

cBioPortal Tutorial #3: Patient View

Investigate individual patients or samples in detail

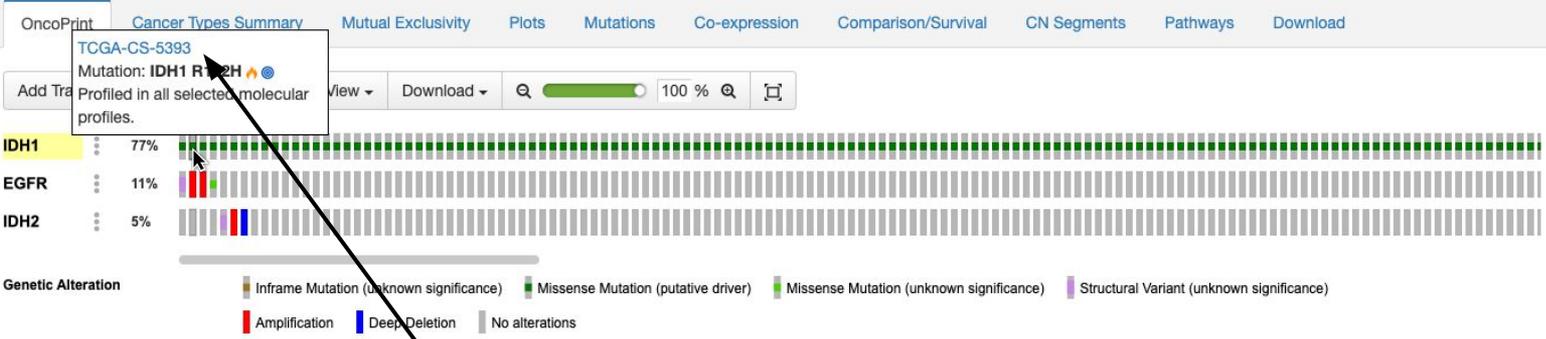
Tutorial Objectives

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



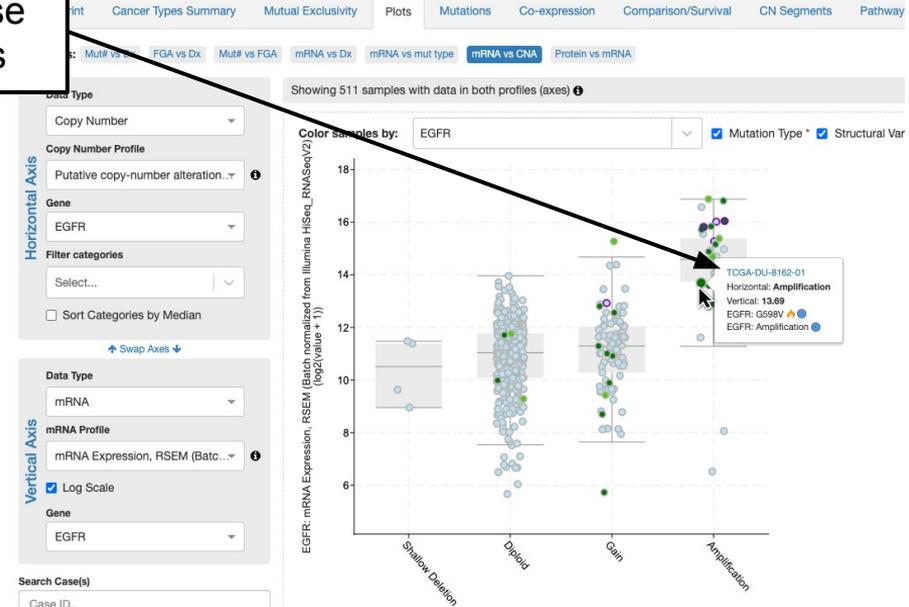
TCGA-CS-5393
 Mutation: IDH1 R132H
 Profiled in all selected molecular profiles.

