

Precision Oncology Course

Exercise: cBioPortal

www.cbioportal.org



Memorial Sloan Kettering
Cancer Center

cBioPortal is a resource for **interactive exploration** of multidimensional
cancer data.

Multidimensional cancer data



cBioPortal integrates different types of data:

- **Epigenomics:** methylation data
- **Genomics:** non-synonymous mutations and CN data
- **Transcriptomics:** mRNA and microRNA expression data
- **Proteomics:** protein and phosphoprotein level data
- **Clinical data (de-identified)**

Main Functionalities



- Stores data from different projects, including TCGA.
- Allows to visualize the data using different types of plots.
- Can perform comparisons between patients (of the same or different projects): expression, mutations, survival, etc.
- Allows to explore the data available for individual patients.
- Users can download the raw data, the plots and the results.
- Users can use their own data to draw some plots (OncoPrints and Lollipop plots).

Query

Quick Search Beta!

Download

Please cite: Cerami et al., 2012 & Gao et al., 2013

Select Studies for Visualization & Analysis:

0 studies selected (0 samples)

Search...

PanCancer Studies	8
Pediatric Cancer Studies	13
Immunogenomic Studies	8
Cell lines	3
Adrenal Gland	3
Ampulla of Vater	1
Biliary Tract	13
Bladder/Urinary Tract	17
Bone	2
Bowel	11
Breast	21
CNS/Brain	20
Cervix	2
Esophagus/Stomach	17
Eye	5

 Select all listed studies matching filter (21)

Breast

- Breast Cancer (MSK, Cancer Cell 2018) 1918 samples   
- Breast Cancer (MSK, Nature Cancer 2020) 141 samples   
- Metastatic Breast Cancer (MSK, Cancer Discovery 2021) 1365 samples   
- Breast Fibroepithelial Neoplasms**
- Breast Fibroepithelial Tumors (Duke-NUS, Nat Genet 2015) 22 samples   
- Invasive Breast Carcinoma**
- Breast Cancer (METABRIC, Nature 2012 & Nat Commun 2016) 2509 samples   
- Breast Cancer (MSK, Clinical Cancer Res 2020) 60 samples   
- Breast Cancer (MSKCC, NPJ Breast Cancer 2019) 70 samples   
- Breast Cancer (SMC 2018) 187 samples   
- Breast Cancer Xenografts (British Columbia, Nature 2015) 117 samples   
- Breast Invasive Carcinoma (British Columbia, Nature 2012) 65 samples   
- Breast Invasive Carcinoma (Broad, Nature 2012) 103 samples   
- Breast Invasive Carcinoma (Sanger, Nature 2012) 100 samples   
- Breast Invasive Carcinoma (TCGA, Cell 2015) 817 samples   
- Breast Invasive Carcinoma (TCGA, Firehose Legacy) 1108 samples   
- Breast Invasive Carcinoma (TCGA, Nature 2012) 825 samples   
- Breast Invasive Carcinoma (TCGA, PanCancer Atlas) 1084 samples   
- Juvenile Papillomatosis and Breast Cancer (MSK, 2020) 5 samples   
- Metastatic Breast Cancer (INSERM, PLoS Med 2016) 216 samples   
- The Metastatic Breast Cancer Project (Provisional, February 2020) 237 samples   

0 studies selected (0 samples)

Query By Gene

OR

 Explore Selected Studies

What's New

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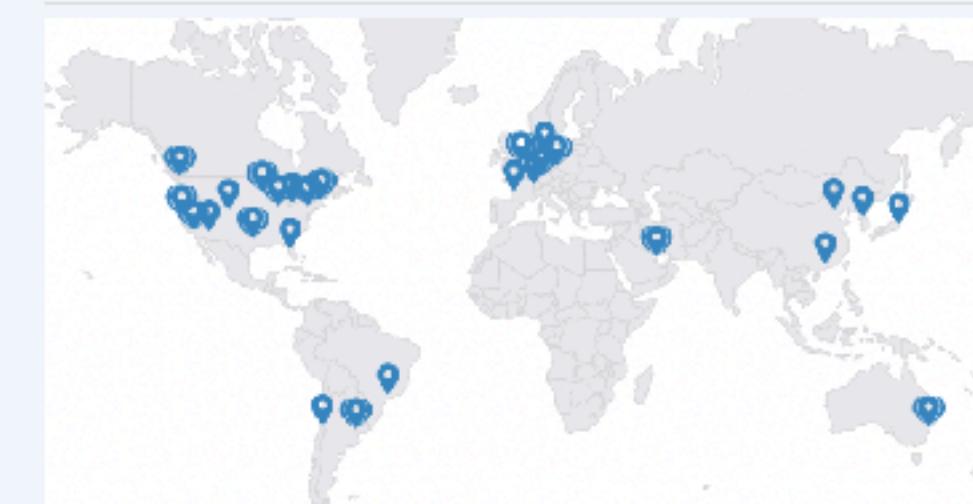

Data for a new study by Stopsack et al. on differences in prostate cancer genomes by self-reported race just published in [@CCR_AACR](#) are available in cBioPortal here: [cbiportal.org/study/summary?... https://twitter.com/DrPhilKantoff/status/1450566128581873666](#)

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Example Queries

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Affected Tissue

0 studies selected (0 samples)

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 1365 samples   

 # samples, info and
link to publication

Invasive Breast Carcinoma

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Whole exome sequencing of 22 phyllodes tumors

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237 samples   

Brief description with
the type of sequencing:
targeted, WES, WGS

0 studies selected (0 samples)
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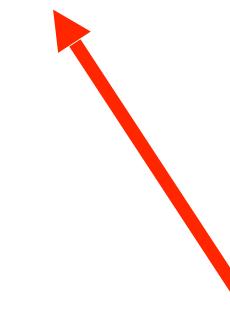
Query

Quick Search **Beta!**

Download

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e.g. Lung, EGFR, TCGA-OR-A5J2



We would love to hear what you think: cbiportal@googlegroups.com

Now, you can make quick queries by type of cancer,
project name, patient ID or genes of interest

Let's learn how to make queries!

What's New

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Types of queries



1. Query across a cohort
2. Cross-cohort query using genes of interest
3. Visualization/interpretation of individual tumor samples

Query across a cohort

Glioma Example

Query 1: Glioma



1 Select "CNS/Brain"

2 Select TCGA cohort

3 Query

Query Quick Search Beta! Download Please cite: Cerami et al., 2012 & Gao et al., 2013

Select Studies for Visualization & Analysis: 1 study selected (514 samples) Deselect all Search...

Ampulla of Vater 1 Select all listed studies matching filter (20)

Biliary Tract 13

CNS/Brain 20 Brain Lower Grade Glioma (TCGA, PanCancer Atlas) 514 samples

Bladder/Urinary Tract 17

Bone 2

Bowel 12

Breast 22

CNS/Brain 20

Cervix 2

Esophagus/Stomach 17

Eye 5

Diffuse Glioma

Brain Lower Grade Glioma (TCGA, Firehose Legacy) 530 samples

Brain Lower Grade Glioma (TCGA, PanCancer Atlas) 514 samples

Glioma (MSK, Nature 2019) 91 samples

Glioma (MSKCC, Clin Cancer Res 2019) 1004 samples

Low-Grade Gliomas (UCSF, Science 2014) 61 samples

Merged Cohort of LGG and GBM (TCGA, Cell 2016) 1102 samples

→ GLIOBLASTOMA

Brain Tumor PDXs (Mayo Clinic, 2019) 97 samples

Glioblastoma (Columbia, Nat Med. 2019) 42 samples

Glioblastoma (TCGA, Cell 2013) 543 samples

Glioblastoma (TCGA, Nature 2008) 206 samples

Glioblastoma Multiforme (TCGA, Firehose Legacy) 619 samples

Glioblastoma Multiforme (TCGA, PanCancer Atlas) 592 samples

1 study selected (514 samples) Deselect all Query By Gene OR Explore Selected Studies

What's New @cbioportal

cBioPortal Retweeted The Hyve @TheHyveNL Adding single-cell data and visualisations to #cBioPortal would reduce the time spent finding, downloading, and processing data for both users new to #singlecell data and users with expertise in single-cell technologies. ow.ly/OyaE50Gb3jT #cancerresearch #genomics

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Query 1: Glioma



Cohort Summary (red arrow pointing to the 'Summary' tab)

Clinical data (red arrow pointing to the 'Clinical Data' tab)

You can remove plots or download the data (red arrow pointing to the 'Selected: 514 patients | 514 samples' button)

Query genes of interest (red arrow pointing to the 'Query' button in the search bar)

Add other types of plots (red arrow pointing to the 'Charts' dropdown menu)

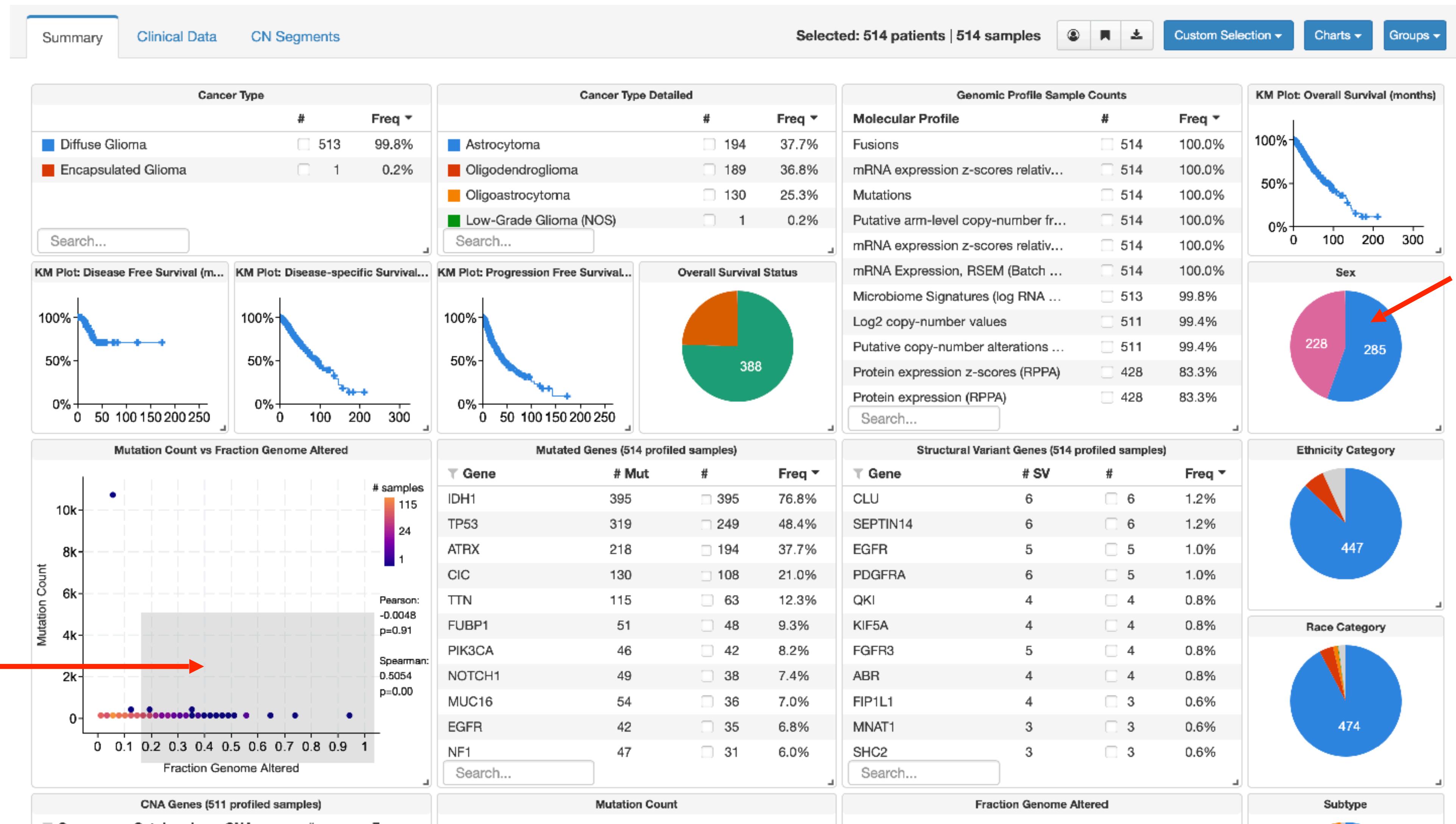
You can rearrange the plots by selecting and dragging (red arrow pointing to the overall layout of the dashboard)

