

cBioPortal Tutorial #3: Patient View

Investigate individual patients or samples in detail

Tutorial Objectives

- Show different routes to get to patient view
- Walk through each of the possible tabs in patient view
 - Summary
 - Clinical Data
 - Genomic Evolution
 - Pathology Report
 - Tissue Image
- Highlight the different types of information available in different studies
- Show an example of the insights that can be found from patient view

Option # 1 to get to patient view:

Anywhere you see a patient or sample ID, that ID is a link to patient view for that case.

See next slide for examples.

[Modify Query](#)

Brain Lower Grade Glioma (TCGA, PanCancer Atlas)

Samples with mutation and CNA data (507 patients/samples) - IDH1, EGFR & IDH2

Queried genes are altered in 465 (92%) of queried patients/samples

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

Add Cl

TCGA-CS-5393

Mutation: IDH1 R132H
Profiled in all selected molecular profiles.

Tracks 4

Sort

Mutations

View

Download



100 %



IDH1

78%

EGFR

11%

IDH2

5%

Genetic Alteration

Inframe Mutation (unknown significance)

Missense Mutation (putative driver)

Missense Mutation (unknown significance)

Fusion

Amplification

Deep Deletion

Click on any of these sample/patient IDs

IDH1

EGFR

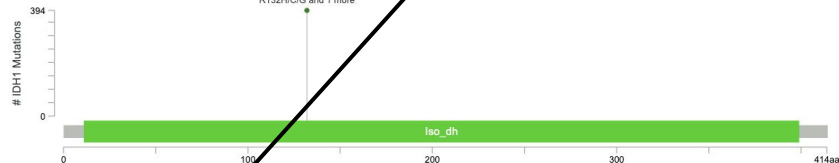
IDH2

Add annotation tracks

Y-Axis Max:

394

Legend



394 Mutations (page 1 of 16)

Sample ID	Cancer Type	Protein Change	Annotation	Mutation Type	Copy #
TCGA-CS-6666-01	Astrocytoma	R132C		Missense	Diploid
TCGA-DB-5240-01	Oligoastrocytoma	R132C		Missense	Diploid
TCGA-DB-A4XD-01	Astrocytoma	R132C		Missense	Diploid

[OncoPrint](#) [Cancer Types Summary](#) [Mutual Exclusivity](#) [Plots](#) [Mutations](#) [Co-expression](#) [Comparison/Survival](#) [CN Segments](#) [Pat](#)[Mut# vs Dx](#)[FGA vs Dx](#)[Mut# vs FGA](#)[mRNA vs Dx](#)[mRNA vs mut type](#)[mRNA vs CNA](#)[Protein vs mRNA](#)

Copy Number

Copy Number Profile

Putative copy-number alteration

Gene

EGFR

Filter categories

Select...

☐ Sort Categories by Median

Swap Axes

Data Type

mRNA

mRNA Profile

mRNA Expression, RSEM (Batch...

☐ Log Scale

Gene

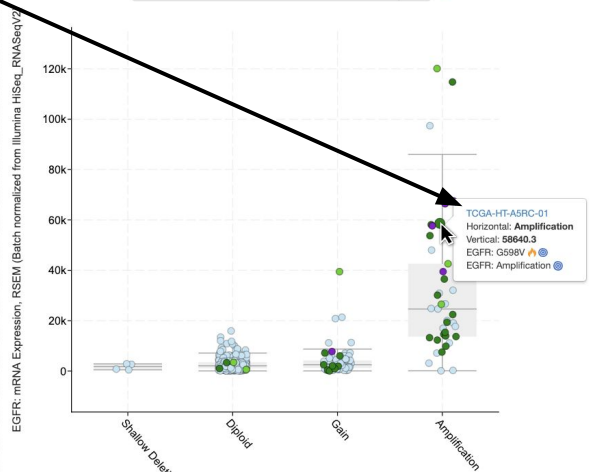
Same gene (EGFR)

Search Case(s)

Case ID...

Color samples by:

EGFR

☒ Mutation Type * ☐ Copy Nu

Option #2 to get to patient view:

Use the study summary page to filter down to cases of interest. Then click the “view the selected patients” button.

See next slide for example.

Cancer Type Detailed : **Oligoastrocytoma**

Clear All Filters



View selected cases

0 samples



Custom Selection ▾

Charts ▾

Groups ▾

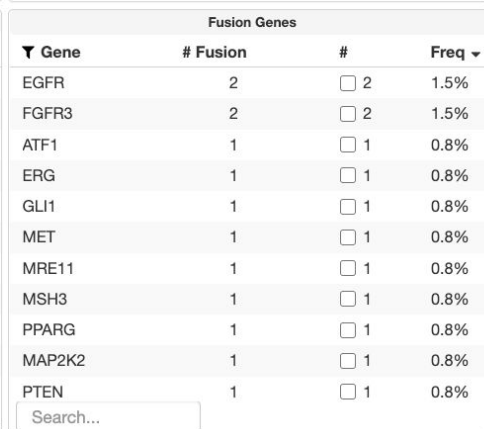
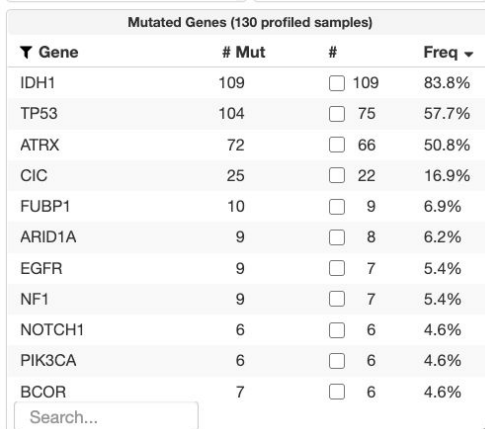
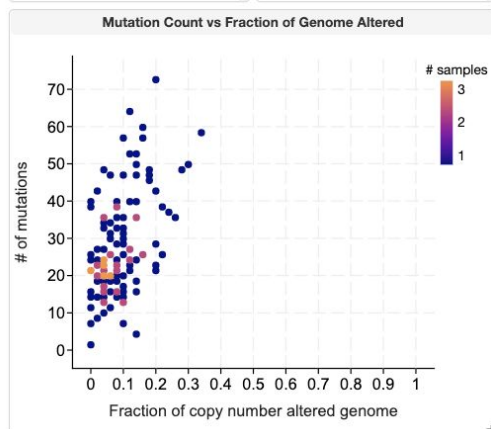
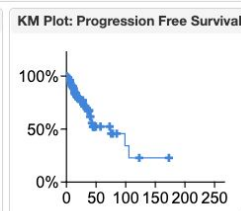
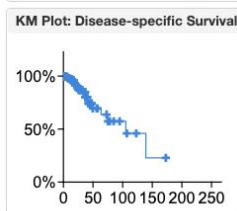
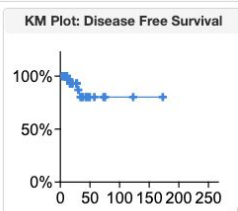
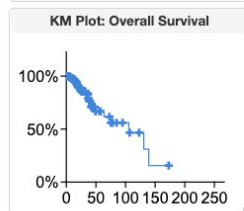
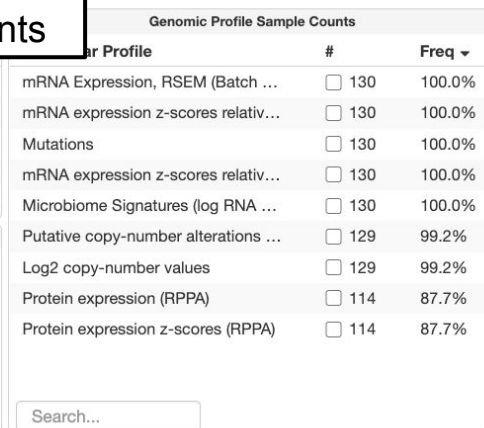
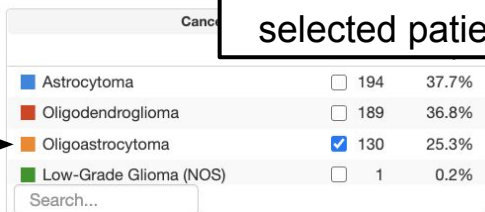
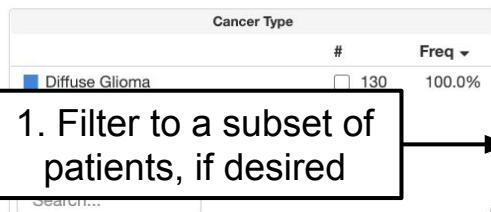
Summary

