



Global Alliance  
for Genomics & Health



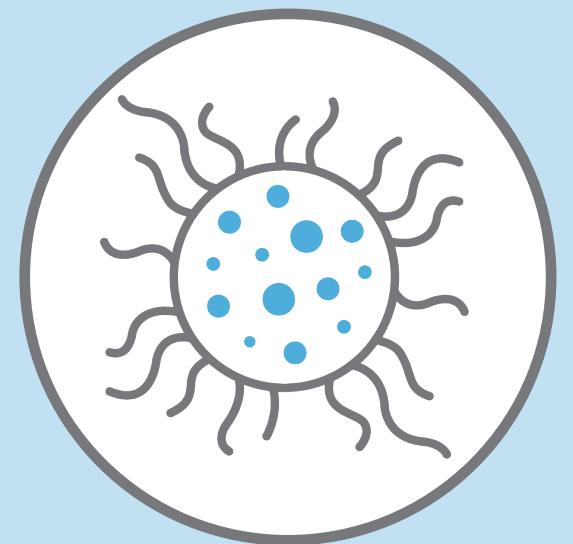
# Open Reference Resource for Oncogenomic Copy Number Aberrations Implementing Emerging Global Standards **Progenetix - From Experiments to APIs**

Michael Baudis | EORTC-PAMM | Firenze December 2022

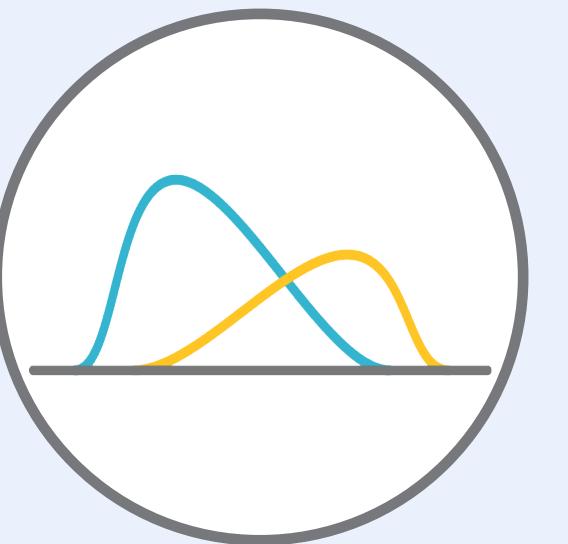
# Genomic Data Aggregation Can...



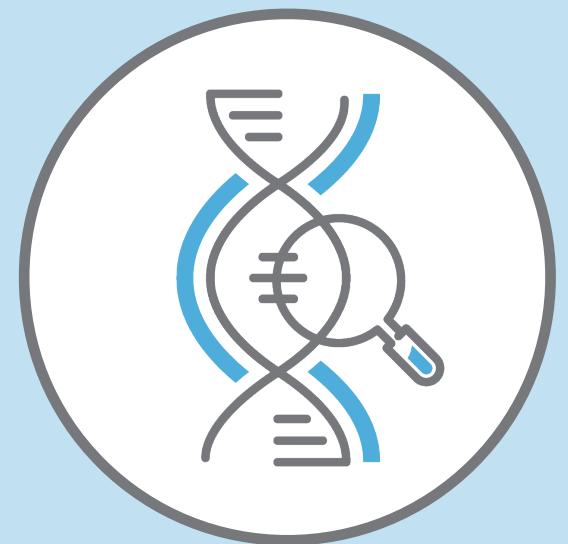
Global Alliance  
for Genomics & Health



Demonstrate  
patterns in health  
& disease



Increase statistical  
significance of  
analyses



Lead to  
“stronger” variant  
interpretations

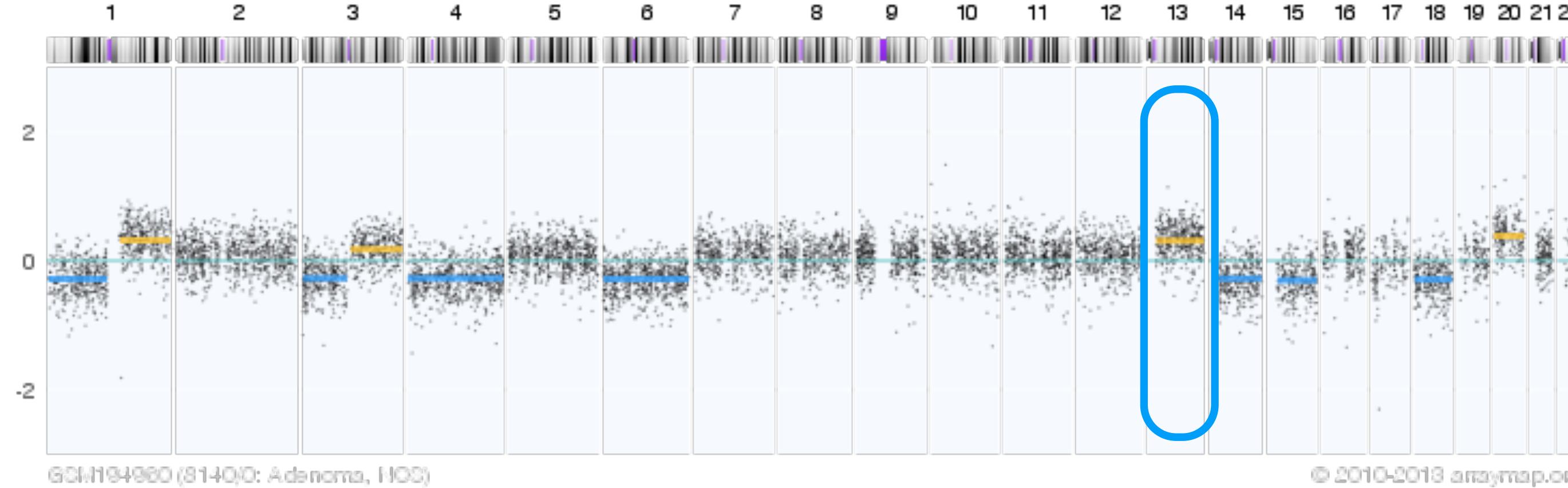


Increase  
accurate  
diagnosis

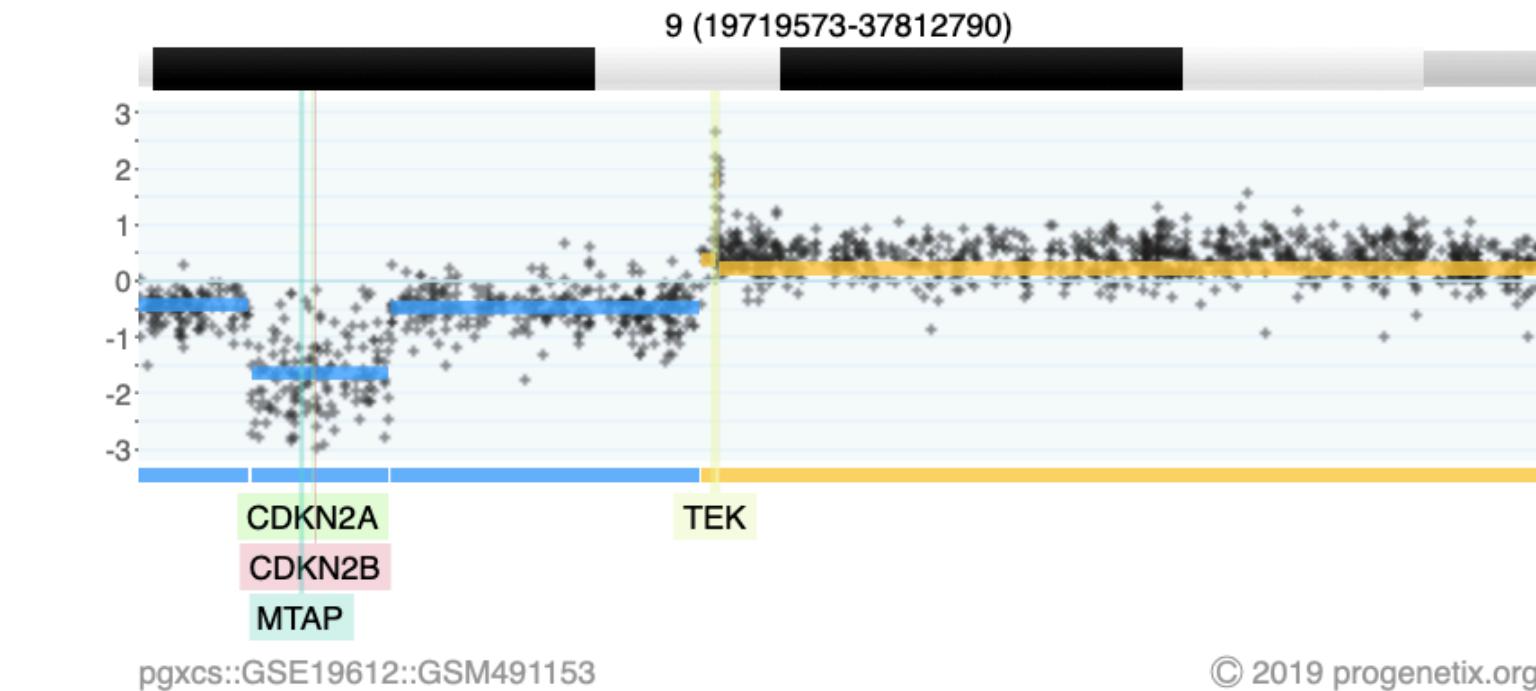


Advance  
precision  
medicine

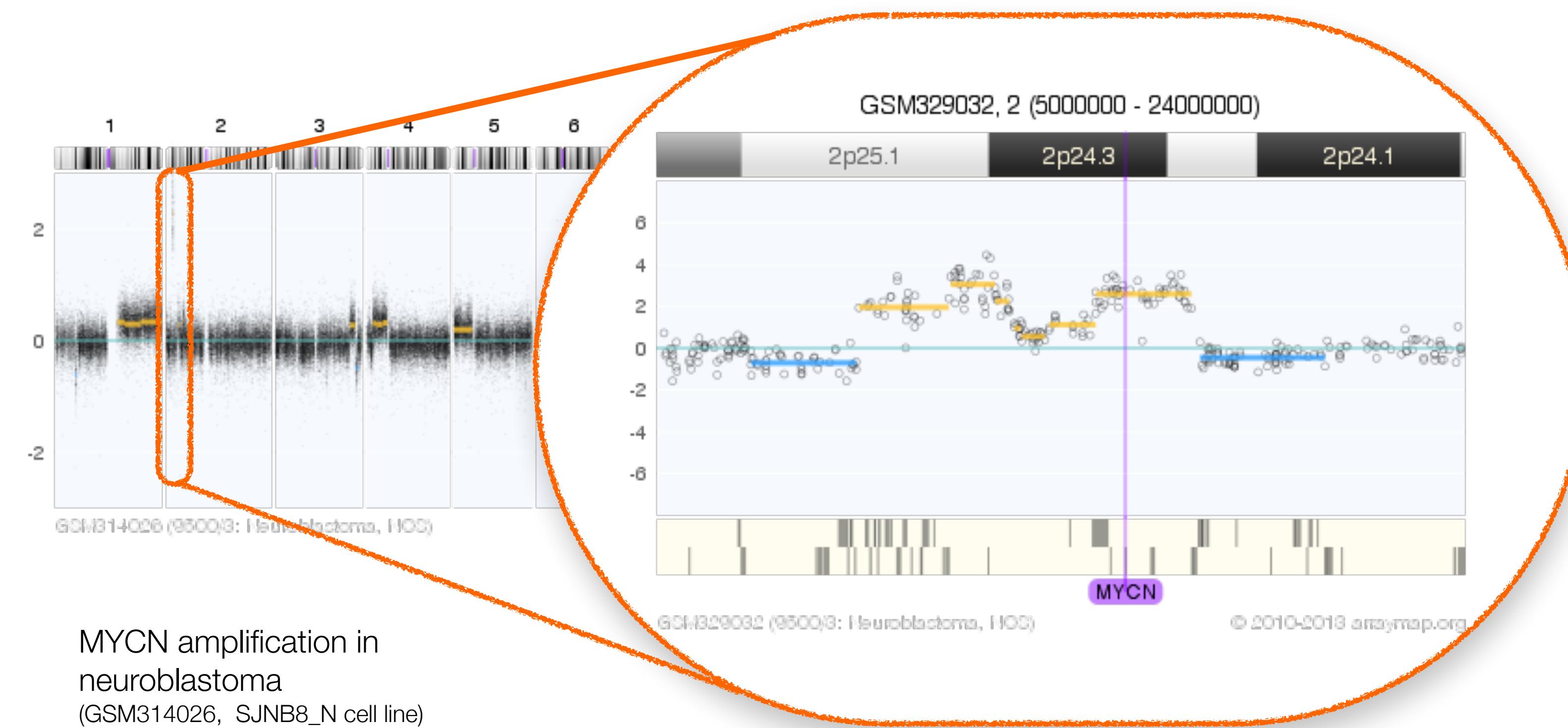
# Somatic Copy Number Variations



Gain of chromosome arm 13q in colorectal carcinoma



2-event, homozygous deletion in a Glioblastoma



MYCN amplification in neuroblastoma  
(GSM314026, SJNB8\_N cell line)

