

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
 - 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

Add clinical

Mutation: IDH1 R132H
 Profiled in all selected molecular profiles.
 TCGA-DH-5142

Diagnosis Age

IDH1

77%

EGFR

9%

IDH2

5%

Genetic Alteration

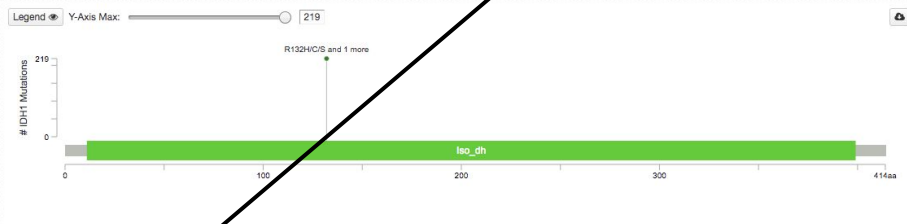
Missense Mutation (putative driver) Missense Mutation (unknown significance) Amplification Deep Deletion No alterations

Diagnosis Age

14 75

Click on any of these sample/patient IDs

IDH1 EGFR IDH2



219 Mutations (page 1 of 9)

Sample ID	Cancer Type	Protein Change	Annotation	Mutation Type	Copy #
TCGA-CS-6666-01	Anaplastic Astrocytoma	R132C	Missense	Missense	Diploid
TCGA-HT-7479-01	Anaplastic Astrocytoma	R132C	Missense	Missense	Diploid
TCGA-FG-8185-01	Anaplastic Astrocytoma	R132C	Missense	Missense	Diploid
TCGA-HT-7693-01	Oligodendroglioma	R132C	Missense	Missense	ShallowDel
TCGA-HT-7855-01	Anaplastic Astrocytoma	R132C	Missense	Missense	Diploid
TCGA-DB-A4XD-01	Anaplastic Astrocytoma	R132C	Missense	Missense	Diploid
TCGA-DB-A4XF-01	Anaplastic Astrocytoma	R132C	Missense	Missense	Diploid
TCGA-P5-A5EZ-01	Anaplastic Astrocytoma	R132C	Missense	Missense	Diploid

Horizontal Axis

Data Type

Copy Number

Copy Number Profile

Putative copy-number alteration

Gene

EGFR

Swap Axes

Vertical Axis

Data Type

mRNA

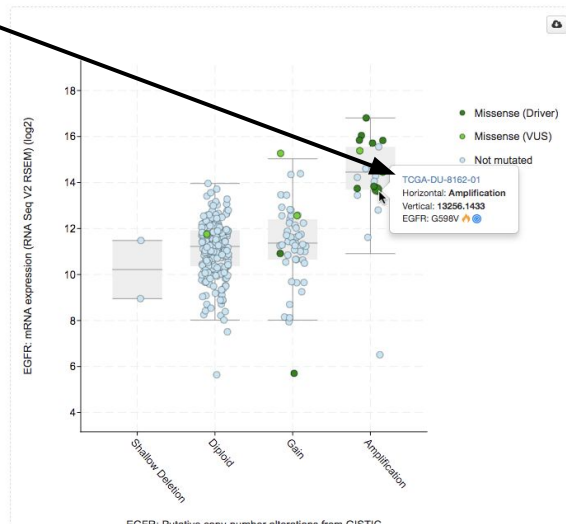
mRNA Profile

mRNA expression (RNA Seq V2)

Apply Log Scale

Gene

Same gene (EGFR)



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.