Clinical Data

CN Segments

2. Click on this button to view the selected patients

1. Filter to a subset of patients, if desired

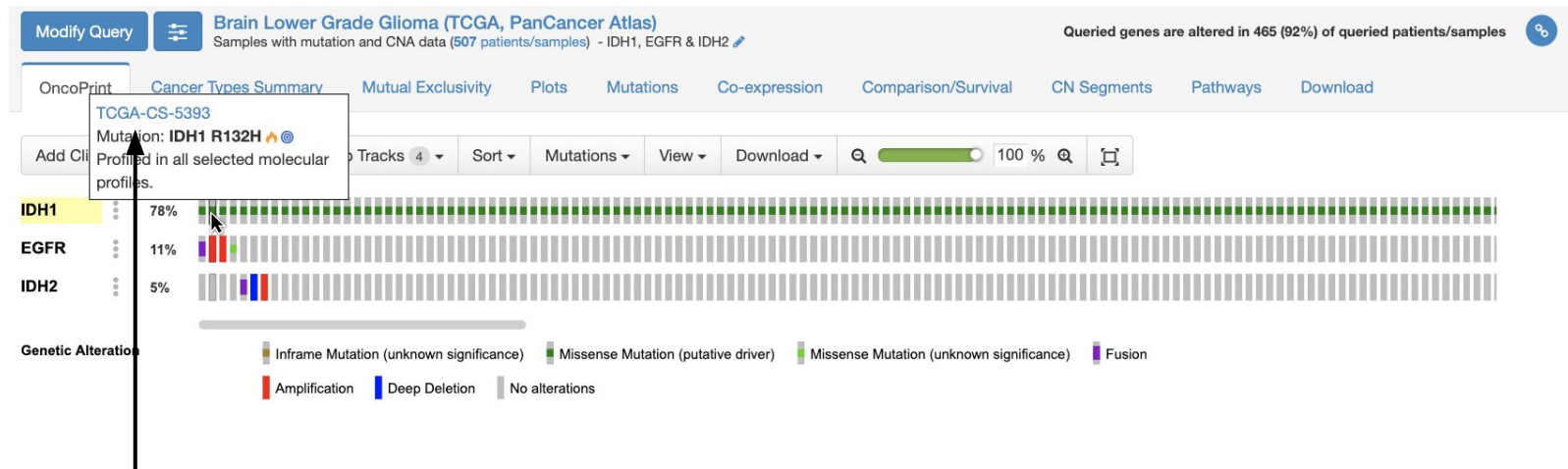


No matter how you get to patient view, you will be taken to the summary tab.

Depending on the study, the other tabs in patient view may or may not be present.

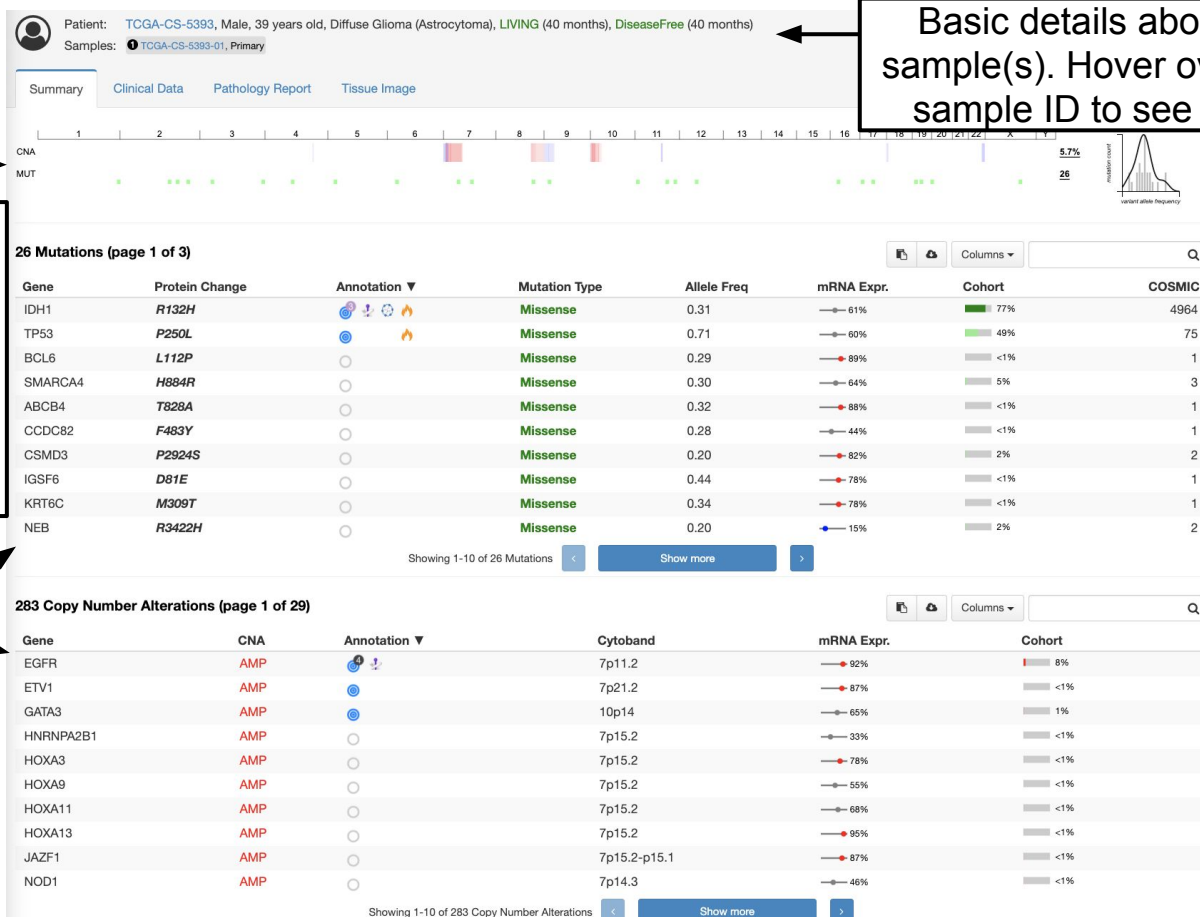
In this tutorial we will look at patient view in two different studies to highlight the different kinds of data that may be available.

