cBioPortal Tutorial #3: Patient View

Investigate individual patients or samples in detail

Last update: June 11, 2019

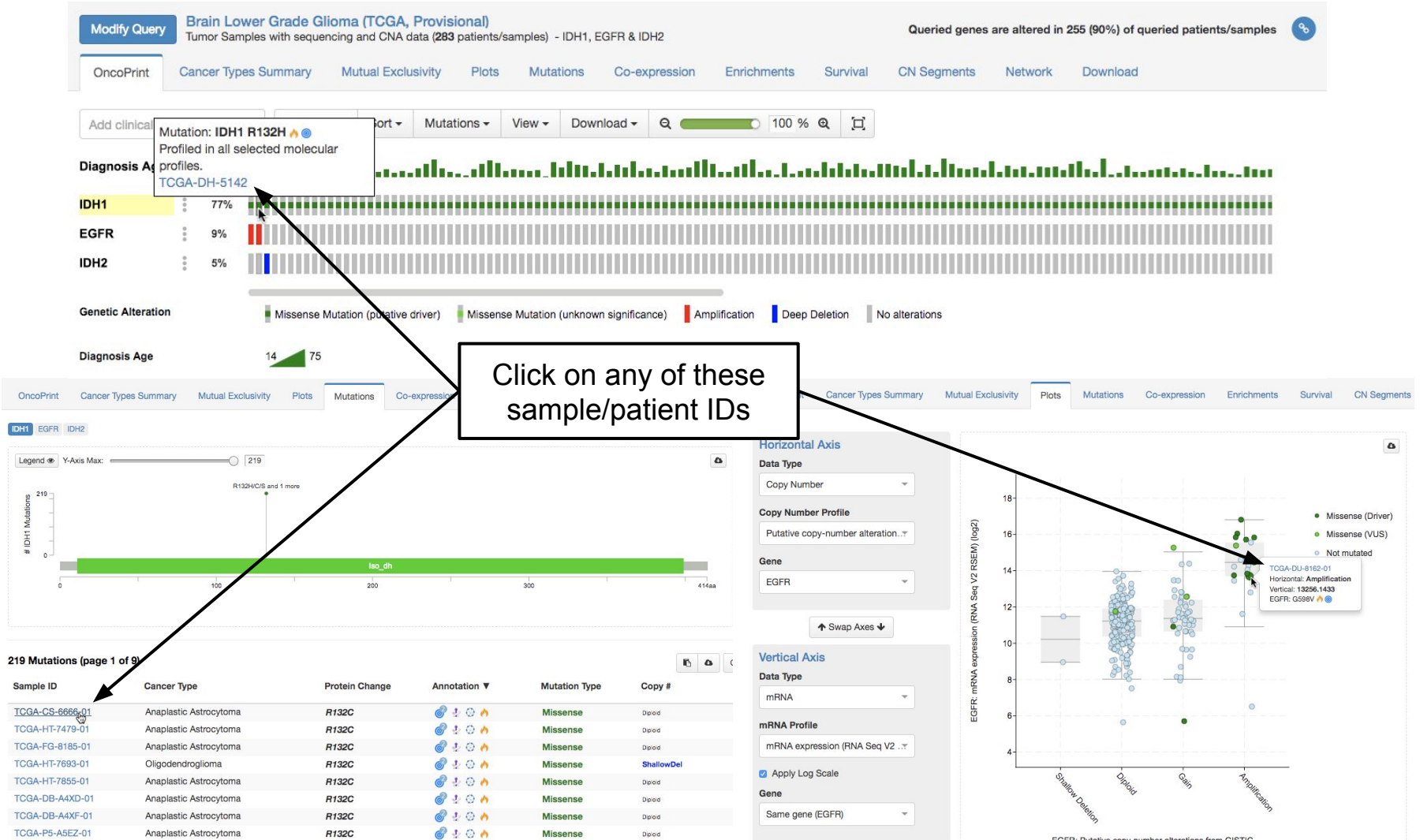
Tutorial Objectives

- Show different routes to get to patient view
- Walk through each of the possible tabs in patient view
  - Summary
  - Clinical Data
  - 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, Provisional)
Tumor Samples with sequencing and CNA data (283 patients/samples) - IDH1, EGFR & IDH2
Queried genes are altered in 255 (90%) of queried patients/samples
OncoPrint
Cancer Types Summary
Mutual Exclusivity
Plots
Mutations
Co-expression
Enrichments
Survival
CN Segments
Network
Download
Mutation: IDH1 R132H
Profiled in all selected molecular profiles.
TCGA-DH-5142
IDH1
77%
EGFR
9%
IDH2
5%
Genetic Alteration
Missense Mutation (putative driver)
Missense Mutation (unknown significance)
Amplification
Deep Deletion
No alterations
Diagnosis Age
14
75
Click on any of these sample/patient IDs
219 Mutations (page 1 of 9)
Sample ID
Cancer Type
Protein Change
Annotation
Mutation Type
Copy #
TCGA-CS-6666-01
TCGA-HT-7479-01
TCGA-FG-8185-01
TCGA-HT-7693-01
TCGA-HT-7855-01
TCGA-DB-A4XD-01
TCGA-DB-A4XF-01
TCGA-P5-A5EZ-01
Anaplastic Astrocytoma
Oligodendroglioma
R132C
Missense
Diploid
ShallowDel
Horizontal Axis
Data Type
Copy Number
Copy Number Profile
Putative copy-number alteration...
Gene
EGFR
Swap Axes
Vertical Axis
Data Type
mRNA
mRNA Profile
mRNA expression (RNA Seq V2 ...
Apply Log Scale
Gene
Same gene (EGFR)
EGFR: mRNA expression (RNA Seq V2 RSEM) (log2)
Missense (Driver)
Missense (VUS)
Not mutated
TCGA-DU-8162-01
Horizontal: Amplification
Vertical: 13256.1433
EGFR: G598V
Shallow Deletion
Diploid
Gain
Amplification

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.

Brain Lower Grade Glioma (TCGA, Provisional)

TCGA Brain Lower Grade Glioma; raw data at the NCI; source mutation data from GDAC Firehose.

