

PO21: 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)

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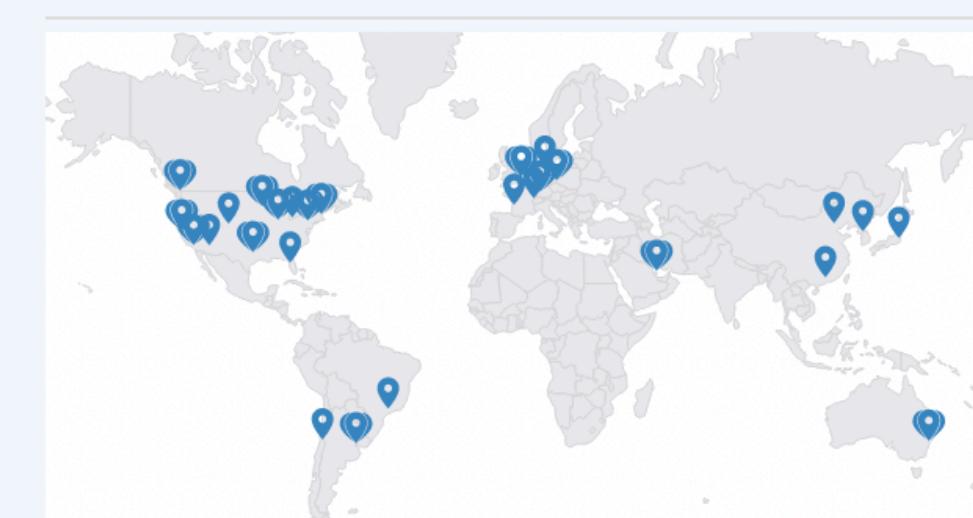
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- Primary vs. metastatic prostate cancer
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Affected Tissue

0 studies selected (0 samples)

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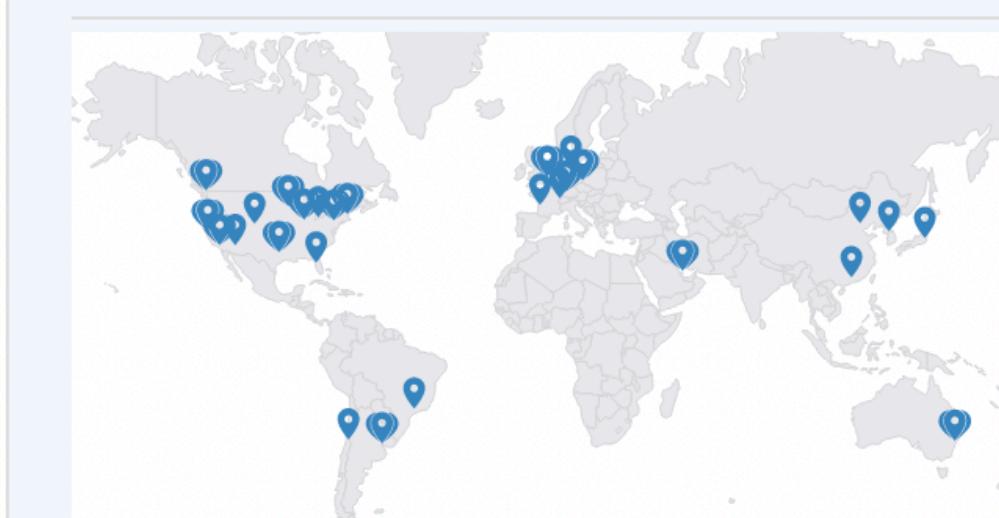
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samples, info and link to publication
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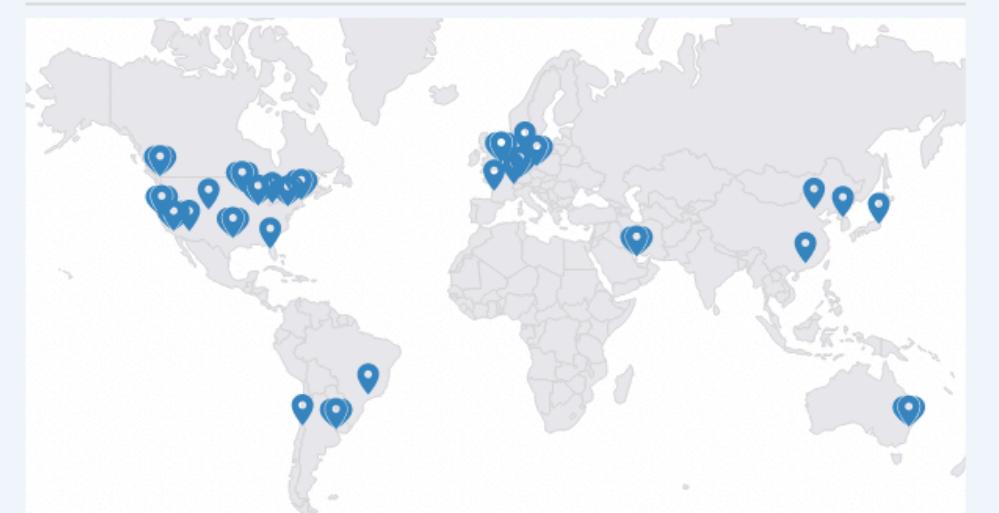
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Brief description with
the type of sequencing:
targeted, WES, WGS

Whole exome sequencing of 22 phyllodes tumors

22 samples   

0 studies selected (0 samples)

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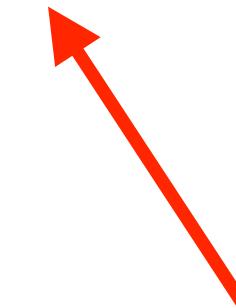
Query

Quick Search **Beta!**

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

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



We would love to hear what you think: cboportal@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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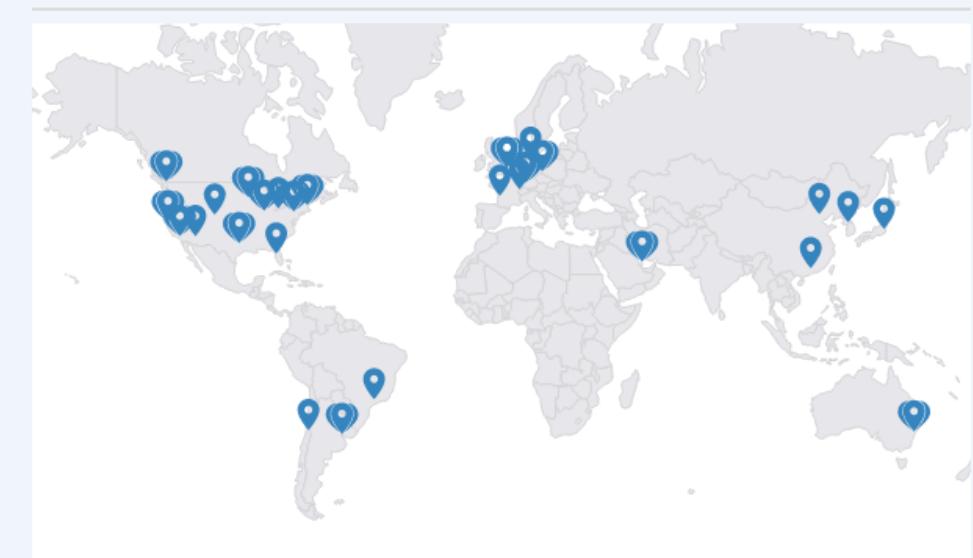
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Types of queries



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...

