

BIO392

Bioinformatics of Genome Variations

Structural Genome Variants in Cancer: Research & Resources

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Computational Oncogenomics



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Theodor Boveri (1914)

(based on observations in sea urchin eggs)

- **Cell-cycle checkpoints** (“Hemmungseinrichtung”)
- **Tumour-suppressor genes** (“Teilungshemmende Chromosomen”), which may be overcome by external signals, and can be eliminated during tumour progression
- **Oncogenes** (“Teilungsfoerdernde Chromosomen”) that become amplified (“im permanenten Übergewicht”)
- **Progression** (benign to malignant), w/ sequential changes of chromosomes
- Clonal origin & Genetic mosaicism
- Cancer **predisposition** through inheritance of “chromosomes” that are less able to suppress malignancy
- Inheritance of the same 'weak chromosome' from both parents leads to **homozygosity** and, consequently, to high-penetrance cancer syndromes - (e.g. xeroderma pigmentosum)
- Wounding and inflammation in tumour promotion; loss of cell adhesion in metastasis; sensitivity of malignant cells to radiation therapy (based on Hertwig *et al.*)

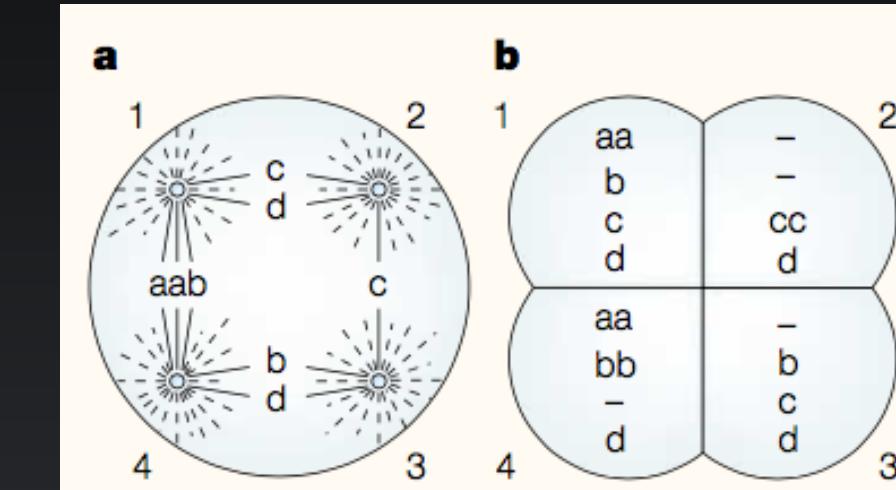
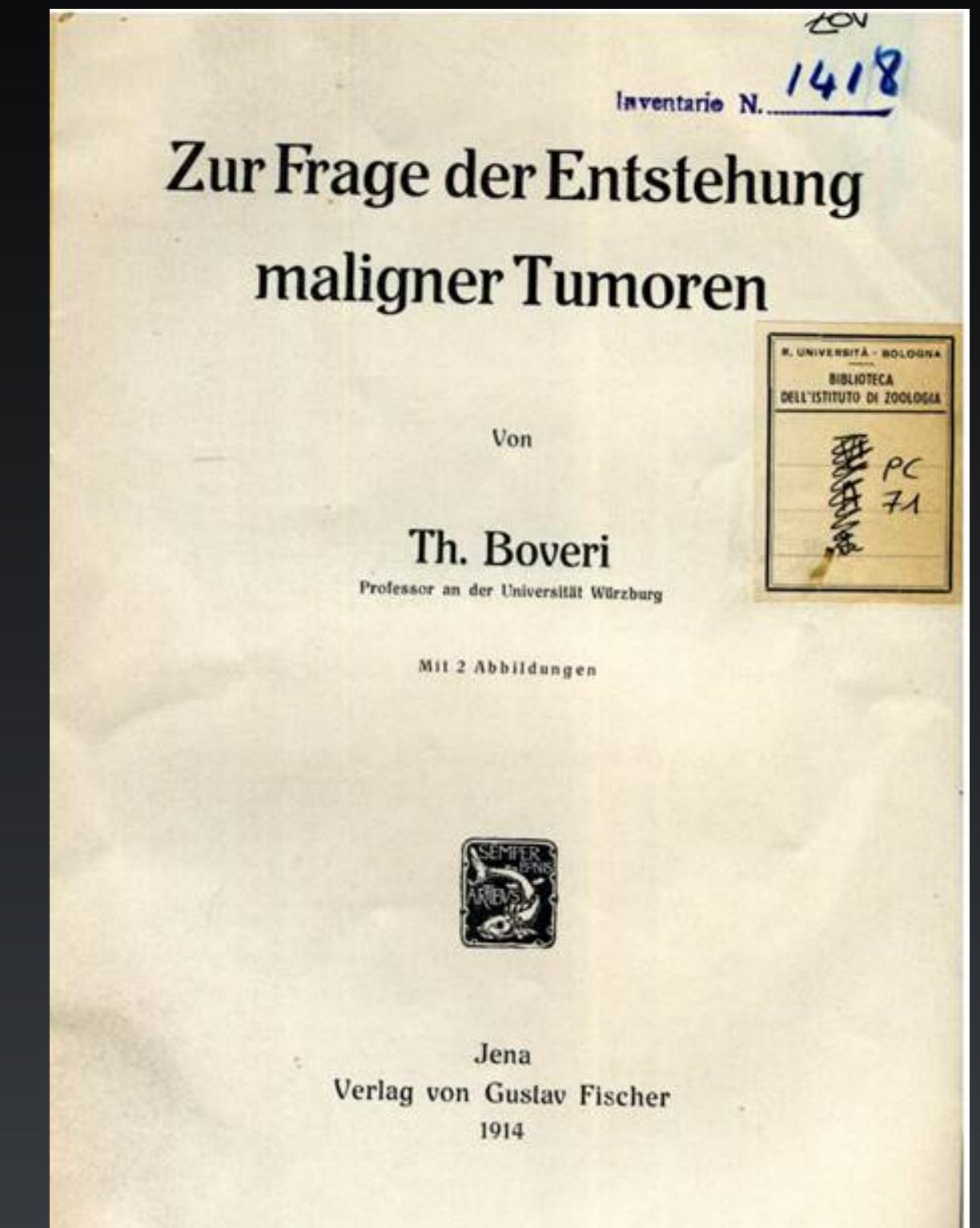
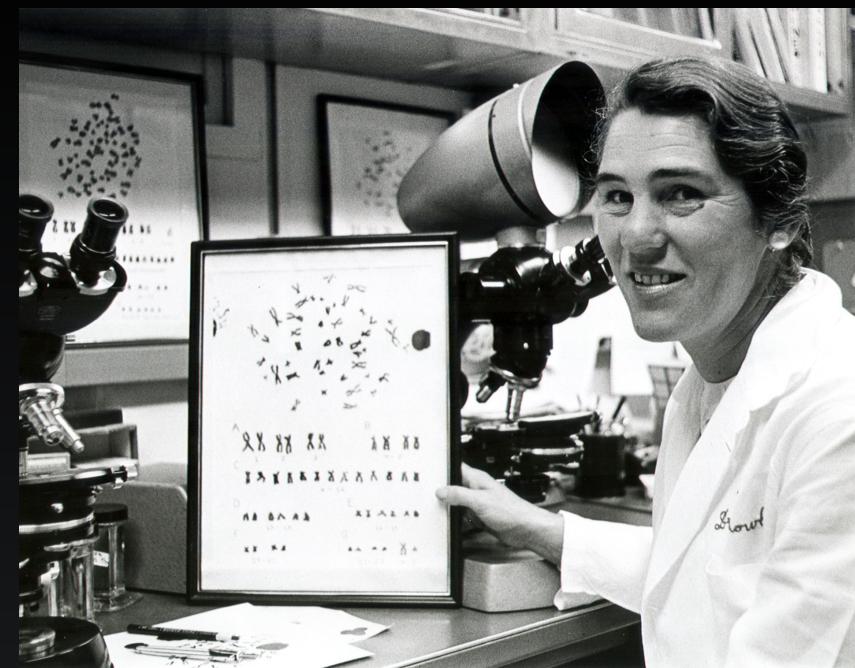


Figure 2 | **Multiple cell poles cause unequal segregation of chromosomes.** **a** | Boveri showed that fertilization of sea-urchin eggs by two sperm results in multiple cell poles. Individual chromosomes then attach to different combinations of poles — for example, one copy of chromosome c is attached to poles 1 and 2, and one copy is attached to poles 2 and 3. **b** | Chromosomes are segregated to the four poles at cell division, leaving some cells with too many copies of the chromosomes and some with too few — for example, cell 2 has two copies of chromosome c and cell 4 has none.



Allan Balmain
Cancer genetics: from Boveri and Mendel to microarrays.
NatRev Cancer (2001); 1: 77-82

Anna Di Lonardo , Sergio Nasi , Simonetta Pulciani
Cancer: We Should Not Forget The Past
Journal of Cancer (2015), Vol. 6: 29-39
(for book cover & summary)



Janet Rowley (1972/73)

Chromosomal translocations in cancer

- Recurrent chromosomal translocations in leukemias and lymphomas
- "Philadelphia chromosome" in CML (Nowell & Hungerford, 1960) represents a reciprocal translocation between chromosomes 9 and 22
- 1972: t(8;21) ALL manuscript rejected by NEJM
- 1973: t(9;22) manuscript rejected by *Nature* "with some reasonable comments and some truly wrong"
- Clinical implications: **Tyrosine Kinase inhibitors** as standard first-line therapy in CML
 - first trials in 1998 (STI-571; Imatinib/Gleevec)
 - cf. Druker BJ, Lydon NB (2000). Lessons learned from the development of an Abl tyrosine kinase inhibitor... *J Clin Invest* 2000;105:3-7

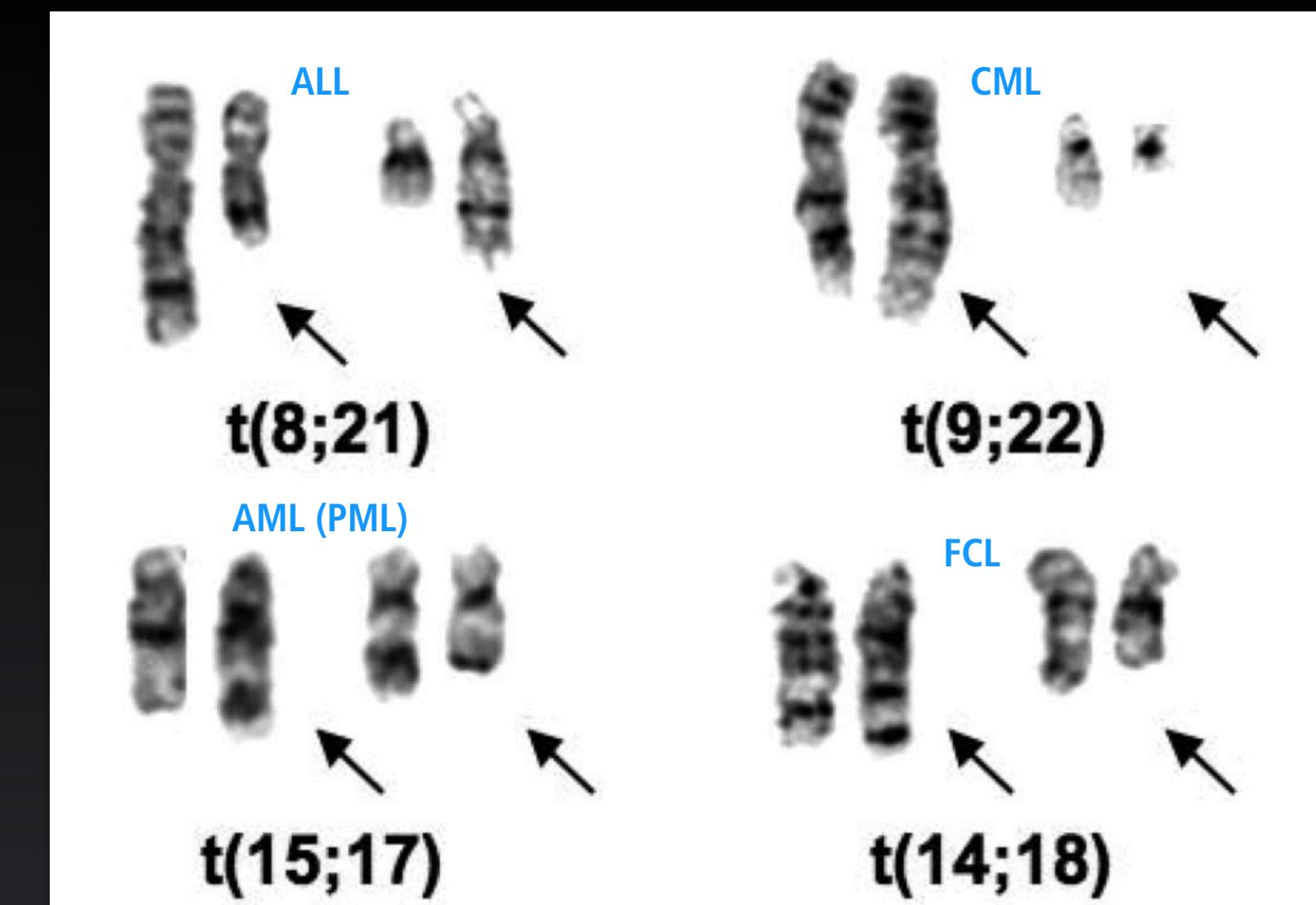
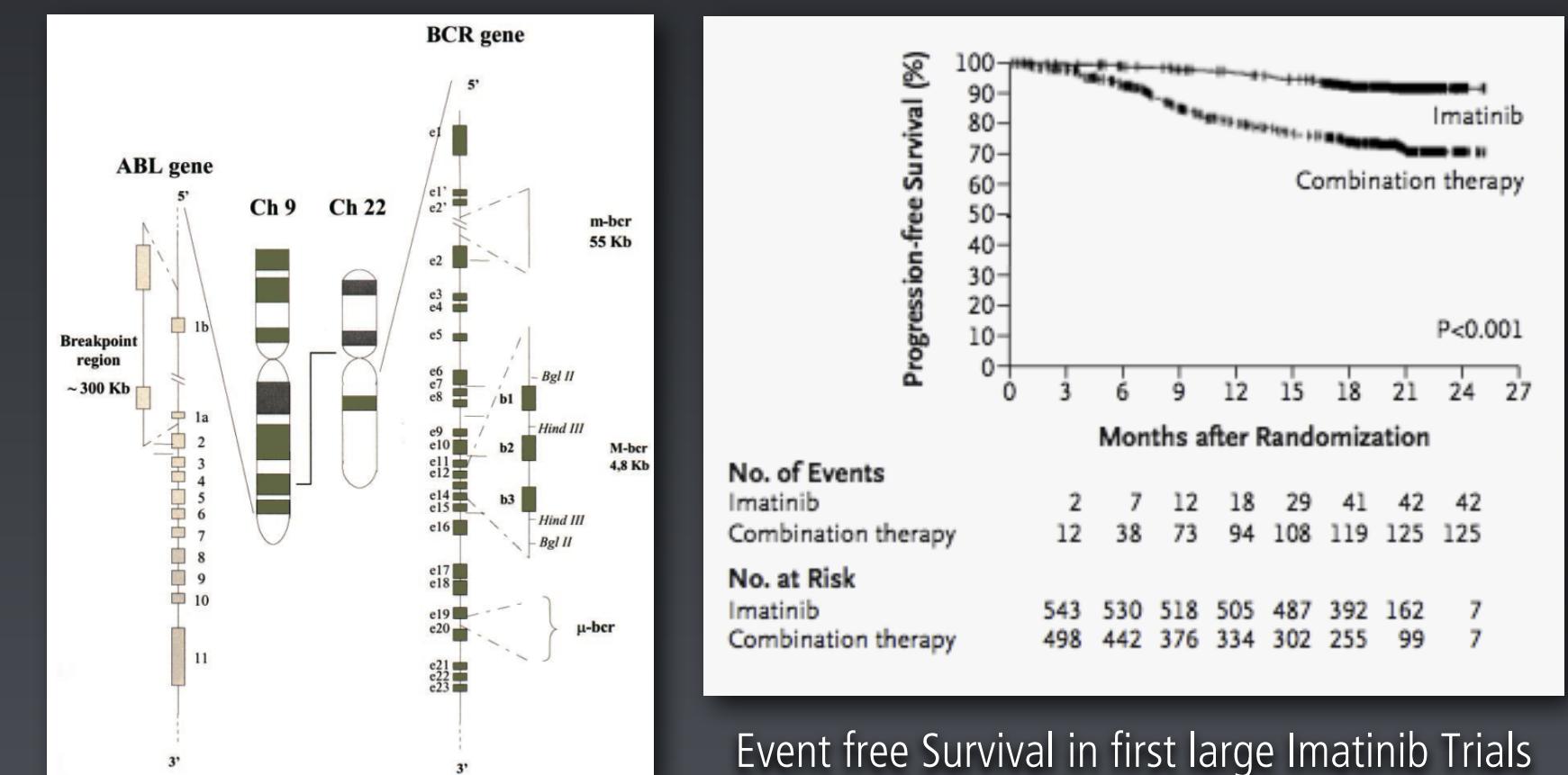


Figure 1. Partial karyotypes of common translocations discovered by Rowley.
The translocations appear in the order in which they were discovered.

Janet D Rowley. Chromosomal translocations: revisited yet again *Blood* (2008), 112(6)



Event free Survival in first large Imatinib Trials

Pane et al. BCR/ABL genes
Oncogene (2002), 21 (56)

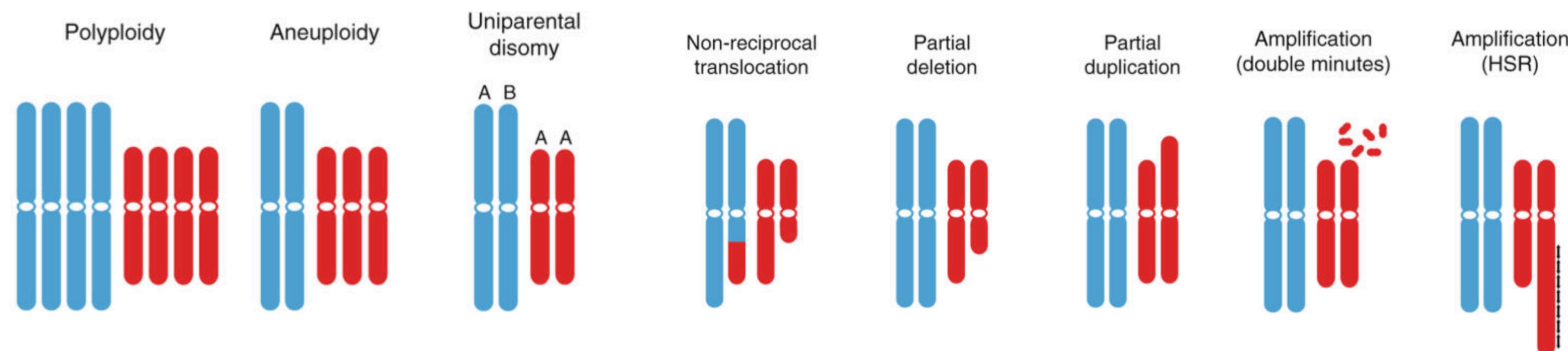
O'Brien et al. Imatinib compared with interferon and low-dose cytarabine... *NEJM* (2003) vol. 348 (11)