Click gene symbols below or enter here | Query

Number of Samples Per Patient : 2 | Clear All Filters

View selected cases

Summary | Clinical Data | Heatmaps | CN Segments | Custom Selection | + Add Chart | Groups

2. Click on this button to view the selected patients

1. Filter to a subset of patients, if desired

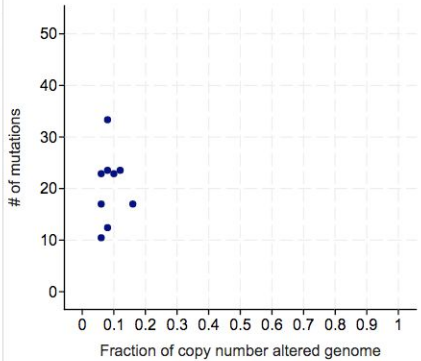
Quick Filters: 9 sam... A data

Cancer Type Detailed

	#	Freq
Oligodendroglioma	11	39.3%
Oligoastrocytoma	8	28.6%
Astrocytoma	5	17.9%
Anaplastic Oligoastrocytoma	3	10.7%
Anaplastic Astrocytoma	1	3.6%

Search...

Mutation Count vs Fraction



Mutated Genes (9 profiled samples)

Gene	# Mut	#	Freq
ACACA	2	2	22.2%
LAMA3	2	2	22.2%
NF1 (M)	3	2	22.2%
TTN	2	2	22.2%
TRRAP	2	2	22.2%
ATG5	2	2	22.2%
CIC (M)	3	2	22.2%
GRK2	1	1	11.1%

Search...

Number of Samples Per Patient

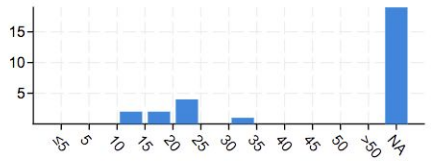
Sex

CNA Genes (14 profiled samples)

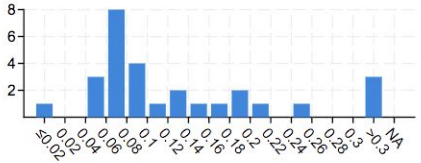
Gene	Cytoband	CNA	#	Freq
MIR-939/9...		AMP	3	21.4%
MIR-661/6...		AMP	3	21.4%
MIR-562/5...		DEL	3	21.4%
MIR-4441/...		DEL	3	21.4%
MIR-4440/...		DEL	3	21.4%
MIR-4269/...		DEL	3	21.4%
MIR-3133/...		DEL	3	21.4%
AGXT	2q37.3	DEL	3	21.4%
ALPI	2q37.1	DEL	3	21.4%
ALPP	2q37.1	DEL	3	21.4%
ALPG	2q37.1	DEL	3	21.4%

Search...

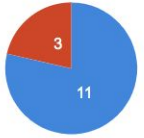
Mutation Count



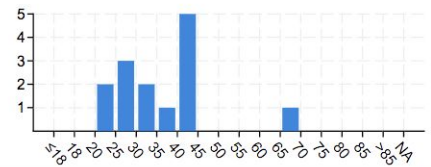
Fraction Genome Altered



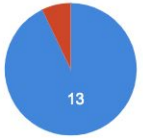
Ethnicity Category



Diagnosis Age



Race Category



Sample Type

Tumor Site

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, Provisional)

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.

Link to this page

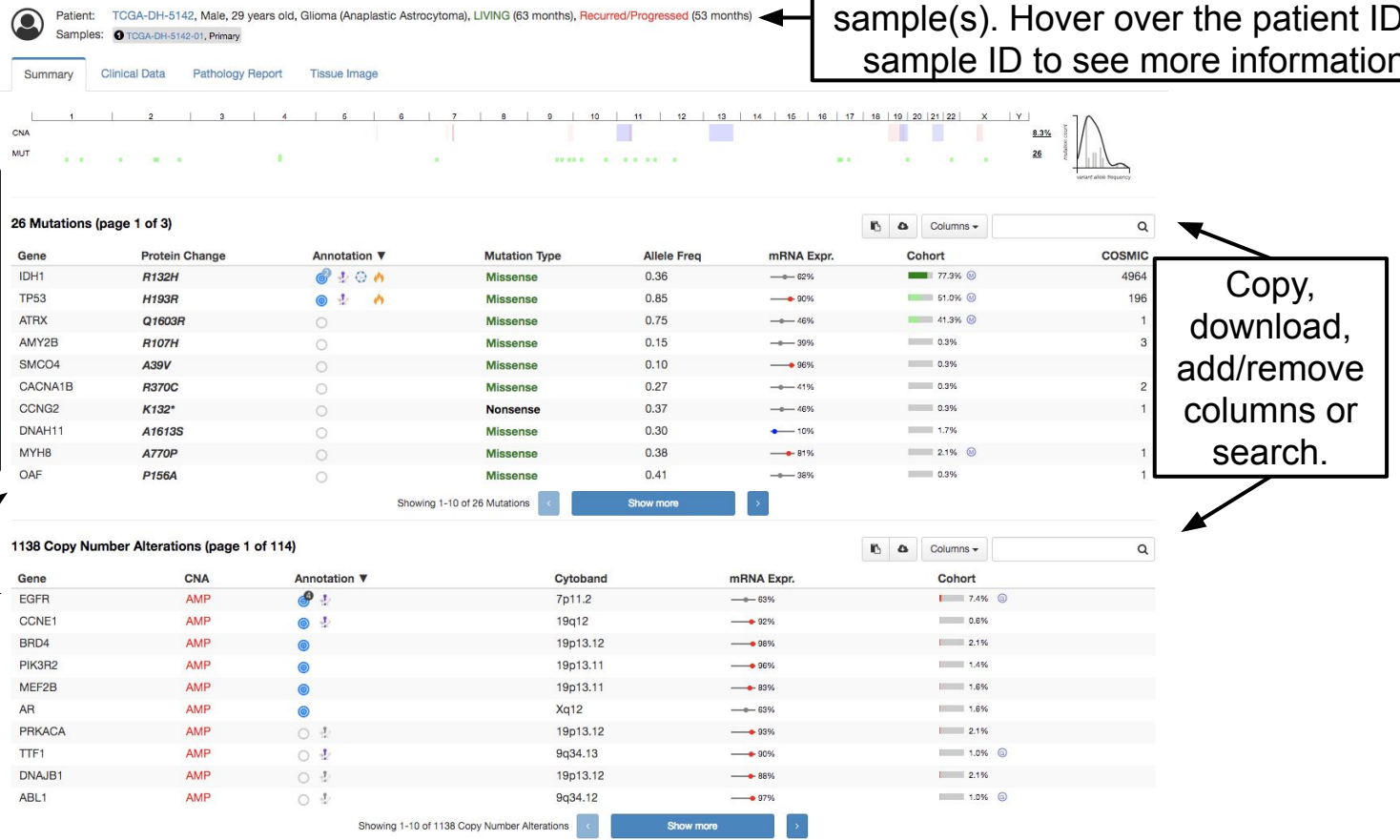
Patient View, Example 1: Summary

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.