Number of Samples Per Patient : **2** [Clear All Filters](#)[View selected cases](#)[Summary](#)[Clinical Data](#)[Heatmaps](#)[CN Segments](#)Quick Filters: ☐ 9 sam

28 sampl

[Custom Selection](#)[+ Add Chart](#)[Groups](#)

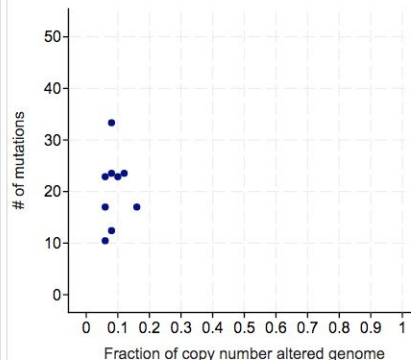
2. Click on this button to view the selected patients

1. Filter to a subset of patients, if desired

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%

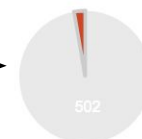
Mutation Count vs Fraction



Mutated Genes (9 profiled samples)

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

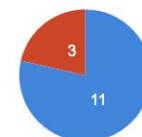
Number of Samples Per Patient



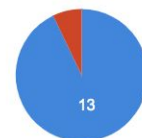
Sex



Ethnicity Category



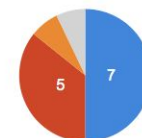
Race Category



Sample Type



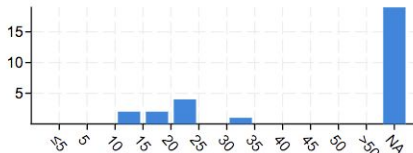
Tumor Site



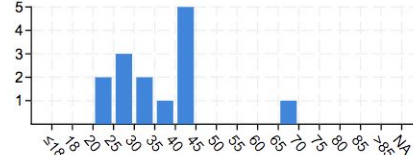
CNA Genes (14 profiled samples)

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%

Mutation Count



Diagnosis Age



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.

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.



26 Mutations (page 1 of 3)

Gene	Protein Change	Annotation	Mutation Type	Allele Freq	mRNA Expr.	Cohort	COSMIC
IDH1	R132H		Missense	0.36			4964
TP53	H193R		Missense	0.85			196
ATRX	K1603R		Missense	0.75			1
AMY2B	R107H		Missense	0.15			3
SMCO4	A39V		Missense	0.10			
CACNA1B	R370C		Missense	0.27			2
CCNG2	K132*		Nonsense	0.37			1
DNAH11	A1613S		Missense	0.30			
MYH8	A770P		Missense	0.38			1
OAF	P156A		Missense	0.41			1

Showing 1-10 of 26 Mutations

Show more

1138 Copy Number Alterations (page 1 of 114)

Gene	CNA	Annotation	Cytoband	mRNA Expr.	Cohort
EGFR	AMP		7p11.2		
CCNE1	AMP		19q12		
BRD4	AMP		19p13.12		
PIK3R2	AMP		19p13.11		
MEF2B	AMP		19p13.11		
AR	AMP		Xq12		
PRKACA	AMP		19p13.12		
TTF1	AMP		9q34.13		
DNAJB1	AMP		19p13.12		
ABL1	AMP		9q34.12		

Showing 1-10 of 1138 Copy Number Alterations

Show more

Copy, download, add/remove columns or search.

Patient View, Example 1: Clinical Data

 Patient: **TCGA-DH-5142**, Male, 29 years old, Glioma (Anaplastic Astrocytoma), **LIVING** (63 months), **Recurred/Progressed** (53 months) Brain Lower Grade Glioma (TCGA, Provisional)
Samples: **1** **TCGA-DH-5142-01**, Primary

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

All available patient-level
clinical information

Patient

Attribute	Value
Animal Insect Allergy History	NO
Asthma History	YES
Birth from Initial Pathologic Diagnosis Date	-10678
Days to initial score performance status scale	10
Days to Last Followup	922
Diagnosis Age	29
Disease Free (Months)	53.45
Disease Free Status	Recurred/Progressed
Eczema History	NO
Ethnicity Category	NOT HISPANIC OR LATINO
Fever History	NO
First symptom longest duration	0 - 30 Days
Food Allergy History	NO
Form completion date	12/8/10
Headache History	
History ionizing rt to head	
ICD-10 Classification	
IDH1 Mutation Test Indicator	
Informed consent verified	
International Classification of Diseases for Oncology, Third Edition ICD-O-3 Histology Code	
International Classification of Diseases for Oncology, Third Edition ICD-O-3 Site Code	
Karnofsky Performance Score	
Last Alive Less Initial Pathologic Diagnosis Date Calculated Day Value	
Mold or Dust Allergy History	
Neoadjuvant Therapy Type Administered Prior To Resection Text	
Neoadjuvant Therapy Type Administered Prior To Resection Text	
Neoplasm Histologic Grade	
Neoplasm Histologic Type Name	