Introduction

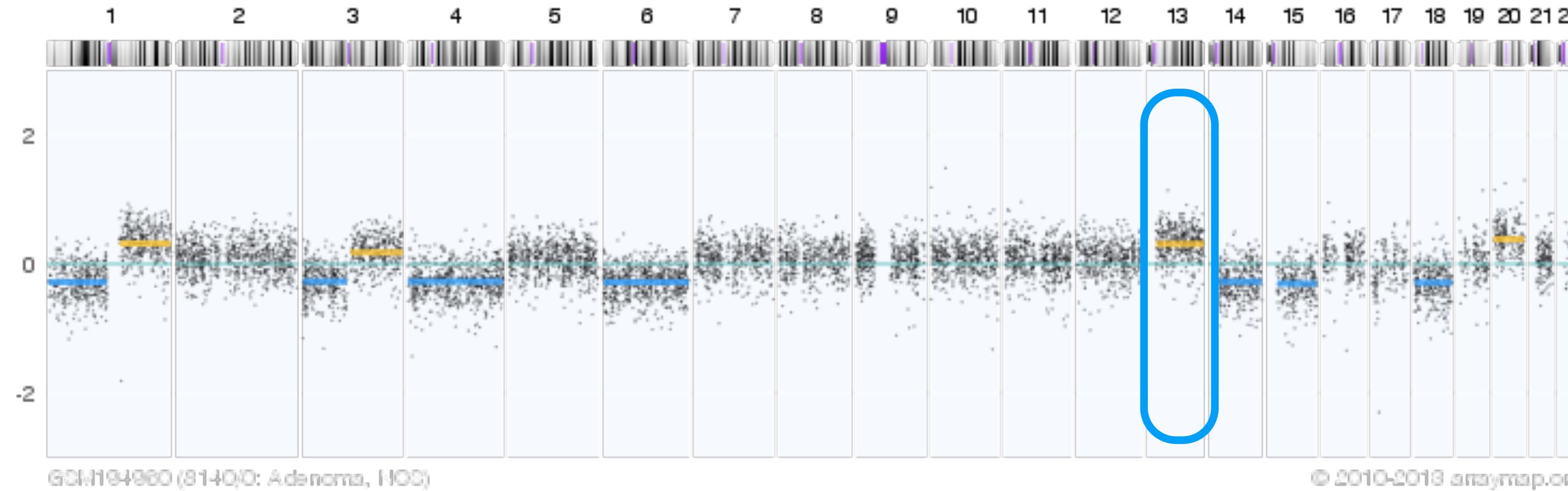
Types of genomic alterations in Cancer

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

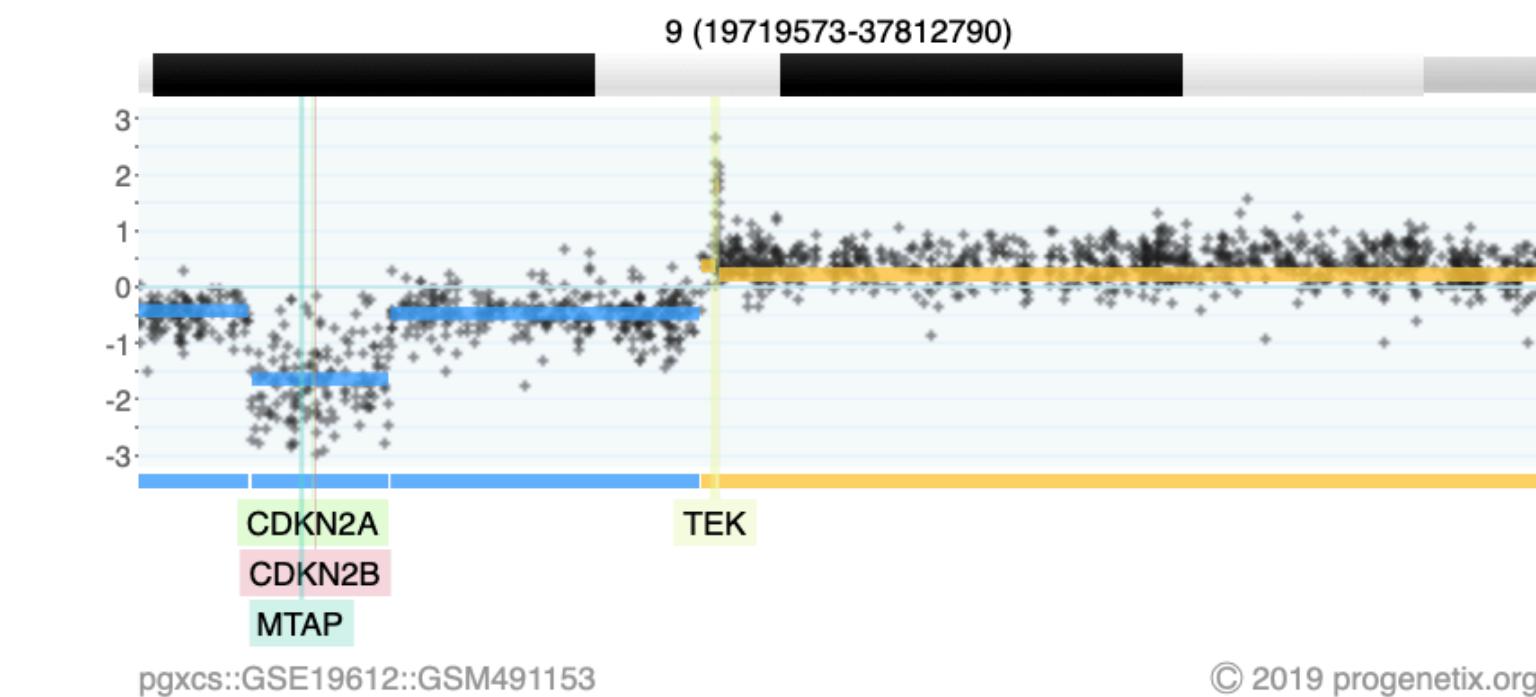
Imbalanced Chromosomal Changes: CNV



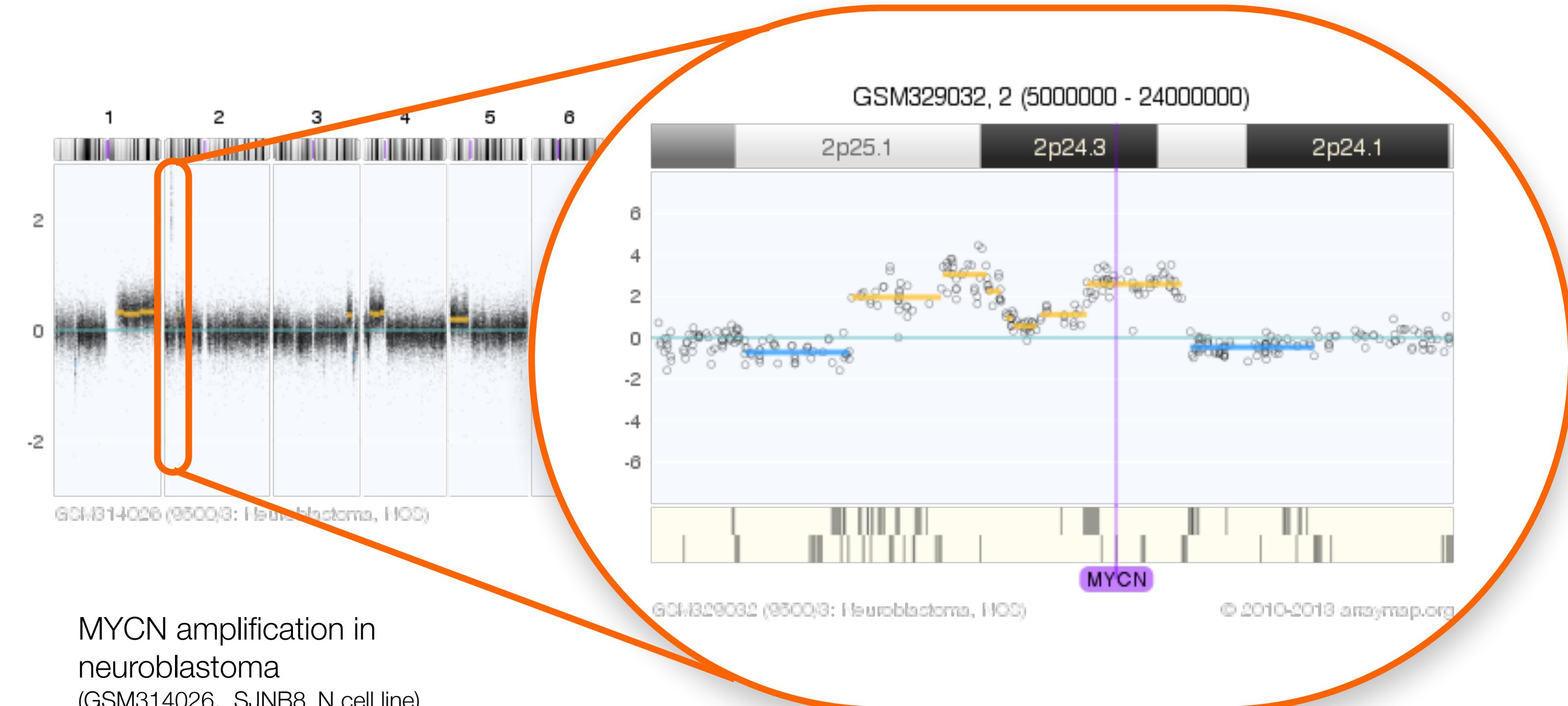
Somatic Copy Number Variations



Gain of chromosome arm 13q in colorectal carcinoma



2-event, homozygous deletion in a Glioblastoma



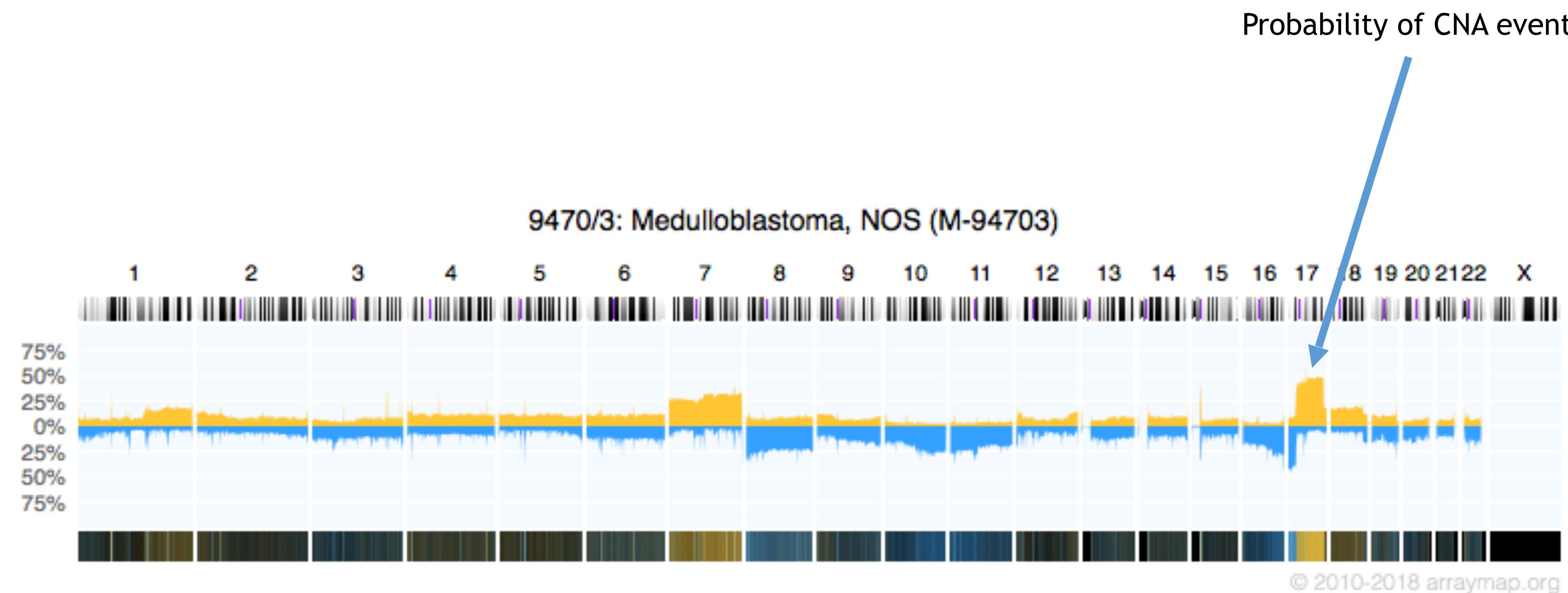
MYCN amplification in neuroblastoma
(GSM314026, SJNB8_N cell line)

low level/high level copy number alterations (CNAs)

arrayMap

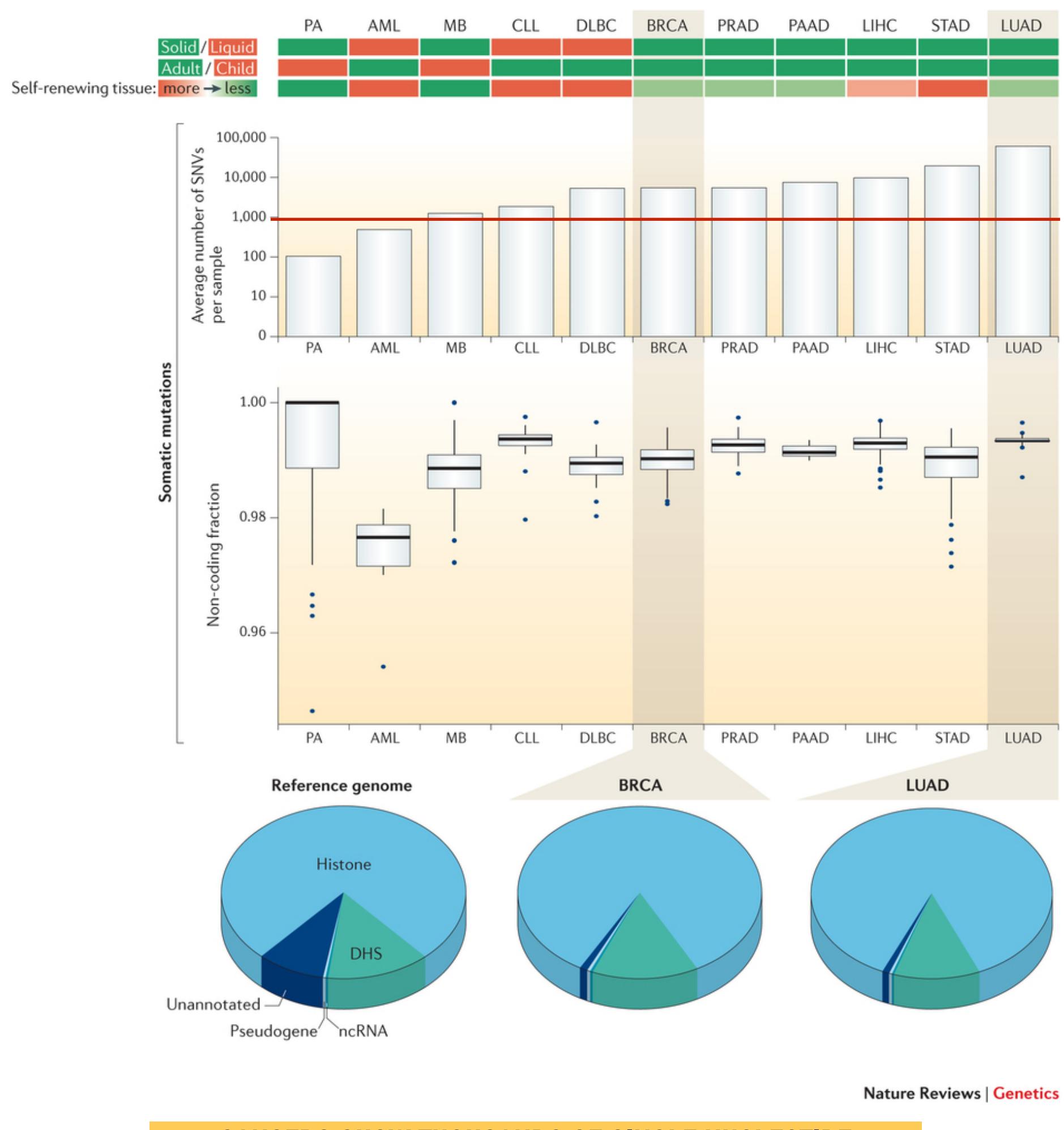


Copy Number Aberrations

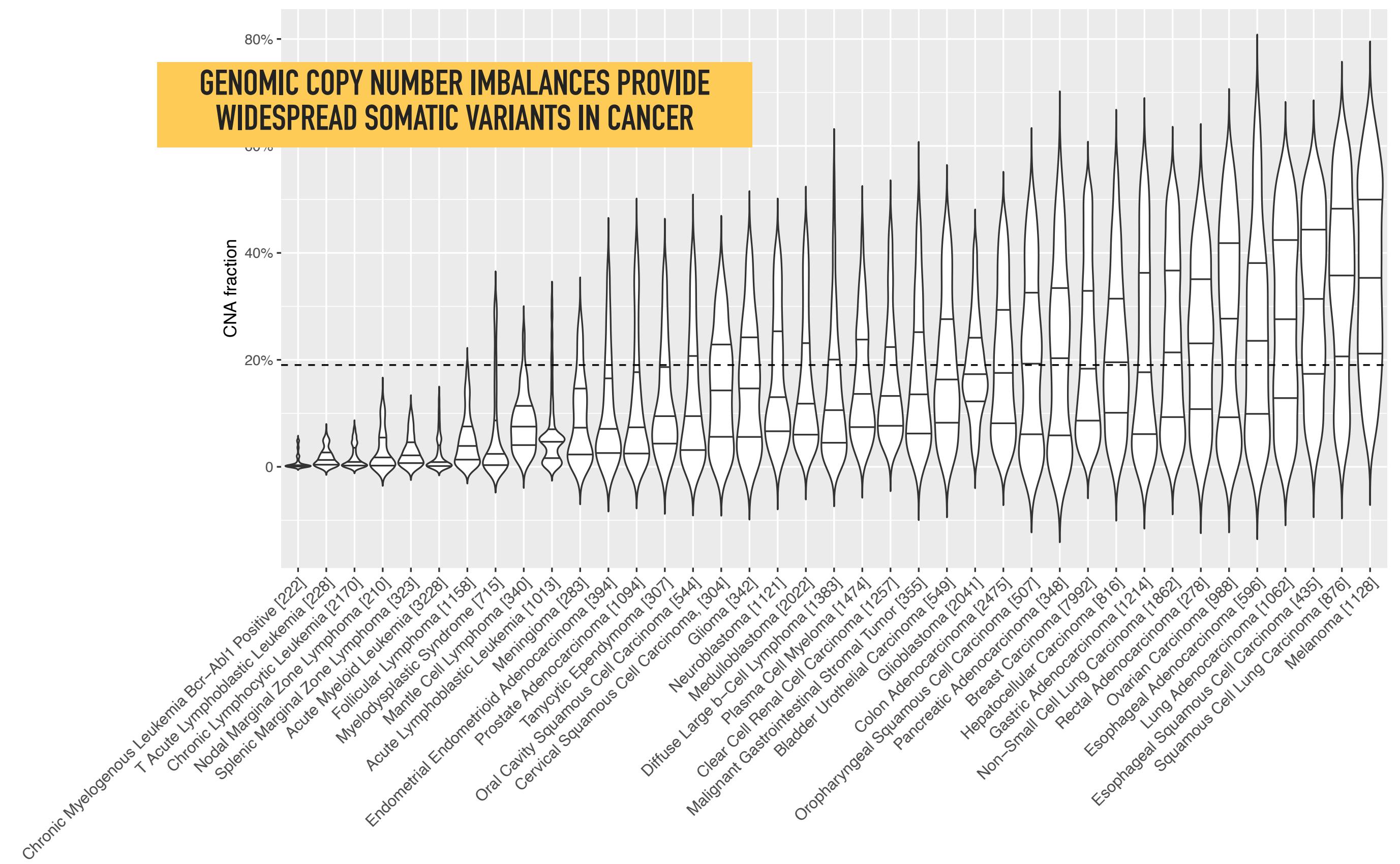


Probability of CNA with the frequency of gains (yellow) and losses (blue) across the chromosomes for 2'021 samples of Medulloblastoma, NOS, extracted from arrayMap database.