Example 1: Brain Lower Grade Glioma (TCGA, PanCancer Atlas)



This is the same query that we used in the single study query tutorial. Hover over a case of interest and then click on the patient ID.

Patient View, Example 1: Summary



Basic details about the patient and sample(s). Hover over the patient ID or sample ID to see more information.

Figure showing where called CNA and mutations are across the genome. Hover over any of these for more details.

Lists of all called mutations and CNAs.

Copy, download, add/remove columns or search.

Patient View, Example 1: Clinical Data

 Patient: [TCGA-CS-5393](#), Male, 39 years old, Diffuse Glioma (Astrocytoma), [LIVING](#) (40 months), [DiseaseFree](#) (40 months) Brain Lower Grade Glioma (TCGA, PanCancer Atlas)

Samples: [1 TCGA-CS-5393-01, Primary](#)

[Summary](#) [Clinical Data](#) [Pathology Report](#) [Tissue Image](#)

All available patient-level clinical information

Patient

Attribute	Value
Overall Survival Status	0:LIVING
Birth from Initial Pathologic Diagnosis Date	-14418.0
Buffa Hypoxia Score	-25
Center of sequencing	Thomas Jefferson University
Diagnosis Age	39.0
Disease Free (Months)	40.174902193
Disease Free Status	0:DiseaseFree
Disease-specific Survival status	0:ALIVE OR DEAD TUMOR FREE
Form completion date	3/16/11
ICD-10 Classification	C71.9
In PanCan Pathway Analysis	Yes
Informed consent verified	Yes
International Classification of Diseases for Oncology, Third Edition ICD-O-3 Histology Code	9401/3

Below the patient-level information is sample-level information. Patients with multiple samples will have multiple columns in this table.

International Classification of Diseases for Oncology, Third Edition ICD-O-3
Last Alive Less Initial Pathologic Diagnosis Date Calculated Day Value
Last Communication Contact from Initial Pathologic Diagnosis Date
Months of disease-specific survival
Neoadjuvant Therapy Type Administered Prior To Resection Text
New Neoplasm Event Post Initial Therapy Indicator
Number of Samples Per Patient
Other Patient ID
Overall Survival (Months)
Person Neoplasm Cancer Status
Prior Diagnosis
Progress Free Survival (Months)
Progression Free Status
Race Category



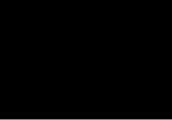
Samples		
Attribute	TCGA-CS-5393-01	
Mutation Count	24	
Fraction Genome Altered	0.0569	
MSI MANTIS Score	0.2715	
MSIsensor Score	0	
Sample Type	Primary	
10q Status	Not Called	
11p Status	Not Called	
11q Status	Not Called	
12p Status	Not Called	
12q Status	Not Called	
13 (13q) Status	Not Called	
14 (14q) Status	Not Called	
15 (15q) Status	Not Called	

Patient View, Example 1: Pathology Report

 Patient: [TCGA-CS-5393](#), Male, 39 years old, Diffuse Glioma (Astrocytoma), [LIVING](#) (40 months), [DiseaseFree](#) (40 months) [Brain Lower Grade Glioma \(TCGA, PanCancer Atlas\)](#)
Samples: [TCGA-CS-5393-01](#), Primary

[Summary](#) [Clinical Data](#) [Pathology Report](#) [Tissue Image](#)

Note: Pathology Reports are only available for TCGA studies.

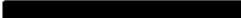


 TCGA-CS-5393 

SURGICAL PATHOLOGY REPORT

FINAL DIAGNOSIS:

1. Left temporal parietal tumor: Anaplastic astrocytoma, grade III of IV (WHO scale), see microscopic description, SEE NOTE

Comment:
The proliferation index of 7.2% is within the expected range for an anaplastic astrocytoma, grade III.

This diagnostic report has been personally interpreted by the signatory of record.

Microscopic Description:
The tumor consists of a moderately pleomorphic and highly infiltrative proliferation of astrocytes.
There are rare mitoses. There is no endothelial proliferation or necrosis. Immunohistochemistry for the proliferation antigen ki67 was performed as follows: Ten 250 x 250 micron fields were counted and the

Original pathology report, de-identified.

Patient View, Example 1: Tissue Image

Patient: [TCGA-CS-5393](#), Male, 39 years old, Diffuse Glioma (Astrocytoma), **LIVING** (40 months), **DiseaseFree** (40 months)
Samples: [TCGA-CS-5393-01](#), Primary

Summary Clinical Data Pathology Report **Tissue Image**

CANCER
Digital Slide Archive

EMORY WINSHIP
CANCER
INSTITUTE

LOGIN HELP

SLIDES

Tree

Thumbnails

acc

TCGA-OR-A5J1

TCGA-OR-A5J1-01A-01-TS1

TCGA-OR-A5...

Slides view Metadata view

Metadata Apply Filters Pathology Report

No Labels Drawing Disabled

Layer: Default Layer

TCGA-OR-A5J1-01A-01-TS1.CFE08710-54B8-45B0-86AE-500D6E36D8A5.svs

< 1 / 1 > 1 slides

This tab integrates the [Cancer Digital Slide Archive](#).

Note: Tissue images are only available for TCGA studies.

Zoomable image of the tissue. When available, additional images can be selected from the list on the left.

Example 2: Low-Grade Gliomas (UCSF, Science 2014)

Low-Grade Gliomas (UCSF, Science 2014) [PubMed](#)

Whole exome sequencing of 23 grade II glioma tumor/normal pairs. [PubMed](#)

Click gene symbols below or enter here

Number of Samples Per Patient : or

Summary Clinical Data

Selected: 2 patients | 12 samples

Cancer Type Detailed

	#	Freq
<input checked="" type="checkbox"/> Glioblastoma	5	41.7%
<input checked="" type="checkbox"/> Oligoastrocytoma	4	33.3%
<input checked="" type="checkbox"/> Oligodendroglioma	3	25.0%

Genomic Profile Sample Counts

Molecular Profile

Mutations

Number of Samples...

	#	Freq
<input type="checkbox"/> 2	17	73.9%
<input type="checkbox"/> 4	3	13.0%
<input type="checkbox"/> 3	1	4.3%
<input type="checkbox"/> 5	1	4.3%
<input checked="" type="checkbox"/> 7	1	4.3%

Search...

Number of Samples Per Patient

Search...

1. Filter the study to a subset of patients, if desired

Low-Grade Gliomas (UCSF, Science 2014) [PubMed](#)

Whole exome sequencing of 23 grade II glioma tumor/normal pairs. [PubMed](#)

Click gene symbols below or enter here

Number of Samples Per Patient : or

Summary Clinical Data

Selected: 2 patients | 12 samples

Cancer Type Detailed

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<input checked="" type="checkbox"/> Oligodendroglioma	3	25.0%

Genomic Profile Sample Counts

Molecular Profile

Mutations

Number of Samples Per Patient

Search...