Click on any of these sample/patient IDs



394 Mutations (page 1 of 16)

Sample ID	Cancer Type Detailed	Protein Change	Annotation	Mutation Type	Copy #
TCGA-DB-S276-01	Astrocytoma	R132C	Missense	Missense	Diploid
TCGA-DB-S278-01	Oligoastrocytoma	R132C	Missense	Missense	Diploid
TCGA-DB-A44X-01	Astrocytoma	R132C	Missense	Missense	Diploid
TCGA-DB-A4XF-01	Astrocytoma	R132C	Missense	Missense	Diploid
TCGA-DB-A64S-01	Oligoastrocytoma	R132C	Missense	Missense	Diploid
TCGA-DB-A75-01	Astrocytoma	R132C	Missense	Missense	Diploid



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



Summary

Clinical Data

CN Segments

View selected cases



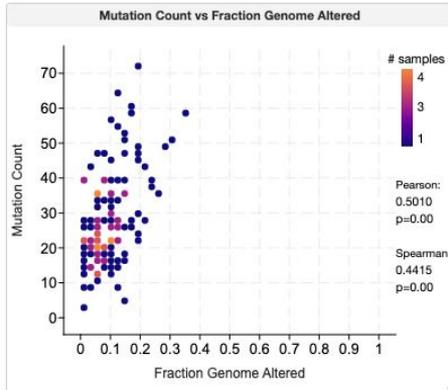
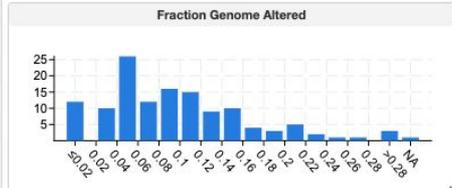
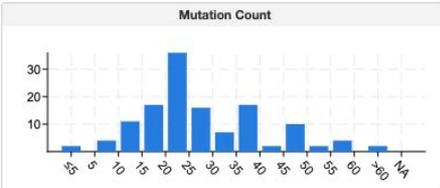
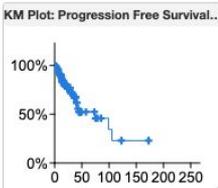
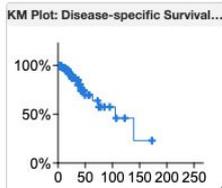
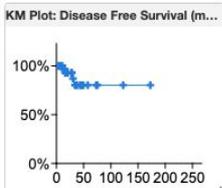
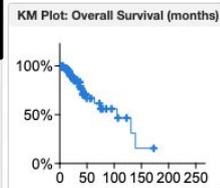
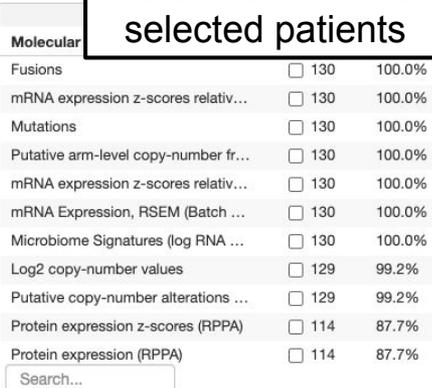
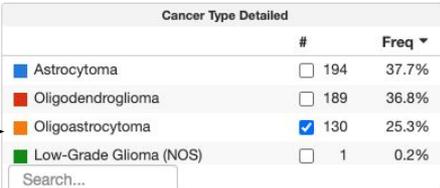
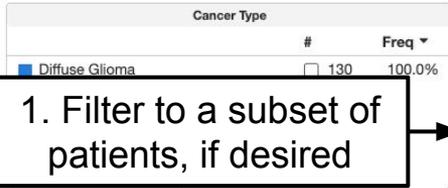
Custom Selection

Charts

Groups

1. Filter to a subset of patients, if desired

2. Click on this button to view the selected patients



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%
TTN	20	18	13.8%
MUC16	13	12	9.2%
FUBP1	10	9	6.9%
ARID1A	9	8	6.2%
EGFR	9	7	5.4%
NF1	9	7	5.4%
SPAG17	7	7	5.4%

Structural Variant Genes (130 profiled samples)

Gene	# SV	#	Freq
SEPTIN14	3	3	2.3%
KIF21B	2	2	1.5%
KCNJ6	2	2	1.5%
UBR1	2	2	1.5%
CNN2	2	2	1.5%
SCAMP2	2	2	1.5%
TTC3	2	2	1.5%
YJU2	2	2	1.5%
EGFR	2	2	1.5%
FGFR3	2	2	1.5%
SHC2	2	2	1.5%

CNA Genes (129 profiled samples)

