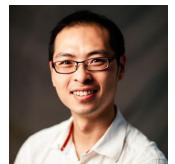
# Open Data Resources for Human Genomics Research



A/Prof Jason Wong Head, Computational Cancer Genomics School of Biomedical Sciences University of Hong Kong



Dr Rebecca Poulos NHMRC Early Career Fellow Children's Medical Research Institute University of Sydney











## Part One Human genomics data resources

1:30pm – 3pm

- UCSC Genome Browser + Exercise
- gnomAd + Exercise
- GTEx + Exercise
- ENCODE database + Exercise
- Exercise: Putting it all together

### Part Two Cancer genomics data resources

3:30pm – 5pm

- Brief introduction to TCGA
- cBioPortal
- Genomic Data Commons
- Xena Browser
- Cancer Genomics Cloud











#### Part Two **Cancer genomics data resources**

3:30pm – 5pm

The Cancer Genome Atlas

Brief introduction to The Cancer Genome Atlas

cbioPortal •

Looking up somatic mutations and any clinical associations

Genomic Data Commons ٠

Accessing the raw and processed data

**XENA** browser •

Downloading tidy processed data

**Cancer Genomics Cloud** •

Accessing and analyzing data in the cloud

Some sections will be accompanied by an exercise to help you test your skills, plus an exercise at the end to put all your knowledge together.

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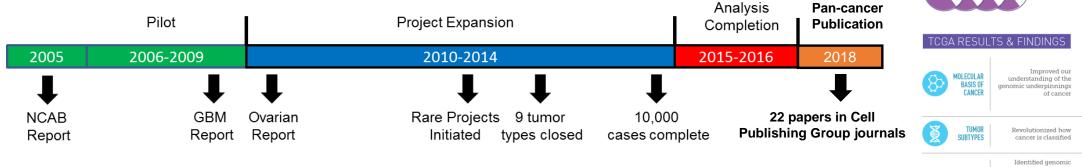






# The Cancer Genome Atlas

• Lunched in 2006 as a pilot and expanded in 2009



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• 11,000 cancer patients across 33 cancer types

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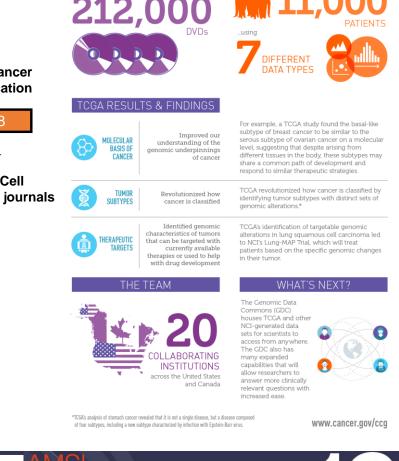
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NATIONAL CANCER INSTITUTE THE CANCER GENOME ATLAS

TCGA BY THE NUMBERS

To put this into perspective, 1 petabyte of data

TCGA data describes

collected from

...based on paired tumor and normal tissue sets

...including

TCGA produced over

of data

is equal to



# Types of TCGA data

- Core dataset:
  - Pathology report
  - Histology images
  - Clinical data
  - Whole exome-seq
  - SNP 6.0 array
  - mRNAseq
  - miRNAseq
  - Methylation array

- Optional datasets:
  - 50x Whole-genome sequencing
  - Bisulfide sequencing
  - Protein Array

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- Radiological images (MRI)

\* Also mRNA/methylation data for ~10-20 matched normal tissue data







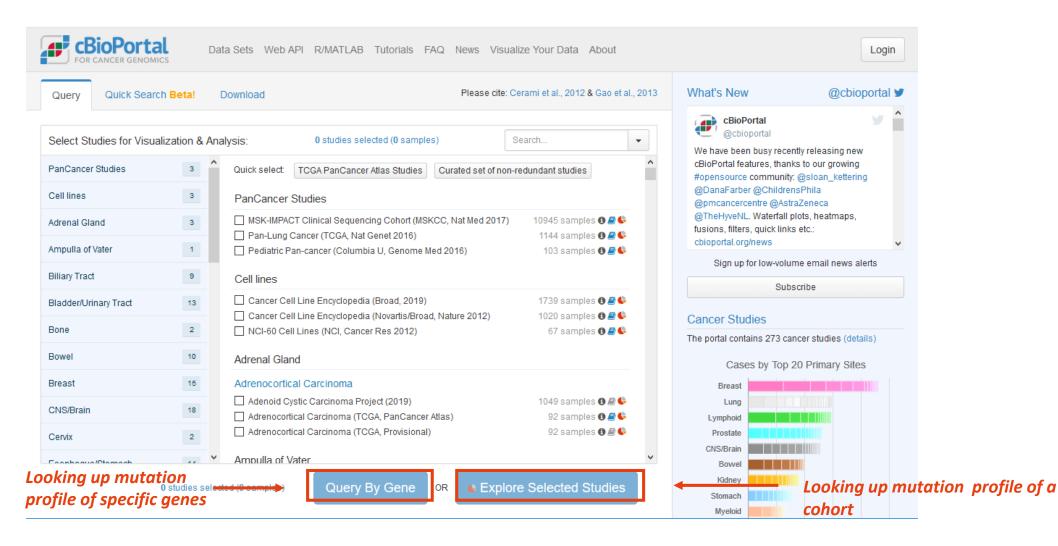
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*Looking up somatic mutations in cancer samples* 













