



# CANCER GENOMICS CONSORTIUM

*Educating for Best Practices in Clinical Cancer Genomics*

## cBioPortal

<http://www.cbioportal.org/>

<https://www.ncbi.nlm.nih.gov/pubmed/23550210>

<http://cancerdiscovery.aacrjournals.org/content/2/5/401>

# Tutorials

- <http://www.cbioportal.org/tutorial.jsp>
- <http://www.cbioportal.org/faq.jsp>
- <https://www.ncbi.nlm.nih.gov/pubmed/23550210>

# Home Page

The cBioPortal for Cancer Genomics provides **visualization, analysis and download** of large-scale **cancer genomics** data sets.  
Please cite Gao et al. *Sci. Signal.* 2013 & Cerami et al. *Cancer Discov.* 2012 when publishing results based on cBioPortal.

Study summary  
and links to  
PubMed

QUERY

DOWNLOAD DATA

Select Studies:

0 studies selected (0 samples) [Select all](#)

PanCancer Studies2

Cell lines2

Adrenal Gland1

Ampulla of Vater1

Biliary Tract5

Bladder/Urinary Tract7

Blood8

Bone2

Bowel5

Breast10

PanCancer Studies

☐ MSK-IMPACT Clinical Sequencing Cohort (MSKCC, Nat Med 2017)

☐ Pan-Lung Cancer (TCGA, Nat Genet 2016)

Cell lines

☐ Cancer Cell Line Encyclopedia (Novartis/Broad, Nature 2012)

☐ NCI-60 Cell Lines (NCI, Cancer Res. 2012)

Adrenal Gland

Adrenocortical Carcinoma

☐ Adrenocortical Carcinoma (TCGA, Provisional)

Ampulla of Vater

Ampullary Carcinoma

☐ Ampullary Carcinoma (Baylor College of Medicine, Cell Reports 2016)

10945 samples

1144 samples

1019 samples

60 samples

92 samples

160 samples

Select Data Type Priority:

☒ Mutation and CNA ☐ Only Mutation ☐ Only CNA

Enter Gene Set:

Advanced: Onco Query Language (OQL)

User-defined List

Enter HUGO Gene Symbols or Gene Aliases

Submit Query

Please select one or more cancer studies.

## What's New

[@cbioportal](#)

Sign up for low-volume email news alerts:

## Cancer Studies

The portal contains 162 cancer studies ([details](#))

## Example Queries

- RAS/RAF alterations in colorectal cancer
- BRCA1 and BRCA2 mutations in ovarian cancer
- POLE hotspot mutations in endometrial cancer
- TP53 and MDM2/4 alterations in GBM
- PTEN mutations in GBM in text format
- BRAF V600E mutations across cancer types
- Patient view of an endometrial cancer case

## Testimonials

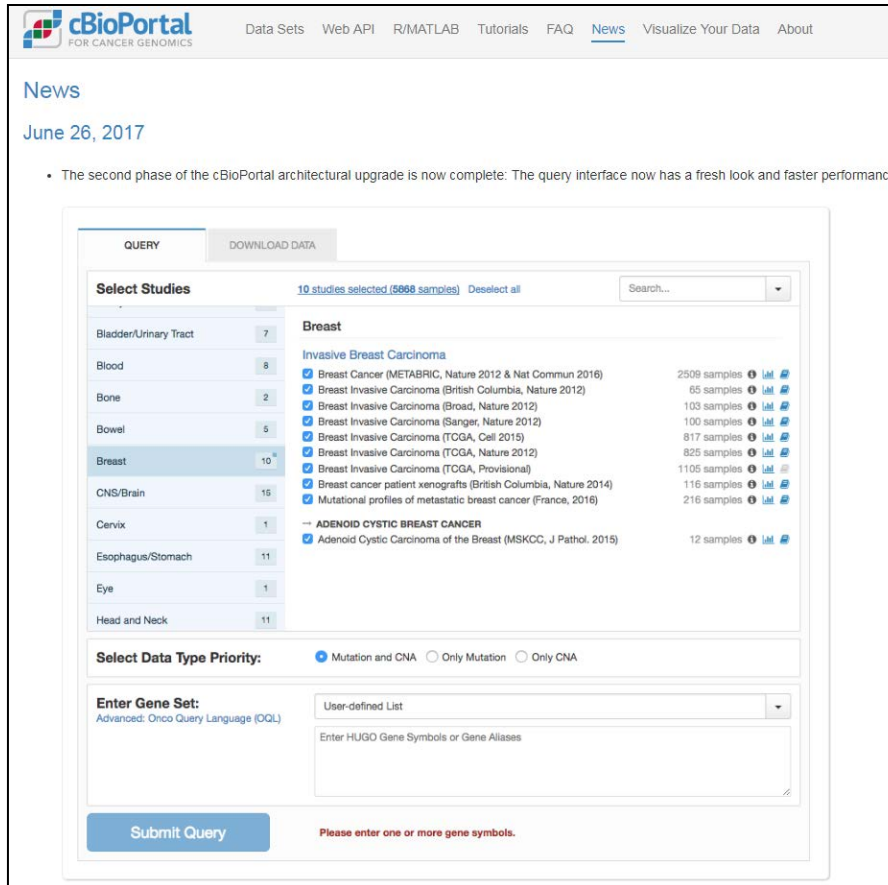
"Whenever bench scientists ask me how they can look at TCGA data, I've never had a good answer for them. Now I do. The cBio Portal meets a critical need—it is the interface that the cancer research community needs to access the wealth of TCGA. Even as a computational biologist, I use it to follow-up on genes of interest. It makes querying the data much less painful."

--Postdoctoral Fellow, Oregon Health & Science University

[View All](#)[Tell Us What You Think](#)

# News

- Click on “News” in top banner on home page.
- Upgrades to cBioPortal and additions to data are listed.



**cBioPortal** FOR CANCER GENOMICS

Data Sets Web API R/MATLAB Tutorials FAQ News Visualize Your Data About

## News

June 26, 2017

- The second phase of the cBioPortal architectural upgrade is now complete: The query interface now has a fresh look and faster performance.

**QUERY** DOWNLOAD DATA

10 studies selected (5098 samples) Deselect all Search...

Select Studies	Breast
Bladder/Urinary Tract 7	<b>Invasive Breast Carcinoma</b>
Blood 8	<input checked="" type="checkbox"/> Breast Cancer (METABRIC, Nature 2012 & Nat Commun 2016) 2509 samples
Bone 2	<input checked="" type="checkbox"/> Breast Invasive Carcinoma (British Columbia, Nature 2012) 65 samples
Bowel 5	<input checked="" type="checkbox"/> Breast Invasive Carcinoma (Broad, Nature 2012) 103 samples
Breast 10	<input checked="" type="checkbox"/> Breast Invasive Carcinoma (Sanger, Nature 2012) 100 samples
CNS/Brain 15	<input checked="" type="checkbox"/> Breast Invasive Carcinoma (TCGA, Cell 2015) 817 samples
Cervix 1	<input checked="" type="checkbox"/> Breast Invasive Carcinoma (TCGA, Nature 2012) 825 samples
Esophagus/Stomach 11	<input checked="" type="checkbox"/> Breast Invasive Carcinoma (TCGA, Provisional) 1105 samples
Eye 1	<input checked="" type="checkbox"/> Breast cancer patient xenografts (British Columbia, Nature 2014) 116 samples
Head and Neck 11	<input checked="" type="checkbox"/> Mutational profiles of metastatic breast cancer (France, 2016) 216 samples
	<b>ADENOID CYSTIC BREAST CANCER</b>
	<input checked="" type="checkbox"/> Adenoid Cystic Carcinoma of the Breast (MSKCC, J Pathol. 2015) 12 samples

**Select Data Type Priority:** ☒ Mutation and CNA ☐ Only Mutation ☐ Only CNA

**Enter Gene Set:** Advanced: Onco Query Language (OQL) User-defined List

Enter HUGO Gene Symbols or Gene Aliases

**Submit Query** Please enter one or more gene symbols.

# Data Sets

- Click on “Data Sets” in top banner on home page.
- All data that are housed in CBioPortal are arranged by Cancer study.
  - Data are available for download for most studies
  - Links to References (PubMed)
  - Summary of data involved in study
    - Total # cases
    - Type of data in study

**Data Sets**

The portal currently contains data from 159 cancer genomics studies. The table below lists the number of available samples per cancer study and data type.