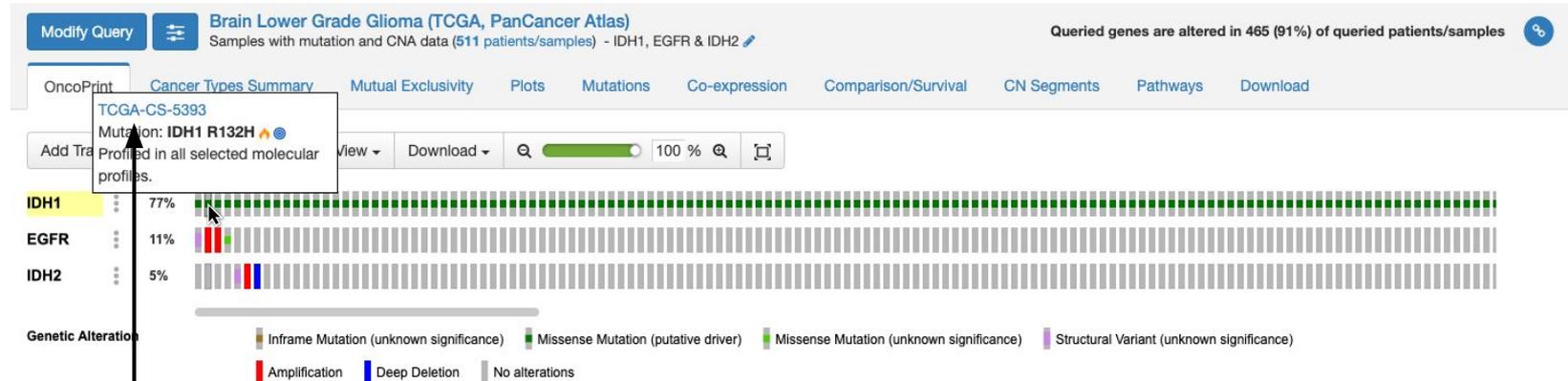
Gene	Cyband	CNA	#	Freq
LINC01238	2q37.3	HOMDEL	10	7.8%
GAL3ST2	2q37.3	HOMDEL	10	7.8%
RTP5	2q37.3	HOMDEL	10	7.8%
PDCD1	2q37.3	HOMDEL	10	7.8%
MTERF4	2q37.3	HOMDEL	10	7.8%
STK25	2q37.3	HOMDEL	10	7.8%
ANO7	2q37.3	HOMDEL	10	7.8%
FARP2	2q37.3	HOMDEL	10	7.8%
LINC01237	2q37.3	HOMDEL	10	7.8%
DTYMK	2q37.3	HOMDEL	10	7.8%
HDLBP	2q37.3	HOMDEL	10	7.8%

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

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.

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

Summary Pathways Clinical Data Pathology Report Tissue Image

CNA
MUT

24 Mutations (page 1 of 3)

Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Copy #	mRNA Expr.	Cohort	COSMIC
IDH1	R132H	⊙ ⊕ ⊖ ⊗ ⚡	Missense	0.31	Diploid	← 61%	→ 77%	4964
TP53	P250L	⊙ ⊕ ⊖ ⊗ ⚡	Missense	0.71	Diploid	← 60%	→ 48%	75
SMARCA4	H884R	⊙ ⊕ ⊖ ⊗ ⚡	Missense	0.30	Diploid	← 64%	→ 5%	3
BCL6	L112P	⊙ ⊕ ⊖ ⊗ ⚡	Missense	0.29	Diploid	← 89%	→ <1%	1
PRDM9	R587W	⊙ ⊕ ⊖ ⊗ ⚡	Missense	0.35	Diploid	← 97%	→ 1%	4
OR51F2	I264V	⊙ ⊕ ⊖ ⊗ ⚡	Missense	0.35	Diploid	← 99%	→ <1%	1
CCDC82	F483Y	⊙ ⊕ ⊖ ⊗ ⚡	Missense	0.28	Diploid	← 44%	→ <1%	1
OR8B4	S287L	⊙ ⊕ ⊖ ⊗ ⚡	Missense	0.37	Diploid	← 100%	→ <1%	1
KRT6G	M309T	⊙ ⊕ ⊖ ⊗ ⚡	Missense	0.34	Diploid	← 78%	→ <1%	1
IGSF6	D81E	⊙ ⊕ ⊖ ⊗ ⚡	Missense	0.44	Diploid	← 78%	→ <1%	1

Showing 1-10 of 24 Mutations

1 Structural Variants (page 1 of 1)

Gene 1	Gene 2	Annotation	Variant Class	Event Info	Connection Type
ADNP2	PAR6G	⊙ ⊕ ⊖ ⊗ ⚡	Fusion	ADNP2-PAR6G	