Quantifying Somatic Mutations In Cancer

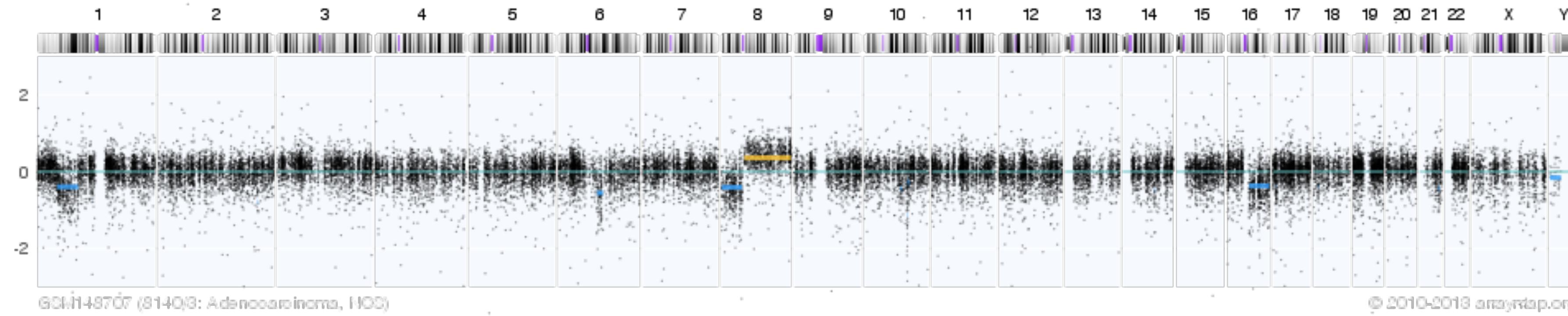
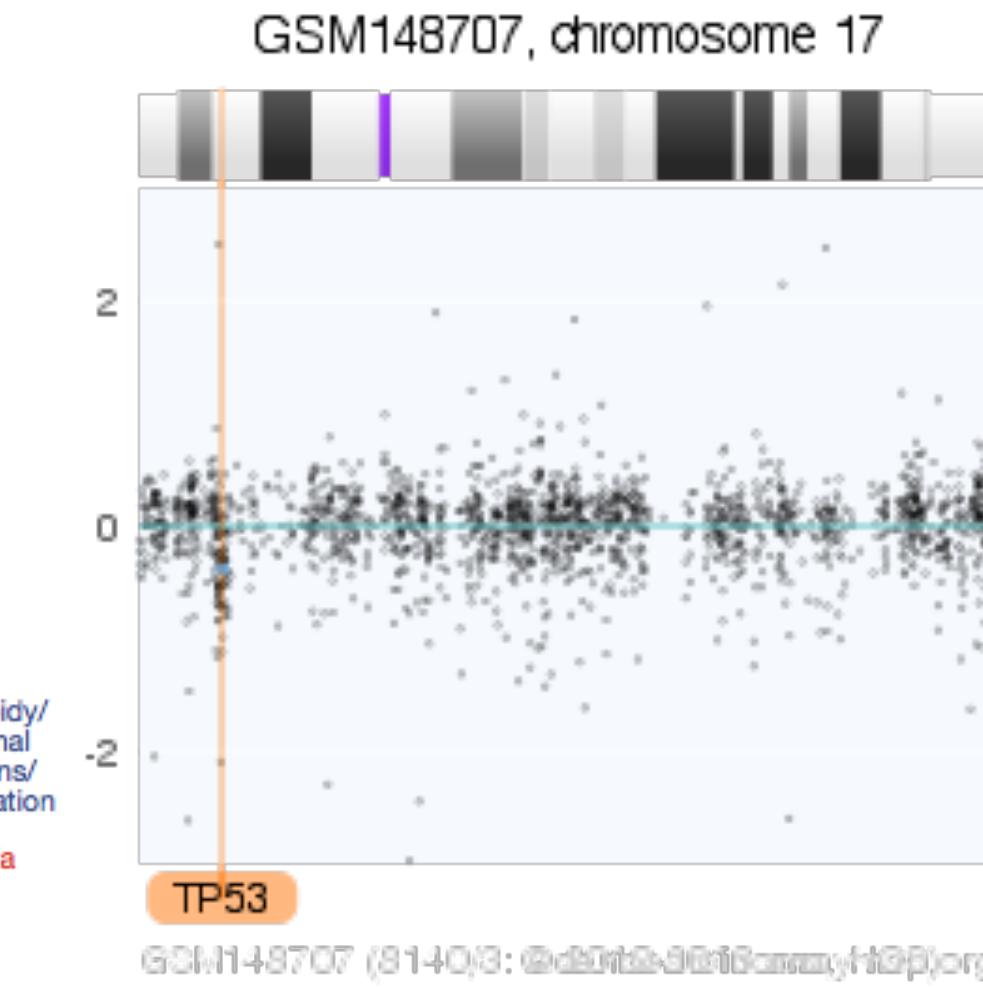
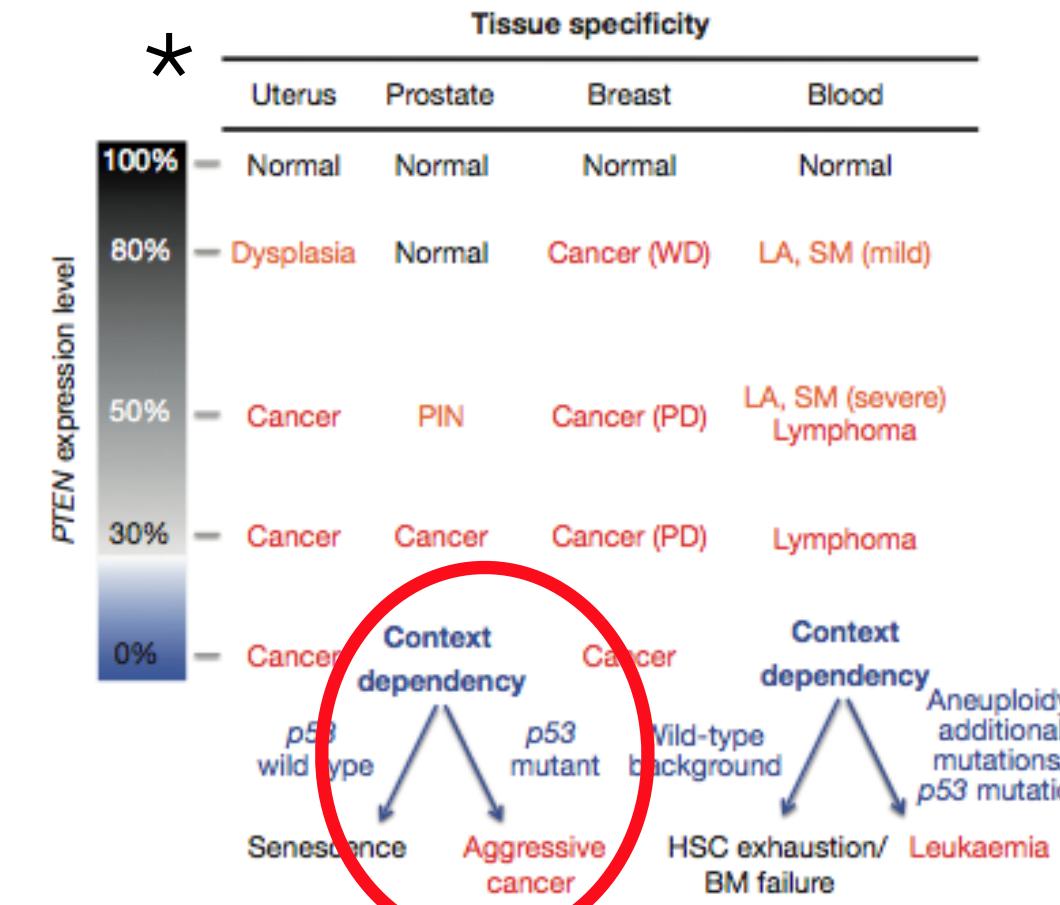
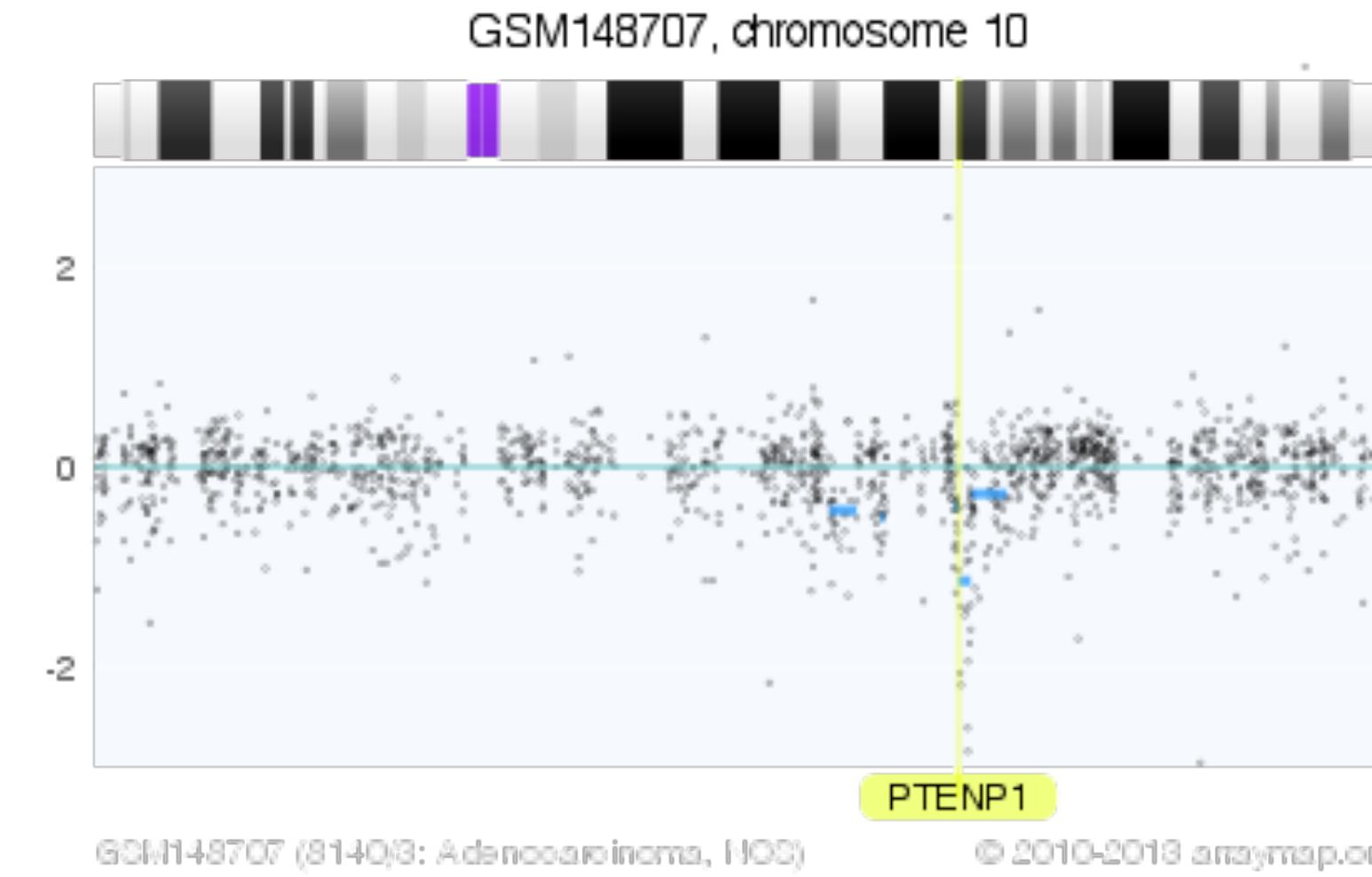


Pan-Cancer Analysis of Whole Genomes (PCAWG) data show widespread mutations in non-coding regions of cancer genomes (Khurana et al., Nat. Rev. Genet. (2016))



On average ~19% of a cancer genome are in an imbalanced state (more/less than 2 alleles); Original data based on 43654 cancer genomes from progenetix.org

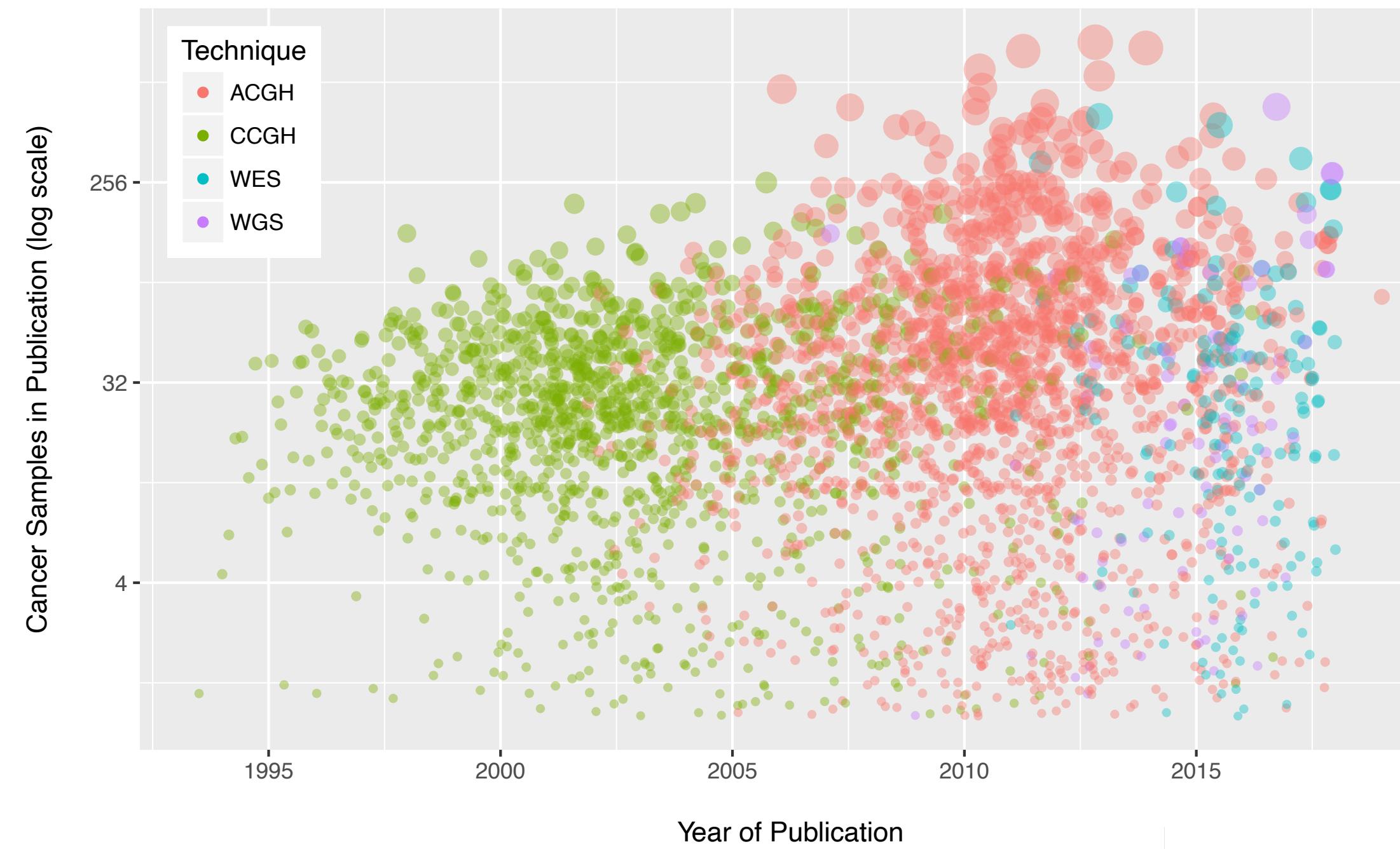
Gene dosage phenomena beyond simple on/off effects



Combined heterozygous deletions involving *PTEN* and *TP53* loci in a case of prostate adenocarcinoma
(GSM148707, PMID 17875689, Lapointe et al., CancRes 2007)

* A. H. Berger, A. G. Knudson, and P. P. Pandolfi, "A continuum model for tumour suppression," *Nature*, vol. 476, no. 7359, pp. 163–169, Aug. 2011.

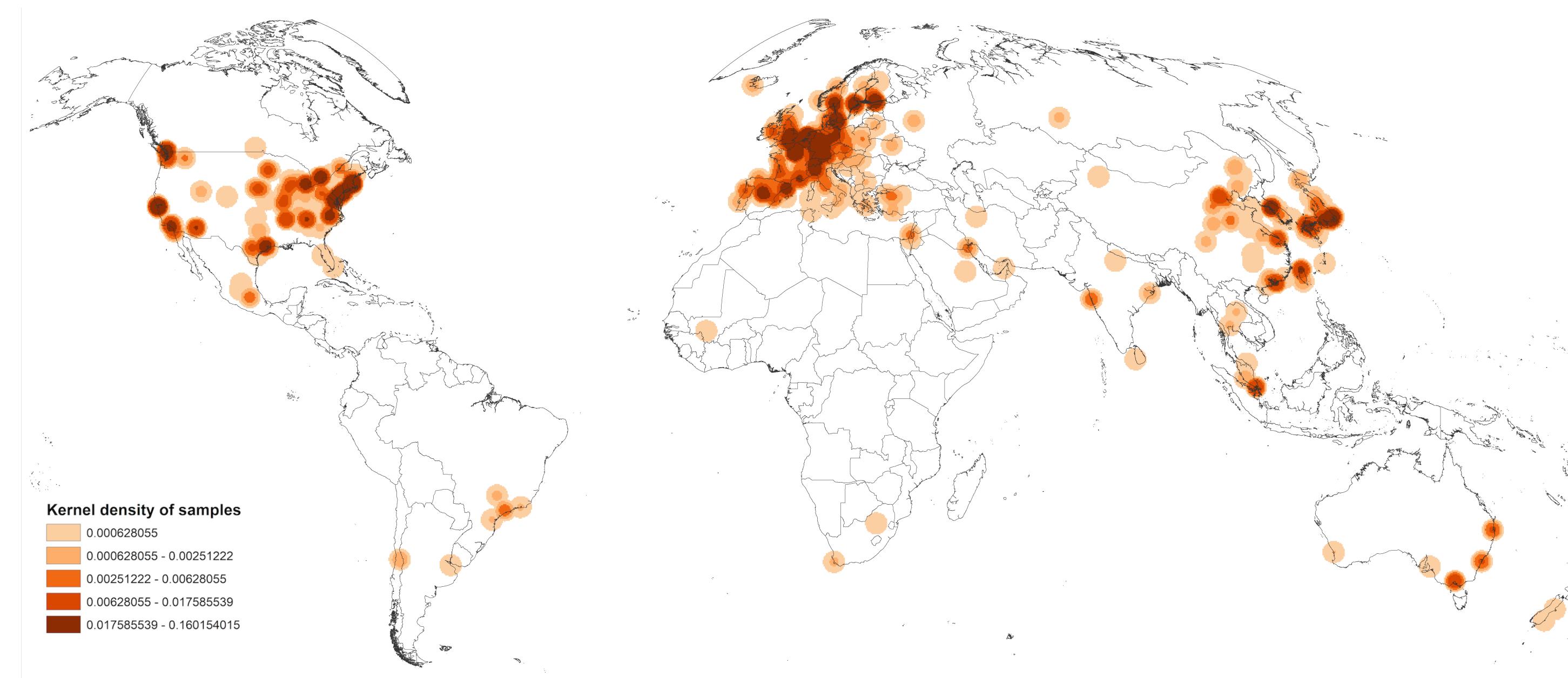
Map of the geographic distribution (by first author affiliation) of the 104'543 genomic array, 36'766 chromosomal CGH and 15'409 whole genome/exome based cancer genome datasets.
The numbers are derived from the 3'240 publications registered in the Progenetix database.



Publication Landscape of Cancer CNV Profiling

Publication statistics for cancer genome screening studies. The graphic shows our assessment of publications reporting whole-genome screening of cancer samples, using molecular detection methods (chromosomal CGH, genomic array technologies, whole exome and genome sequencing).

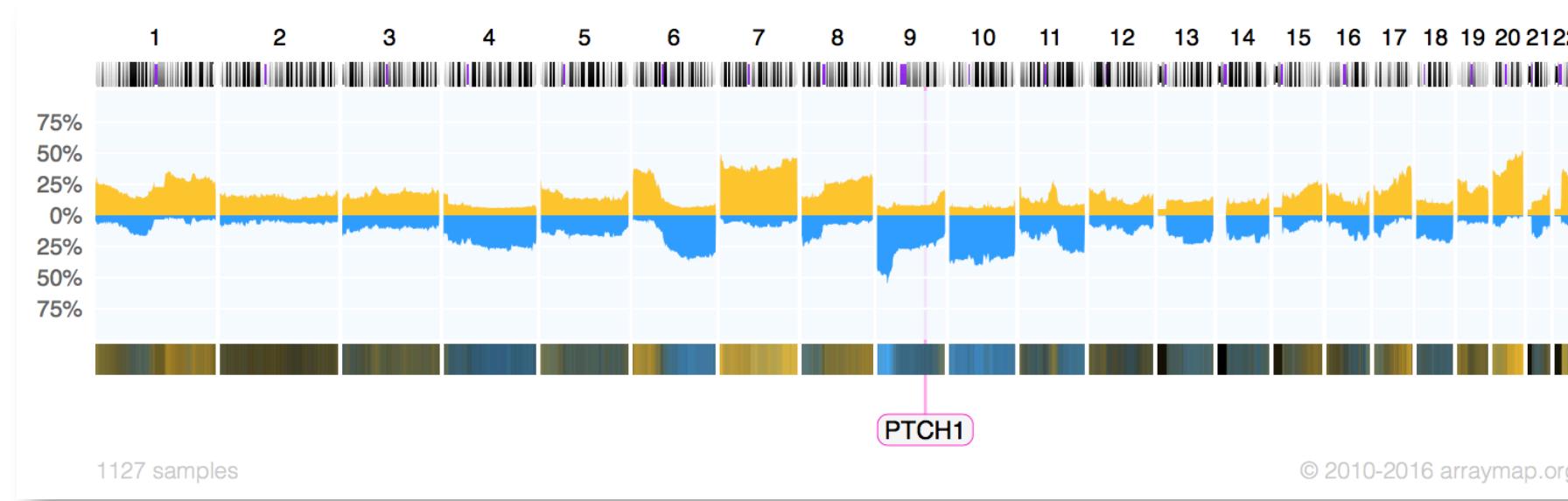
For the years 1993-2018, we found 3'229 publications reporting 174'530 individual samples in single series from 1 to more than 1000 samples. Y-axis and size of the dots correspond to the sample number; the color codes indicate the technology used.