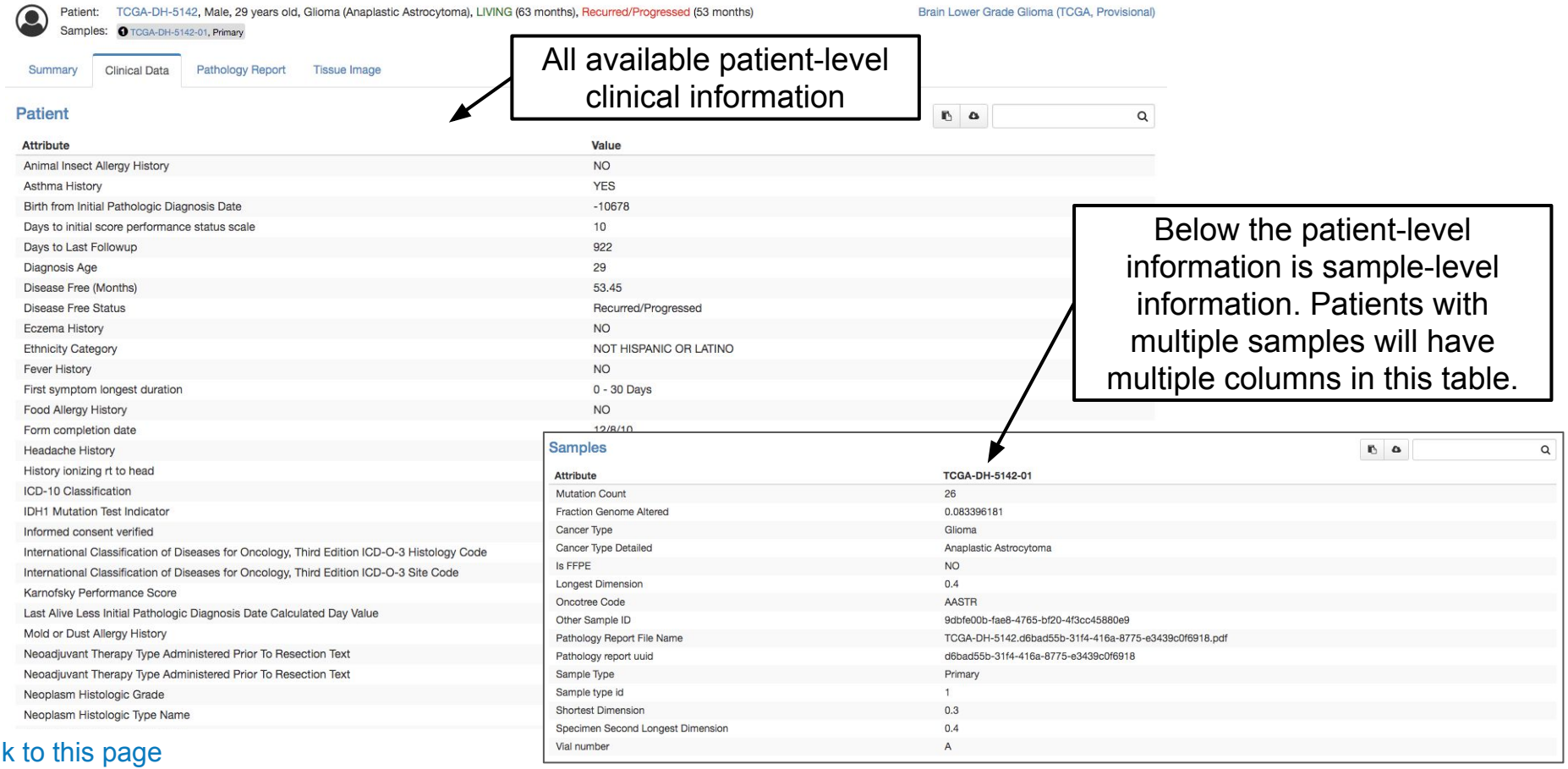
Lists of all called mutations and CNAs.

Copy, download, add/remove columns or search.