Showing 1-1 of 1 Structural Variants

271 Copy Number Alterations (page 1 of 28)

Gene	CNA	Annotation	Cytoband	mRNA Expr.	Cohort
EGFR	AMP	⊙ ⊕ ⊖ ⊗ ⚡	7p11.2	← 92%	→ 8%
ETV1	AMP	⊙ ⊕ ⊖ ⊗ ⚡	7p21.2	← 87%	→ <1%
GATA3	AMP	⊙ ⊕ ⊖ ⊗ ⚡	10p14	← 65%	→ 1%
HNRNPA2B1	AMP	⊙ ⊕ ⊖ ⊗ ⚡	7p15.2	← 33%	→ <1%
HOXA3	AMP	⊙ ⊕ ⊖ ⊗ ⚡	7p15.2	← 78%	→ <1%
HOXA9	AMP	⊙ ⊕ ⊖ ⊗ ⚡	7p15.2	← 56%	→ <1%
HOXA11	AMP	⊙ ⊕ ⊖ ⊗ ⚡	7p15.2	← 68%	→ <1%
HOXA13	AMP	⊙ ⊕ ⊖ ⊗ ⚡	7p15.2	← 95%	→ <1%
JAZF1	AMP	⊙ ⊕ ⊖ ⊗ ⚡	7p15.2-p15.1	← 87%	→ <1%
NOD1	AMP	⊙ ⊕ ⊖ ⊗ ⚡	7p14.3	← 46%	→ <1%

Showing 1-10 of 271 Copy Number Alterations

Patient View, Example 1: Pathways

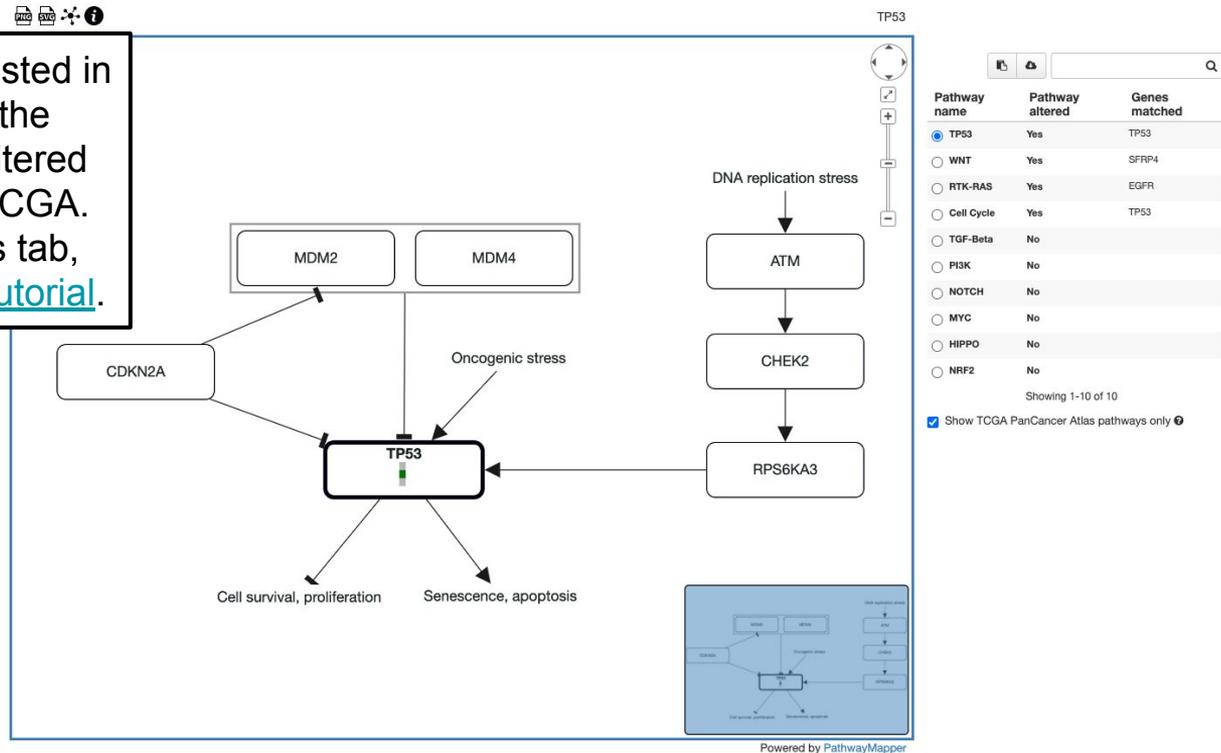
Patient: [TCGA-CS-5393](#), Male, 39 years old, Diffuse Glioma (Astrocytoma), [LIVING](#) (40 months), [DiseaseFree](#) (40 months) [Brain Lower Grade Glioma \(TCGA, PanCancer Atlas\)](#)

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

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



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](#).