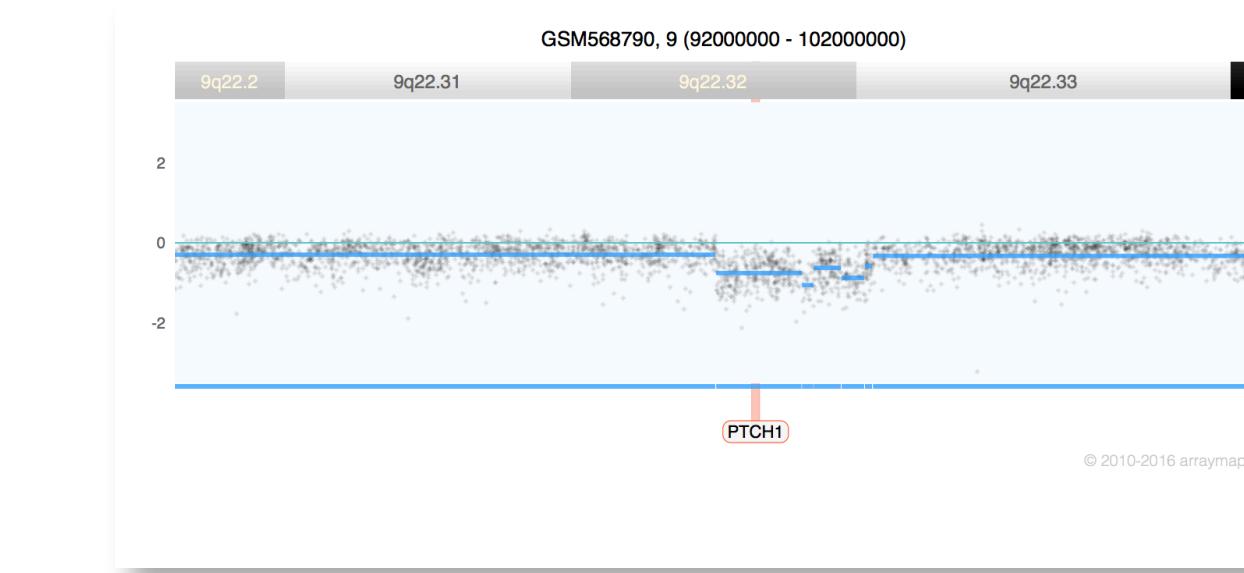
Rare CNV Events & Hidden Therapeutic Options?

Example: PTCH1 deletions in malignant melanomas

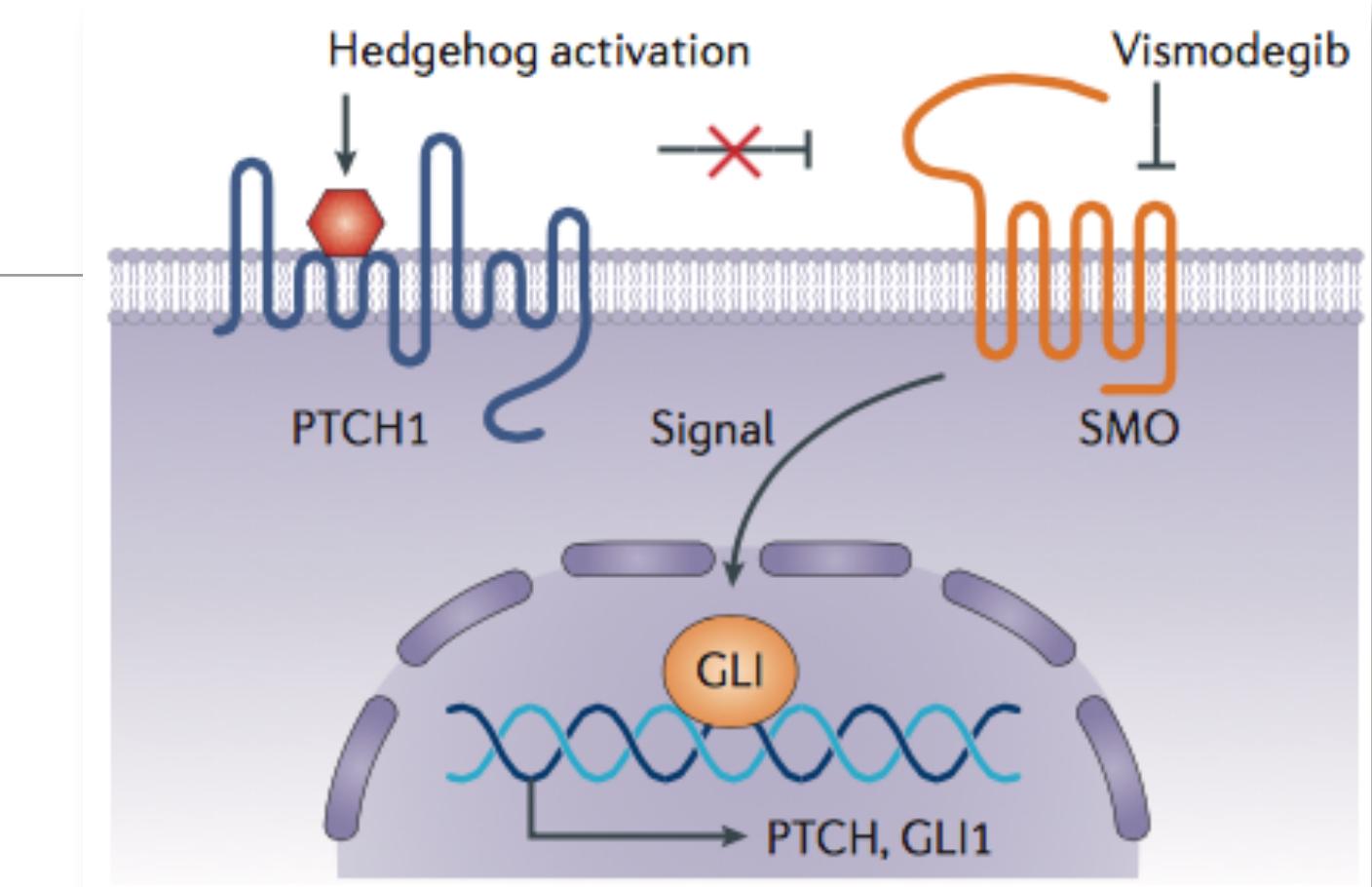
PTCH1 is a actionable tumor suppressor gene, which has been demonstrated in e.g. basalomas and medulloblastomas analysis of 1127 samples from 26 different publications could identify **focal** deletions in 4 samples a current project addresses the focal involvement of all mapped genes, in >50'000 cancer genome profiles



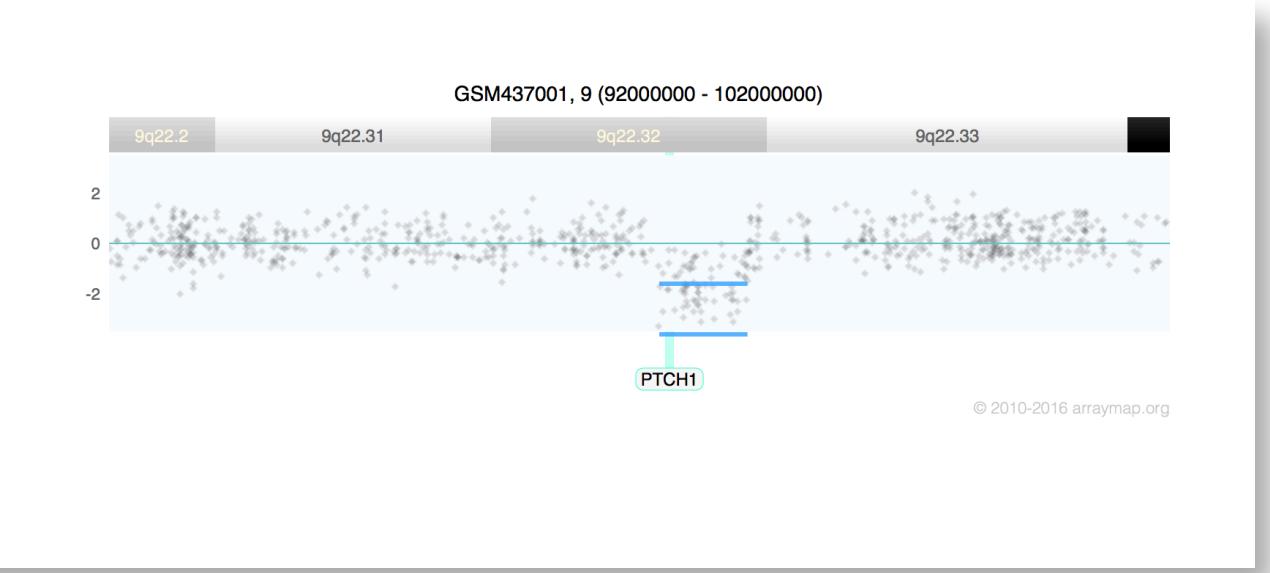
Summary of somatic copy number aberrations from the analysis of 1127 genome profiles of malignant melanomas, collected in our arraymap.org cancer genome resource. While PTCH1 does not represent a deletion hotspot, the genomic locus is part of larger deletions in ~25% of melanoma samples.



Examples of focal / homozygous PTCH1 deletions detected in the analysis of 1127 genomic array datasets. Focal somatic imbalance events are considered an indicator for oncogenic involvement of the affected target genes.



In its normal function, PTCH1 is a tumor suppressor gene in the sonic hedgehog pathway and inhibits SMO driven transcriptional activation. A loss of PTCH1 function (mutation, deletion) can be mitigated through drugs antagonistic to SMO activation.



Progenetix - Reference Resource for Oncogenomic Profiling Data

- launched in 2001 as progenetix.net with 999 samples (September 2001)
- curated CNV data from chromosomal CGH studies
- now containing > 137'902 single sample CNV tracks from ~1600 publications
- **aCGH, cCGH, WES, WGS**
- additionally tracking and annotating of publications reporting compatible original data (more than 3305 articles as of 2020)



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.

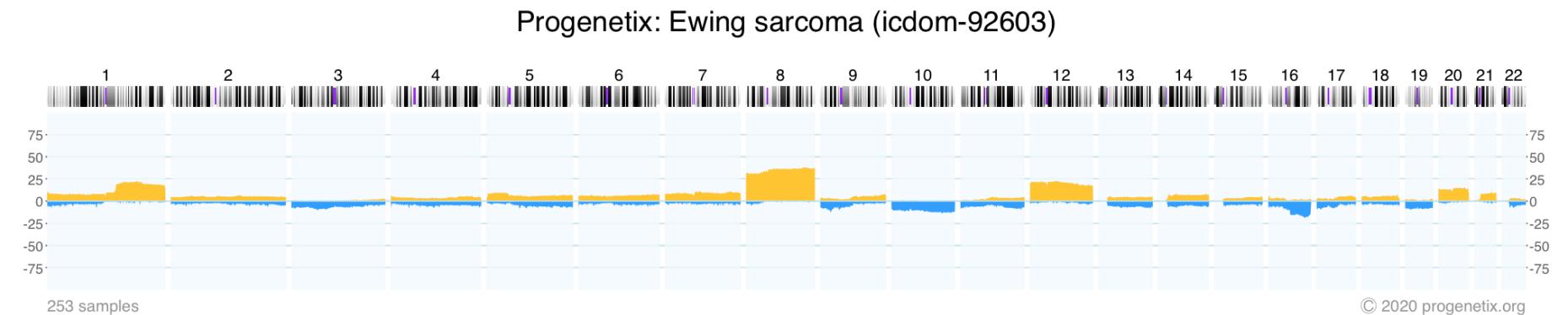
Data Content and Provenance

The resource currently contains genome profiles of 113322 individual experiments. The genomic profiling data was derived from array and chromosomal Comparative Genomic Hybridization (CGH) experiments as well as Whole Genome or Whole Exome Sequencing (WGS, WES) studies. Genomic profiles are either processed from various raw data formats or are extracted from published experimental results.

Besides genomic profiling data, Progenetix contains sample specific biological, technical and provenance information which so far has been curated from 1600 articles.

Original diagnoses are mapped to (hierarchical) classification systems and represents 420 and 542 different cancer types, according to the International classification of Diseases in Oncology (ICD-O) and NCIt "neoplasm" classification, respectively.

Progenetix: Ewing sarcoma (icdom-92603)



A genomic profile plot showing copy number variations (CNVs) across chromosomes 1 through 22. The y-axis represents the log2 ratio, ranging from -75 to 75. The x-axis shows 253 samples. The plot displays several distinct regions of gain (yellow) and loss (blue) across the chromosomes, with a notable peak in gain on chromosome 8.

For exploration of the resource it is suggested to either start with [Cancer Types](#) or by [searching](#) for CNVs genes of interest.

Additionally to genome profiles and associated metadata, the website present information about publications (currently 3962 articles) referring to cancer genome profiling experiments.

Access, Maintenance and Contributions

The content of the progenetix resource is freely accessible for research and commercial purposes, with attribution.

The database & software are developed by the [group of Michael Baudis](#) at the [University of Zurich](#) and the Swiss Institute of Bioinformatics [SIB](#).

Many previous members and external collaborators have contributed to data content and resource features. Participation (features, data, comments) by volunteers are welcome.

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No responsibility is taken for the correctness of the data presented nor the results achieved with the Progenetix tools.

arrayMap

Accessing Probe-Level Genomic Array Data in Cancer



Search Samples

Search Publications

Progenetix



Citation & Licensing

User Guide

People

Beacon+



162.158.150.56

visualizing cancer genome array data @ arraymap.org

arrayMap is a curated reference database and bioinformatics resource targeting copy number profiling data in human cancer. The arrayMap database provides an entry point for meta-analysis and systems level data integration of high-resolution oncogenomic CNA data.

The current data reflects:

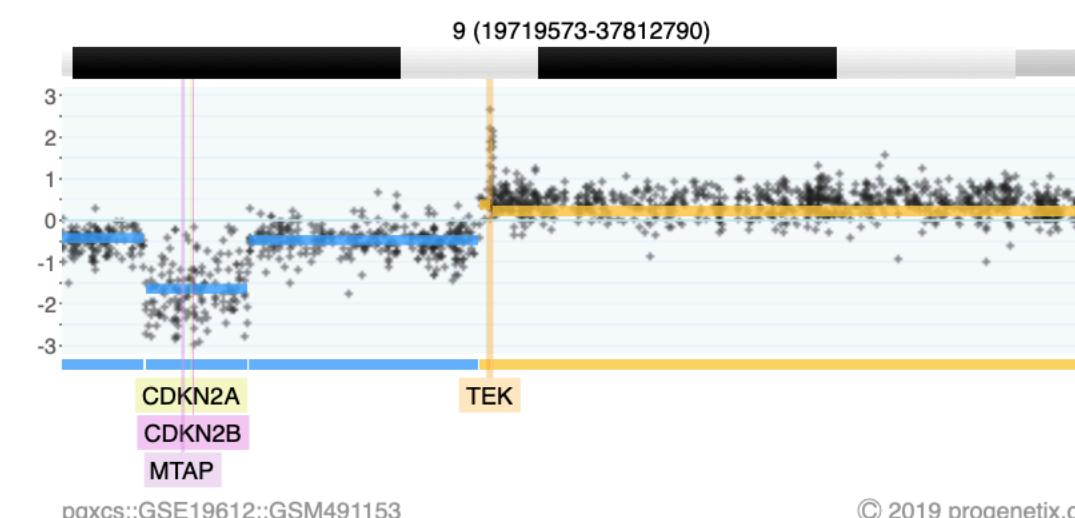
72724 genomic array profiles

898 experimental series

257 array platforms

341 ICD-O cancer entities

795 publications (Pubmed entries)



Genomic copy number imbalances on chromosome 9 in a case of Glioblastoma ([GSM491153](#)), indicating, among others, a homozygous deletion involving CDKN2A/B.

For the majority of the samples, probe level visualization as well as customized data representation facilitate gene level and genome wide data review. Results from multi-case selections can be connected to downstream data analysis and visualization tools, as we provide through our Progenetix project.

arrayMap is developed by the group "Theoretical Cytogenetics and Oncogenomics" at the Institute of Molecular Life Sciences of the University of Zurich.

RELATED PUBLICATIONS



Cai H, Gupta S, Rath P, Ai N, Baudis M. arrayMap 2014: an updated cancer genome resource. *Nucleic Acids Res.* 2015 Jan;43(Database issue). Epub 2014 Nov 26. [\[PubMed\]](#)

Cai, H., Kumar, N., & Baudis, M. 2012. arrayMap: A Reference Resource for Genomic Copy Number Imbalances in Human Malignancies. *PLoS One* 7(5), e36944. [\[PubMed\]](#)

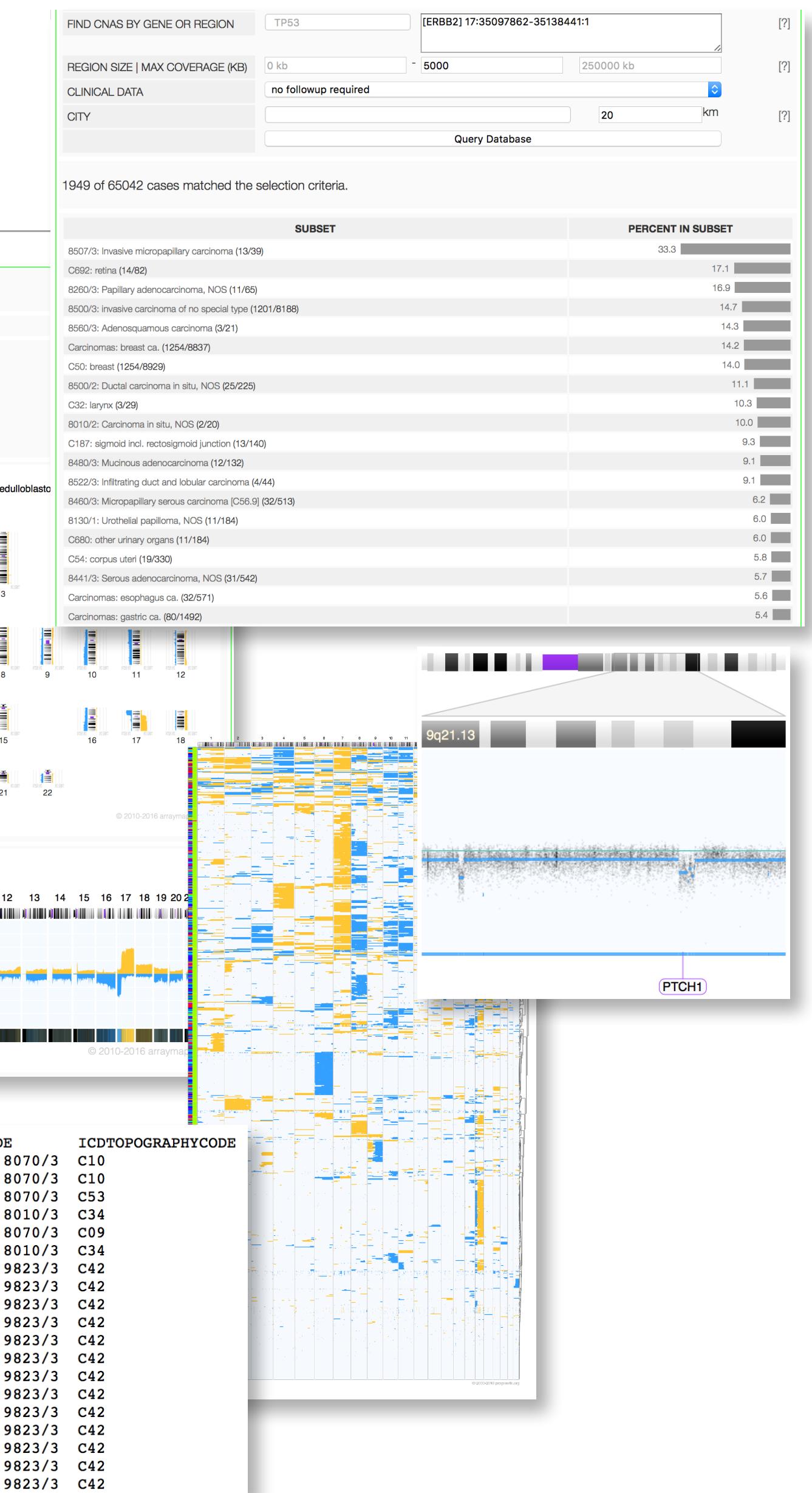
Baudis, M. 2007. Genomic imbalances in 5918 malignant epithelial tumors: An explorative meta-analysis of chromosomal CGH data. *BMC Cancer* 7:226. [\[PubMed\]](#)

Baudis, M. 2006. Online database and bioinformatics toolbox to support data mining in cancer cytogenetics. *Biotechniques* 40, no. 3: 296-272. [\[PubMed\]](#)

Baudis, M, and ML Cleary. 2001. Progenetix.net: an online repository for molecular cytogenetic aberration data. *Bioinformatics* 12, no. 17: 1228-1229. [\[PubMed\]](#)

Feel free to use the data and tools for academic research projects and other applications. If more support and/or custom analysis is needed, please contact Michael Baudis regarding a collaborative project.