<img alt="Screenshot of the cBioPortal interface for Brain Lower Grade Glioma (TCGA, PanCancer Atlas). The interface includes tabs for Summary, Clinical Data, and CN Segments. The Summary tab is active, showing a 'Cancer Type' table with Diffuse Glioma (99.8%) and Encapsulated Glioma (0.2%). It also features a 'Cancer Type Detailed' panel with a pie chart and options to show pie, compare groups, or download data. Below these are several survival plots (e.g., KM Plot: Disease-Free Survival, KM Plot: Overall Survival) and a pie chart for Overall Survival Status (388). The Clinical Data tab shows a scatter plot of Mutation Count vs Fraction Genome Altered and a table of Mutated Genes (514 samples). The CN Segments tab shows a table of Structural Variant Genes (514 samples). On the right, there are tables for Genomic Profile Sample Counts (e.g., Fusions, mRNA expression z-scores) and pie charts for Sex (228 pink, 285 blue) and Ethnicity Category (447).</p>

Query 1: Glioma



You can filter the samples by selecting features you are interested in



Query 1: Glioma



cBioPortal
FOR CANCER GENOMICS

The plots are redrawn with the selected samples

The filters you applied appear here

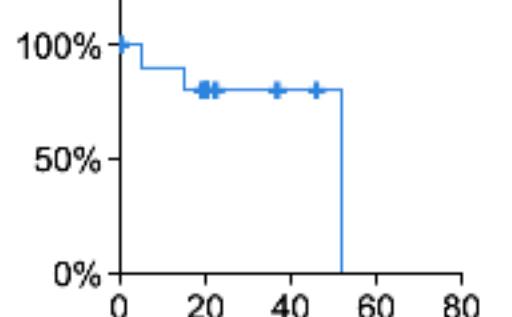
You can create a new group from the selected samples

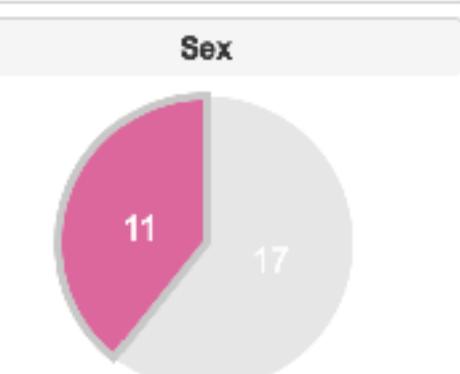
Summary Clinical Data CN Segments Selected: 11 patients | 11 samples Custom Selection Charts Groups

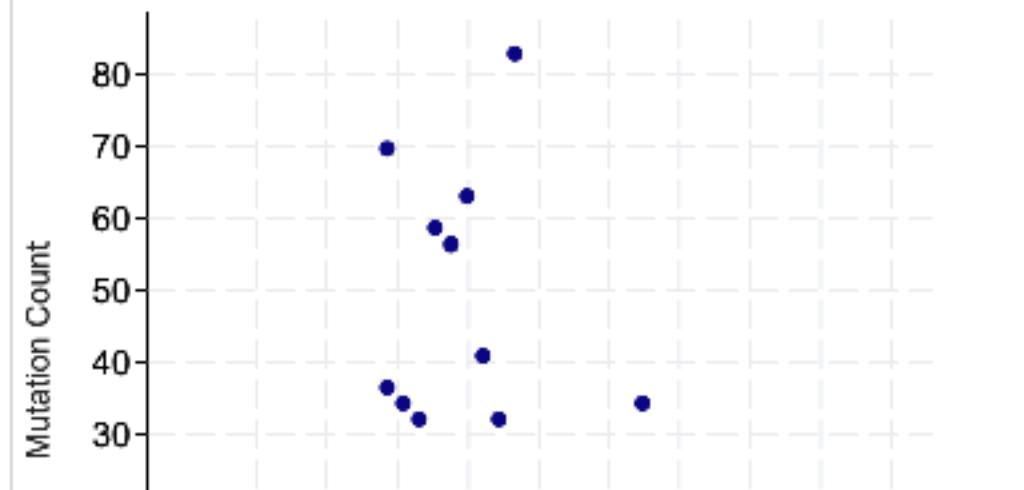
Cancer Type	#	Freq
Diffuse Glioma	11	100.0%

Cancer Type Detailed	#	Freq
Astrocytoma	6	54.5%
Oligodendrogloma	4	36.4%
Oligoastrocytoma	1	9.1%

Genomic Profile Sample Counts	#	Freq
Fusions	11	100.0%
mRNA expression z-scores relativ...	11	100.0%
Mutations	11	100.0%
Putative arm-level copy-number fr...	11	100.0%
Log2 copy-number values	11	100.0%
mRNA expression z-scores relativ...	11	100.0%
mRNA Expression, RSEM (Batch ...	11	100.0%
Putative copy-number alterations ...	11	100.0%
Microbiome Signatures (log RNA ...	11	100.0%
Protein expression z-scores (RPPA)	7	63.6%
Protein expression (RPPA)	7	63.6%

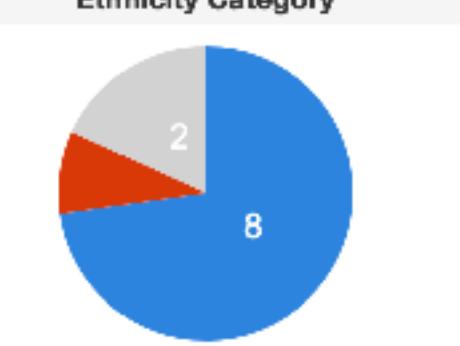
KM Plot: Overall Survival (months)


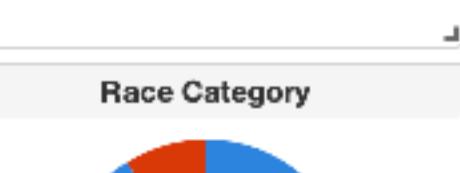
Sex


Mutation Count vs Fraction Genome Altered


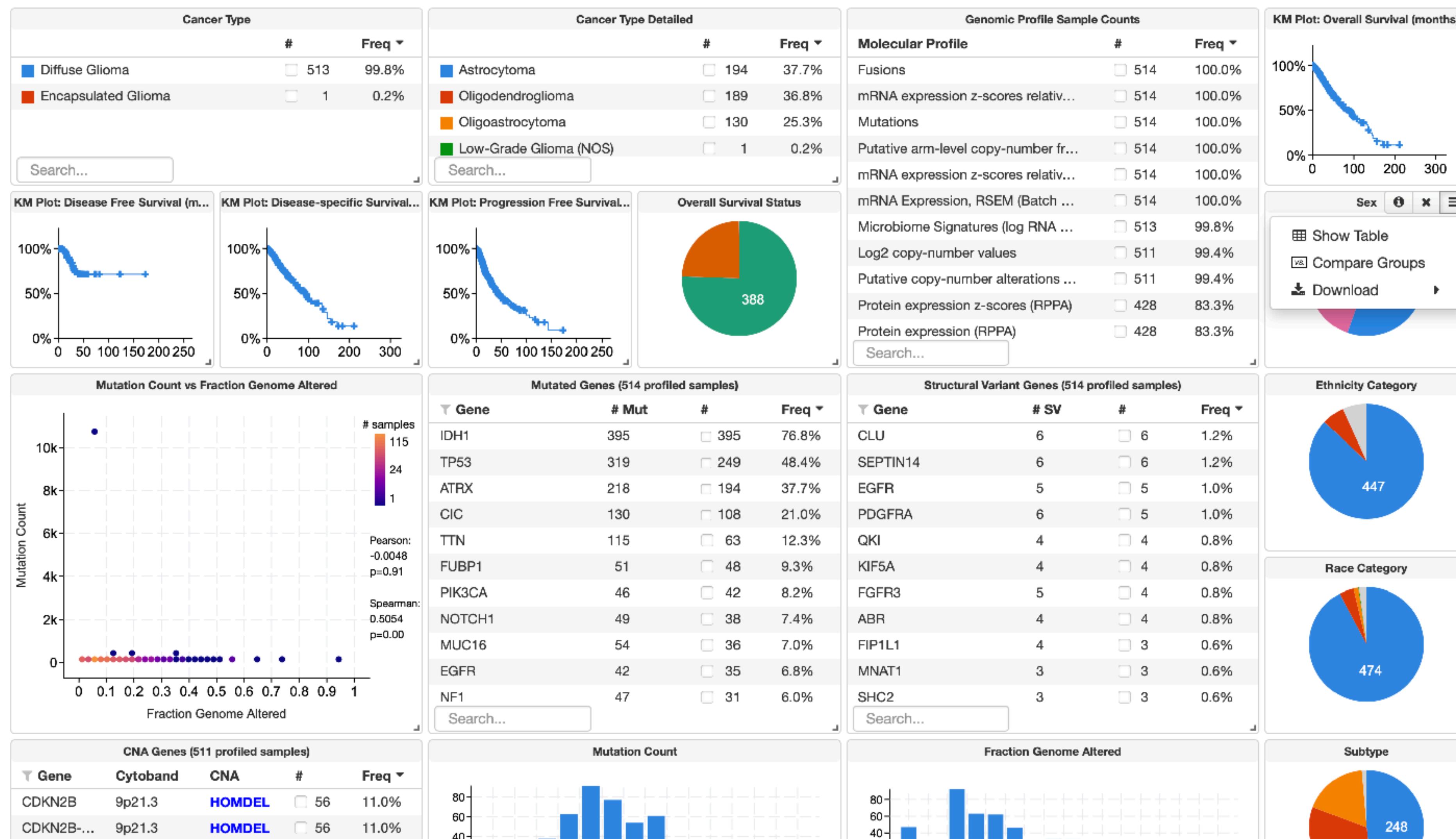
Mutated Genes (11 profiled samples)																																
<table border="1"><thead><tr><th>Gene</th><th># Mut</th><th>#</th><th>Freq</th></tr></thead><tbody><tr><td>TP53</td><td>9</td><td>8</td><td>72.7%</td></tr><tr><td>IDH1</td><td>6</td><td>6</td><td>54.5%</td></tr><tr><td>KRT86</td><td>2</td><td>2</td><td>18.2%</td></tr><tr><td>FLG</td><td>2</td><td>2</td><td>18.2%</td></tr><tr><td>SULT2B1</td><td>2</td><td>2</td><td>18.2%</td></tr><tr><td>PTPRH</td><td>2</td><td>2</td><td>18.2%</td></tr><tr><td>PTPRM</td><td>2</td><td>2</td><td>18.2%</td></tr></tbody></table>	Gene	# Mut	#	Freq	TP53	9	8	72.7%	IDH1	6	6	54.5%	KRT86	2	2	18.2%	FLG	2	2	18.2%	SULT2B1	2	2	18.2%	PTPRH	2	2	18.2%	PTPRM	2	2	18.2%
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Structural Variant Genes (11 profiled samples)																																
<table border="1"><thead><tr><th>Gene</th><th># SV</th><th>#</th><th>Freq</th></tr></thead><tbody><tr><td>EPB41L4A</td><td>1</td><td>1</td><td>9.1%</td></tr><tr><td>TTC3</td><td>1</td><td>1</td><td>9.1%</td></tr><tr><td>SCN9A</td><td>1</td><td>1</td><td>9.1%</td></tr><tr><td>FANCD2</td><td>1</td><td>1</td><td>9.1%</td></tr><tr><td>EXOC7</td><td>1</td><td>1</td><td>9.1%</td></tr><tr><td>OSBPL10</td><td>1</td><td>1</td><td>9.1%</td></tr><tr><td>CCDC112</td><td>1</td><td>1</td><td>9.1%</td></tr></tbody></table>	Gene	# SV	#	Freq	EPB41L4A	1	1	9.1%	TTC3	1	1	9.1%	SCN9A	1	1	9.1%	FANCD2	1	1	9.1%	EXOC7	1	1	9.1%	OSBPL10	1	1	9.1%	CCDC112	1	1	9.1%
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OSBPL10	1	1	9.1%																													
CCDC112	1	1	9.1%																													