Select all listed studies matching filter (20)

CNS/Brain

Diffuse Glioma

Study Type	Number of Samples	Action Buttons
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 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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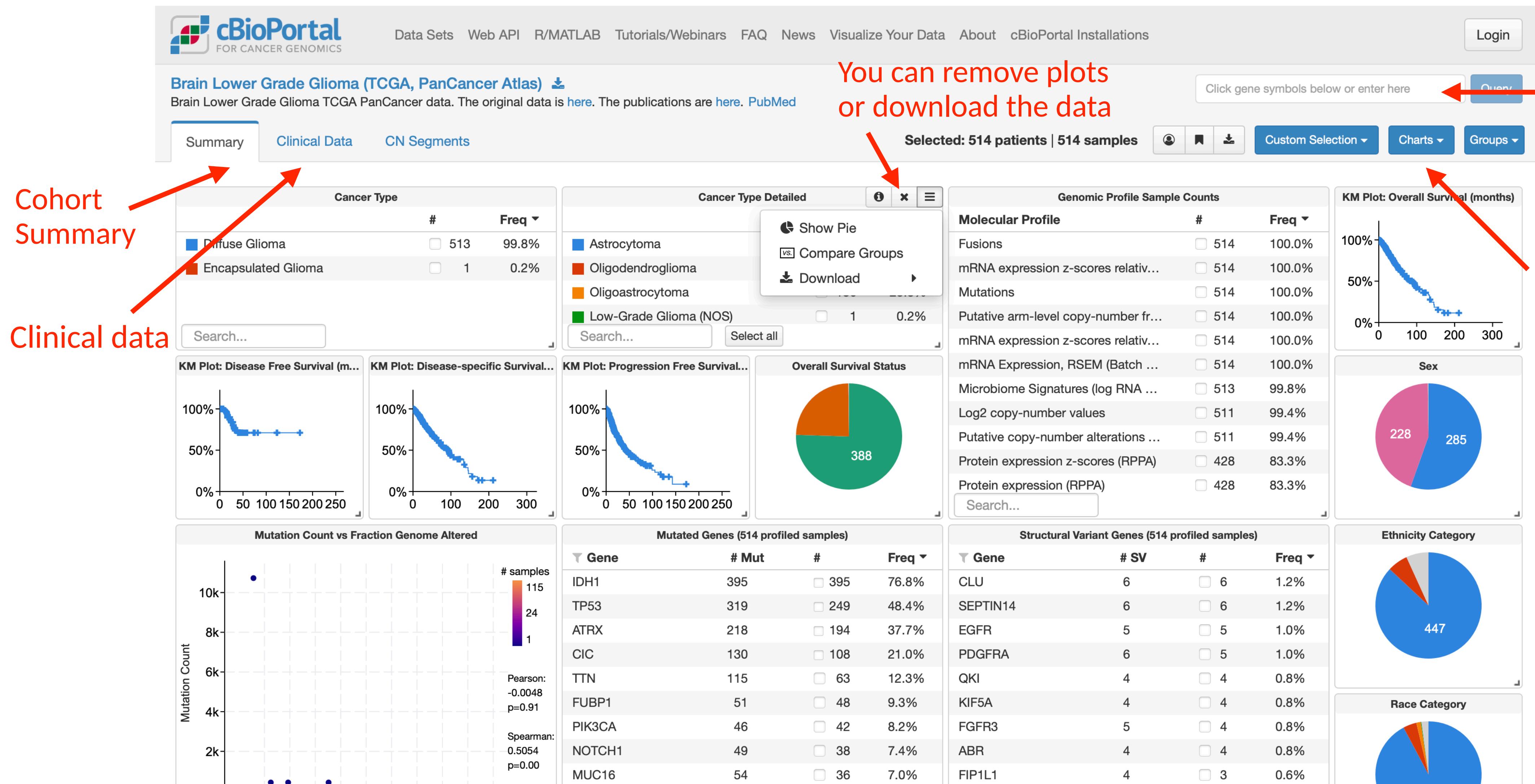
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Query 1: Glioma

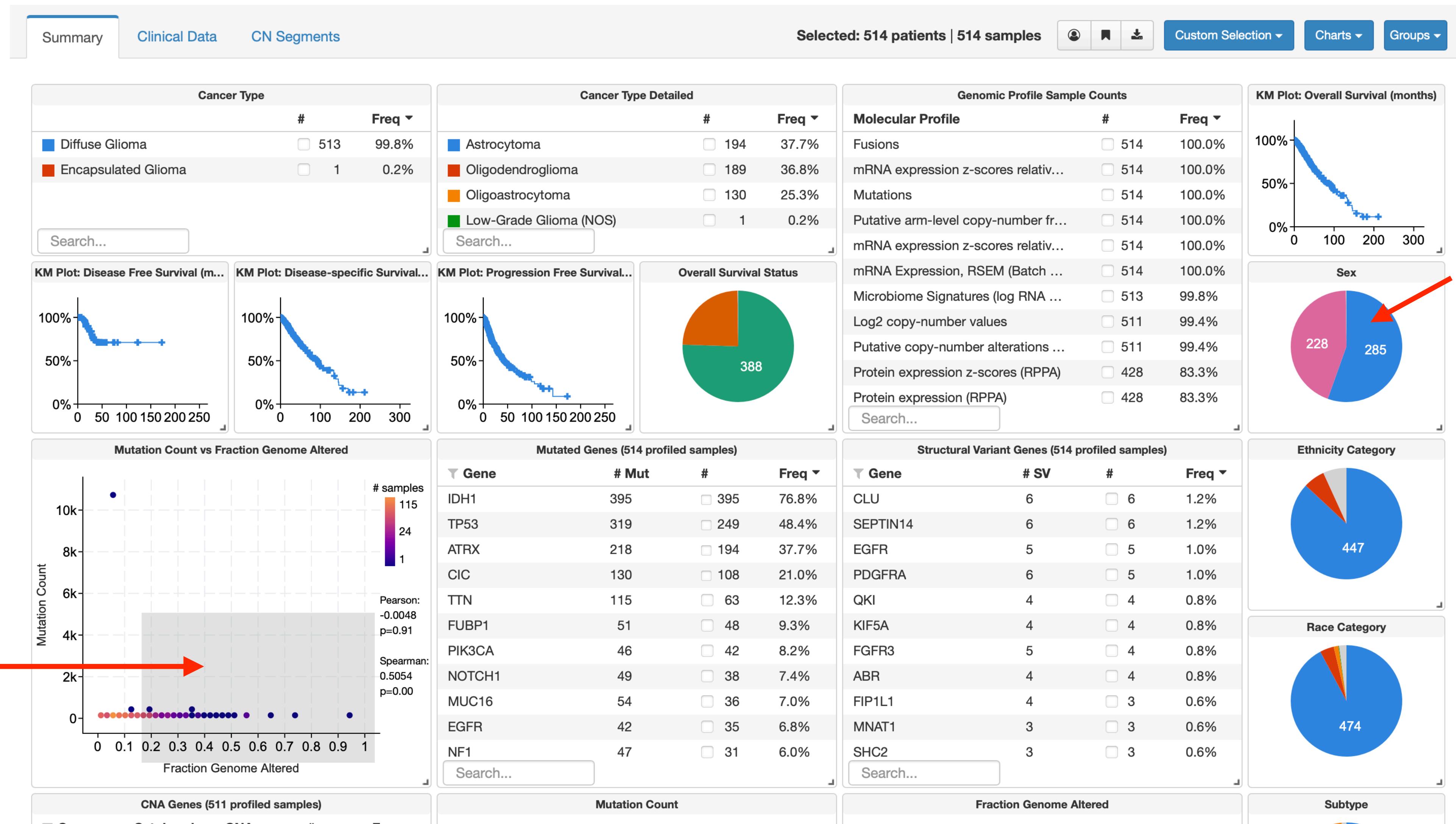


You can rearrange the plots by selecting and dragging

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%

KM Plot: Disease Free Survival (m...)
No data to plot.

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

KM Plot: Progression Free Survival...