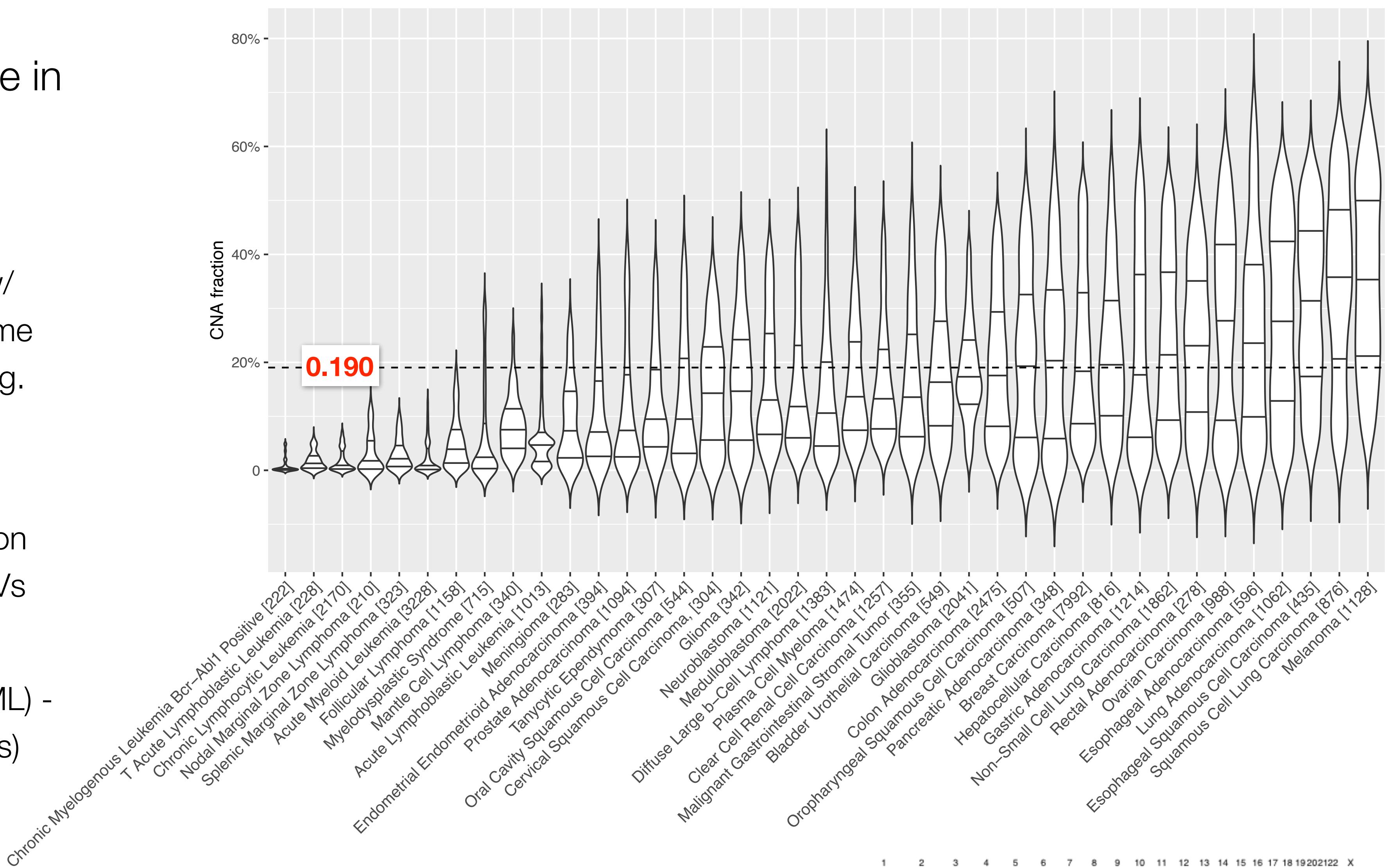
low level/high level copy number alterations (CNAs)

arrayMap

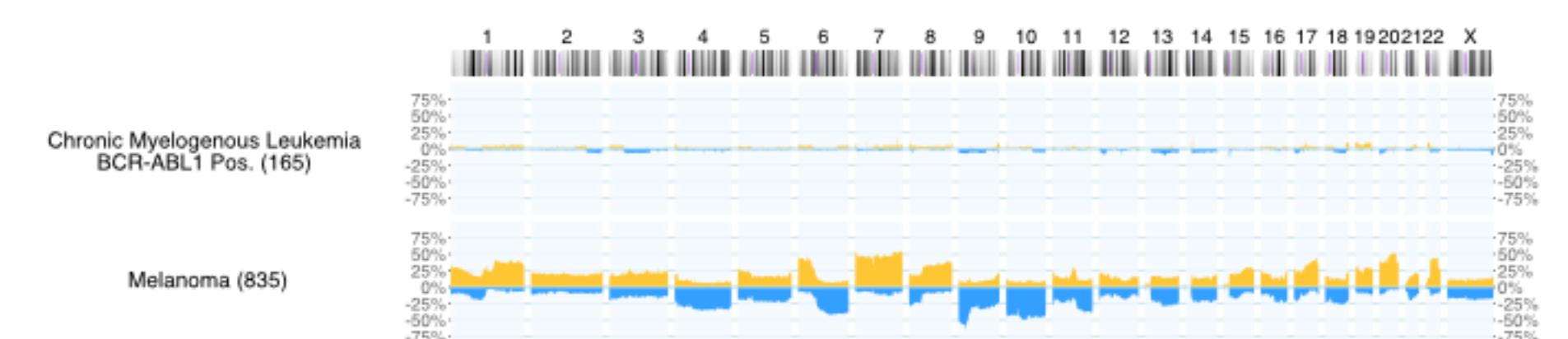


# Genome CNV coverage in Cancer Classes

- 43654 out of 93640 CNV profiles; filtered for entities w/ >200 samples (removed some entities w/ high CNV rate, e.g. sarcoma subtypes)
- Single-sample CNV profiles were assessed for the fraction of the genome showing CNVs (relative gains, losses)
- range of medians 0.001 (CML) - 0.358 (malignant melanomas)



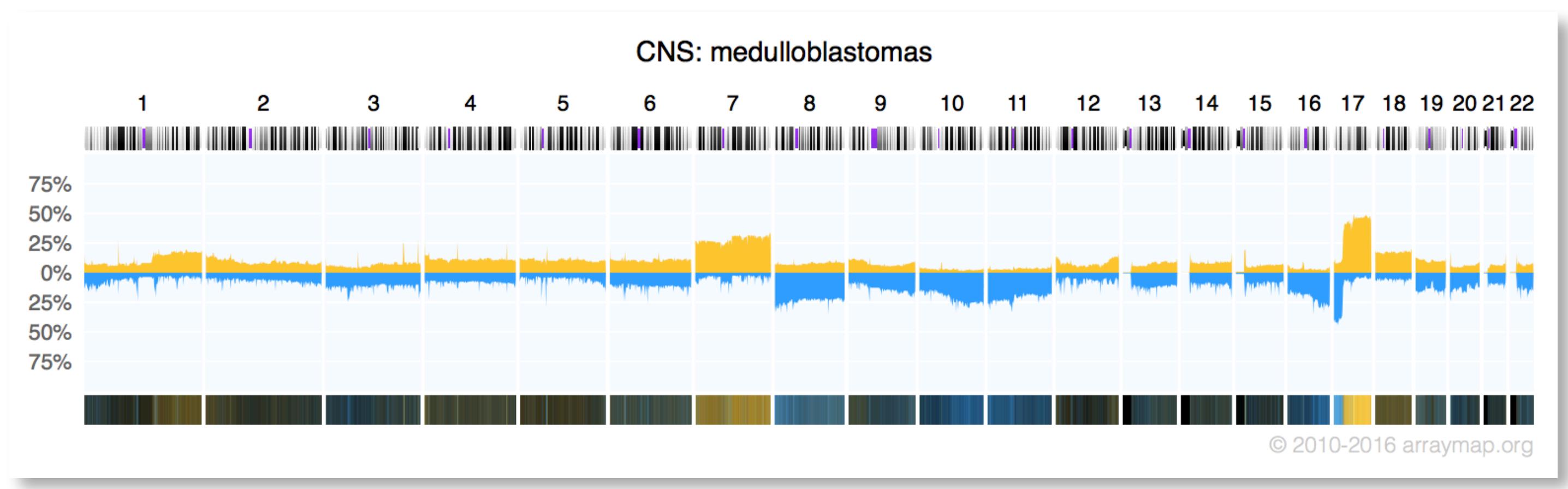
Lowest / Highest CNV fractions =>



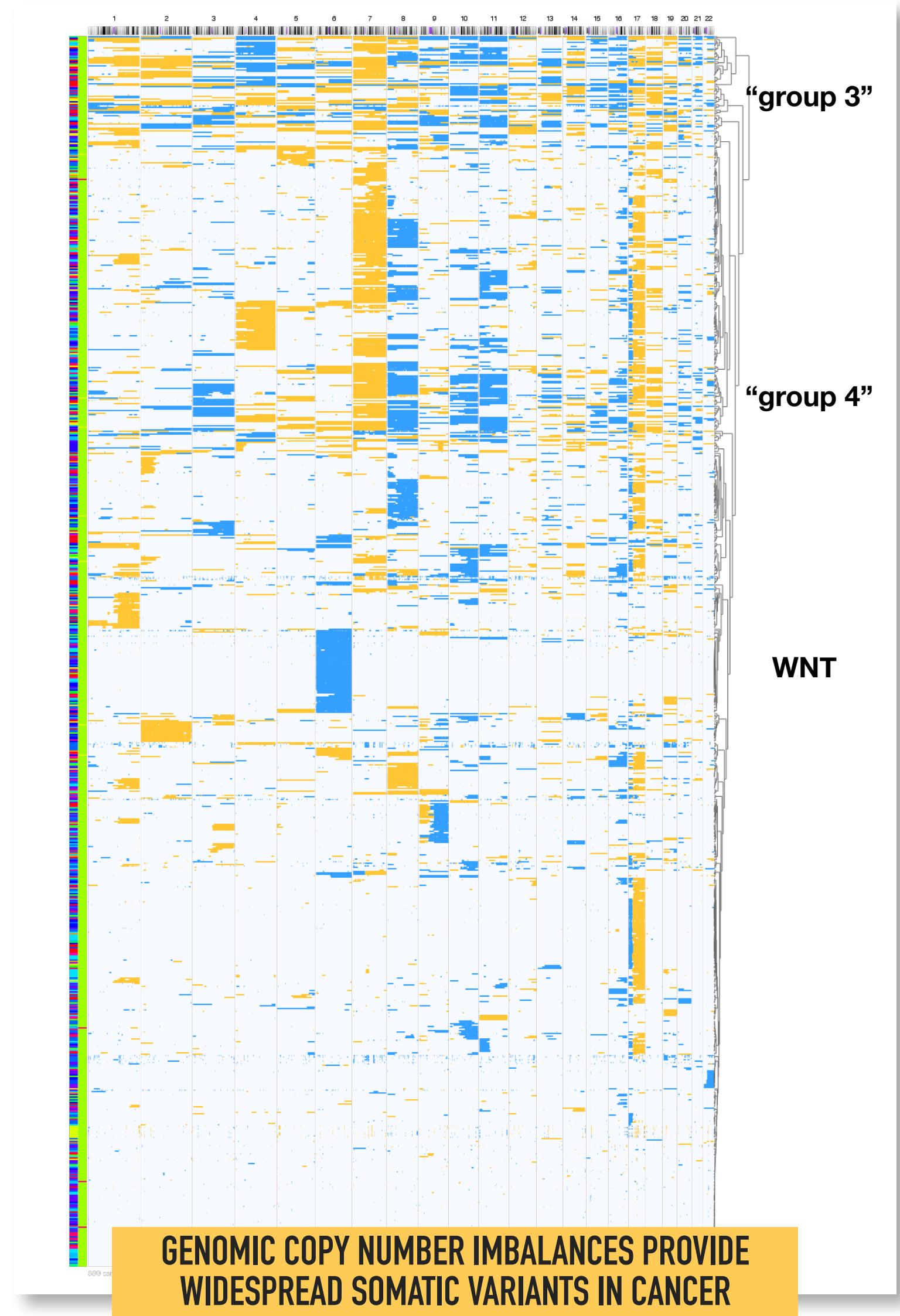
# Somatic CNVs In Cancer

## 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.



# CNVs Come in a Variety of Formats

## Text conversion from ISCN

- articles and supplements with **cytoband-based rev ish CGH** results are a great source of CNV data
- conversion by mapping cytoband locations (e.g. UCSC annotation files) to genome coordinates and assigning CNV types (enh, dim, amp are standard)

## CGH AND FISH OF METASTATIC COLORECTAL CANCER

TABLE 3. Comparison of Primary Tumors and Metastases by CGH

Case	Gain in common	Loss in common	Primary tumor only	Metastasis only
108		18		
113	7, 8q24-qter, 13q11-qter, 20q11-qter, Xq11-Xter	1p33-pter, 2p21-pter, 4q24-qter, 15q11-q15, 17p11-pter, 18		
LM	12q22-qter, 15q23-qter, 17q11-ter, 20p11-p12, 20q11-ter, 22q11-ter	1p11-p32, 1q24-31, 4, 13q11-pter, 17p11-pter, 18, 20p11-ter	11p11-pter-	12+
145	4q26-q28, 6p11-p13, 8p11-p12, 920q11-qter	1p11-pter, 4q31-qter, 6q11-qter, 8p12-pter, 11, 15q11-qter, 16q11-qter, 17p11-pter, 18, 21q11-qter	13q21-qter+, 20p11-pter-	8q11-qter+, 10-, 6p21-pter-
53	7, 8q11-qter, 9q33-qter, 13q11-qter, 20p11-p12, 20q11-qter	4p13-pter, 4q21-qter, 8p12-pter, 15q14-qter, 18q11-qter, 20p12-pter	5p11-pter-, 5q13-qter-, 14q11-qter-	11+, 16p11-pter+, 17q11-qter+, 19+, 21q11-qter+, 22q11-qter+
147	7, 13q11-qter, 20q11-qter	8p21-pter, 18	4p14-pter-, 4q28-qter+, 8p11-21-, 17q11-q2+, 21q11-qter-	11q22-qter+, 16+, 1p11-33-

TABLE 1. Clinical Data

Case number	Age	Sex	Site	Stage <sup>a</sup>	Grade <sup>b</sup>	Diagnosis of metastatic disease <sup>c</sup>
2	40	M	Transverse colon	IV	3	Synchronous
6	79	M	Ascending colon	IV	2	Synchronous
9	73	M	Transverse colon	II	2	N/A
11	56	M	Rectosigmoid	IV	2	Metachronous
12	70	F	Sigmoid colon	IV	2	Synchronous
13	65	M	Descending colon	II	9	Synchronous
14	60	M	Rectum	III	3	Metachronous
15	51	F	Rectum	III	2	Metachronous
19	63	M	Rectosigmoid Junction	III	2	Synchronous
20	63	M	Rectum	IV	9	Metachronous
21	64	F	Sigmoid colon	IV	2	Synchronous
35	71	M	Rectum	III	9	Metachronous
49	72	M	Cecum	IV	3	Synchronous
53	72	F	Sigmoid colon	IV	2	Synchronous
104	61	M	Sigmoid colon	IV	2	Metachronous
105	58	M	Ascending colon	II	2	Metachronous
107	77	F	Cecum	IV	2	Metachronous
108	53	F	Splenic flexure	IV	2	Synchronous
112	68	M	Rectum	III	3	Synchronous
113	41	M	Splenic flexure	IV	2	Synchronous
114	49	M	Splenic flexure	IV	3	Synchronous
116	73	M	Rectosigmoid	III	9	Metachronous
120	24	F	Descending colon	IV	2	Synchronous
123	62	F	Rectum	III	2	Metachronous
124	42	M	Rectum	IV	9	Synchronous
145	70	M	Rectosigmoid	IV	2	Synchronous
147	86	F	Cecum	IV	2	Synchronous

<sup>a</sup>AJCC/UICC staging system (Hutter and Sabin, 1986).

<sup>b</sup>Grade of primary tumor: 1–3, low, moderate, high grade; 9, grading unknown.

<sup>c</sup>Synchronous, diagnosis of metastatic disease within 12 months following diagnosis of primary tumor; metachronous, diagnosis of metastatic disease after 12 months or later.