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		ightarrow SMALL CELL LUNG CANCER	
east	15	Small Cell Lung Cancer (CLCGP, Nat Genet 2012)	29 samples 🚯 🗐 😓
		Small Cell Lung Cancer (Johns Hopkins, Nat Genet 2012)	51 samples 🚯 🗐 😓
S/Brain	18	Small Cell Lung Cancer (U Cologne, Nature 2015)	110 samples 🚯 🗐 😓
vix	2	Small-Cell Lung Cancer (Multi-Institute, Cancer Cell 2017)	20 samples 🕄 🖨 📞
		Non-Small Cell Lung Cancer	
ophagus/Stomach	14	Non-Small Cell Lung Cancer (MSK, Cancer Cell 2018)	75 samples 🔀 🛃 😓
e	3	Non-Small Cell Lung Cancer (MSKCC, J Clin Oncol 2018)	240 samples 🔀 🛃 😓
	3	Non-Small Cell Lung Cancer (TRACERx, NEJM 2017)	327 samples 🔂 🗾 😓
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		Non-small cell lung cancer (MSK, Science 2015)	16 samples 🕄 릗 😓
ney	17	Pan-Lung Cancer (TCGA, Nat Genet 2016)	1144 samples 🔀 🛃 📞
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		Lung Adenocarcinoma (Broad, Cell 2012)	183 samples 🔂 릗 📞
ıg	21	Lung Adenocarcinoma (MSKCC, Science 2015)	35 samples 🔂 릗 🌭
		Lung Adenocarcinoma (TCGA, Nature 2014)	230 samples 🔂 🗾 📞
nphoid	19	Lung Adenocarcinoma (TCGA, PanCancer Atlas)	566 samples 🕄 🛃 😓
		Lung Adenocarcinoma (TCGA, Provisional)	586 samples 🕄 릗 📞
reloid	9	Lung Adenocarcinoma (TSP, Nature 2008)	163 samples 🕄 🗐 崎
ner	13	Non-Small Cell Cancer (MSKCC, Cancer Discov 2017)	915 samples 🕄 🗐 🈓
		ightarrow LUNG SQUAMOUS CELL CARCINOMA	
ary/Fallopian Tube	4	Lung Squamous Cell Carcinoma (TCGA, Nature 2012)	178 samples 🚯 🚄 🔩
		Lung Squamous Cell Carcinoma (TCGA, PanCancer Atlas)	487 samples 🚯 🚄 🔩
ncreas	10	Lung Squamous Cell Carcinoma (TCGA, Provisional)	511 samples 🛈 <i> </i> 🖶
ipheral Nervous System	5		

1 study selected (230 samples) Deselect all

Query By Gene

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Explore Selected Studies OR









#### **cBioPortal** Please cite: Cerami et al., 2012 & Gao et al., 2013 Quick Search Beta! Query Download Selected Studies: Modify Lung Adenocarcinoma (TCGA, Nature 2014) (230 total samples) Select Genomic Profiles: Mutations ② Putative copy-number alterations from GISTIC 0 mRNA Expression z-Scores (RNA Seq V2 RSEM) 3 Select Patient/Case Set: Samples with mutation data (230) **X T** To build your own case set, try out our enhanced Study View. Enter Genes: User-defined List х 🔻 **Hint:** Learn Onco Query Language (OQL) <u>TP53</u> to write more powerful queries 🗹 Type in "TP53" and "KRAS" one per line KRAS ✓ All gene symbols are valid. Submit Query



















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Query Quick Search Beta! Download Please cite: Cerami et al., 2012 & Gao et al., 2013

elect Studies for Visu		Analysis: 1 study selected (230 samples) Deselect all	Search
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ung	21	Non-small cell lung cancer (MSK, Science 2015)	16 samples 🛈 🗾 🤤
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vary/Fallopian Tube	4	Lung Adenocarcinoma (TCGA, PanCancer Atlas)	566 samples 🕄 🛃 🤤
wary/ralloplan rube	4	Lung Adenocarcinoma (TCGA, Provisional)	586 samples 🛈 <i> </i> 🖶
Pancreas	10	Lung Adenocarcinoma (TSP, Nature 2008)	163 samples 🚯 <i>叁</i> 📞

1 study selected (230 samples) Deselect all

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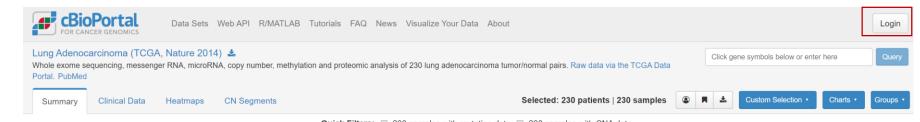
Explore Selected Studies

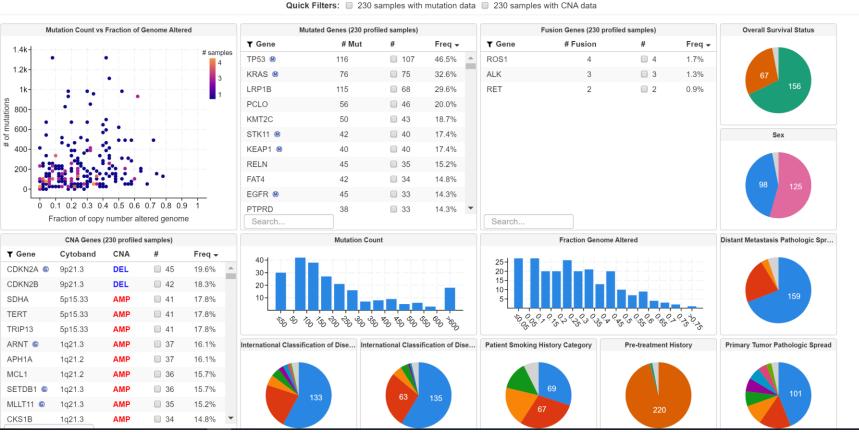






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#### Lung Adenocarcinoma (TCGA, Nature 2014) 🕹

