cBioPortal Tutorial #3: Patient View

Investigate individual patients or samples in detail

Last update: December 30, 2020

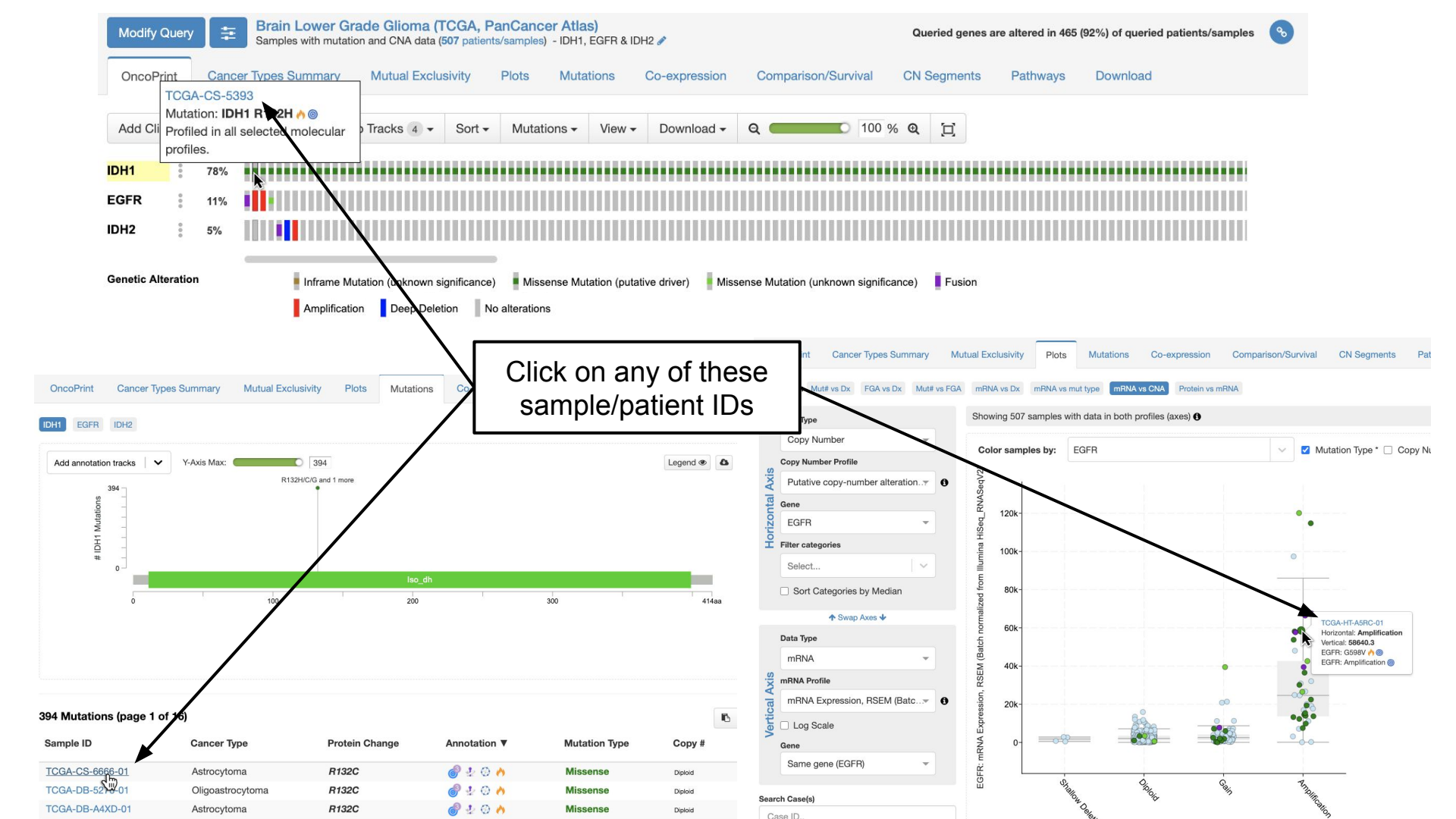
Tutorial Objectives

- 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
TCGA-CS-5393
Mutation: IDH1 R132H
Profiled in all selected molecular profiles.
Add 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
No alterations
Click on any of these sample/patient IDs
Add annotation tracks
Y-Axis Max:
394
Legend
R132H/C/G and 1 more
IDH1 Mutations
Iso_dh
414aa
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-5271-01
Oligoastrocytoma
R132C
Missense
Diploid
TCGA-DB-A4XD-01
Astrocytoma
R132C
Missense
Diploid
Horizontal Axis
Data Type
Copy Number
Copy Number Profile
Putative copy-number alteration...
Gene
EGFR
Filter categories
Select...
Sort Categories by Median
Swap Axes
Vertical Axis
Data Type
mRNA
mRNA Profile
mRNA Expression, RSEM (Batc...
Log Scale
Gene
Same gene (EGFR)
Search Case(s)
Case ID...
Mut# vs Dx
FGA vs Dx
Mut# vs FGA
mRNA vs Dx
mRNA vs mut type
mRNA vs CNA
Protein vs mRNA
Showing 507 samples with data in both profiles (axes)
Color samples by:
EGFR
Mutation Type *
Copy Nu
EGFR: mRNA Expression, RSEM (Batch normalized from Illumina HiSeq_RNASeqV2)
120k
100k
80k
60k
40k
20k
0
Shallow Deler...
Diploid
Gain
Amplification
TCGA-HT-A5RC-01
Horizontal: Amplification
Vertical: 58640.3
EGFR: G598V
EGFR: 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, 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

Cancer Type Detailed : Oligoastrocytoma | Clear All Filters

View selected cases

Summary | Clinical Data | CN Segments | Custom Selection | Charts | Groups

1. Filter to a subset of patients, if desired

2. Click on this button to view the selected patients

Cancer Type

	#	Freq
Diffuse Glioma	130	100.0%

Cancer Type Detailed

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

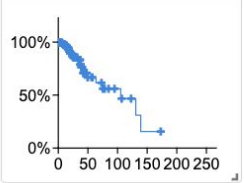
Search...

Genomic Profile Sample Counts

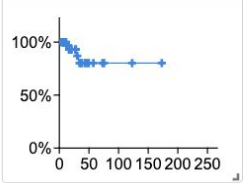
Profile	#	Freq
mRNA Expression, RSEM (Batch ...	130	100.0%
mRNA expression z-scores relativ...	130	100.0%
Mutations	130	100.0%
mRNA expression z-scores relativ...	130	100.0%
Microbiome Signatures (log RNA ...	130	100.0%
Putative copy-number alterations ...	129	99.2%
Log2 copy-number values	129	99.2%
Protein expression (RPPA)	114	87.7%
Protein expression z-scores (RPPA)	114	87.7%

Search...

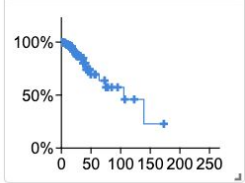
KM Plot: Overall Survival



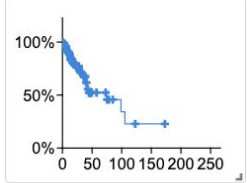
KM Plot: Disease Free Survival



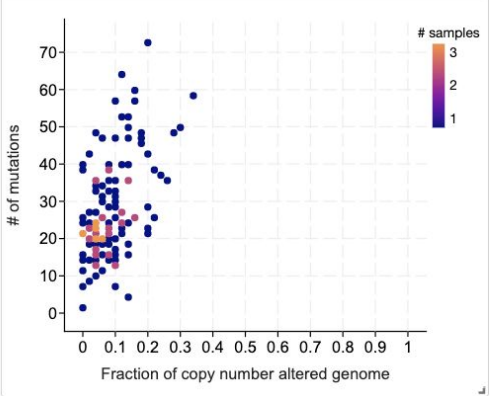
KM Plot: Disease-specific Survival



KM Plot: Progression Free Survival



Mutation Count vs Fraction of Genome Altered



Mutated Genes (130 profiled samples)

Gene	# Mut	#	Freq
IDH1	109	109	83.8%
TP53	104	75	57.7%
ATRX	72	66	50.8%
CIC	25	22	16.9%
FUBP1	10	9	6.9%
ARID1A	9	8	6.2%
EGFR	9	7	5.4%
NF1	9	7	5.4%
NOTCH1	6	6	4.6%
PIK3CA	6	6	4.6%
BCOR	7	6	4.6%

Search...