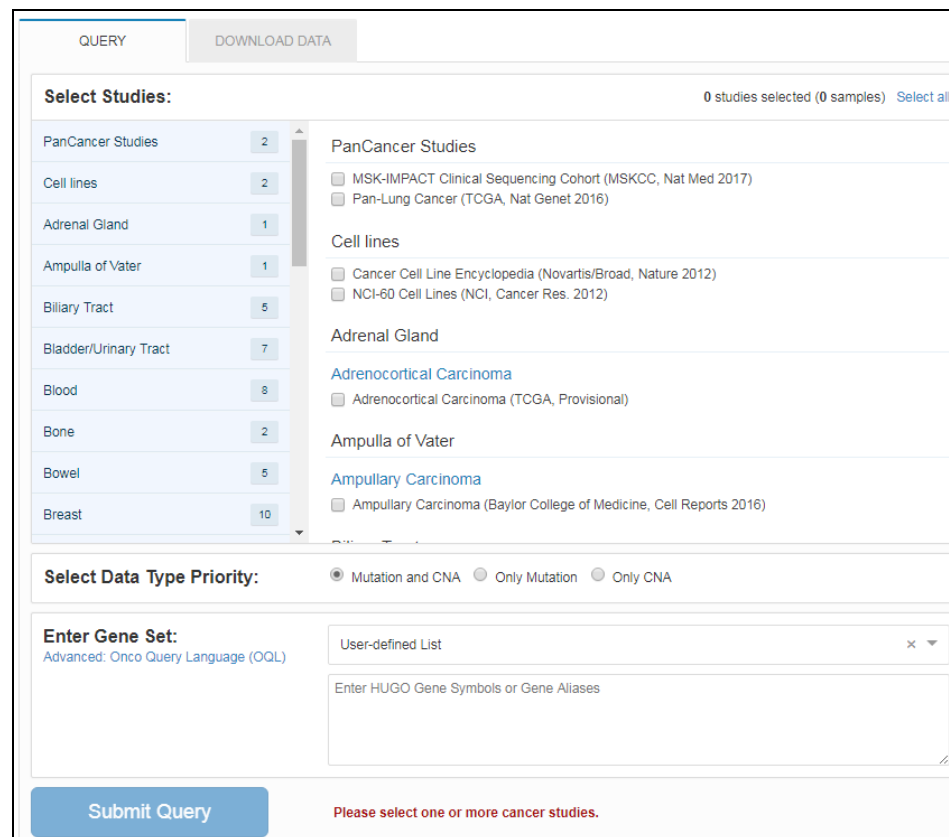
CancerStudy	Reference	All	Sequenced	CNA	Tumor mRNA (RNA-Seq)	Tumor mRNA (microarray)	Tumor mRNA	Methylation (HM27)	RPPA	Complete
Acinar Cell Carcinoma of the Pancreas (Johns Hopkins, J Pathol 2014)	Jin et al. J Pathol 2014	23	23							
Acute Myeloid Leukemia (TCGA, NEJM 2013)	TCGA, NEJM 2013	200	200	191	172			194		166
Acute Myeloid Leukemia (TCGA, Provisional)		200	197	181	172			194		163
Adenoid Cystic Carcinoma (FHM, Am J Surg Pathol 2014)	Ross et al. Am J Surg Pathol 2014	28	13	28						
Adenoid Cystic Carcinoma (HDA, Clin Cancer Res 2015)	Hilal et al. Clin Cancer Res 2015	102	102							
Adenoid Cystic Carcinoma (MSKCC, Nat Genet 2013)	Ho et al. Nat Genet 2013	60	60	60						
Adenoid Cystic Carcinoma (Sanger/HDA, JCI 2013)	Stephens et al. JCI 2013	24	24							
Adenoid Cystic Carcinoma of the Breast (MSKCC, J Pathol 2015)	Hartelotto et al. J Pathol 2015	12	12	12						
Adenocarcinoma (TCGA, Provisional)		92	90	90	79				46	75
Ampullary Carcinoma (Baylor College of Medicine, Cell Reports 2016)		160	160							
Bladder Cancer (MSKCC, Eur Urol 2014)	Kim et al. Eur Urol 2014	109	109	109						
Bladder Cancer (MSKCC, JCO 2013)	Iyer et al. JCO 2013	97	97	97		50		24		58
Bladder Cancer, Plasmacytoid Variant (MSKCC, Nat Genet 2016)		34	34	33						
Bladder Urothelial Carcinoma (RGI, Nat Genet 2013)	Guo et al. Nat Genet 2013	99	99							
Bladder Urothelial Carcinoma (Dana Farber & MSKCC, Cancer Discov 2014)	Van Allen et al. Cancer Discov 2014	50	50							
Bladder Urothelial Carcinoma (TCGA, Nature 2014)	TCGA, Nature 2014	131	130	120	129				120	125
Bladder Urothelial Carcinoma (TCGA, Provisional)		413	130	408	408				344	126
Brain Lower Grade Glioma (TCGA, Provisional)		530	286	513	530				425	283
Breast Cancer (HETARRIC, Nature 2012 & Nat Commun 2016)	Pereira et al. Nat Commun 2016	2569	2565	2173						
Breast cancer patient xenografts (British Columbia, Nature 2014)	Eisew et al. Nature 2014	116	116	30						
Breast Invasive Carcinoma (British Columbia, Nature 2012)	Shah et al. Nature 2012	65	65							
Breast Invasive Carcinoma (Broad, Nature 2012)	Banerjee et al. Nature 2012	103	103							
Breast Invasive Carcinoma (Sanger, Nature 2012)	Stephens et al. Nature 2012	100	100							
Breast Invasive Carcinoma (TCGA, Cell 2015)	TCGA, Cell 2015	817	817	816	817	421		264	672	816
Breast Invasive Carcinoma (TCGA, Nature 2012)	TCGA, Nature 2012	825	507	778	778	526	302	311	410	
Breast Invasive Carcinoma (TCGA, Provisional)		1105	982	1080	1100	529		316	892	960
Cancer Cell Line Encyclopedia (Novartis/Broad, Nature 2012)	Barretina et al. Nature 2012	1019	905	995		967				877
Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA, Provisional)		309	194	255	306				173	190

# Web API and R/MATLAB Packages

- Click on “Web API” in top banner on home page.
- Web interface API code is provided to connect with the Cancer Genomic Data Server (CGDS)
- Can also access CGDS through R or MATLAB statistical software
  - Installation instructions for packages available under “R/MATLAB” in top banner on home page.
- Create a local instance of cBioPortal for your laboratory data through GitHub (<https://github.com/cBioPortal/cbioportal>) or Wiki pages (<https://github.com/cBioPortal/cbioportal/wiki>)

# Setting up a Query


- Select Cancer Study / Data Set(s) to query
- Select Data Type(s) of interest
- Select/Enter Gene Set to interrogate
  - User Defined List
    - Can be copied/pasted into window.
    - Can follow Onco Query Language if you are looking for specific alterations in genes.
  - Pre-defined list
    - Will populate in window if selected from drop-down

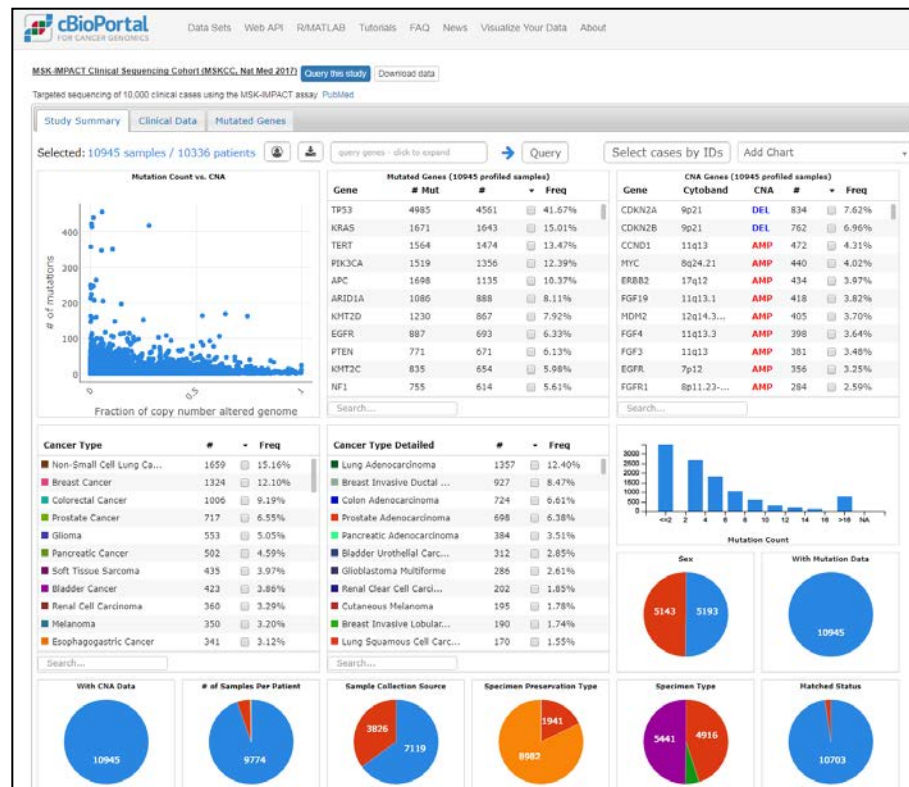


The screenshot shows the cBioPortal Query interface. At the top, there are tabs for 'QUERY' and 'DOWNLOAD DATA'. Below the tabs, the 'Select Studies:' section shows a list of studies on the left and a detailed view on the right. The left list includes categories like PanCancer Studies, Cell lines, Adrenal Gland, Ampulla of Vater, Biliary Tract, Bladder/Urinary Tract, Blood, Bone, Bowel, and Breast, each with a count. The right view shows the details for the selected studies, including checkboxes for 'MSK-IMPACT Clinical Sequencing Cohort (MSKCC, Nat Med 2017)', 'Pan-Lung Cancer (TCGA, Nat Genet 2016)', 'Cancer Cell Line Encyclopedia (Novartis/Broad, Nature 2012)', 'NCI-60 Cell Lines (NCI, Cancer Res. 2012)', 'Adrenocortical Carcinoma (TCGA, Provisional)', and 'Ampullary Carcinoma (Baylor College of Medicine, Cell Reports 2016)'. Below the studies section, the 'Select Data Type Priority:' section has radio buttons for 'Mutation and CNA' (selected), 'Only Mutation', and 'Only CNA'. The 'Enter Gene Set:' section has a dropdown menu set to 'User-defined List' and a text input field for 'Enter HUGO Gene Symbols or Gene Aliases'. At the bottom, there is a 'Submit Query' button and a red error message: 'Please select one or more cancer studies.'



# Study Summary

- Prior to setting up a query based on specific studies, you might want to look at a summary of the data from those studies.
  - Click on  button on home page next to desired study.





# Onco Query Language

- Guide is visible in separate window when link is selected from home page near Gene Set window

Data Type	Keyword	Categories and Levels	Default
Copy Number Alterations	CNA	AMP Amplified HOMDEL Deep Deletion GAIN Gained HETLOSS Shallow Deletion	AMP and HOMDEL
Mutations	MUT	MUT Show mutated cases MUT = X Specific mutations or mutation types.	All somatic, non-synonymous mutations
Fusions	FUSION	FUSION	Show cases with fusions
mRNA Expression	EXP	EXP < -x Under-expression is less than x SDs below the mean. EXP > x Over-expression is greater than x SDs above the mean. The comparison operators <= and >= also work.	At least 2 standard deviations (SD) from the mean.
Protein/phosphoprotein level (RPPA)	PROT	PROT < -x Protein-level under-expression is less than x SDs below the mean. PROT > x Protein-level over-expression is greater than x SDs above the mean. The comparison operators <= and >= also work.	At least 2 standard deviations (SD) from the mean.