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

Samples

Attribute	TCGA-DH-5142-01
Mutation Count	26
Fraction Genome Altered	0.083396181
Cancer Type	Glioma
Cancer Type Detailed	Anaplastic Astrocytoma
Is FFPE	NO
Longest Dimension	0.4
Oncotree Code	AASTR
Other Sample ID	9dbfe00b-fae6-4765-bf20-4f3cc45880e9
Pathology Report File Name	TCGA-DH-5142.d6bad55b-3114-416a-8775-e3439c0f6918.pdf
Pathology report uuid	d6bad55b-3114-416a-8775-e3439c0f6918
Sample Type	Primary
Sample type id	1
Shortest Dimension	0.3
Specimen Second Longest Dimension	0.4
Vial number	A

Patient View, Example 1: Pathology Report

Patient: TCGA-DH-5142, Male, 29 years old, Glioma (Anaplastic Astrocytoma), LIVING (63 months), Recurred/Progressed (53 months)
Samples: TCGA-DH-5142-01, Primary

Brain Lower Grade Glioma (TCGA, Provisional)

Summary Clinical Data Pathology Report Tissue Image

Note: Pathology Reports are only available for TCGA studies.

Original pathology report, de-identified.

SPECIMEN(S) SUBMITTED/ PROCEDURES ORDERED:

CLINICAL HISTORY: [REDACTED] male, here for left side craniotomy for tumor.

GROSS DESCRIPTION: Received the following specimens in the Department of Pathology, labeled with the patient's name and [REDACTED]

- A. Brain tumor
- B. Brain tumor

A. The specimen is received fresh and consists of a dome-shaped portion of edematous beige to gray-tan homogenous tissue that is 3.2 x 2.5 x 2.0cm. A representative portion is submitted for frozen section. Frozen section diagnosis is "Infiltrating glioma, most likely astrocytic, with features suspicious for anaplasia," by Dr. [REDACTED]. The frozen tissue is submitted in cassette FSA1, and representative tissue is submitted in cassettes A2-A4. A representative portion is submitted for the [REDACTED]

B. The specimen is received fresh and consists of a 3.6 x 3.3 x 0.9 cm portion of homogenous beige to gray-tan soft tissue with focal areas of yellow to white softening. A representative portion is submitted in cassettes B1-B4, and a portion is submitted to [REDACTED]

DIAGNOSIS:



- A. "Brain tumor":

Anaplastic astrocytoma (WHO Grade III) (see comment)

Page 1 / 2



Patient View, Example 1: Tissue Images



Note: Tissue images are only available for TCGA studies.

 Patient: TCGA-DH-5142, Male, 29 years old, Glioma (Anaplastic Astrocytoma), **LIVING** (63 months), **Recurred/Progressed** (53 months)
Samples:  TCGA-DH-5142-01, Primary

Summary Clinical Data Pathology Report **Tissue Image**

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TCGA-DH-5142

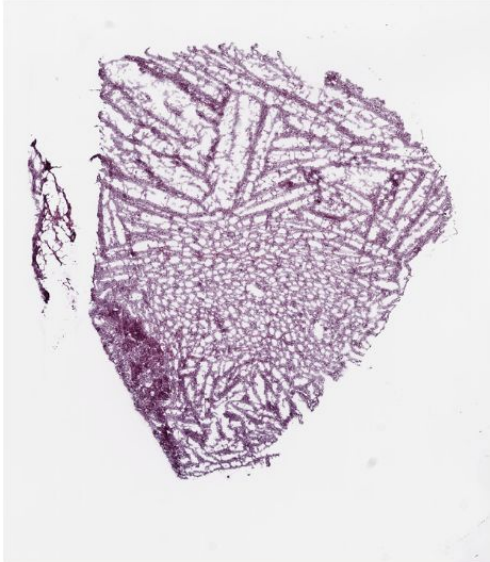
< 1/3 >
(5 slides)