Genomic Profile Sample Counts		
Molecular Profile	#	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%

Overall Survival Status

KM Plot: Overall Survival (months)		
Sex	11	17

Mutation Count vs Fraction Genome Altered

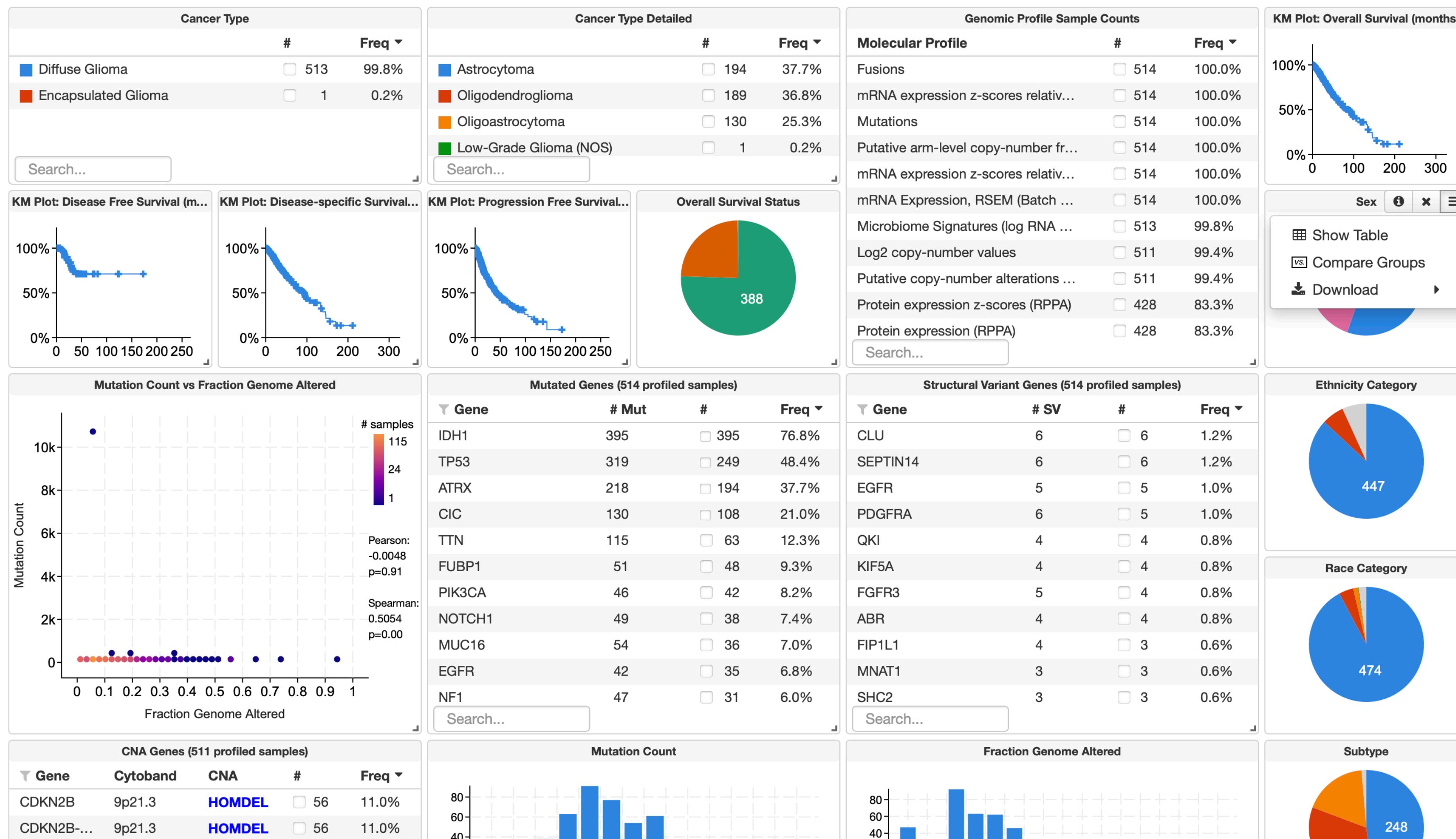
Mutated Genes (11 profiled samples)			
Gene	# Mut	#	Freq ▾
TP53	9	8	72.7%
IDH1	6	6	54.5%
KRT86	2	2	18.2%
FLG	2	2	18.2%
SULT2B1	2	2	18.2%
PTPRH	2	2	18.2%
PTPRM	2	2	18.2%

Structural Variant Genes (11 profiled samples)			
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%