Fusion Genes

Gene	# Fusion	#	Freq
EGFR	2	2	1.5%
FGFR3	2	2	1.5%
ATF1	1	1	0.8%
ERG	1	1	0.8%
GLI1	1	1	0.8%
MET	1	1	0.8%
MRE11	1	1	0.8%
MSH3	1	1	0.8%
PPARG	1	1	0.8%
MAP2K2	1	1	0.8%
PTEN	1	1	0.8%

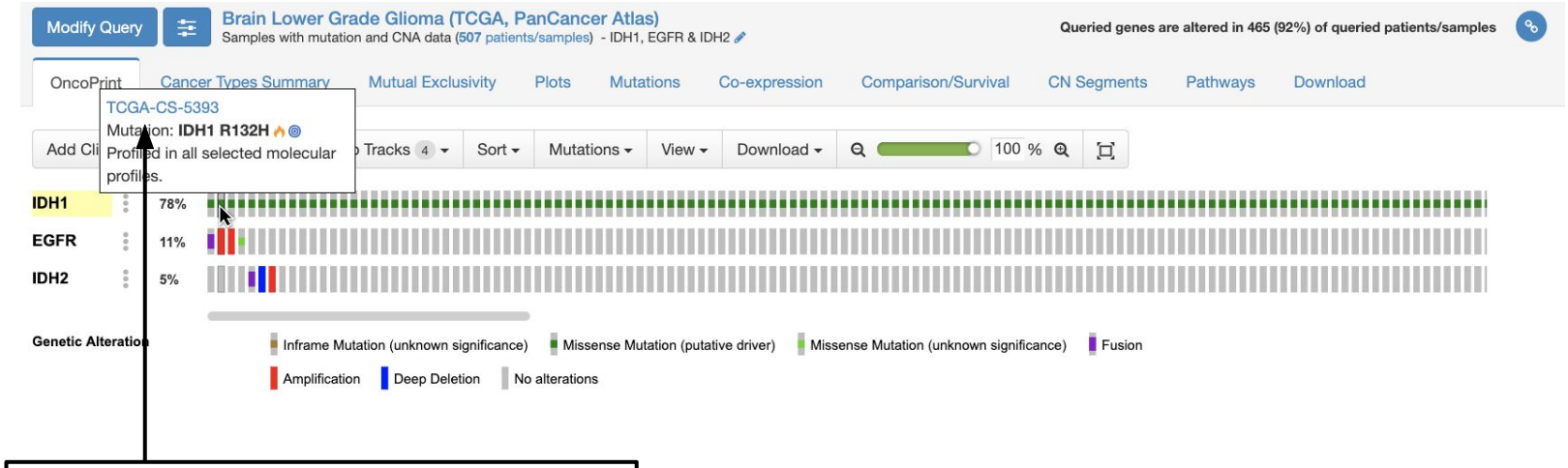
Search...

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.

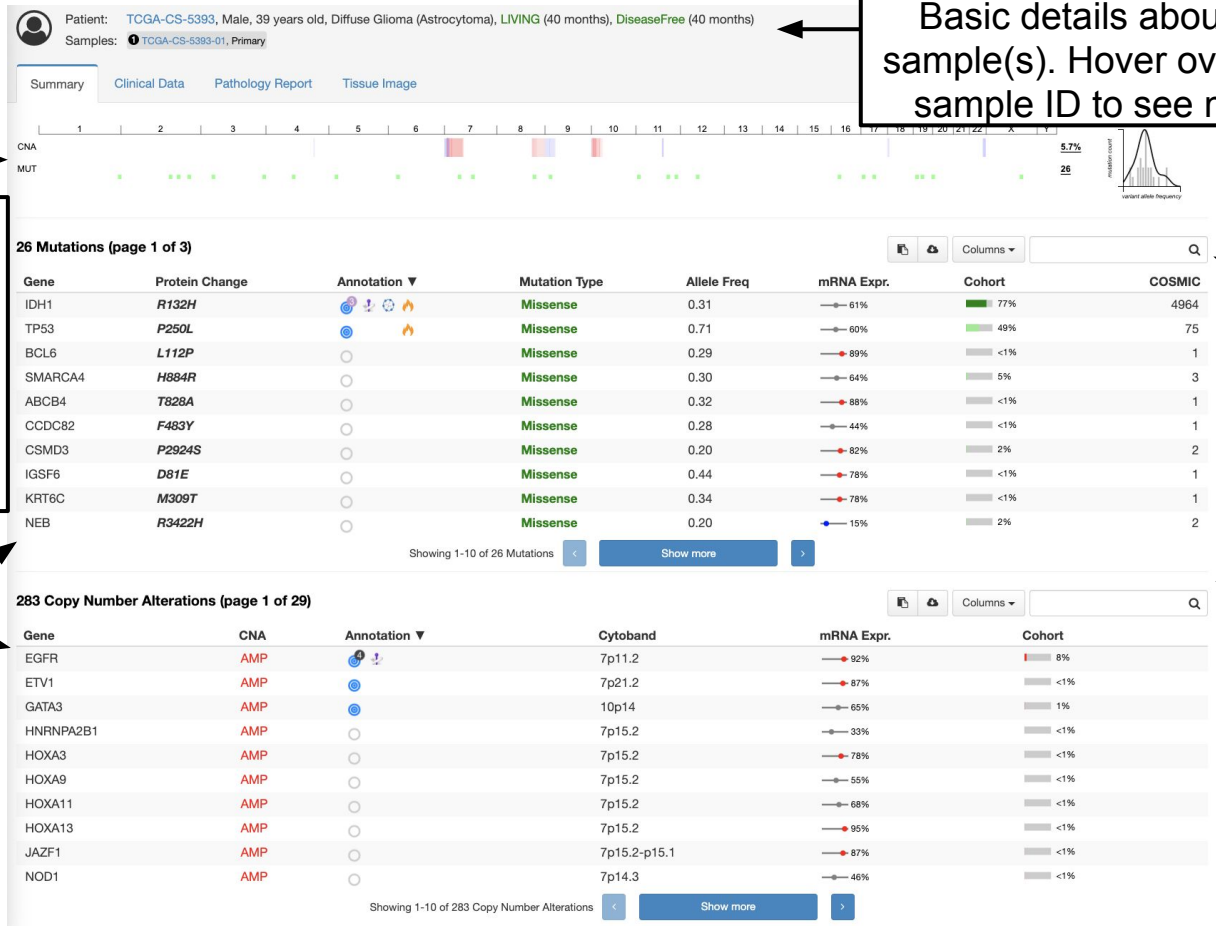
Example 1: Brain Lower Grade Glioma (TCGA, PanCancer Atlas)



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.

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.