GENES, CHROMOSOMES & CANCER 25:82–90 (1999)  
Chromosome Arm 20q Gains and Other Genomic Alterations in Colorectal Cancer Metastatic to Liver, as Analyzed by Comparative Genomic Hybridization and Fluorescence In Situ Hybridization

W. Michael Korn,<sup>1</sup>\* Toru Yasutake,<sup>2</sup> Wen-Lin Kuo,<sup>1</sup> Robert S. Warren,<sup>3</sup> Colin Collins,<sup>1</sup> Masao Tomita,<sup>2</sup> Joe Gray,<sup>1</sup> and Frederic M. Waidman<sup>1</sup>

# CNVs Come in a Variety of Formats: VCF

## Issue 1: There are two fields to specify SV/CNV

#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO
1	2827694	rs2376870	CGTGGATGCGGGAC	C	.	PASS	SVTYPE=DEL;END=282770
2	321682	.	T	<DEL>	6	PASS	SVTYPE=DEL;END=321887
2	14477084	.	C	<DEL:ME:ALU>	12	PASS	SVTYPE=DEL;END=144773
3	9425916	.	C	<INS:ME:L1>	23	PASS	SVTYPE=INS;END=942591
3	12665100	.	A	<DUP>	14	PASS	SVTYPE=DUP;END=126862
4	18665128	.	T	<DUP:TANDEM>	11	PASS	SVTYPE=DUP;END=186652

1) Symbolic allele (SA)

2) SVTYPE

- DEL Deletion relative to the reference
  - INS Insertion of novel sequence relative to the reference
  - DUP Region of elevated copy number relative to the reference
  - INV Inversion of reference sequence
  - CNV Copy number variable region (may be both deletion and duplication)
  - BND Breakend
- The CNV category should not be used when a more specific category can be applied. Reserved subtypes include:
- DUP:TANDEM Tandem duplication
  - DEL:ME Deletion of mobile element relative to the reference
  - INS:ME Insertion of a mobile element relative to the reference



- DEL: Deletion relative to the reference
- INS: Insertion of novel sequence relative to the reference
- DUP: Region of elevated copy number relative to the reference
- INV: Inversion of reference sequence
- CNV: Copy number variable region (may be both deletion and duplication)
- BND: Breakend

## VCF v4.4 deprecate SVTYPE

#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO	FOR
chrA	2	.	TGC	T	.	.	EVENT=DEL_seq	
chrA	2	.	T	<DEL>	.	.	SVLEN=2;SVCLAIM=DJ;EVENT=DEL_symbolic;END=4	
chrA	2	delbp1	T	T[chrA:5[ .	.	.	MATEID=delbp2;EVENT=DEL_split_bp_cn	
chrA	2	delbp2	A	]chrA:2]A	.	.	MATEID=delbp1;EVENT=DEL_split_bp_cn	
chrA	2	.	T	<DEL>	.	.	SVLEN=2;SVCLAIM=D;EVENT=DEL_split_bp_cn;END=4	
chrA	5	.	G	GAAA	.	.	EVENT=homology_seq	
chrA	5	.	G	<DUP>	.	.	SVLEN=3;CIPOS=0,5;EVENT=homology_dup	
chrA	14	.	T	<INS>	.	.	IMPRECISE;SVLEN=100;CILEN=-50,50;CIPOS=-10,10;END=14	
chrA	14	.	G	.CCCCCG	.	.	EVENT=single_breakend	

Symbolic allele (SA)

- DEL Region of lowered copy number relative to the reference, or a deletion breakpoint
  - INS Insertion of novel sequence relative to the reference
  - DUP Region of elevated copy number relative to the reference, or a tandem duplication breakpoint
  - INV Inversion of reference sequence
  - CNV Copy number variable region (may be both deletion and duplication)
- The CNV category should not be used when a more specific category can be applied. Implementations are free to define their own subtypes. The presence of a subtype does not change either the copy number or breakpoint interpretation of a symbolic structural variant allele. The following subtypes are recommended:
- DUP:TANDEM Tandem duplication
  - DEL:ME Deletion of mobile element relative to the reference
  - INS:ME Insertion of a mobile element relative to the reference

Note that the position of symbolic structural variant alleles is the position of the base immediately preceding the variant.

Reserved specific subtypes



- using genome positions (POS, INFO.END) for start, end mappings
- treatment of markers for imprecision during matching is left to the implementer
- DUP, DEL are interpreted as indicators for the type of copy number change

Use Subtype to define new structural variant

- <DUP:TANDEM> precise form of duplication
- <DEL:ME:LINE>

Subtypes do not change the meaning of symbolic allele.



# Progenetix Oncogenomics Resource

## CNV Profiles for Most Cancer Entities from Individual Experiments

# 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

**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<sup>+</sup>**

**Documentation**

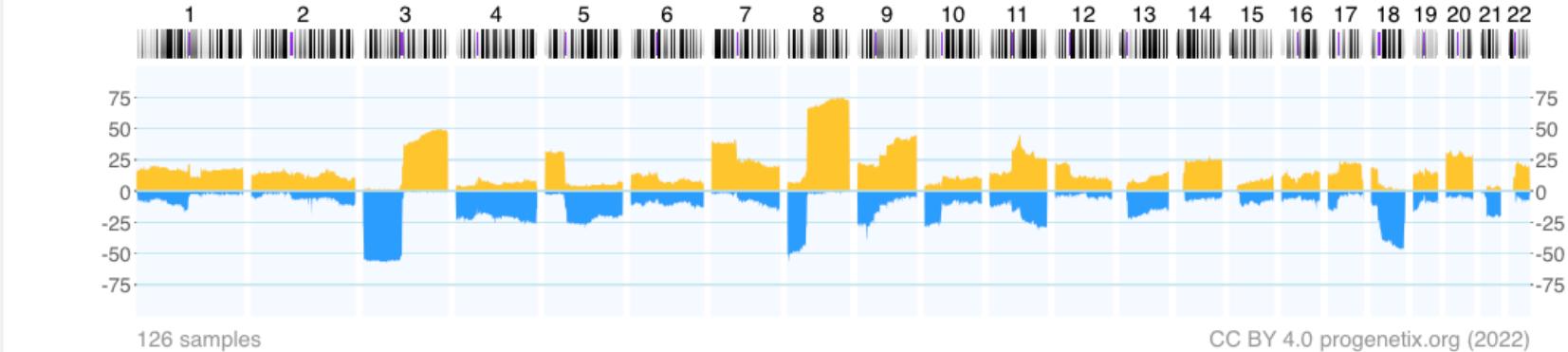
- News
- Downloads & Use Cases
- Sevices & API

**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.

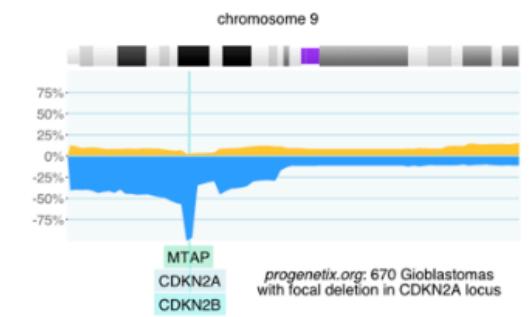
### Floor of the Mouth Neoplasm (NCIT:C4401)



[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 in 2022

## Cancer Genomics Reference Resource

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- publication database and code mapping services

progenetix

Search Samples Modify Query

Assembly: GRCh38 Chro: 9 Start: 21500001-21975098 End: 21967753-22500000

Type: DEL Filters: NCIT:C3058

progenetix

Samples: 668 Variants: 286 Calls: 675

Found Variants (.pgxseg) All Sample Variants (.json) All Sample Variants (.pgxseg) Show Variants in UCSC

UCSC region JSON Response Visualization options

Results Biosamples Biosamples Map Variants

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22

75% 50% 25% 0% -25% -50% -75%

progenetix: 670 samples

CC BY 4.0 progenetix.org (2021)

Matched Subset Codes	Subset Samples	Matched Samples	Subset Match Frequencies
UBERON:0002021	4	1	0.250
icdot-C71.4	4	1	0.250
icdom-94403	4291	664	0.155
NCIT:C3058	4375	664	0.152
UBERON:0016525	14	2	0.143
icdot-C71.1	14	2	0.143
UBERON:0000955	7068	651	0.092
icdot-C71.9	7066	651	0.092
icdom-94423	84	4	0.048
NCIT:C3796	84	4	0.048
UBERON:0001869	1712	14	0.008
icdot-C71.0	1712	14	0.008

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Cancer CNV Profiles

Search Samples

Studies & Cohorts

arrayMap

TCGA Samples

DIPG Samples

Gao & Baudis, 2021

Cancer Cell Lines

Publication DB

Genome Profiling

Progenetix Use

Services

NCIt Mappings

UBERON Mappings

Upload & Plot

Download Data

Beacon<sup>+</sup>

Progenetix Info

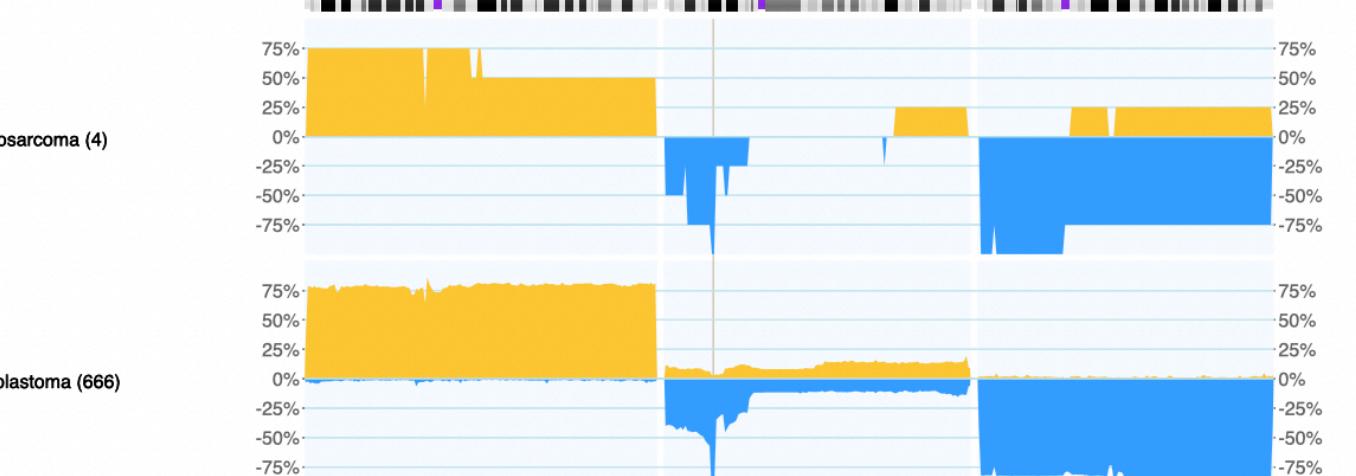
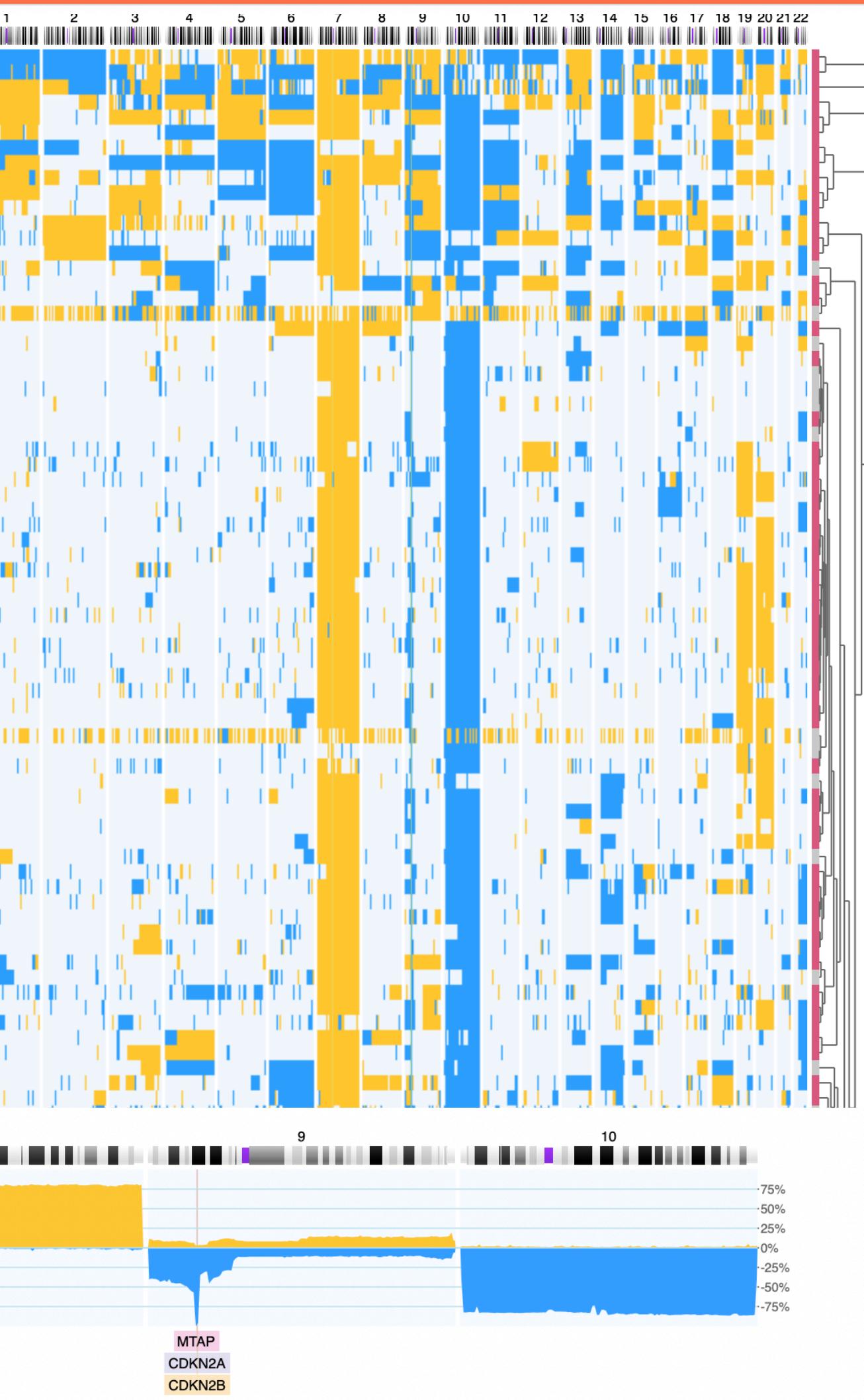
About Progenetix

Use Cases

Documentation

Baudisgroup @ UZH

pgxcs-kftwze6  
pgxcs-kftwlz7d  
pgxcs-kftwzab  
pgxcs-kftwzum  
pgxcs-kftw55qo  
pgxcs-kftwluql  
pgxcs-kftwluvf  
pgxcs-kftw3hd8  
pgxcs-kftw4v2q  
pgxcs-kftw3596  
pgxcs-kftwzkd4  
pgxcs-kftw4gw1  
pgxcs-kftwybv  
pgxcs-kftwuisse  
pgxcs-kftwejw (20215515)  
pgxcs-kftvh60  
pgxcs-kftwuj1y  
pgxcs-kftvmnxn (23079654)  
pgxcs-kftw60u2 (18167341)  
pgxcs-kftwluia  
pgxcs-kftwn4q (18077431)  
pgxcs-kftwcov7 (21102433)  
pgxcs-kftw6gv (19139420)  
pgxcs-kftwur8g  
pgxcs-kftw7w91 (18828157)  
pgxcs-kftw1odk  
pgxcs-kftw1zb  
pgxcs-kftw3oev  
pgxcs-kftwlu0t  
pgxcs-kftwukhg  
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pgxcs-kftwxez9  
pgxcs-kftwukjd  
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pgxcs-kftwluqd  
pgxcs-kftwyefi  
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pgxcs-kftvazp  
pgxcs-kftwx0a  
pgxcs-kftwuk38  
pgxcs-kftwuke1  
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pgxcs-kftvmoav (23079654)  
pgxcs-kftw52ga  
pgxcs-kftw52d8  
pgxcs-kftwupf9  
pgxcs-kftwuk2a  
pgxcs-kftwuiub  
pgxcs-kftw8ja (21884817)  
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pgxcs-kftv40u  
pgxcs-kftvhueo  
pgxcs-kftvx29g  
pgxcs-kftwummg  
pgxcs-kftwzmk (23468990)  
pgxcs-kftw16vy  
pgxcs-kftw52z3  
pgxcs-kftvz31  
pgxcs-kftwugqy  
pgxcs-kftwifv  
pgxcs-kftw08qx  
pgxcs-kftwuihs



# Progenetix in 2022

## Cancer Genomics Reference Resource

- contains special data subsets, identified using the "cohorts" concept
  - ▶ TCGA CNV data
  - ▶ 1000Genomes germline CNVs (WGS)
  - ▶ Cancer cell line CNVs with upcoming addition of annotated SNV ... data
  - ▶ cBioPortal studies
  - ▶ ...