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

Summary Pathways **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
International Classification of Diseases for Oncology, Third Edition ICD-O-3 Site	
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	

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

Attribute	TCGA-CS-5393-01
Mutation Count	24
Fraction Genome Altered	0.0569
MSI MANTIS Score	0.2715
MSIsensor Score	0
Sample Type	Primary
Aneuploidy Score	0
Cancer Type	Diffuse Glioma
Cancer Type Detailed	Astrocytoma
Neoplasm Histologic Grade	G3
Oncotree Code	DIFG
Somatic Status	Matched
Tissue Prospective Collection Indicator	No

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 Pathways 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 percentage of labeled nuclei determined. Over 1,000 cells were counted. The proliferation index ranged from 4.4% to 12.5% with an

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) Brain Lower Grade G
Samples: [TCGA-CS-5393-01](#), Primary

Summary Pathways Clinical Data Pathology Report **Tissue Image**

CANCER
Digital Slide Archive



LOGIN

HELP

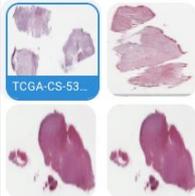
SLIDES

Tree

Thumbnails

Igg

TCGA-CS-5393



Slides view Metadata view

Metadata

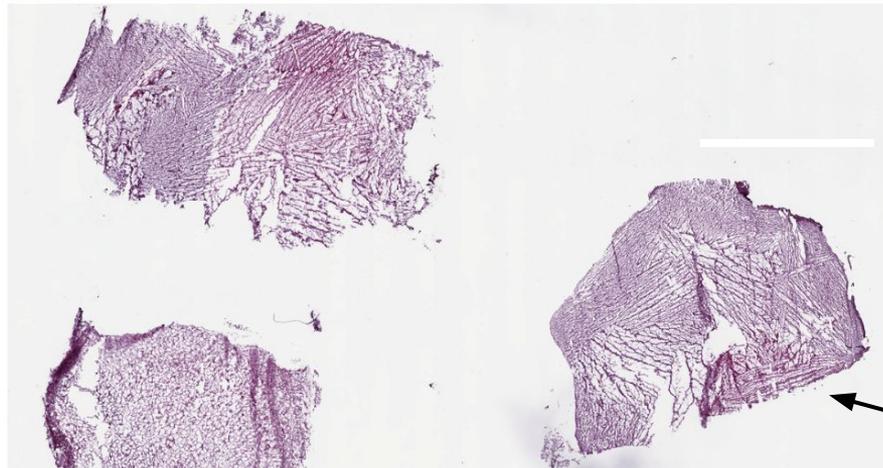
Apply Filters

Pathology Report



No Labels Drawing Disabled

Layer: Default Layer



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. [Click gene symbols below or enter here](#)

Number of Samples Per Patient : or

Summary Clinical Data Selected: 2 patients | 12 samples

Cancer Type Detailed

Cancer Type	#	Freq
Glioblastoma	5	41.7%
Oligoastrocytoma	4	33.3%
Oligodendroglioma	3	25.0%

Genomic Profile Sample Counts

Number of Samples...	#	Freq
2	17	73.9%
4	3	13.0%
3	1	4.3%
5	1	4.3%
7	1	4.3%

Molecular Profile

Mutations

Number of Samples Per Patient

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. [Click gene symbols below or enter here](#)

Number of Samples Per Patient : or

Summary Clinical Data Selected: 2 patients | 12 samples

Cancer Type Detailed

Cancer Type	#	Freq
Glioblastoma	5	41.7%
Oligoastrocytoma	4	33.3%
Oligodendroglioma	3	25.0%

Genomic Profile Sample Counts

Number of Samples...	#	Freq
2	12	100.0%

Molecular Profile

Mutations

Number of Samples Per Patient

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

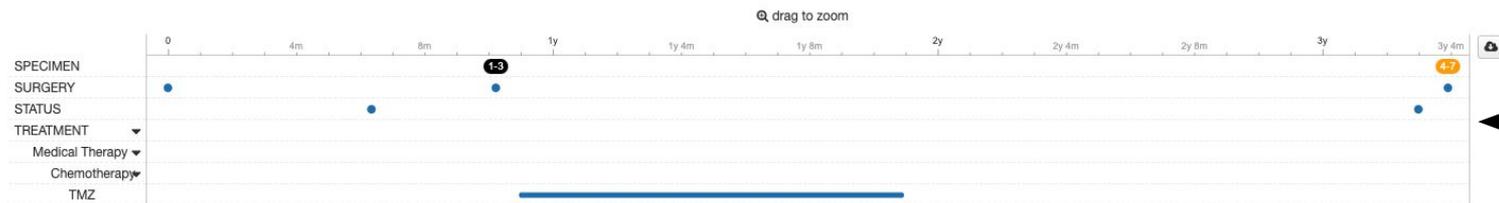
Patient View, Example 2: Patient Summary