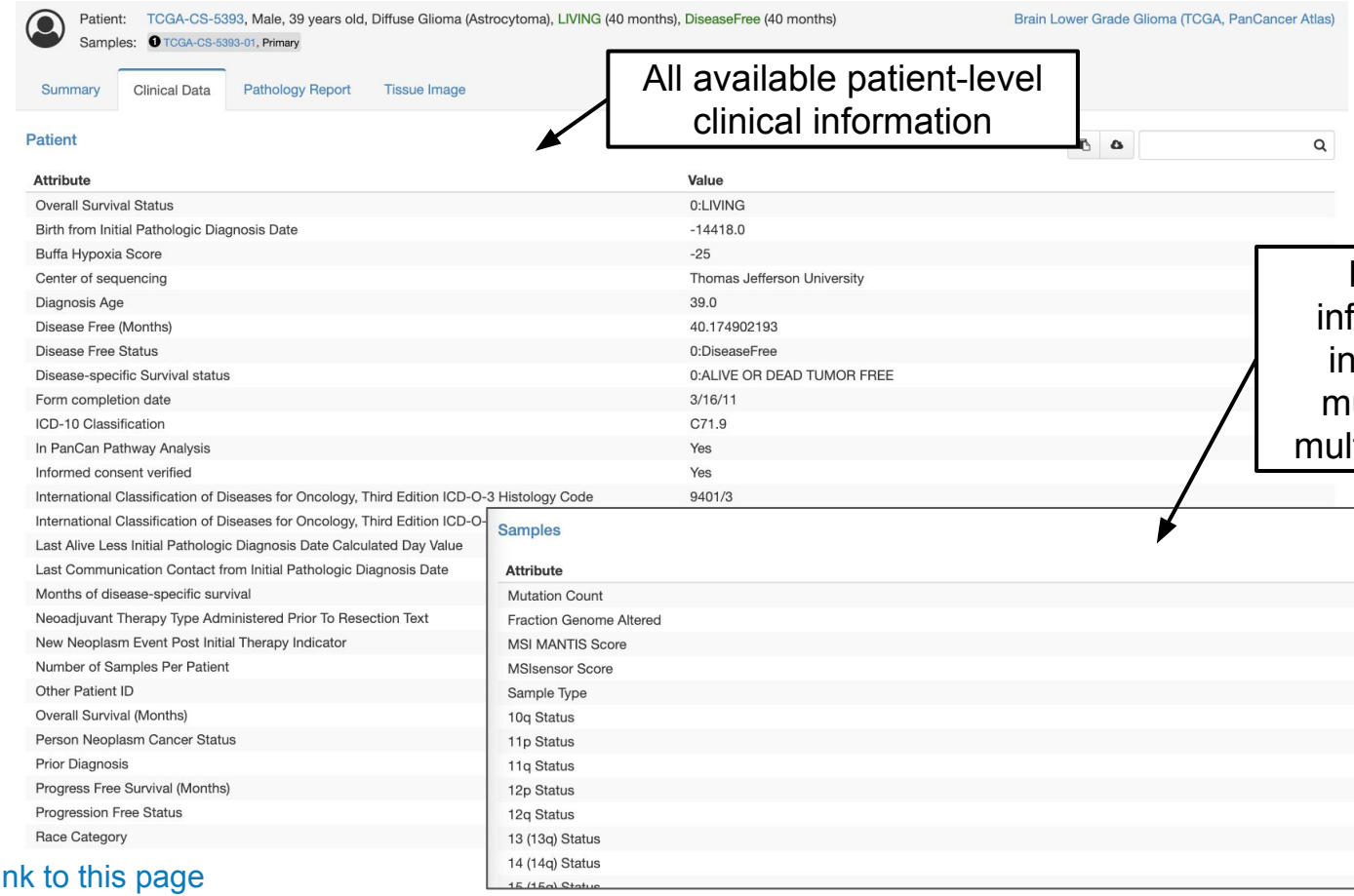
Figure showing where called CNA and mutations are across the genome. Hover over any of these for more details.

Copy, download, add/remove columns or search.

Lists of all called mutations and CNAs.