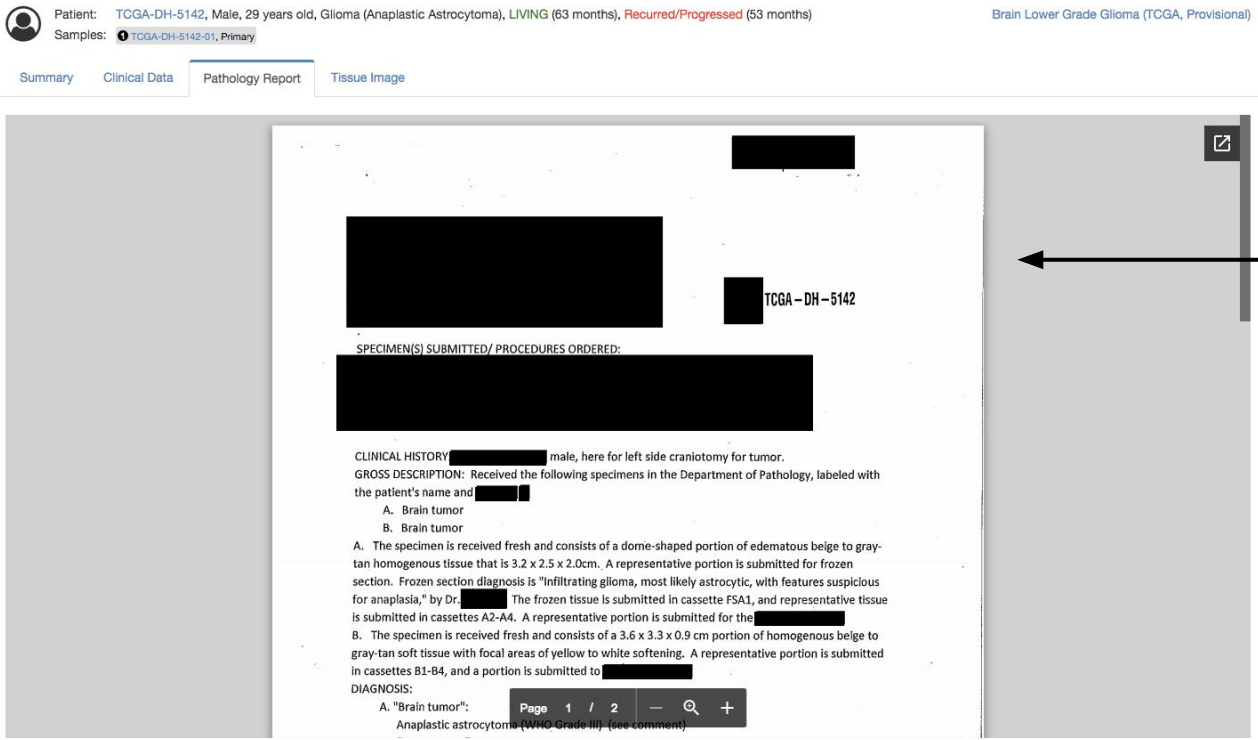


Link to this page

Patient View, Example 1: Clinical Data



Patient View, Example 1: Pathology Report



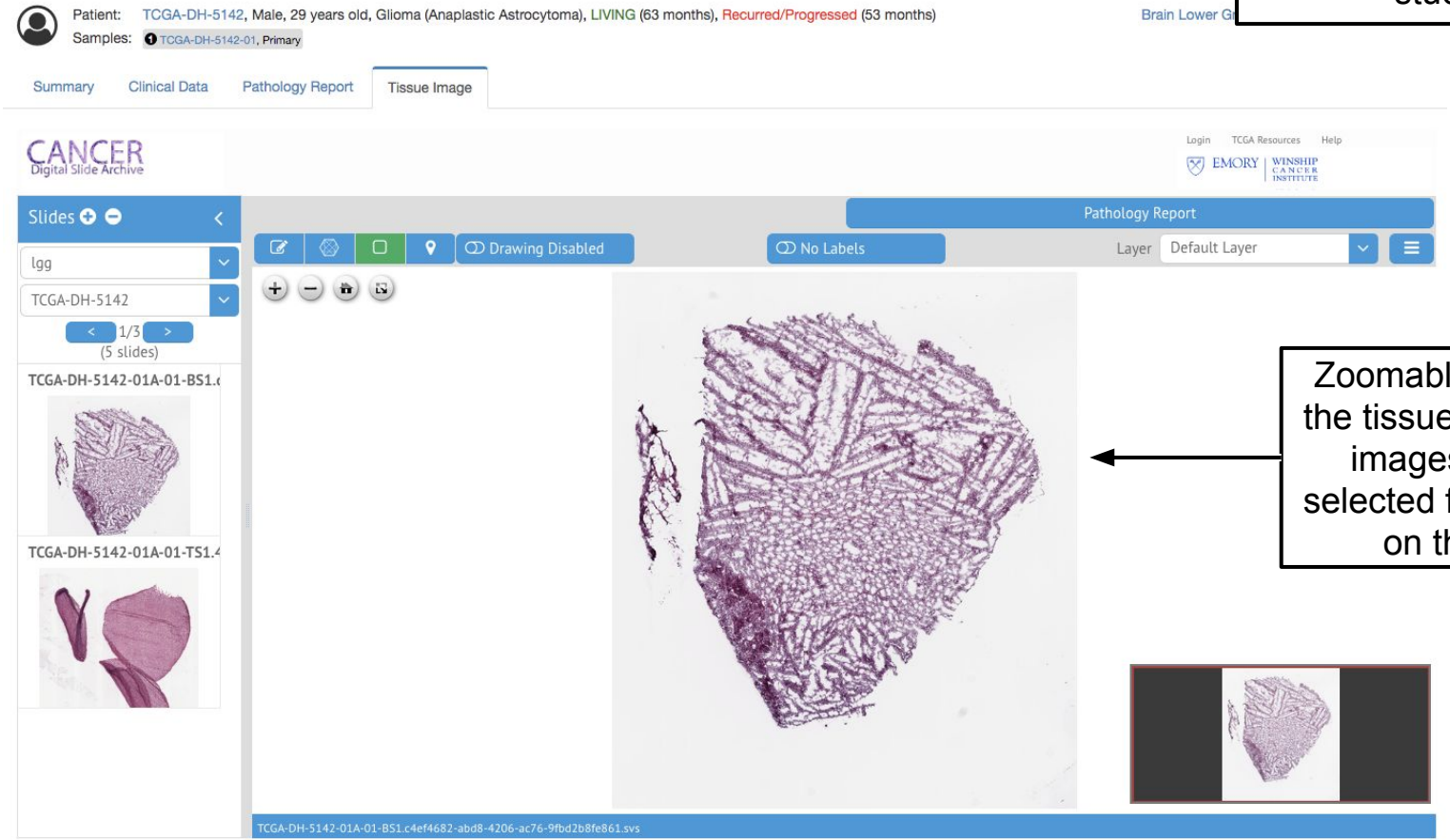
Note: Pathology Reports are only available for TCGA studies.

Original pathology report, de-identified.

Link to this page

Patient View, Example 1: Tissue Images

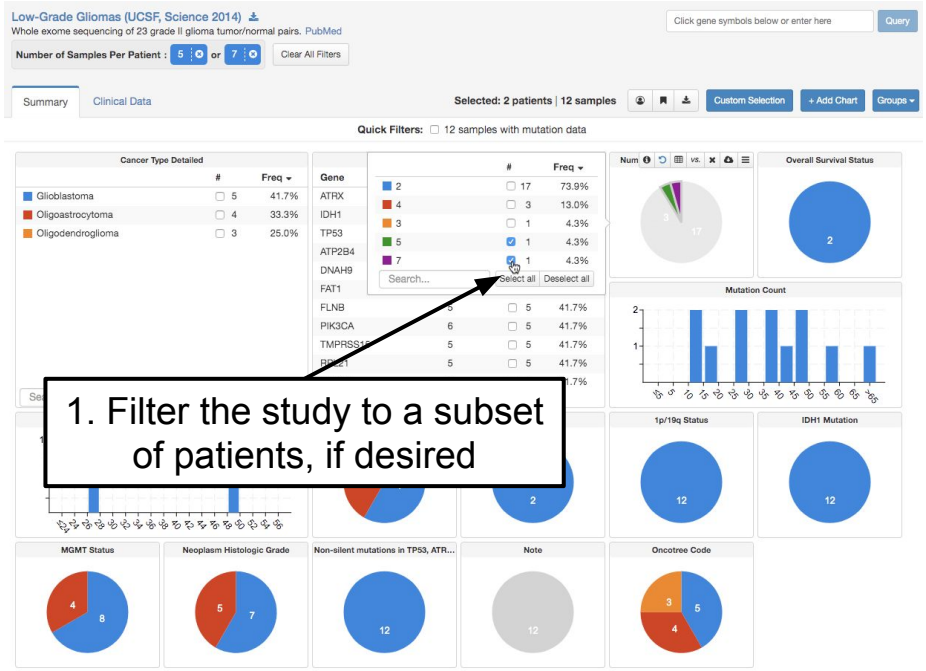
Note: Tissue images are only available for TCGA studies.