2. Click on this button to "View selected cases"

Patient View, Example 2: Patient Summary

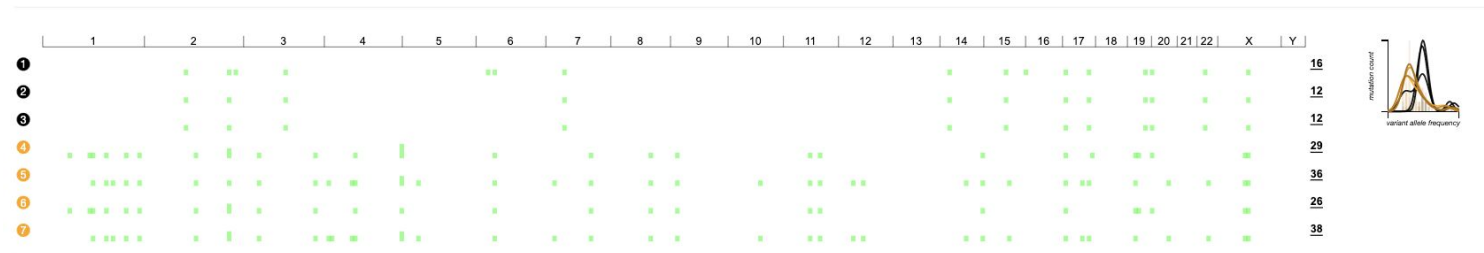
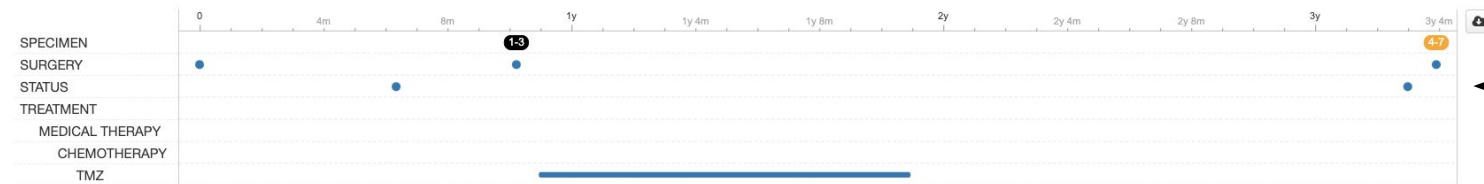
Patient: **P17**, Male, 27 years old, Glioma, **LIVING** (59 months)

Samples: **1** P17_Pri_A, Primary (Oligodendroglioma) **2** P17_Pri_B, Primary (Oligodendroglioma) **3** P17_Pri_C, Primary (Oligodendroglioma) **4** P17_Rec1_A, Recurrence (Glioblastoma)
5 P17_Rec1_B, Recurrence (Glioblastoma) **6** P17_Rec1_C, Recurrence (Glioblastoma) **7** P17_Rec1_D, Recurrence (Glioblastoma)

Low-Grade Gliomas (UCSF, Science 2014)

« < 1 of 2 patients > »

Summary Genomic Evolution Clinical Data



62 Mutations (page 1 of 7)

Samples	Gene	Protein Change	Annotation ▼	Mutation Type	Allele Freq	Cohort	COSMIC
1 2 3 4 5 6 7	IDH1	R132H		Missense	■■■ ■■■ ■■■	100%	4964
4 5 6 7	PIK3CA	H1047R		Missense	■■■■	8%	1983
1 2 3	TP53	C176F		Missense	■■■	90%	261
4 5 6 7	TP53	C176F		Missense	■■■■	90%	261

This study has multiple samples per patient and extensive clinical data to generate this enhanced patient timeline.

Patient View, Example 2: Patient Summary

Click to view
the next patient

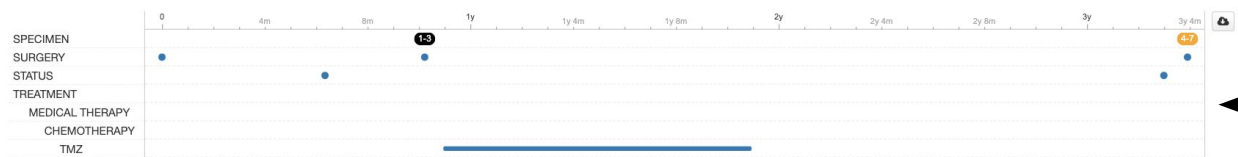
Patient: P17, Male, 27 years old, Glioma, **LIVING** (59 months)

Samples: P17_Pr1_A, Primary (Oligodendroglioma) P17_Pr1_B, Primary (Oligodendroglioma) P17_Pr1_C, Primary (Oligodendroglioma) P17_Rec1_A, Recurrence (Glioblastoma)
 P17_Rec1_B, Recurrence (Glioblastoma) P17_Rec1_C, Recurrence (Glioblastoma) P17_Rec1_D, Recurrence (Glioblastoma)

Low-Grade Gliomas **UCSF, Science (2014)**

<< < 1 of 2 patients > >>

Summary Genomic Evolution Clinical Data



Patient timeline showing surgeries, radiographic progression and treatments. Hover over any feature for additional information. Click the to expand the timeline.

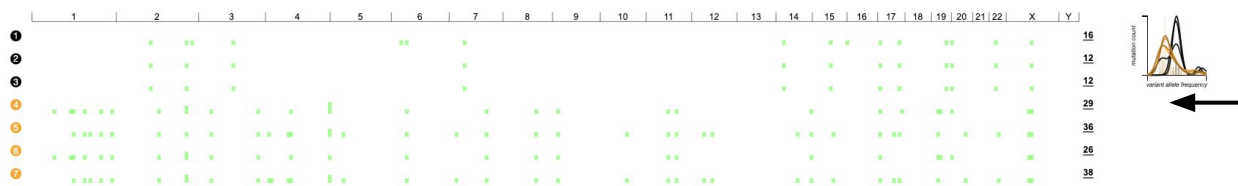


Figure showing distribution of mutations across the genome for each sample.

62 Mutations (page 1 of 7)

Samples	Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Cohort	COSMIC
	IDH1	R132H		Missense		100%	25
	PIK3CA	H1047R		Missense		8%	1983
	TP53	C176F		Missense		90%	261
	TP53	G129E		Missense		90%	261

List of all mutations called. The first column ("Samples") shows which samples had a particular mutation. The Allele Freq column depicts the mutation frequency in each sample by the height of the bar.

Patient View, Example 2: Patient Summary

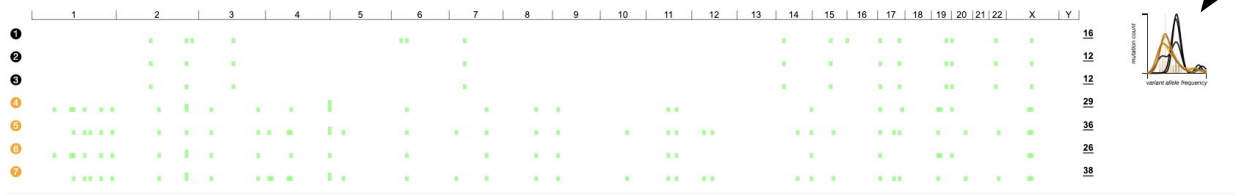
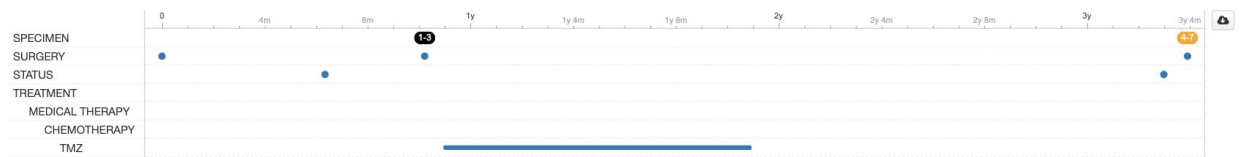
List of all samples for this patient. Hover on a sample ID for more details or click to get to a sample summary page (we'll do this in a few slides)