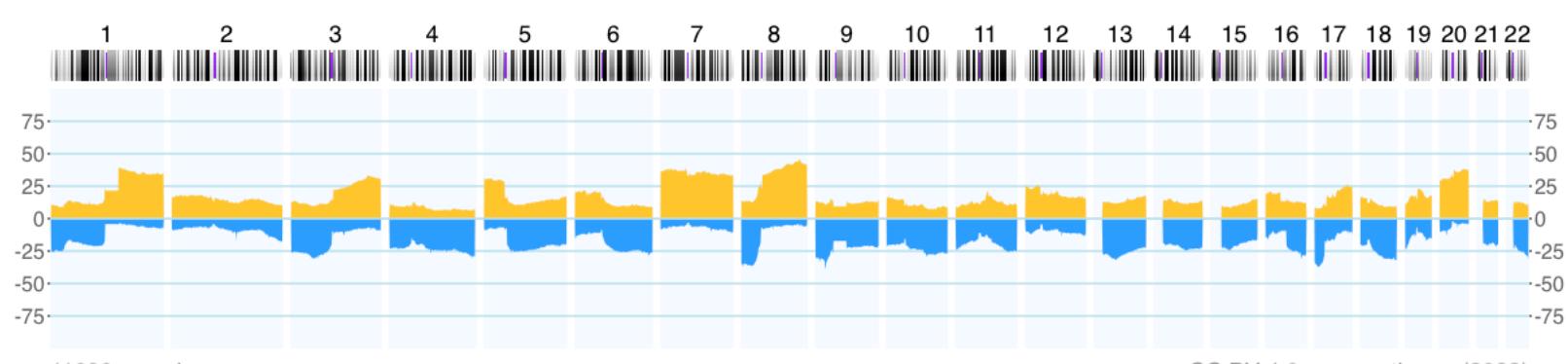


TCGA CNV Data

Search Genomic CNV Data from TCGA

This search page accesses the TCGA subset of the Progenetix collection, based on 22142 samples (tumor and references) from The Cancer Genome Atlas project. The results are based upon data generated by the [TCGA Research Network](#). Disease-specific subsets of TCGA data (aka. projects) can be accessed below.

TCGA Cancer samples (pgx:cohort-TCGAcancers)



1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22

75  
50  
25  
0  
-25  
-50  
-75

11090 samples

CC BY 4.0 progenetix.org (2022)

[Download SVG](#) | [Go to pgx:cohort-TCGAcancers](#) | [Download CNV Frequencies](#)

[Edit Query](#)

Publication DB

- Genome Profiling
- Progenetix Use

Services

- NCIt Mappings
- UBERON Mappings

Upload & Plot

Beacon<sup>+</sup>

Documentation

News

Filter subsets e.g. by prefix

Hierarchy Depth: 2 levels

No Selection

- pgx:TCGA-ACC: TCGA ACC project (180 samples)
- pgx:TCGA-BLCA: TCGA BLCA project (810 samples)
- pgx:TCGA-BRCA: TCGA BRCA project (2219 samples)



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progenetix Cellosaurus

Cell Lines

Progenetix

Publication DB

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News

Downloads & Use Cases

Services & API

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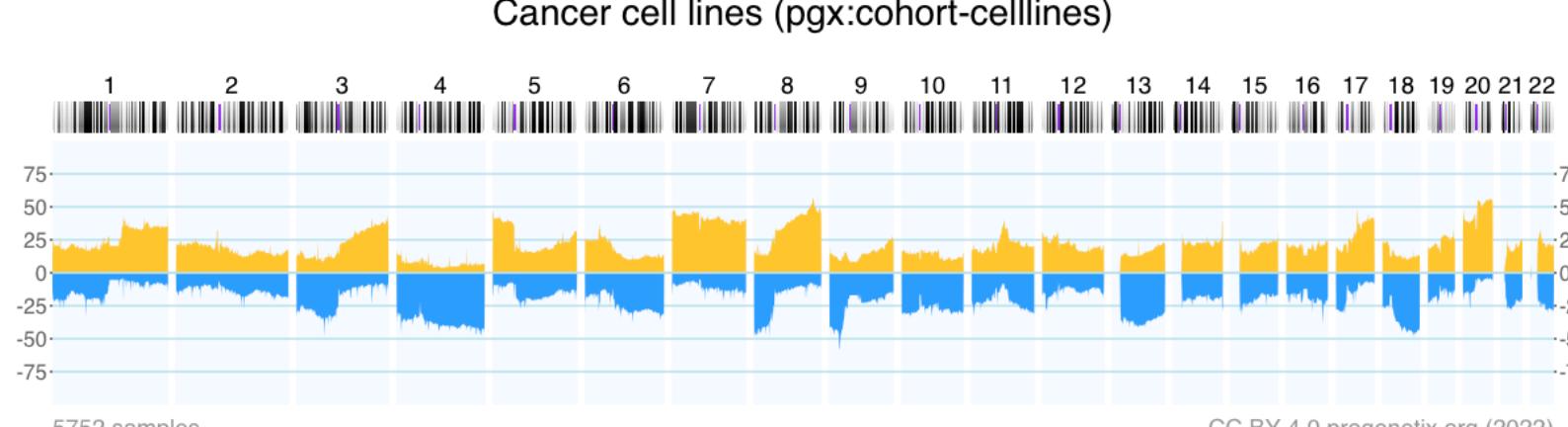
Cancer Cell Lines

Cancer cell line CNVs

This search page uses Progenetix cell line copy number variation data. These data include cancer cell lines that have been mapped to [Cellosaurus](#) – a knowledge resource on cell lines.

CNV Frequency Plot

Cancer cell lines (pgx:cohort-celllines)



5752 samples

CC BY 4.0 progenetix.org (2022)

[Download SVG](#) | [Go to pgx:cohort-celllines](#) | [Download CNV Frequencies](#)

Edit Query

Cell Lines (with parental/derived hierarchies)

Filter subsets e.g. by prefix

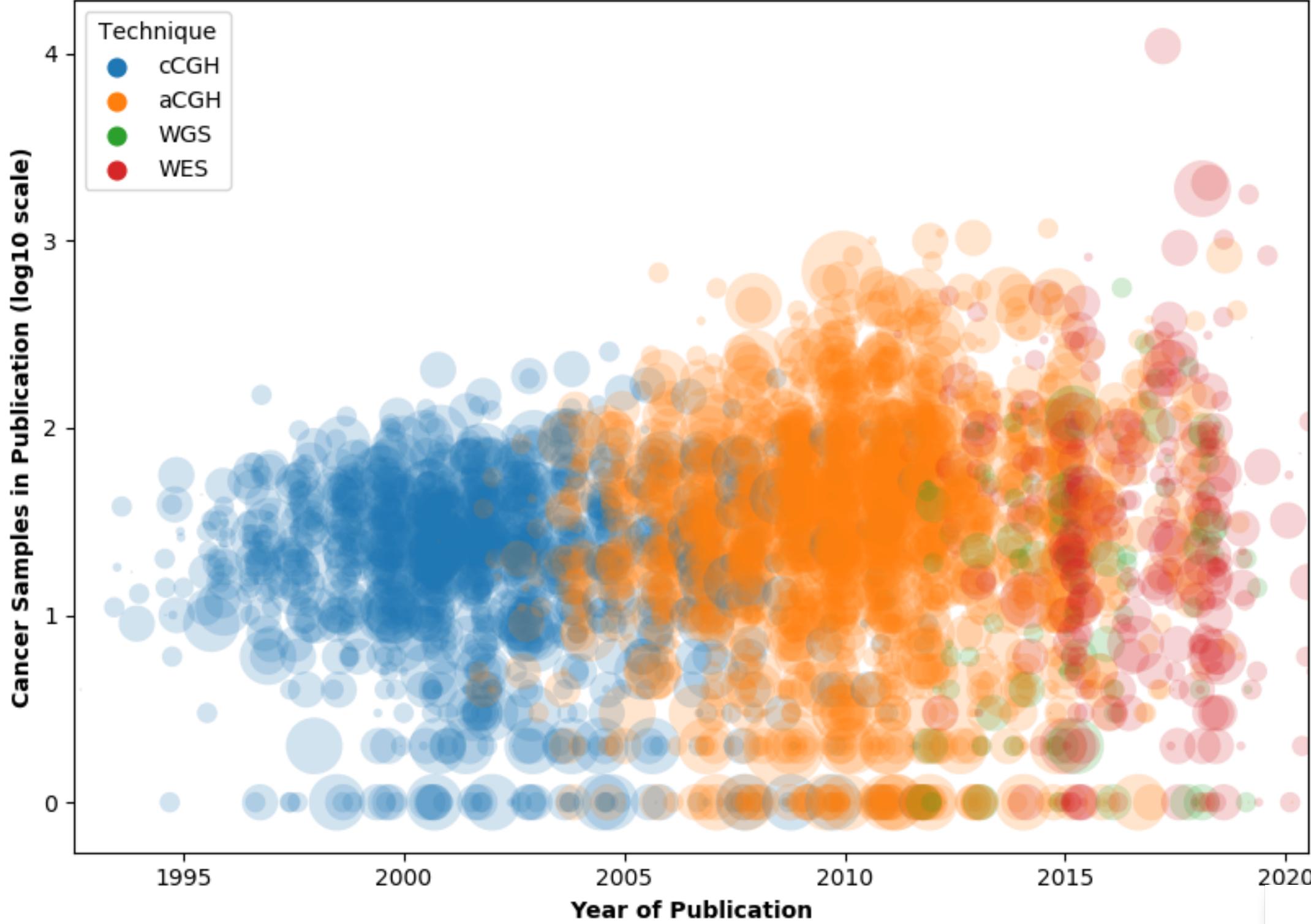
Hierarchy Depth: 2 levels

No Selection

cellosaurus:CVCL\_0001: HEL (5 samples)

cellosaurus:CVCL\_2481: HEL 92.1.7 (3 samples)

## Number of tumor samples for each publication across the years



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.



## 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.

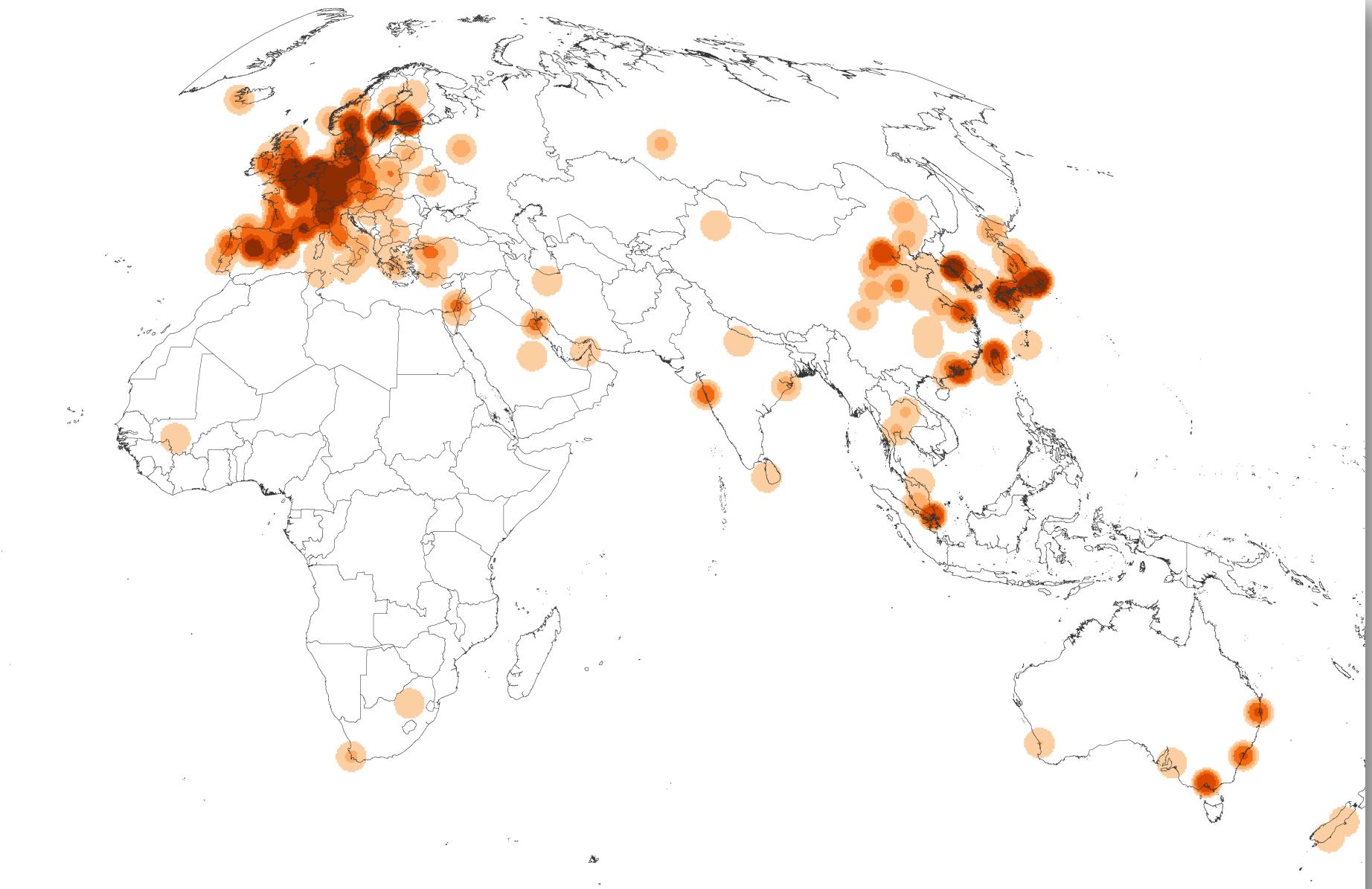
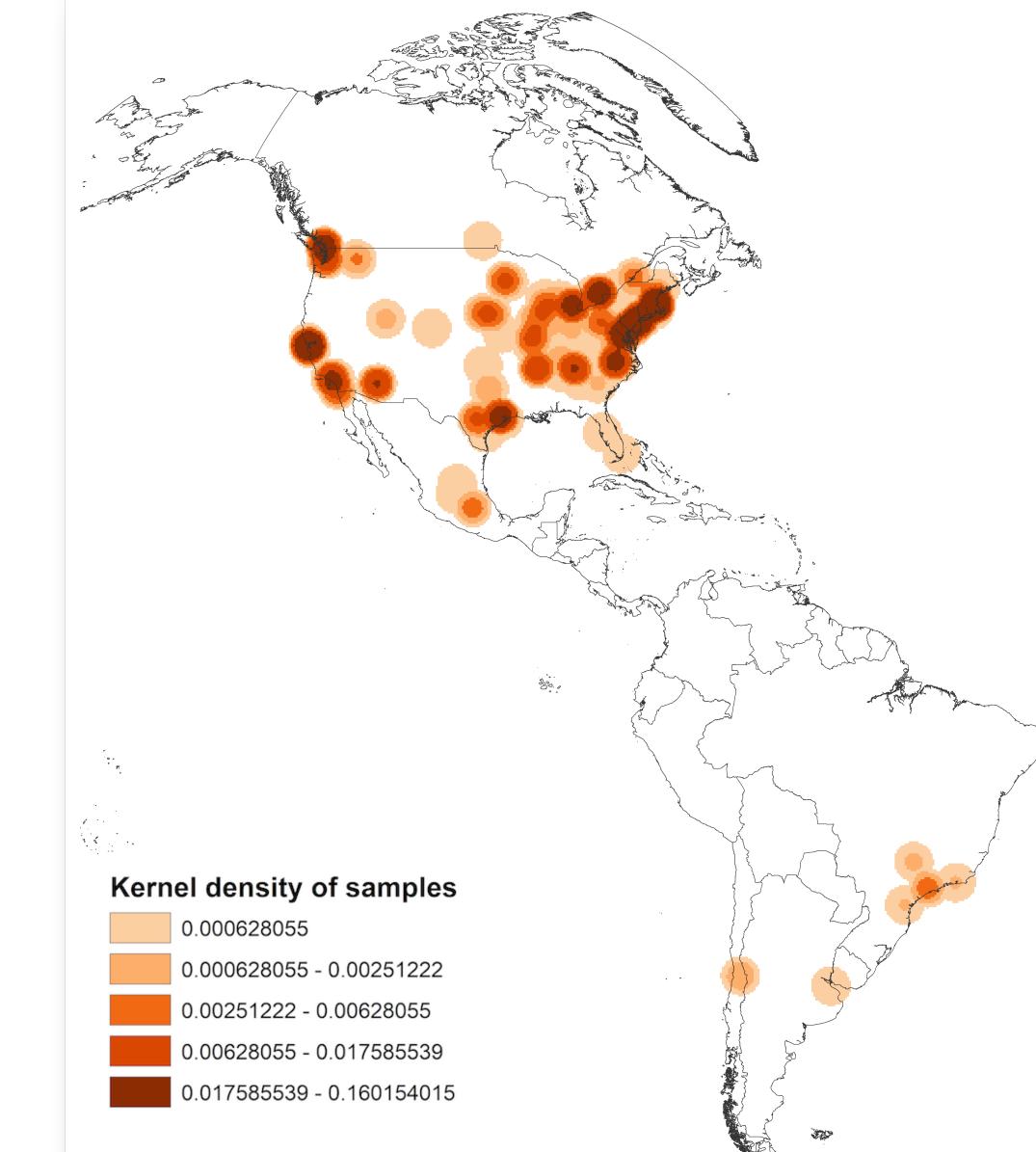
Please [contact us](#) to alert us about additional articles you are aware of. The inclusion criteria are described in the documentation [🔗](#).