Ethnicity Category

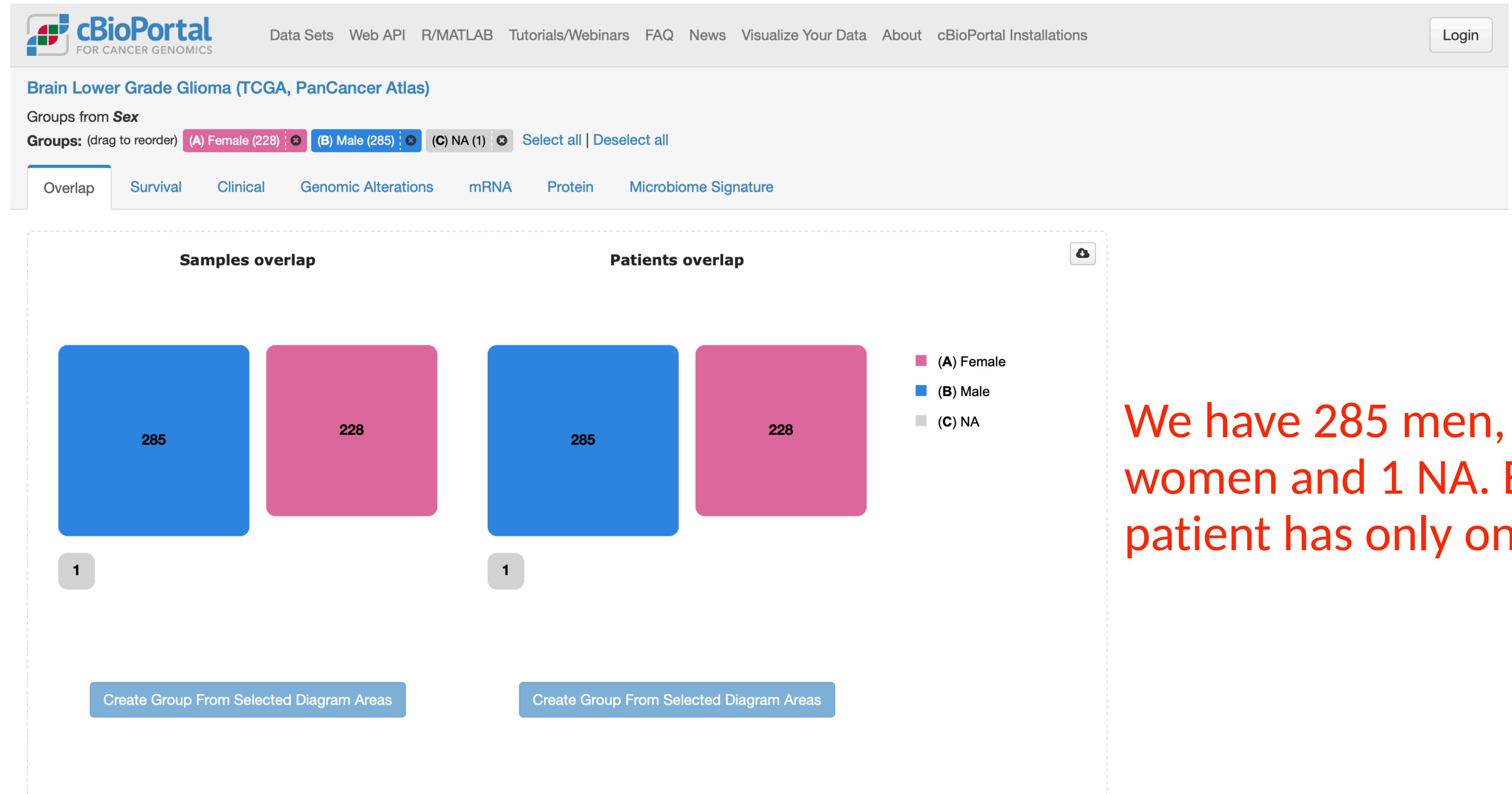
Race Category

Query 1: Glioma

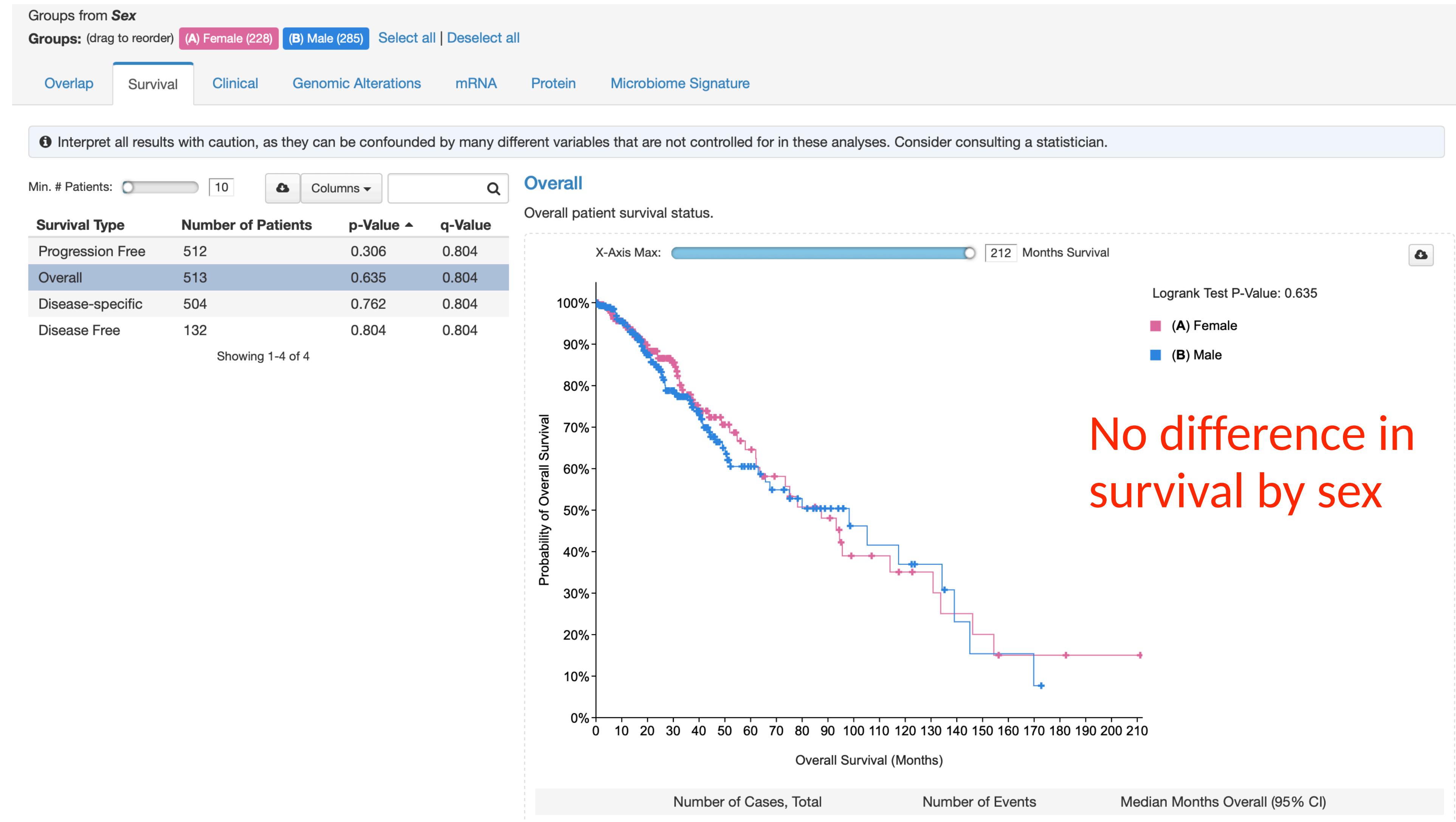


You can also select pre-existent clinical groups to perform comparisons

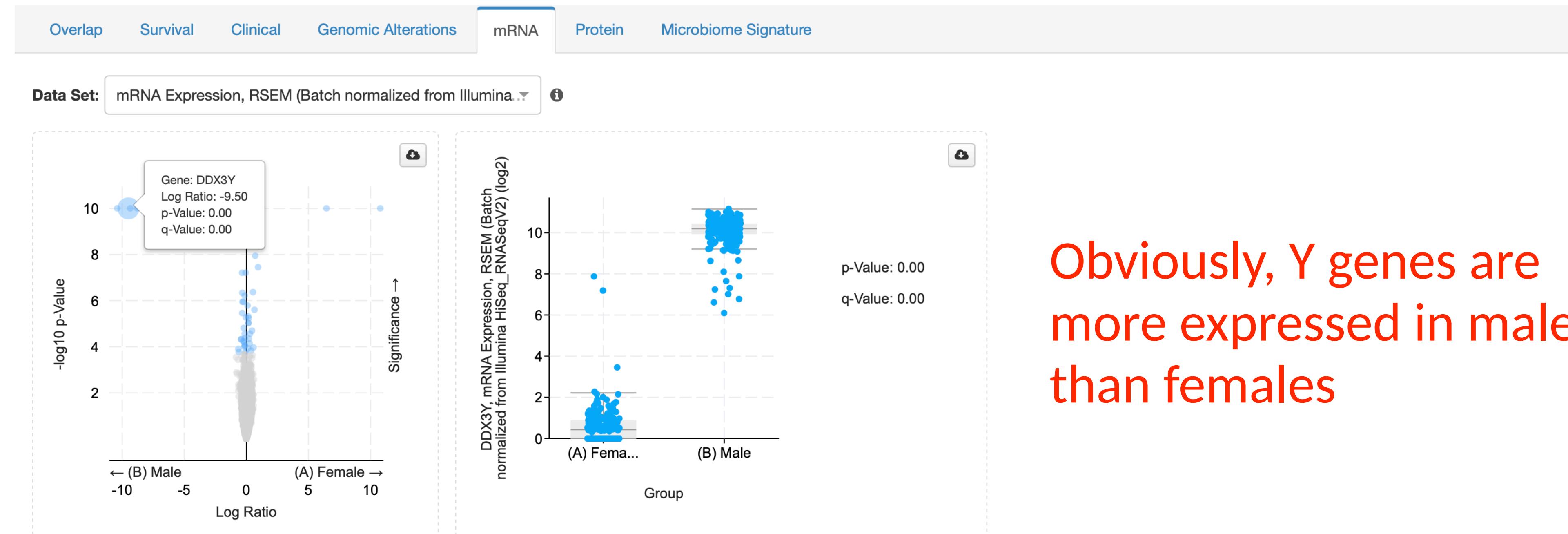
Query 1: Glioma



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



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

Summary Clinical Data CN Segments Selected: 514 patients | 514 samples [User, Bookmarks, Download] Custom Selection ▾ Charts ▾ Groups ▾

Cancer Type Detailed

- Show Pie
- vs Compare Groups
- Download

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

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%

Genomic Profile Sample Counts	
Overall Survival Status	388

KM Plot: Overall Survival (months)

Sex

Ethnicity Category

Mutation Count vs Fraction Genome Altered

Mutated Genes (514 profiled samples)																																								
<table border="1"><thead><tr><th>Gene</th><th># Mut</th><th>#</th><th>Freq</th></tr></thead><tbody><tr><td>MUC17</td><td>23</td><td>16</td><td>3.1%</td></tr><tr><td>APOB</td><td>16</td><td>15</td><td>2.9%</td></tr><tr><td>BCOR</td><td>16</td><td>15</td><td>2.9%</td></tr><tr><td>LRP2</td><td>20</td><td>15</td><td>2.9%</td></tr><tr><td>ADGRV1</td><td>20</td><td>14</td><td>2.7%</td></tr><tr><td>TCF12</td><td>15</td><td>14</td><td>2.7%</td></tr><tr><td>ZNF292</td><td>17</td><td>13</td><td>2.5%</td></tr><tr><td>FAT2</td><td>23</td><td>13</td><td>2.5%</td></tr><tr><td>COL6A3</td><td>15</td><td>12</td><td>2.3%</td></tr></tbody></table>	