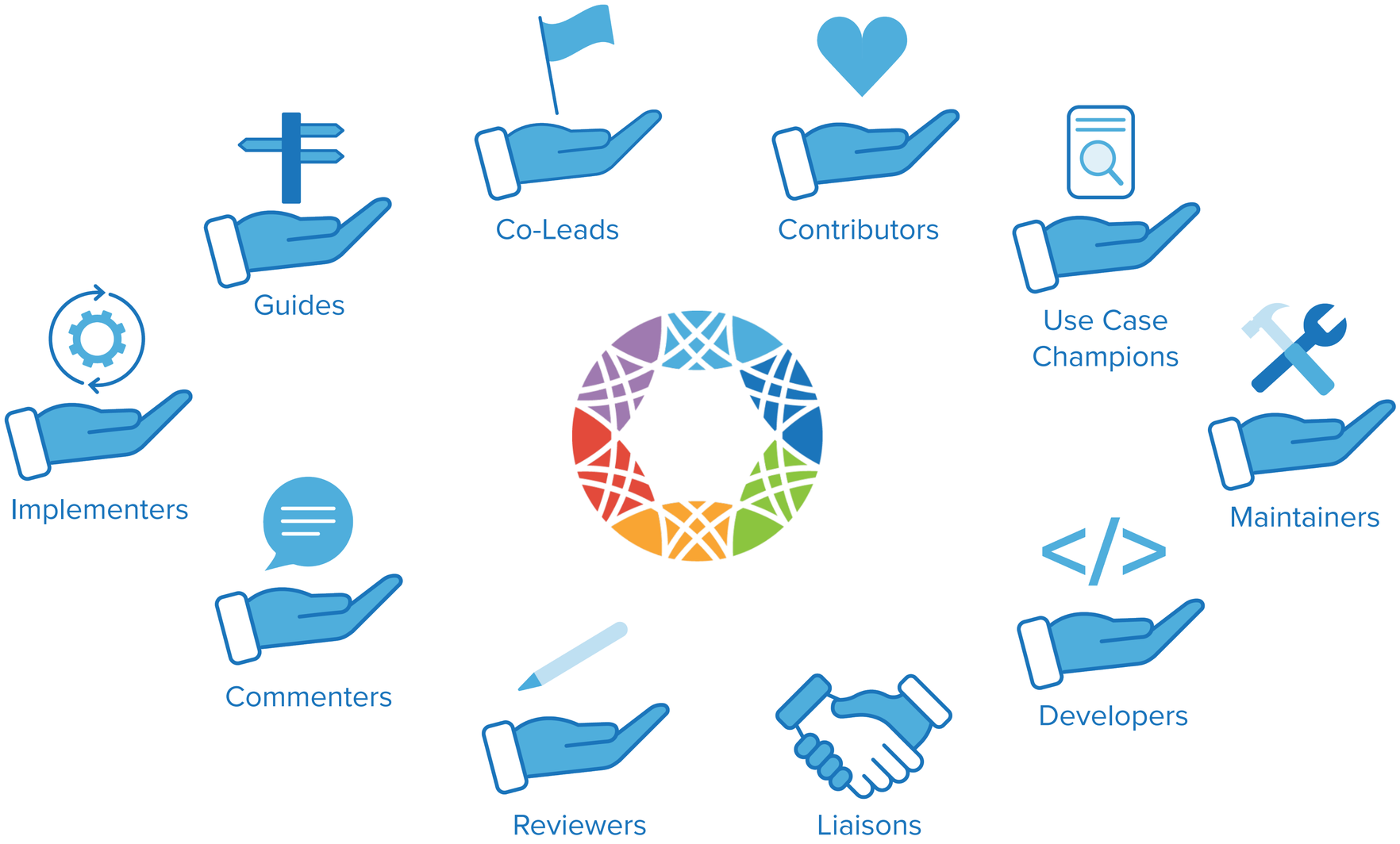

Progenetix Needs & Offers

What we have ...

✓ collection of >4000 articles assessed for scope
- training set for NLP & search engine generation

✓ cancer specific ontologies with cross-mappings (ICD-O vs. NCIt) based on >100k samples
- existing service API

✓ metadata ontology mappings for some 10k samples, with varying coverage for grade / stage / survival / ...

✓ CNV profiles for >110k samples, >700 entities with disease codes and metadata

✓ cell line CNV profiles together with mapped variants with clinical evidences

What we're working on...

➡ (semi-)automated detection of additional articles for scope (genome screening technologies, cancer samples, geographies)

➡ generation of a complete ICD-O terminology tree with NCIT (?) correspondence
- improved service API & publication

➡ improved annotations using smarter source (article, annotation files) pre-/processing

➡ correlation between individual profiles, profile heterogeneity and external parameters

➡ relation between cell lines and native tumor types, with consideration of non-CNV parameters and publication use

info.baudisgroup.org | progenetix.org

Universität Zürich UZH

SIB

elixir

Universität Zürich UZH

info.baudisgroup.org