Ethnicity Category


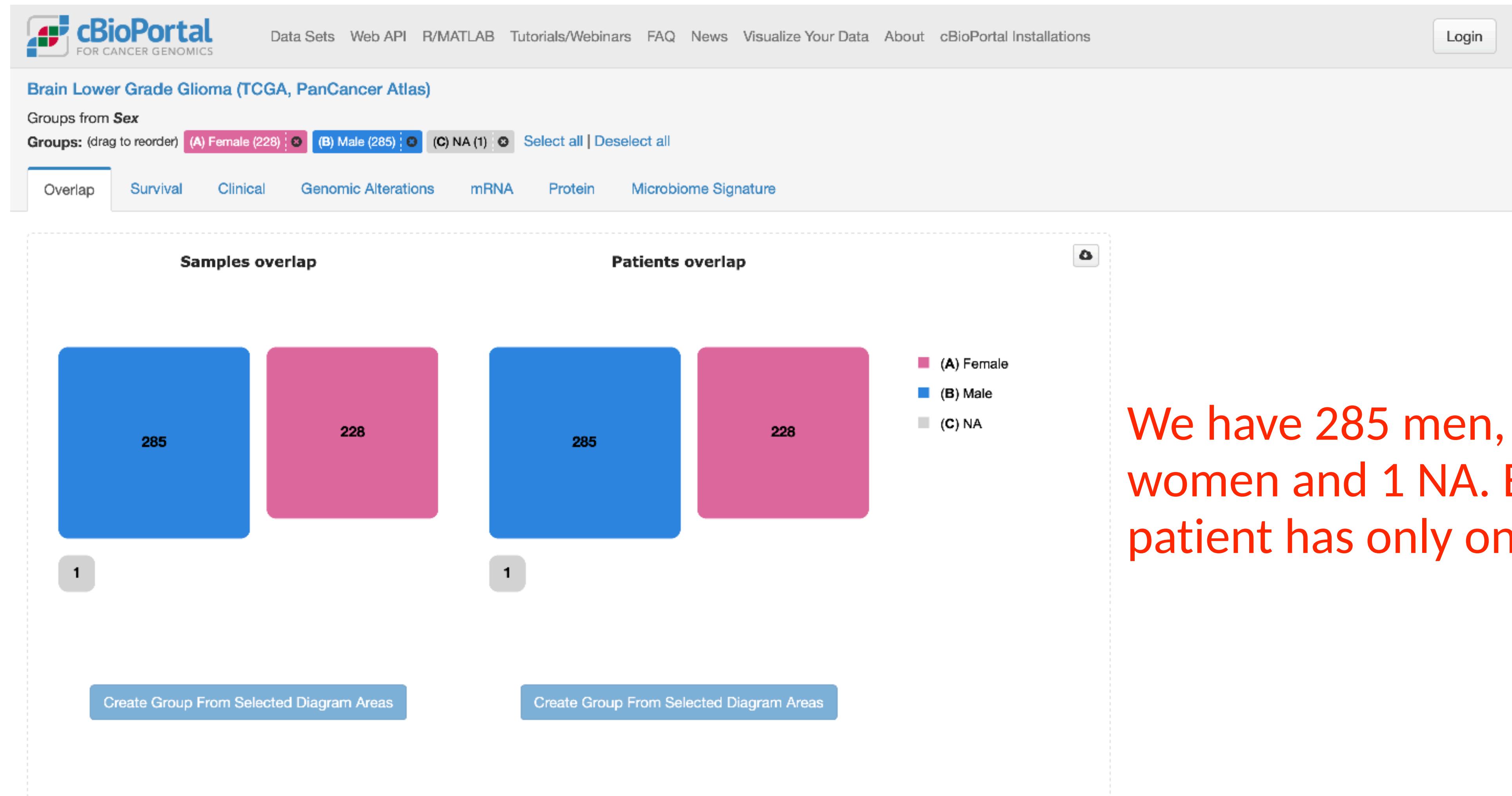
Race Category


Query 1: Glioma

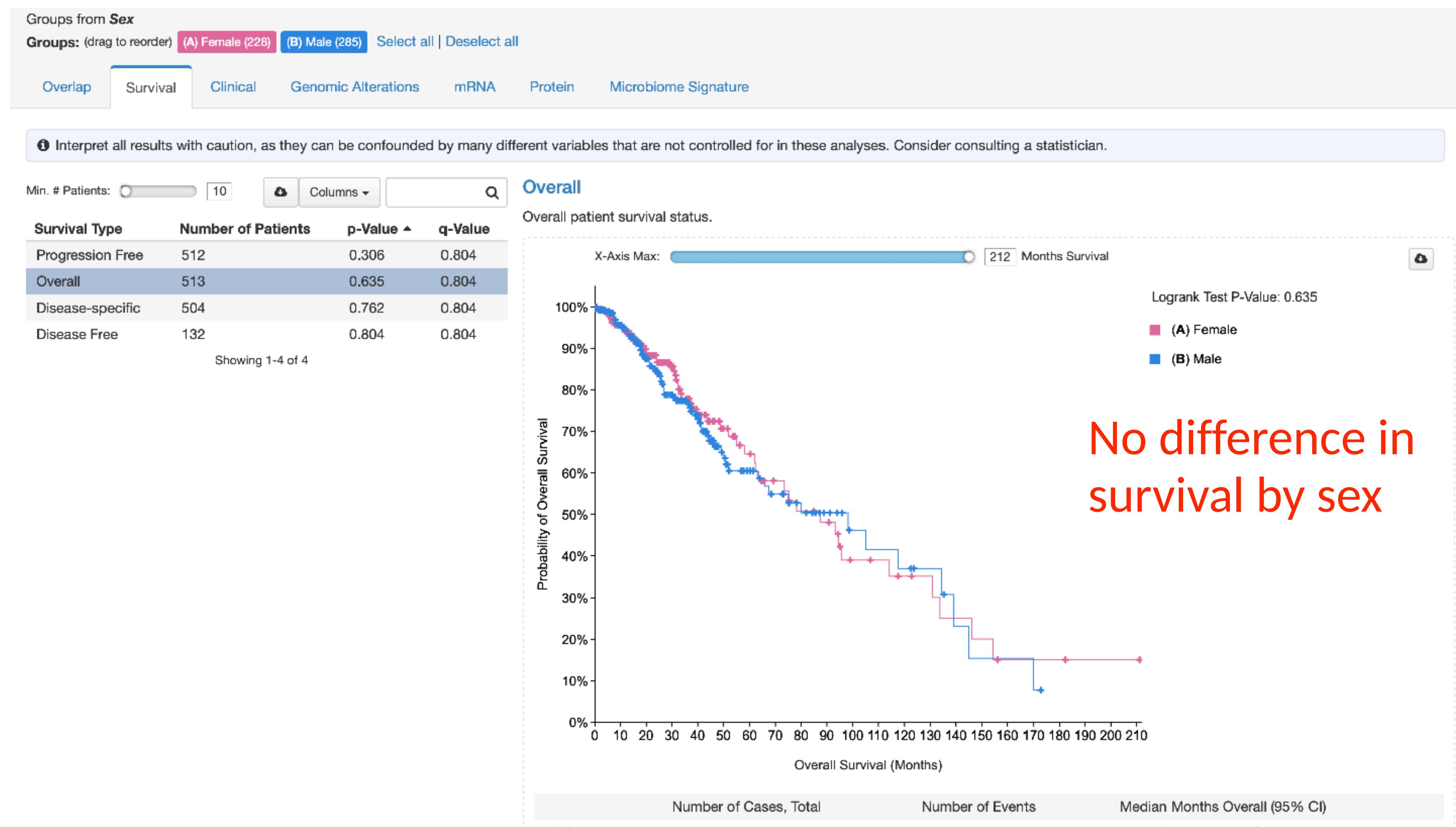


You can also select pre-existent clinical groups to perform comparisons

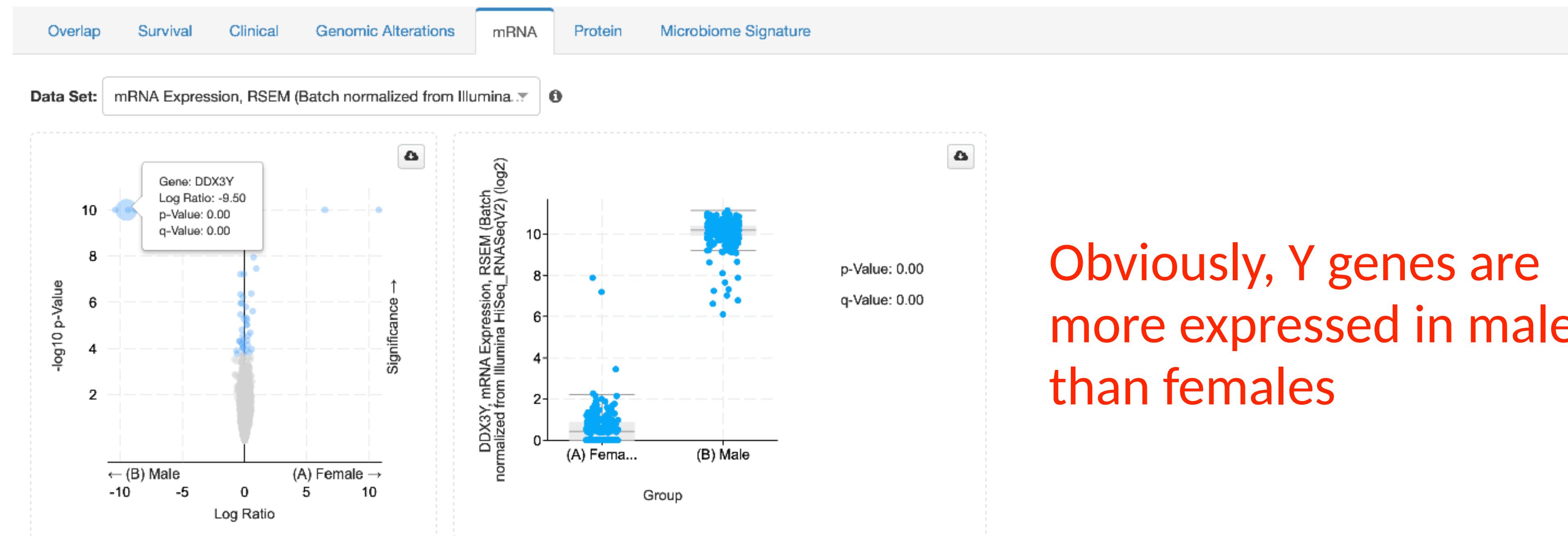
Query 1: Glioma



Query 1: Glioma



Query 1: Glioma



Obviously, Y genes are more expressed in males than females

mRNA Expression, RSEM (Batch normalized from Illumina HiSeq_RNASeqV2)

High expression in ... | ▾ Significant only

Columns ▾ Q

Gene	Cytoband	μ in (A) Female	μ in (B) Male	σ in (A) Female	σ in (B) Male	Log Ratio	p-Value	q-Value ▾	Higher expression in
DDX3Y	Yq11.221	0.57	10.07	0.88	0.67	-9.50	0.00	0.00	(B) Male
EIF1AY	Yq11.223	0.21	8.59	0.69	0.86	-8.38	0.00	0.00	(B) Male
KDM5D	Yq11.223	0.43	9.78	0.79	0.83	-9.35	0.00	0.00	(B) Male
TTY15	Yq11.221	0.15	7.88	0.55	0.80	-7.73	0.00	0.00	(B) Male
TXLNGY	Yq11.222-q11.223	0.18	7.28	0.50	0.78	-7.11	0.00	0.00	(B) Male
USP9Y	Yq11.221	0.32	9.11	0.66	0.72	-8.79	0.00	0.00	(B) Male
UTY	Yq11.221	0.17	8.15	0.54	0.76	-7.98	0.00	0.00	(B) Male

Query 1: Glioma



cBioPortal FOR CANCER GENOMICS

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Brain Lower Grade Glioma (TCGA, PanCancer Atlas) ↴
Brain Lower Grade Glioma TCGA PanCancer data. The original data is [here](#). The publications are [here](#). PubMed

Click gene symbols below or enter here Query

Selected: 514 patients | 514 samples Custom Selection ▾ Charts ▾ Groups ▾

Summary Clinical Data CN Segments

Cancer Type

	#	Freq
Diffuse Glioma	513	99.8%
Encapsulated Glioma	1	0.2%

Search...

Cancer Type Detailed

- Astrocytoma
- Oligodendrogloma
- Oligoastrocytoma
- Low-Grade Glioma (NOS)

Show Pie
Compare Groups
Download

Search... Select all

Genomic Profile Sample Counts

Molecular Profile	#	Freq
Fusions	514	100.0%
mRNA expression z-scores relativ...	514	100.0%
Mutations	514	100.0%
Putative arm-level copy-number fr...	514	100.0%
mRNA expression z-scores relativ...	514	100.0%
mRNA Expression, RSEM (Batch ...	514	100.0%
Microbiome Signatures (log RNA ...	513	99.8%
Log2 copy-number values	511	99.4%
Putative copy-number alterations ...	511	99.4%
Protein expression z-scores (RPPA)	428	83.3%
Protein expression (RPPA)	428	83.3%

Search...

KM Plot: Overall Survival (months)

Sex

228 285

Ethnicity Category

447

Race Category

447

KM Plot: Disease Free Survival (months)

KM Plot: Disease-specific Survival (months)

KM Plot: Progression Free Survival (months)

Overall Survival Status

388

Mutation Count vs Fraction Genome Altered

Mutation Count # samples 115 24 1

Pearson: -0.0048 p=0.91
Spearman: 0.5054 p=0.00

Mutated Genes (514 profiled samples)

Gene	# Mut	#	Freq
MUC17	23	16	3.1%
APOB	16	15	2.9%
BCOR	16	15	2.9%
LRP2	20	15	2.9%
ADGRV1	20	14	2.7%
TCF12	15	14	2.7%
ZNF292	17	13	2.5%
FAT2	23	13	2.5%
COL6A3	15	12	2.3%