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%
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COL6A3	15	12	2.3%																																					

Structural Variant Genes (514 profiled samples)																																								
<table border="1"><thead><tr><th>Gene</th><th># SV</th><th>#</th><th>Freq</th></tr></thead><tbody><tr><td>CLU</td><td>6</td><td>6</td><td>1.2%</td></tr><tr><td>SEPTIN14</td><td>6</td><td>6</td><td>1.2%</td></tr><tr><td>EGFR</td><td>5</td><td>5</td><td>1.0%</td></tr><tr><td>PDGFRA</td><td>6</td><td>5</td><td>1.0%</td></tr><tr><td>QKI</td><td>4</td><td>4</td><td>0.8%</td></tr><tr><td>KIF5A</td><td>4</td><td>4</td><td>0.8%</td></tr><tr><td>FGFR3</td><td>5</td><td>4</td><td>0.8%</td></tr><tr><td>ABR</td><td>4</td><td>4</td><td>0.8%</td></tr><tr><td>FIP1L1</td><td>4</td><td>3</td><td>0.6%</td></tr></tbody></table>	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%
Gene	# SV	#	Freq																																					
CLU	6	6	1.2%																																					
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FGFR3	5	4	0.8%																																					
ABR	4	4	0.8%																																					
FIP1L1	4	3	0.6%																																					