Filter [i](#) City [i](#)

  Type to search... [▼](#)

### Publications (3324)

id <a href="#">i</a> ▾	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 ... <i>BMC Med Genomics</i>	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. ... <i>bioRxiv</i>	0	0	5	113	0



# 200+ Genomic Data Initiatives Globally

Clinical/Genomic  
Medicine



Research



National



Cohorts



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





# Enabling genomic data sharing for the benefit of human health

The Global Alliance for Genomics and Health (GA4GH) is a policy-framing and technical standards-setting organization, seeking to enable responsible genomic data sharing within a **human rights framework**



**Genomic Data  
Toolkit**



**Regulatory & Ethics  
Toolkit**



**Data Security  
Toolkit**



[VIEW OUR LEADERSHIP](#)

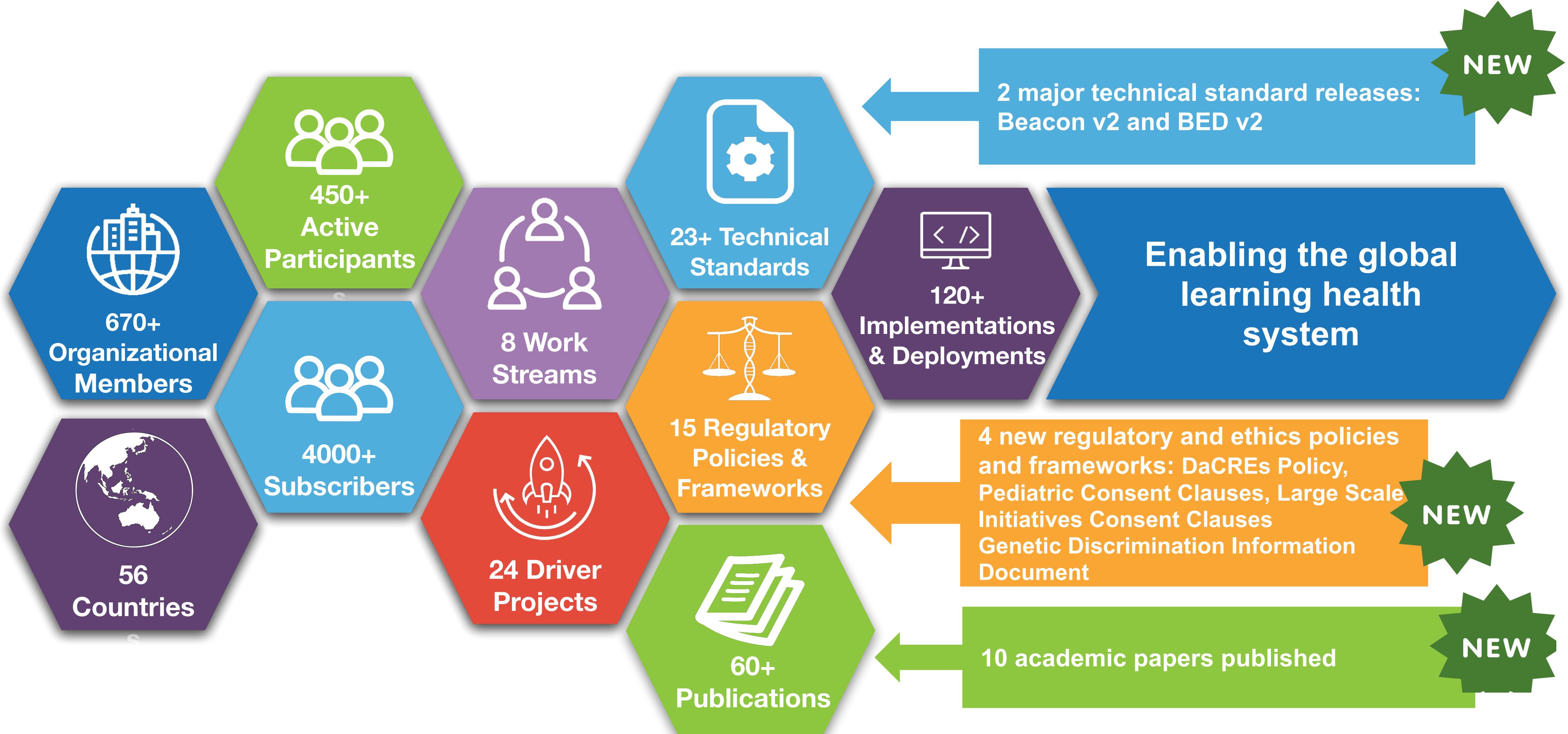
[MORE ABOUT US](#)

[BECOME A MEMBER](#)

# The GA4GH ecosystem and outputs



Global Alliance  
for Genomics & Health



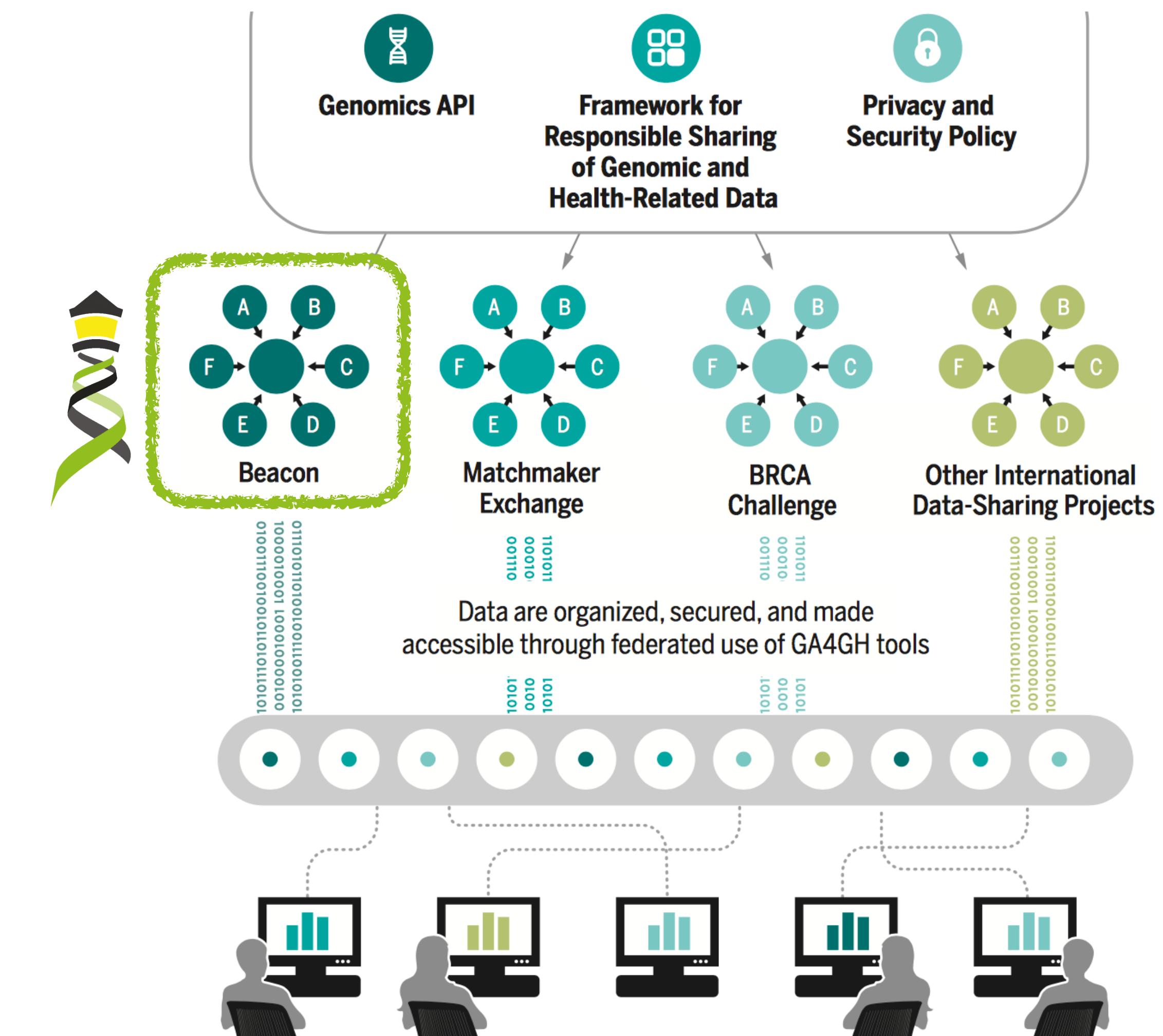


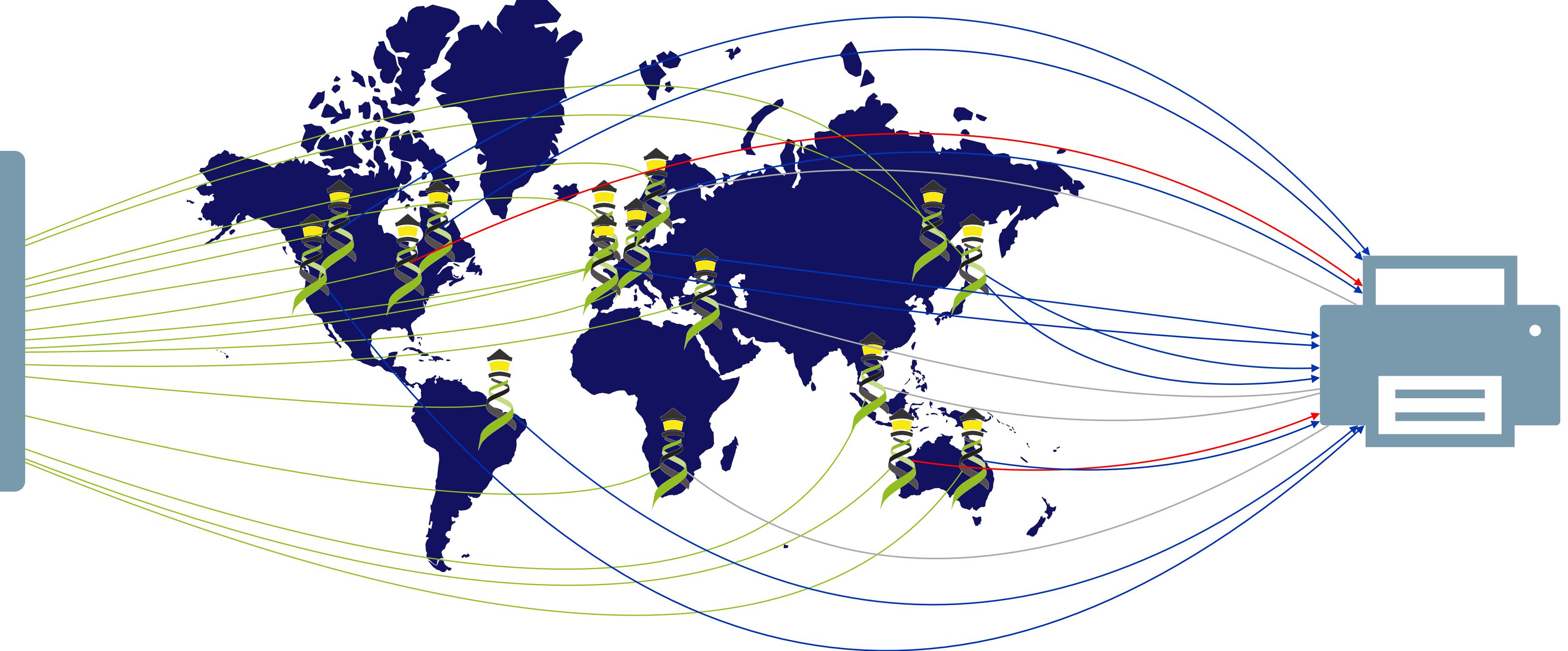
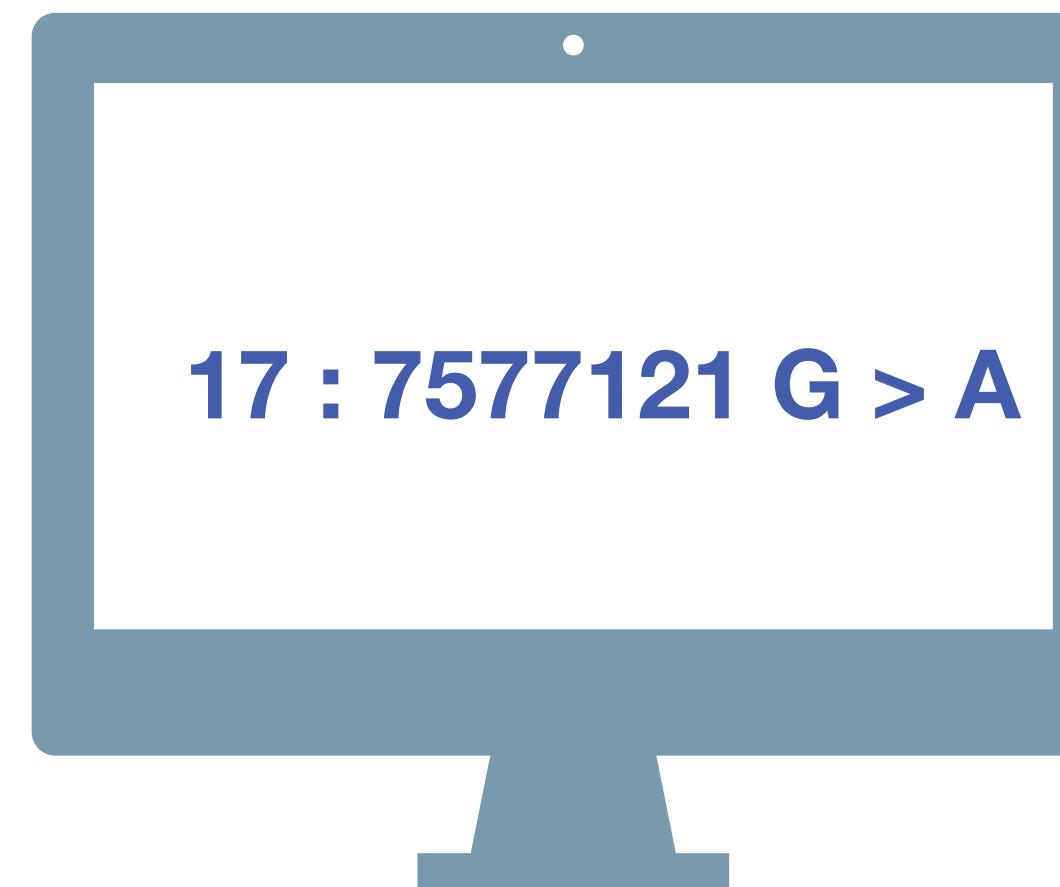
## GENOMICS

# *A federated ecosystem for sharing genomic, clinical data*

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

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





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**.