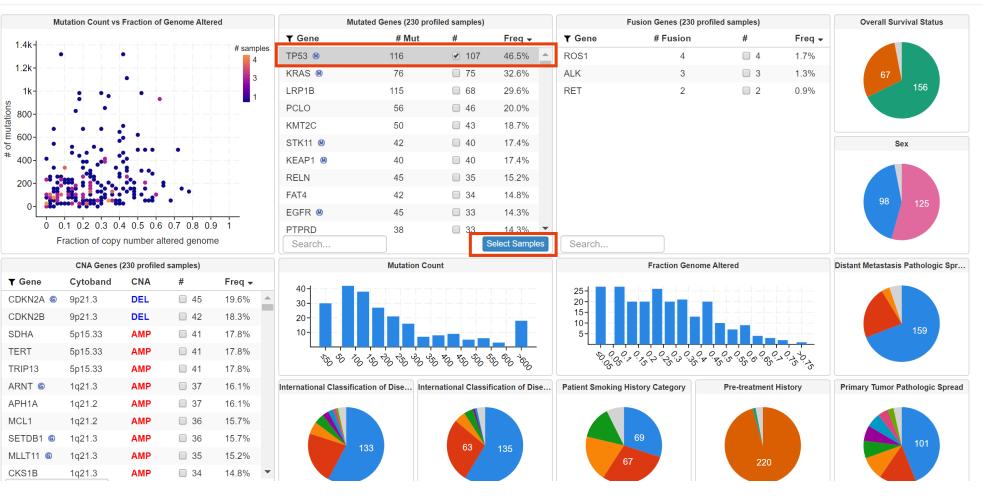
Whole exome sequencing, messenger RNA, microRNA, copy number, methylation and proteomic analysis of 230 lung adenocarcinoma tumor/normal pairs. Raw data via the TCGA Data Portal. PubMed

\*

٩  Custom Selection • Charts •

Clinical Data Heatmaps **CN** Segments Summary

Selected: 230 patients | 230 samples



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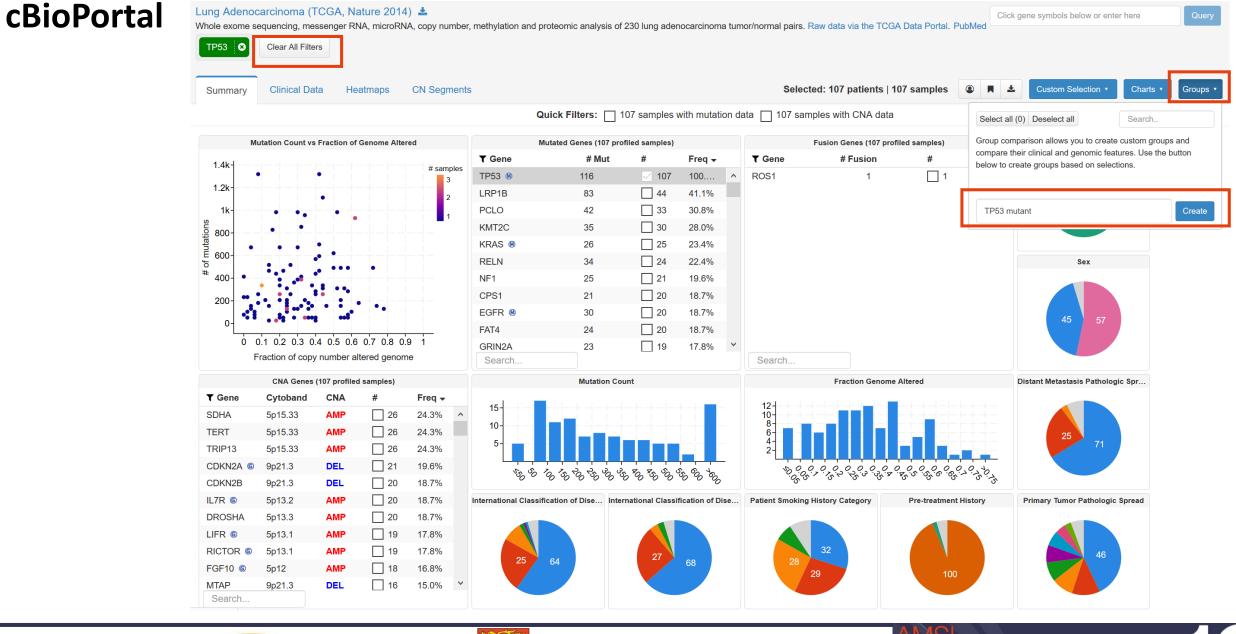
Quick Filters: 230 samples with mutation data 230 samples with CNA data

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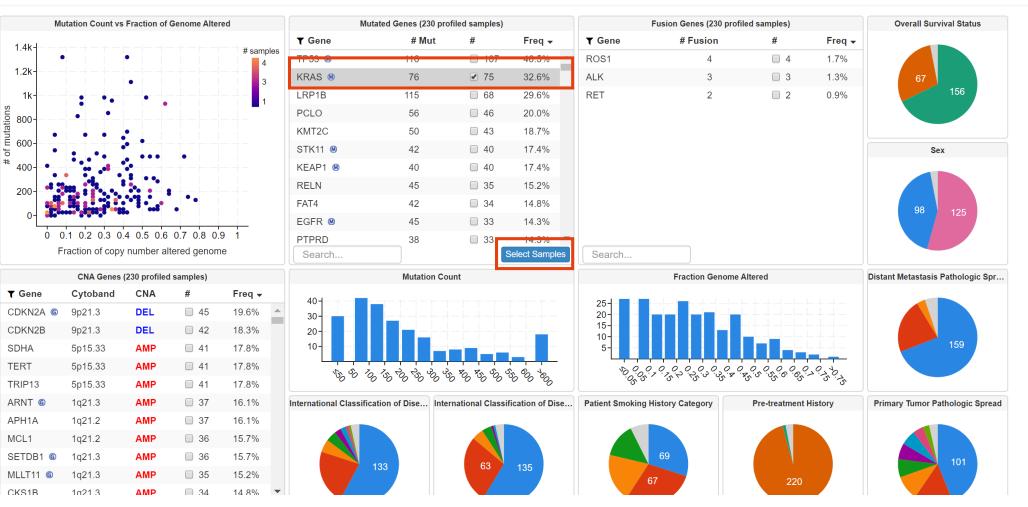
#### Lung Adenocarcinoma (TCGA, Nature 2014) 📥