# Example Queries

- Example Queries are available on bottom right panel of home page.
- Clicking on these links will bring you straight to the data and will not show you the query screen unless you click “Modify Query” on resulting screen.
- Examples are useful if you are running a similar query.

## Example Queries

- [RAS/RAF alterations in colorectal cancer](#)
- [BRCA1 and BRCA2 mutations in ovarian cancer](#)
- [POLE hotspot mutations in endometrial cancer](#)
- [TP53 and MDM2/4 alterations in GBM](#)
- [PTEN mutations in GBM in text format](#)
- [BRAF V600E mutations across cancer types](#)
- [Patient view of an endometrial cancer case](#)

# Query

- Simple Query for mutations and CNAs in several B-ALL genes across all ALL studies in cBioPortal.

Query
Download Data

Select Cancer Study:

Search...
2 studies selected. Deselect all

- Blood (8)
  - Leukemia (6)
    - ☒ Acute Lymphoid Leukemia (2)
      - ☒ Hypodiploid Acute Lymphoid Leukemia (St Jude, Nat Genet 2013) 44 samples
      - ☒ Infant MLL-Rearranged Acute Lymphoblastic Leukemia (St Jude, Nat Genet 2015) 62 samples
    - ☐ Acute Myeloid Leukemia (2)
      - ☐ Acute Myeloid Leukemia (TCGA, Provisional) 200 samples
      - ☐ Acute Myeloid Leukemia (TCGA, NEJM 2013) 200 samples
    - ☐ Chronic Lymphocytic Leukemia (2)

Select Data Type Priority:
☒ Mutation and CNA
☐ Only Mutation
☐ Only CNA

Enter Gene Set:
Advanced: Onco Query Language (OQL)

User-defined List

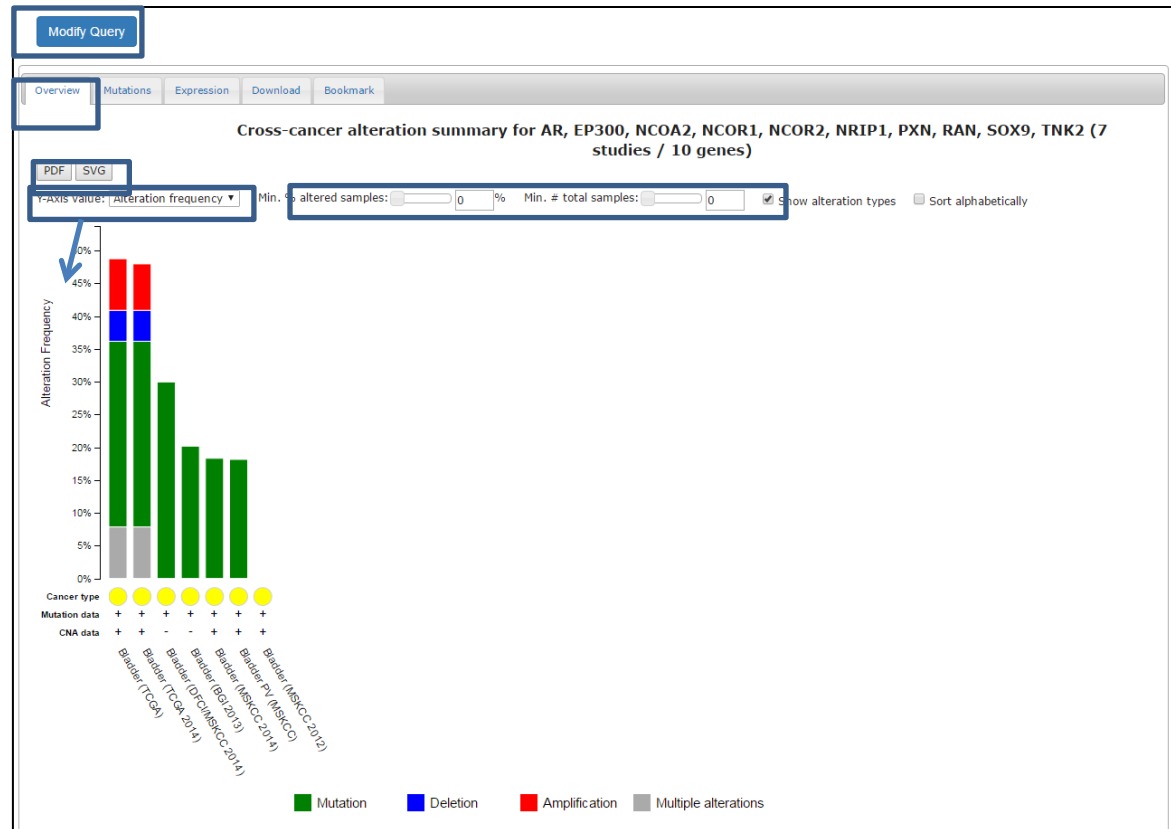
PBX1  
PDGFRA  
PDGFRB  
PIK3AP1  
PML

☒ All gene symbols are valid.

Submit

# Query Results: Overview

- Overview of alterations in your desired data set are viewed as bar graph and can be downloaded as PDF or SVG.
- Y-axis can be changed to illustrate absolute counts rather than alteration frequency.
- If interrogating many data sets, the minimum % altered samples and minimum total samples can be adjusted to show only the most pertinent data.
- Alteration types are color-coded
- Query can be modified to produce more/less results.



# Query Results: Download and Bookmark

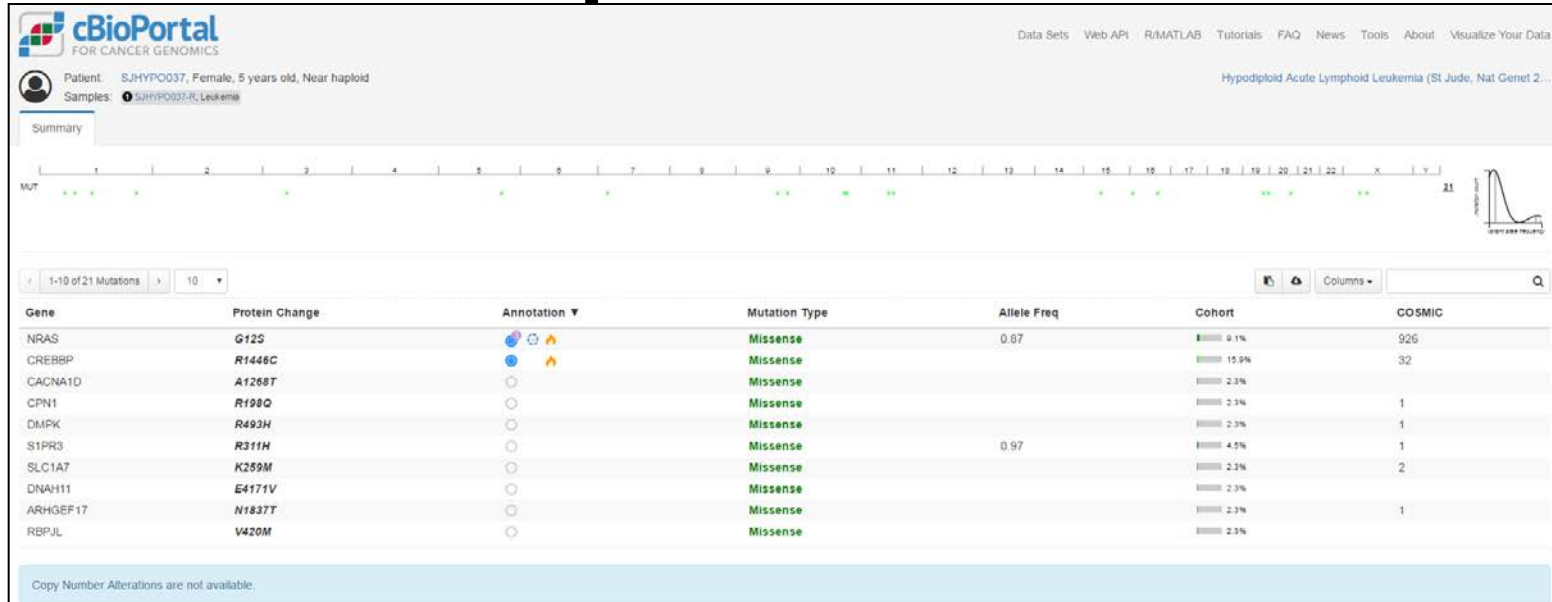
- Click on “Download” tab to copy and paste summary results into Excel.
- Click on “Bookmark” for links you can bookmark to save your query results

# Query Results: Mutations

- Each gene in your query will have its own tab.
  - Within the tab will be
    - Gene schematic with visualization of domains and observed mutations
      - 3D protein structure visualization available
    - Table of all observed mutations with sample and mutation information.
      - Available for download
  - If no mutations found, tab will be empty



# Mutation Information: Sample ID



- Click on Sample ID to see
  - Patient demographics
  - All mutations within a given sample by chromosomes and in a table (download available)
  - Mutation annotation – OncoKB, My Cancer Genome, Cancer HotSpots
  - % samples in study with same/similar mutations
  - COSMIC hits