TCGA-DH-5142-01A-01-BS1.4

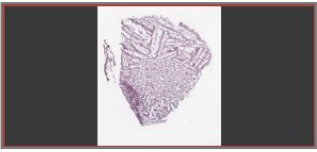
TCGA-DH-5142-01A-01-TS1.4

Pathology Report

Drawing Disabled No Labels Layer Default Layer

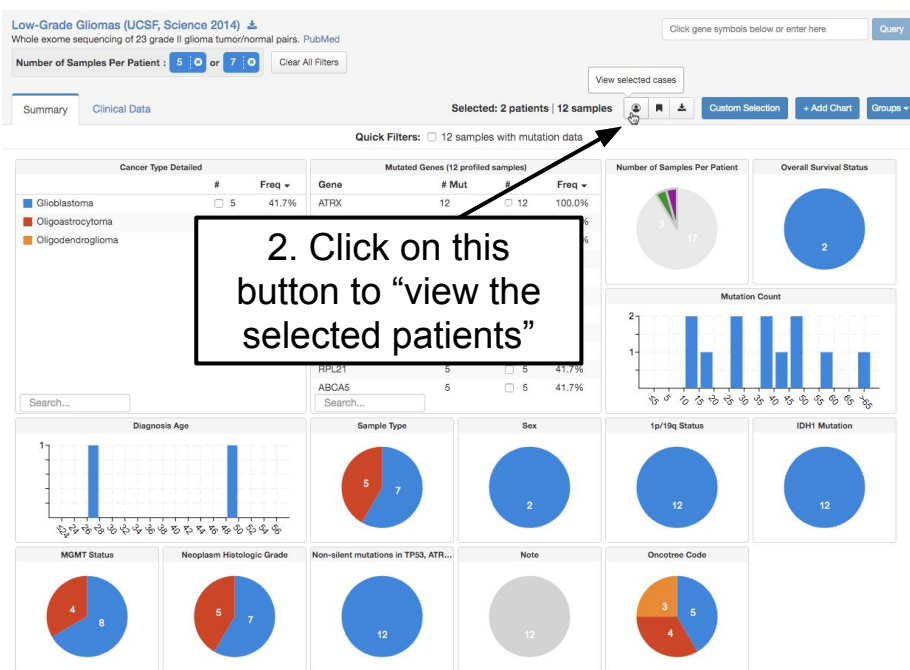
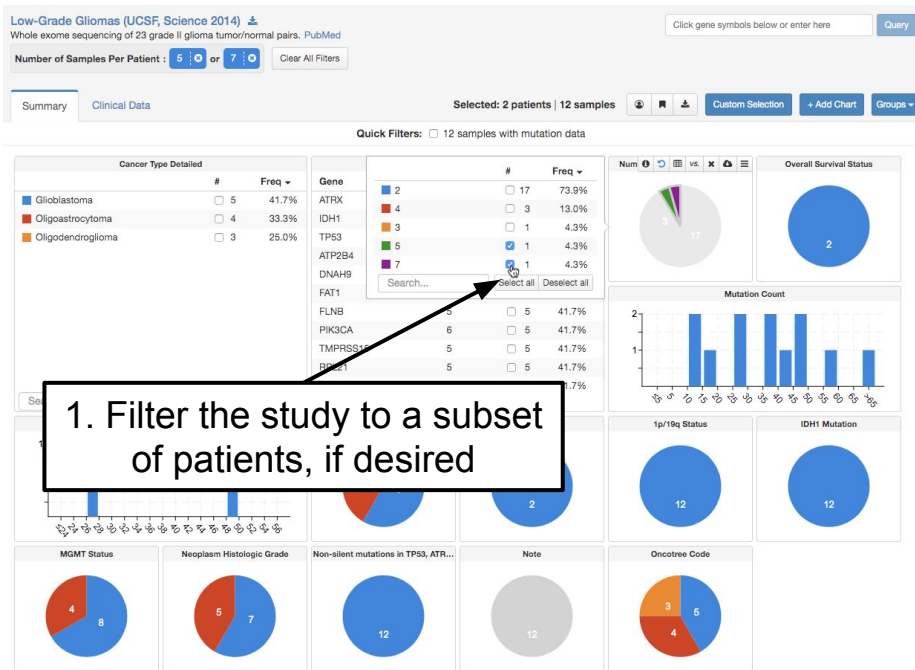