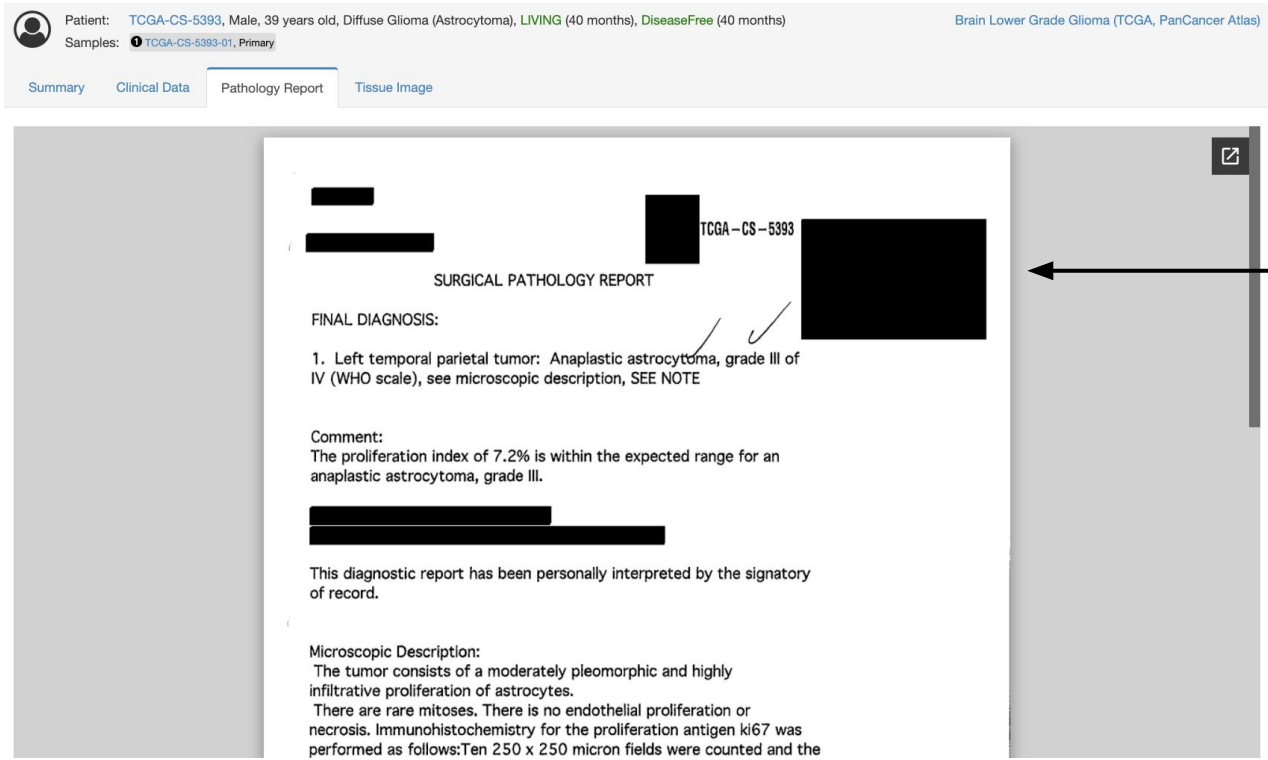
Link to this page

Patient View, Example 1: Clinical Data



Link to this page

Patient View, Example 1: Pathology Report

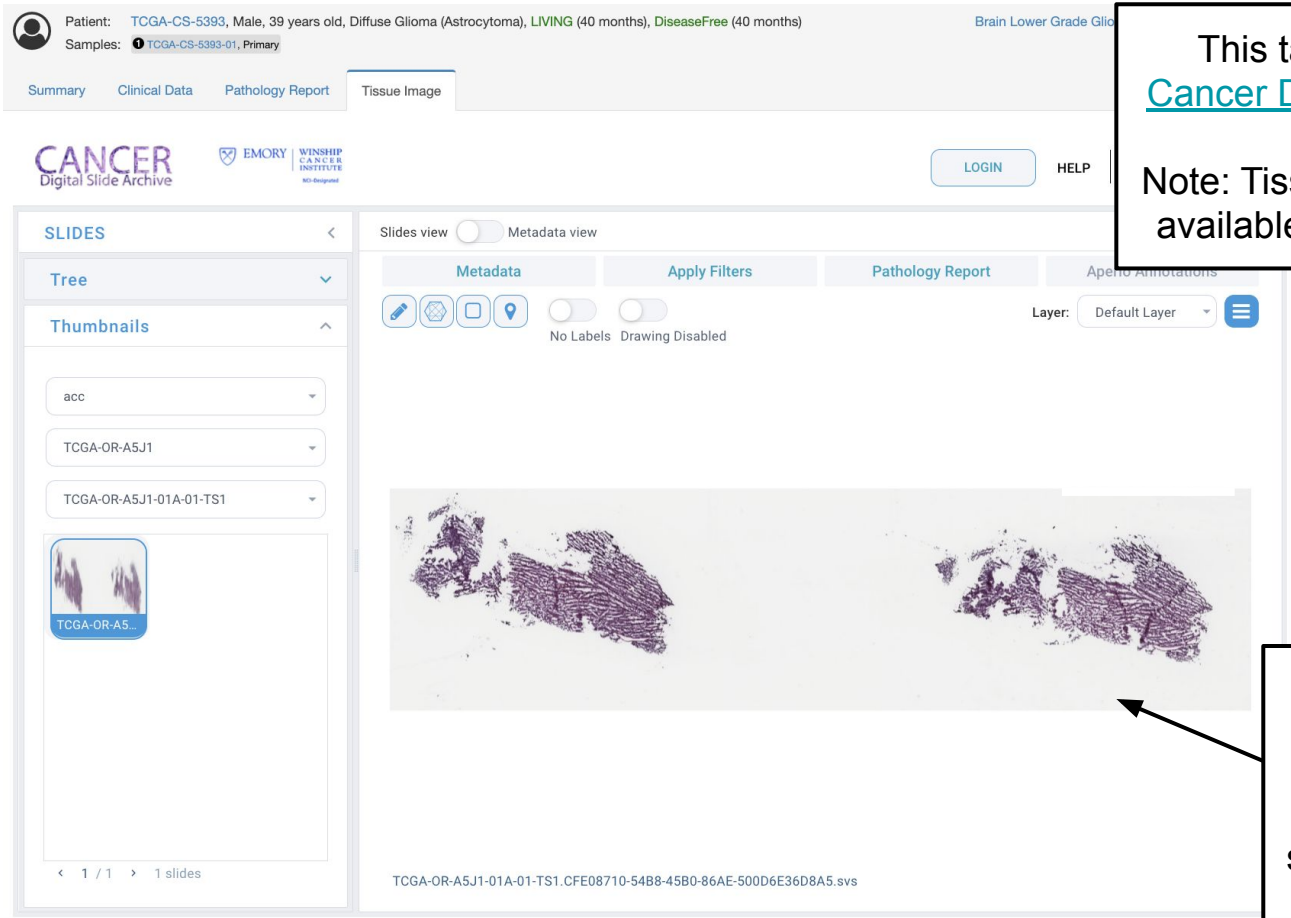


Note: Pathology Reports are only available for TCGA studies.

Original pathology report, de-identified.

Link to this page

Patient View, Example 1: Tissue Image



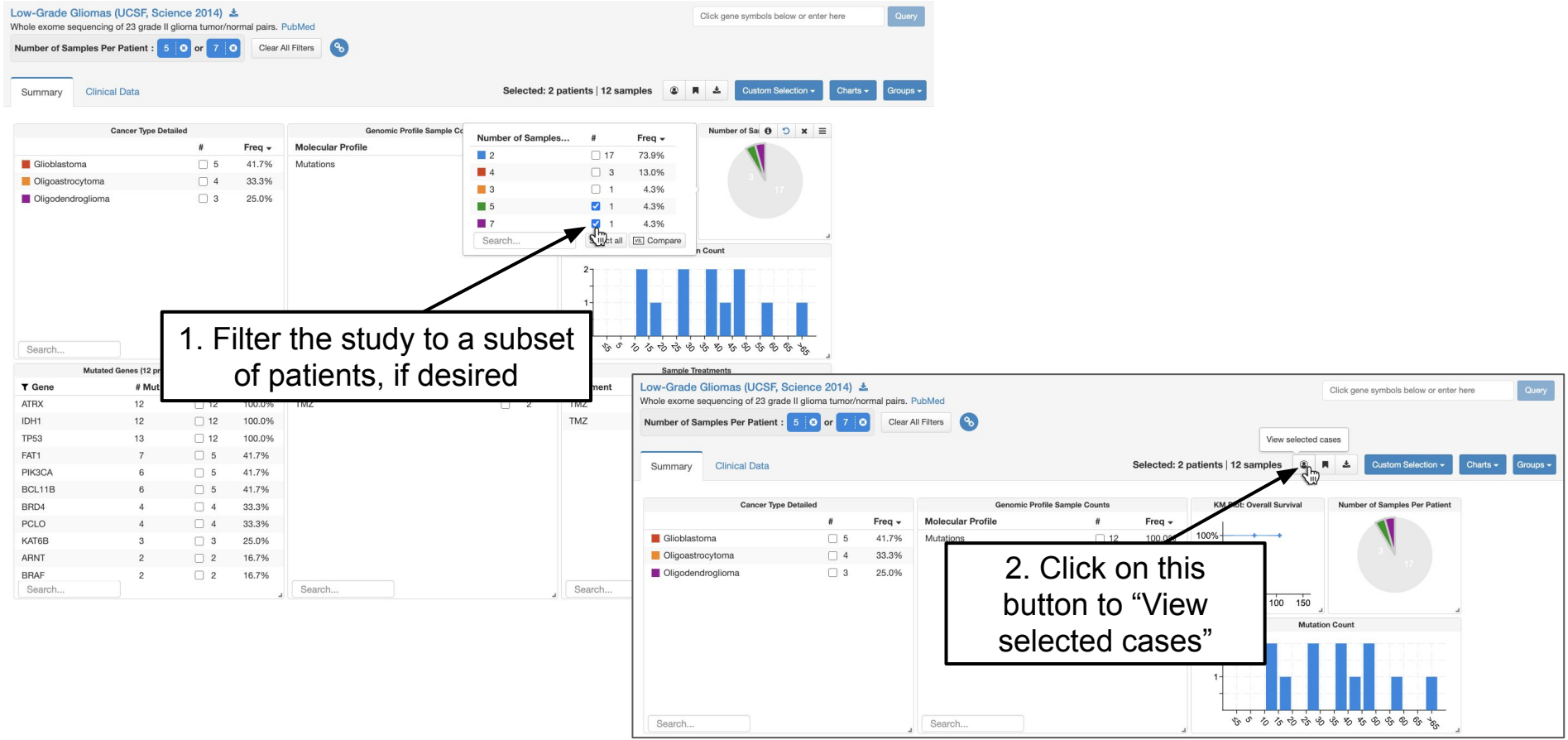
This tab integrates the Cancer Digital Slide Archive.

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.