© 2000 - 2019 Michael Baudis, refreshed 2019-06-12T21:00:19Z in 6.00s on server 130.60.240.68. No responsibility is taken for the correctness of the data presented nor the results achieved with the Progenetix tools.



arrayMap

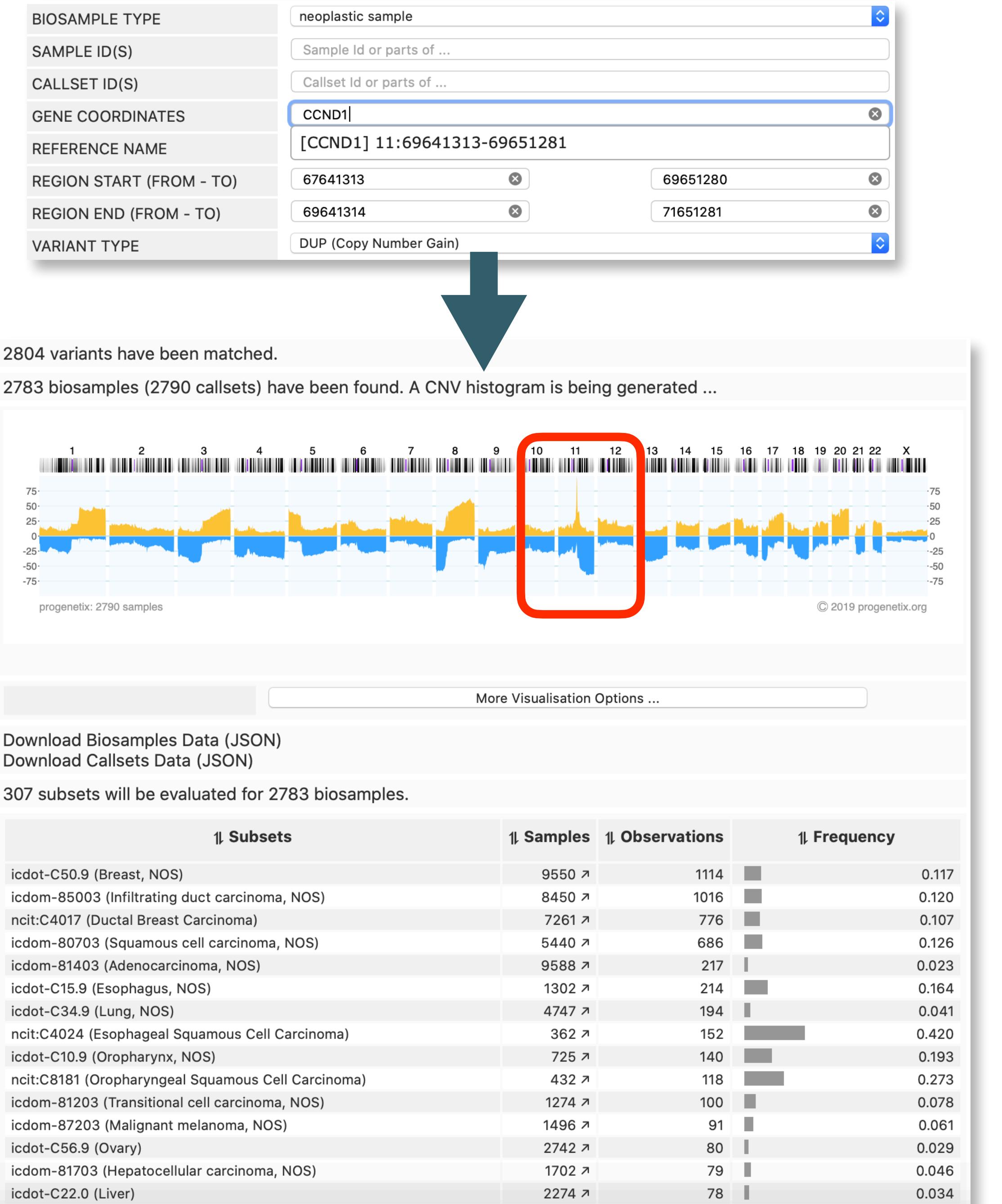


Progenetix & arrayMap: available data

Data Source	GEO	ArrayExpress	cBioPortal	TCGA	Total
No. Studies	898	44	38	33	1'932
No. samples	63'568	3887	19'712	22'142	137'870
Tumor	52'090	3887	19'712	11'090	114'730
Normal	11'478	0	0	11'052	23'133
Classifications					
ICD-O (Topography)	100	57	88	157	216
ICD-O (Morphology)	246	88	265	140	498
NCIt	346	143	422	182	789
Collections					
Individuals	63'568	3'887	19'712	10'995	126'755
Biosamples	63'568	3'887	19'712	22'142	137'870
Callsets	63'568	3'887	19'712	22'376	138'136
Variants	5'514'126	1'083'415	1'778'096	2'654'065	11'507'273

Progenetix - Reference Resource for Oncogenomic Profiling Data

- Progenetix is based on the single-sample CNV tracks of cancer samples from 402/469 (ICD-O/NCIt) diagnostic categories
- typical applications include
 - ▶ reference CNV patterns in given diagnoses (e.g. "does my analysis match the diagnosis/prediction")
 - ▶ target gene entity mapping (e.g. "in which tumour type is this gene frequently gained/lost?")



Progenetix API

- the Progenetix API provides access to a growing number of database features
 - biosample data listings
 - code translations (ICD-O <-> NCIIt)
 - publication data

Progenetix :: Info

Structural Cancer Genomics Resource Documentation and Example Pages

New

About.

Documentation

Publications

Related Sites

arrayMap
Baudisgroup @ UZH
Beacon+
SchemaBlocks {S}[B]
ELIXIR Beacon
Baudisgroup Interna

Github Projects

baudisgroup
progenetix
ELIXIR Beacon

Tags

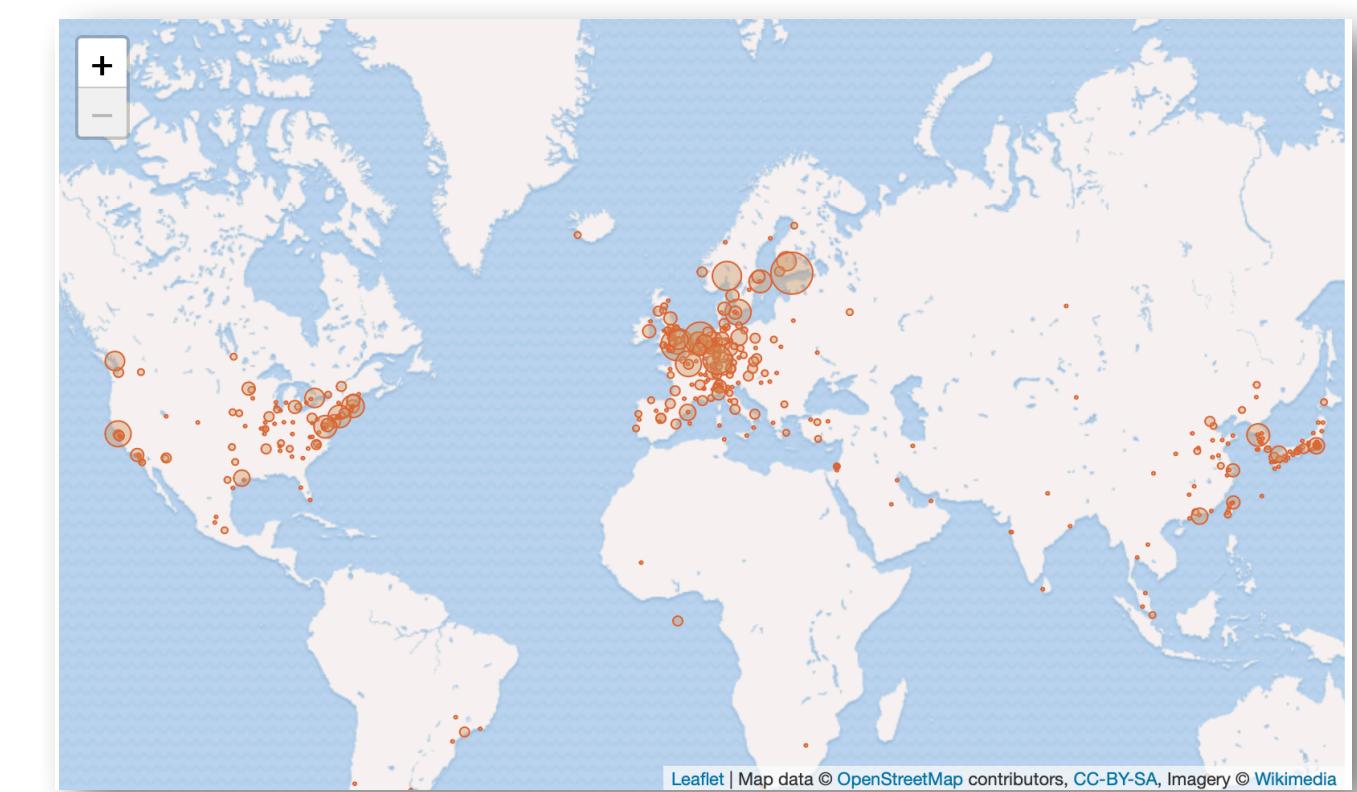
API **Beacon** **BeaconPlus** **JavaScript**
Perl **article** **code** **documentation**

API::api.cgi Code Documentation

API syntax

The query elements are ordered in the URL

1. **api**
 - fixed, required; i.e. the request has to start with `/api/`
 2. database (parameter **apidb**)
 - mostly “progenetix”
 3. collection (**apiscope**)
 4. method (**apimethod**)
 - see examples/documentation below
 5. filters (**filters**)
 - essentially query parameters in a simplified format
 - comma-concatenated
 - can also be omitted for query string
 6. output parameter (**apioutput**)
 - optional
 7. query string
 - optional
 - can be used for any parameter; e.g. a query can be formatted completely as standard query string:
 - `progenetix.org/api/?apidb=progenetix&apiscope=publications ...`



Standardized Data

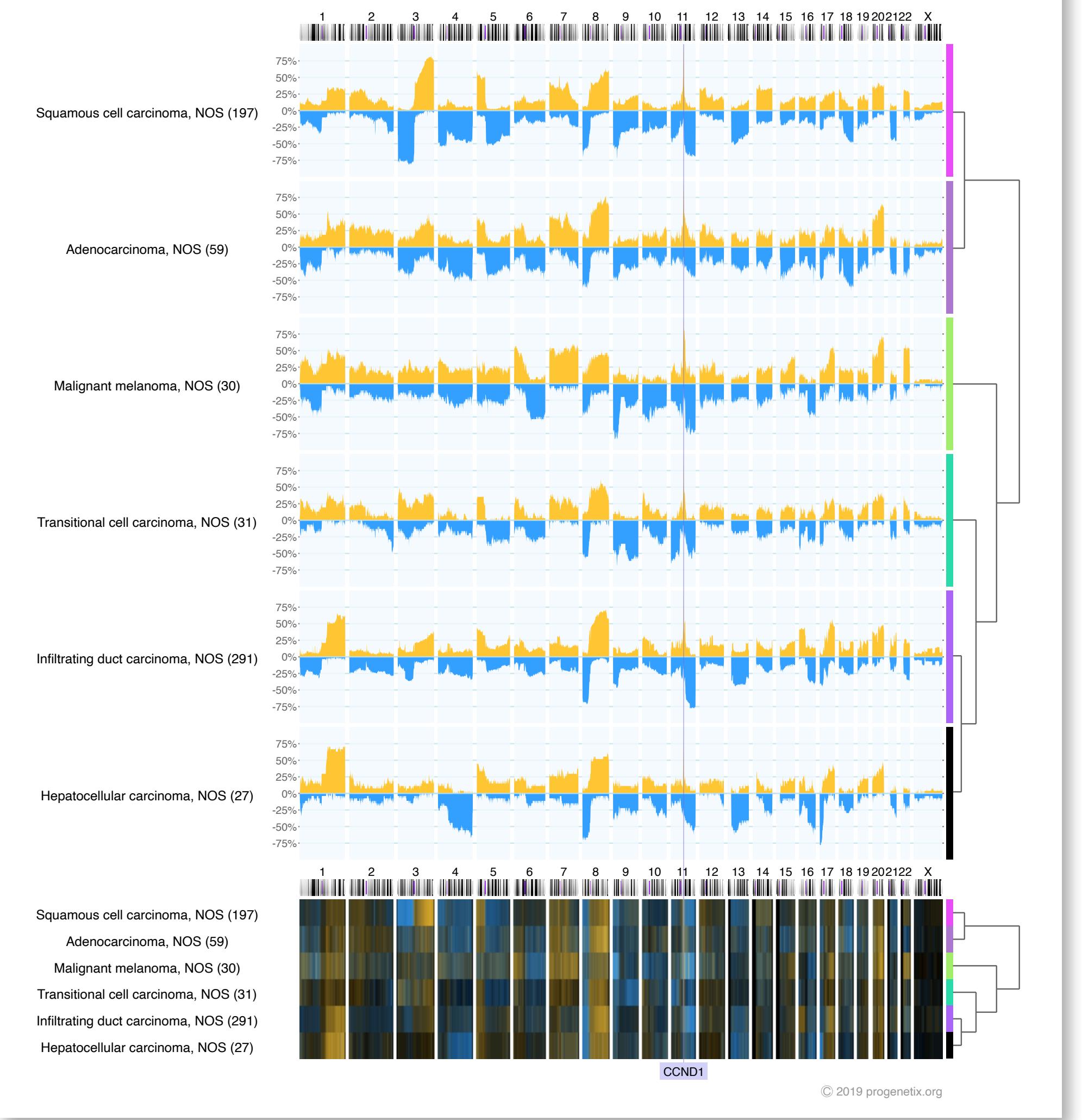
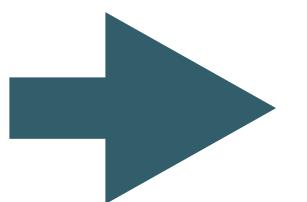
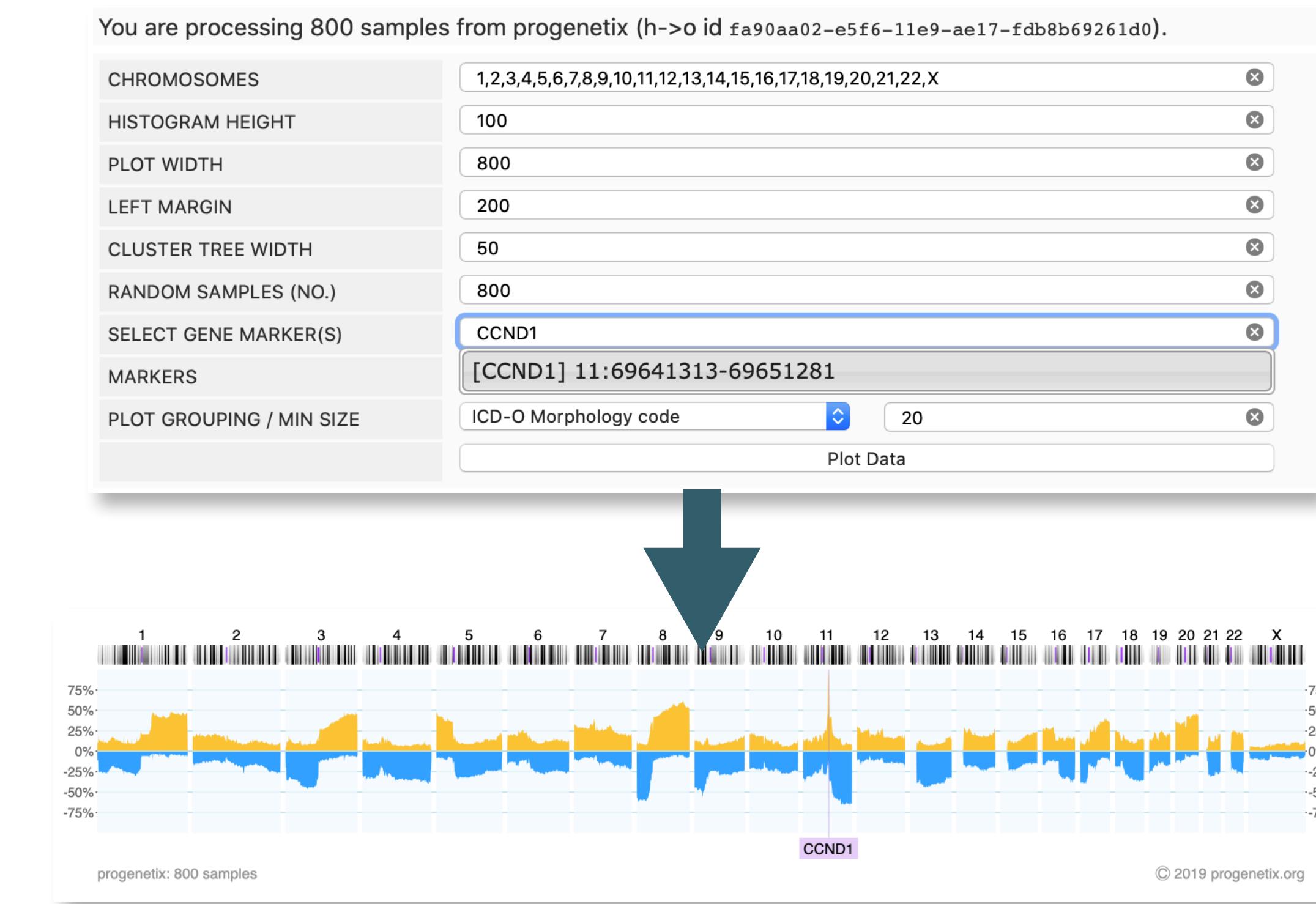
Data re-use depends on standardized, machine-readable metadata

- Multiple international initiatives (ELIXIR, GA4GH, MONARCH...) and resource providers (EBI, NCBI ...) work on the generation and implementation of data annotation standards
- emerging / established principles are the use of hierarchical coding systems where individual codes are represented as CURIEs
- other formats for non-categorical annotations based on international standards, e.g.
 - ISO (ISO 8601 time & period, ISO 3166 country codes ...)
 - IETF (GeoJSON ...)
 - W3C (CURIE ...)
- these standards become pervasive throughout GA4GH's ecosystem (e.g. Phenopackets ...)

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    },  
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            ]  
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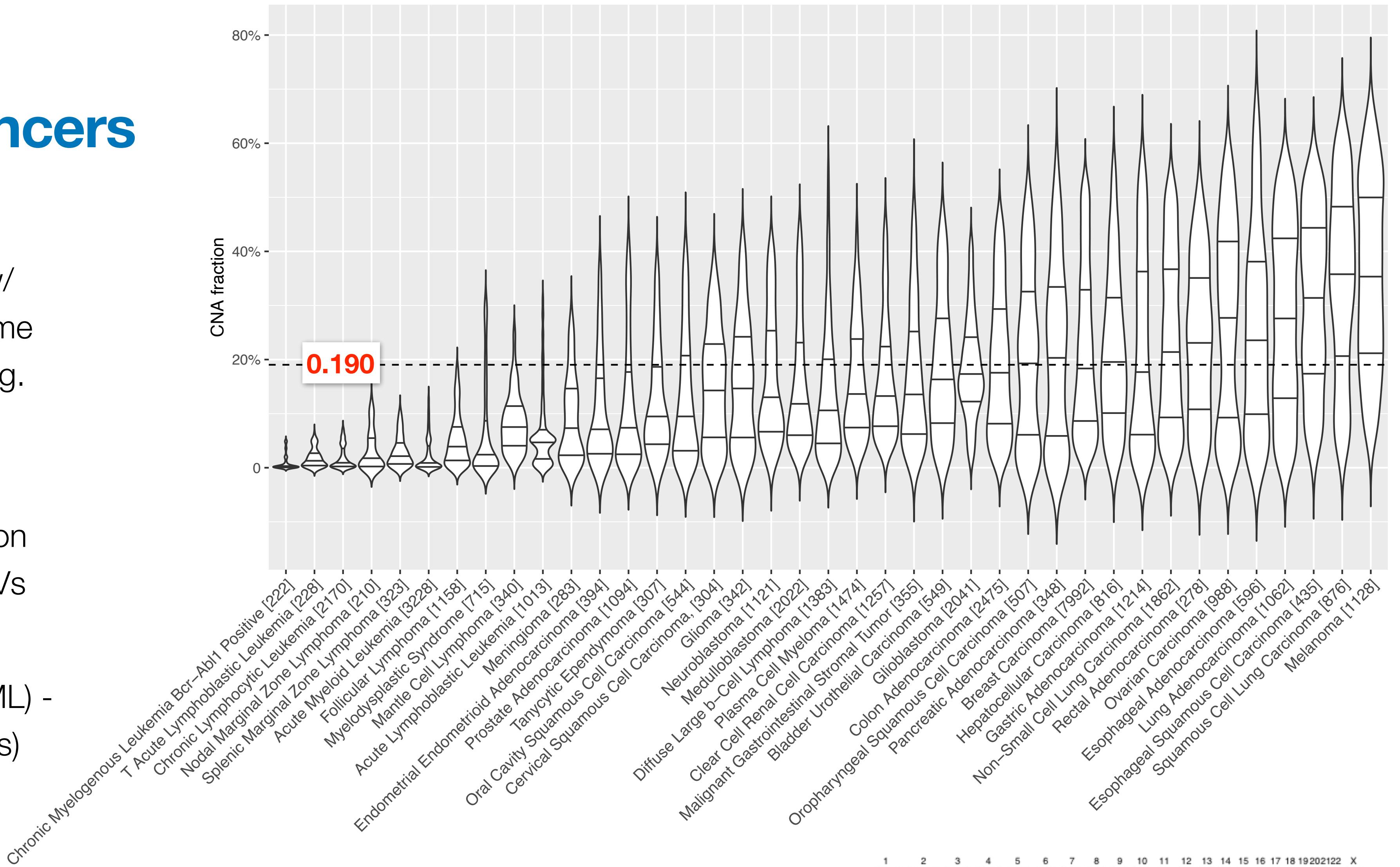
Progenetix - Reference Resource for Oncogenomic Profiling Data

- Group histogram and heatmap representation of CNV profiles by external labels (disease codes, publications ...)