Patient: P17, Male, 27 years old, Glioma, LIVING (59 months)

Samples: P17_Pr1_A, Primary (Oligodendroglioma) P17_Pr1_B, Primary (Oligodendroglioma) P17_Pr1_C, Primary (Oligodendroglioma) P17_Rec1_A, Recurrence (Glioblastoma) P17_Rec1_B, Recurrence (Glioblastoma) P17_Rec1_C, Recurrence (Glioblastoma) P17_Rec1_D, Recurrence (Glioblastoma)

Low-Grade Gliomas (UCSF, Science 2014)

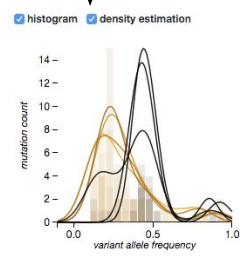
Summary Genomic Evolution Clinical Data



62 Mutations (page 1 of 7)

Samples	Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Cohort	COSMIC
P17_Pr1_A, P17_Pr1_B, P17_Pr1_C, P17_Rec1_A, P17_Rec1_B, P17_Rec1_C, P17_Rec1_D	IDH1	R132H	Missense	Missense	100%	100%	4964
P17_Pr1_A, P17_Pr1_B, P17_Pr1_C, P17_Rec1_A, P17_Rec1_B, P17_Rec1_C, P17_Rec1_D	PIK3CA	H1047R	Missense	Missense	8%	8%	1983
P17_Pr1_A, P17_Pr1_B, P17_Pr1_C, P17_Rec1_A, P17_Rec1_B, P17_Rec1_C, P17_Rec1_D	TP53	C176F	Missense	Missense	90%	90%	261
P17_Pr1_A, P17_Pr1_B, P17_Pr1_C, P17_Rec1_A, P17_Rec1_B, P17_Rec1_C, P17_Rec1_D	TP53	G147E	Missense	Missense	90%	90%	261

Hover to see an enlarged version. It shows a histogram with overlaid density estimation of the allele frequency in each tumor sample.



Hover over a sample ID to see the plot for just that sample

[Link to this page](#)

Patient View, Example 2: Genomic Evolution

Patient: P17, Male, 27 years old, Glioma, LIVING (59 months)

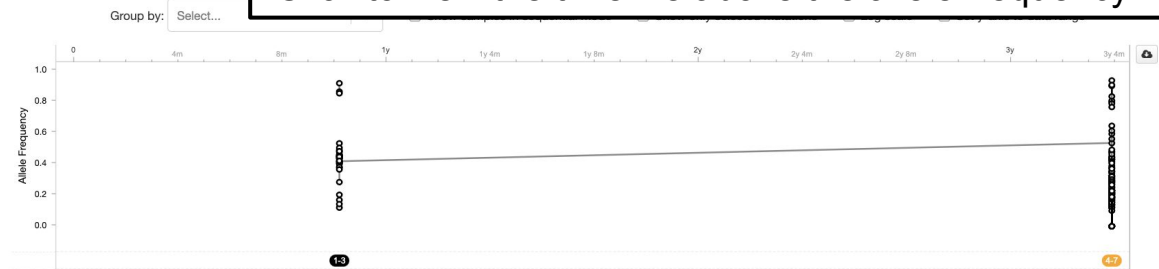
Low-Grade Gliomas (UCSF, Science 2014)

Samples: 1 P17_Ph_A, Primary (Oligodendroglioma) 2 P17_Ph_B, Primary (Oligodendroglioma) 3 P17_Ph_C, Primary (Oligodendroglioma) 4 P17_Rec1_A, Recurrence (Glioblastoma) 5 P17_Rec1_B, Recurrence (Glioblastoma) 6 P17_Rec1_C, Recurrence (Glioblastoma) 7 P17_Rec1_D, Recurrence (Glioblastoma)

< 1 of 2 patients >

Allele frequencies can be displayed as a Line Chart or Heatmap

Click to view the timeline above the allele frequency visualization



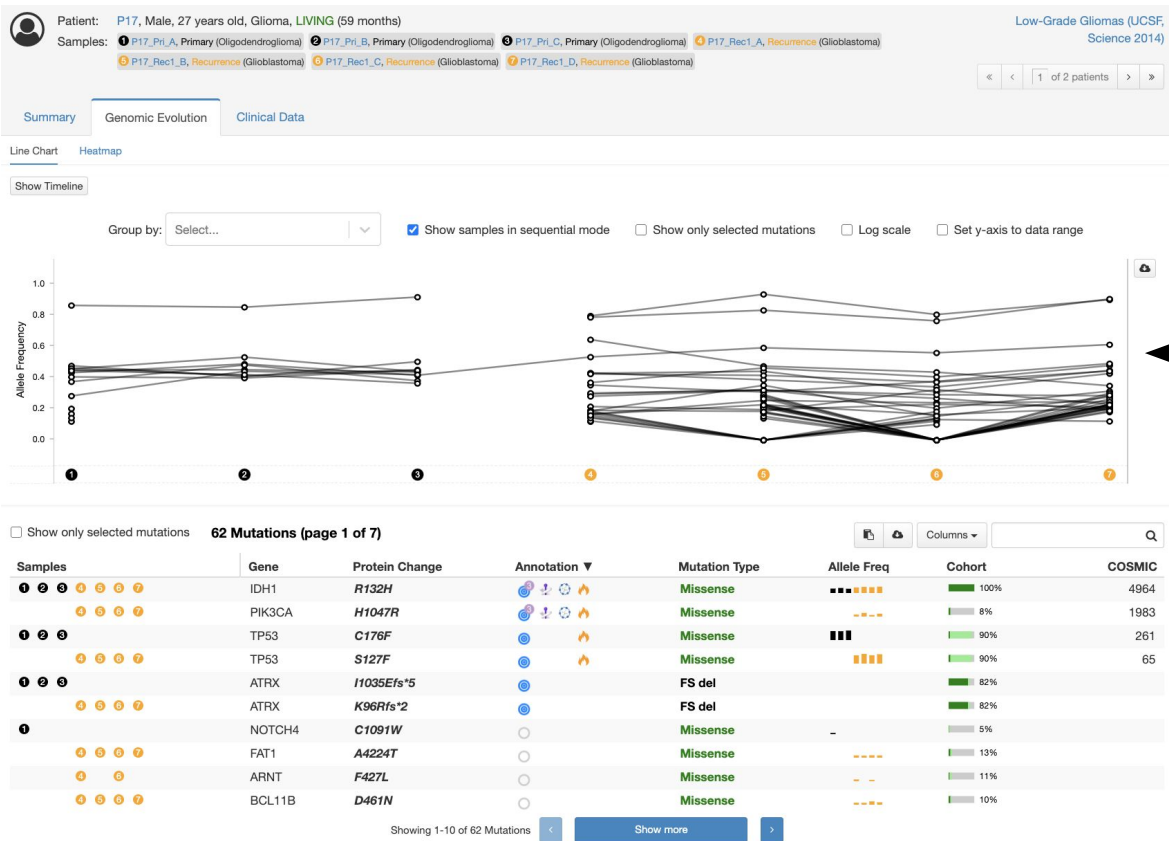
☐ Show only selected mutations 62 Mutations (page 1 of 7)