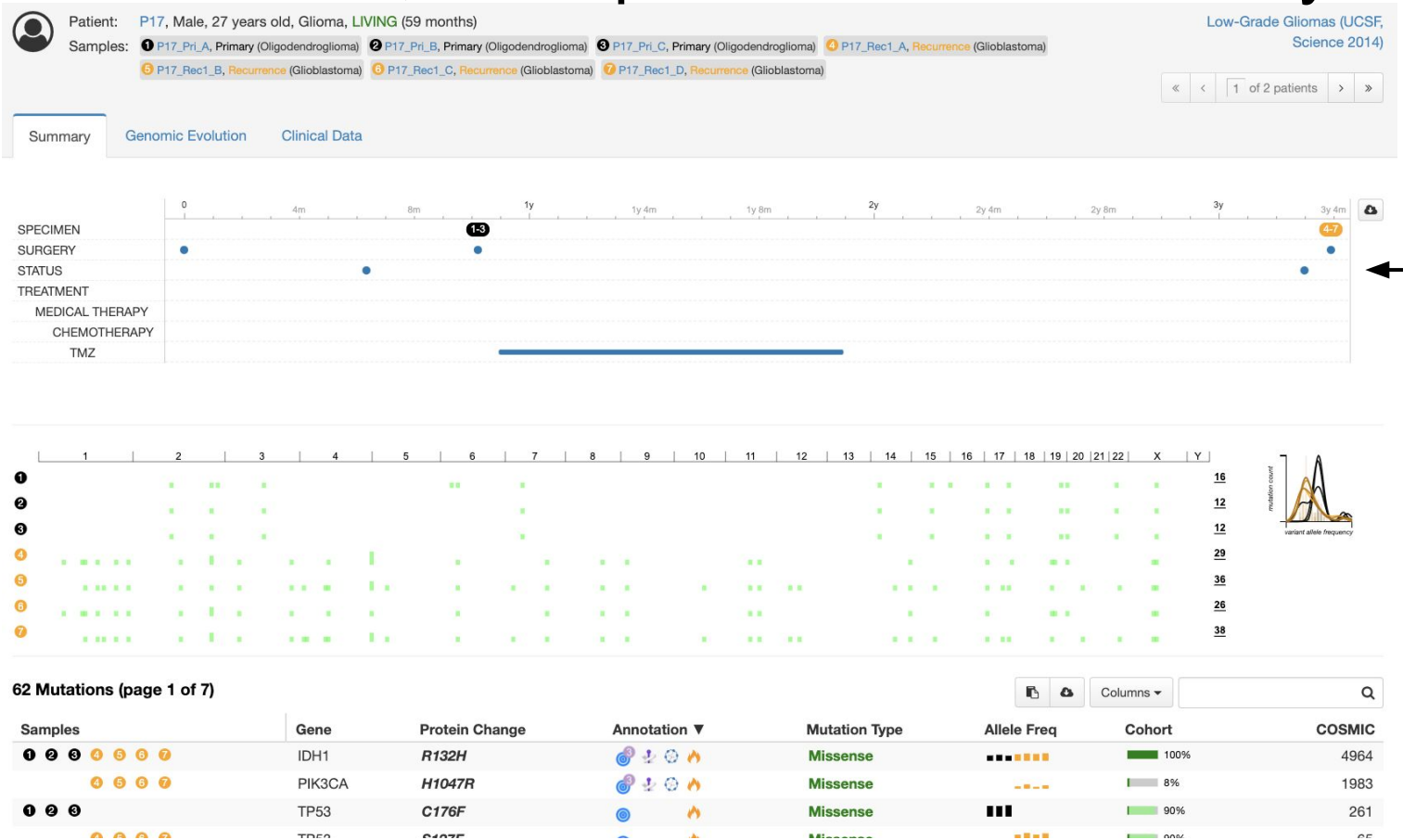
Link to this page

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



Link to this page

Patient View, Example 2: Patient Summary

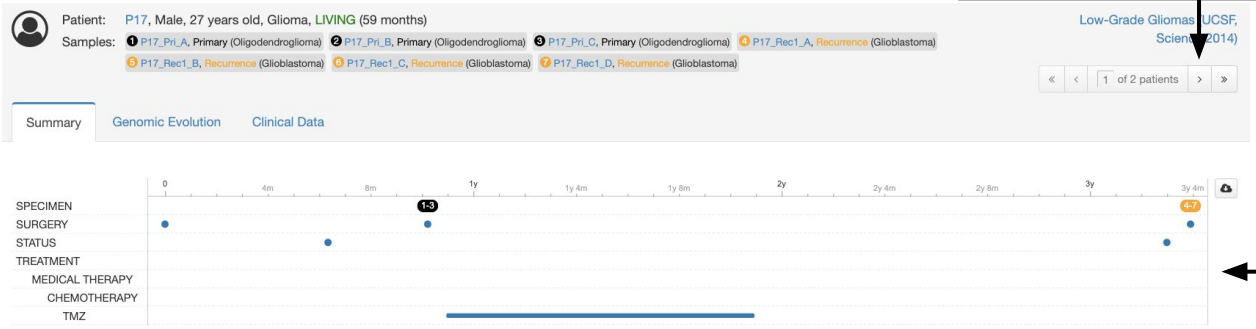


This study has multiple samples per patient and extensive clinical data to generate this enhanced patient timeline.

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.

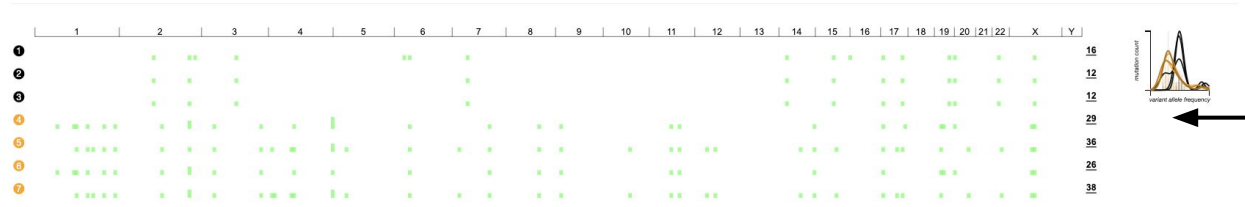
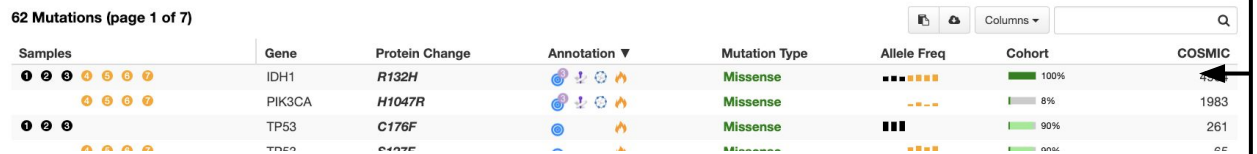


Figure showing distribution of mutations across the genome for each sample.