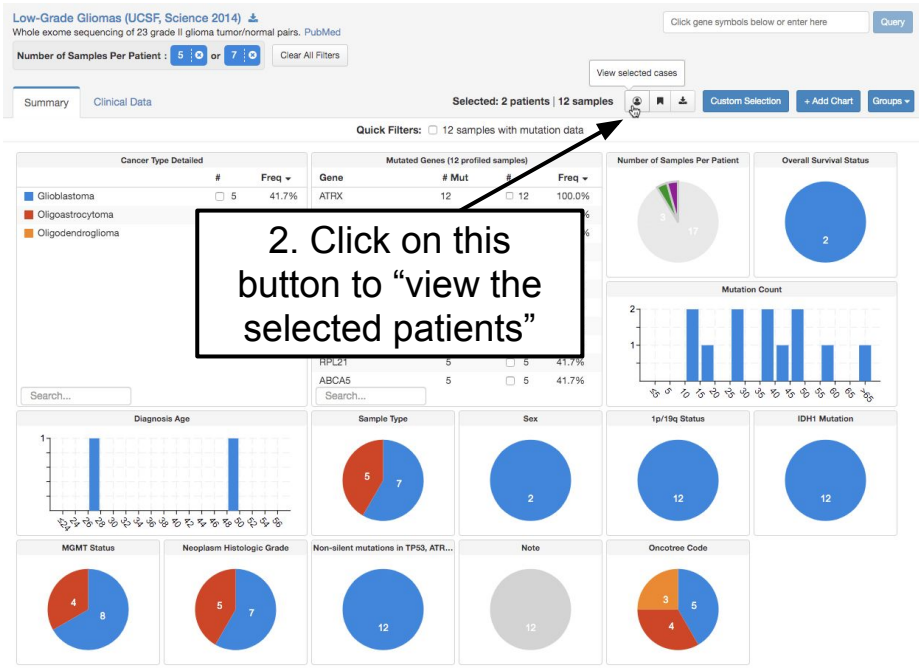


Zoomable image of the tissue. Additional images can be selected from the list on the left.

Link to this page

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





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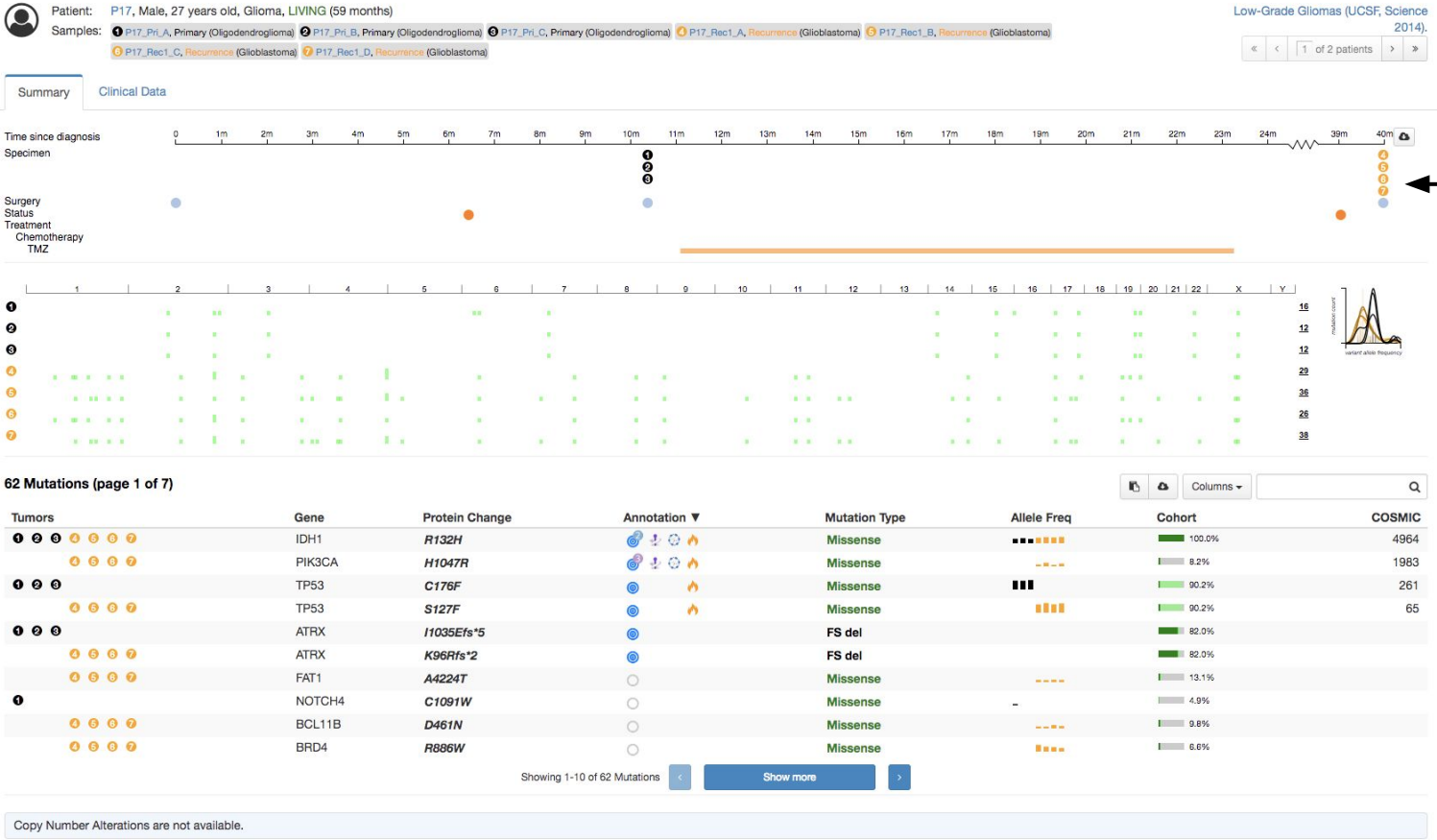
Patient View, Example 2: Patient Summary

