cBioPortal Tutorial #3: Patient View

Investigate individual patients or samples in detail

Last update: December 22, 2021

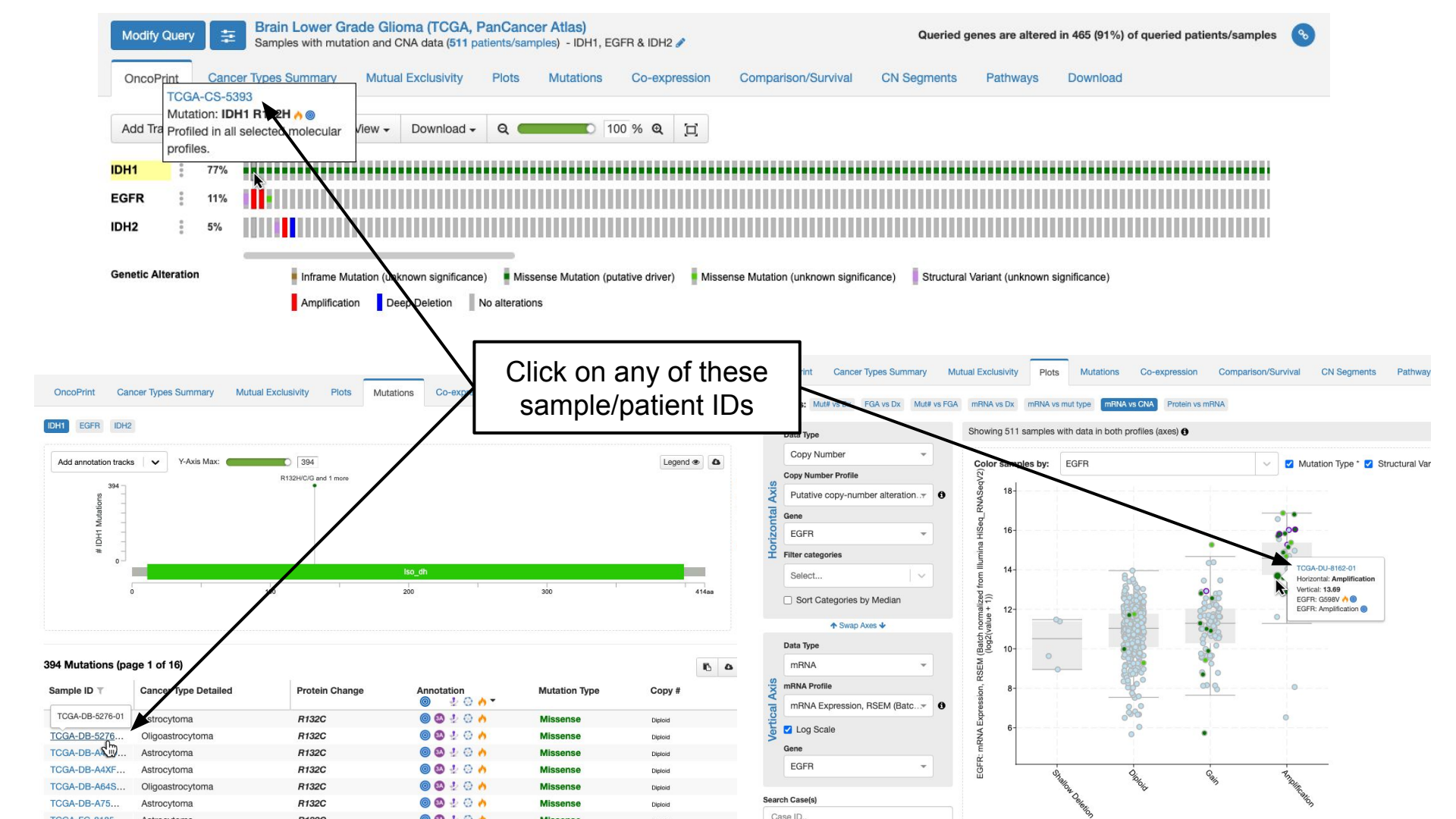
Tutorial Objectives

- Show different routes to get to patient view
- Walk through each of the possible tabs in patient view
  - Summary
  - Pathways
  - 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.

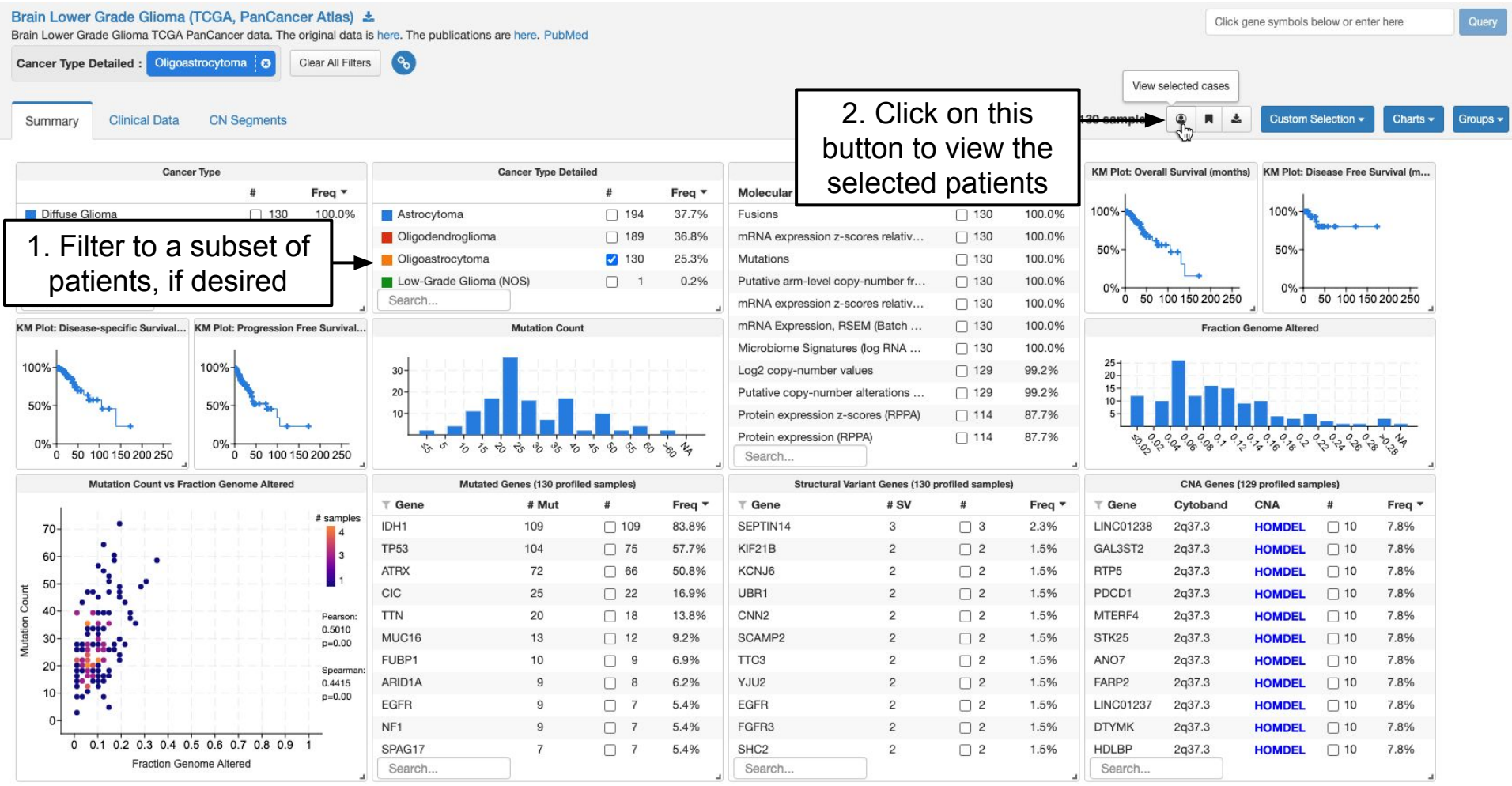
See next slide for examples.


Modify Query
Brain Lower Grade Glioma (TCGA, PanCancer Atlas)
Samples with mutation and CNA data (511 patients/samples) - IDH1, EGFR & IDH2
Queried genes are altered in 465 (91%) 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.
Download
100 %
IDH1 77%
EGFR 11%
IDH2 5%
Genetic Alteration
Inframe Mutation (unknown significance)
Missense Mutation (putative driver)
Missense Mutation (unknown significance)
Structural Variant (unknown significance)
Amplification
Deep Deletion
No alterations
Click on any of these sample/patient IDs
Add annotation tracks
Y-Axis Max:
394
Legend
IDH1 Mutations
R132H/C/G and 1 more
Iso_dh
414aa
394 Mutations (page 1 of 16)
Sample ID
Cancer Type Detailed
Protein Change
Annotation
Mutation Type
Copy #
TCGA-DB-5276-01
Astrocytoma R132C Missense Diploid
TCGA-DB-5276... Oligoastrocytoma R132C Missense Diploid
TCGA-DB-A4XF... Astrocytoma R132C Missense Diploid
TCGA-DB-A64S... Oligoastrocytoma R132C Missense Diploid
TCGA-DB-A75... Astrocytoma R132C Missense Diploid
Showing 511 samples with data in both profiles (axes)
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
EGFR
Search Case(s)
Case ID
Color samples by:
EGFR
Mutation Type
Structural Var
EGFR: mRNA Expression, RSEM (Batch normalized from Illumina HiSeq_RNASeqV2) (log2(value + 1))
Shallow Deletion
Diploid
Gain
Amplification
TCGA-DU-8162-01
Horizontal: Amplification
Vertical: 13.69
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.