# Mutation Information: Cancer Study: Study Summary

- Can Filter by Any of the criteria on Study Summary Page

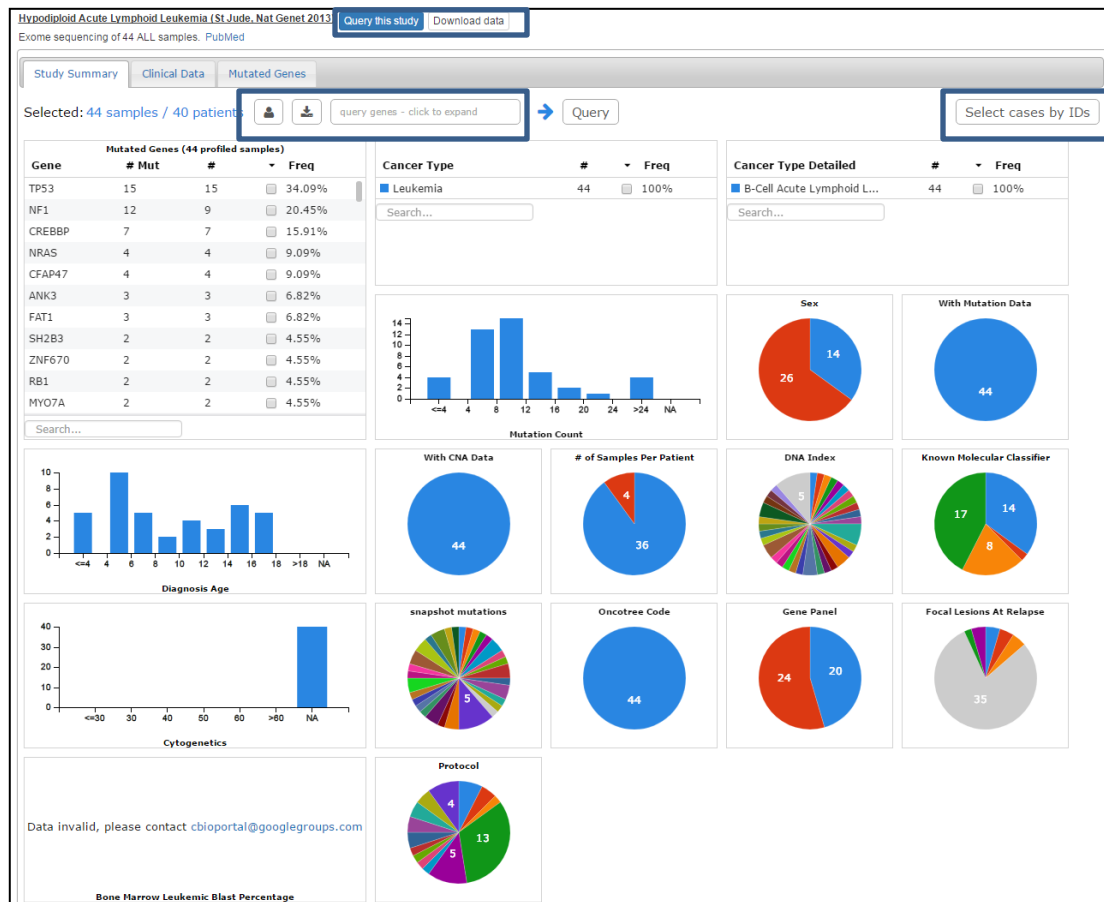
- Mutated Genes: number and % frequency
- Diagnosis Age
- Cytogenetics – chromosome count
- Cancer type – Broad, Detailed, or Oncotree
- Sex
- % and # Cases with Mutation data and CNA data
- Molecular Classification
- Gene Panel (if applicable)
- Focal Lesions at Relapse
- Snapshot Mutations
- Treatment Protocol
- DNA Index
- Blast % (if applicable)

- Link to PubMed reference

- Can Download information

- Can Select Patients to view

- Can query specific gene(s) within study

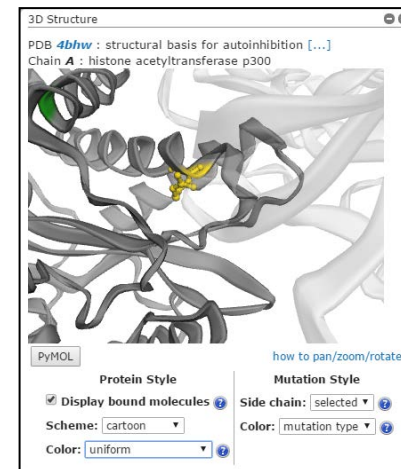
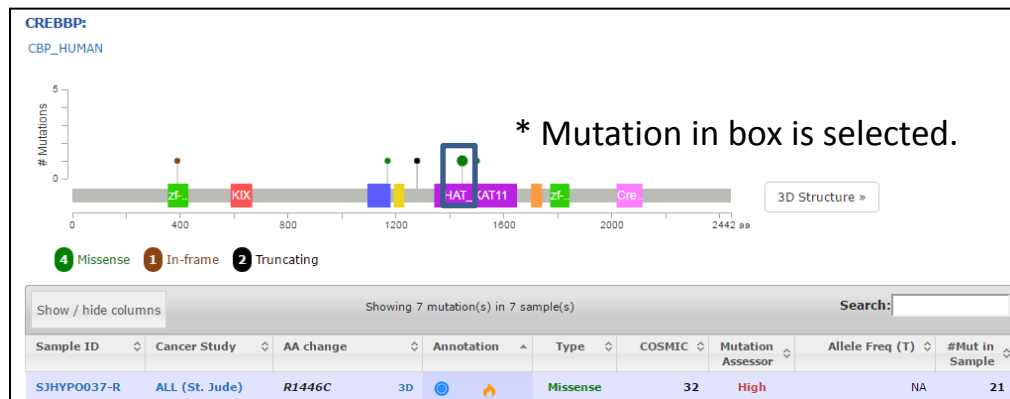


# Mutation Information: Cancer Study: Clinical Data and Mutated Genes




- Clinical data table is available for each study in cBioPortal. Can filter by:
  - Patient or Sample ID
  - Blast %
  - Name of cancer study or cancer type
- Mutated genes table contains information about gene
  - Cytoband
  - Gene size (nts)
  - # mutations observed in study
  - # mutations per nucleotide

# Mutation Information: AA Change

- Click on this row to highlight mutation marker on gene schematic.
- Click on 3D link to view mutation in context of protein structure.
  - Several visualization options for protein structure.



# Mutation Information: Annotation

- Click on Blue/Gray Target for OncoKB annotations 
  - Numbers attached to target indicates therapeutic information available – See levels of evidence in OncoKB presentation
  - Citations included in pop-up window
- Click on Mv Cancer Genome symbol for MCG links 
- Flame icon indicates recurrent cancer hotspot (from <http://cancerhotspots.org/>) 

# Other Mutation Information

- Mutation tab will contain
  - Type of mutation
  - # COSMIC entries
  - Mutation Assessor
    - Predicts functional impact
    - Link to <http://mutationassessor.org>
    - Allele Frequency (mutated reads/total reads)
  - # Mutations in sample

Sample(s) <span>Search: <input type="text"/></span>				
Type ▾	COSMIC ▾	Mutation Assessor ▾	Allele Freq (T) ▾	#Mut in Sample ▾
Missense	32	High	NA	21
Splice			0.68	5
Splice			NA	9
Missense	3	Medium	0.98	4
Missense	3	Medium	0.98	6
IF del			1.00	4
Missense	3	Medium	0.34	19

# Expression Data

- Can be used to provide evidence for mutations affecting expression levels.
- Can view one gene at a time
- Data is downloadable

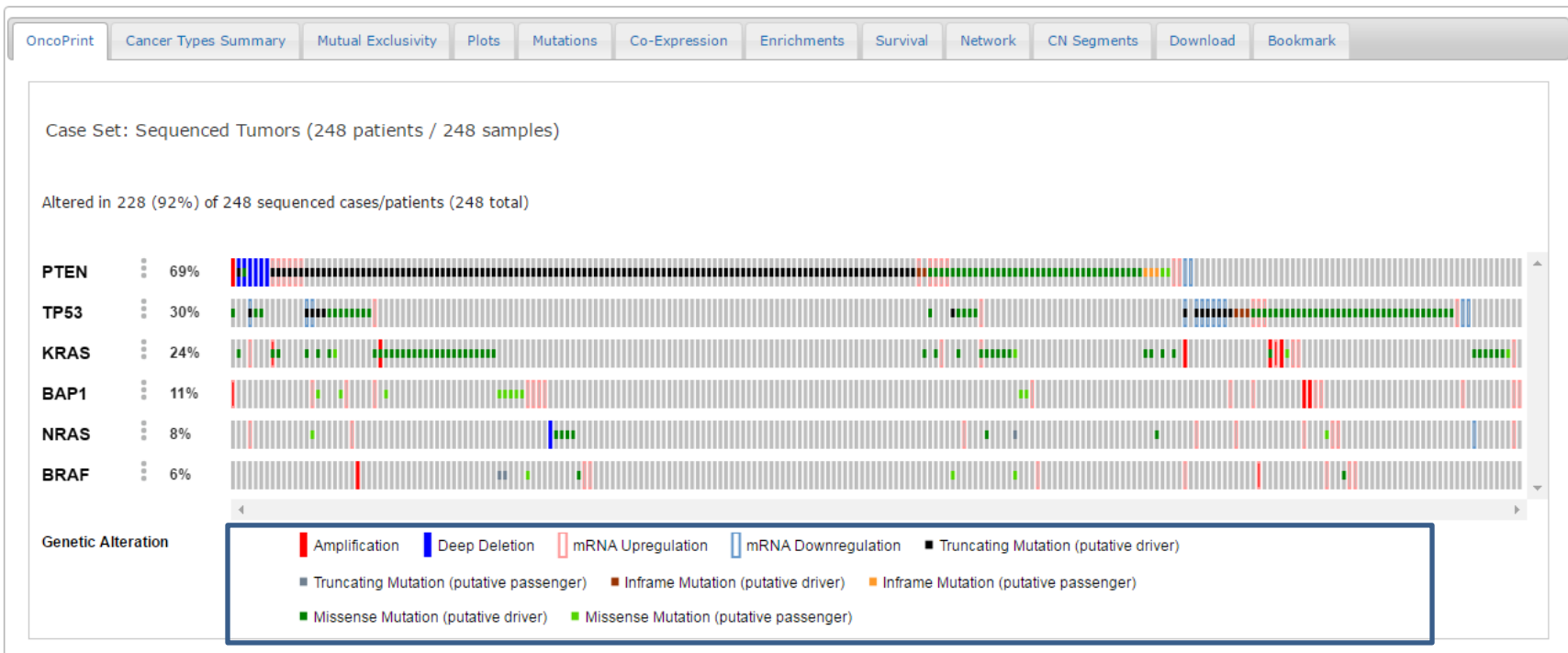


# OncoPrint as Query Result

[Modify Query](#)