 Patient: **P17**, Male, 27 years old, Glioma, **LIVING** (59 months)
 Low-Grade Gliomas (UCSF, Science 2014)

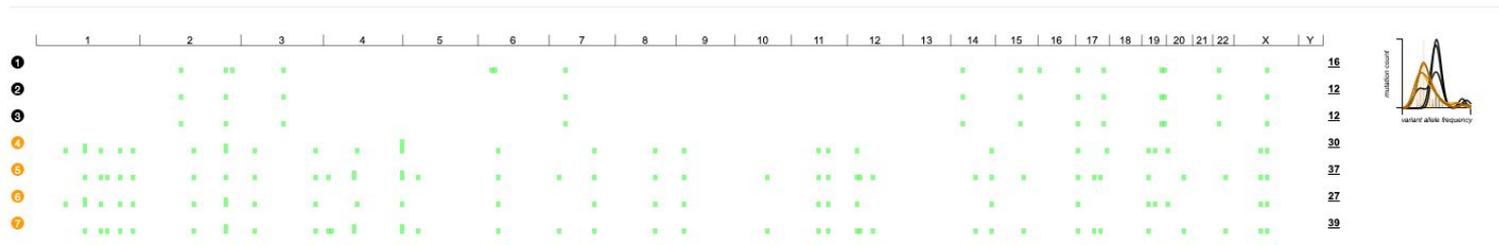
Samples:
 1 P17_Pr1_A, Primary (Oligodendroglioma)
2 P17_Pr1_B, Primary (Oligodendroglioma)
3 P17_Pr1_C, Primary (Oligodendroglioma)
4 P17_Rec1_A, Recurrence (Glioblastoma)
5 P17_Rec1_B, Recurrence (Glioblastoma)
6 P17_Rec1_C, Recurrence (Glioblastoma)
7 P17_Rec1_D, Recurrence (Glioblastoma)

<< < 1 of 2 patients > >>

Summary
Genomic Evolution
Pathways
Clinical Data



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



63 Mutations (page 1 of 7)

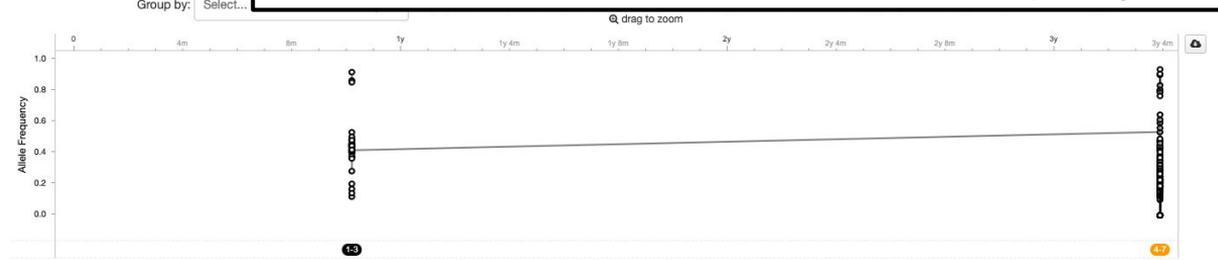
Samples	Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Cohort	COSMIC
1 2 3 4 5 6 7	IDH1	R132H	⊙ ⚙ ⚙ ⚙ ⚙ ⚙ ⚙	Missense	■■■■■	100%	4964
4 5 6 7	PIK3CA	H1047R	⊙ ⚙ ⚙ ⚙ ⚙ ⚙ ⚙	Missense	---	8%	1983
1 2 3	TP53	C176F	⊙ ⚙ ⚙ ⚙ ⚙ ⚙ ⚙	Missense	■■■	90%	261
4 5 6 7	TP53	S127F	⊙ ⚙ ⚙ ⚙ ⚙ ⚙ ⚙	Missense	■■■	90%	65
1 2 3	ATRX	I1035Efs*5	⊙	FS del	■■■	82%	

Patient View, Example 2: Genomic Evolution

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)