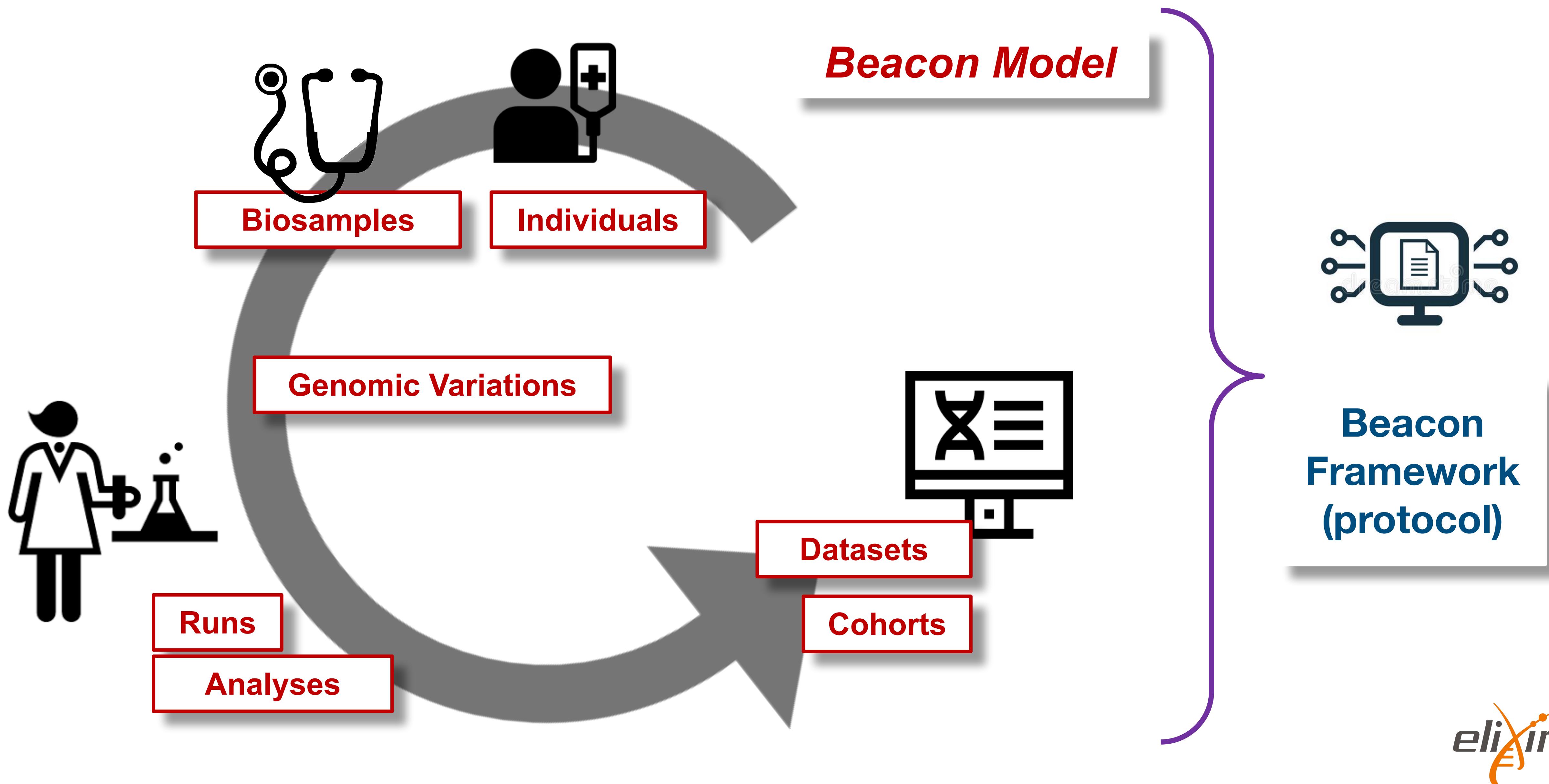


# Making Beacons Biomedical - Beacon v2

- Scoping queries through "biodata" parameters
- Extending the queries towards clinically ubiquitous variant formats
  - ▶ cytogenetic annotations, named variants, variant effects
- Beacon queries as entry for **data delivery**
  - ▶ Beacon v2 permissive to respond with variety of data types
    - Phenopackets, biosample data, cohort information ...
  - ▶ handover to stream and download using htsgt, VCF, EHRs
- Interacting with EHR standards
  - ▶ FHIR translations for queries and handover ...
- Beacons as part of local, secure environments
- Authentication to enable non-aggregate, patient derived datasets
  - ▶ ELIXIR AAI with compatibility to other providers (OAuth...)

# Beacon v2

docs.genomebeacons.org



# Progenetix & GA4GH Beacon v2

**A custom "full stack" implementation of a genomics resource  
around Beacon data model & API**



# 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

Beacon v2 GA4GH Approval Registry

Beacons:  European Genome-Phenome Archive |  progenetix |  cnag |  UNIVERSITY OF LEICESTER

 European Genome-Phenome Archive (EGA)

[Visit us](#) [Beacon API](#) [Contact us](#)

GA4GH Approval Beacon Test

This [Beacon](#) is based on the GA4GH Beacon [v2.0](#)

BeaconMap	██████████
Bioinformatics analysis	██████████
Biological Sample	██████████
Cohort	██████████
Configuration	██████████
Dataset	██████████
EntryTypes	██████████
Genomic Variants	██████████
Individual	██████████
Info	██████████
Sequencing run	██████████

 progenetix+

[Visit us](#) [Beacon UI](#) [Beacon API](#) [Contact us](#)

Theoretical Cytogenetics and Oncogenomics group at UZH and SIB

Progenetix Cancer Genomics Beacon+ provides a forward looking implementation of the Beacon v2 API, with focus on structural genome variants and metadata based on the...

BeaconMap	██████████
Bioinformatics analysis	██████████
Biological Sample	██████████
Cohort	██████████
Configuration	██████████
Dataset	██████████
EntryTypes	██████████
Genomic Variants	██████████
Individual	██████████
Info	██████████
Sequencing run	██████████

 Centre Nacional Analisis Genomica (CNAG-CRG)

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Beacon @ RD-Connect

This [Beacon](#) is based on the GA4GH Beacon [v2.0](#)

BeaconMap	██████████
Bioinformatics analysis	██████████
Biological Sample	██████████
Cohort	██████████
Configuration	██████████
Dataset	██████████
EntryTypes	██████████
Genomic Variants	██████████
Individual	██████████
Info	██████████
Sequencing run	██████████

 University of Leicester

[Beacon UI](#) [Beacon API](#) [Contact us](#)

Cafe Variome Beacon v2

This [Beacon](#) is based on the GA4GH Beacon [v2.0](#)

BeaconMap	██████████
Bioinformatics analysis	██████████
Biological Sample	██████████
Cohort	██████████
Configuration	██████████
Dataset	██████████
EntryTypes	██████████
Genomic Variants	██████████
Individual	██████████
Info	██████████
Sequencing run	██████████

Matches the Spec Not Match the Spec Not Implemented


# Progenetix

## Genomic resource utilizing Beacon v2 calls

- Progenetix uses Beacon v2 queries to drive its UI
- all individuals, biosamples, variants, analyses matched by a given query are stored by their object ids
- handovers for variant purposes (e.g. to retrieve all matched variants) are returned in the original response and asynchronously retrieved by the front end app

Edit Query

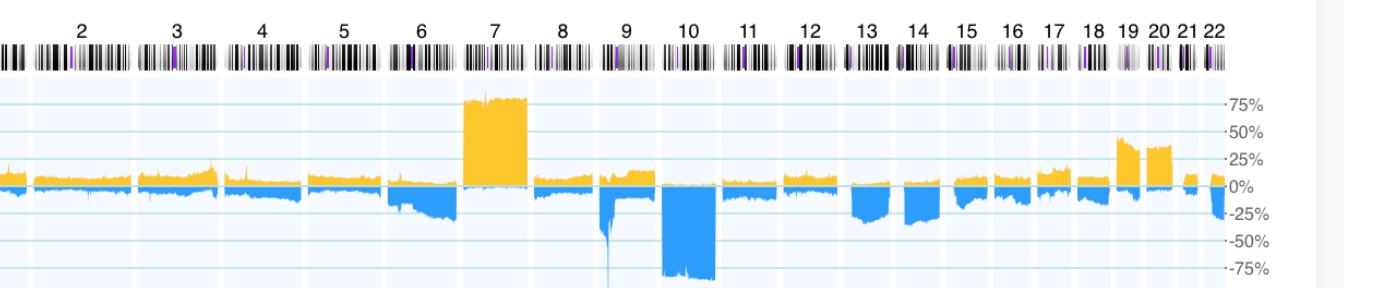
Assembly: GRCh38 Chro: 9 Start: 21500001-21975098 End: 21967753-22500000  
Type: EFO:0030067 Filters: NCIT:C3058

progenetix

Matched Samples: 660 Retrieved Samples: 660 Variants: 279 Calls: 667

UCSC region [↗](#) Variants in UCSC [↗](#) Dataset Response (JSON) [↗](#) Visualization options

Results Biosamples Biosamples Map Variants Annotated Variants



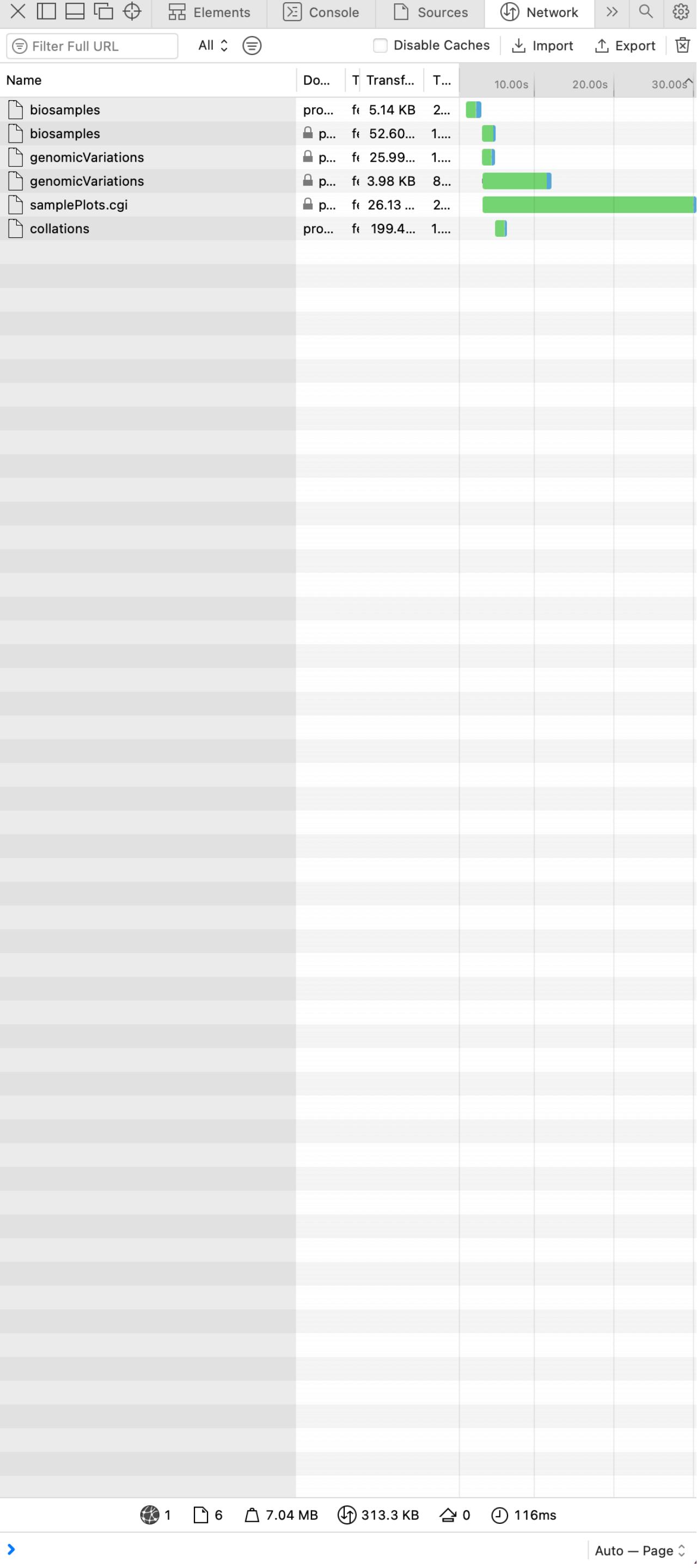
progenetix: 662 samples CC BY 4.0 progenetix.org (2022)

Matched Subset Codes	Subset Samples	Matched Samples	Subset Match Frequencies
UBERON:0002021	4	1	0.250
pgx:icdot-C71.4	4	1	0.250
pgx:icdom-94403	4286	656	0.153
NCIT:C3058	4370	656	0.150
UBERON:0016525	14	2	0.143
pgx:icdot-C71.1	14	2	0.143
UBERON:0000955	7199	643	0.089
pgx:icdot-C71.9	7204	643	0.089
pgx:icdom-94423	84	4	0.048
NCIT:C3796	84	4	0.048
UBERON:0001869	1714	14	0.008
pgx:icdot-C71.0	1714	14	0.008

Download Sample Data (TSV)  
1-660 [↗](#)

Download Sample Data (JSON)  
1-660 [↗](#)

Download Sample Variants (JSON)  
1-660 [↗](#)



# Progenetix

## Genomic resource utilizing Beacon v2 calls

- Progenetix uses Beacon v2 queries to drive its UI
- all individuals, biosamples, variants, analyses matched by a given query are stored by their object ids
- handovers for variant purposes (e.g. to retrieve all matched variants) are returned in the original response and asynchronously retrieved by the front end app

The screenshot shows the Progenetix UI with a search bar at the top. Below it, a summary box displays assembly (GRCh38), chromosome (9), start (21500001-21975098), end (21967753-22500000), type (EFO:0030067), and filters (NCIT:C3058). A 'progenetix' logo is visible. The main area has tabs for Results, Biosamples, Biosamples Map, Variants, and Annotated Variants. A yellow callout highlights a 'Results' section with a histogram and a complex API query:

```
/beacon/biosamples/?  
requestedGranularity=record&limit=1000&skip=0  
&assemblyId=GRCh38&referenceName=9&variantType=EFO:0030067  
&start=21500000,21975098&end=21967753,22500000  
&filters=NCIT:C3058
```

A cyan callout highlights a 'Biosamples' section with another API query:

```
/beacon/biosamples/?  
skip=0&limit=1000  
&accessid=fbffda57-0f41-4d6a-99fc-41d4cfdea9f6&requestedSchema=biosample
```

A pink callout highlights a 'Variants' section with a third API query:

```
/beacon/genomicVariations/?  
accessid=e2dadd91-9326-46de-97e4-6b88413b6bfe  
&requestedSchema=genomicVariant
```

At the bottom, download links are shown for sample plots and variants, each with its own API query:

Download Sample Plots (JSON)  
1-660

Download Sample Data (UCSC)  
1-660 ↗

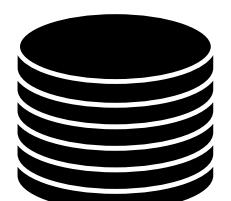
Download Sample Variants (JSON)  
1-660 ↗

On the right, a Network tab in the browser developer tools shows several requests for 'biosamples' and 'genomicVariations' resources.