TCGA-DH-5142-01A-01-BS1.c4ef4682-abd8-4206-ac76-9fbd2b8fe861.svs



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

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



Patient View, Example 2: Patient Summary

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 >



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

62 Mutations (page 1 of 7)

Tumors	Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Cohort	COSMIC
P17 P17 P17 P17 P17	IDH1	R132H		Missense	■■■■■	100.0%	4964
P17 P17 P17 P17 P17	PIK3CA	H1047R		Missense	■■■■■	8.2%	1983
P17 P17 P17 P17 P17	TP53	C176F		Missense	■■■■■	90.2%	261
P17 P17 P17 P17 P17	TP53	S127F		Missense	■■■■■	90.2%	65
P17 P17 P17 P17 P17	ATRX	I103SEfs*5		FS del	■■■■■	82.0%	
P17 P17 P17 P17 P17	ATRX	K96Rfs*2		FS del	■■■■■	82.0%	
P17 P17 P17 P17 P17	FAT1	A4224T		Missense	■■■■■	13.1%	
P17 P17 P17 P17 P17	NOTCH4	C1091W		Missense	■■■■■	4.9%	
P17 P17 P17 P17 P17	BCL11B	D461N		Missense	■■■■■	9.8%	
P17 P17 P17 P17 P17	BRD4	R886W		Missense	■■■■■	6.6%	

Showing 1-10 of 62 Mutations < Show more >

Copy Number Alterations are not available.

Patient View, Example 2: Patient Summary

Click to view
the next patient


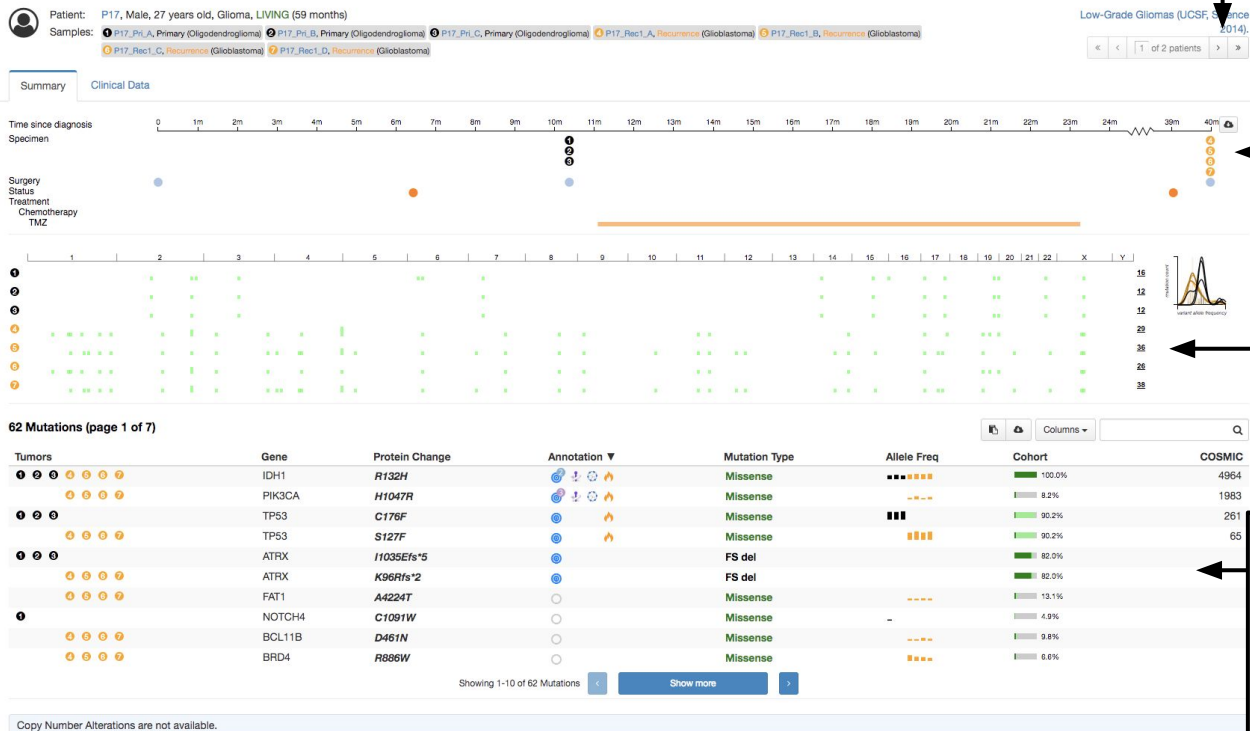
Patient timeline showing surgeries, radiographic progression and treatments. Hover over any feature for additional information. Click the  to expand the timeline.