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 63 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	100%	100%	261
P17_Pr1_A, P17_Pr1_B, P17_Pr1_C, P17_Rec1_A, P17_Rec1_B, P17_Rec1_C, P17_Rec1_D	TP53	S127F	Missense	Missense	90%	90%	65
P17_Pr1_A, P17_Pr1_B, P17_Pr1_C, P17_Rec1_A, P17_Rec1_B, P17_Rec1_C, P17_Rec1_D	ATRX	I1035Efs*5	FS del	FS del	82%	82%	
P17_Pr1_A, P17_Pr1_B, P17_Pr1_C, P17_Rec1_A, P17_Rec1_B, P17_Rec1_C, P17_Rec1_D	ATRX	K96Rfs*2	FS del	FS del	82%	82%	
P17_Pr1_A, P17_Pr1_B, P17_Pr1_C, P17_Rec1_A, P17_Rec1_B, P17_Rec1_C, P17_Rec1_D	ARNT	F427L	Missense	Missense	11%	11%	
P17_Pr1_A, P17_Pr1_B, P17_Pr1_C, P17_Rec1_A, P17_Rec1_B, P17_Rec1_C, P17_Rec1_D	KAT6B	M1961V	Missense	Missense	7%	7%	
P17_Pr1_A, P17_Pr1_B, P17_Pr1_C, P17_Rec1_A, P17_Rec1_B, P17_Rec1_C, P17_Rec1_D	NOTCH4	C1091W	Missense	Missense	5%	5%	
P17_Pr1_A, P17_Pr1_B, P17_Pr1_C, P17_Rec1_A, P17_Rec1_B, P17_Rec1_C, P17_Rec1_D	FAT1	A4224T	Missense	Missense	13%	13%	

Showing 1-10 of 63 Mutations Show more

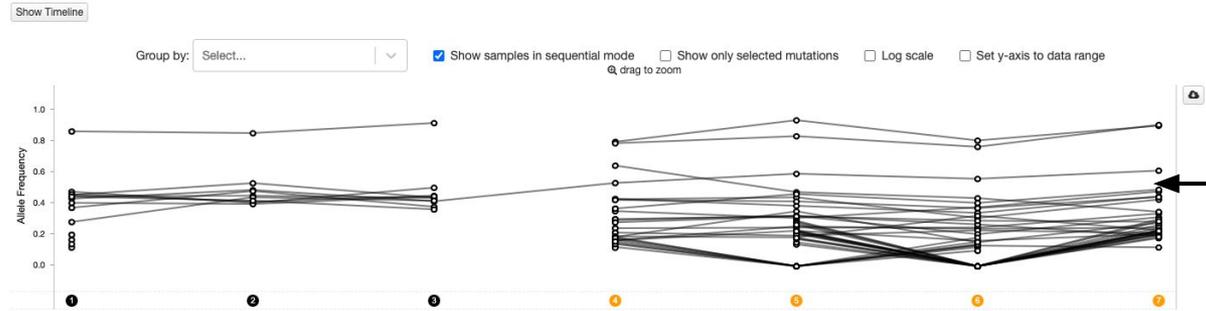
The Genomic Evolution tab is present for any patient with 2 or more samples. This tab provides visualizations to examine how mutation allele frequencies vary among samples and change over time. The Timeline (on the Summary tab) can also be shown on this tab to put allele frequency changes in context.

Patient View, Example 2: Genomic Evolution - Line Chart

Patient: P17, Male, 27 years old, Glioma, LIVING (59 months) Low-Grade Gliomas (UCSF, Science 2014)
 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)
 5 P17_Rec1_B, Recurrence (Glioblastoma)
 6 P17_Rec1_C, Recurrence (Glioblastoma)
 7 P17_Rec1_D, Recurrence (Glioblastoma)

[Summary](#) [Genomic Evolution](#) [Pathways](#) [Clinical Data](#)

[Line Chart](#) [Heatmap](#)



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.