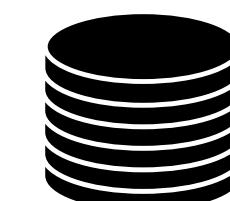
# Progenetix Stack



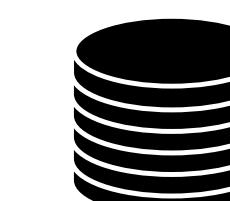
- JavaScript front-end is populated for query results using asynchronous access to multiple handover objects
  - ▶ biosamples and variants tables, CNV histogram, UCSC .bed loader, .pgxseg variant downloads...
- the complete middleware / CGI stack is provided through the *bycon* package 
  - ▶ schemas, 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



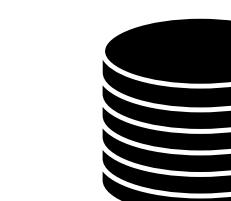
variants



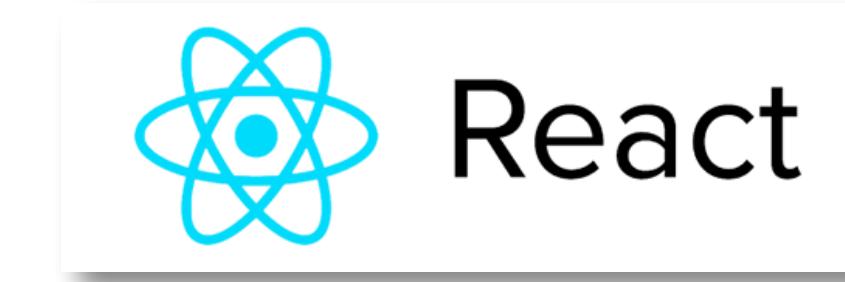
analyses



biosamples

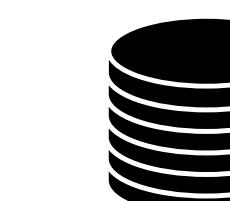


individuals

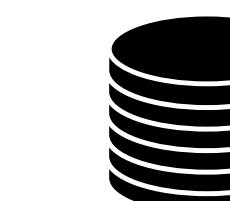


- *collations* contain pre-computed data (e.g. CNV frequencies, statistics) and information for all grouping entity instances and correspond to **filter values**
  - ▶ PMID:10027410, NCIT:C3222, pgx:cohort-TCGA, pgx:icdom-94703...
- *querybuffer* stores id values of all entities matched by a query and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

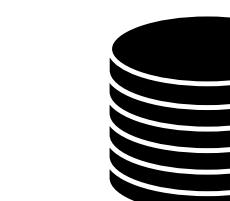
```
_id: ObjectId("6249bb654f8f8d67eb94953b"),
_id: '0765ee26-5029-4f28-b01d-9759abf5bf14',
source_collection: 'variants',
source_db: 'progenetix',
source_key: '_id',
target_collection: 'variants',
target_count: 667,
target_key: '_id',
target_values: [
  ObjectId("5bab578b727983b2e00ca99e"),
  ObjectId("5bab578d727983b2e00cb505")]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



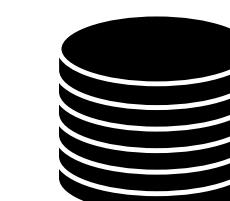
collations



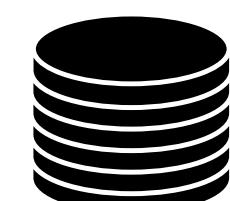
geolocs



genespans



publications



qBuffer

Entity collections

Utility collections

# Beacon v1 => v2

## Genomic variation queries

- Beacon v2 defines query schemas through JSON Schema documents for POST requests and REST paths in OpenAPI documents
- Additional variant parameters:
  - **variantType**, mateName (existing in v1)
  - geneld
  - variantMinLength, variantMaxLength
  - aminoacidChange
  - genomicAlleleShortForm