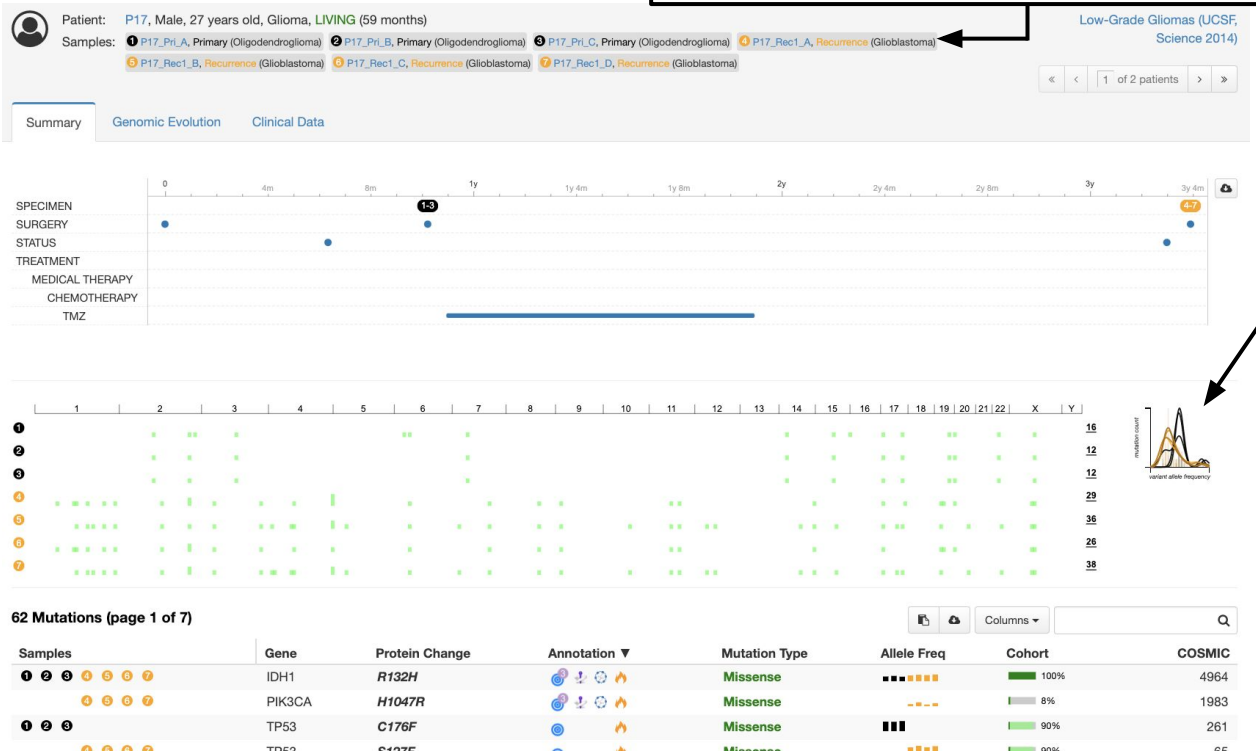


List of all mutations called. The first column (“Samples”) shows which samples had a particular mutation. The Allele Freq column depicts the mutation frequency in each sample by the height of the bar.

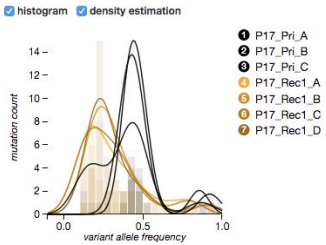
Link to this page

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: Genomic Evolution

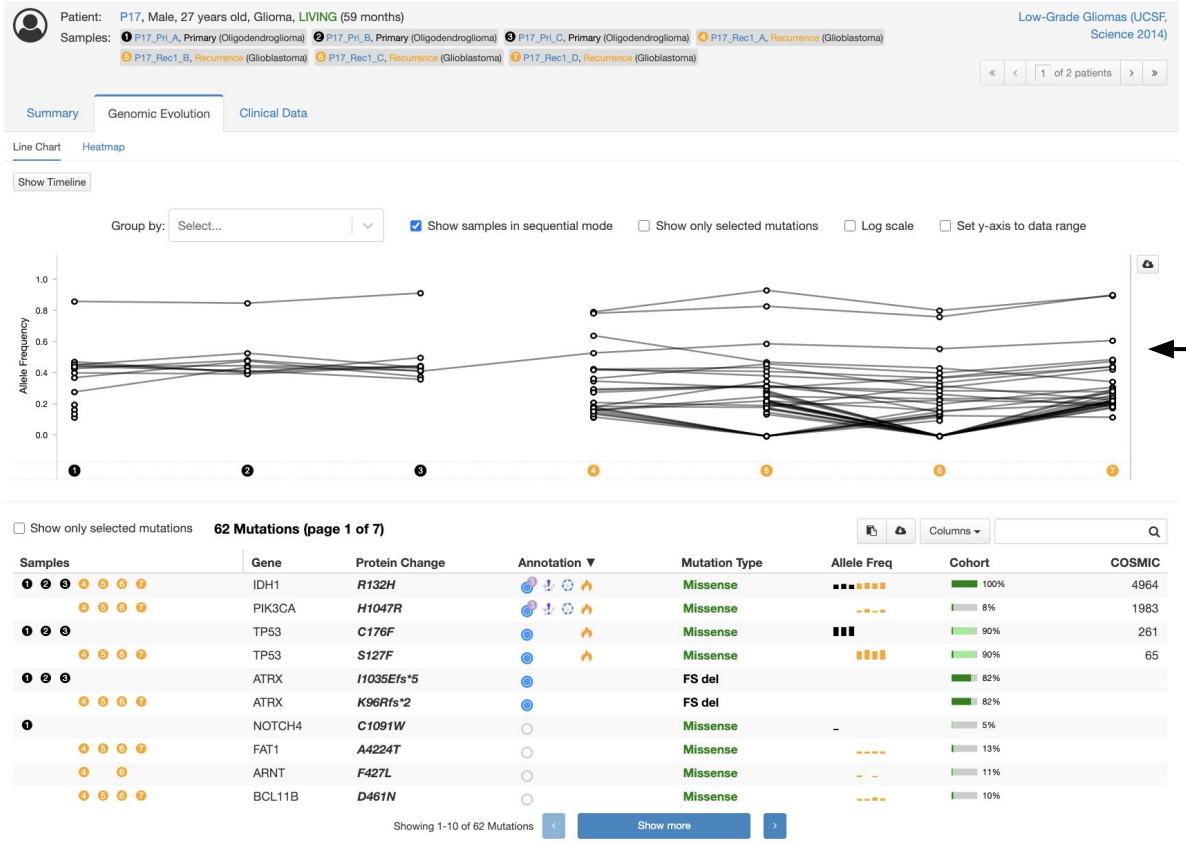
Allele frequencies can be displayed as a Line Chart or Heatmap

Click to view the timeline above the allele frequency visualization

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.

Link to this page

Patient View, Example 2: Genomic Evolution - Line Chart



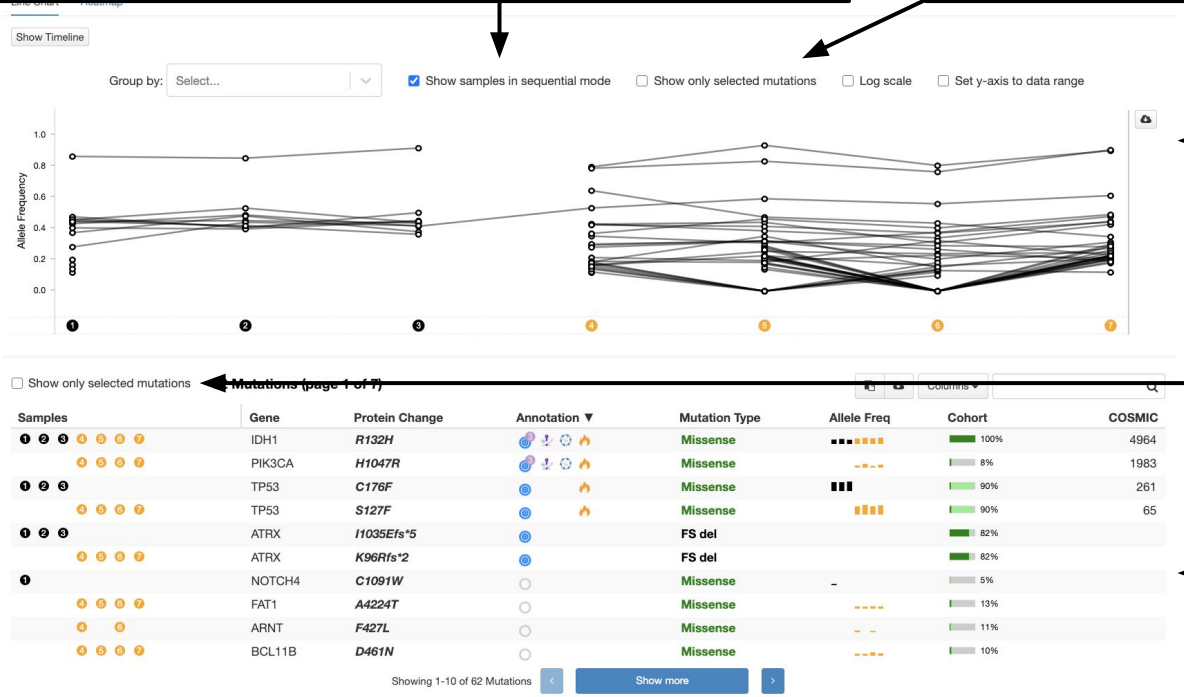
Each dot represents the allele frequency of a mutation in a sample. Lines connect mutations that are detected in multiple samples. Options above the chart enable customization.