Show only selected mutations **63 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
4 5 6 7	TP53	S127F		Missense		90%	65
1 2 3 4 5 6 7	ATRX	I1035Efs*5		FS del		82%	
4 5 6 7	ATRX	K96Rfs*2		FS del		82%	
4 5 6 7	ARNT	F427L		Missense		11%	
4 5 6 7	KAT6B	M1961V		Missense		7%	
1 4 5 6 7	NOTCH4	C1091W		Missense		5%	
4 5 6 7	FAT1	A4224T		Missense		13%	

Showing 1-10 of 63 Mutations

Patient View, Example 2: Clinical Data

Patient: P17, Male, 27 years old, Glioma, LIVING (59 months) Low-Grade Gliomas (UCSF, Science 2014)

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)

Summary Genomic Evolution Pathways **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	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)	0.5333333333333333	0.4	0.4	0.9666666666666666			

All available sample-level information

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

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

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: 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 all samples (log 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) 
- Protein expression z-scores (RPPA) 

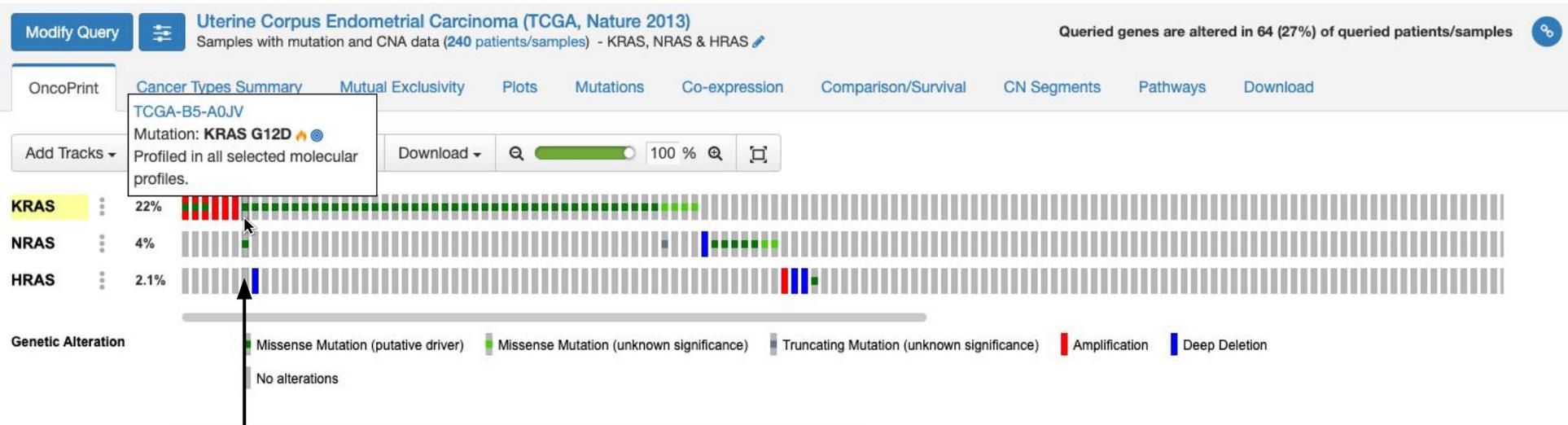
Select Patient/Case Set:

Enter Genes:

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

All gene symbols are valid.

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

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.

Patient: TCGA-B5-A0JV, Female, 63 years old, Endometrial Adenocarcinoma
Samples: TCGA-B5-A0JV-01, Primary, Stage I

Summary Pathways Clinical Data Pathology Reports



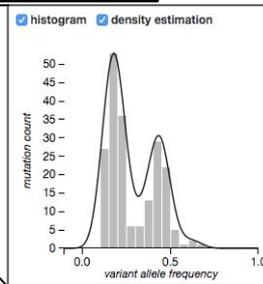
209 Mutations (page 1 of 21)

Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Copy #	mRNA Expr.	Cohort	COSMIC
NRAS	Q61K	Missense	Missense	0.19	Diploid	68%	4%	2297
PIK3CA	E542K	Missense	Missense	0.38	Diploid	74%	53%	787
KRAS	G12D	Missense	Missense	0.21	Diploid	55%	21%	25876
PTEN	P96T	Missense	Missense	0.23	Diploid	29%	65%	11
ARID1A	R693*	Nonsense	Nonsense	0.44	Diploid	34%	33%	8
CCND1	T286I	Missense	Missense	0.50	Diploid	22%	6%	4
SLX4	L1056Cfs*60	FS del	FS del		Diploid		7%	1
ARHGEF12	E37K	Missense	Missense	0.48	Diploid	54%	7%	1
PDE4DIP	E354K	Missense	Missense	0.18	Diploid	57%	6%	1
TPM3	E116D	Missense	Missense	0.30	Diploid	50%	13%	

Structural Variants are not available.

30 Copy Number Alterations (page 1 of 3)

Gene	CNA	Annotation	Cytoband	mRNA Expr.	Cohort
CUL3	DeepDel		2q36.2	2%	<1%
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%



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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