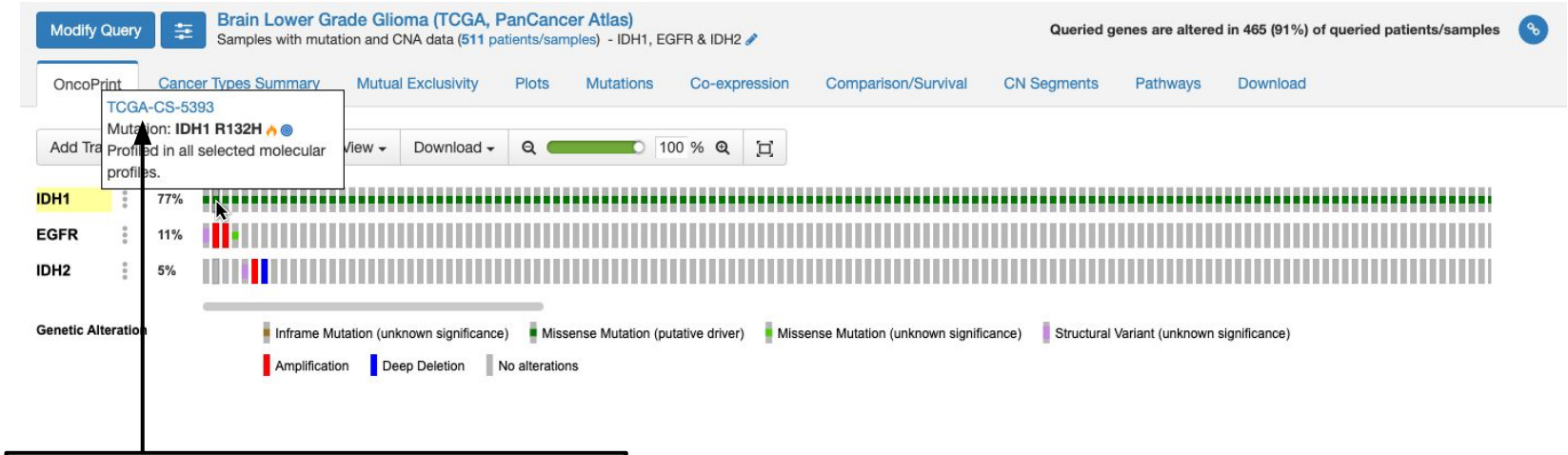

Brain Lower Grade Glioma (TCGA, PanCancer Atlas)
Brain Lower Grade Glioma TCGA PanCancer data. The original data is here. The publications are here. PubMed
Click gene symbols below or enter here
Query
Cancer Type Detailed :
Oligoastrocytoma
Clear All Filters
View selected cases
Summary
Clinical Data
CN Segments
130 samples
Custom Selection
Charts
Groups
2. Click on this button to view the selected patients
1. Filter to a subset of patients, if desired
Cancer Type
Diffuse Glioma
Cancer Type Detailed
Astrocytoma
Oligodendroglioma
Oligoastrocytoma
Low-Grade Glioma (NOS)
Molecular
Fusions
mRNA expression z-scores relativ…
Mutations
Putative arm-level copy-number fr…
mRNA expression z-scores relativ…
mRNA Expression, RSEM (Batch …
Microbiome Signatures (log RNA …
Log2 copy-number values
Putative copy-number alterations …
Protein expression z-scores (RPPA)
KM Plot: Overall Survival (months)
KM Plot: Disease Free Survival (m…
KM Plot: Disease-specific Survival…
KM Plot: Progression Free Survival…
Mutation Count
Fraction Genome Altered
Mutation Count vs Fraction Genome Altered
samples
Pearson:
0.5010
p=0.00
Spearman:
0.4415
p=0.00
Mutation Count
Fraction Genome Altered
Mutated Genes (130 profiled samples)
Gene
Mut
IDH1
TP53
ATRX
CIC
TTN
MUC16
FUBP1
ARID1A
EGFR
NF1
SPAG17
Structural Variant Genes (130 profiled samples)
SV
SEPTIN14
KIF21B
KCNJ6
UBR1
CNN2
SCAMP2
TTC3
YJU2
EGFR
FGFR3
SHC2
CNA Genes (129 profiled samples)
Cytoband
CNA
LINC01238
GAL3ST2
RTP5
PDCD1
MTERF4
STK25
ANO7
FARP2
LINC01237
DTYMK
HDLBP
HOMDEL
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

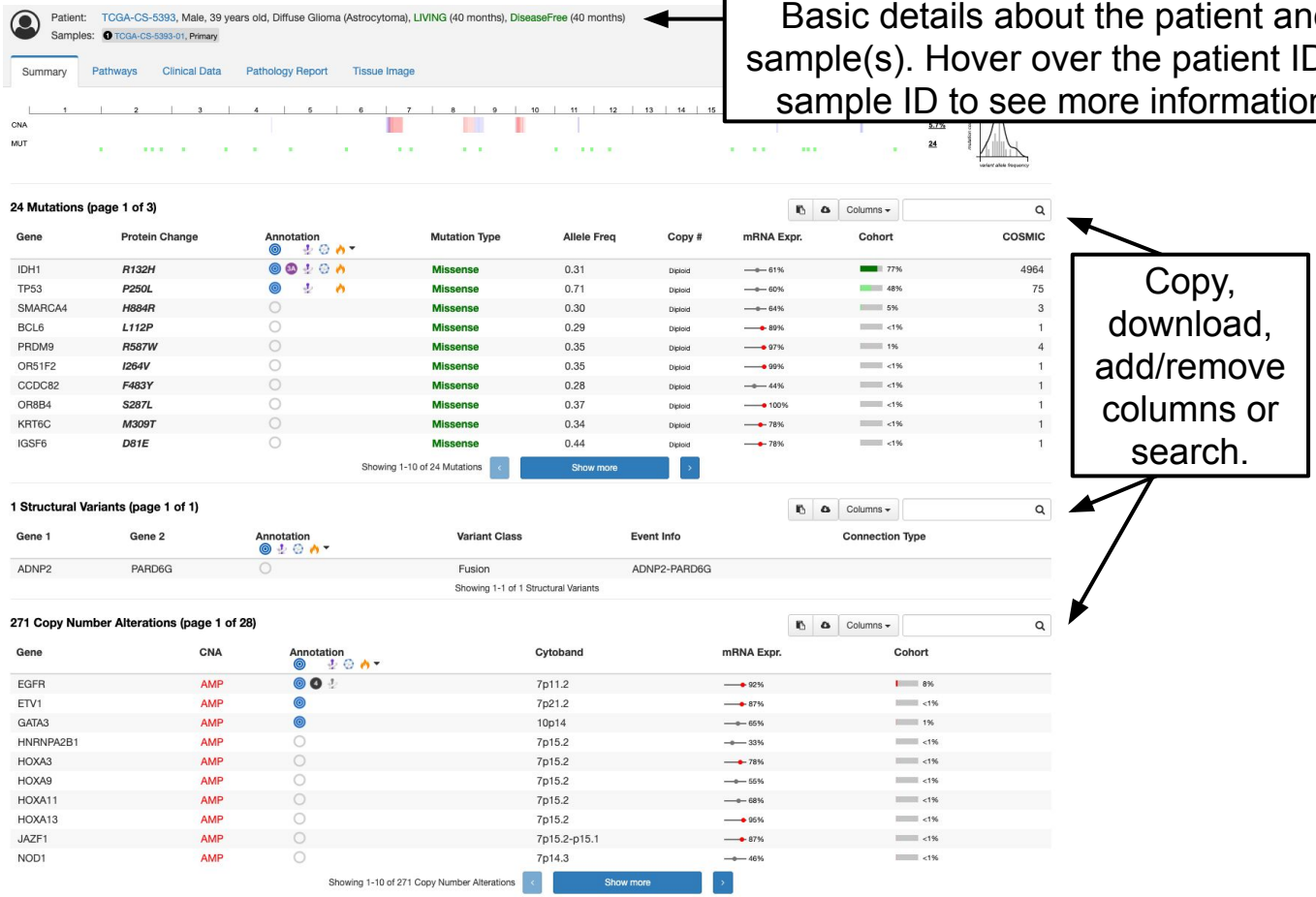
Patient View, Example 1: Summary

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

Lists of all called mutations, structural variants and CNAs (amplifications and deep deletions only).

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

Copy, download, add/remove columns or search.