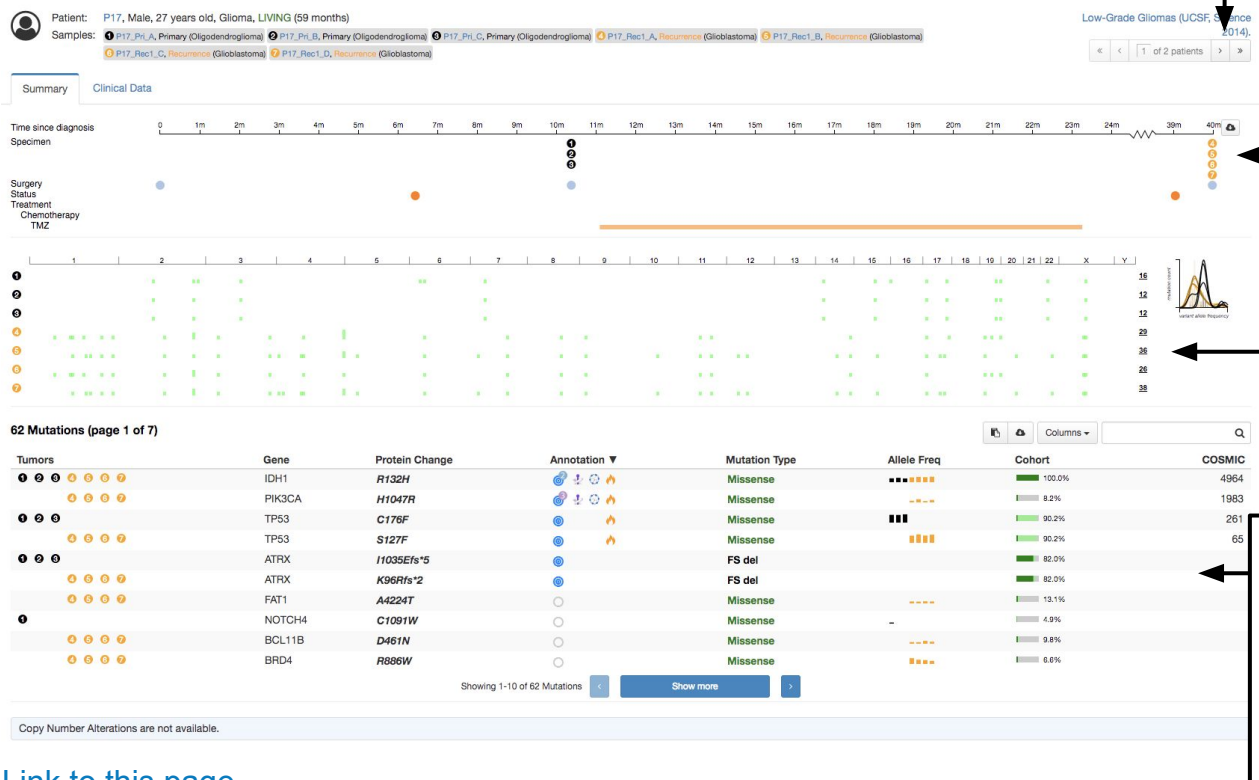


This study has multiple samples per patient and extensive clinical data to generate this enhanced patient timeline.

Link to this page

Patient View, Example 2: Patient Summary

Click to view the next patient



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

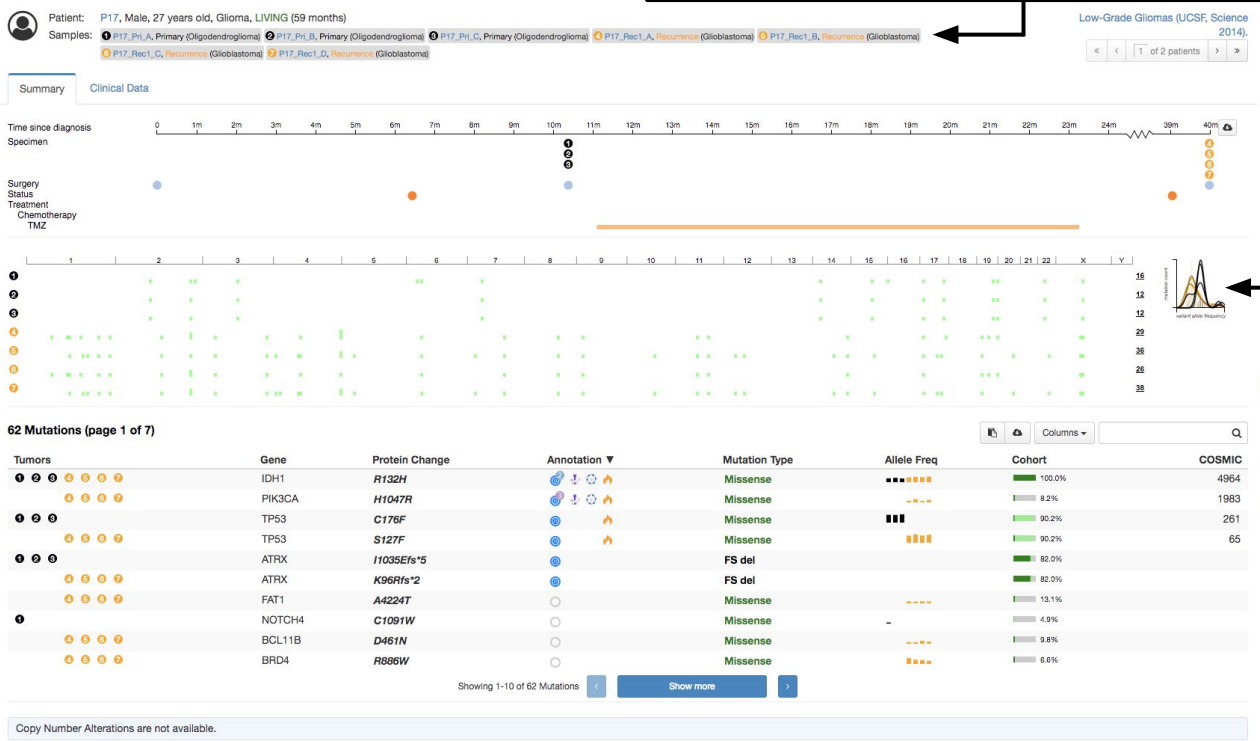
Figure showing distribution of mutations across the genome for each sample.