Samples	Gene	Protein Change	Annotation ▼	Mutation Type	Allele Freq	Cohort	COSMIC
1 2 3 4 5 6 7	IDH1	R132H	⚡ 🔥	Missense	■■■■■	100%	4964
1 2 3 4 5 6 7	PIK3CA	H1047R	⚡ 🔥	Missense	■■■■■	8%	1983
1 2 3 4 5 6 7	TP53	C176F	⚡ 🔥	Missense	■■■■■	90%	261
1 2 3 4 5 6 7	TP53	S127F	⚡ 🔥	Missense	■■■■■	90%	65
1 2 3 4 5 6 7	ATRX	I1035Efs*5	⚡	FS del	■■■■■	82%	
1 2 3 4 5 6 7	ATRX	K96Rfs*2	⚡	FS del	■■■■■	82%	
1 2 3 4 5 6 7	NOTCH4	C1091W	○	Missense	■■■■■	5%	
1 2 3 4 5 6 7	FAT1	A4224T	○	Missense	■■■■■	13%	
1 2 3 4 5 6 7	ARNT	F427L	○	Missense	■■■■■	11%	
1 2 3 4 5 6 7	BCL11B	D461N	○	Missense	■■■■■	10%	

Showing 1-10 of 62 Mutations [Show more](#)

The Genomic Evolution tab is present for any patient with 2 or more samples. This tab provides visualizations to examine how mutation allele frequencies vary among samples and change over time. The Timeline (on the Summary tab) can also be shown on this tab to put allele frequency changes in context.

Patient View, Example 2: Genomic Evolution - Line Chart

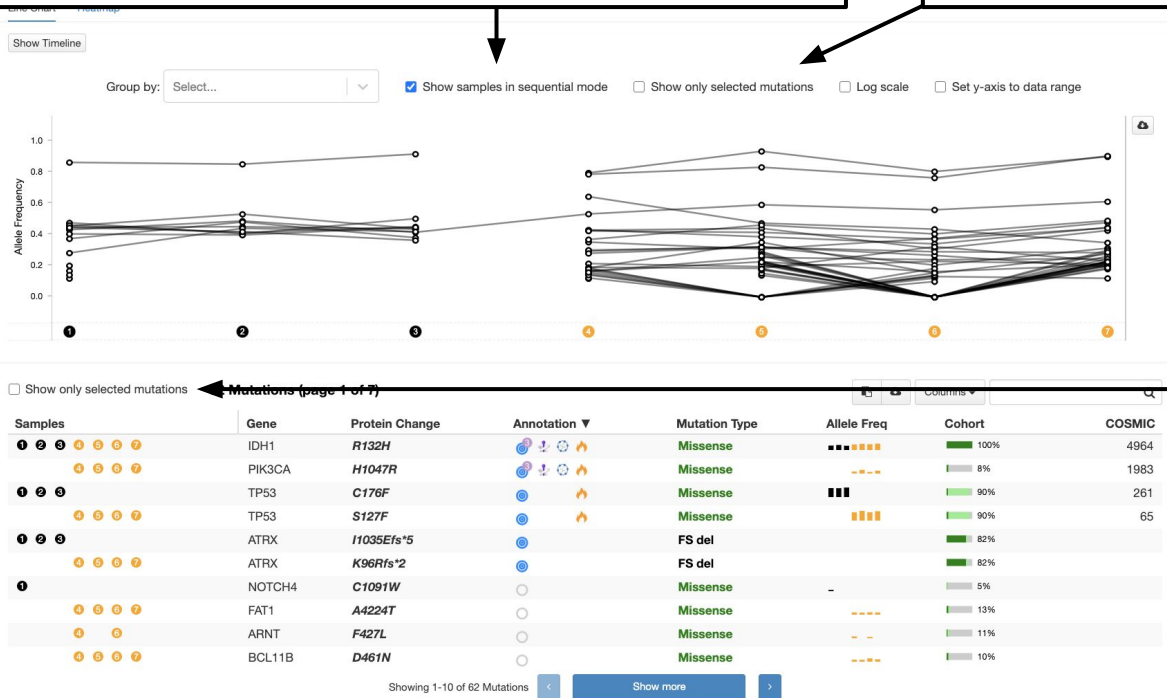


Each dot represents the allele frequency of a mutation in a sample. Lines connect mutations that are detected in multiple samples. Options above the chart enable customization.

Patient View, Example 2: Genomic Evolution - Line Chart

Change x-axis to show all samples equally spaced (below) or samples in real time (aligned with timeline, see first Genomic Evolution slide)

Click on mutations in the table below and then check this box to only see those mutations in the chart.

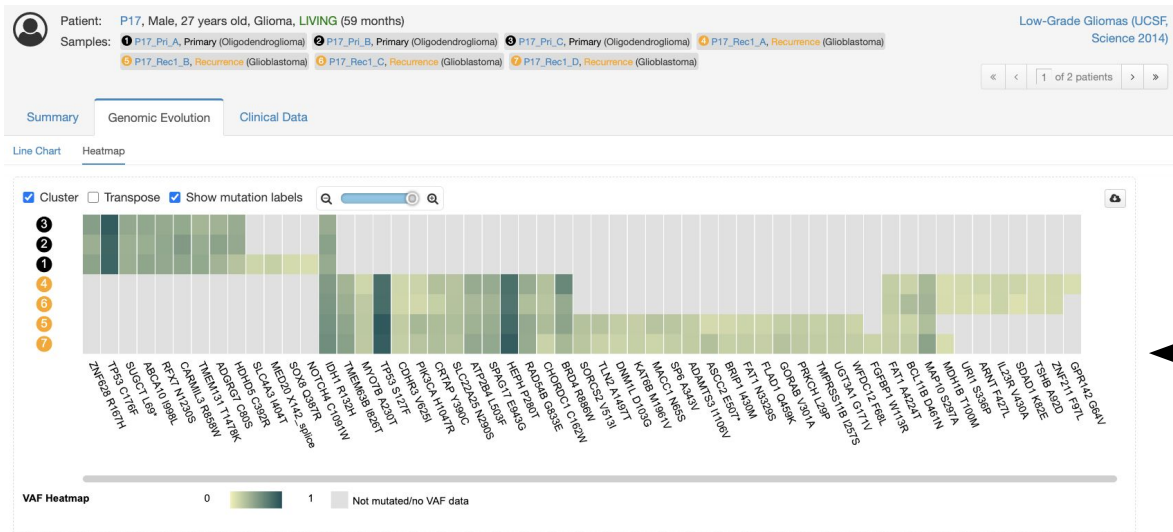


This chart and the mutation table are linked - hover or click on a mutation in the chart to see it highlighted in the table below.

Click on mutations in the chart above and then check this box to see only those mutations in the table.

Hover or click on a mutation in the table to see it highlighted in the chart above.

Patient View, Example 2: Genomic Evolution - Heatmap



Each box is colored according to the allele frequency of a mutation in a sample. Options above the chart enable customization.

Show only selected mutations 62 Mutations (page 1 of 7)

Samples	Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Cohort	COSMIC
1 2 3 4 5 6 7	IDH1	R132H		Missense	100%	100%	4964
1 2 3 4 5 6 7	PIK3CA	H1047R		Missense	8%	8%	1983
1 2 3 4 5 6 7	TP53	C176F		Missense	90%	90%	261
1 2 3 4 5 6 7	TP53	S127F		Missense	90%	90%	65
1 2 3 4 5 6 7	ATRX	I1035Efs*5		FS del	82%	82%	
1 2 3 4 5 6 7	ATRX	K96Rfs*2		FS del	82%	82%	
1 2 3 4 5 6 7	NOTCH4	C1091W		Missense	5%	5%	
1 2 3 4 5 6 7	FAT1	A4224T		Missense	13%	13%	
1 2 3 4 5 6 7	ARNT	F427L		Missense	11%	11%	
1 2 3 4 5 6 7	BCL11B	D461N		Missense	10%	10%	

Hover or click on a mutation in the table to see it highlighted in the chart above.