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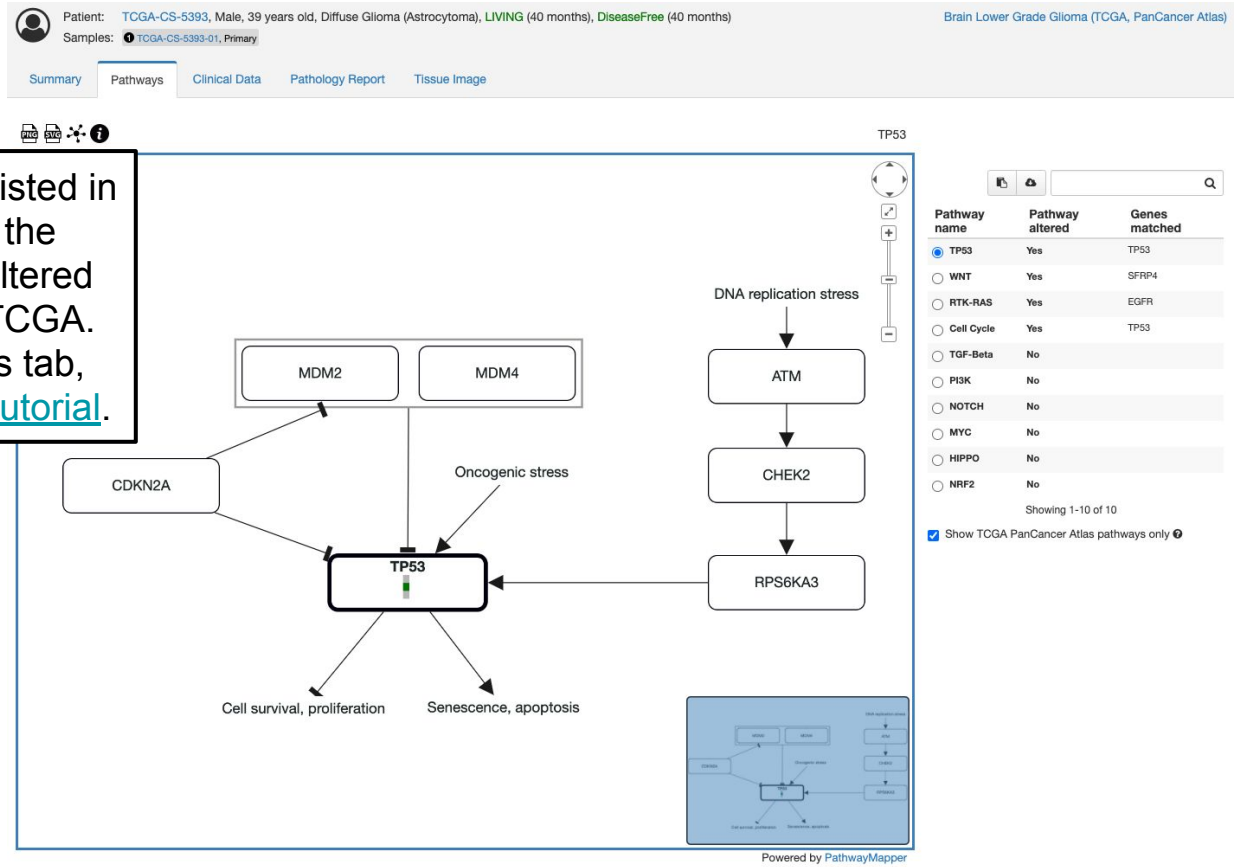
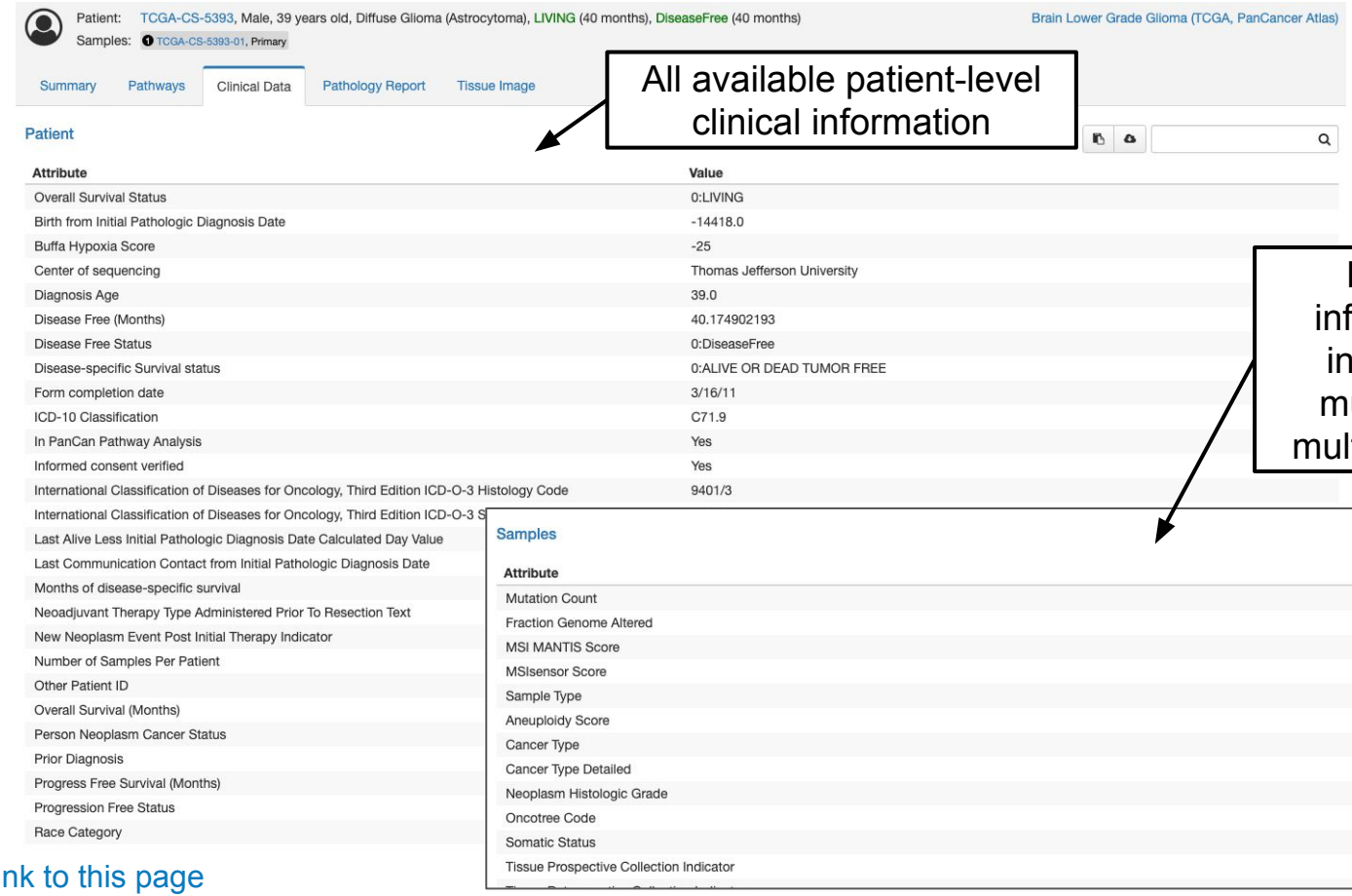
Patient View, Example 1: Pathways

Explore the alterations listed in the Summary tab in the context of frequently altered pathways defined by TCGA. For more detail on this tab, refer to the Pathways Tutorial.



Link to this page

Patient View, Example 1: Clinical Data



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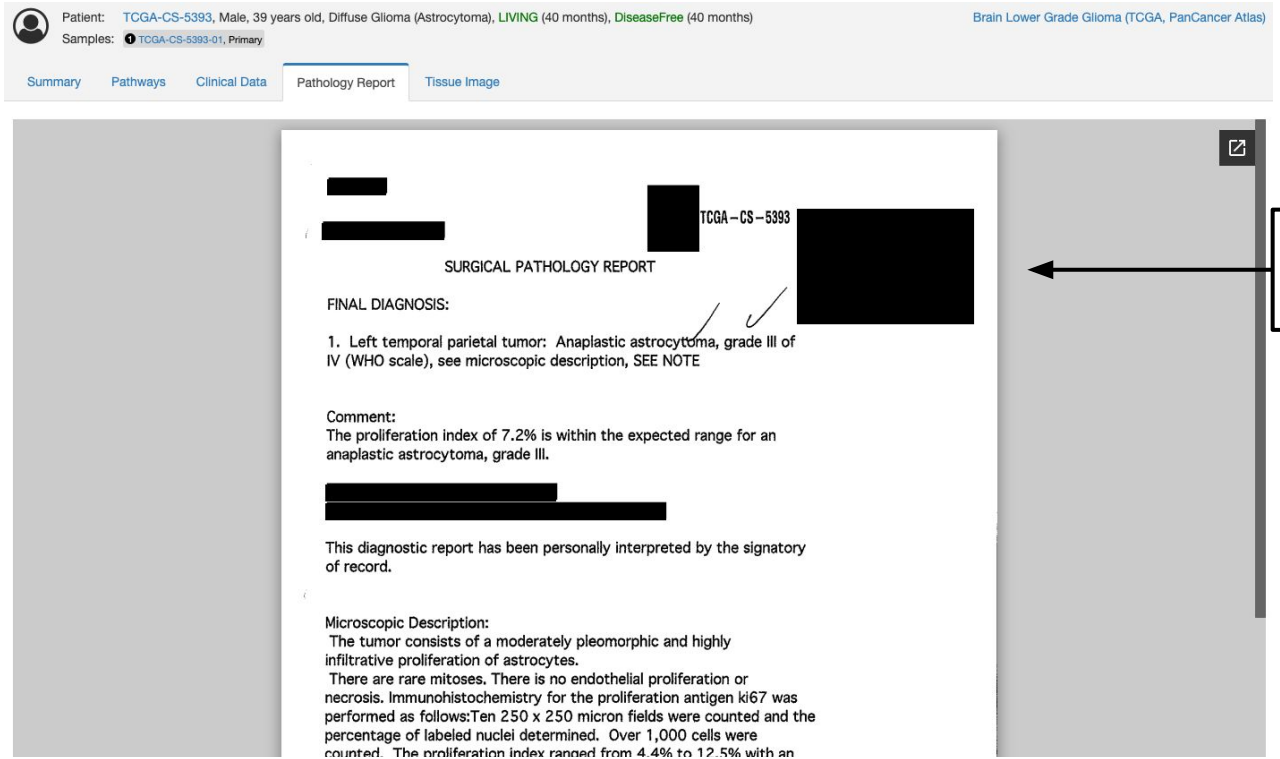
Patient View, Example 1: Pathology Report