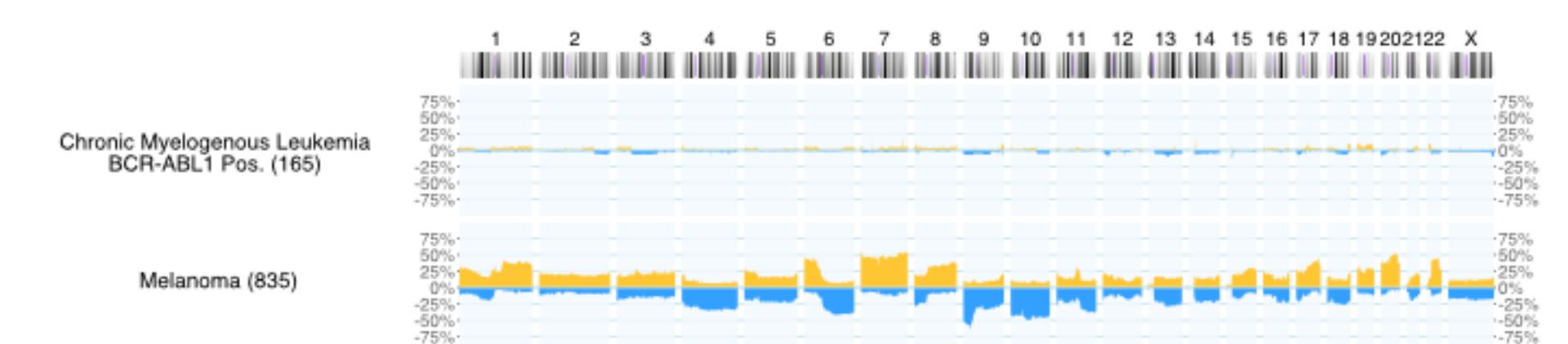


Genome CNV coverage in Cancers

- 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)



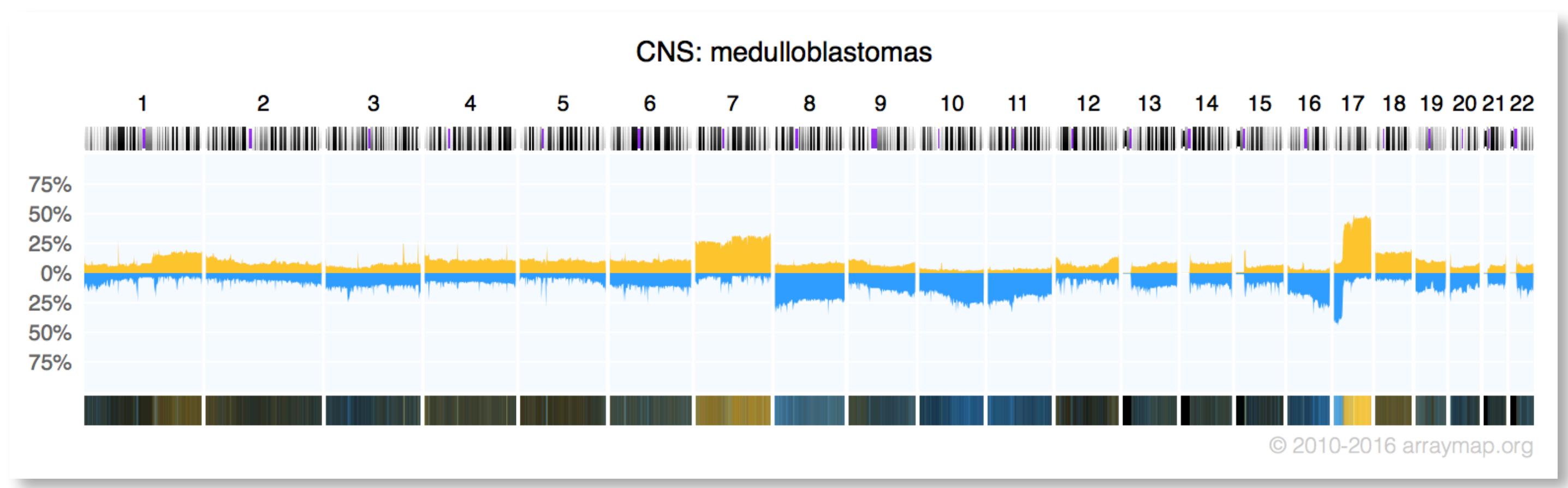
Lowest / Highest CNV fractions =>



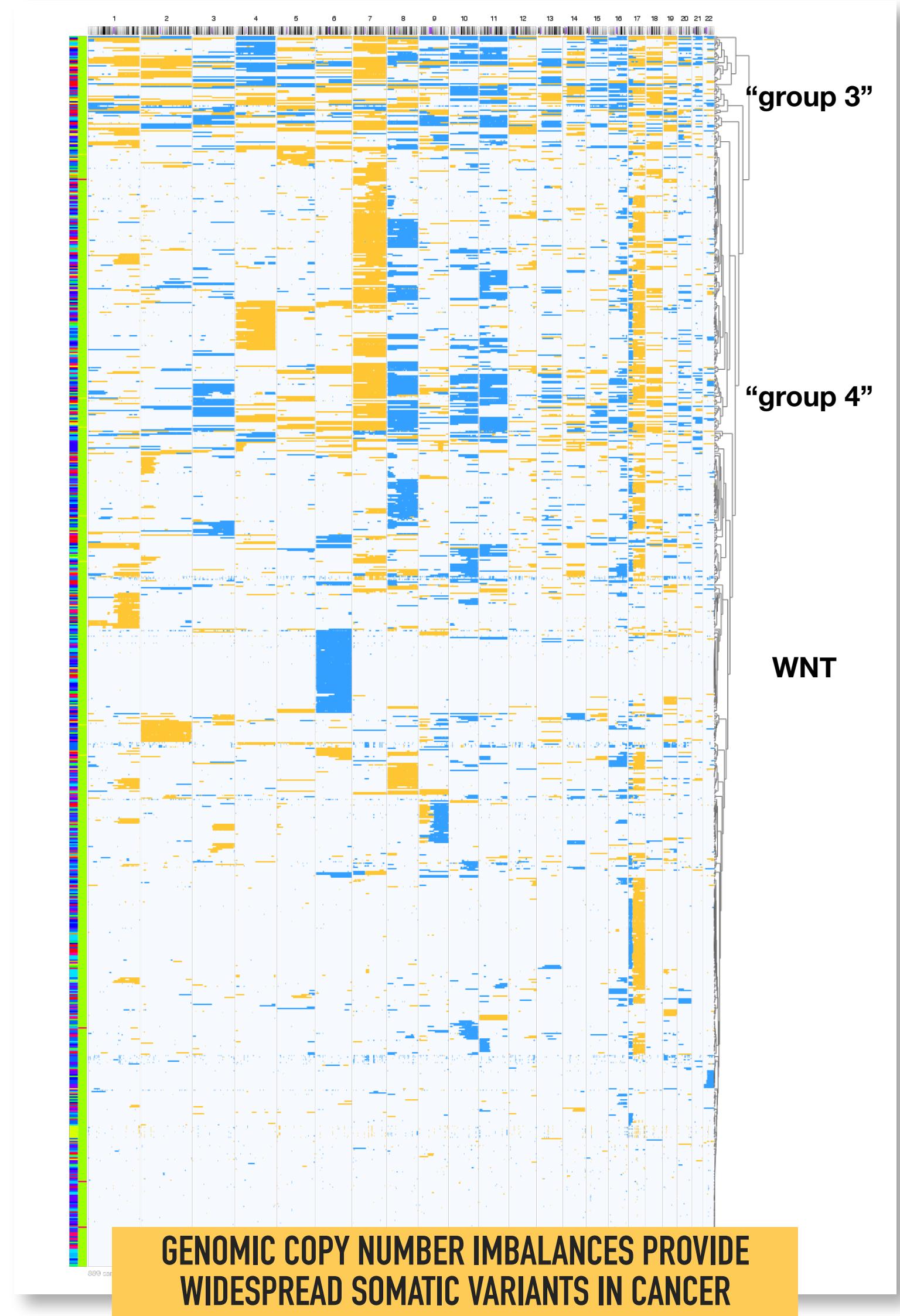
Somatic CNVs In Cancer

Recurrent mutation patterns

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



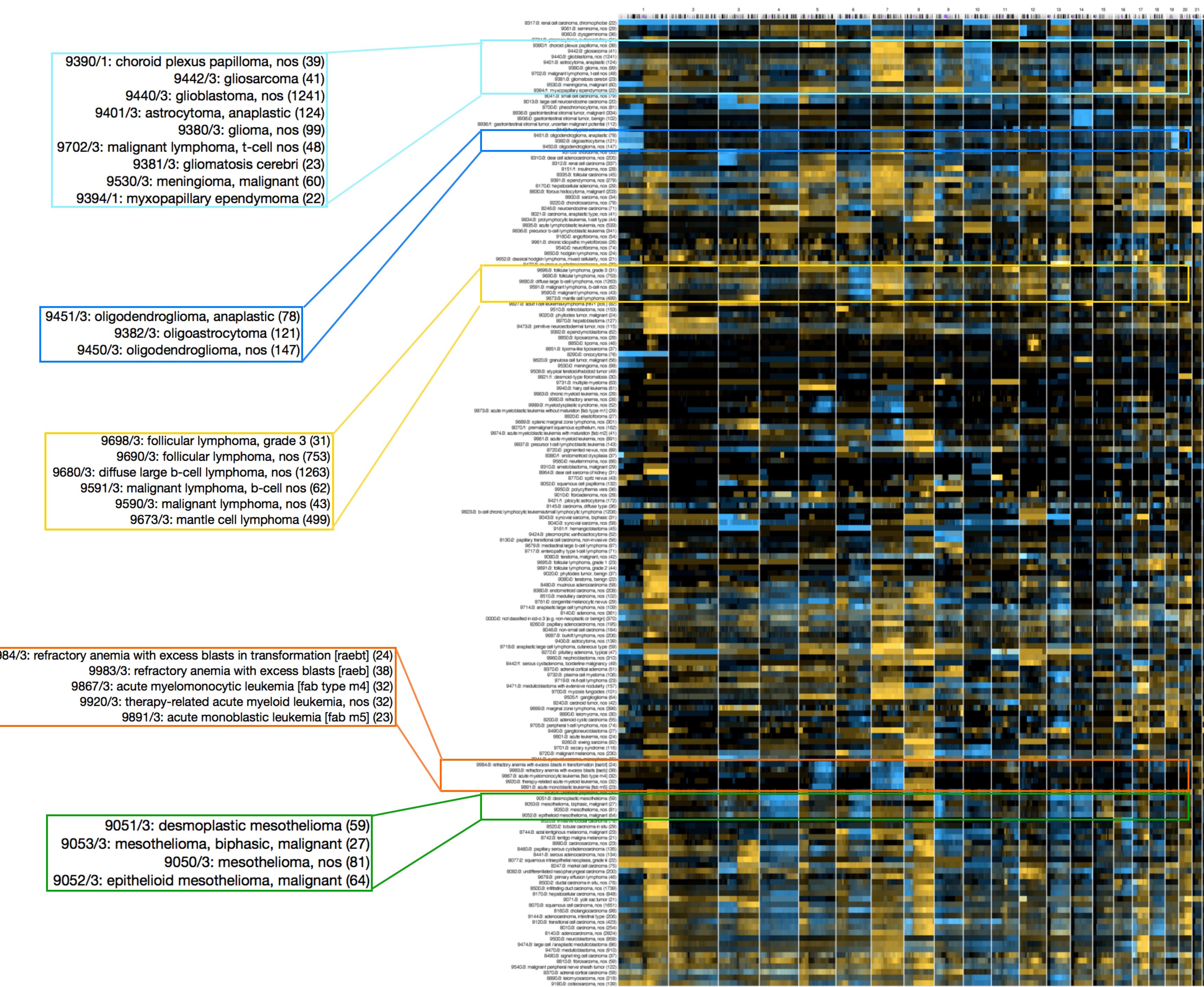
A genomic copy number histogram for malignant medulloblastomas, the most frequent type of pediatric brain tumors, displaying regions of genomic duplications and deletions. These can be decomposed into individual tumor profiles which segregate into several clusters of related mutation patterns with functional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



Somatic Mutations In Cancer: Patterns

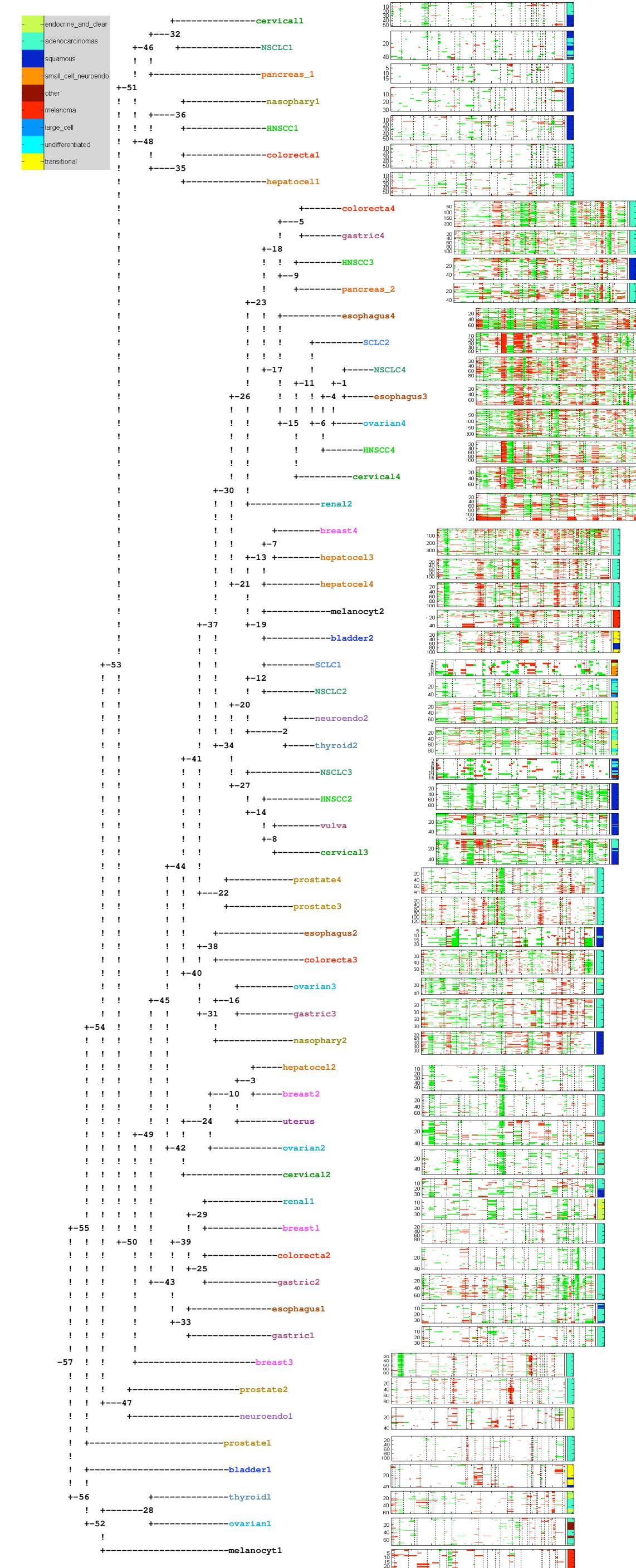
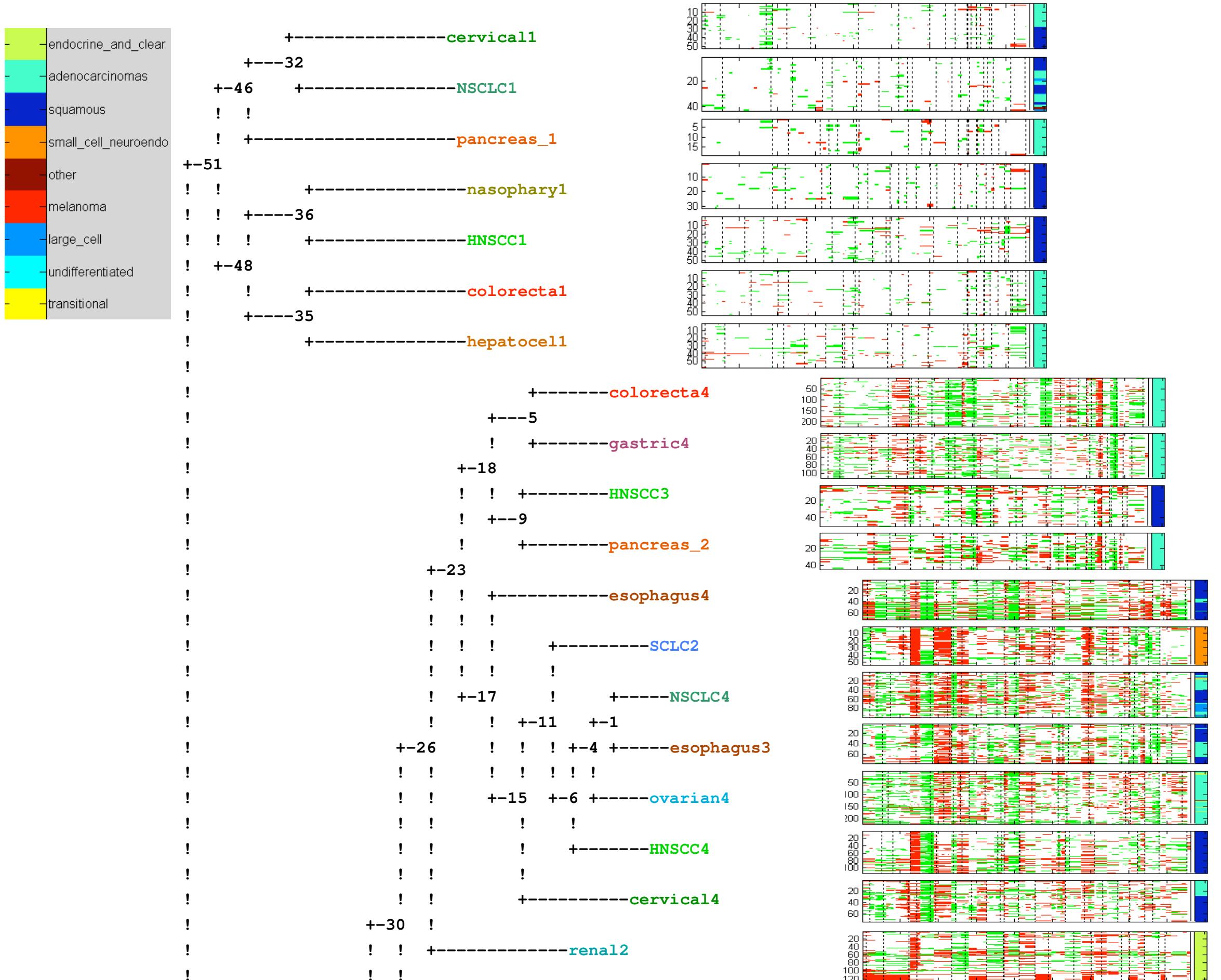
Making the case for genomic classifications