Patient View, Example 2: Clinical Data

 Patient: P17, Male, 27 years old, Glioma, LIVING (59 months)

Samples:  P17_Pri_A, Primary (Oligodendroglioma)  P17_Pri_B, Primary (Oligodendroglioma)  P17_Pri_C, Primary (Oligodendroglioma)  P17_Rec1_A, Recurrence (Glioblastoma)

 P17_Rec1_B, Recurrence (Glioblastoma)  P17_Rec1_C, Recurrence (Glioblastoma)  P17_Rec1_D, Recurrence (Glioblastoma)

Low-Grade Gliomas (UCSF, Science 2014)

<< < 1 of 2 patients > >>

Summary Genomic Evolution Clinical Data

Patient

Attribute	Value
Overall Survival Status	0:LIVING
Diagnosis Age	27
Number of Samples Per Patient	7
Overall Survival (Months)	59
Sex	Male

All available patient-level clinical information

Samples

Attribute	P17_Pri_A	P17_Pri_B	P17_Pri_C	P17_Rec1_A	P17_Rec1_B	P17_Rec1_C	P17_Rec1_D
Mutation Count	16	12	12	29	36	26	38
Sample Type	Primary	Primary	Primary	Recurrence	Recurrence	Recurrence	Recurrence
1p/19q Status	Intact	Intact	Intact	Intact	Intact	Intact	Intact
Cancer Type	Glioma	Glioma	Glioma	Glioma	Glioma	Glioma	Glioma
Cancer Type Detailed	Oligodendroglioma	Oligodendroglioma	Oligodendroglioma	Glioblastoma	Glioblastoma	Glioblastoma	Glioblastoma
IDH1 Mutation	R132H	R132H	R132H	R132H	R132H	R132H	R132H
MGMT Status	Methylated	Methylated	Methylated	Unmethylated	Unmethylated	Unmethylated	Unmethylated
Neoplasm Histologic Grade	II	II	II	IV	IV	IV	IV
Non-silent mutations in TP53, ATRX, CIC, FUBP1	TP53, ATRX	TP53, ATRX	TP53, ATRX	TP53, ATRX	TP53, ATRX	TP53, ATRX	TP53, ATRX
Oncotree Code	ODG	ODG	ODG	GB			
Somatic Status	Matched	Matched	Matched	Matched			

All available sample-level information

Timeline Data

Specimen

START_DATE	STOP_DATE	EVENT_TYPE	SURGERY	SAMPLE_ID
311		SPECIMEN	Oligo II Initial	P17_Pri_A
1214		SPECIMEN	GBM Recurrence1	P17_Rec1_A
311		SPECIMEN	Oligo II Initial	P17_Pri_B

When available, the data used to populate the timeline in the Summary tab is shown here.

Patient View, Example 2: Sample Summary

Patient: P17, Male, 27 years old, Glioma, **LIVING** (59 months)

Samples: 1 P17_Pr1_A, Primary (Oligodendroglioma) 2 P17_Pr1_B, Primary (Oligodendroglioma) 3 P17_Pr1_C, Primary (Oligodendroglioma) 4 P17_Rec1_A, Recurrence (Glioblastoma)

1 P17_Pr1_A

Attribute	Value
Mutation Count	16
Sample Type	Primary
1p/19q Status	Intact
Cancer Type	Glioma
Cancer Type Detailed	Oligodendroglioma
IDH1 Mutation	R132H
MGMT Status	Methylated
Neoplasm Histologic Grade	II
Non-silent mutations in TP53, ATRX, CIC, FUBP1	TP53, ATRX
Oncotree Code	ODG
Somatic Status	Matched

SPECIMEN
SURGERY
STATUS
TREATMENT
MEDICAL THERAPY
CHEMOTHERAPY
TMZ

Clicking on a sample ID on one of the previous pages brings up this sample summary page.

Patient: P17, Male, 27 years old, Glioma, **LIVING** (59 months)

Samples: 1 P17_Pr1_A, Primary (Oligodendroglioma) Show all 7 samples

Low-Grade Gliomas (UCSF, Science 2014)

Summary Clinical Data

0 4m 8m 1y 1y 4m 1y 8m 2y 2y 4m 2y 8m 3y 3y 4m

SPECIMEN
SURGERY
STATUS
TREATMENT
MEDICAL THERAPY
CHEMOTHERAPY
TMZ

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

16 Mutations (page 1 of 2)

Samples	Gene	Protein Change	Annotation ▼	Mutation Type	Allele Freq	Cohort	COSMIC
1	IDH1	R132H	🔵 🔽 🔵 🔴	Missense	■	100%	4964
1	TP53	C176F	🔵 🔴	Missense	■	90%	261
1	ATRX	I1035Efs*5	🔵	FS del	■	82%	
1	NOTCH4	C1091W	○	Missense	■	5%	
1	CARMIL3	R858W	○	Missense	■	8%	
1	RFX7	N1230S	○	Missense	■	8%	
1	ZNF628	R167H	○	Missense	■	7%	
1	ABCA10	I998L	○	Missense	■	7%	
1	SUGCT	L69*	○	Nonsense	■	5%	

Ok, now that we've seen what data is present in Patient View, we can start asking some fun question!

Let's look at RAS mutations in Uterine Corpus Endometrial Carcinoma (TCGA, Nature 2013).

Example 3: Run the query

Query

Quick Search **Beta!**

Download

Please cite: [Cerami et al., 2012](#) & [Gao et al., 2013](#)

Selected Studies:

Modify

Uterine Corpus Endometrial Carcinoma (TCGA, Nature 2013) (373 total samples)

Select Genomic Profiles:

☒ Mutations [?](#)

☒ Putative copy-number alterations from GISTIC [?](#)

☐ mRNA Expression. Select one of the profiles below:

☐ mRNA expression z-scores relative to diploid samples (microarray) [?](#)

☐ mRNA expression z-scores relative to diploid samples (RNA Seq V2 RSEM) [?](#)

☐ mRNA expression z-scores relative to all samples (log RNA Seq V2 RSEM) [?](#)

☐ mRNA expression z-scores relative to all samples (log microarray) [?](#)

Select Patient/Case Set:

To build your own case set,
try out our enhanced Study View.