**Uterine Corpus Endometrial Carcinoma (TCGA, Nature 2013)**  
Sequenced Tumors (248 samples) / 6 Genes

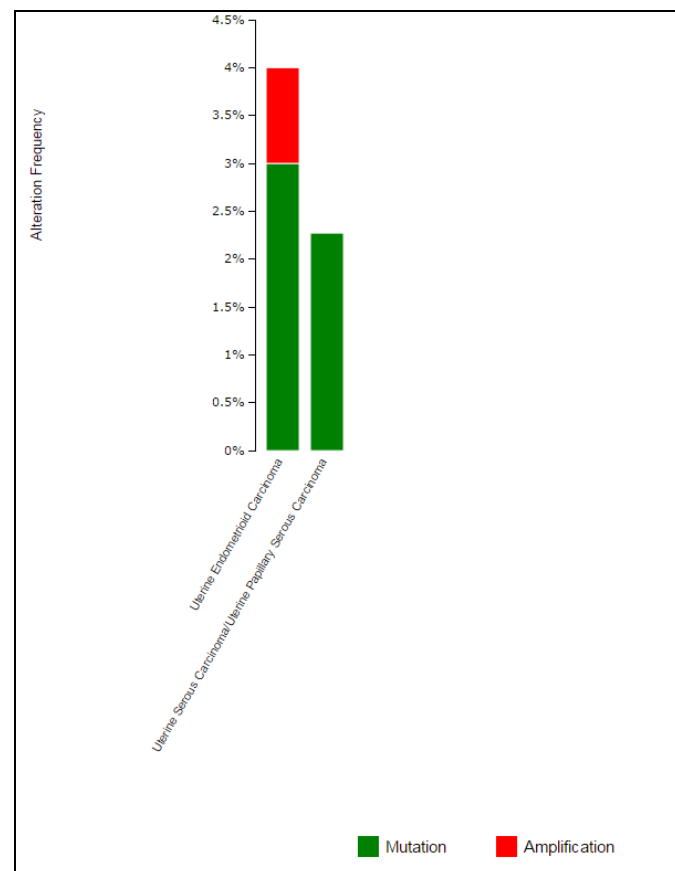
Gene Set / Pathway is altered in 228 (91.9%) of queried samples





# Cancer Types Summary

- If study contains multiple cancer types, data will stratify mutation by cancer type per gene in this tab resulting from query.



# Mutual Exclusivity

- Tab in query result will show which gene pairs have mutually exclusive mutations and which gene pairs have co-occurring mutations
- P-value of less than 0.05 = significant
- Results are downloadable

The query contains 6 gene pairs with mutually exclusive alterations (1 significant), and 9 gene pairs with co-occurrent alterations (2 significant).

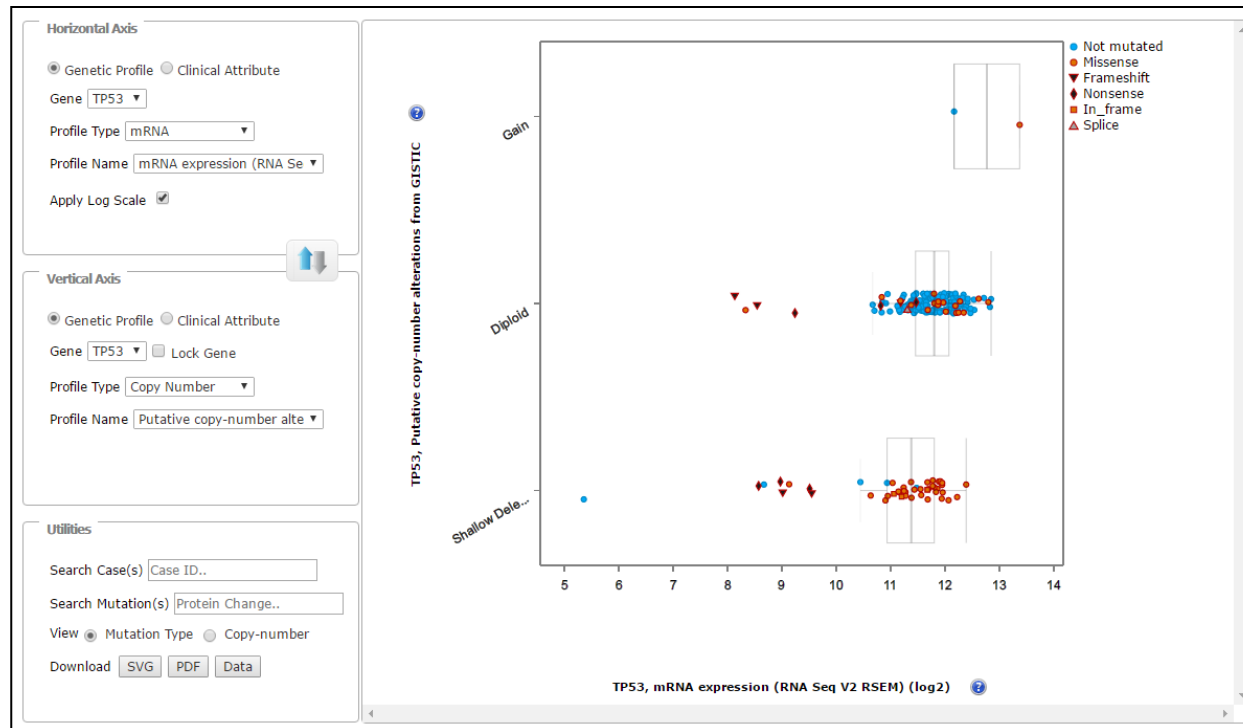
<input checked="" type="checkbox"/> Mutual exclusivity <input checked="" type="checkbox"/> Co-occurrence <input type="checkbox"/> Significant pairs <span>Search Gene</span>				
Gene A	Gene B	p-Value	Log Odds Ratio	Association
PTEN	TP53	<0.001	-2.282	Tendency towards mutual exclusivity <b>Significant</b>
TP53	BAP1	0.038	0.815	Tendency towards co-occurrence <b>Significant</b>
NRAS	TP53	0.040	0.941	Tendency towards co-occurrence <b>Significant</b>
BRAF	TP53	0.066	0.922	Tendency towards co-occurrence
KRAS	PTEN	0.080	0.528	Tendency towards co-occurrence
KRAS	BAP1	0.142	-0.717	Tendency towards mutual exclusivity
BRAF	KRAS	0.209	-0.847	Tendency towards mutual exclusivity
KRAS	TP53	0.219	-0.316	Tendency towards mutual exclusivity
BRAF	BAP1	0.265	0.648	Tendency towards co-occurrence
PTEN	NRAS	0.267	-0.409	Tendency towards mutual exclusivity
BRAF	NRAS	0.377	0.530	Tendency towards co-occurrence
NRAS	BAP1	0.398	0.360	Tendency towards co-occurrence
PTEN	BAP1	0.544	-0.036	Tendency towards mutual exclusivity
KRAS	NRAS	0.557	0.047	Tendency towards co-occurrence
BRAF	PTEN	0.613	0.010	Tendency towards co-occurrence

Showing 1 to 15 of 15 entries

[Download Full Result](#)

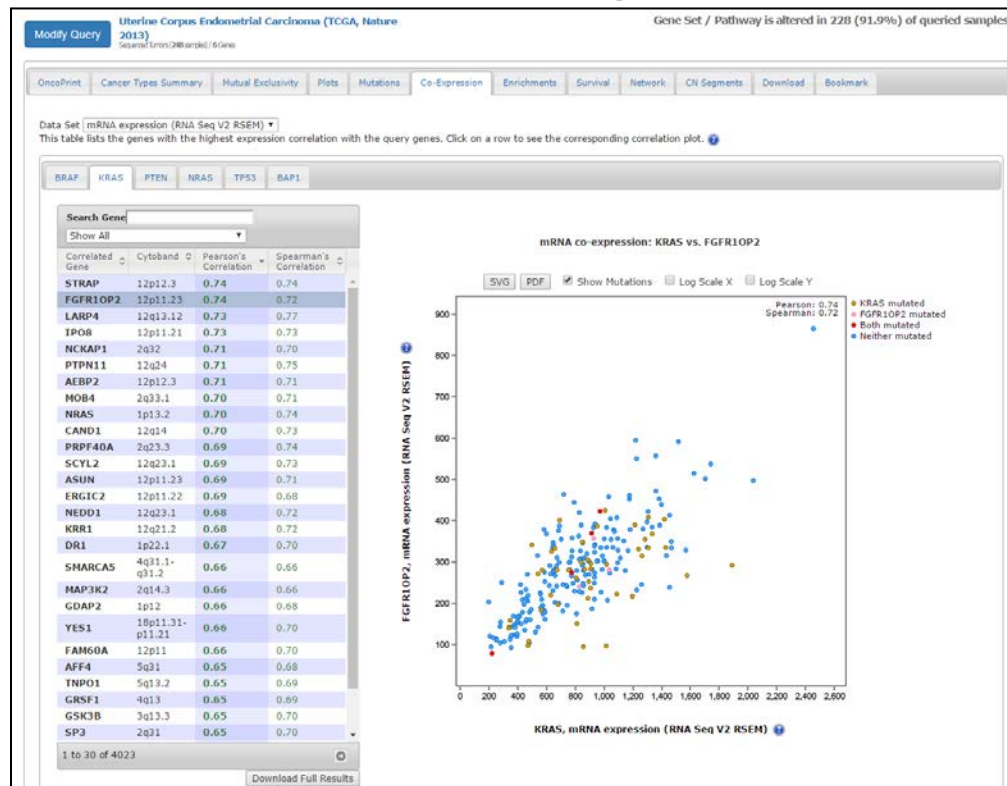
# Plots

- Can choose data type and gene for both horizontal and vertical axes.
- Can also search case IDs if you are curious where data will lie in context of cohort
- Plots are available for download