Structural Variant Genes (514 profiled samples)

Gene	# SV	#	Freq
CLU	6	6	1.2%
SEPTIN14	6	6	1.2%
EGFR	5	5	1.0%
PDGFRA	6	5	1.0%
QKI	4	4	0.8%
KIF5A	4	4	0.8%
FGFR3	5	4	0.8%
ABR	4	4	0.8%
FIP1L1	4	3	0.6%

Query 1: Glioma



Brain Lower Grade Glioma (TCGA, PanCancer Atlas)

Groups from **Cancer Type Detailed**

Groups: (drag to reorder) (A) Astrocytoma (194) (B) Oligoastrocytoma (130) (C) Oligodendrogloma (189) Select all | Deselect all

Overlap Survival Clinical Genomic Alterations mRNA Protein Microbiome Signature

Alteration Types

Mutations

- Missense
- Inframe
 - Inframe Insertion
 - Inframe Deletion
- Truncating
 - Nonsense
 - Frameshift
 - Frameshift Insertion
 - Frameshift Deletion
- Nonstart
- Nonstop
- Splice
- Other

Structural Variants / Fusions

Copy Number Alterations

- Amplification
- Deletion

Select

Genomic Alterations

Genes with highest frequency in any group

Alteration event frequency (%)

Legend: (A) Astrocytoma, (B) Oligoastrocytoma, (C) Oligodendrogloma

Genomic Alterations

Gene	Cytoband	(A) Astrocytoma	(B) Oligoastrocytoma	(C) Oligodendrogloma	p-Value	q-Value	Most enriched in
CIC	19q13.2	6 (3.09%)	24 (16.46%)	85 (44.97%)	< 10 ⁻¹⁰	< 10 ⁻¹⁰	(C) Oligodendrogloma
TP53	17p13.1	129 (66.49%)	75 (57.69%)	44 (23.28%)	< 10 ⁻¹⁰	< 10 ⁻¹⁰	(A) Astrocytoma
ATRX	Xq21.1	105 (54.12%)	67 (51.54%)	33 (17.46%)	< 10 ⁻¹⁰	1.19e-10	(A) Astrocytoma
FUBP1	1p31.1	2 (1.03%)	9 (6.92%)	39 (20.63%)	3.76e-10	1.926e-6	(C) Oligodendrogloma
CDKN2B	9p21.3	42 (21.65%)	6 (4.62%)	8 (4.23%)	9.37e-9	3.201e-5	(A) Astrocytoma
CDKN2B-AS1	9p21.3	42 (21.65%)	6 (4.62%)	8 (4.23%)	9.37e-9	3.201e-5	(A) Astrocytoma
CDKN2A	9p21.3	42 (21.65%)	7 (5.38%)	8 (4.23%)	2.29e-8	6.703e-5	(A) Astrocytoma
CDKN2A-DT	9p21.3	38 (19.59%)	6 (4.62%)	7 (3.70%)	8.79e-8	2.251e-4	(A) Astrocytoma
MIR31HG	9p21.3	25 (12.89%)	3 (2.31%)	2 (1.06%)	7.19e-7	1.637e-3	(A) Astrocytoma

You can see the genes that have different genomic alterations between groups

We are going to search some of these genes: **IDH1**, **TP53** and **EGFR**

Query 1: Glioma



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Brain Lower Grade Glioma TCGA PanCancer data. The original data is [here](#). The publications are [here](#). PubMed

EGFR IDH1 TP53 Query

Selected: 514 patients | 514 samples

Summary Clinical Data CN Segments

Cancer Type

	#	Freq
Diffuse Glioma	513	99.8%
Encapsulated Glioma	1	0.2%

Search...

Cancer Type Detailed

	#	Freq
Astrocytoma	194	37.7%
Oligodendrogloma	189	36.8%
Oligoastrocytoma	130	25.3%
Low-Grade Glioma (NOS)	1	0.2%

Search...

Genomic Profile Sample Counts

Molecular Profile	#	Freq
Fusions	514	100.0%
mRNA expression z-scores relativ...	514	100.0%
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mRNA Expression, RSEM (Batch ...	514	100.0%
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Protein expression (RPPA)	428	83.3%

Search...