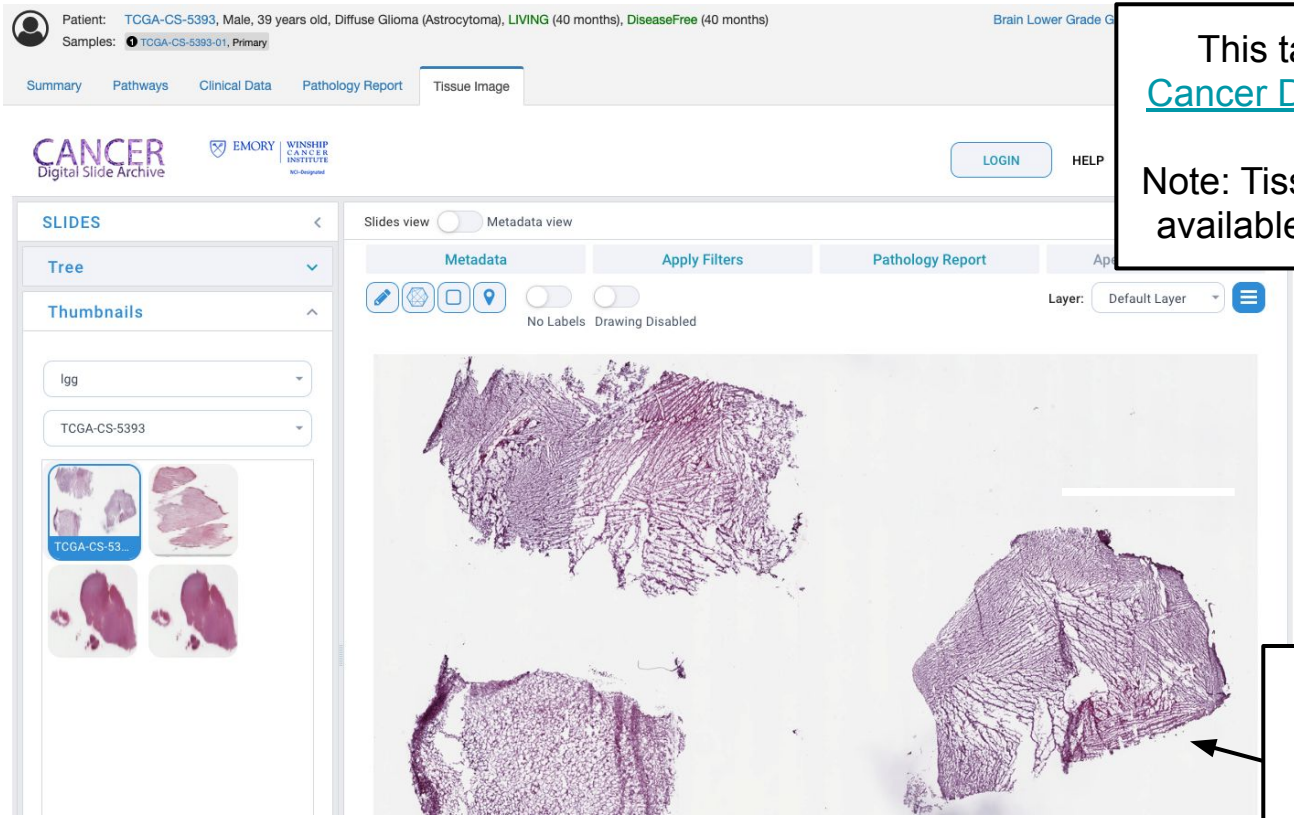


Note: Pathology Reports are only available for TCGA studies.

Original pathology report, de-identified.

Link to this page

Patient View, Example 1: Tissue Image



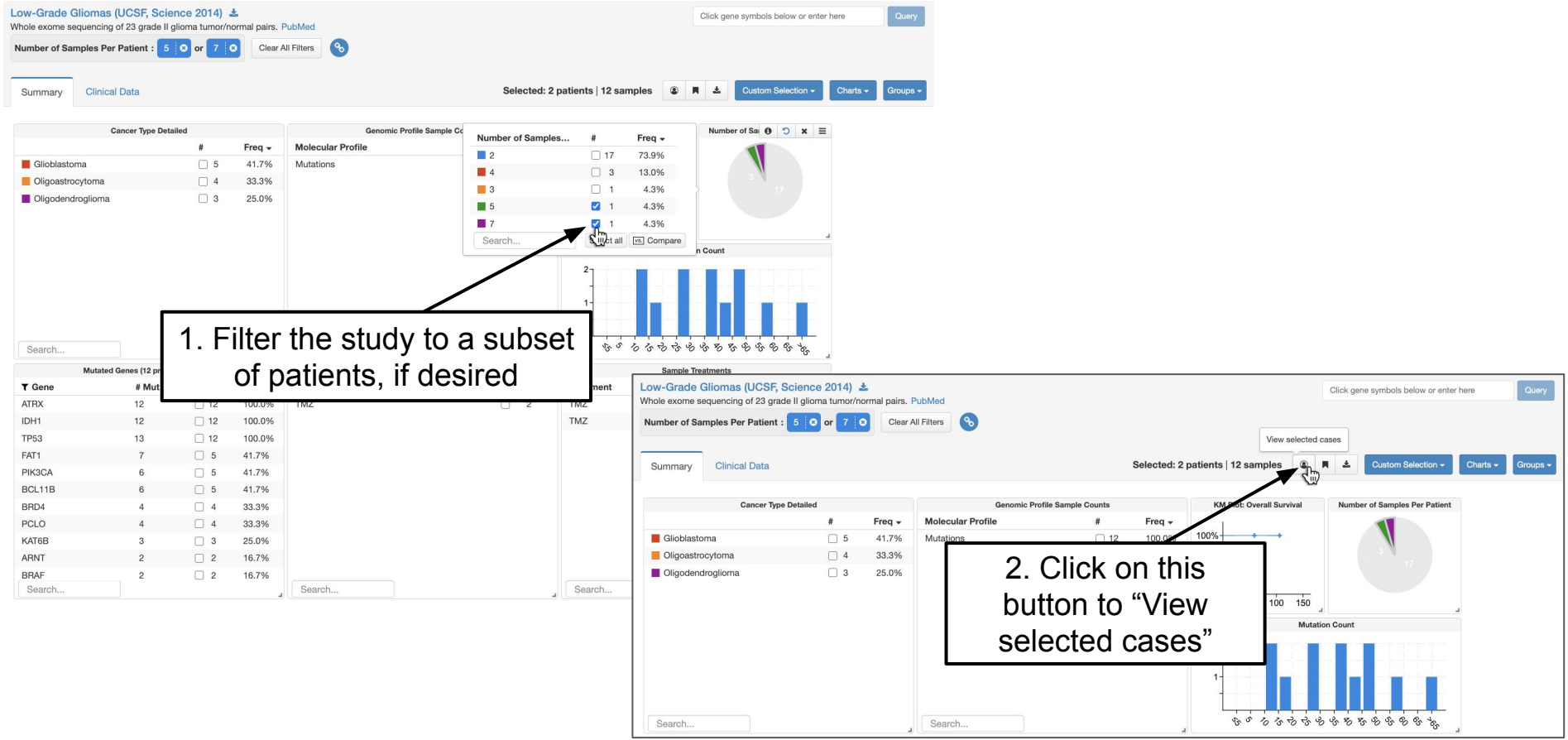
This tab integrates the Cancer Digital Slide Archive.

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.