Link to this page

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.



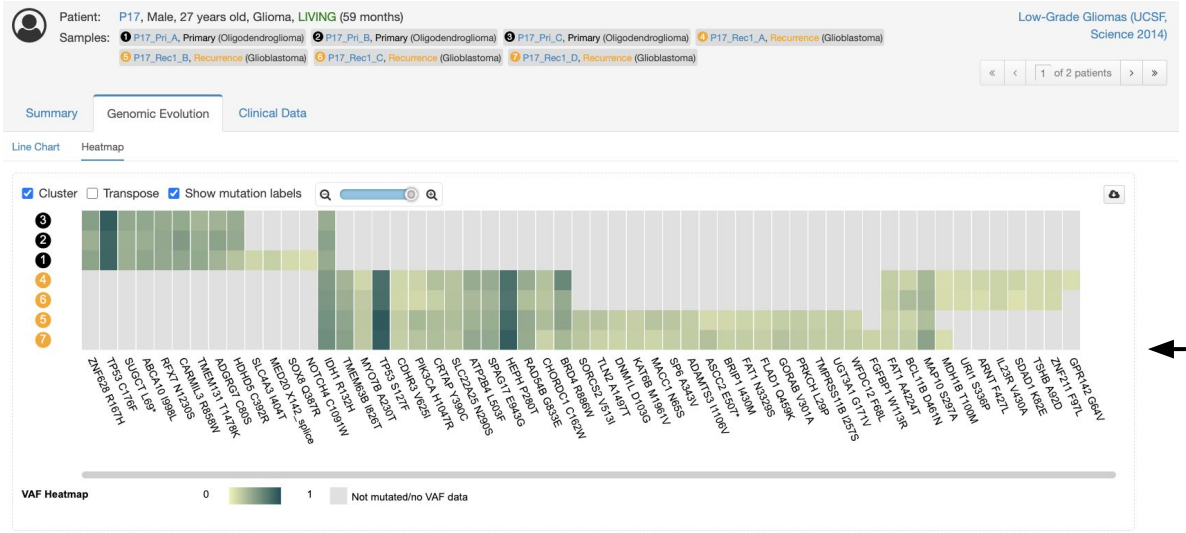
This chart and the mutation table are linked - hover or click on a mutation in the chart to see it highlighted in the table below.

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.

Link to this page

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)**

Columns

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	*S127F*		Missense		90%	65
1 2 3	ATRX	*I1035Efs*5*		FS del		82%	
4 5 6 7	ATRX	*K96Rfs*2*		FS del		82%	
1	NOTCH4	*C1091W*		Missense		5%	
4 5 6 7	FAT1	*A4224T*		Missense		13%	
4 5	ARNT	*F427L*		Missense		11%	
4 5 6 7	BCL11B	*D461N*		Missense		10%	

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

Link to this page

Patient View, Example 2: 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

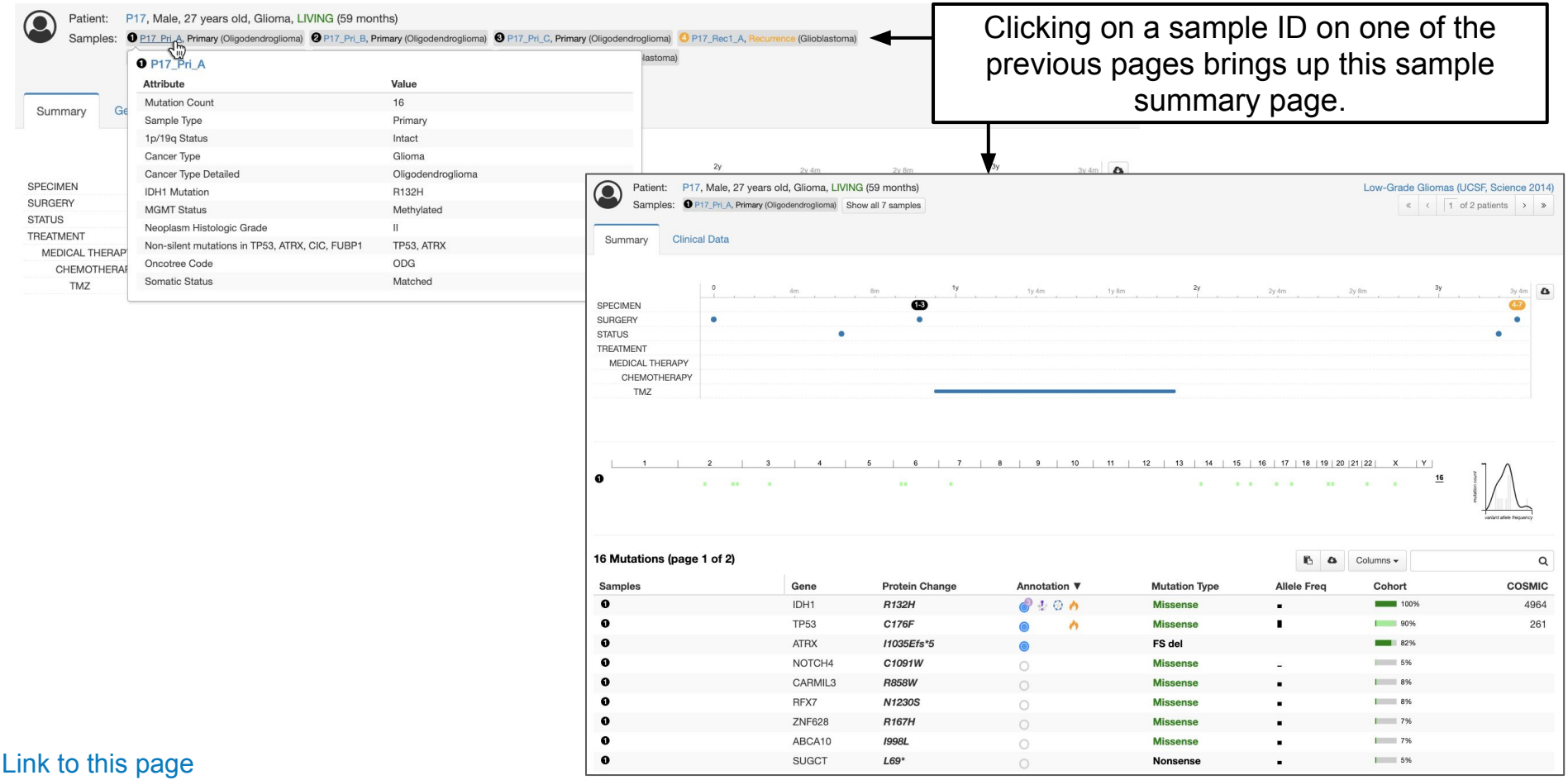
When available, the data used to populate the timeline in the Summary tab is shown here.