KM Plot: Overall Survival (months)

Sex

228 285

Mutation Count vs Fraction Genome Altered

samples 115 24 1

Pearson: -0.0048
p=0.91

Spearman: 0.5054

Mutated Genes (514 profiled samples)

Gene	# Mut	#	Freq
IDH1	395	395	76.8%
TP53	319	249	48.4%
ATRX	218	194	37.7%
CIC	130	108	21.0%
TTN	115	63	12.3%
FUBP1	51	48	9.3%
PIK3CA	46	42	8.2%
NOTCH1	49	38	7.4%

Structural Variant Genes (514 profiled samples)

Gene	# SV	#	Freq
CLU	6	6	1.2%
SEPTIN14	6	6	1.2%
EGFR	5	5	1.0%
PDGFRA	6	5	1.0%
QKI	4	4	0.8%
KIF5A	4	4	0.8%
FGFR3	5	4	0.8%
ABR	4	4	0.8%

Ethnicity Category

447

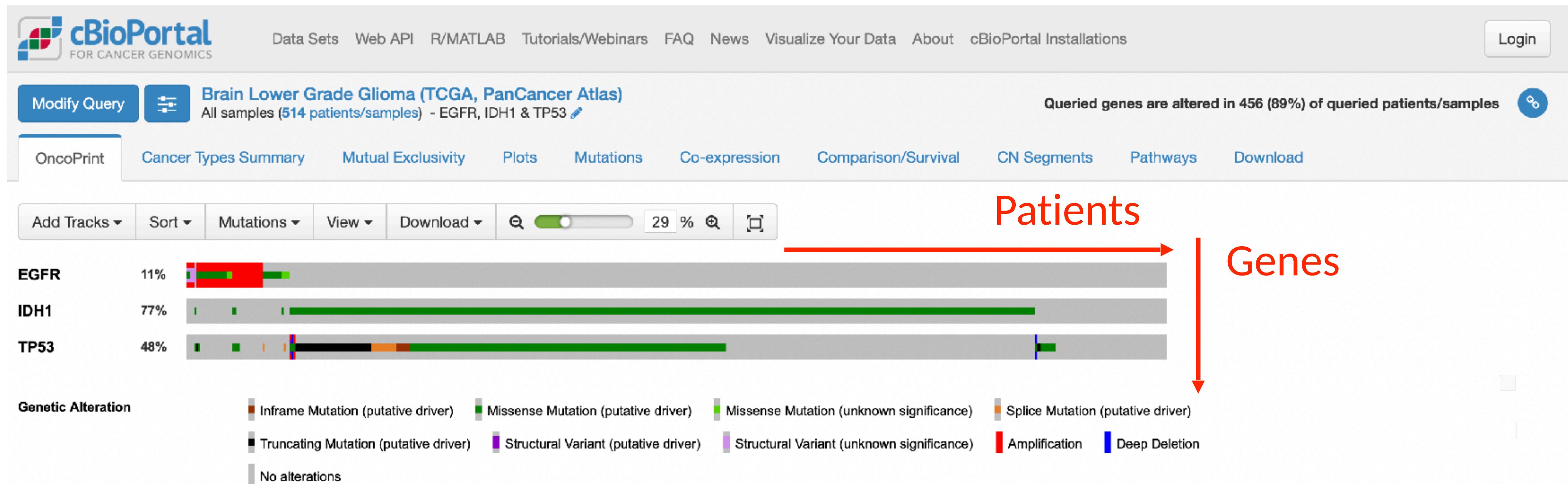
Race Category

447

Query genes
of interest

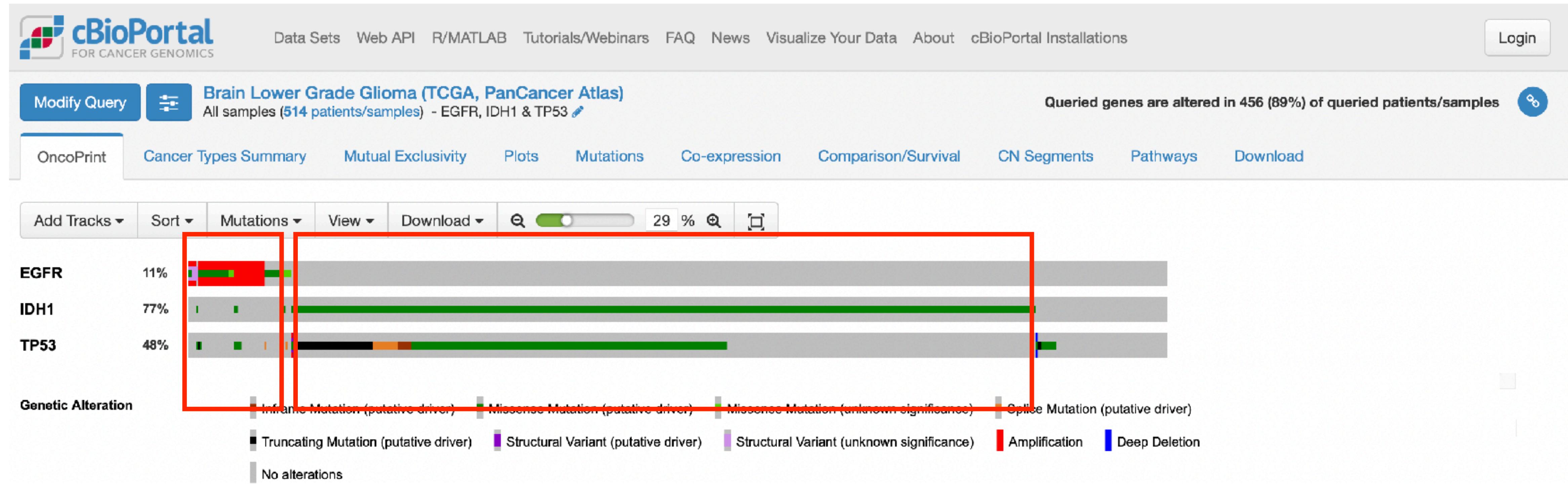
Query 1: Glioma

ONCOPRINT



You can plot this plot with your own data

Query 1: Glioma



You may have the intuition that *EGFR* and *IDH1/TP53* mutations are exclusive and *IDH1* and *TP53* mutations co-occur

Query 1: Glioma



cBioPortal FOR CANCER GENOMICS

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[Modify Query](#) **Brain Lower Grade Glioma (TCGA, PanCancer Atlas)**
All samples (514 patients/samples) - EGFR, IDH1 & TP53

[Queried genes are altered in 456 \(89%\) of queried patients/samples](#)

[OncoPrint](#) [Cancer Types Summary](#) [Mutual Exclusivity](#) [Plots](#) [Mutations](#) [Co-expression](#) [Comparison/Survival](#) [CN Segments](#) [Pathways](#) [Download](#)

The analysis tested 3 pairs between the 3 tracks in the OncoPrint.

Mutual exclusivity Co-occurrence Significant only

[Columns ▾](#)

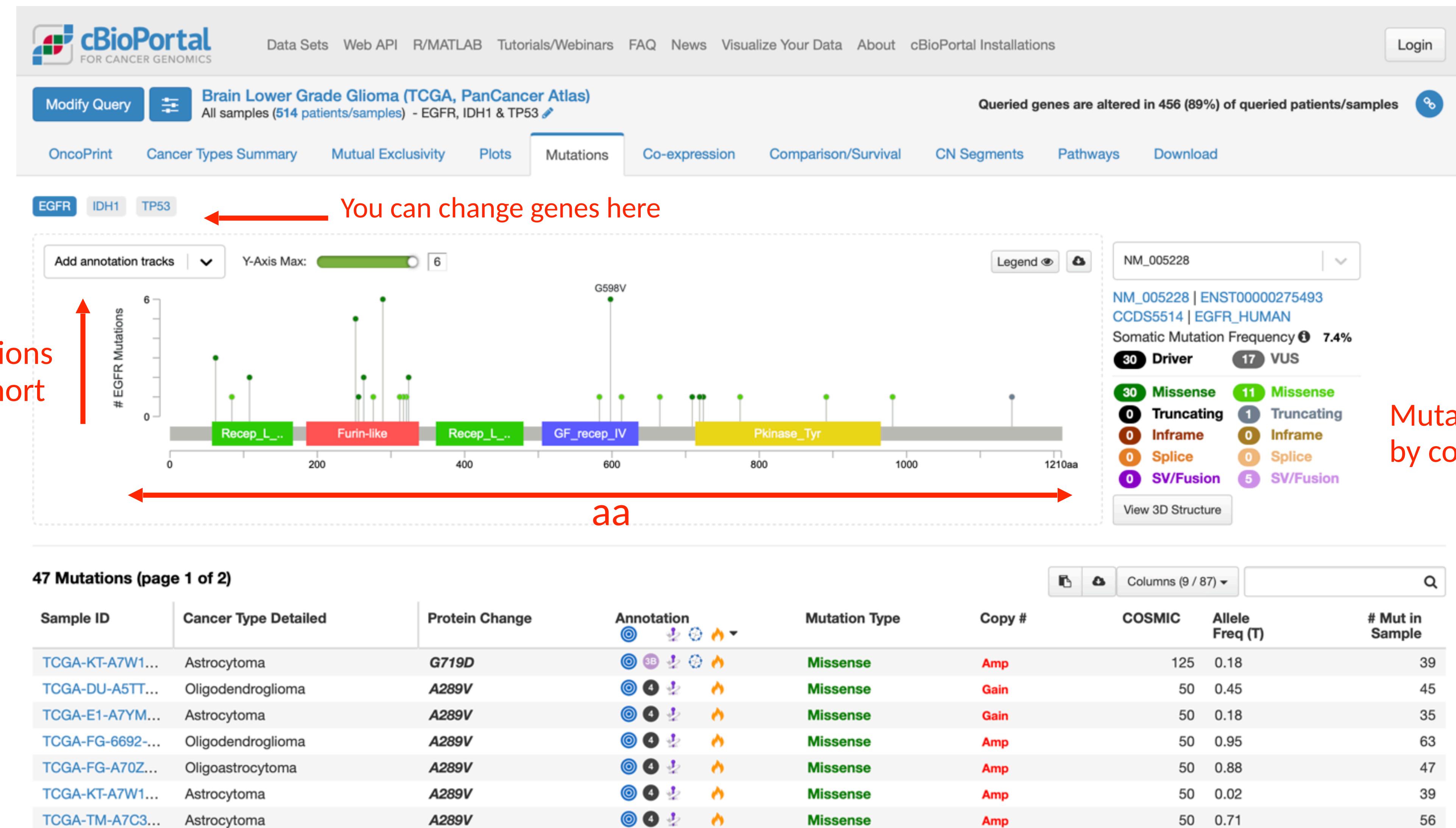
A	B	Neither	A Not B	B Not A	Both	Log2 Odds Ratio	p-Value	q-Value ▾	Tendency
EGFR	IDH1	67	50	390	4	<-3	<0.001	<0.001	Mutual exclusivity
IDH1	TP53	100	162	17	232	>3	<0.001	<0.001	Co-occurrence
EGFR	TP53	217	45	240	9	-2.467	<0.001	<0.001	Mutual exclusivity