```
{  
    "$$schema": "beaconRequestBody.json",  
    "meta": {  
        "apiVersion": "2.0",  
        "requestedSchemas": [  
            {  
                "entityType": "genomicVariation",  
                "schema": "https://  
raw.githubusercontent.com/ga4gh-beacon/beacon-v2/  
main/models/json/beacon-v2-default-model/  
genomicVariations/defaultSchema.json"  
            }  
        ]  
    },  
    "query": {  
        "requestParameters": {  
            "g_variant": {  
                "referenceName": "NC_000017.11",  
                "start": [7577120],  
                "referenceBases": "G",  
                "alternateBases": "A"  
            }  
        }  
    },  
    "requestedGranularity": "record",  
    "pagination": {  
        "skip": 0,  
        "limit": 5  
    }  
}
```

# Beacon & CNVs

## Open types w/ some definitions

- Beacon supports structural variant queries through the **variantType** parameter
- The default model **does not prescribe** which types can be used (but documents VCF derived DUP & DEL)
- CNV values are not (yet) supported but EFO offers common classes
- Progenetix supports EFO *relative CN terms* (but accepts & interpolates DUP & DEL)

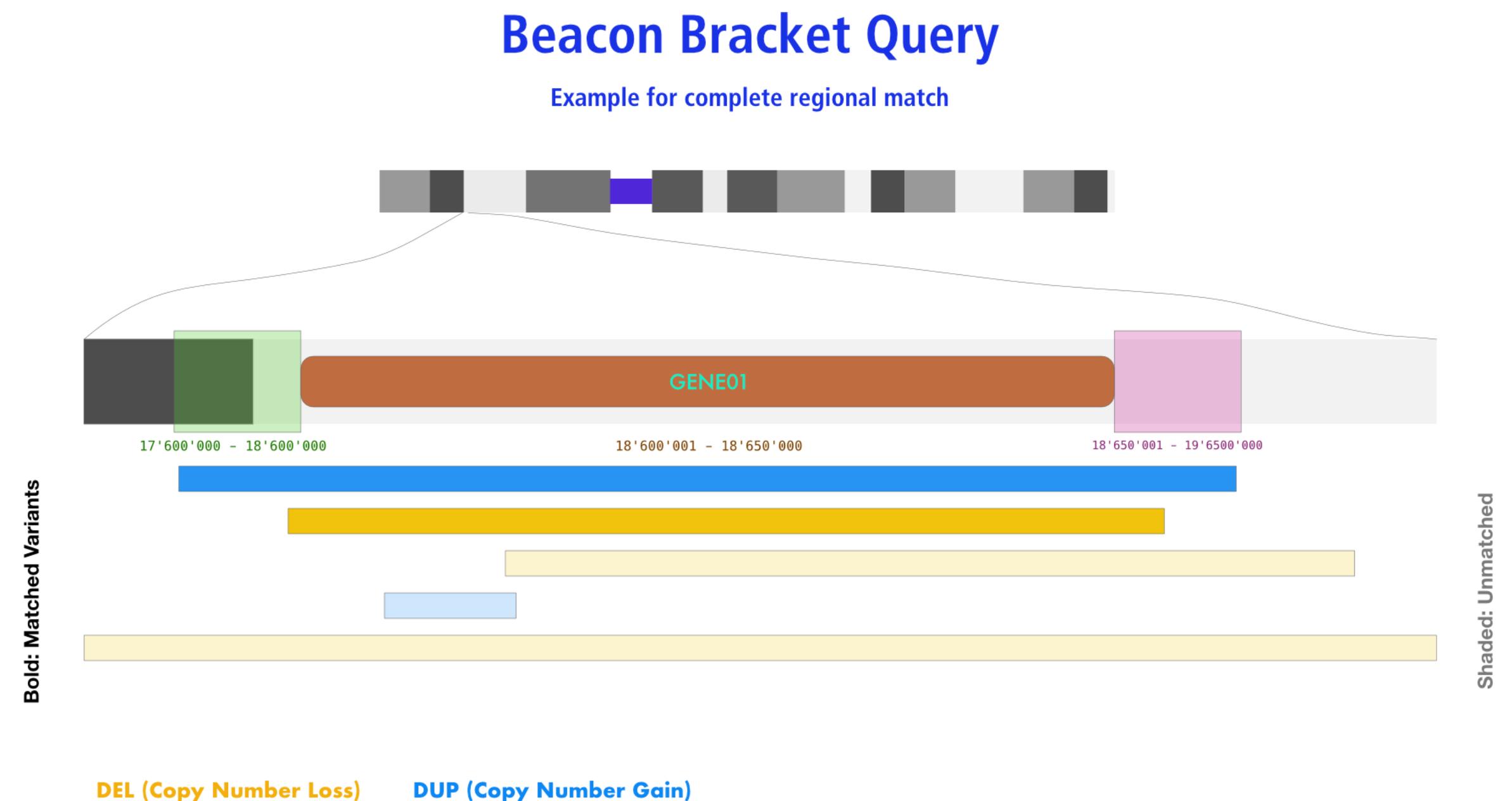
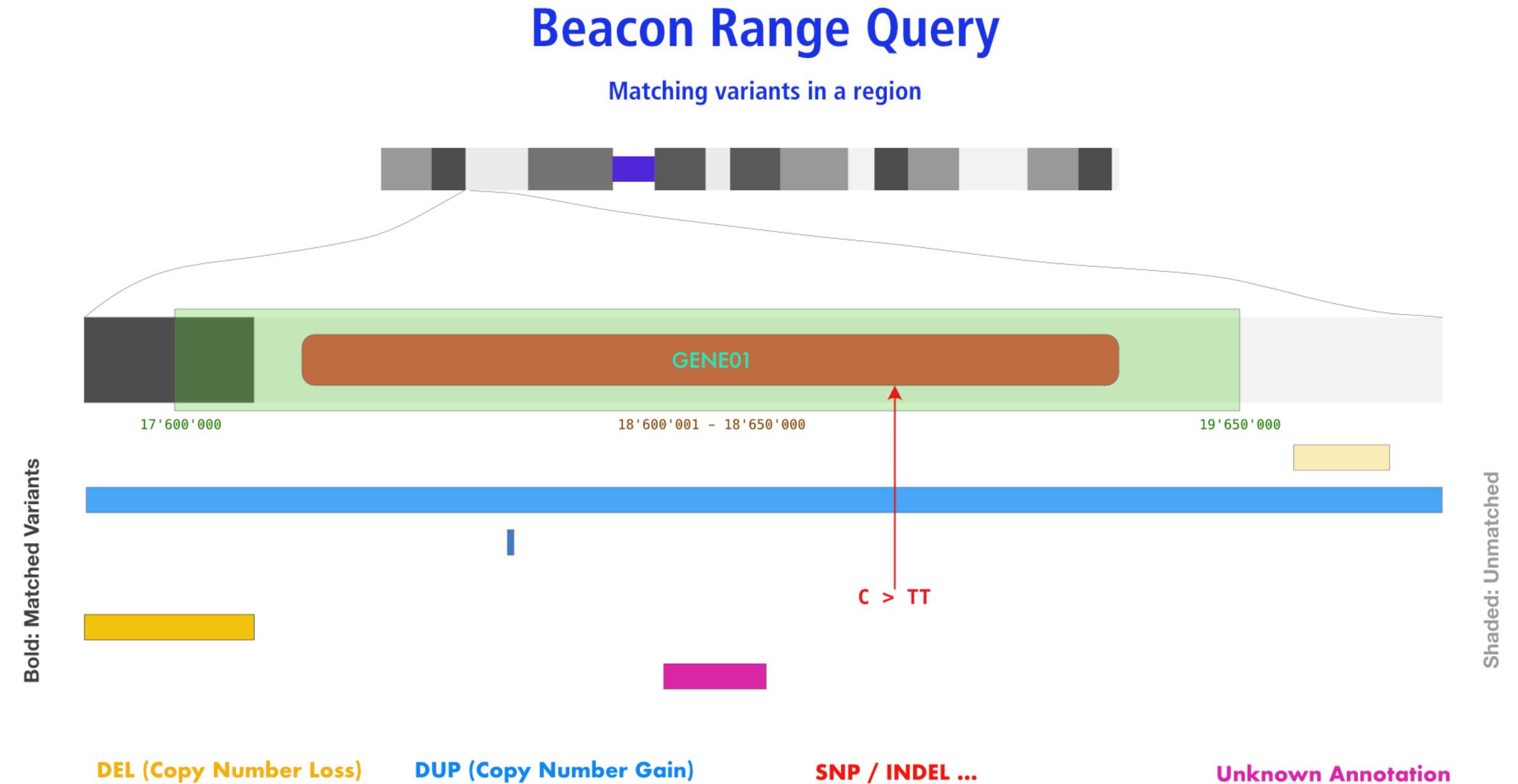
Beacon	VCF	SO	EFO	VRS	Notes
DUP	DUP <sup>1</sup>	SO:0001742 copy_number_gain	EFO:0030070 copy number gain	low-level gain (implicit)	a sequence alteration whereby the copy number of a given genomic region is greater than the reference sequence
DUP	DUP <sup>1</sup>	SO:0001742 copy_number_gain	EFO:0030071 low-level copy number gain	low-level gain	
DUP	DUP <sup>1</sup>	SO:0001742 copy_number_gain	EFO:0030072 high-level copy number gain	high-level gain	commonly but not consistently used for >=5 copies on a bi-allelic genome region
DUP	DUP <sup>1</sup>	SO:0001742 copy_number_gain	EFO:0030073 focal genome amplification	high-level gain	commonly but not consistently used for >=5 copies on a bi-allelic genome region, of limited size (operationally max. 1-5Mb)
DEL	DEL <sup>1</sup>	SO:0001743 copy_number_loss	EFO:0030067 copy number loss	partial loss (implicit)	a sequence alteration whereby the copy number of a given genomic region is smaller than the reference sequence
DEL	DEL <sup>1</sup>	SO:0001743 copy_number_loss	EFO:0030068 low-level copy number loss	partial loss	
DEL	DEL <sup>1</sup>	SO:0001743 copy_number_loss	EFO:0030069 complete genomic deletion	complete loss	complete genomic deletion (e.g. homozygous deletion on a bi-allelic genome region)

<sup>1</sup> VCFv4.4 introduces an SVCLAIM field to disambiguate between in situ events (such as tandem duplications; known adjacency/ break junction: SVCLAIM=J) and events where e.g. only the change in abundance / read depth (SVCLAIM=D) has been determined. Both J and D flags can be combined.

# Positional Queries

## Going beyond single positions...

- Beacon v1 already provided support for "bracket" queries, e.g. for CNV queries - v2 improves documentation
- Use cases w/ focus on structural variants were evaluated by a Beacon "scout" team
- new "range" option
  - anything w/ overlap
  - matched variants can optionally be filtered by type, size, sequence
- query options are not hard defined but derived from parameters
  - Strong wish for defined types?



# bycon

## Progenetix' Beacon Stack

- Python-based software stack
- originally developed for in-house use
- public code base & increasingly sophisticated documentation

→ Happy about adoption & contributions!

progenetix / bycon Public

Code Issues Pull requests Actions Projects Wiki Security Insights Settings

main 1 branch 0 tags Go to file Add file Code About

mbaudis datatable parameter refinements 1d808c1 2 days ago 649 commits

beaconServer some refactoring 16 days ago

config datatable parameter refinements 2 days ago

lib datatable parameter refinements 2 days ago

rsrc/genomes reshuffle & variantType fix 3 months ago

schemas cytoband now in intervals 2 months ago

services datatable parameter refinements 2 days ago

.gitignore lib to package root 11 months ago

LICENSE Create LICENSE 2 years ago

README.md reshuffling and some args are back 7 months ago

\_\_init\_\_.py mostly handover stub for UCSC... 6 months ago

requirements.txt annotatedvariants handover 7 days ago

tests.md ... 12 days ago

README.md

License CC0 1.0

**Bycon - a Python-based environment for the Beacon v2 genomics API**

The `bycon` project - at least at its current stage - is a mix of *Progenetix* (i.e. GA4GH object model derived, *MongoDB* implemented) - data management, and the implementation of middleware & server for the Beacon API.

More information about the current status of the package can be found in the inline documentation which is also presented in an accessible format on the *Progenetix* website.

**More Documentation**

**Services**

The `bycon` environment - together with the *Progenetix* resource - provide a growing number of data services in (cancer) genomics and disease ontologies. `bycon`'s services are tools to enable the APIs.

**Directory Structure**

**beaconServer**

- web applications for data access
- Python modules for Beacon query and response functions in `lib`

**services**

Readme CC0-1.0 license 5 stars 4 watching 4 forks

No releases published Create a new release

No packages published Publish your first package

Contributors 4

mbaudis Michael Baudis

sofiapfund Sofia

qingyao

KyleGao Bo Gao

Languages

Python 99.5% Shell 0.5%

This screenshot shows the GitHub repository page for 'bycon'. The repository is public and has 649 commits. The README.md file is open, providing an overview of the project as a Python-based environment for the Beacon v2 genomics API. It mentions the integration of Progenetix and MongoDB, and provides links to inline documentation and the Progenetix website. The repository has 4 forks and no releases published. Contributors include mbaudis, sofiapfund, qingyao, and KyleGao. The languages used are Python (99.5%) and Shell (0.5%).

## 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](#)

**Rapidly evolving documentation of both the Beacon API itself and its use and technical implementation on [docs.genomebeacons.org](#) [docs.progenetix.org](#)**

- 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](#)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 Beacon+ is implemented in Python.

**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 services (e.g. the [publications](#) service)
- 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 48**Base /biosamples**[/BIOSAMPLES + QUERY](#)

- [/biosamples?filters=cellosaurus:CVCL\\_0004](#)

• this example retrieves all biosamples having an annotation for the Cellosaurus CVCL\_0004 identifier (K562)

[/BIOSAMPLES/{ID}](#)[/BIOSAMPLES/{ID}/TEST\\_MODE](#)[/BIOSAMPLES/{ID}/TEST\\_MODE=true](#)[/BIOSAMPLES/{ID}/TEST\\_MODE=false](#)[/BIOSAMPLES/{ID}/TEST\\_MODE=false?testMode=true](#)[/BIOSAMPLES/{ID}/TEST\\_MODE=false?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

**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 48

## Org.progenetix

Progenetix & Beacon<sup>+</sup>

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

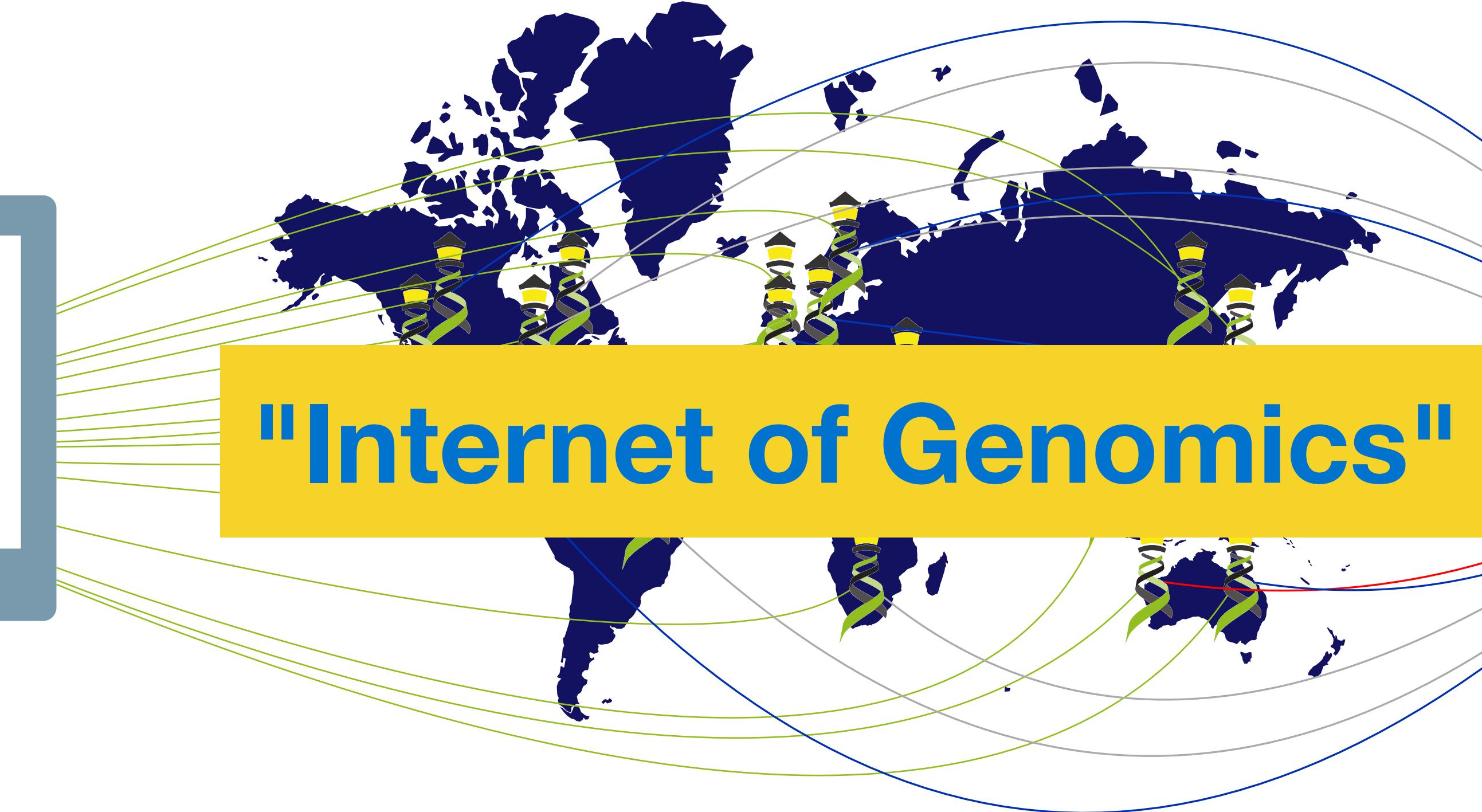
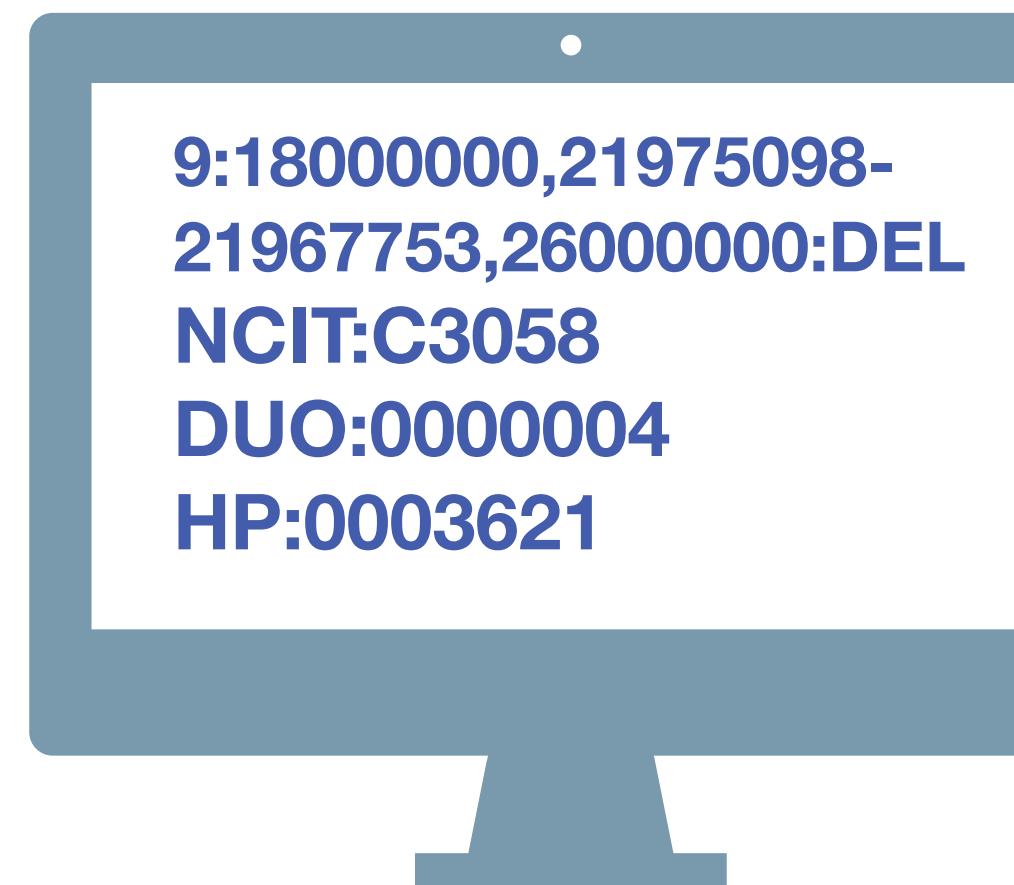
In queries with a complete [beaconRequestBody](#) the type of the delivered data is independent of the path and determined in the [requestedSchemas](#). So far, Beacon+ will compare the first of those to its supported responses and provide the results accordingly; it doesn't matter if the endpoint was [/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 can 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": {
        ...
      }
    }
  }
}
```



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".

# Progenetix & Beacon v2<sup>+</sup>



A cancer genomics reference resource powered by GA4GH standards

## Copy Number Variations (CNV / CNA)

- complex, exciting and still poorly understood area in cancer and rare diseases

## Progenetix

- largest public resource for CNV in cancers (w/ increasing germline data)

## Global Alliance for Genomics and Health (GA4GH)

- policies & technical standards with focus on ***genomic data sharing***

## Federated Data Access

- genomic complexity requires ***data access beyond individual resources***

## Beacon v2

- main GA4GH ***data discovery*** and sharing protocol, developed with support from the European bioinformatics organization **ELIXIR**

# Progenetix & Beacon v2<sup>+</sup>



A cancer genomics reference resource powered by GA4GH standards

## Copy Number Variations (CNV / CNA)

- complex, exciting and still poorly understood area in cancer and rare diseases

Progenetix serves as a testbed for the early implementation of  
GA4GH standards such as  
Beacon extensions, Phenopackets and VRS

## Federated Data Access

- genomic complexity requires *data access beyond individual resources*

## Beacon v2

- main GA4GH data discovery and sharing protocol, developed with support from the European bioinformatics organization **ELIXIR**

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

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Nitin Kumar

### Rahel Paloots

Ziying Yang

Hangjia Zhao

Pierre-Henri Toussaint  
Sofia Pfund

**Beacon Protocol for Genomic Data Sharing**  
Beacons provide discovery services for genomic data using the Beacon API developed by the Global Alliance for Genomics and Health (GA4GH). The Beacon protocol itself is a standard for genomic data discovery. To provide a framework for publishing genomic data, the GA4GH specification defines a set of rules for publishing genomic data.

**Beacon+** About Progenetix

**Samples**  
Request Allele Request

**Example**

This example shows the query for CNV deletion variants overlapping the CDKN2A gene's coding region. The query is limited to hits containing at least a single base, but limited to "focal" hits (here i.e.  $\leq \sim 2\text{Mbp}$  in size). The query is against the Beacon+ collection and can be modified e.g. through changing the assembly, the gene, or the type of variant.

**Variant Spans** Cytoband(s)

**Assembly** hg38 / hg38

**Gene name** CDKN2A

**Start Position** 21000001-21975098

**Classification(s)** C3058: Glioblastoma (2119)

**City** Select...  
21000001 21975098  
21967753 23000000

**Query Beacon**

beacon.progenetix.org/beaconPlus/

## Samples

Request Allele Request

Example

This example shows the query for CNV deletion variants overlapping the CDKN2A gene's coding region. The query is limited to hits containing at least a single base, but limited to "focal" hits (here i.e.  $\leq \sim 2\text{Mbp}$  in size). The query is against the Beacon+ collection and can be modified e.g. through changing the assembly, the gene, or the type of variant.

This query type is for copy number queries ("variantCNVrequest"). It uses a list of genomic positions to capture a set of similar variants.

Variant Spans

Cytoband(s)

Assembly

hg38 / hg38

Gene name

CDKN2A

Start Position

21000001

21975098

Classification(s)

C3058: Glioblastoma (2119)

City

Select...

21000001 21975098

21967753 23000000

Query Beacon

Display a menu

**ELIXIR h-CNV**  
Christophe Béroux  
David Salgado  
many more ...

## Beacon API Leads

Jordi Rambla

Anthony Brooks

## Discovery WS

Michael Baudis (Beacon)

Marc Fiume (Networks)

## {S}[B] and GA4GH

Melanie Courtot

Helen Parkinson

many more ...

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bin Update README.md 7 days ago

docs Update ComplexValue.md 3 days ago

StringTerms description refinement 6 days ago

commonDefinitions.json 3 days ago

structuring intro pages 3 months ago

Revert "Update .gitignore" 3 days ago

repository changes moved to docs page 3 months ago

chatting 3 months ago

Initial commit 4 months ago

Update README.md 7 days ago

Creating the REST page last month

Switch to mermaid2 plugin 3 months ago

**About**  
Unified repository for the GA4GH Beacon v2 API standard

**schema** json-schema

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**Unified repository for Beacon v2 Code & Documentation**

**Description**

This repository is a unified repository representing the different parts of the Beacon API:

- framework
- models
- Beacon v2 Documentation
  - authoritative source already in this repository /docs
  - rendered version through here (alternative address is docs.genomebeacons.org)

github.com/ga4gh-beacon/

**Get Involved! Visit [GA4GH.ORG](http://GA4GH.ORG)**



**Global Alliance**  
for Genomics & Health

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