Whole exome sequencing, messenger RNA, microRNA, copy number, methylation and proteomic analysis of 230 lung adenocarcinoma tumor/normal pairs. Raw data via the TCGA Data Portal. PubMed

**Clinical Data** Heatmaps **CN** Segments Summarv

Selected: 230 patients | 230 samples ٩ **.** 

Custom Selection Charts • Groups •



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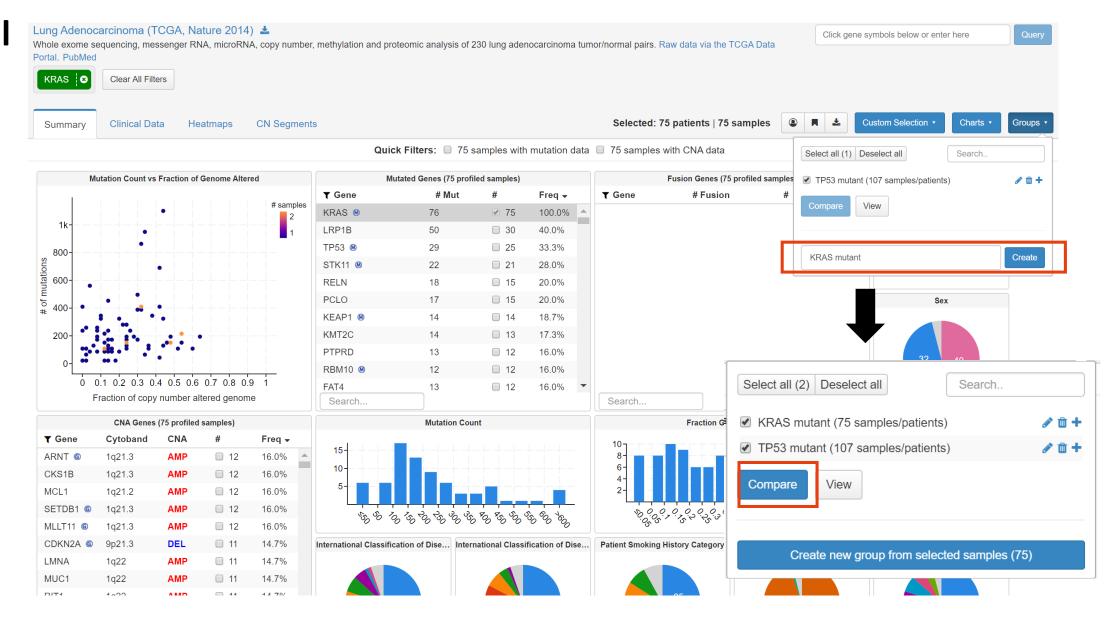
Quick Filters: 230 samples with mutation data 230 samples with CNA data











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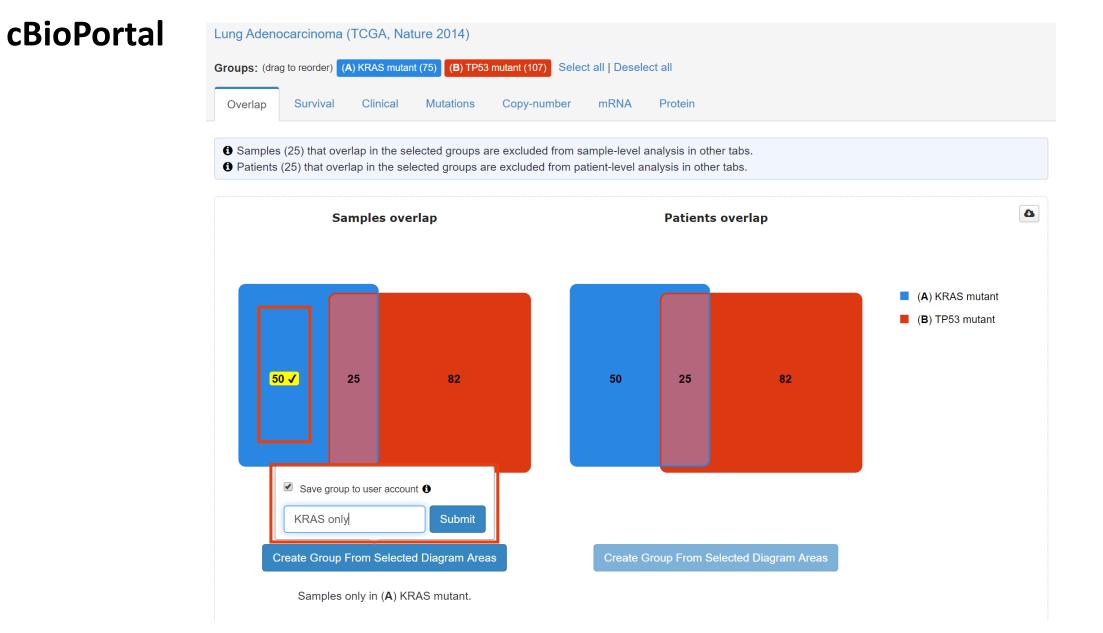
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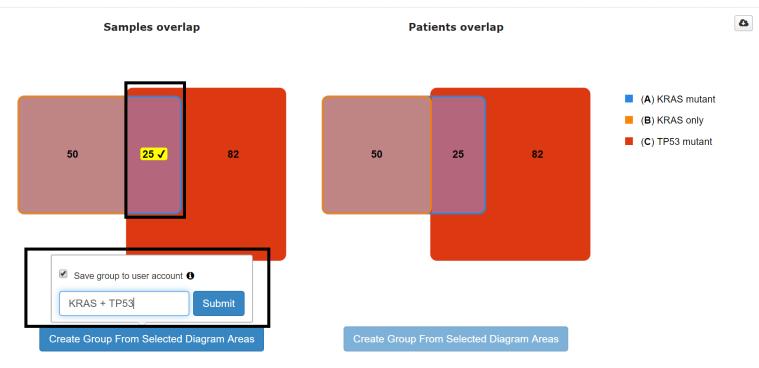
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(B) KRAS only is completely overlapping with other selected groups, so is excluded from analysis in other tabs.
(B) KRAS only is completely overlap between (A) KRAS mutant and (C) TP53 mutant are excluded from sample-level analysis in other tabs.
(C) Patients (25) that overlap between (A) KRAS mutant and (C) TP53 mutant are excluded from patient-level analysis in other tabs.