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

Link to this page

Patient View, Example 2: Sample Summary

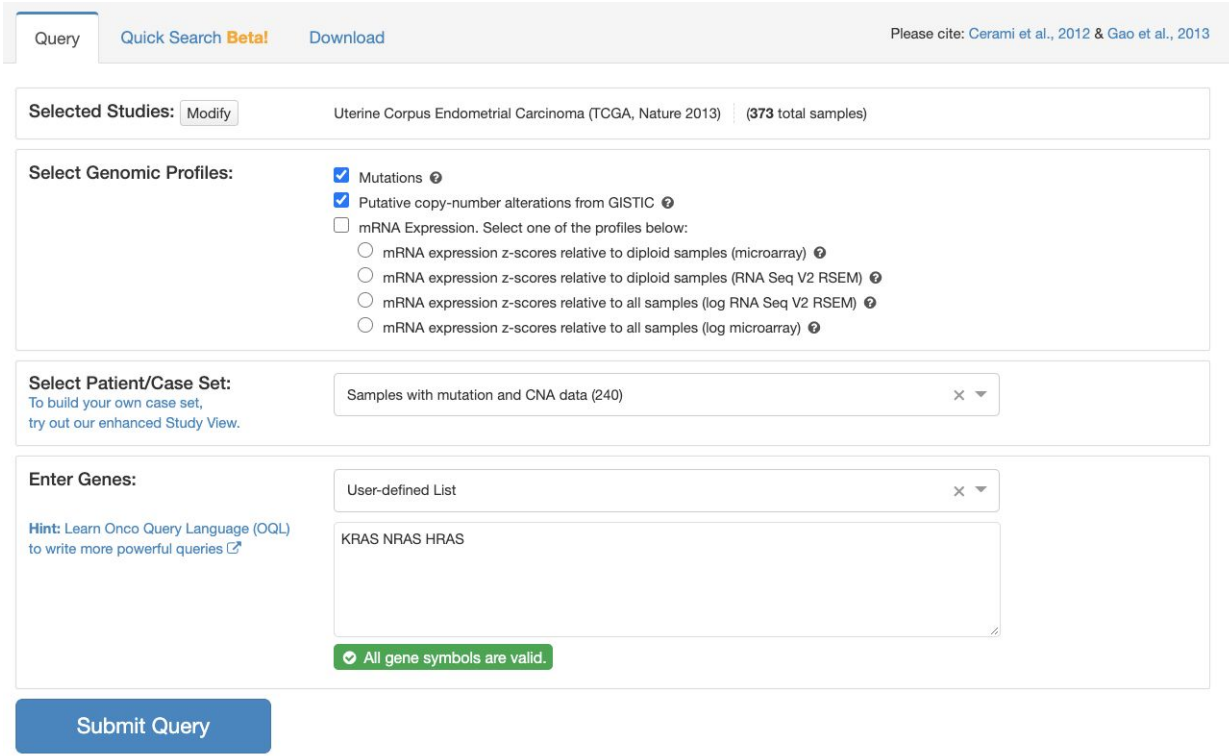


Link to this page

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

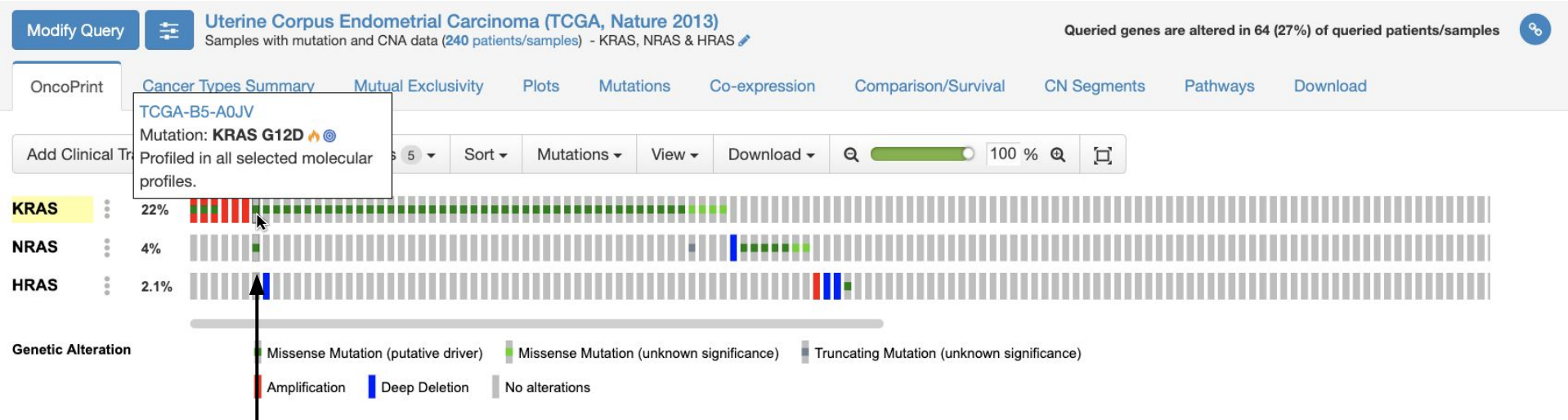
Let's look at RAS mutations in Uterine Corpus Endometrial Carcinoma (TCGA, Nature 2013).

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%).

Patient: TCGA-B5-A0JV, Female, 63 years old, End
(22 months)
Samples: TCGA-B5-A0JV-01, Primary, Stage I

Summary | Clinical Data | Pathology Report | Tissu

CNA 3.6%
MUT 208

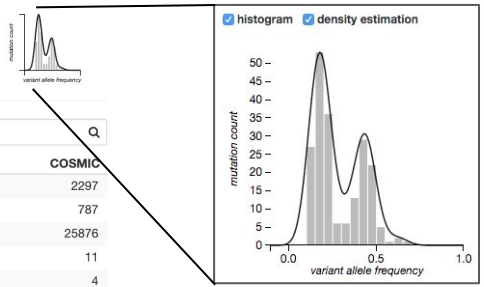
208 Mutations (page 1 of 21)

Columns

Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Copy #	mRNA Expr.	Cohort	COSMIC
NRAS	*Q61K*		Missense	0.19	Diploid	68%	4%	2297
PIK3CA	*E542K*		Missense	0.38	Diploid	74%	53%	787
KRAS	*G12D*		Missense	0.21	Diploid	55%	21%	25876
PTEN	*P96T*		Missense	0.23	Diploid	29%	65%	11
CCND1	*T286I*		Missense	0.50	Diploid	22%	6%	4
ARID1A	*R693**		Nonsense	0.44	Diploid	34%	33%	8
SLX4	*L1056Cfs*60*		FS del		Diploid		7%	1
PMS2	*N432S*		Missense	0.19	Diploid	17%	4%	1
PTPRB	*L853P*		Missense	0.21	Diploid	26%	8%	1
PTPRS	*S1104G*							1

30 Copy Number Alterations (page 1 of 3)

Gene	CNA				
CUL3	DeepDel				
IRS1	DeepDel		2q36.3	35%	<1%
CCNB3	DeepDel		Xp11.22	2%	<1%
DOCK10	DeepDel		2q36.2	7%	<1%
NYAP2	DeepDel		2q36.3		<1%
RHBDD1	DeepDel		2q36.3	3%	<1%
COL4A4	DeepDel		2q36.3	20%	<1%
COL4A3	DeepDel		2q36.3	13%	<1%



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?
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cbioportal@googlegroups.com