Race Category

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 (blue), (B) Oligoastrocytoma (orange), (C) Oligodendrogloma (red)

Genomic Alterations

Gene	Cytoband	(A) Astrocytoma	(B) Oligoastrocytoma	(C) Oligodendrogloma	p-Value	q-Value	Most enriched in
CIC	19q13.2	6 (3.09%)	24 (18.46%)	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



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

EGFR IDH1 TP53 Query

Selected: 514 patients | 514 samples

Clinical Data CN Segments

Summary

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%
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

Mutation Count vs Fraction Genome Altered

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%

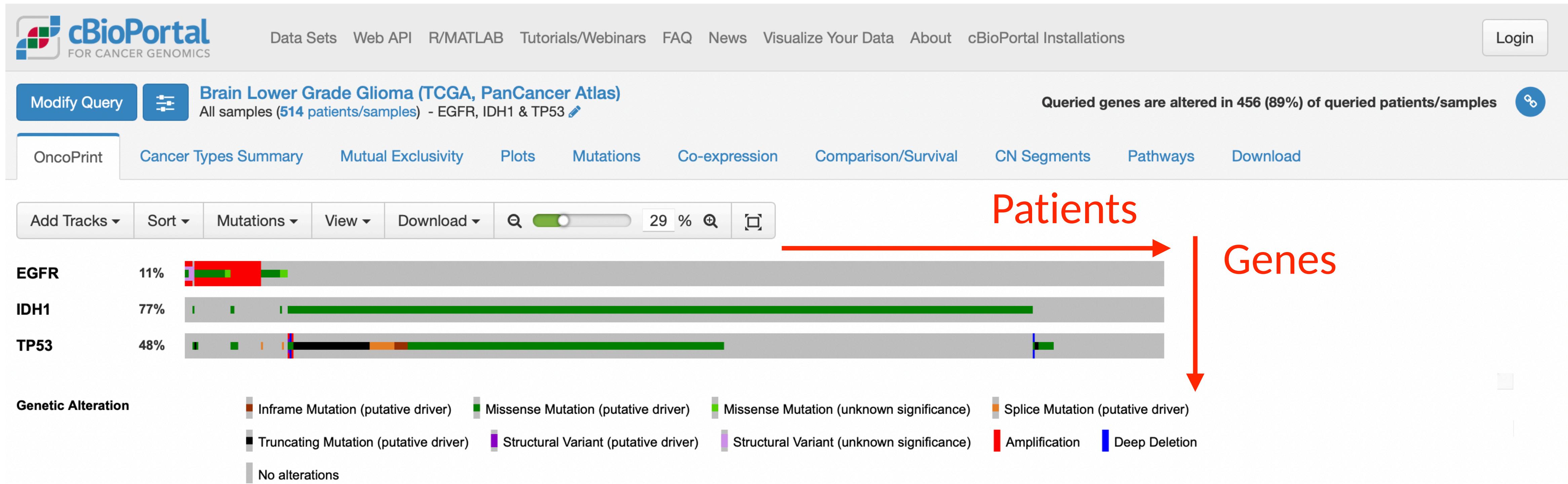
Ethnicity Category

Race Category

Query genes
of interest

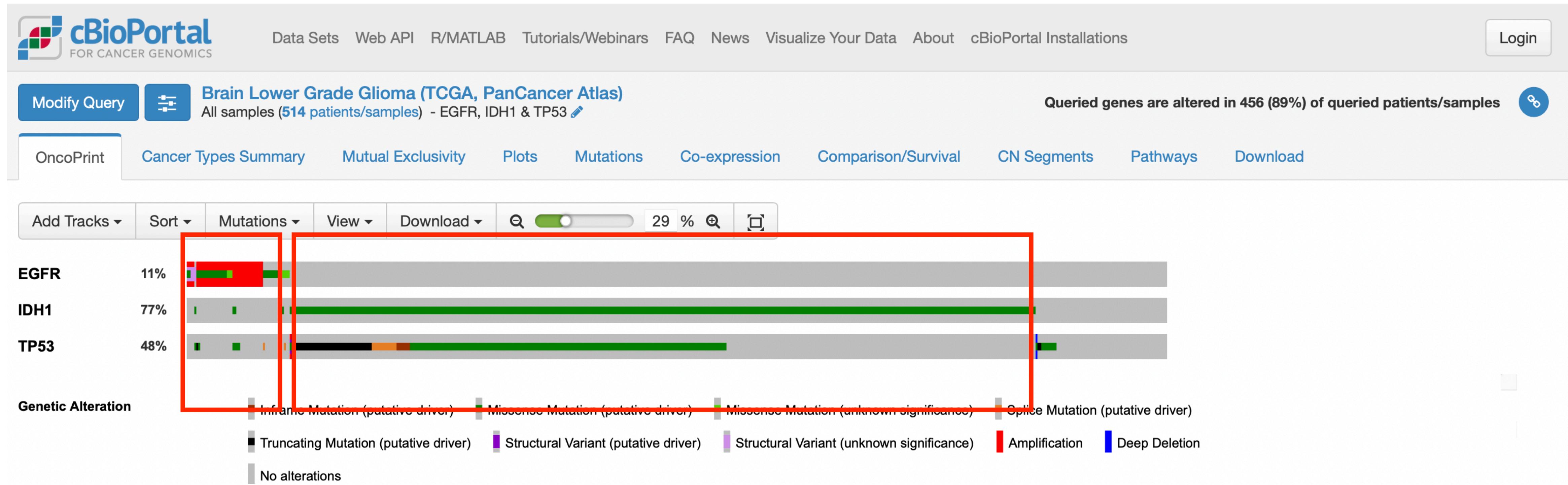
Query 1: Glioma

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



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

Mutual exclusivity Co-occurrence Significant only

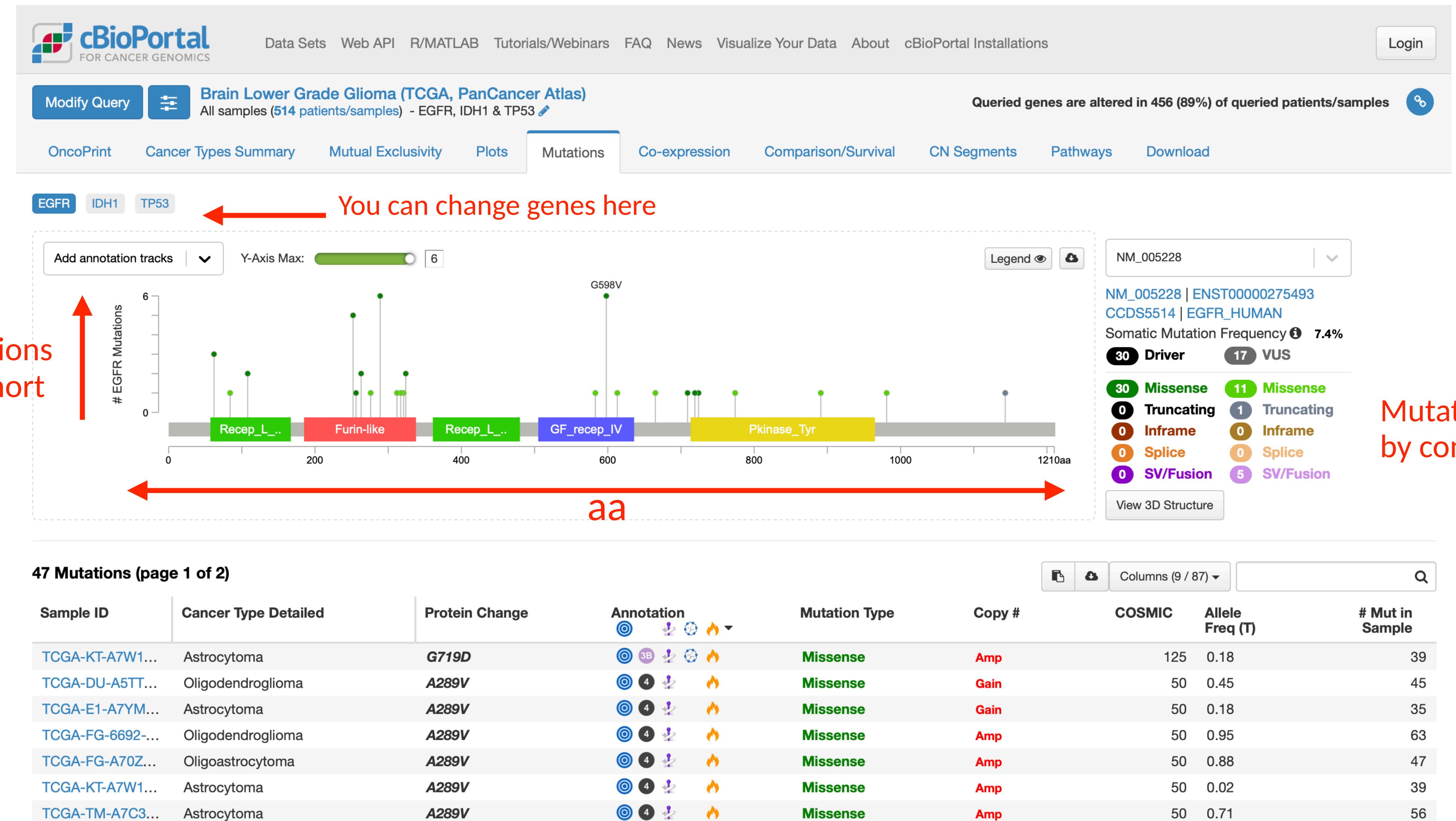
A	B	Neither	A Not B	B Not A	Both	Log2 Odds Ratio	p-Value	q-Value ▲	Tendency
EGFR	IDH1	67	50	390	4	<-3	<0.001	<0.001	Mutual exclusivity
IDH1	TP53	100	162	17	232	>3	<0.001	<0.001	Co-occurrence
EGFR	TP53	217	45	240	9	-2.467	<0.001	<0.001	Mutual exclusivity