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

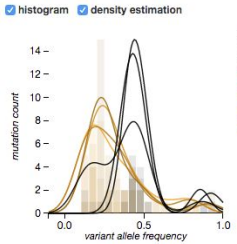
Link to this page

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)



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: Clinical Data

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 | Clinical Data

All available patient-level clinical information

Patient

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

All available sample-level 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	GB	GB	GB

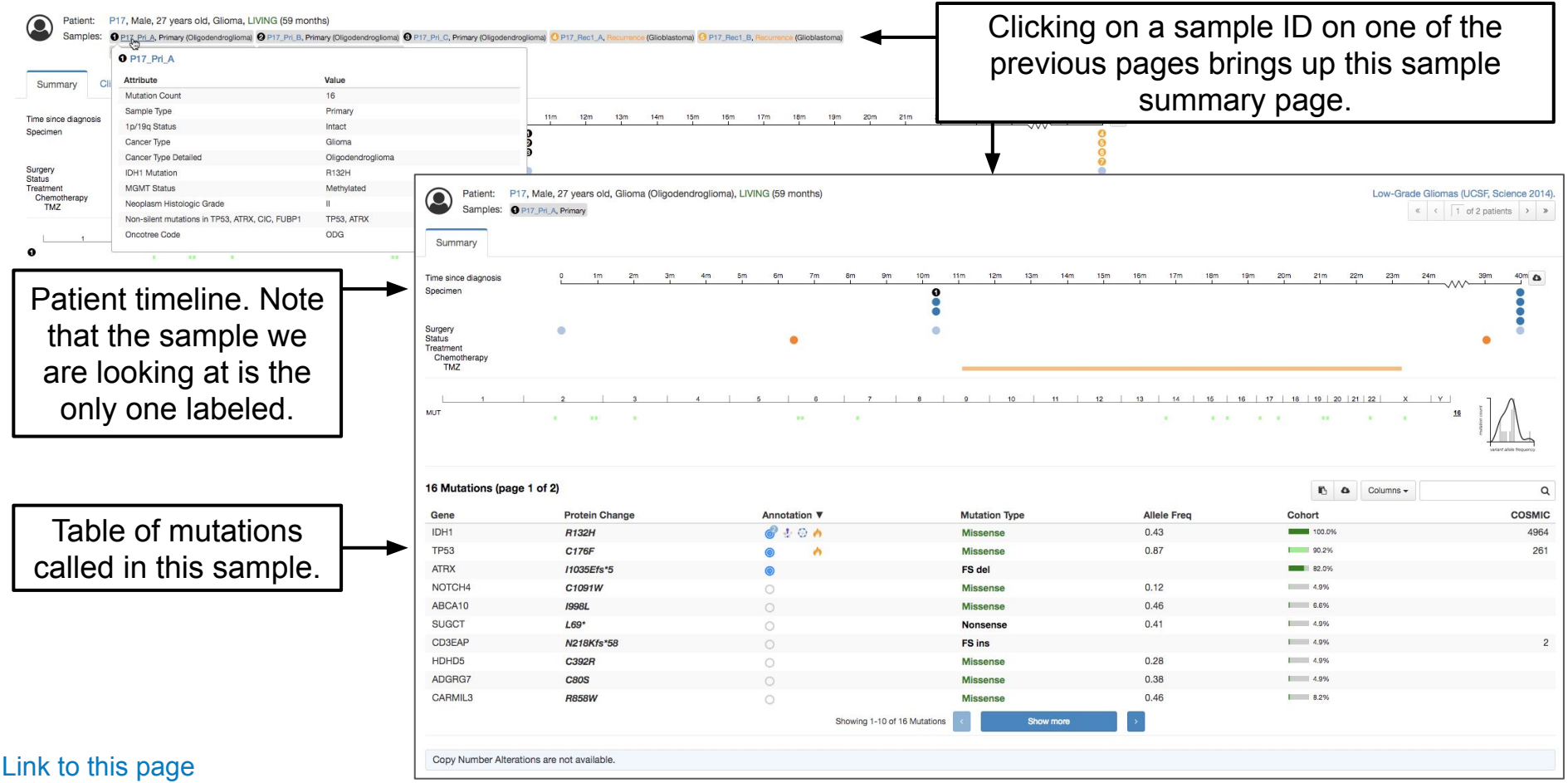
Link to this page

Patient View, Example 2: Sample Summary

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

Patient timeline. Note that the sample we are looking at is the only one labeled.

Table of mutations called in this sample.