Samples only in (A) KRAS mutant and (C) TP53 mutant.

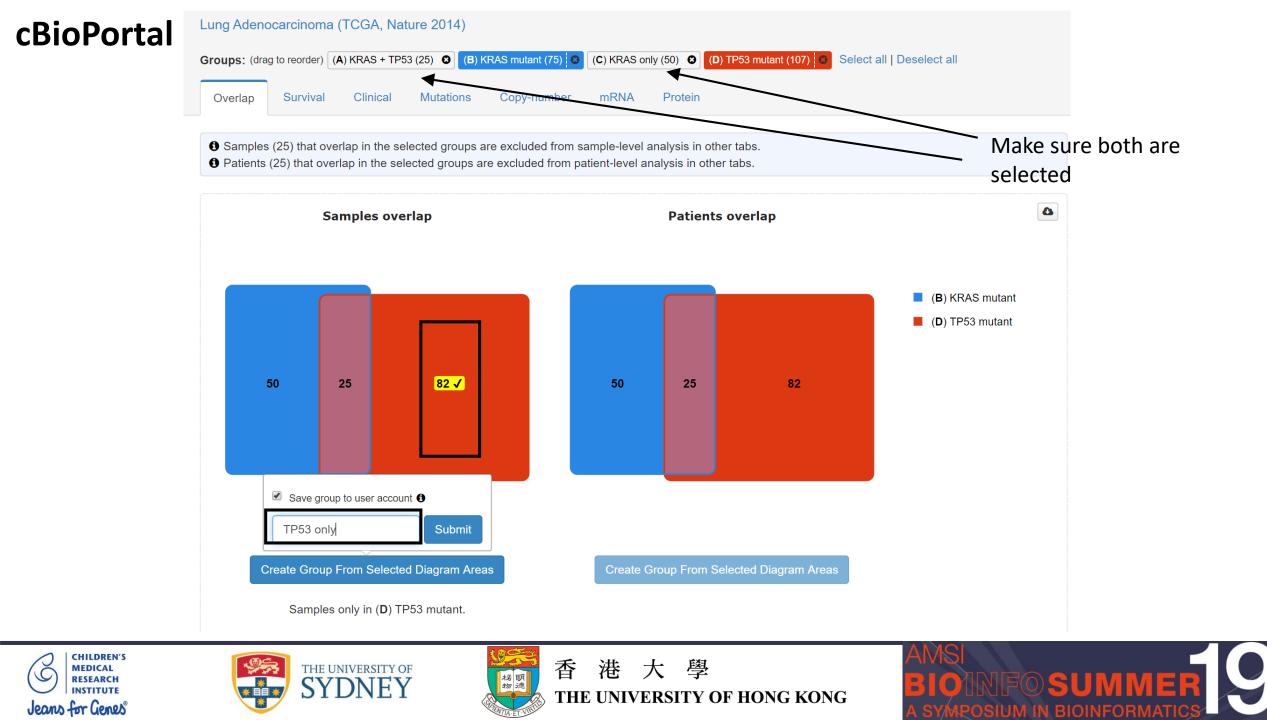






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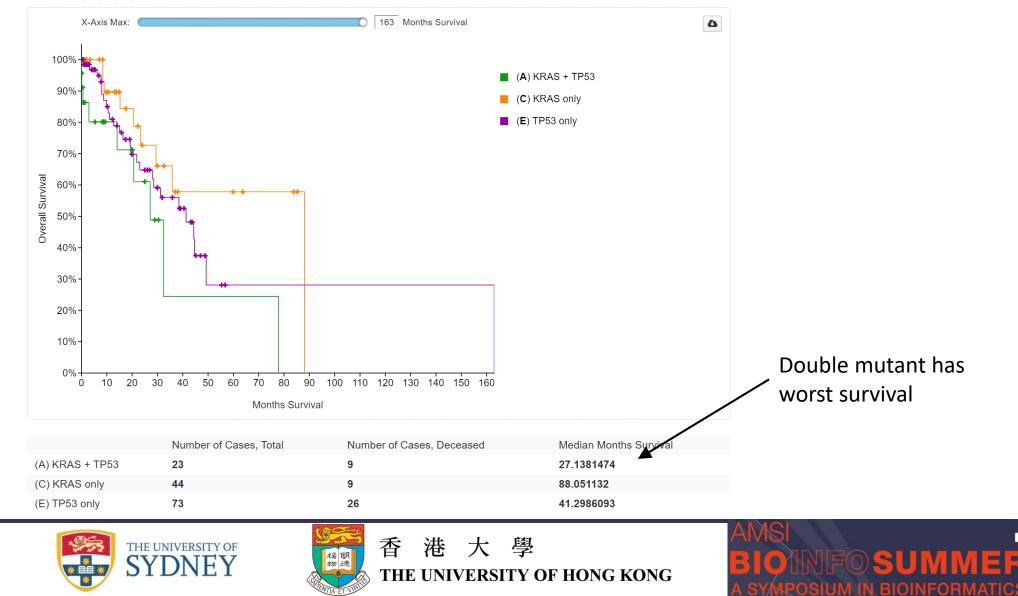
#### Overall Survival Kaplan-Meier Estimate (Overall patient survival status.)