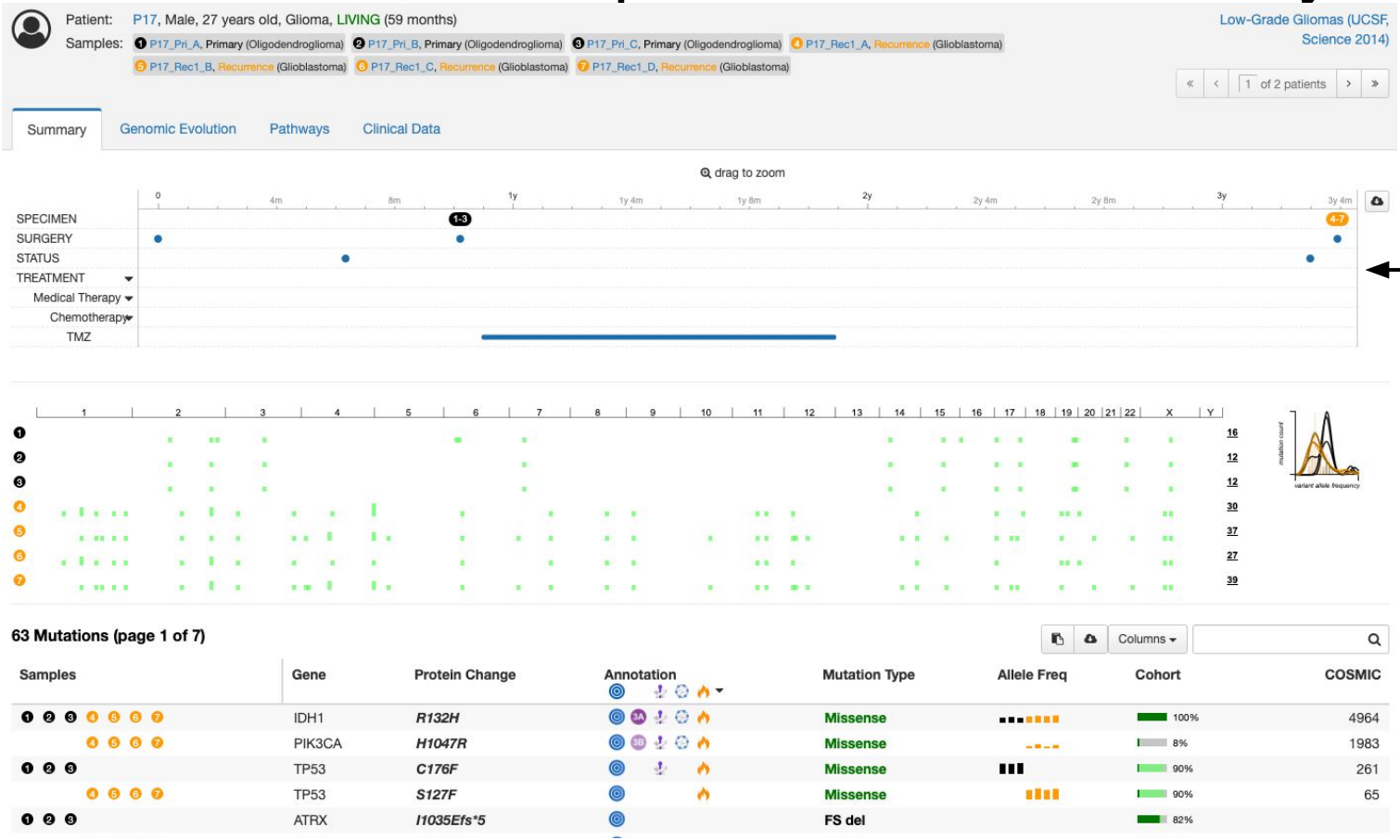
Link to this page

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



Link to this page

Patient View, Example 2: Patient Summary

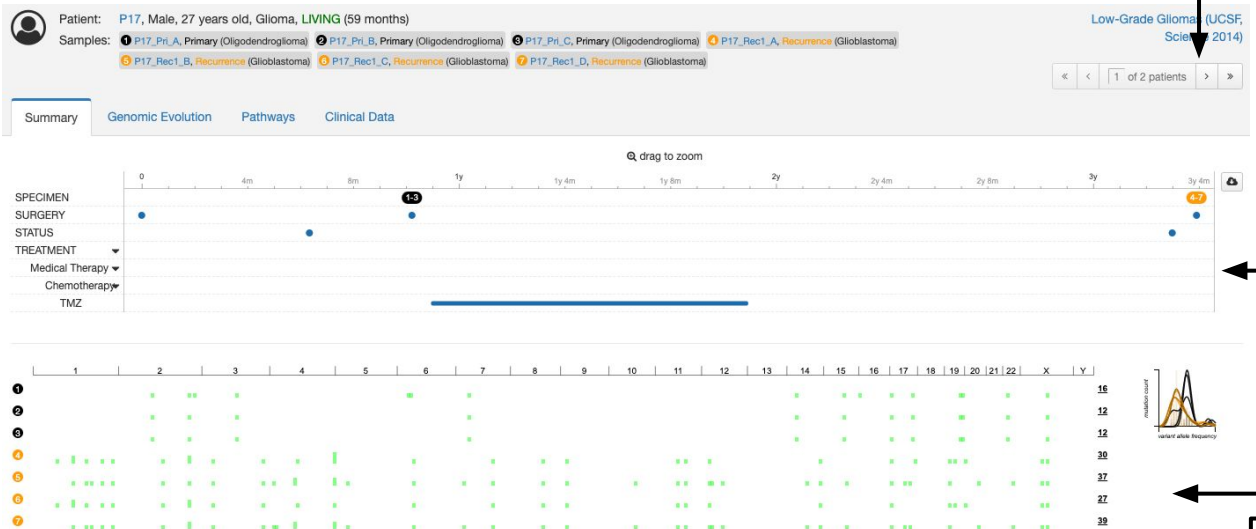


This study has multiple samples per patient and extensive clinical data to generate this enhanced patient timeline.

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.

63 Mutations (page 1 of 7)

Columns

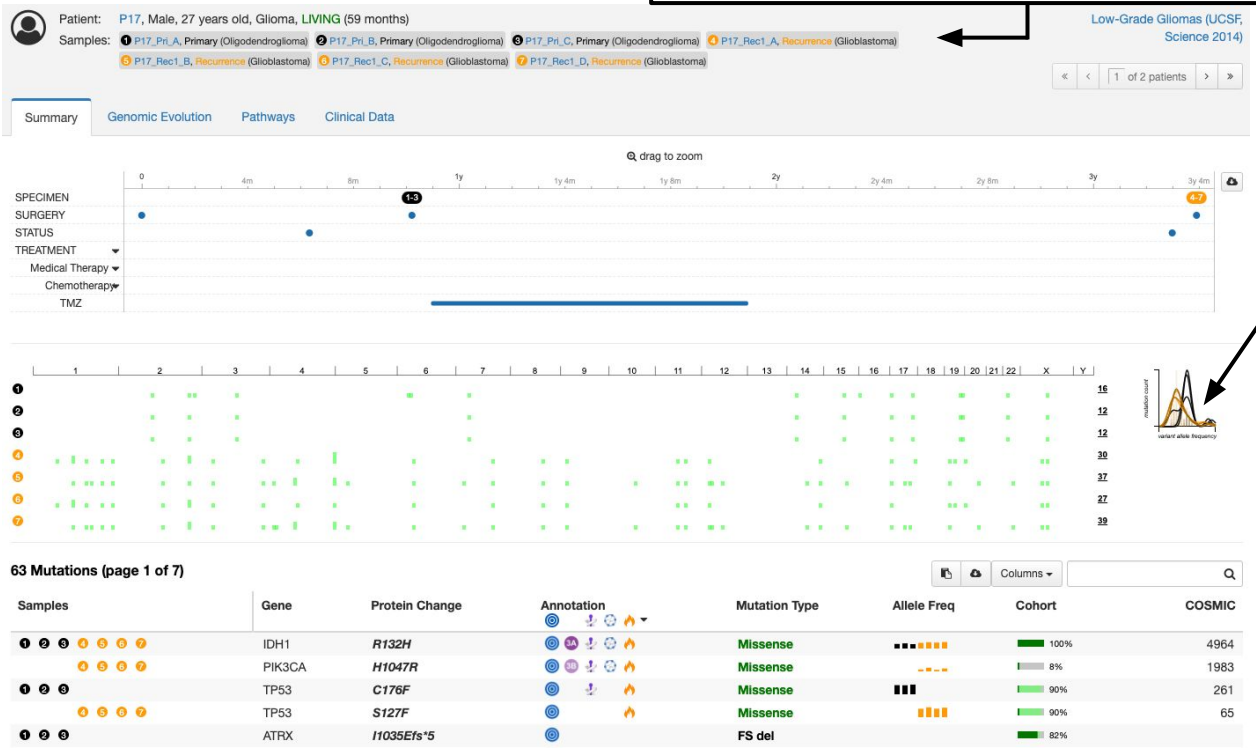
Samples	Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Cohort	COSMIC
1 2 3 4 5 6 7	IDH1	*R132H*		Missense		100%	4964
4 5 6 7	PIK3CA	*H1047R*		Missense		8%	1013
1 2 3	TP53	*C176F*		Missense		90%	261
4 5 6 7	TP53	*S127F*		Missense		90%	65
1 2 3	ATRX	*I1035Efs*5*		FS del		82%	

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.