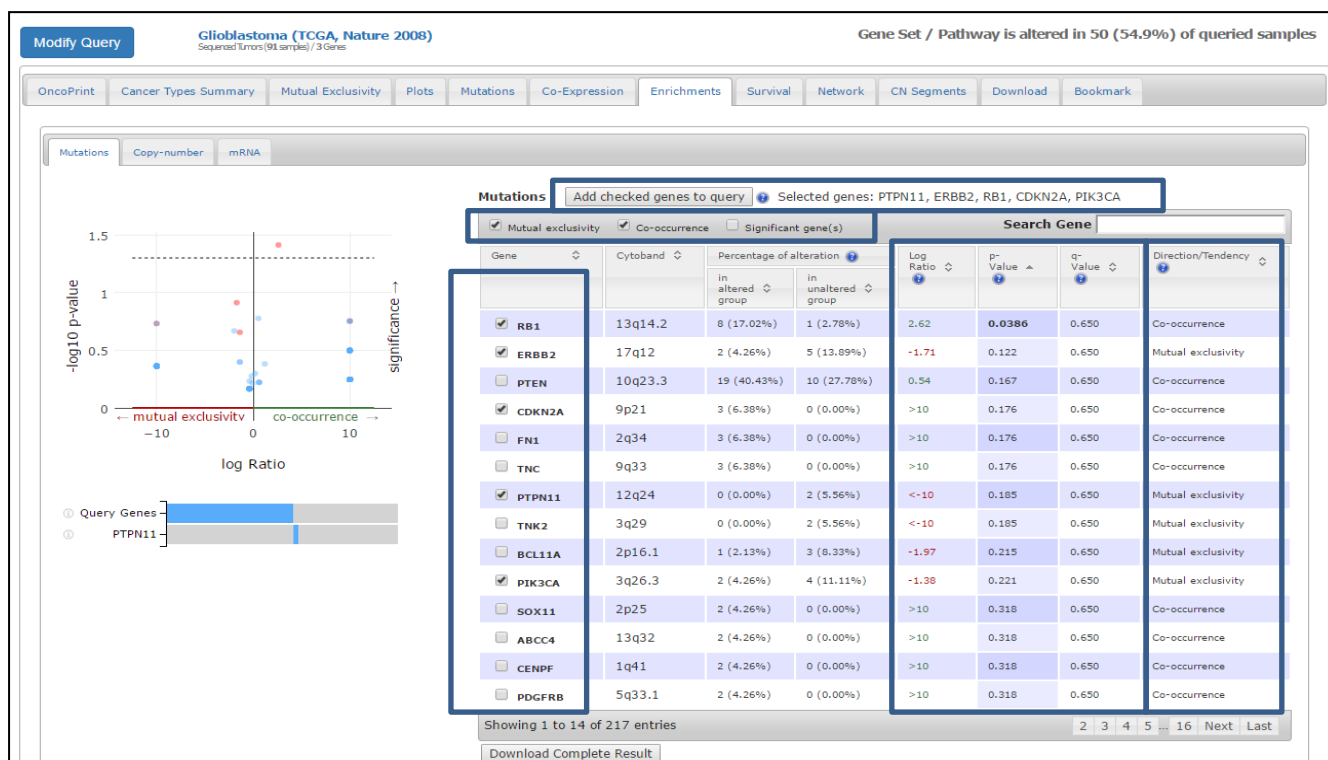
# Co-Expression

- Separate tab for each gene in query.
- Can see co-expression data for many genes.
  - Use 'Search Gene' function or sort columns to find desired target.
  - Can download full results per gene



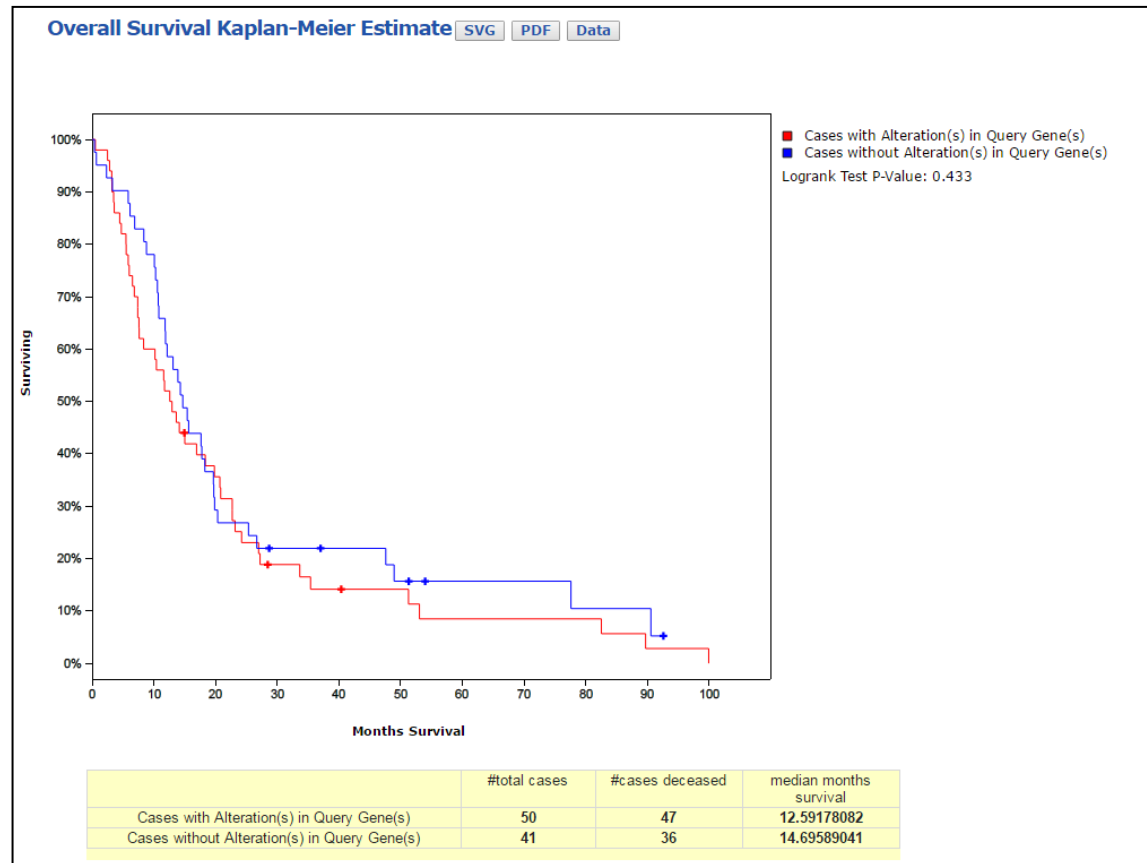
# Enrichments

- Separate tabs for identifying mutations, copy number, and changes in mRNA/microRNA expression that either co-occur or are mutually exclusive to your query.



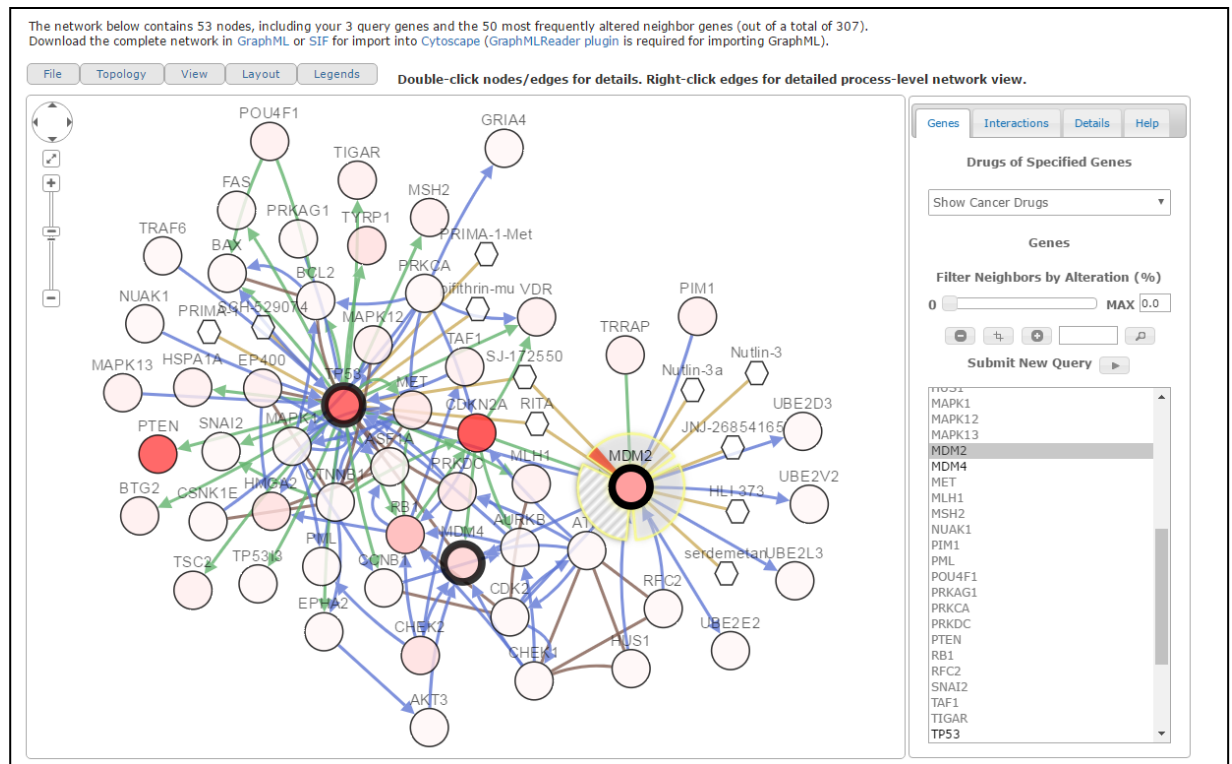
# Survival

- If the query selected contains studies with survival information, Kaplan-Meier Estimates for disease free survival and overall survival are displayed.