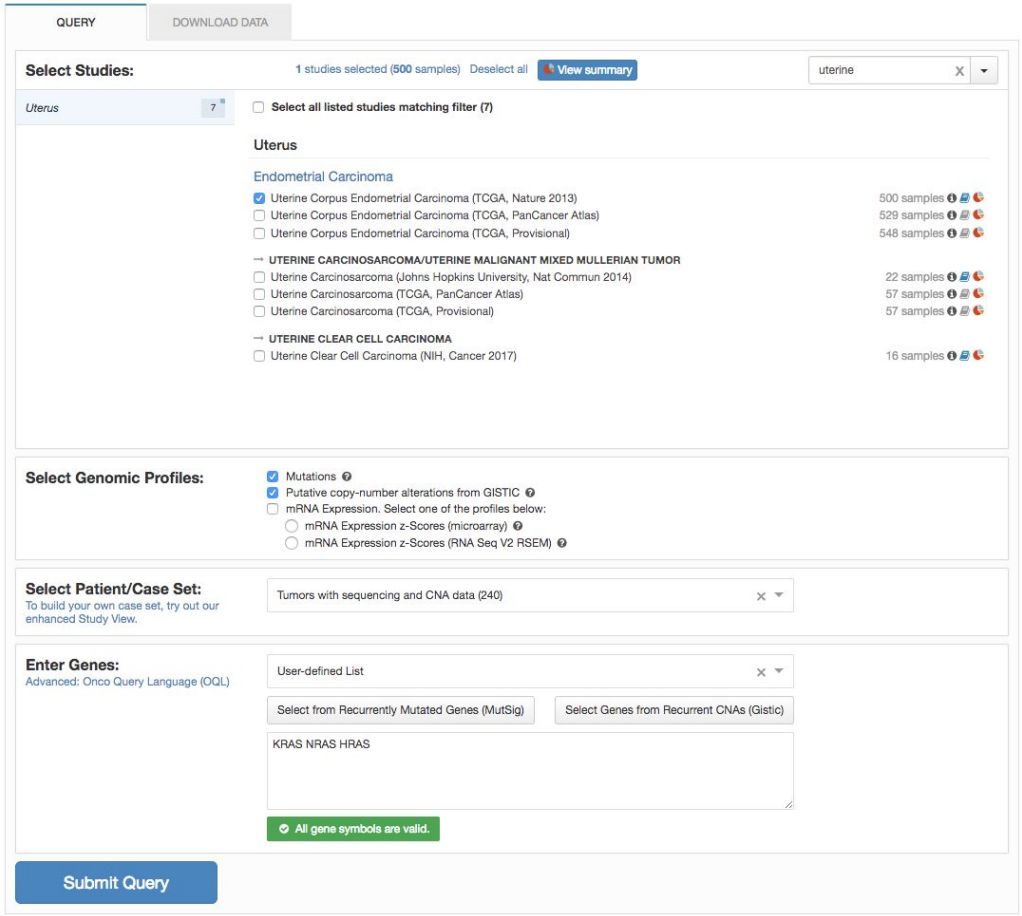


Link to this page

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

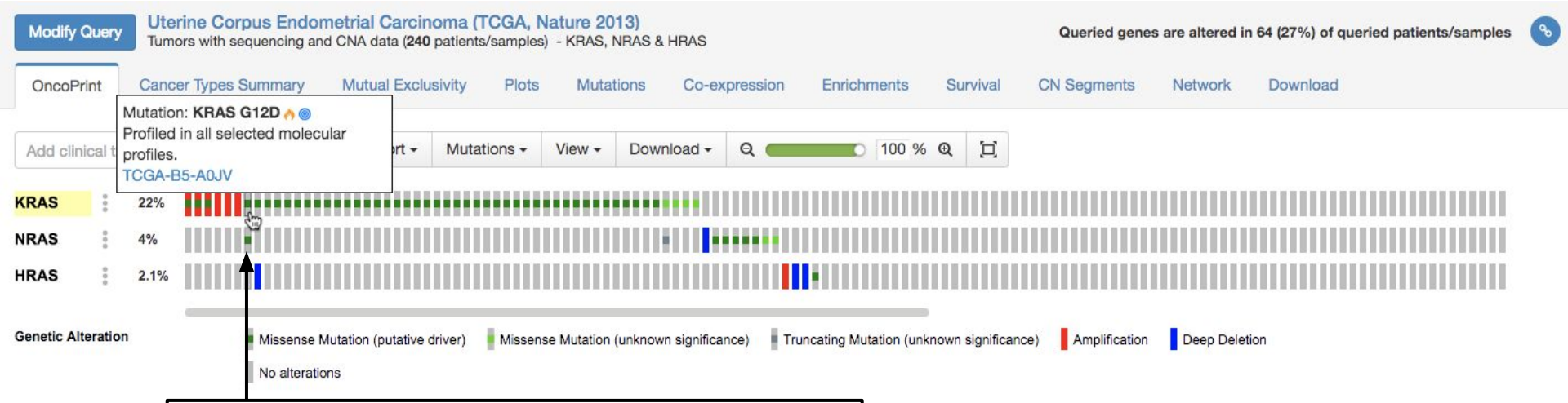
Let's look at RAS mutations in Uterine Corpus Endometrial Carcinoma (TCGA, Nature 2013).

Example 3: Run the query



Link to this page

Example 3: OncoPrint

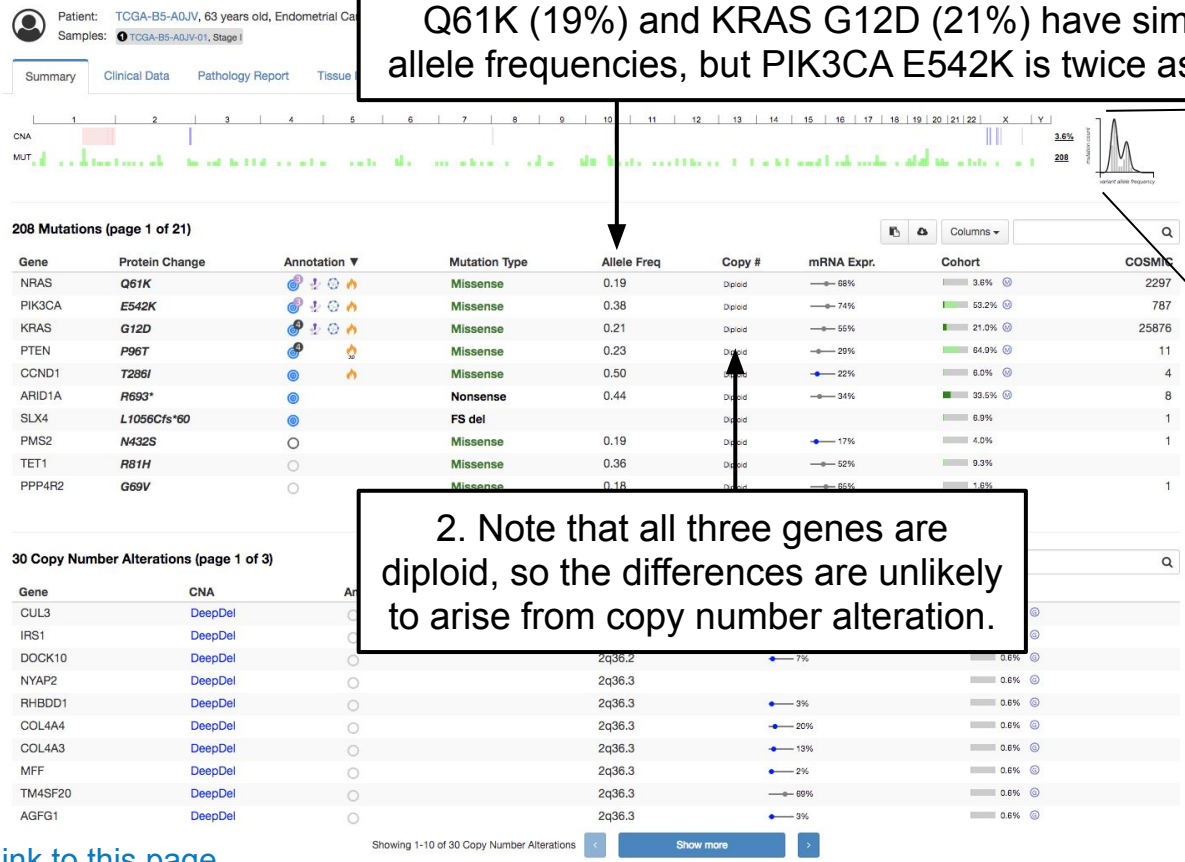


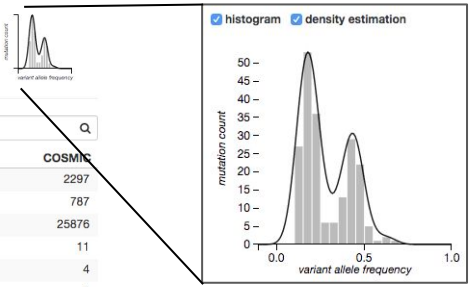
In general, mutations in these genes are mutually exclusive. However, there's one case with driver mutations in both KRAS and NRAS. Let's look at that patient in greater detail by clicking on the patient ID ("TCGA-B5-A0JV").

Link to this page

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.

Link to this page

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