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.



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_Pt_A, Primary (Oligodendroglioma) P17_Pt_B, Primary (Oligodendroglioma) P17_Pt_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

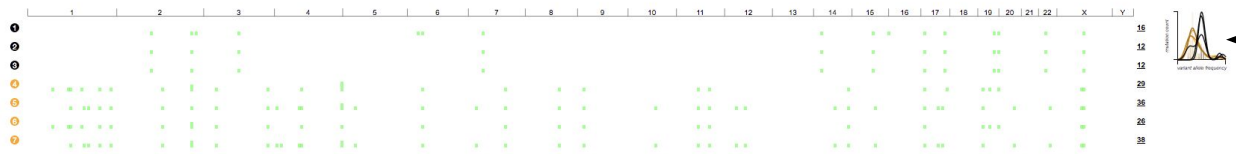
2014).

< < 1 of 2 patients > >

Summary Clinical Data

Time since diagnosis Specimen 0 1m 2m 3m 4m 5m 6m 7m 8m 9m 10m 11m 12m 13m 14m 15m 16m 17m 18m 19m 20m 21m 22m 23m 24m 30m 40m

Surgery
Status
Treatment
Chemotherapy
TMZ



62 Mutations (page 1 of 7)

Tumors	Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Cohort	COSMIC
P17_Pt_A	IDH1	R132H	Missense	Missense	100.0%	4964	
P17_Pt_A	PIK3CA	H1047R	Missense	Missense	8.2%	1983	
P17_Pt_A	TP53	C178F	Missense	Missense	90.2%	261	
P17_Pt_A	TP53	S127F	Missense	Missense	90.2%	65	
P17_Pt_A	ATRX	I1035Efs*5	FS del	FS del	82.0%		
P17_Pt_A	ATRX	K96Rfs*2	FS del	FS del	82.0%		
P17_Pt_A	FAT1	A4224T	Missense	Missense	13.1%		
P17_Pt_A	NOTCH4	C1091W	Missense	Missense	4.9%		
P17_Pt_A	BCL11B	D461N	Missense	Missense	9.8%		
P17_Pt_A	BRD4	R886W	Missense	Missense	6.6%		

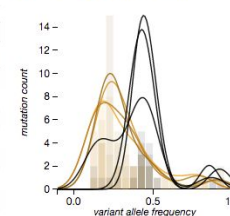
Showing 1-10 of 62 Mutations

Show more

Copy Number Alterations are not available.