Showing 1-3 of 3

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

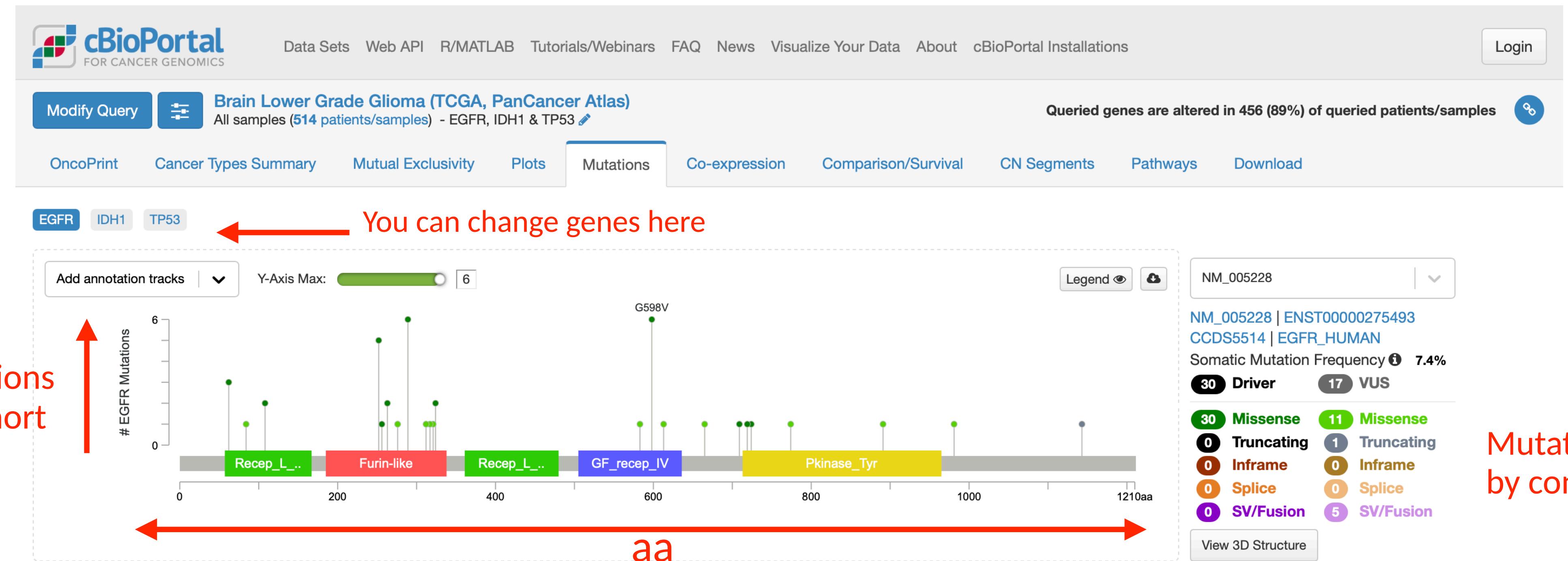
Query 1: Glioma

LOLLIPOP



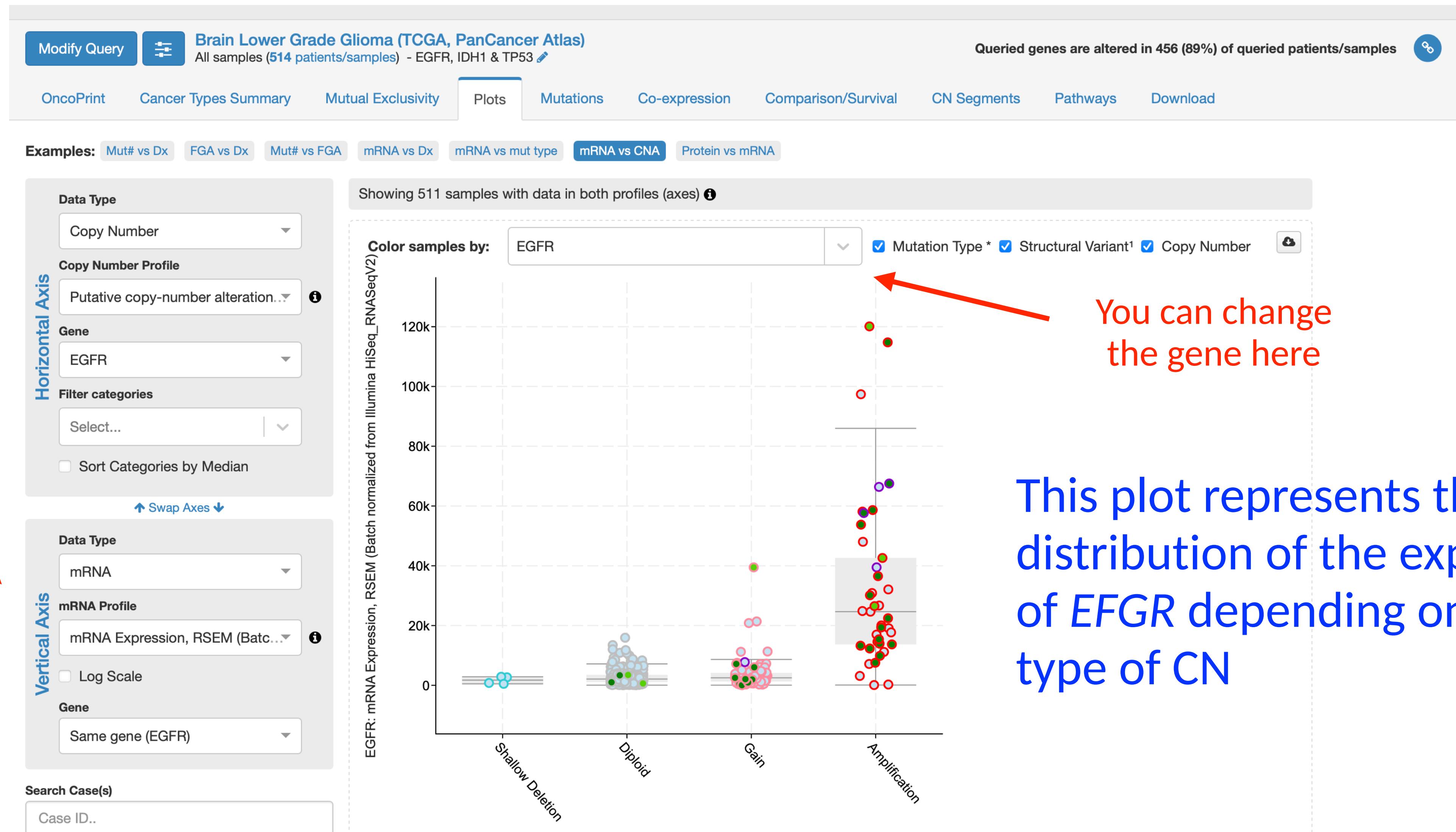
Query 1: Glioma

LOLLIPOP

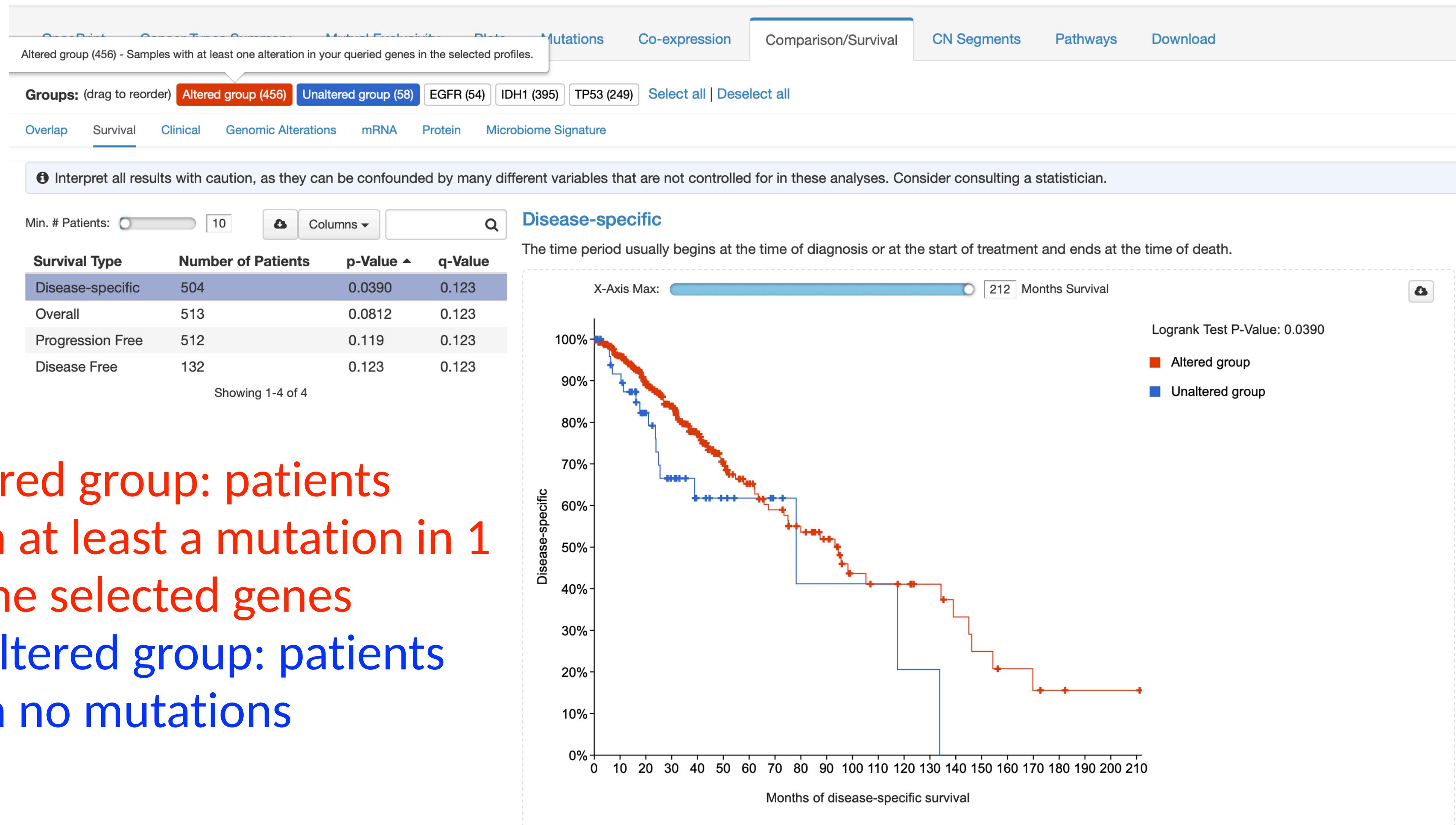


You can plot this plot with your own data

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)

High expression in ...		<input type="checkbox"/> Significant only								Columns ▾	<input type="text"/>
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)

-log10 p-value

Log Ratio

Significance →

Altered group Unaltered group

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
RBP1	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

Select Studies for Visualization & Analysis: 1 32 studies selected (10967 samples) Deselect all Search...

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

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 ⓘ 🔍

What's New @cbioportal 🔍

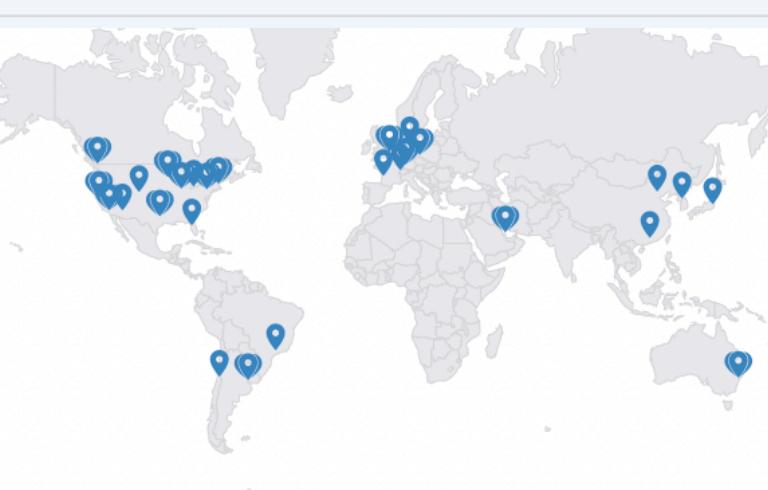
cBioPortal Retweeted
The Hyve
@TheHyveNL
Adding single-cell data and visualisations to #cBioPortal would reduce the time spent finding, downloading, and processing data for both users new to #singlecell data and users with expertise in single-cell technologies.
ow.ly/OyaE50Gb3jT #cancerresearch #genomics

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Example Queries

- 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



2 Query by gene

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 [🔗](#)
User-defined List
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

- 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
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- Patient view of an endometrial cancer case
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- MSK-IMPACT clinical cohort, Zehir et al. 2017
- Histone mutations across cancer types