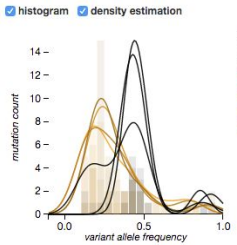
Link to this page

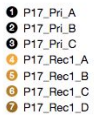
Patient View, Example 2: Patient Summary

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.

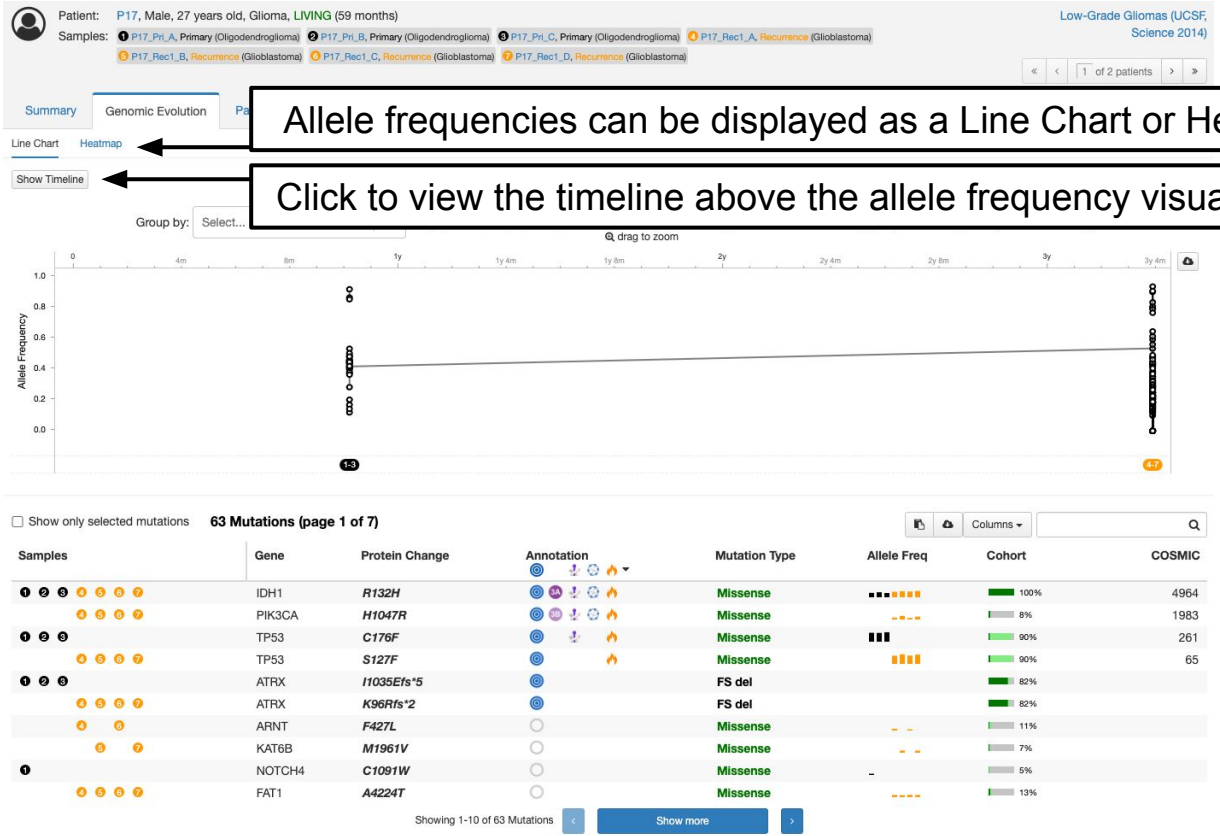




Hover over a sample ID to see the plot for just that sample

Link to this page

Patient View, Example 2: Genomic Evolution



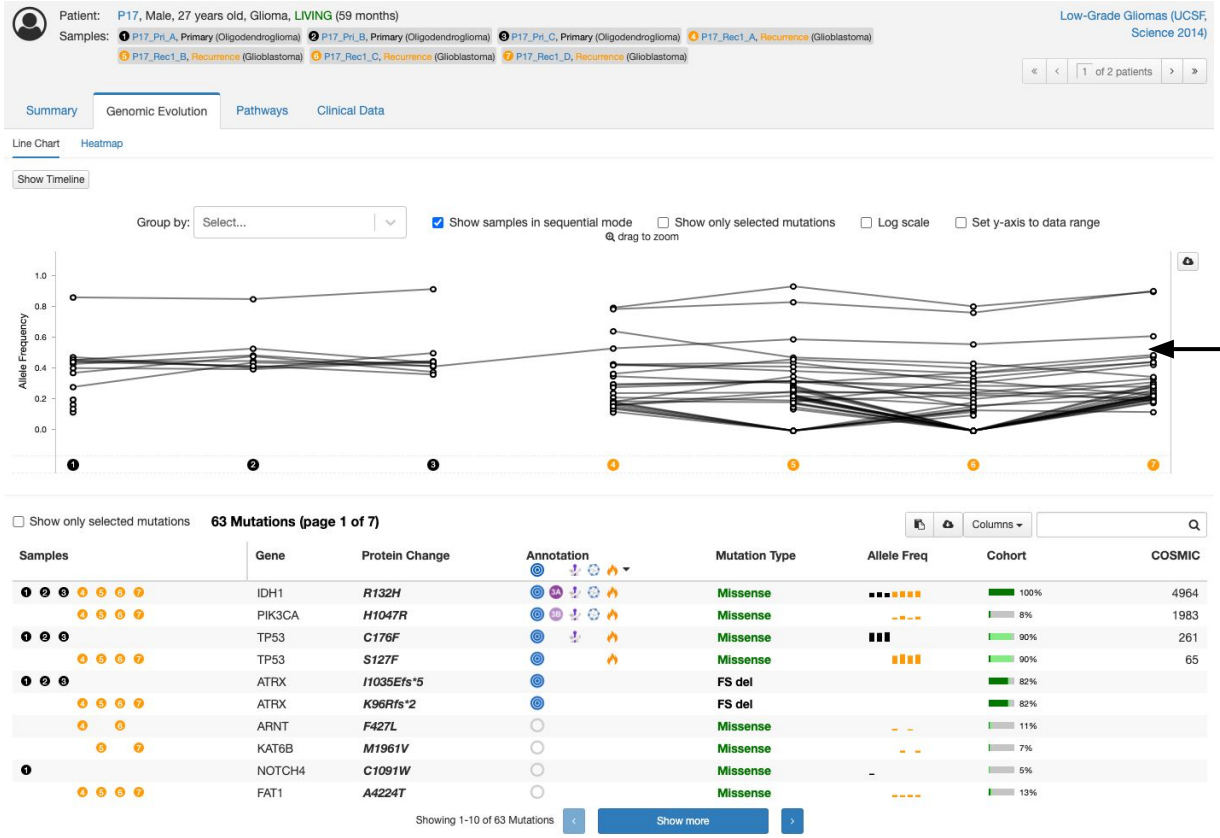
Allele frequencies can be displayed as a Line Chart or Heatmap

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



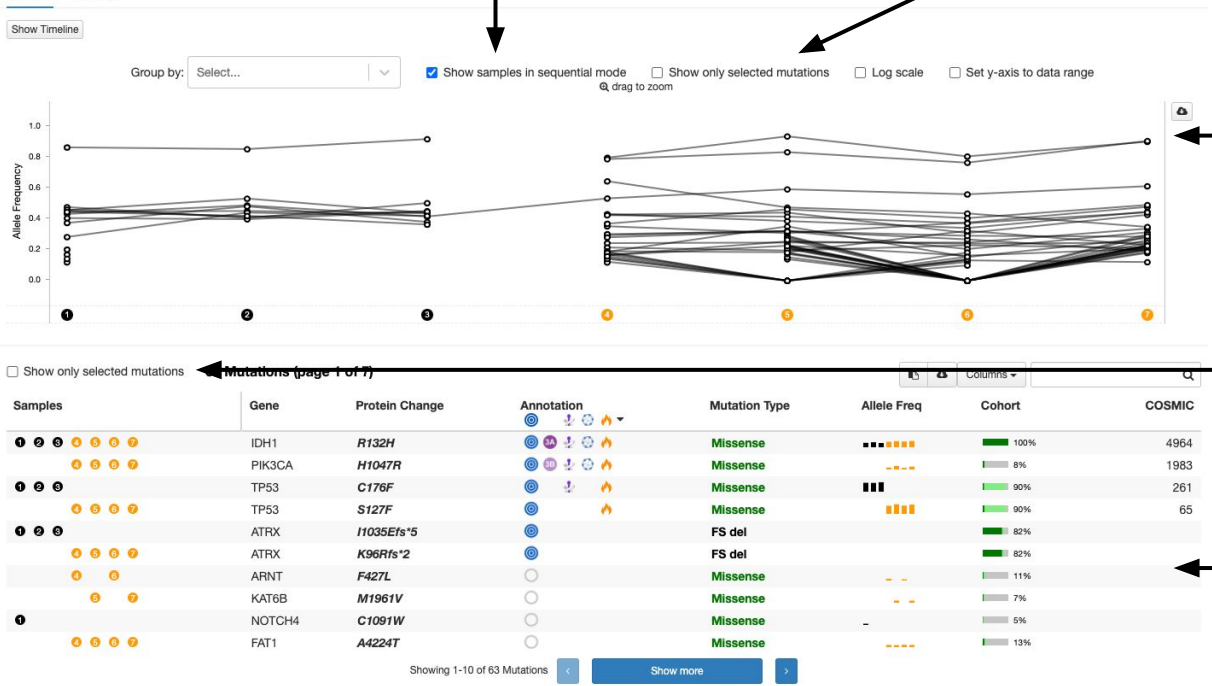
Each dot represents the allele frequency of a mutation in a sample. Lines connect mutations that are detected in multiple samples. Options above the chart enable customization.