Showing 1-3 of 3

Actually, you are right! All the mutual exclusivity results are significative

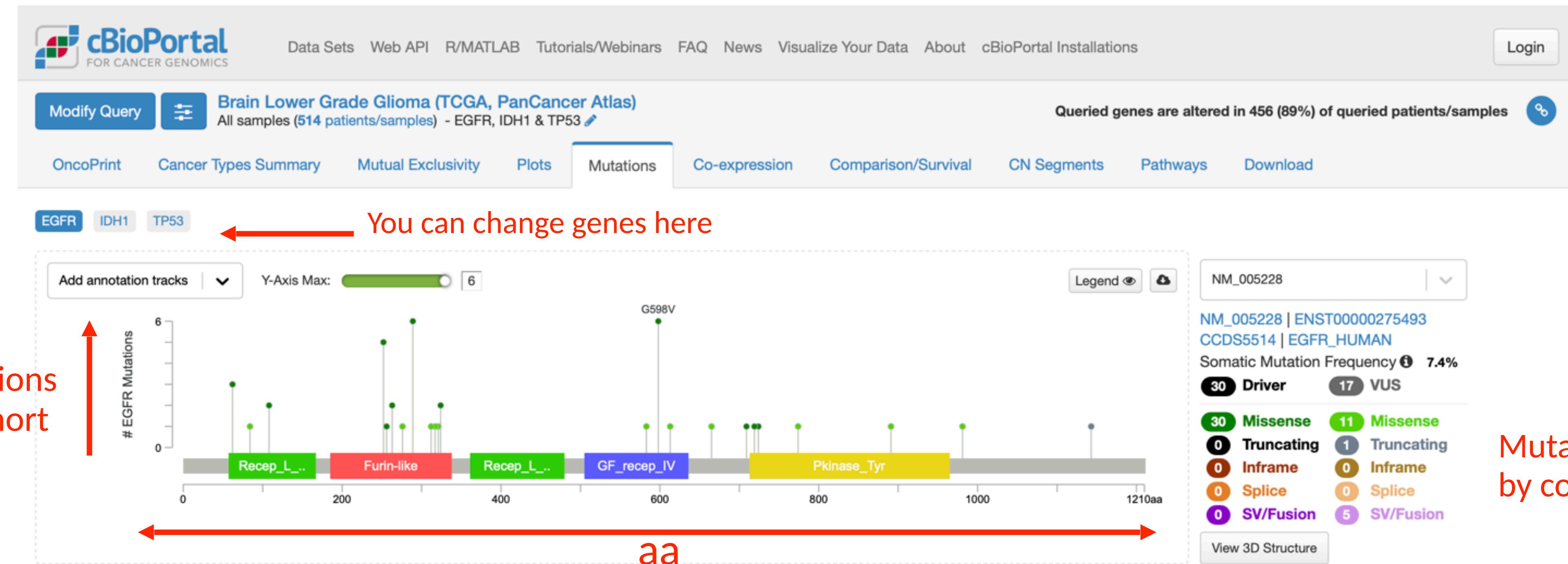
Query 1: Glioma

LOLLIPOP



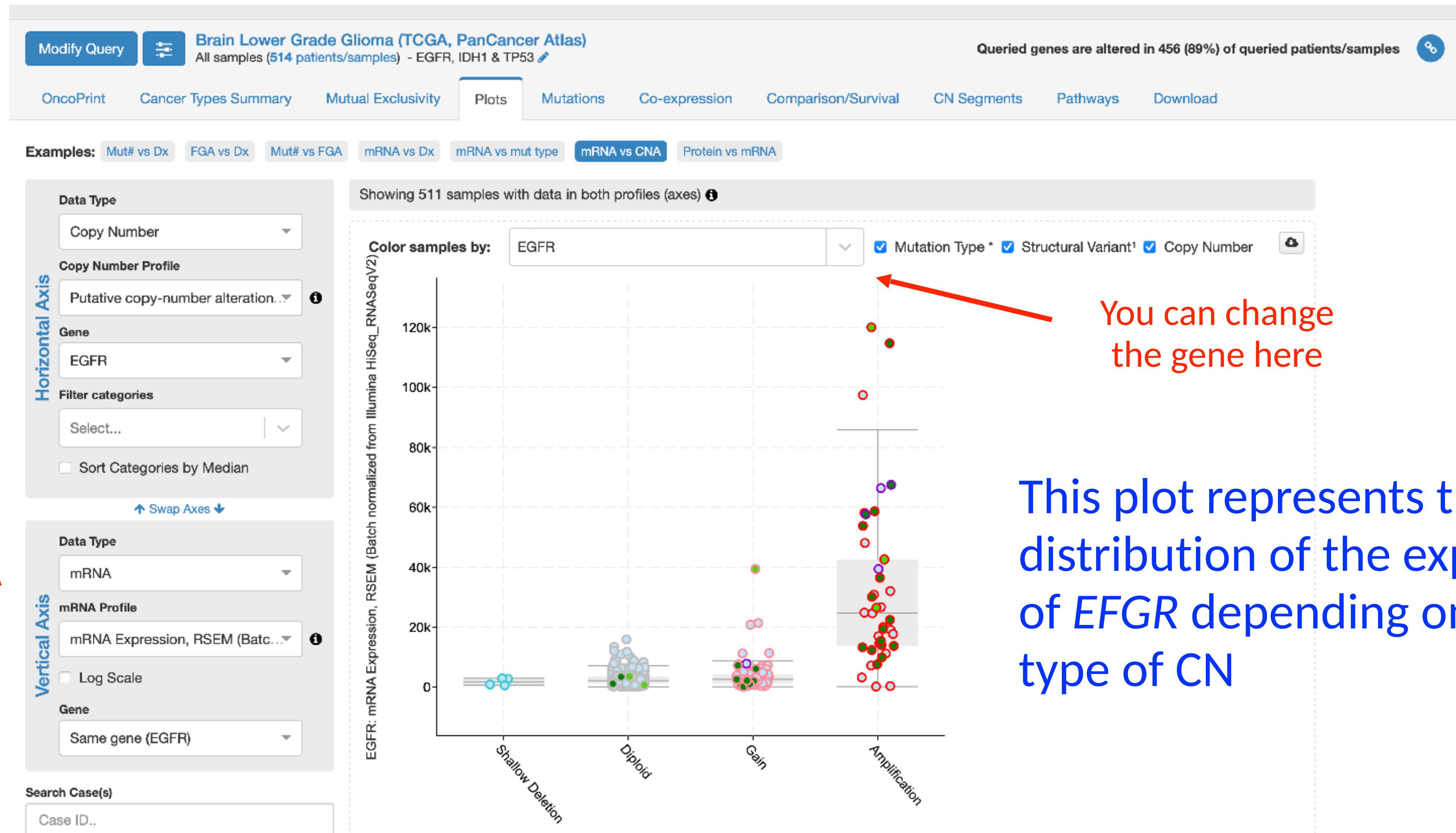
Query 1: Glioma

LOLLIPOP

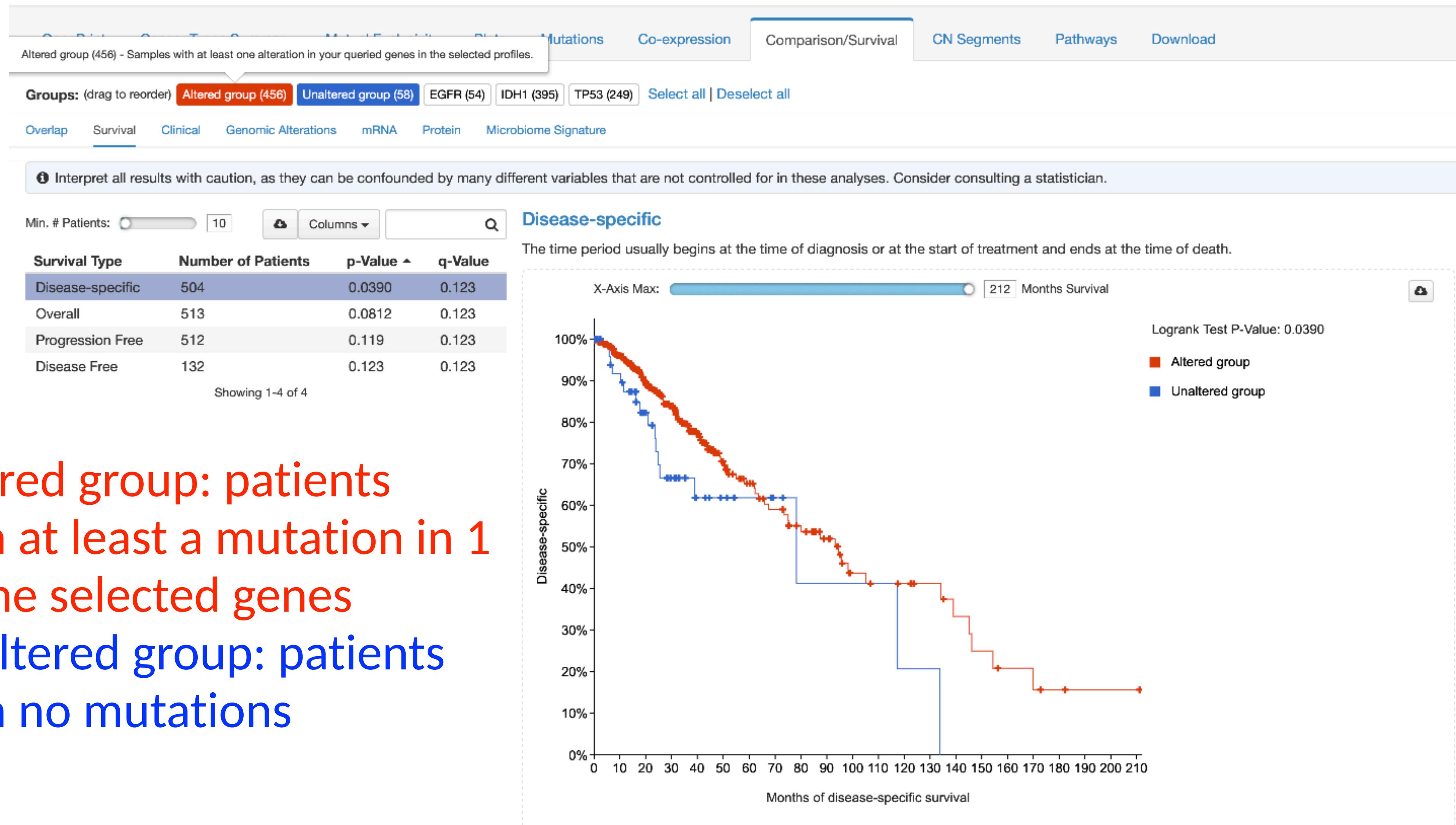


You can plot this plot with your own data

Query 1: Glioma



Query 1: Glioma



Altered group: patients with at least a mutation in 1 of the selected genes
Unaltered group: patients with no mutations

Query 1: Glioma



Comparisons/Survival > mRNA

mRNA Expression, RSEM (Batch normalized from Illumina HiSeq_RNASeqV2)

Add checked genes to query (none checked)