Local Installations Host your own

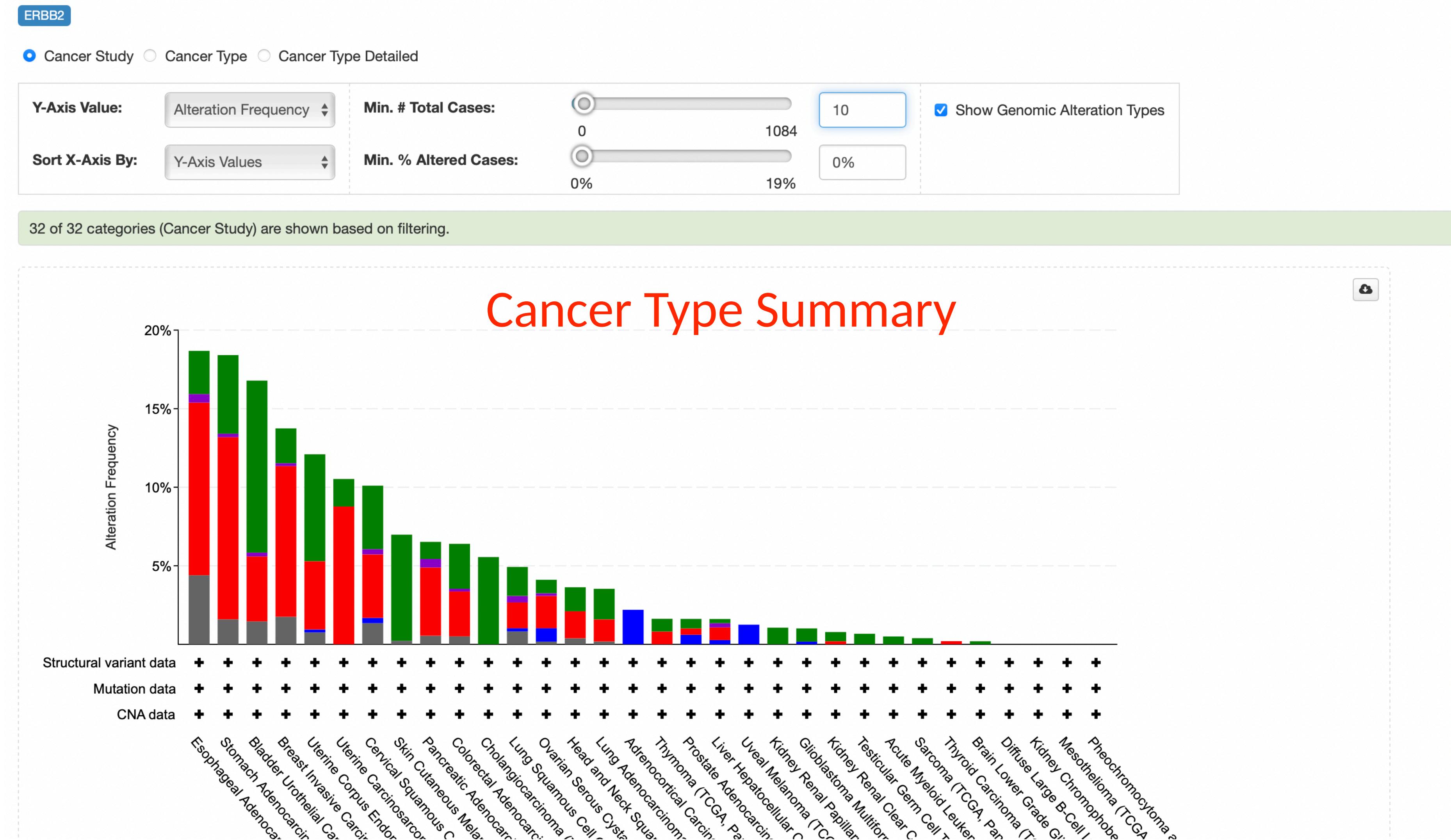


Enter the genes of interest
ERBB2

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-2GUGK'. Below the search bar, there are two sections: 'PATIENT' and 'SAMPLE'. The 'PATIENT' section lists 'LUAD-2GUGK' (Lung Adenocarcinoma (Broad, Cell 2012)) and 'LUAD-2GUGK' (Pan-Lung Cancer (TCGA, Nat Genet 2016)). The 'SAMPLE' section lists 'LUAD-2GUGK' (Lung Adenocarcinoma (Broad, Cell 2012)) and 'LUAD-2GUGK-Tumor' (Pan-Lung Cancer (TCGA, Nat Genet 2016)). The right side of the page includes a 'What's New' section with a tweet from The Hyve (@TheHyveNL) about adding single-cell data and visualizations to cBioPortal, and a 'Example Queries' section with a list of various cancer-related queries.

Quick Search Beta!

LUAD-2GUGK

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)

Click on a patient to see a summary

Click on a sample to open its summary

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

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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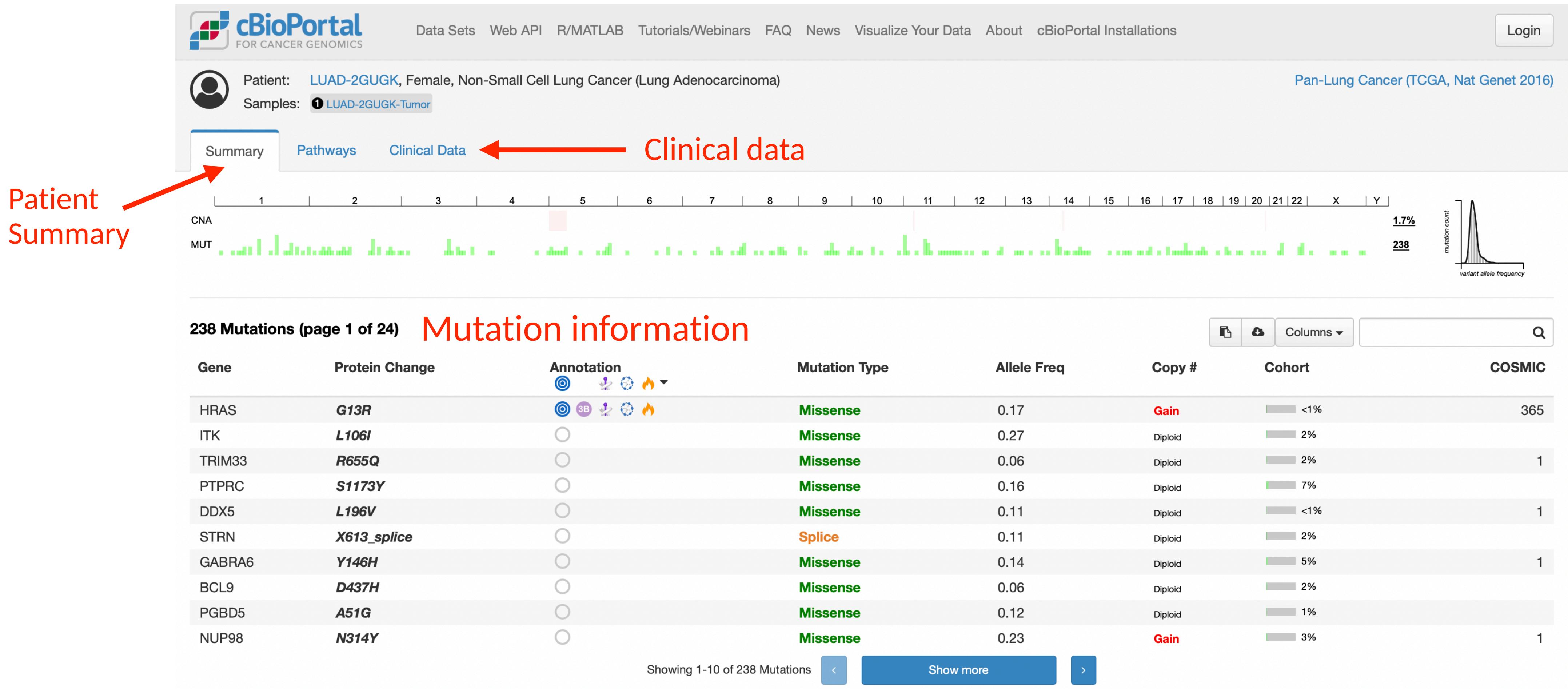
Example Queries

- 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

Map showing the locations of cBioPortal installations around the world, with pins placed in North America, Europe, Asia, and Australia.

Query 3: Individual samples



Query 3: Individual samples



Gene	Protein	Chromosome	Variant Class	Frequency	Diploid	Cohort	Sample ID
PTPRC	S1173Y	11q13.3	Missense	0.16	Diploid	7%	1
DDX5	L196V	11q13.3	Missense	0.11	Diploid	<1%	1
STRN	X613_splice	11q13.3	Splice	0.11	Diploid	2%	1
GABRA6	Y146H	11q13.3	Missense	0.14	Diploid	5%	1
BCL9	D437H	11q13.3	Missense	0.06	Diploid	2%	1
PGBD5	A51G	11q13.3	Missense	0.12	Diploid	1%	1
NUP98	N314Y	11q13.3	Missense	0.23	Gain	3%	1

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.cbiportal.org/tutorials**