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.



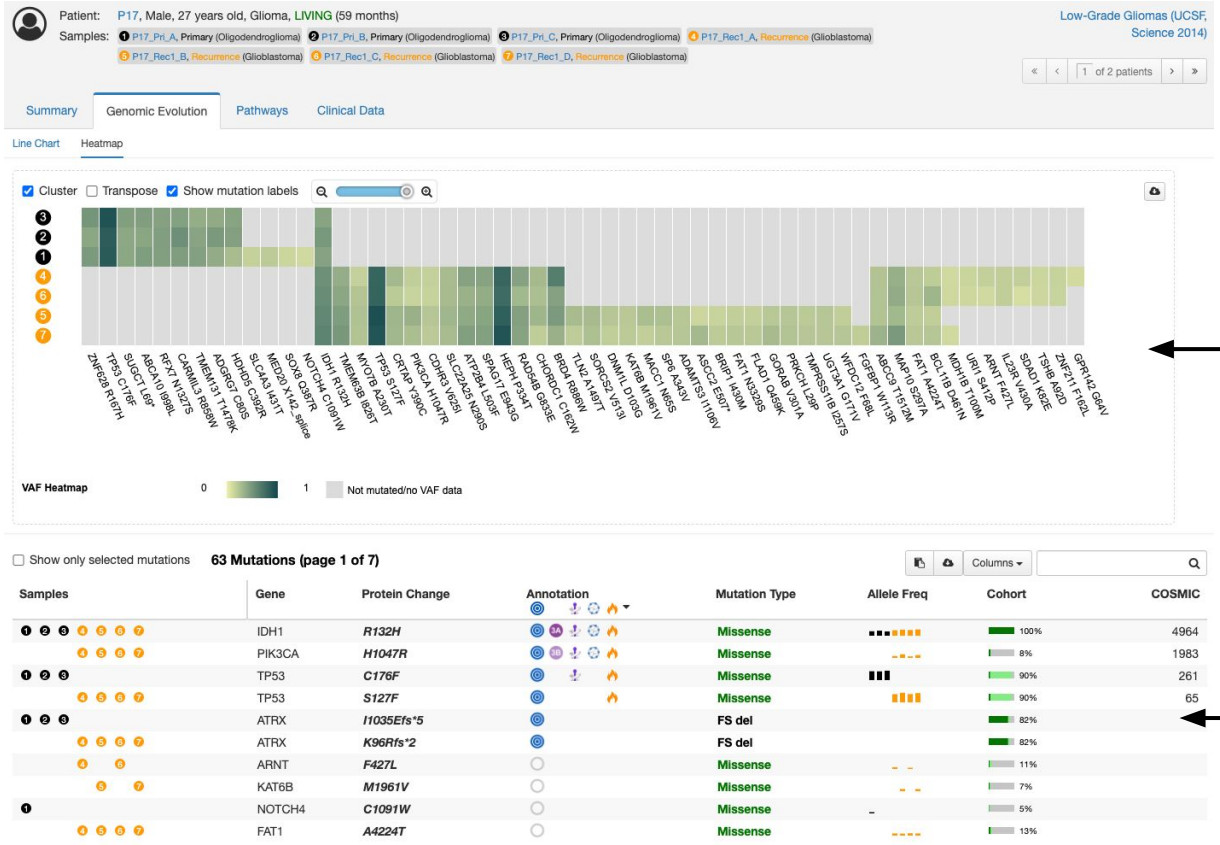
This chart and the mutation table are linked - hover or click on a mutation in the chart to see it highlighted in the table below.

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.

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

All available patient-level clinical information

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 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	30	37	27	39
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
Somatic Status	Matched	Matched	Matched	Matched			
TMB (nonsynonymous)	0.533333333333	0.4	0.4	0.9666666666			

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

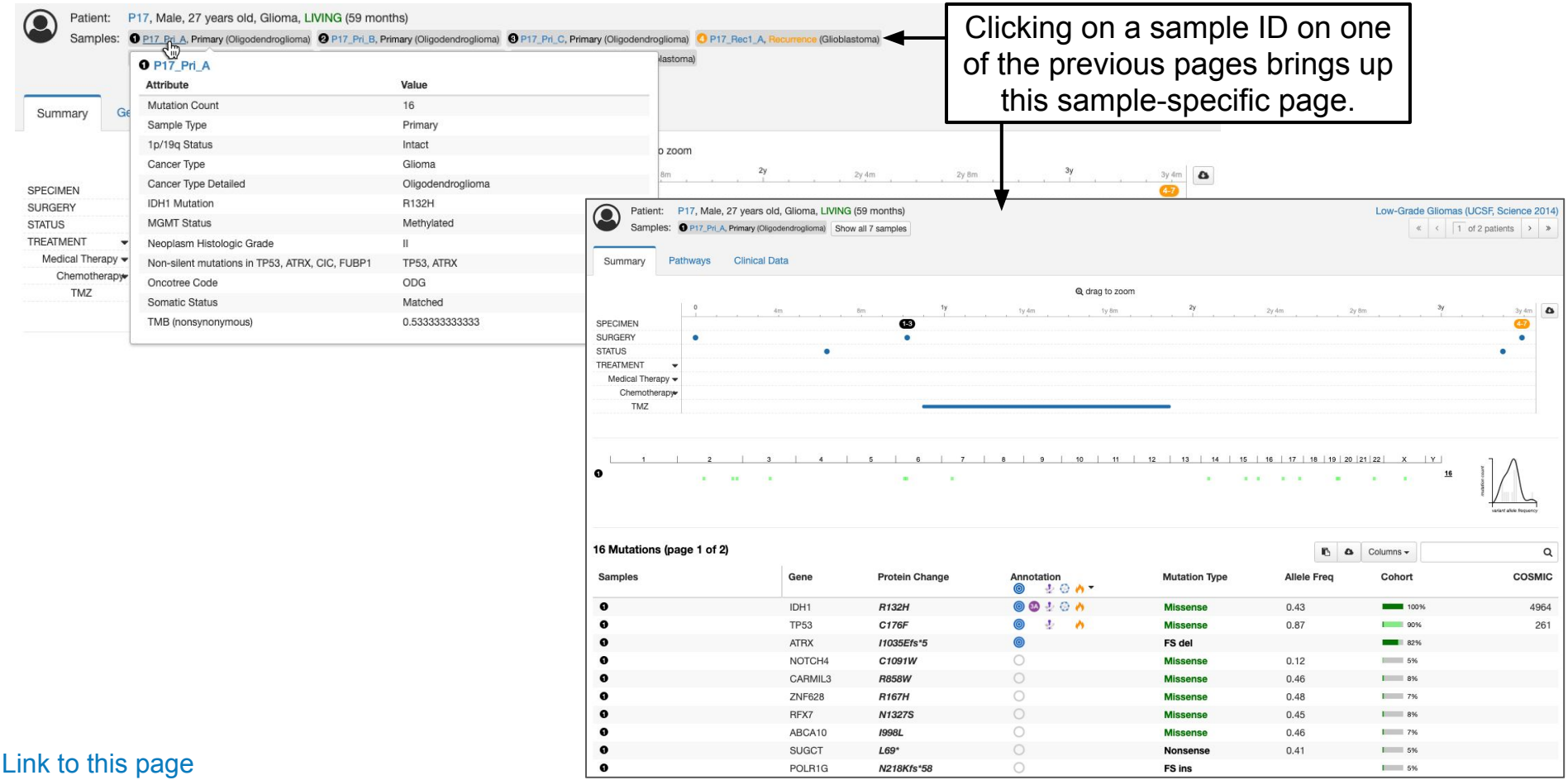
Timeline Data

Surgery

START_DATE	STOP_DATE	EVENT_TYPE	EVENT_TYPE_DETAILED
0		Surgery	OA II biopsy
311		Surgery	Oligo II Initial
1214		Surgery	GBM Recurrence1