Samples with mutation and CNA data (240)

×

▼

Enter Genes:

Hint: Learn Onco Query Language (OQL)
to write more powerful queries [↗](#)

User-defined List

×

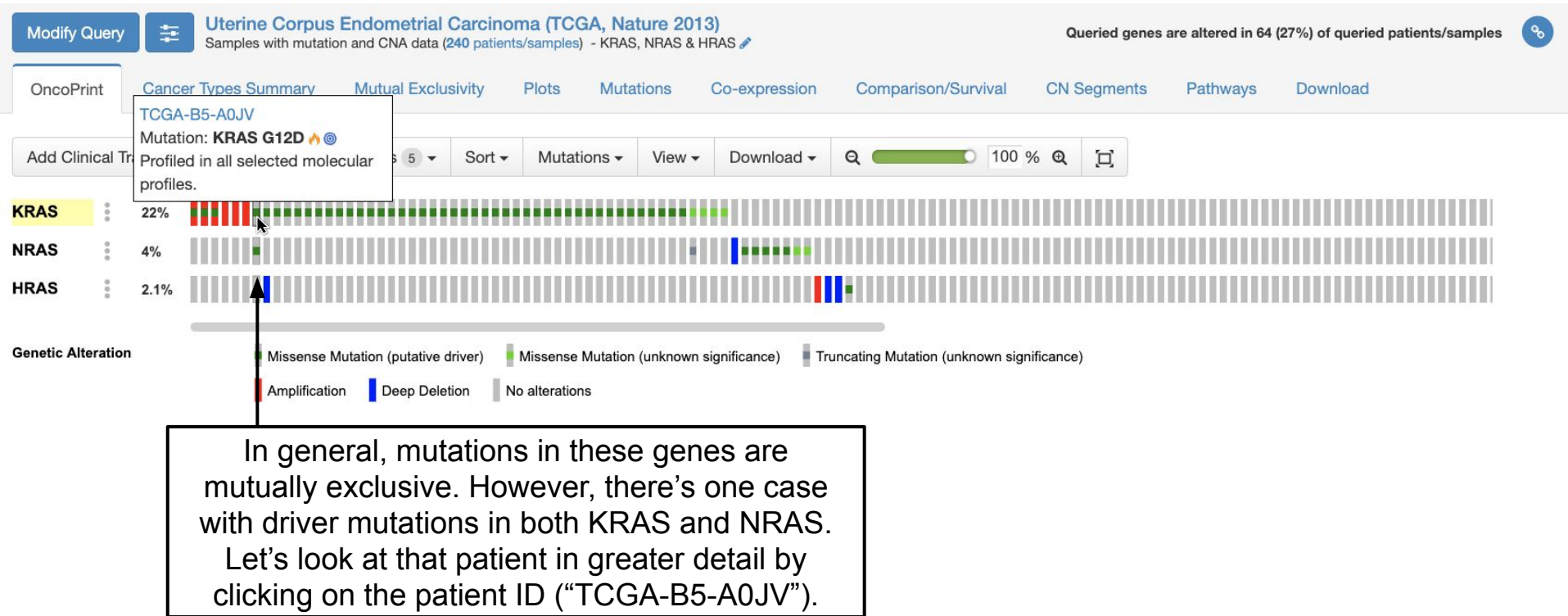
▼

KRAS NRAS HRAS

✔ All gene symbols are valid.

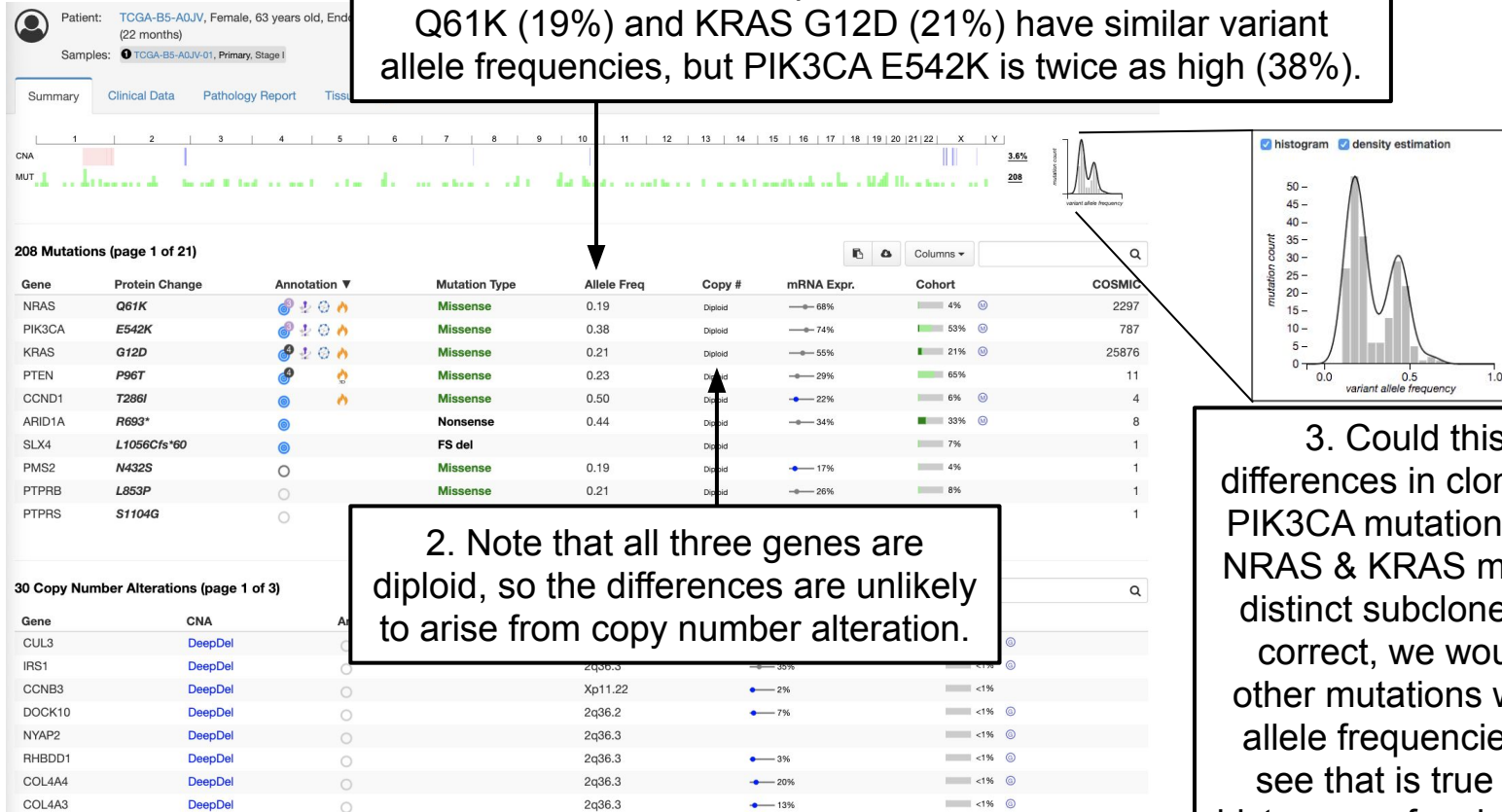
Submit Query

Example 3: OncoPrint



Example 3: Patient View

1. Look at the Allele Freq column for each mutation. NRAS Q61K (19%) and KRAS G12D (21%) have similar variant allele frequencies, but PIK3CA E542K is twice as high (38%).



2. Note that all three genes are diploid, so the differences are unlikely to arise from copy number alteration.

3. Could this be related to differences in clonality? Perhaps the PIK3CA mutation is clonal while the NRAS & KRAS mutations are in two distinct subclones. If that theory is correct, we would expect to see other mutations with similar variant allele frequencies. Indeed, we can see that is true by looking at the histogram of variant allele frequency.

Summary of Example 3: Using Patient View, we can infer the clonality of mutations and understand how two mutations, which are usually mutually exclusive, can be present in the same tumor sample. In this case, the KRAS and NRAS mutations appear to be present in two distinct subclones of a single tumor.

Questions?

Check out our other tutorials
or email us at:

cbioportal@googlegroups.com