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


☒ histogram ☒ density estimation



P17_Pt_A
P17_Pt_B
P17_Pt_C
P17_Rec1_A
P17_Rec1_B
P17_Rec1_C
P17_Rec1_D

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

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

Patient

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

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

All available patient-level clinical information

All available sample-level information

Patient View, Example 2: Sample Summary

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

Samples: 1 P17_Pr_A, Primary (Oligodendroglioma) 2 P17_Pr_B, Primary (Oligodendroglioma) 3 P17_Pr_C, Primary (Oligodendroglioma) 4 P17_Recl_A, Recurrence (Glioblastoma) 5 P17_Recl_B, Recurrence (Glioblastoma)

Summary

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

Time since diagnosis Specimen

Surgery Status

Treatment

Chemotherapy TMZ

0 1

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.

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

Samples: 1 P17_Pr_A, Primary

Low-Grade Gliomas (UCSF, Science 2014).

Summary

Time since diagnosis Specimen

Surgery Status

Treatment

Chemotherapy TMZ

MUT

16 Mutations (page 1 of 2)

Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Cohort	COSMIC
IDH1	R132H	🔵 🔴 🔥	Missense	0.43	100.0%	4964
TP53	C176F	🔵 🔴 🔥	Missense	0.87	90.2%	261
ATRX	I1035Efs*5	🔵	FS del		82.0%	
NOTCH4	C1091W	○	Missense	0.12	4.9%	
ABCA10	I998L	○	Missense	0.46	6.0%	
SUGCT	L69*	○	Nonsense	0.41	4.9%	
CD3EAP	N218Kfs*58	○	FS ins		4.9%	2
HDHD5	C392R	○	Missense	0.28	4.9%	
ADGRG7	C80S	○	Missense	0.38	4.9%	
CARMIL3	R858W	○	Missense	0.46	8.2%	

Showing 1-10 of 16 Mutations

Copy Number Alterations are not available.

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

DOWNLOAD DATA

Select Studies:

1 studies selected (500 samples) Deselect all View summary

uterine X

Uterus ?

☐ Select all listed studies matching filter (7)

Uterus

Endometrial Carcinoma

☒ Uterine Corpus Endometrial Carcinoma (TCGA, Nature 2013) 500 samples

☐ Uterine Corpus Endometrial Carcinoma (TCGA, PanCancer Atlas) 529 samples

☐ Uterine Corpus Endometrial Carcinoma (TCGA, Provisional) 548 samples

→ UTERINE CARCINOSARCOMA/UTERINE MALIGNANT MIXED MULLERIAN TUMOR

☐ Uterine Carcinosarcoma (Johns Hopkins University, Nat Commun 2014) 22 samples

☐ Uterine Carcinosarcoma (TCGA, PanCancer Atlas) 57 samples

☐ Uterine Carcinosarcoma (TCGA, Provisional) 57 samples

→ UTERINE CLEAR CELL CARCINOMA

☐ Uterine Clear Cell Carcinoma (NIH, Cancer 2017) 16 samples

Select Genomic Profiles:

☒ Mutations

☒ Putative copy-number alterations from GISTIC

☐ mRNA Expression. Select one of the profiles below:

☐ mRNA Expression z-Scores (microarray)

☐ mRNA Expression z-Scores (RNA Seq V2 RSEM)

Select Patient/Case Set:

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

Tumors with sequencing and CNA data (240) X

Enter Genes:

Advanced: Onco Query Language (OQL)

User-defined List X

Select from Recurrently Mutated Genes (MutSig)