# Network

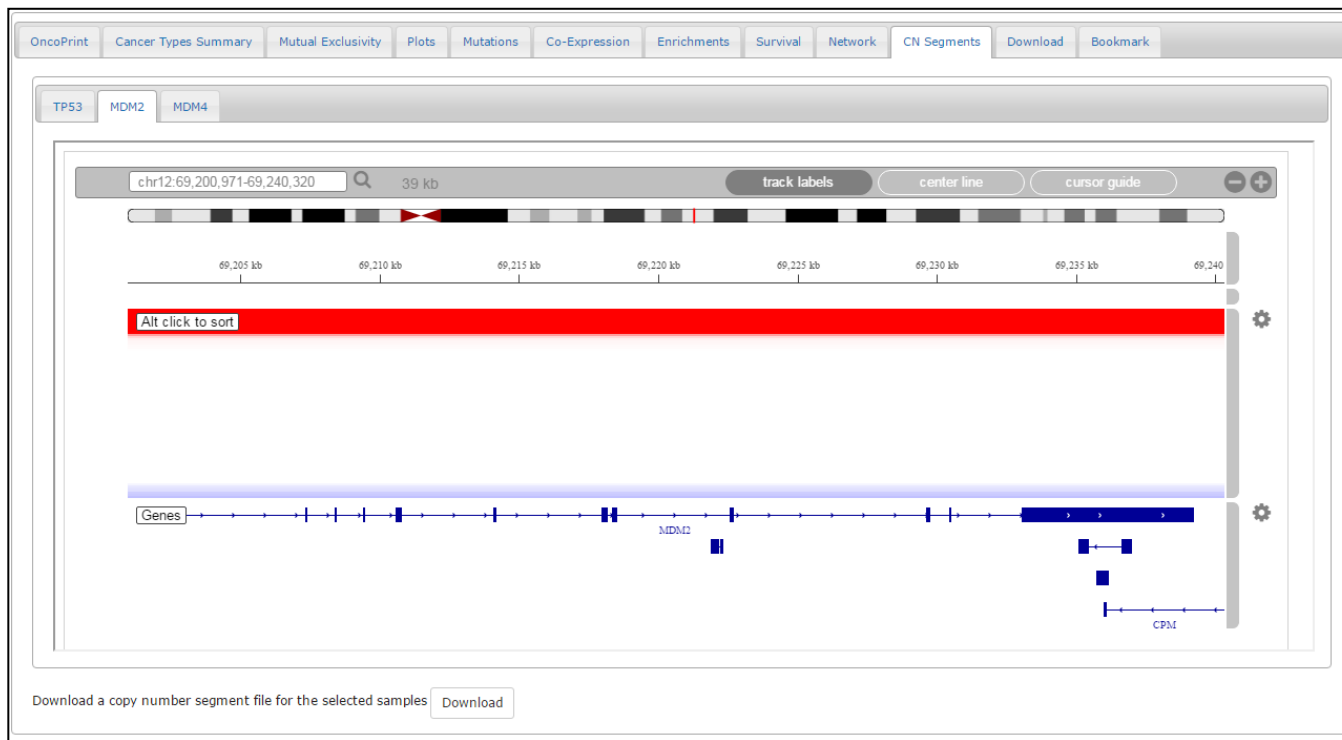
- Will display your queried genes and their 50 most frequently altered neighbors.
- Can alter image by filtering neighbors by alteration % on Gene subtab on right panel and buttons along top of network window





# CN Segments

- Will show Copy number data on all genes in your query within your selected studies.
  - Each gene is on a separate tab.
    - Red = CN Gain
    - Blue = CN Loss



# OncoPrinter Visualization Tool

- This tool can be used to visualize your own data.
- <http://www.cbioportal.org/oncoprinter.jsp>
- Click on link above and view link for “Data Format” (or see next page) and Format a tab-delimited file to the specifications listed.
- Copy and paste data into “Input Mutation Data” window or “Choose File” to select tab-delimited file from saved location.
- If desired, order the genes and/or samples as you would like them to be shown on the graph.

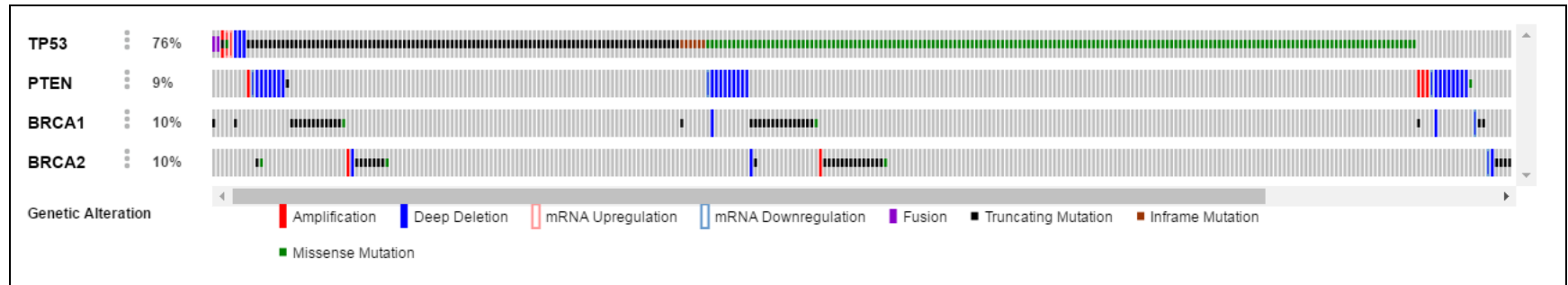
# OncoPrinter: Data Format

## Data format

The data should contain three tab-delimited columns. The first row is a header row, which contains: **Sample Gene Alteration Type**. Each following row contains a single genomic event in a single sample. You can also list samples without any events at the end of the list so that the percentages can be properly calculated. Note: Any row which has an entry in the Gene (2nd) column must also have an entry in the Alteration (3rd) and Type (4th) columns

1. Sample: Sample ID
2. Gene: Gene symbol (or other gene identifier)
3. Alteration: Definition of the alteration event
  - Mutation event: amino acid change or any other information about the mutation
  - Fusion event: fusion information
  - Copy number alteration (CNA) - please use one of the four events below:
    - AMP: high level amplification
    - GAIN: low level gain
    - HETLOSS: shallow deletion
    - HOMDEL: deep deletion
  - mRNA expression - please use one of the two events below:
    - UP: expression up
    - DOWN: expression down
4. Type: Definition of the alteration type. It has to be one of the following.
  - For a mutation event, please use one of the three mutation types below:
    - MISSENSE: a missense mutation
    - INFRAME: a inframe mutation
    - TRUNC: a truncation mutation
  - FUSION: a fusion event
  - CNA: a copy number alteration event
  - EXP: a expression event

# OncoPrinter Results



- Percent mutated samples per gene displayed adjacent to table
- Genetics alterations are color-coded
- Options for visualization appear in a menu near upper right-hand side of graph when you hover over graph
  - Color-code mutations and sort by type OR show all mutations with same color
  - Hide unaltered cases
  - Remove white spaces between columns
  - Zoom in/out – by buttons or slider bar
  - Zoom to fit altered cases in screen
  - Downloads: PDF, PNG, SVG, or .txt file with order of patients in graph

# MutationMapper

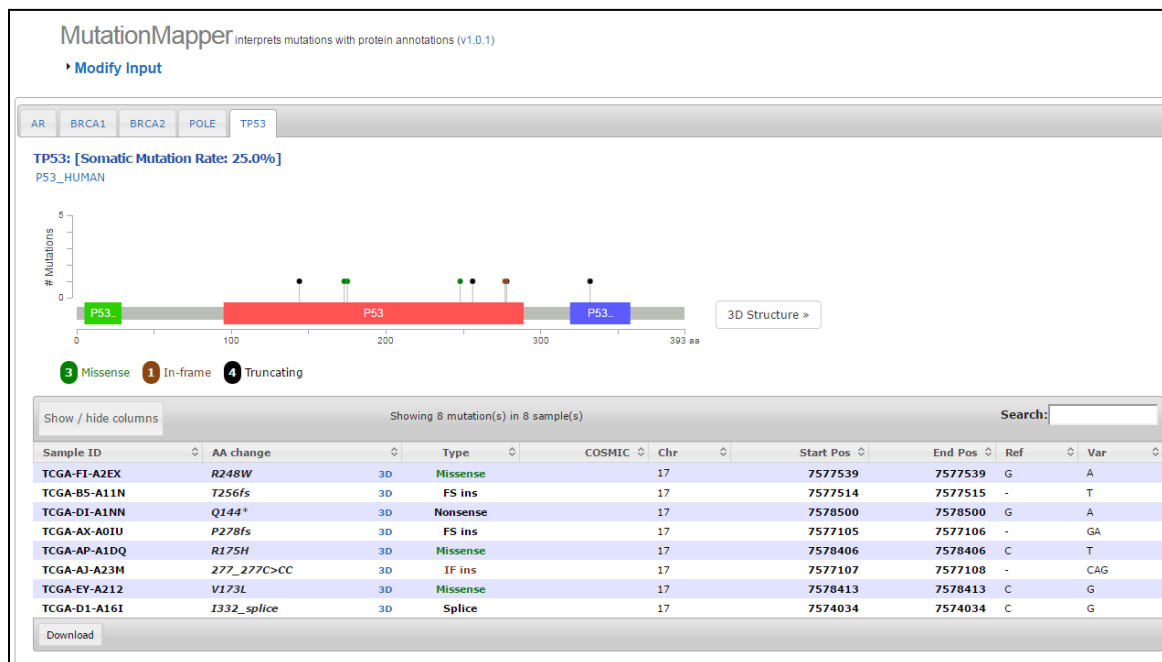
- Interprets mutations with protein annotations
- Input tab-delimited file needs at least two columns
  - HUGO gene name
  - Protein change

List of valid input headers:

Column Header	Description	Example
Hugo_Symbol	HUGO symbol for the gene	TP53
Protein_Change	Amino acid change	V600E
Sample_ID	Tumor sample ID	TCGA-B5-A11E
Mutation_Type	Translational effect of variant allele	Missense_Mutation, Nonsense_Mutation, etc.
Chromosome	Chromosome number	X, Y, M, 1, 2, etc.
Start_Position	Lowest numeric position of the reported variant on the genomic reference sequence	666
End_Position	Highest numeric position of the reported variant on the genomic reference sequence	667
Reference_Allele	The plus strand reference allele at this position	A
Variant_Allele	Tumor sequencing (discovery) allele	C
Validation_Status	Second pass results from orthogonal technology	Valid
Mutation_Status	Mutation status	Somatic, Germline, etc.
Center	Center/Institute reporting the variant	mskcc.org

# MutationMapper: Resulting Data

- Similar to Mutation window in Query results
  - Does not have annotations like query results
- Can modify input data
- Table can be downloaded as TSV



# Scenario #1

- You would like to validate one of your lab's disease specific gene lists against the data in cBioPortal.
  - Run Query by selecting relevant studies and enter all genes in Gene Set window.