Link to this page

Patient View, Example 2: Sample Summary

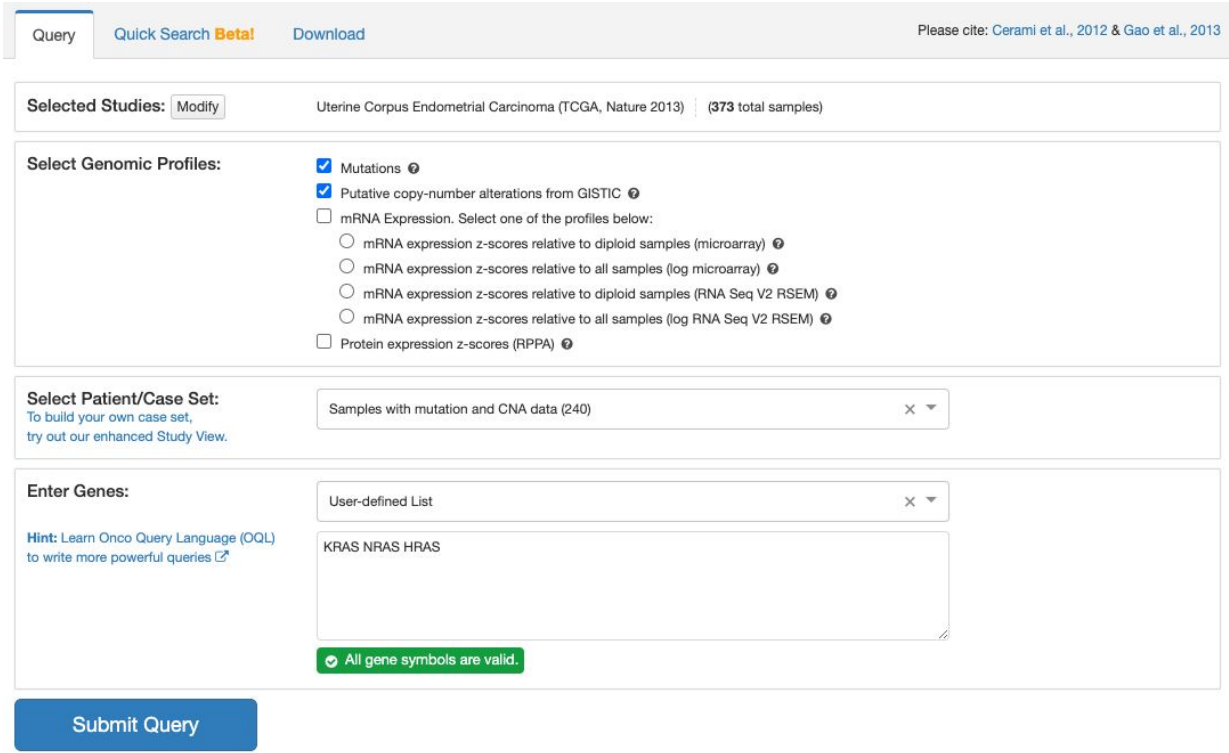


Link to this page

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

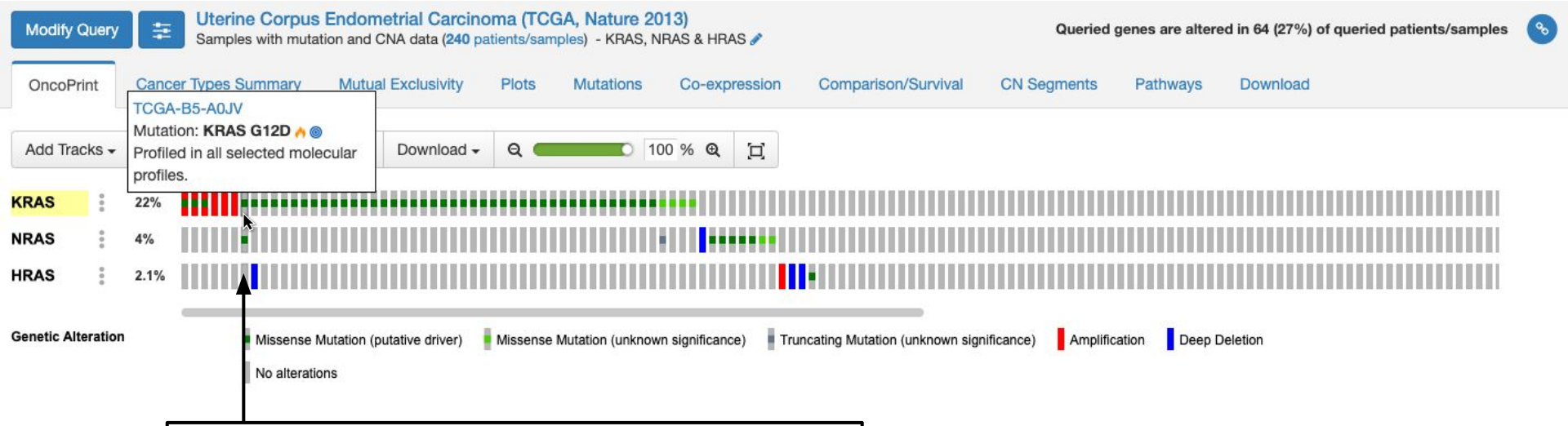
Let's look at RAS mutations in Uterine Corpus Endometrial Carcinoma (TCGA, Nature 2013).

Example 3: Run the query



Link to this page

Example 3: OncoPrint

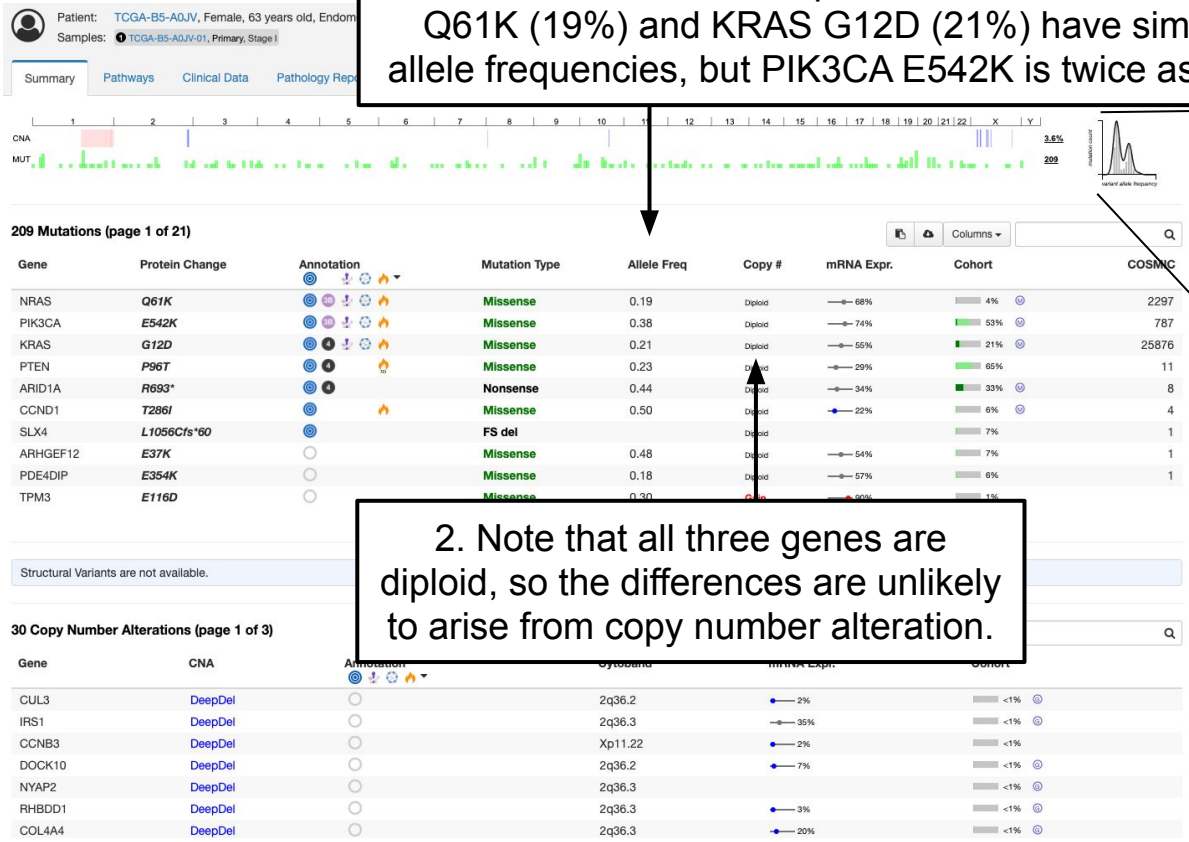


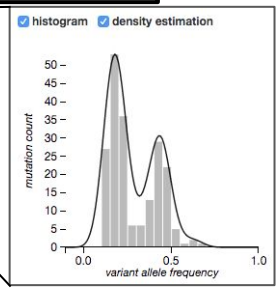
In general, mutations in these genes are mutually exclusive. However, there's one case with driver mutations in both KRAS and NRAS. Let's look at that patient in greater detail by clicking on the patient ID ("TCGA-B5-A0JV").

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%).





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