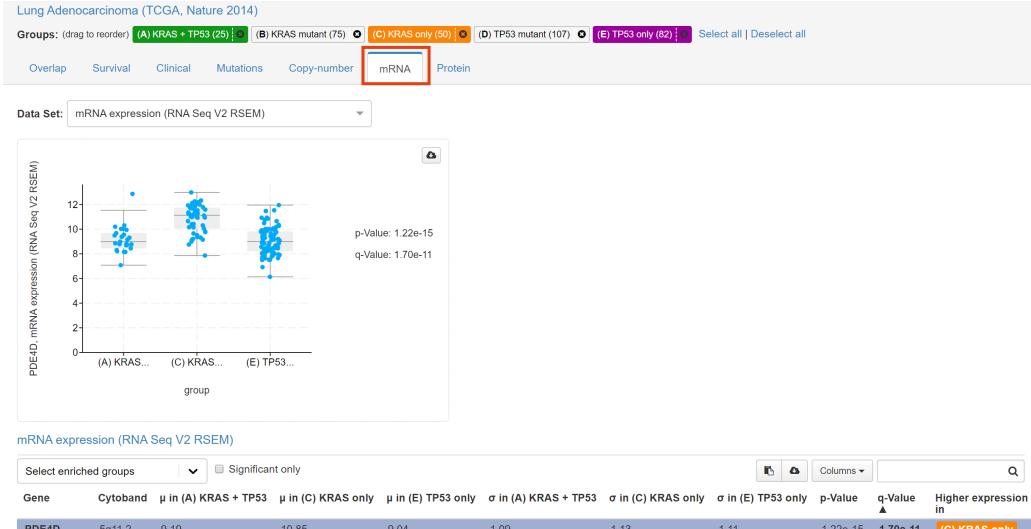
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PDE4D	5q11.2- q12.1	9.19	10.85	9.04	1.09	1.13	1.11	1.22e-15	1.70e-11	(C) KRAS only
APOBEC3B	22q13.1	7.66	5.90	8.11	1.11	1.21	1.51	3.89e-15	2.71e-11	(E) TP53 only
GPSM2	1p13.3	9.09	8.23	9.27	0.68	0.63	0.72	1.01e-13	4.69e-10	(E) TP53 only

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Exercise...

What is the most common hotspot point mutation in ERCC2 in bladder cancer?

N238S

Challenge...

What is the most significantly higher expressed gene in ERCC2 mutant versus WT bladder cancer?