Some related cancer entities show similar copy number profiles



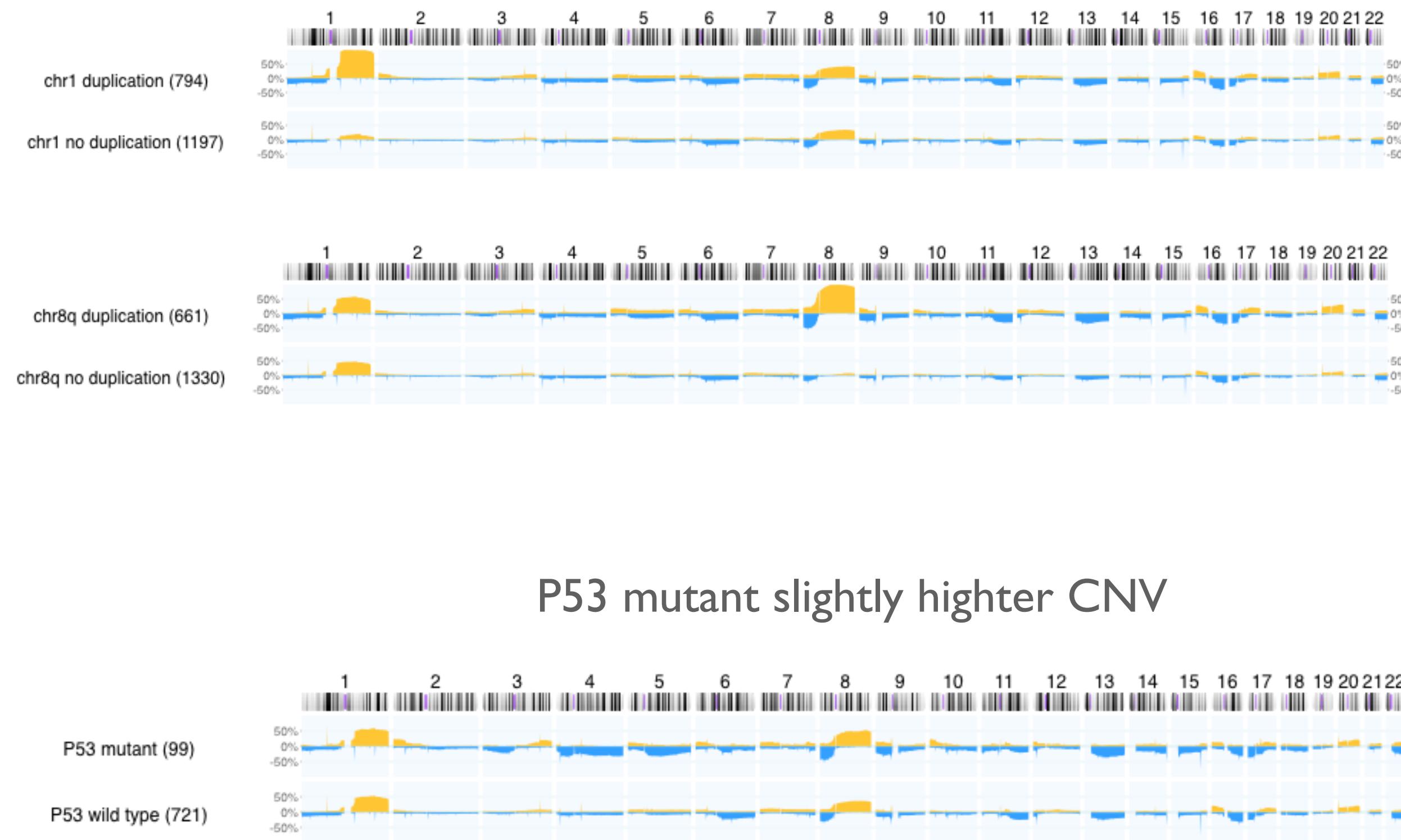
Gene expression

Inferring progression models for CGH data

Jun Liu¹, Nirmalya Bandyopadhyay^{1,*}, Sanjay Ranka¹, M. Baudis² and Tamer Kahveci^{1,*}¹Computer and Information Science and Engineering, University of Florida, Gainesville, FL, USA and ²Institute for Molecular Biology, University of Zurich, Zurich, Switzerland

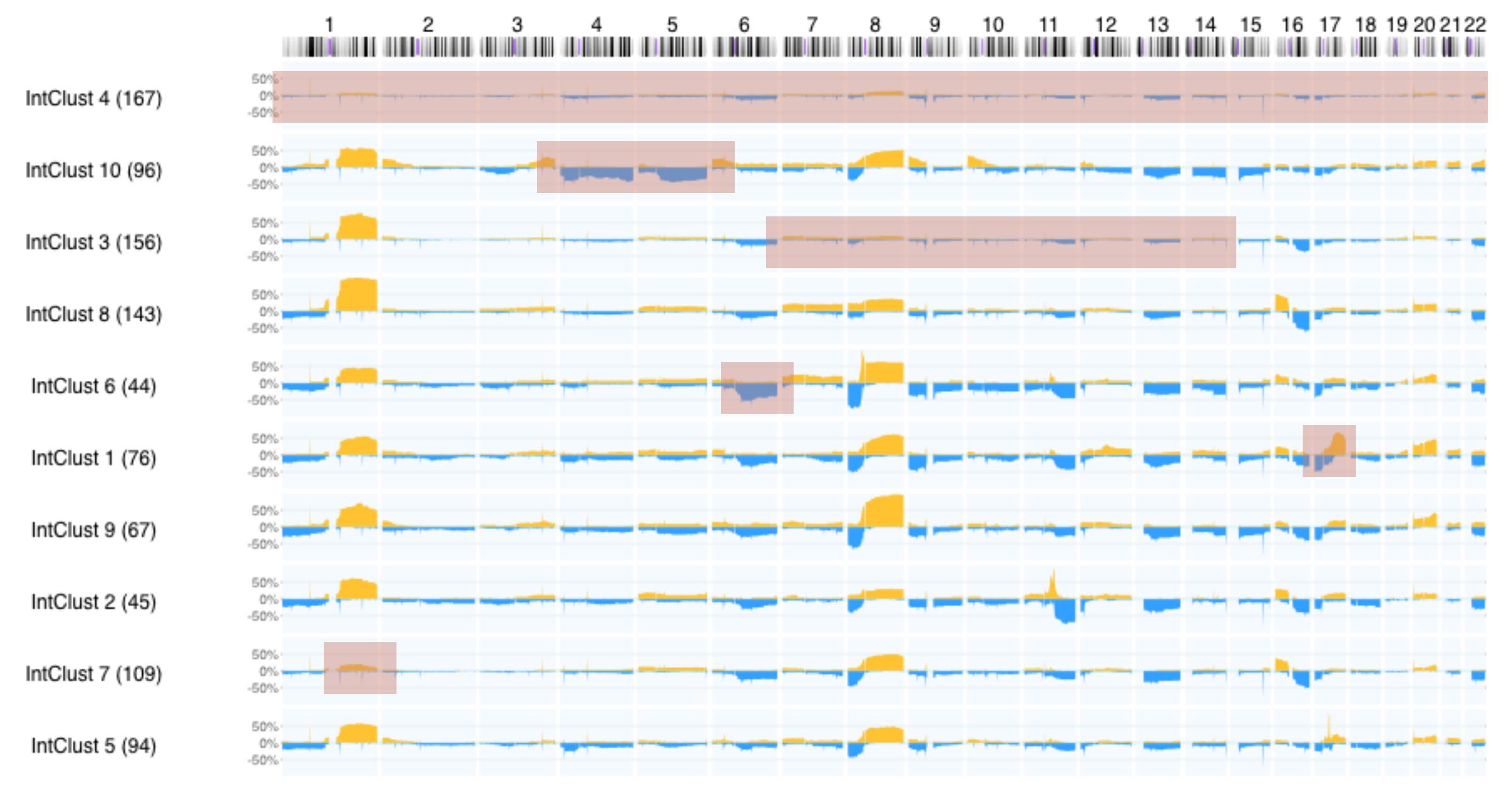
Interpret CNV by Association to Molecular/Clinical Information

Chr1q and chr8q duplication unlinked



P53 mutant slightly higher CNV

Some IntClust groups define Distinctive CNV patterns



METABRIC

Interpret CNV by Association to Molecular/Clinical Information

ER status

ER neg (440)

ER pos (1508)

not specified (44)

HER2 level

HER2 level 0 (5)

HER2 level 2 (27)

HER2 level 3 (121)

not specified (1168)

HER2 level 1 (671)

Pam50 category

→ Pam50 Normal (202)

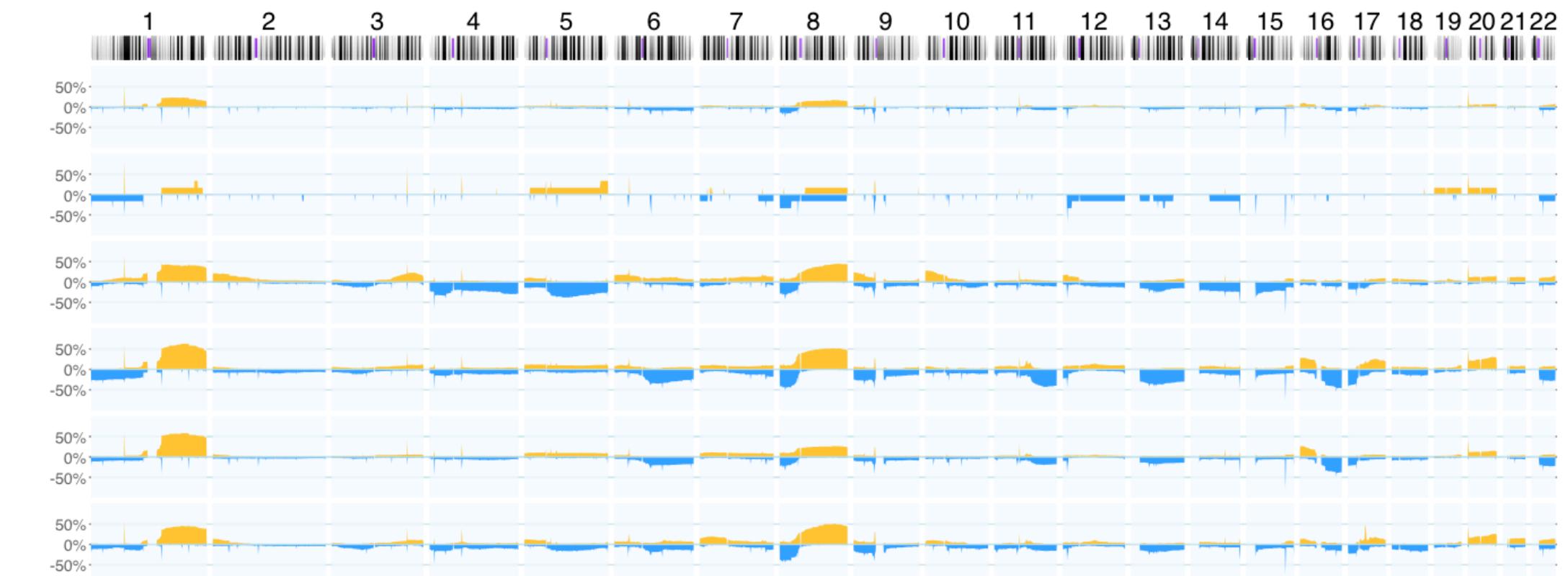
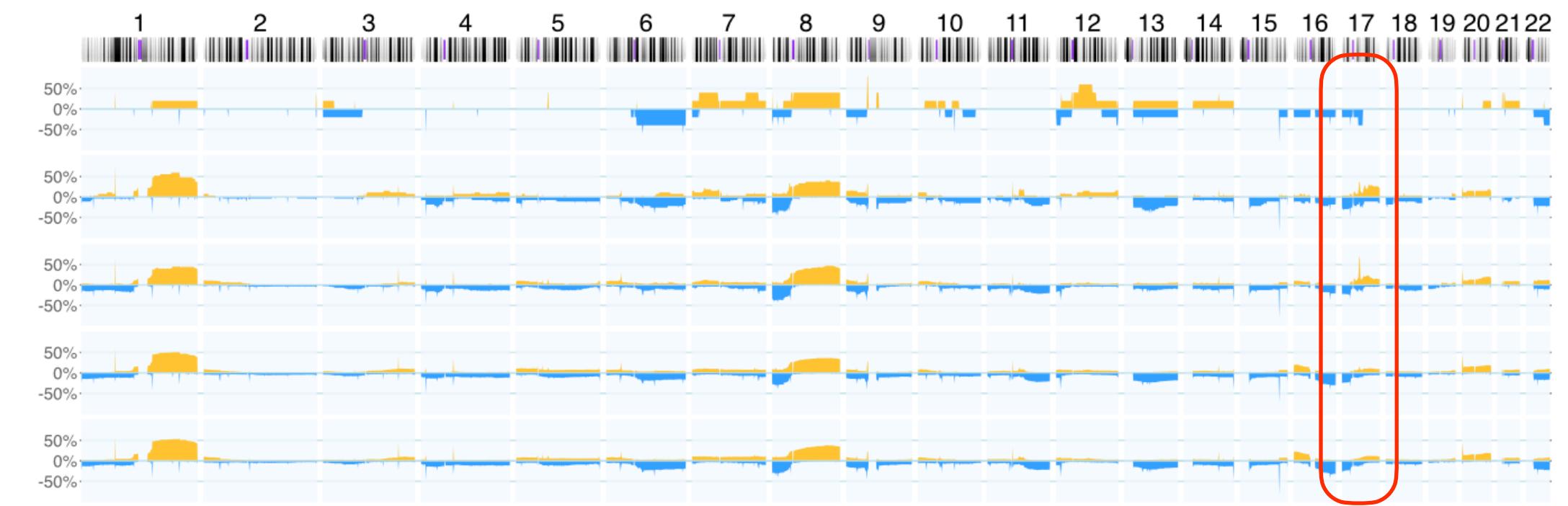
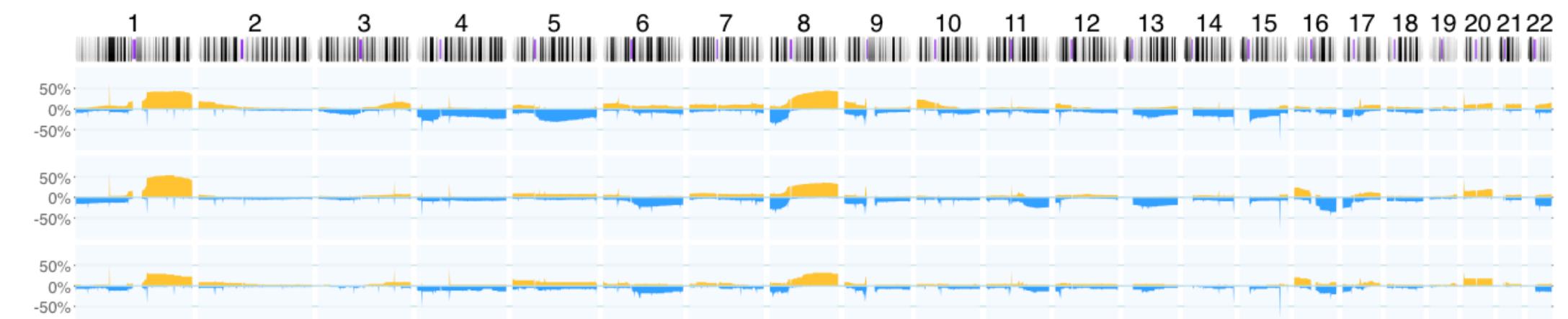
not specified (6)

Pam50 Basal (331)

Pam50 LumB (492)

Pam50 LumA (721)

Pam50 Her2 (240)



Progenetix & arrayMap: Available formats

- DB dumps (variants, biosamples, callsets)
 - status values for all CNV variants
 - 4'606'156 | 3'264'504 DEL
 - 3'916'360 | 2'602'169 DUP
 - pre-computed 1MB binned status levels for all callsets
 - (so far) uncalibrated log2 intensity values for all intervals hit by a CNV
 - pre-calculated 1MB CNV frequencies for all mapped entities (NCIT, ICDOM, ICDOT, PMID, GSE, CVCL ...)

Inference of integer copy number states

- Cancer samples are made of heterogeneous cells
 - Normal tissue contamination
 - Cancer evolves as it acquires extra mutations
 - The majority of current copy number segmentations methods do not distinguish between heterogeneous cell components
- It is of interest to know the exact copy number
 - e.g. full deletion (0), single copy deletion (1), normal copy number (2), single copy duplication (3), double copy duplication (4), amplification (5+), etc.
 - Full deletion - tumour suppressor genes
 - amplification - oncogenes

intCNA - Estimating copy number counts

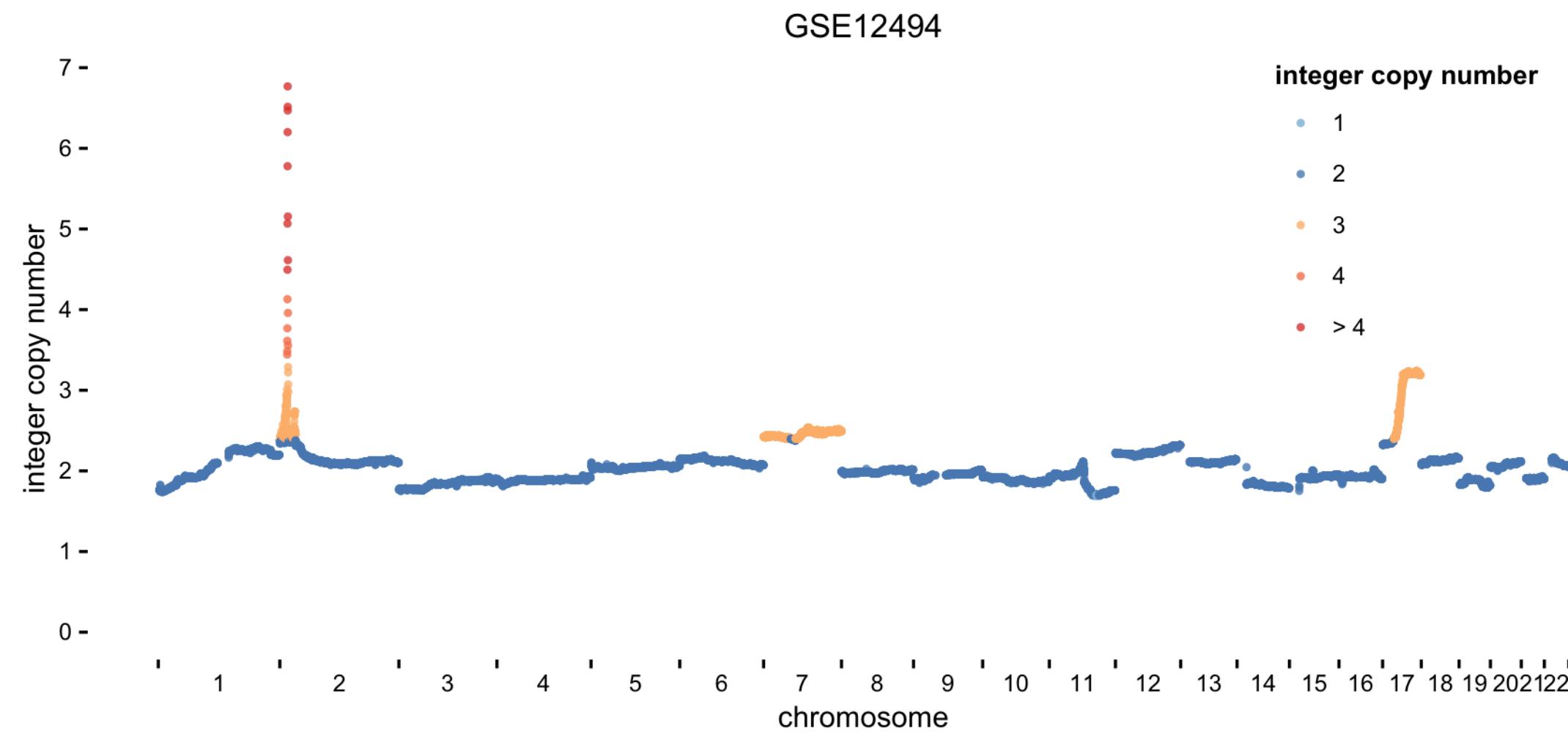


Figure 5: Mean values of integer copy number for 194 neuroblastoma samples containing discernible CNAs from GSE12494 [17]. Recurrent CNAs including MYCN amplification at 2p24, chromosome 17 gain, chromosome 7 gain, 1p and 11q loss.