Gene	Cytoband	μ in Altered group	μ in Unaltered group	σ in Altered group	σ in Unaltered group	Log Ratio	p-Value	q-Value ▲	Higher expression in
<input type="checkbox"/> OR4N2	14q11.2	3.26	0.96	2.37	1.48	2.30	3.26e-17	3.27e-13	Altered group
<input type="checkbox"/> LINC00698	3p14.2	3.14	1.21	1.94	1.33	1.93	7.04e-16	4.70e-12	Altered group
<input type="checkbox"/> TSSK3	1p35.1	5.21	4.16	1.00	0.74	1.05	1.41e-15	7.08e-12	Altered group
<input type="checkbox"/> C10ORF120	10q26.13	0.47	0.10	0.74	0.22	0.37	9.67e-15	3.54e-11	Altered group
<input type="checkbox"/> DEFB119	20q11.21	1.31	0.16	2.24	0.71	1.15	1.06e-14	3.54e-11	Altered group
<input type="checkbox"/> ZNF560	19p13.2	4.27	1.72	2.60	1.95	2.55	4.73e-14	1.35e-10	Altered group
<input type="checkbox"/> RAB3D	19p13.2	7.55	8.46	0.91	0.72	-0.91	1.70e-13	4.24e-10	Unaltered group
<input type="checkbox"/> SSTR5	16p13.3	2.46	1.04	1.32	1.13	1.42	2.10e-13	4.29e-10	Altered group
<input type="checkbox"/> LINC00115	1p36.33	5.02	4.25	0.75	0.61	0.76	2.14e-13	4.29e-10	Altered group
<input type="checkbox"/> MTARC2	1q41	6.24	7.75	1.47	1.22	-1.52	3.36e-13	5.69e-10	Unaltered group
<input type="checkbox"/> TMEM213	7q34	1.10	0.49	0.81	0.47	0.60	3.41e-13	5.69e-10	Altered group
<input type="checkbox"/> KIAA1614	1q25.3	6.12	7.19	1.09	0.86	-1.07	3.94e-13	6.07e-10	Unaltered group
<input type="checkbox"/> TFDP3	Xq26.2	0.18	0.02	0.35	0.10	0.16	1.38e-12	1.97e-9	Altered group
<input type="checkbox"/> TOM1L1	17q22	4.25	6.79	1.90	2.16	-2.53	2.31e-12	3.09e-9	Unaltered group
<input type="checkbox"/> DLC1	8p22	9.12	10.23	0.83	0.95	-1.11	2.70e-12	3.37e-9	Unaltered group
<input type="checkbox"/> RBP1	3q23	7.34	9.59	1.61	1.94	-2.25	3.08e-12	3.63e-9	Unaltered group
<input type="checkbox"/> ZSCAN2	15q25.2	7.86	7.37	0.38	0.42	0.49	4.10e-12	4.55e-9	Altered group
<input type="checkbox"/> FBXO17	19q13.2	6.02	7.82	1.61	1.56	-1.80	5.01e-12	4.89e-9	Unaltered group
<input type="checkbox"/> RAB36	22q11.23	6.21	8.01	1.61	1.56	-1.80	5.02e-12	4.89e-9	Unaltered group
<input type="checkbox"/> MYOZ2	4q26	3.89	2.55	1.70	1.14	1.33	5.46e-12	4.89e-9	Altered group

Showing 1-20 of 19679

Show more

Query 1: Glioma



Comparisons/Survival > mRNA

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Brain Lower Grade Glioma (TCGA, PanCancer Atlas)
All samples (514 patients/samples) - EGFR, IDH1 & TP53

Queried genes are altered in 456 (89%) of queried patients/samples

Modify Query Comparison/Survival

OncoPrint Cancer Types Summary Mutual Exclusivity Plots Mutations Co-expression Comparison/Survival CN Segments Pathways Download

Groups: (drag to reorder) Altered group (456) Unaltered group (58) EGFR (54) IDH1 (395) TP53 (249) Select all | Deselect all

Overlap Survival Clinical Genomic Alterations mRNA Protein Microbiome Signature

Data Set: mRNA Expression, RSEM (Batch normalized from Illumina...

mRNA Expression, RSEM (Batch normalized from Illumina HiSeq_RNASeqV2) (log2)

-log10 p-Value

Significance →

← Unaltered group Altered group →

Log Ratio

OR4N2, mRNA Expression, RSEM (Batch normalized from Illumina HiSeq_RNASeqV2) (log2)

p-Value: 3.26e-17
q-Value: 3.27e-13

Group

mRNA Expression, RSEM (Batch normalized from Illumina HiSeq_RNASeqV2)

Add checked genes to query (none checked)

Gene	Cytoband	μ in Altered group	μ in Unaltered group	σ in Altered group	σ in Unaltered group	Log Ratio	p-Value	q-Value	Higher expression in
OR4N2	14q11.2	3.26	0.96	2.37	1.48	2.30	3.26e-17	3.27e-13	Altered group
LINC00698	3p14.2	3.14	1.21	1.94	1.33	1.93	7.04e-16	4.70e-12	Altered group
TSSK3	1p35.1	5.21	4.16	1.00	0.74	1.05	1.41e-15	7.08e-12	Altered group
C100RF120	10q26.13	0.47	0.10	0.74	0.22	0.37	9.67e-15	3.54e-11	Altered group
DEFB119	20q11.21	1.31	0.16	2.24	0.71	1.15	1.06e-14	3.54e-11	Altered group
ZNF560	19p13.2	4.27	1.72	2.60	1.95	2.55	4.73e-14	1.35e-10	Altered group
RAB3D	19p13.2	7.55	8.46	0.91	0.72	-0.91	1.70e-13	4.24e-10	Unaltered group
SSTR5	16p13.3	2.46	1.04	1.32	1.13	1.42	2.10e-13	4.29e-10	Altered group
LINC00115	1p36.33	5.02	4.25	0.75	0.61	0.76	2.14e-13	4.29e-10	Altered group
MTARC2	1q41	6.24	7.75	1.47	1.22	-1.52	3.36e-13	5.69e-10	Unaltered group
TMEM213	7q34	1.10	0.49	0.81	0.47	0.60	3.41e-13	5.69e-10	Altered group
KIAA1614	1q25.3	6.12	7.19	1.09	0.86	-1.07	3.94e-13	6.07e-10	Unaltered group
TFDP3	Xq26.2	0.18	0.02	0.35	0.10	0.16	1.38e-12	1.97e-9	Altered group
TOM1L1	17q22	4.25	6.79	1.90	2.16	-2.53	2.31e-12	3.09e-9	Unaltered group
DLC1	8p22	9.12	10.23	0.83	0.95	-1.11	2.70e-12	3.37e-9	Unaltered group
RBPI	3q23	7.34	9.59	1.61	1.94	-2.25	3.08e-12	3.63e-9	Unaltered group
ZSCAN2	15q25.2	7.86	7.37	0.38	0.42	0.49	4.10e-12	4.55e-9	Altered group
FBXO17	19q13.2	6.02	7.82	1.61	1.56	-1.80	5.01e-12	4.89e-9	Unaltered group
RAB36	22q11.23	6.21	8.01	1.61	1.56	-1.80	5.02e-12	4.89e-9	Unaltered group
MYOZ2	4q26	3.89	2.55	1.70	1.14	1.33	5.46e-12	4.89e-9	Altered group

Showing 1-20 of 19679 [Show more](#) [>](#)

mRNA Expression, RSEM (Batch normalized from Illumina HiSeq_RNASeqV2)

Add checked genes to query (none checked)

High expression in ... Significant only

Columns

Cross-cohort query using genes of interest

ERBB2

Query 2: Cross-cohort + genes



Select all TCGA studies

Please cite: Cerami et al., 2012 & Gao et al., 2013

1

Select Studies for Visualization & Analysis: 32 studies selected (10967 samples) Deselect all Search...

PanCancer Studies 8 Quick select: TCGA PanCancer Atlas Studies Curated set of non-redundant studies

PanCancer Studies	8	Quick select: TCGA PanCancer Atlas Studies Curated set of non-redundant studies
Pediatric Cancer Studies	13	
Immunogenomic Studies	8	
Cell lines	3	
Adrenal Gland	3	
Ampulla of Vater	1	
Biliary Tract	13	
Bladder/Urinary Tract	17	
Bone	2	
Bowel	12	
Breast	22	
CNS/Brain	20	
Cervix	2	
Esophagus/Stomach	17	
Eye	5	

PanCancer Studies

<input type="checkbox"/> MSK-IMPACT Clinical Sequencing Cohort (MSKCC, Nat Med 2017)	10945 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Metastatic Solid Cancers (UMich, Nature 2017)	500 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> MSS Mixed Solid Tumors (Broad/Dana-Farber, Nat Genet 2018)	249 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> SUMMIT - Neratinib Basket Study (Multi-Institute, Nature 2018)	141 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> TMB and Immunotherapy (MSKCC, Nat Genet 2019)	1661 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Tumors with TRK fusions (MSK, Clin Cancer Res 2020)	106 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Cancer Therapy and Clonal Hematopoiesis (MSK, Nat Genet 2020)	24146 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> China Pan-cancer (OriMed2020)	10194 samples	ⓘ ⓘ ⓘ

Pediatric Cancer Studies

<input type="checkbox"/> Pediatric Preclinical Testing Consortium (CHOP, Cell Rep 2019)	261 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Pediatric Acute Lymphoid Leukemia - Phase II (TARGET, 2018)	1978 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Pediatric Rhabdoid Tumor (TARGET, 2018)	72 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Pediatric Wilms' Tumor (TARGET, 2018)	657 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Pediatric Acute Myeloid Leukemia (TARGET, 2018)	1025 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Pediatric Neuroblastoma (TARGET, 2018)	1089 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Pediatric Pan-Cancer (DKFZ, Nature 2017)	961 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Pediatric Pan-cancer (Columbia U, Genome Med 2016)	103 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Acute Lymphoblastic Leukemia (St Jude, Nat Genet 2016)	73 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Acute Lymphoblastic Leukemia (St Jude, Nat Genet 2015)	93 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Pediatric Ewing Sarcoma (DFCI, Cancer Discov 2014)	107 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Ewing Sarcoma (Institut Curie, Cancer Discov 2014)	112 samples	ⓘ ⓘ ⓘ
<input type="checkbox"/> Medulloblastoma (PCGP, Nature 2012)	37 samples	ⓘ ⓘ ⓘ

32 studies selected (10967 samples) Deselect all

Query By Gene OR Explore Selected Studies

2 Query by gene

What's New @cbiportal

cBioPortal Retweeted The Hyve @TheHyveNL Adding single-cell data and visualisations to #cBioPortal would reduce the time spent finding, downloading, and processing data for both users new to #singlecell data and users with expertise in single-cell technologies. ow.ly/OyaE50Gb3JT #cancerresearch #genomics

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Example Queries

- Primary vs. metastatic prostate cancer
- RAS/RAF alterations in colorectal cancer
- BRCA1 and BRCA2 mutations in ovarian cancer
- POLE hotspot mutations in endometrial cancer
- TP53 and MDM2/4 alterations in GBM
- PTEN mutations in GBM in text format
- Patient view of an endometrial cancer case
- All TCGA Pan-Cancer
- MSK-IMPACT clinical cohort, Zehir et al. 2017
- Histone mutations across cancer types

Local Installations Host your own



Query 2: Cross-cohort + genes



Query Quick Search Beta! Download Please cite: Cerami et al., 2012 & Gao et al., 2013

Selected Studies: [Modify](#)
Acute Myeloid Leukemia (TCGA, PanCancer Atlas) | Adrenocortical Carcinoma (TCGA, PanCancer Atlas)
Bladder Urothelial Carcinoma (TCGA, PanCancer Atlas) | Brain Lower Grade Glioma (TCGA, PanCancer Atlas) | and 28 more (10967 total samples)

Select Molecular Profiles:
 Mutations Structural variants Copy number alterations

Select Patient/Case Set:
To build your own case set, try out our enhanced Study View.
All (10967)

Enter Genes:
Hint: Learn Onco Query Language (OQL) to write more powerful queries
erbB2
All gene symbols are valid.