SGO1





Accessing raw and processed data









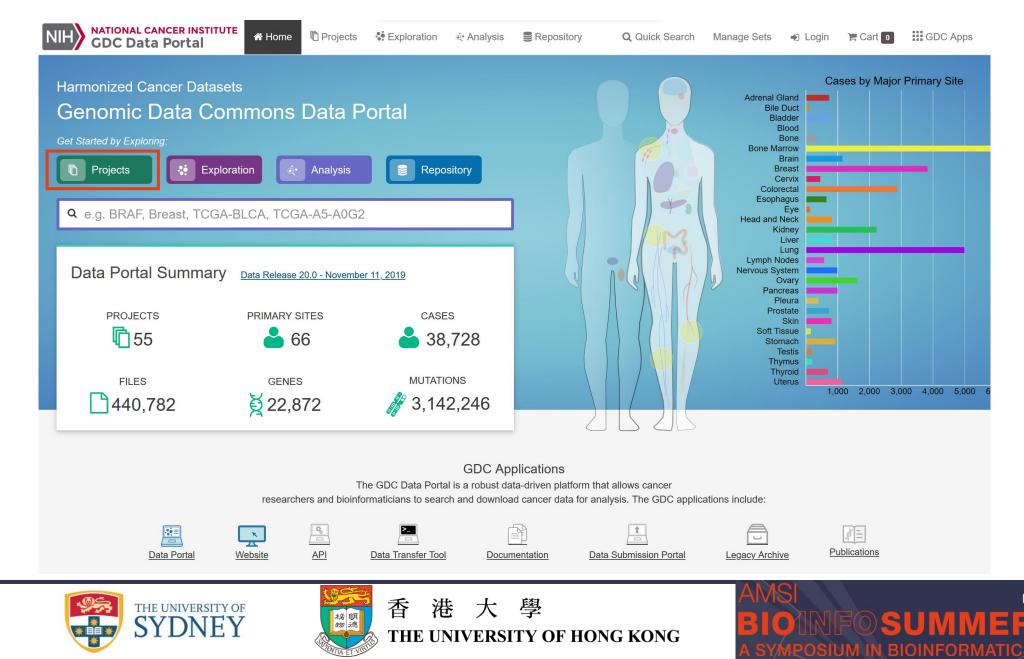


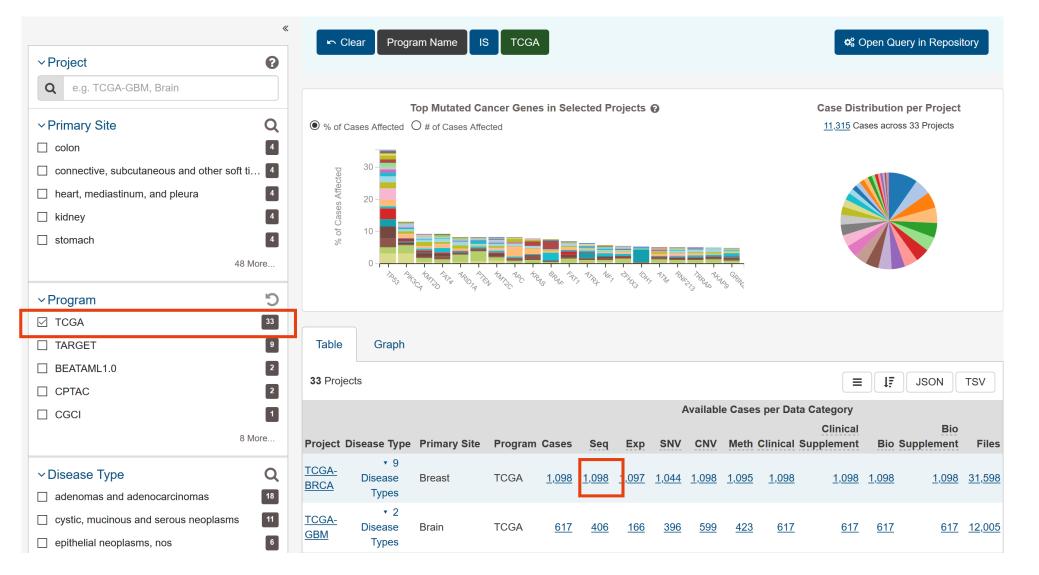
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WXS	2,175		TCGA-A8-A08F	TCGA-BRCA	Breast	Female	<u>54</u>	<u>3</u>	_	<u>16</u>	<u>5</u>	1	<u>11</u>		_	<u></u> (3)
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			TCGA-A2-A0YT	TCGA-BRCA	Breast	Female	<u>57</u>	<u>5</u>		<u>16</u>	<u>5</u>	1	<u>10</u>			<b>b</b> (3)
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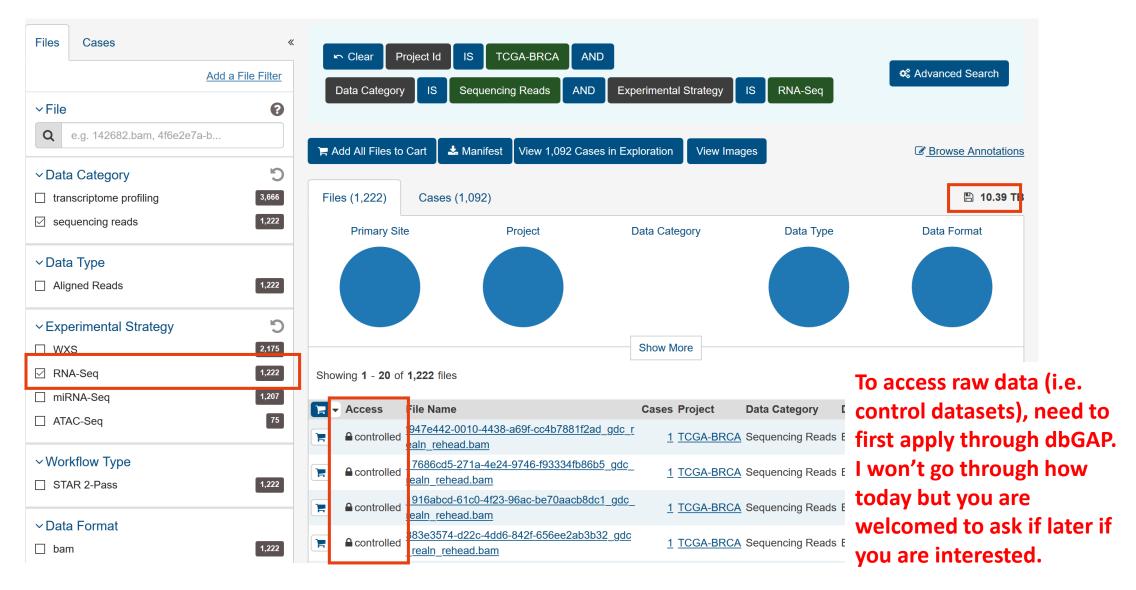
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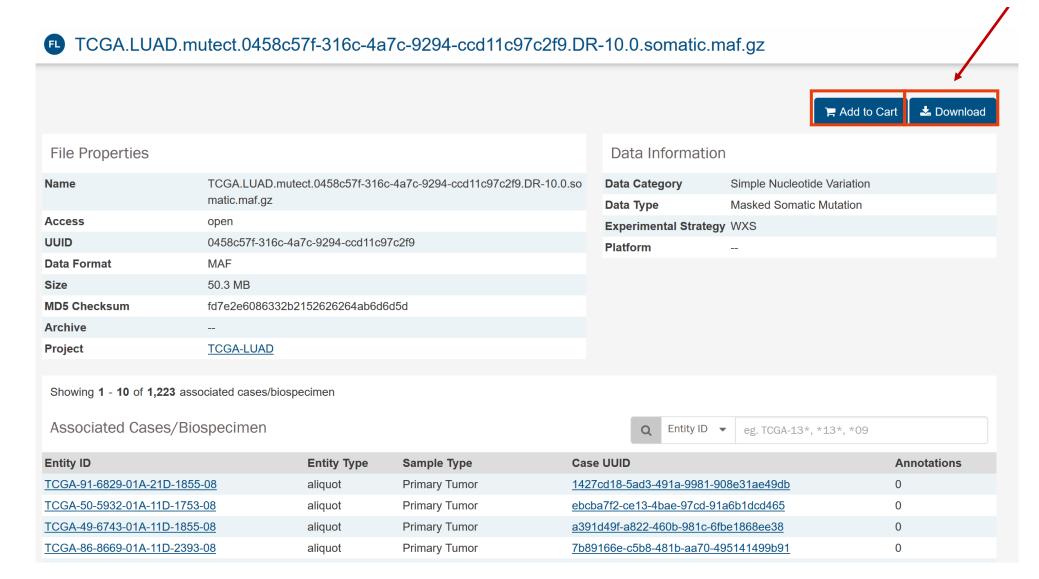