intCNA - Estimating copy number counts

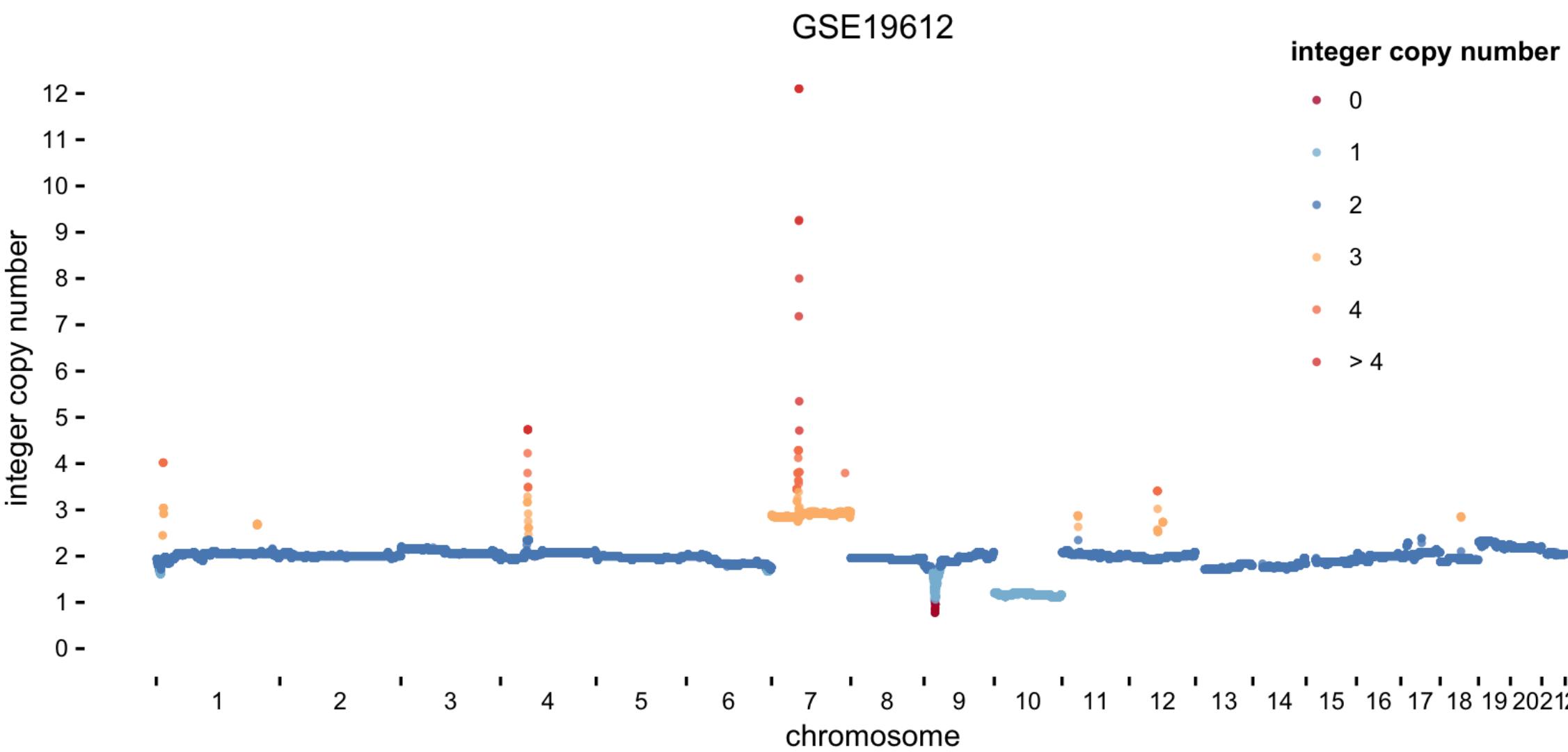
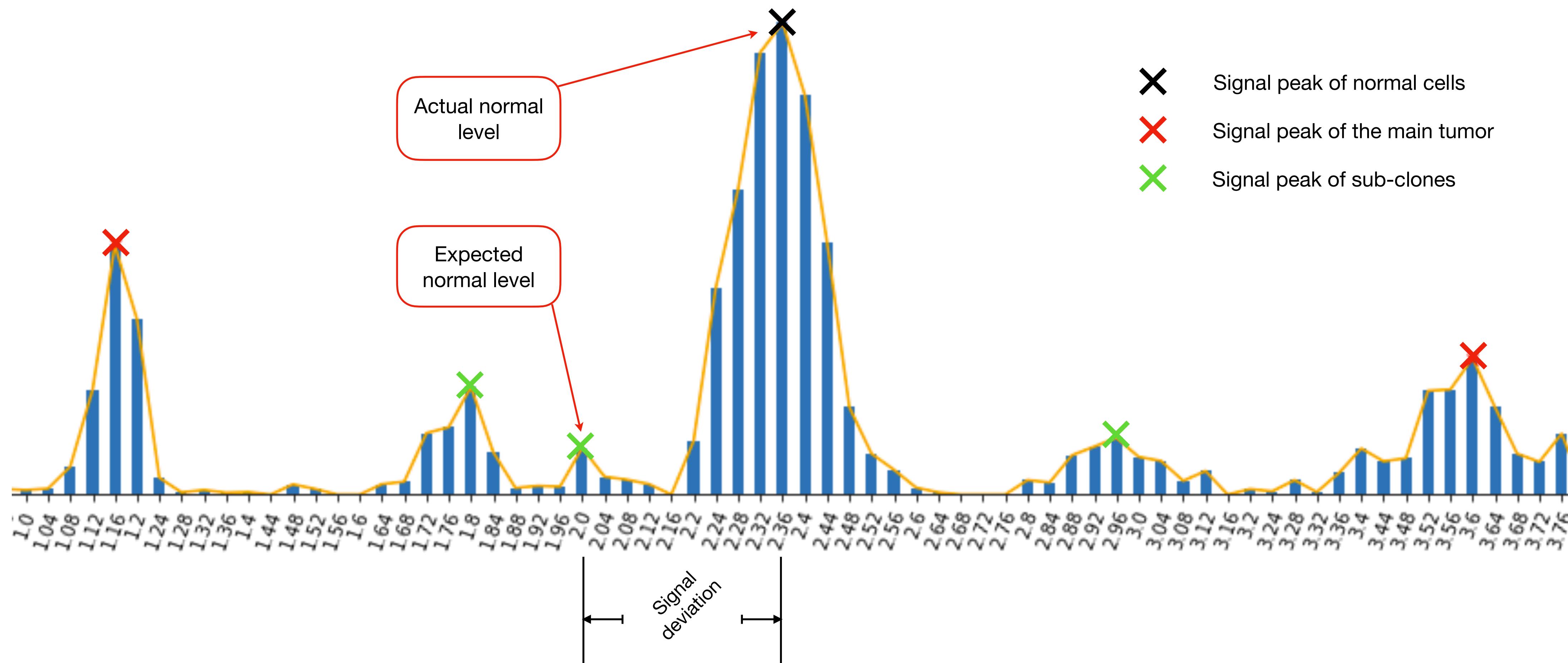


Figure 6: Mean values of integer copy number for 49 glioblastoma samples containing discernible CNAs from GSE19612 [13]. Recurrent CNAs including chromosome 7 gain, chromosome 10 loss, EGFR amplification at 7p11.2, PDGFRA amplification at 4q12, homozygous and heterozygous focal deletion of 9p21 where CDKN2A, CDKN2B and MTAP are located.

A visual illustration

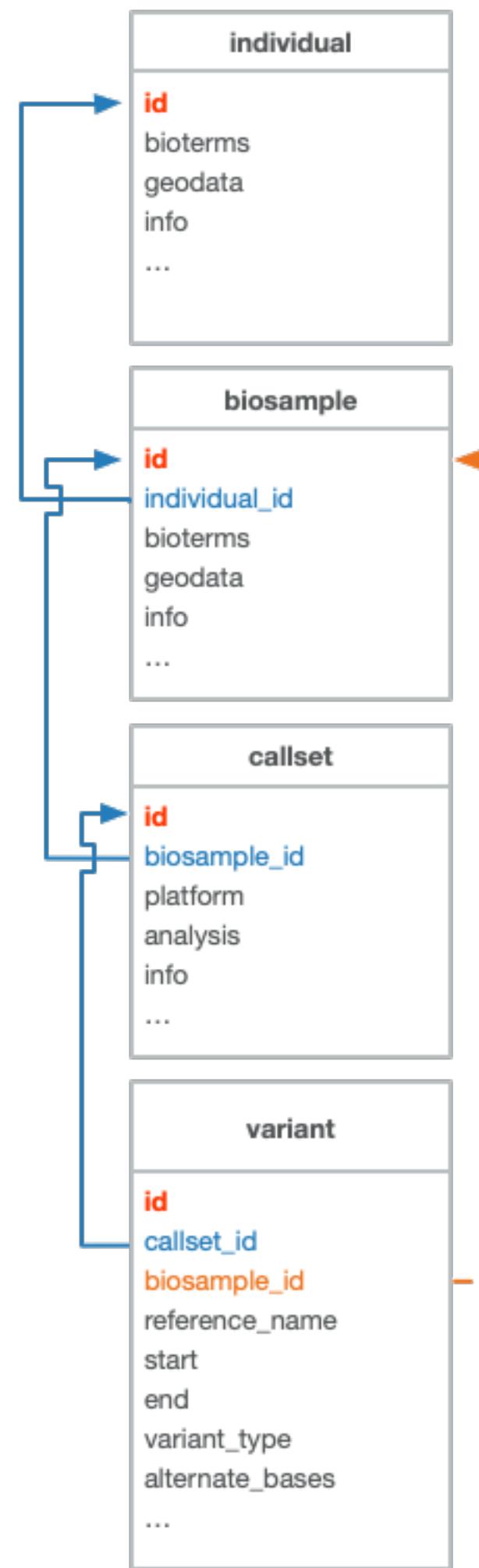


Progenetix & arrayMap: Data Notes

- currently ongoing data calibration
 - baseline
 - QC & purging
 - "calibrated log2" generation for comparable values across samples
- instead of 1MB mapping -> any binning or pre-selected set (gene CDRs ...) can be generated
- more/cleaner metadata always achievable (but work/interest ...)
- population background for SNP datasets ...



- Progenetix uses document storage databases, implemented in **MongoDB**
- the schemas are aligned with the original GA4GH data model and track ongoing GA4GH projects (PXF, Beacon, VR), as represented through {S}[B]
- **variants**
 - The variant object includes attributes and examples for both structural (DUP, DEL, BRK) and precise genome variants.
- **callsets**
 - The callset object is for technical data and series information (e.g. used platform and analysis methods). It is not strictly needed for querying combined variant + biosample aspects, since in the current implementation the variant object contains a reference to the biosample it was derived from.
- **biosamples**
 - Most relevant "bio" data (such as diagnoses, phenotypes ...) is stored in the biosample object.
- **individuals**
 - The individual object contains information which pertains to the whole biological entity biosamples are derived from (e.g. sex, heritable phenotypes...).



A Python Based Beacon API Implementation for the Progenetix Data Model

Directory Structure

bin

- applications for data access & processing

bycon

- Python modules

cgi

- web server apps - "beacon" et al.

config

- configuration files, separated for topic/scope
- YAML ...

data/in, data/out, data/out/yaml

- input and output for example and test data

Usage Examples

- `bin/pgxport.py -d progenetix -a 0.5 -j '{ "biosamples": { "biocharacteristics.type.id": "$regex\":\"icdom-94403\" } }'`
 - uses the dataset `progenetix`
 - the provided JSON query string for the `MongoDB` backend will retrieve all samples from `progenetix.biosamples` which have an *ICD-O* 3 code of "9440/3" (pgx version is "icdom-94403") corresponding to a "Glioblastoma, NOS" diagnosis
 - unspecified output methods will use the current defaults
 - plot dots will have an opacity of "0.5"
- `bin/pgx_update_mappings.py -f rsrc/progenetix-icdo-to-ncit.ods -d arraymap,progenetix`
 - updates NCIt mappings in the `arraymap` and `progenetix` databases from the specified mapping file
- `bin/pgx_update_mappings.py -f rsrc/progenetix-icdo-to-ncit.ods -d arraymap,progenetix -y ~/switchdrive/work/GitHub/progenetix/ICDOntologies/current`
 - this version of the ICD => NCIT mapping updater will read from the specified input table and - besides updating the NCIt codes which have different/missing values for the same ICD-O M+T combinations - write the YAML files to the local GitHub repository for `ICDOntologies` (obviously YDMV)



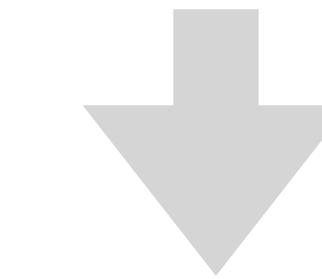
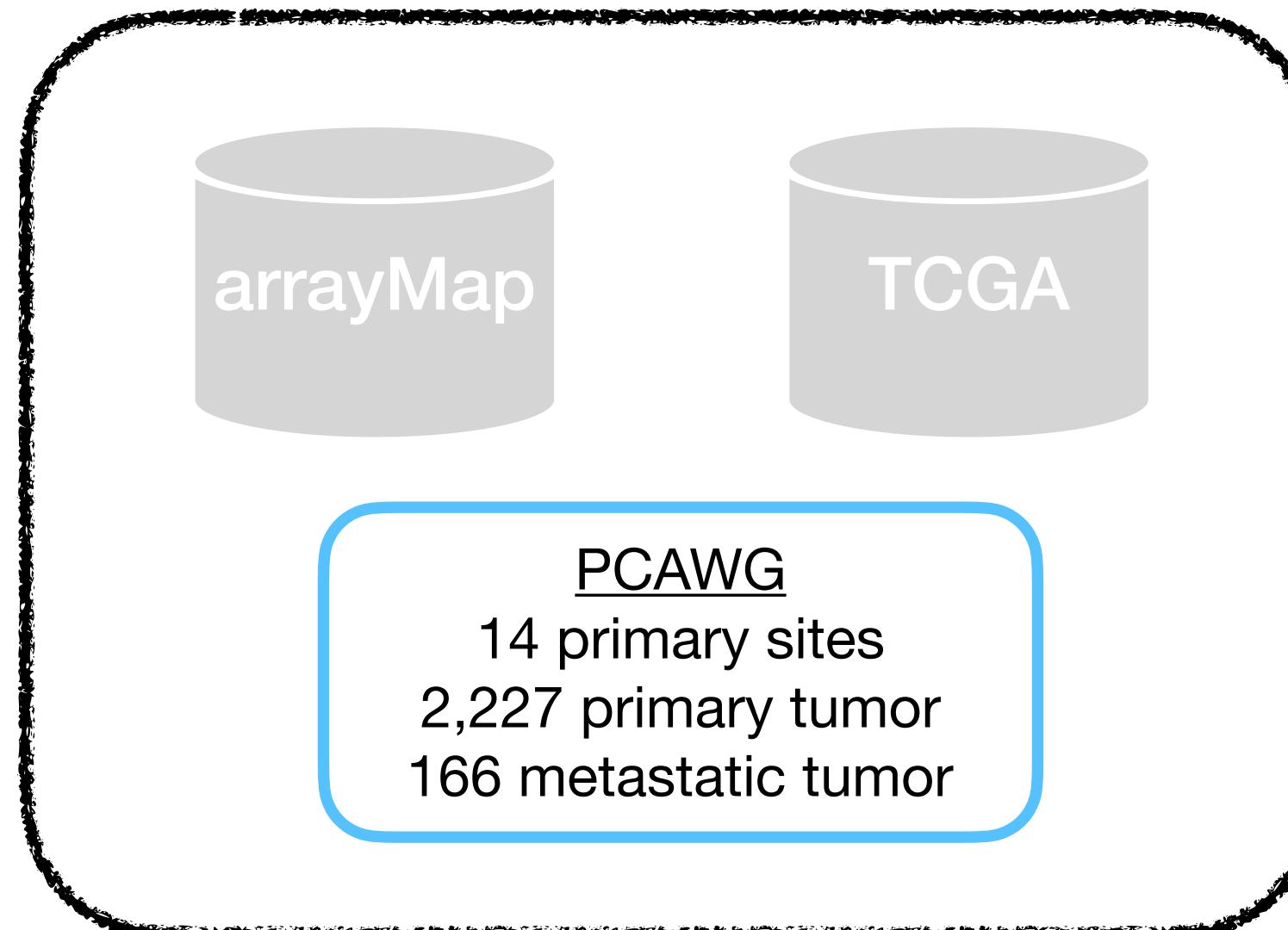
PCAWG - Pancancer Analysis Of Whole Genomes

Available Data

Data type	Donors	Files	Strategy	Access
Simple Somatic Mutation	2715	25'501	WGS	Controlled
Structural Somatic Mutation	2715	14'195	WGS	Controlled
Aligned Reads	2793	12'169	RNA-Seq	Controlled
Simple Germline Variation	2715	8'505	WGS	Controlled
Copy Number Somatic Muation	2715	5'671	WGS	Controlled
Structural Germline Variants	2715	8'505	WGS	Controlled
Clinical Metadata	2715	-	-	Public
Experiment Metadata	2715	-	-	Public

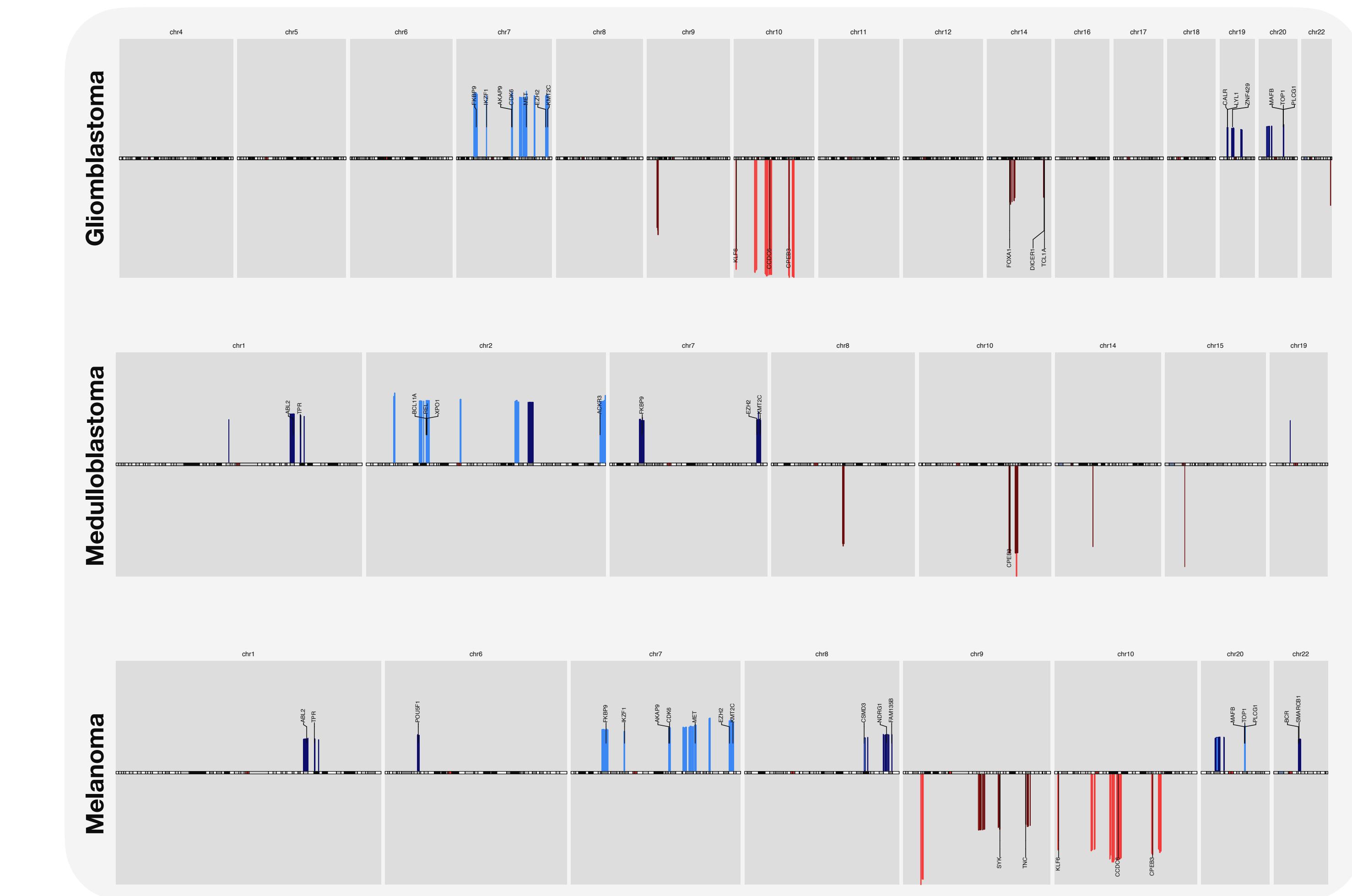
Unique Patterns of Copy Number Mutations Across Cancer Types

An extensive collection of tumor CNV



- Unique & distinctive patterns
- Patterns of sites and disease
- Identify the origin of a tumor

Examples of unique CNV patterns



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