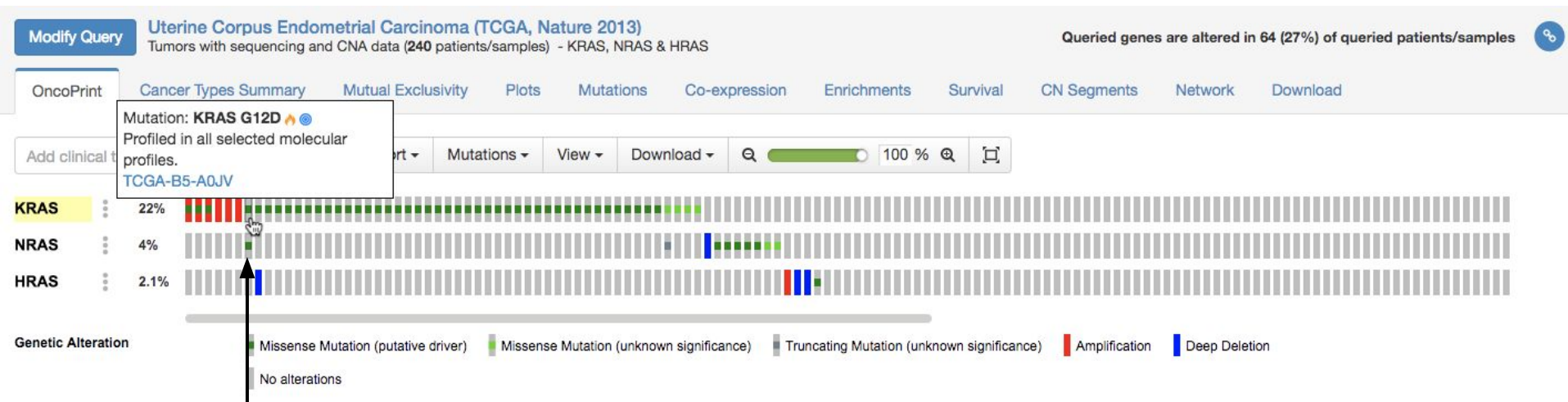
Select Genes from Recurrent CNAs (Gistic)

KRAS NRAS HRAS

All gene symbols are valid.

Submit Query

Example 3: OncoPrint

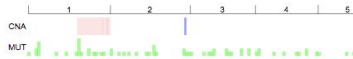


In general, mutations in these genes are mutually exclusive. However, there's one case with driver mutations in both KRAS and NRAS. Let's look at that patient in greater detail by clicking on the patient ID ("TCGA-B5-A0JV").

Example 3: Patient View

Patient: TCGA-B5-A0JV, 63 years old, Endometrial Ca
Samples: TCGA-B5-A0JV-01, Stage I

Summary Clinical Data Pathology Report Tissue



208 Mutations (page 1 of 21)

Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Copy #	mRNA Expr.	Cohort	COSMIC
NRAS	Q61K		Missense	0.19	Diploid	88%	3.6%	2297
PIK3CA	E542K		Missense	0.38	Diploid	74%	53.2%	787
KRAS	G12D		Missense	0.21	Diploid	55%	21.0%	25876
PTEN	P96T		Missense	0.23	Diploid	29%	64.3%	11
CCND1	T286I		Missense	0.50	Diploid	22%	6.0%	4
ARID1A	R693*		Nonsense	0.44	Diploid	34%	33.5%	8
SLX4	L1056Cfs*60		FS del		Diploid		6.9%	1
PMS2	N432S		Missense	0.19	Diploid	17%	4.0%	1
TET1	R81H		Missense	0.36	Diploid	52%	0.3%	
PPP4R2	G69V		Missense	0.18	Diploid	55%	1.6%	1

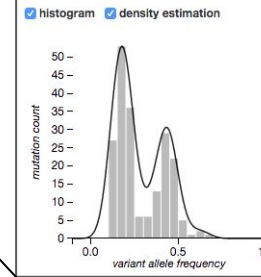
30 Copy Number Alterations (page 1 of 3)

Gene	CNA	Alt
CUL3	DeepDel	
IRS1	DeepDel	
DOCK10	DeepDel	2q36.2 7%
NYAP2	DeepDel	
RHBDD1	DeepDel	2q36.3 3%
COL4A4	DeepDel	2q36.3 20%
COL4A3	DeepDel	2q36.3 13%
MF1	DeepDel	2q36.3 2%
TM4SF20	DeepDel	2q36.3 89%
AGFG1	DeepDel	2q36.3 3%

Showing 1-10 of 30 Copy Number Alterations

Show more

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:

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