Submit Query

What's New @cbioportal

cBioPortal Retweeted
The Hyve @TheHyveNL
Adding single-cell data and visualisations to #cBioPortal would reduce the time spent finding, downloading, and processing data for both users new to #singlecell data and users with expertise in single-cell technologies.
ow.ly/OyaE50Gb3jT #cancerresearch #genomics

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Example Queries

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- RAS/RAF alterations in colorectal cancer
- BRCA1 and BRCA2 mutations in ovarian cancer
- POLE hotspot mutations in endometrial cancer
- TP53 and MDM2/4 alterations in GBM
- PTEN mutations in GBM in text format
- Patient view of an endometrial cancer case
- All TCGA Pan-Cancer
- MSK-IMPACT clinical cohort, Zehir et al. 2017
- Histone mutations across cancer types

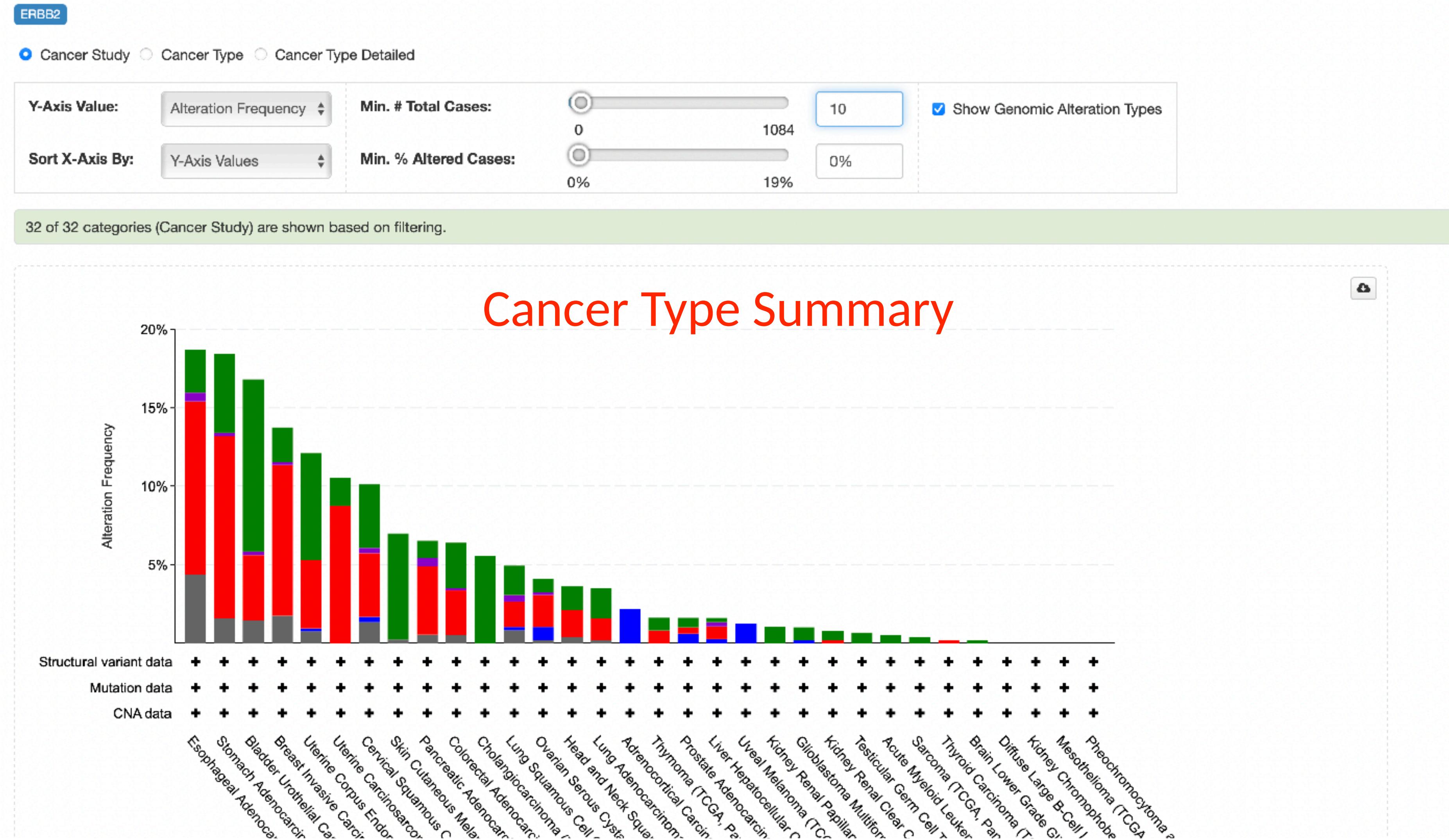
Local Installations Host your own

Enter the genes of interest
ERBB2

Query 2: Cross-cohort + genes



Query 2: Cross-cohort + genes



Individual tumor samples

LUAD-2GUGK

Query 3: Individual samples



Go to Quick Search tab and introduce the name of the sample

LUAD-2GUGK

The screenshot shows the cBioPortal homepage with the search bar containing 'LUAD-2G'. Below the search bar, there are two tabs: 'Quick Search Beta!' (highlighted with a red arrow) and 'Download'. The main content area displays search results for 'LUAD-2G'. It includes a header 'Click on a patient to see a summary' and a list of results:

- PATIENT** LUAD-2GUGK (Lung Adenocarcinoma (Broad, Cell 2012))
- PATIENT** LUAD-2GUGK (Pan-Lung Cancer (TCGA, Nat Genet 2016))
- SAMPLE** LUAD-2GUGK (Lung Adenocarcinoma (Broad, Cell 2012))
- SAMPLE** LUAD-2GUGK-Tumor (Pan-Lung Cancer (TCGA, Nat Genet 2016))

A message at the top right says 'Please cite: Cerami et al., 2012 & Gao et al., 2013'. On the right side, there's a 'What's New' section with a tweet from 'The Hyve' (@TheHyveNL) about adding single-cell data and visualizations, and a 'Example Queries' section with a list of various cancer-related queries.

Query 3: Individual samples



Patient Summary → Clinical data

238 Mutations (page 1 of 24) Mutation information

Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Copy #	Cohort	COSMIC
HRAS	G13R	① ③B ④ ⑤ ⑥	Missense	0.17	Gain	<1%	365
ITK	L106I	○	Missense	0.27	Diploid	2%	
TRIM33	R655Q	○	Missense	0.06	Diploid	2%	1
PTPRC	S1173Y	○	Missense	0.16	Diploid	7%	
DDX5	L196V	○	Missense	0.11	Diploid	<1%	1
STRN	X613_splice	○	Splice	0.11	Diploid	2%	
GABRA6	Y146H	○	Missense	0.14	Diploid	5%	1
BCL9	D437H	○	Missense	0.06	Diploid	2%	
PGBD5	A51G	○	Missense	0.12	Diploid	1%	
NUP98	N314Y	○	Missense	0.23	Gain	3%	1

Showing 1-10 of 238 Mutations < Show more >

CNA

MUT

1.7% 238

relative count

variant allele frequency

Query 3: Individual samples



Gene	Protein	Annotation	Mutation Class	Frequency	Diploid	Cohort	Count
PTPRC	S1173Y	○	Missense	0.00	Diploid	7%	1
DDX5	L196V	○	Missense	0.16	Diploid	<1%	1
STRN	X613_splice	○	Splice	0.11	Diploid	2%	1
GABRA6	Y146H	○	Missense	0.11	Diploid	5%	1
BCL9	D437H	○	Missense	0.14	Diploid	2%	1
PGBD5	A51G	○	Missense	0.06	Diploid	1%	1
NUP98	N314Y	○	Missense	0.12	Diploid	3%	1
				0.23	Gain		

Showing 1-10 of 238 Mutations Show more

0 Structural Variants (page 1 of 1)					
Gene 1	Gene 2	Annotation	Variant Class	Event Info	Connection Type
There are no results.					

Showing 0-0 of 0 Structural Variants

58 Copy Number Alterations (page 1 of 6) CNV information					
Gene	CNA	Annotation	Cytoband	Cohort	
ANKRD11	DeepDel	○	16q24.3	1%	
FANCA	DeepDel	○	16q24.3	1%	
CBFA2T3	DeepDel	○	16q24.3	1%	
SLC37A2	DeepDel	○	11q24.2	1%	
SACS	DeepDel	○	13q12.12	<1%	
JPH3	DeepDel	○	16q24.2	1%	
KLHDC4	DeepDel	○	16q24.2	1%	
SLC7A5	DeepDel	○	16q24.2	1%	
CA5A	DeepDel	○	16q24.2	1%	
BANP	DeepDel	○	16q24.2	1%	

Showing 1-10 of 58 Copy Number Alterations Show more

Exercise



1. Select Cancer Study:

Glioblastoma — Glioblastoma (TCGA, Cell 2013)

2. Select Genomic Profiles:

Mutations

Putative copy-number alterations from GISTIC

mRNA Expression z-Scores (threshold: 2.0)

3. Select Patient/Case Set:

Complete Samples (141)

4. Enter Gene Set:

IDH1 CDK4 TP53 CDKN2A EGFR RB1

Questions



- What gene is the most frequently deleted?
- Can you identify significant mutual exclusivity or co-occurrent alterations with *CDK4*?
- Which is the most frequent point mutation in *EGFR*?
- Is this group of genes a molecular marker of overall survival?
- Search the patient TCGA-06-0650. Based on her molecular alterations, which treatment they might receive?

Further Info



**Five detailed tutorials (total time 5h) at [www.cbioportal.org/
tutorials](http://www.cbioportal.org/tutorials)**