# Design Query

Modify Query

Select Cancer Study:

Search... 4 studies selected. [Deselect all](#)

☒ Cholangiocarcinoma (4)

- ☒ Intrahepatic Cholangiocarcinoma (Johns Hopkins University, Nat Genet 2013) 40 samples
- ☒ Cholangiocarcinoma (National Cancer Centre of Singapore, Nat Genet 2013) 15 samples
- ☒ Cholangiocarcinoma (National University of Singapore, Nat Genet 2012) 8 samples
- ☒ Cholangiocarcinoma (TCGA, Provisional) 36 samples

☐ Gallbladder Cancer (1)

- ☐ Gallbladder Carcinoma (Shanghai, Nat Genet 2014) 32 samples

☐ Bladder/Urinary Tract (7)

- ☐ Bladder Urothelial Carcinoma (7)

Select Data Type Priority:
☒ Mutation and CNA
☐ Only Mutation
☐ Only CNA

Enter Gene Set:
Advanced: Onco Query Language (OQL)

User-defined List

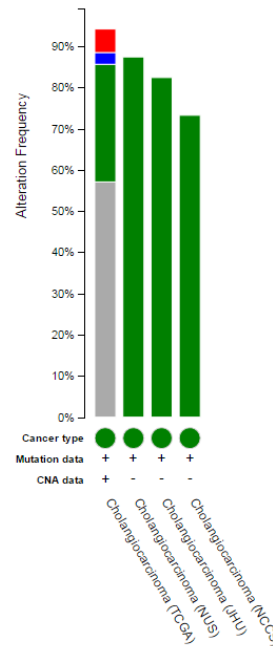
AKT1  
ALB  
APC  
ARAF  
ARID1A

☒ All gene symbols are valid.

Submit

# View Overview

- Is your gene list sensitive enough to have a high number and percentage of alterations?
  - Qualitatively, yes.



# View Mutations

- How many genes have observed mutations out of your gene list?
  - 41/62 genes have hits and are validated through cBioPortal. Look through literature or other knowledgebase resources to validate (or not validate) the other 21.

# Scenario #2

- You are studying KRAS mutation frequency and co-mutations in glioblastoma.
  - What genes are commonly co-mutated in KRAS mutant GBM?

# Query KRAS mutations in GBM

Modify Query

Select Cancer Study:
 

Search...
 3 studies selected. [Deselect all](#)

- ☒ Glioblastoma (3)
  - ☒ Glioblastoma Multiforme (3)
    - ☒ Glioblastoma Multiforme (TCGA, Provisional) 604 samples
 ☒ Glioblastoma (TCGA, Nature 2008) 206 samples
 ☒ Glioblastoma (TCGA, Cell 2013) 580 samples
- ☐ Sellar Tumor (1)
  - ☐ Granular Cell Tumor (1)
    - ☐ Genomic Profile of Patients with Advanced Germ Cell Tumors (MSK, JCO 2016). 180 samples
 ☐ Miscellaneous Neuroepithelial Tumor (1)

Select Data Type Priority:
 ☐ Mutation and CNA
 ☒ Only Mutation
 ☐ Only CNA

Enter Gene Set:
 Advanced: Onco Query Language (OQL)
 

User-defined List

KRAS

☒ All gene symbols are valid.

Submit

# Go to Mutations Page and Click on Sample ID in list

- Repeat for all patients
- If necessary, download CSV files for CNAs and/or mutations
- Identify recurring mutations in cohort besides KRAS
  - 4/5 GBM patients with KRAS mutation also have PTEN mutation
  - 4/5 GBM patients with KRAS mutation also have MDM4 amplification, CDKN2A and CDKN2B deep deletions

# Scenario #2B

- Same scenario – instead of GBM, look for co-mutations in uterine endometrial carcinoma via query.
  - OncoPrint as Query Result
  - Enrichments using Modified Query

# OncoPrint as Query Resu



Modify Query

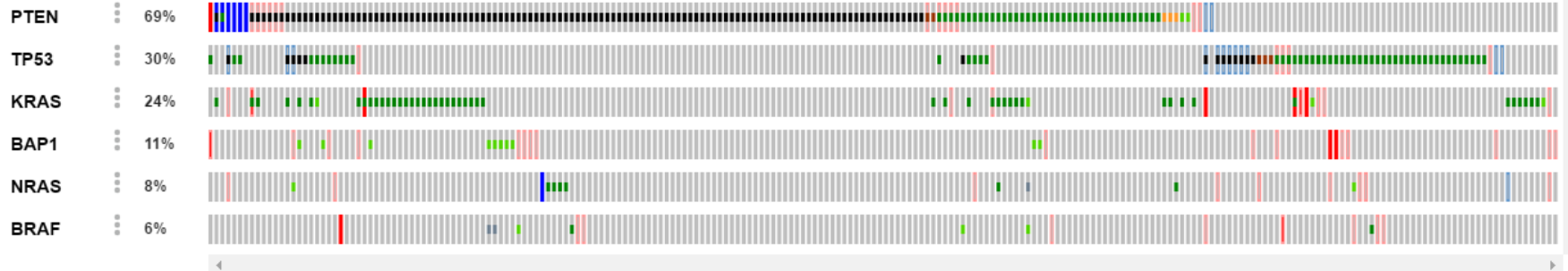
Uterine Corpus Endometrial Carcinoma (TCGA, Nature 2013)  
Sequenced Tumors (248 samples) / 6 Genes

Gene Set / Pathway is altered in 228 (91.9%) of queried samples

OncoPrint Cancer Types Summary Mutual Exclusivity Plots Mutations Co-Expression Enrichments Survival Network CN Segments Download Bookmark

Case Set: Sequenced Tumors (248 patients / 248 samples)

Altered in 228 (92%) of 248 sequenced cases/patients (248 total)



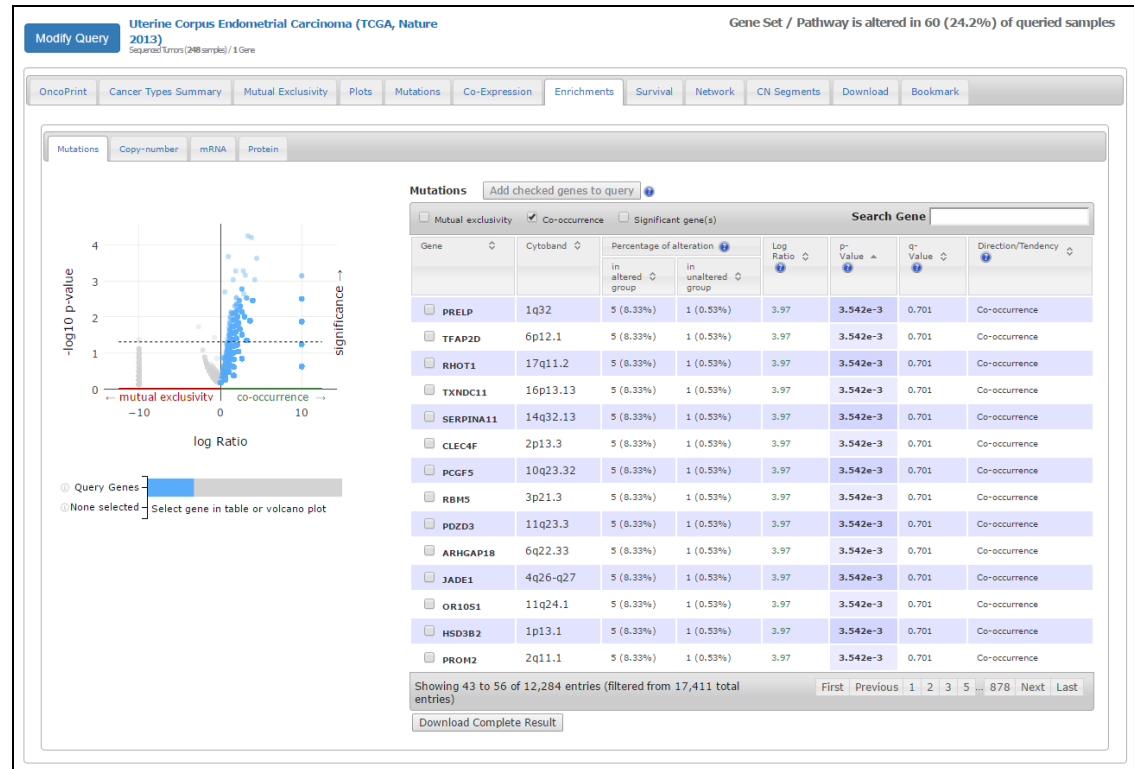
Genetic Alteration

Amplification Deep Deletion mRNA Upregulation mRNA Downregulation Truncating Mutation (putative driver)  
Truncating Mutation (putative passenger) Inframe Mutation (putative driver) Inframe Mutation (putative passenger)  
Missense Mutation (putative driver) Missense Mutation (putative passenger)



# Enrichments

- Modify query to only include gene of interest (KRAS)
- Click on Enrichments tab in Query Results
- Make sure you are on Mutations sub-tab within Enrichments tab.
- Download complete result by pressing button at bottom of window.
  - Determine p-value to use for data acceptance.
  - Can also toggle through data points on log Ratio graphs on left side of window.



# Scenario #3

- You would like to contribute to cBioPortal efforts.
  - cBioPortal accepts coding contributions
    - <https://github.com/cBioPortal/cbioportal/blob/master/CONTRIBUTING.md>
    - See link above to learn how to contribute through GitHub.

# Contacts

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