**Only works for small files** 

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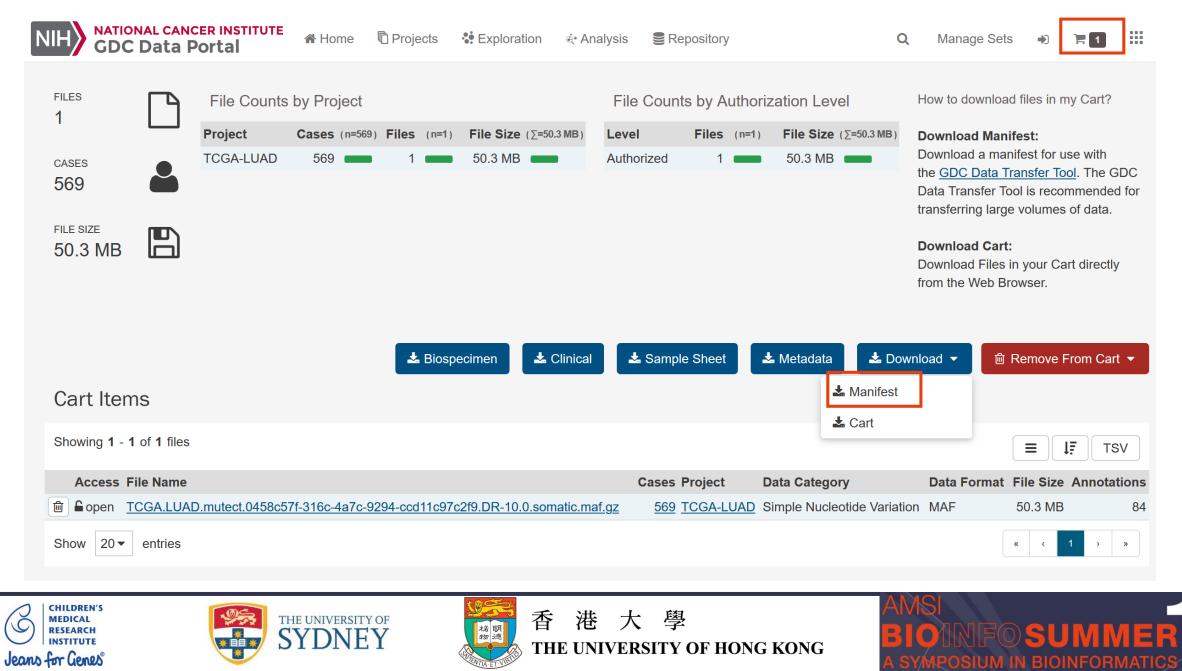
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#### https://gdc.cancer.gov/access-data/gdc-data-transfer-tool

#### GDC Data Transfer Tool



The GDC provides a standard client-based mechanism in support of highperformance data downloads and submission.

The raw sequence files, typically stored as BAM or FASTQ, make up the bulk of data. The size for a single file can vary greatly depending on the specific analysis; However, some of the whole genome BAM files in The Cancer Genome Atlas (TCGA) reach sizes of 200-300 GB. In such cases, a high-performance data download and submission tool is essential.

Below are basic instructions and links for downloading the GDC Data Transfer Tool client-based interface for data downloads and submission and user interface (beta version) for data downloads. For additional instructions, please visit the GDC Data Transfer Tool User's Guide.











#### Genomic Data Commons – download WXS BAM files for all ERCC2 N238S mutants

NIH GDC Data Portal	🖀 Home 🛛 🖺	] Projects 😽 Expl	oration 🔌	Analysis	Reposit	ory				(	<b>Q</b> Mai	nage Sets	•		
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modifier	116,194	Primary Site		Project		Dise	ase T	Гуре			Gender		Vita	al Status	
moderate	76,158														
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☐ tolerated_low_confidence	4,976	<u>TCGA-A5-A0G2</u>	TCGA-UCEC	Corpus uteri	Female	<u>58</u>	<u>4</u>	<u>5</u>	<u>16</u>	<u>5</u>	<u>1 10</u>	<u>17</u>	<u>1,856</u>	<u>481</u>	<b>b</b> (3)
	[	TCGA-EO-A22U	TCGA-UCEC	Corpus uteri	Female	<u>57</u>	<u>4</u>	<u>5</u>	<u>16</u>	<u>5</u>	<u>1 10</u>	<u>16</u>	<u>1,234</u>	<u>445</u>	<b>b</b> (2)
∽ Polyphen Impact	[	TCGA-FI-A2D5	TCGA-UCEC	Corpus uteri	Female	<u>58</u>	<u>4</u>	<u>5</u>	<u>16</u>	<u>5</u>	<u>1 11</u>	<u>16</u>	<u>1,103</u>	<u>437</u>	<b>b</b> (2)
probably_damaging	35,559	TCGA-B5-A3FC	TCGA-UCEC	Corpus uteri	Female	<u>57</u>	<u>4</u>	<u>5</u>	<u>16</u>	<u>5</u>	<u>1 10</u>	<u>16</u>	<u>1,180</u>	<u>436</u>	<b>上</b> (2)
🗌 benign	34,726	TCGA-AX-A2HC	TCGA-UCEC	Corpus uteri	Female	<u>65</u>	<u>6</u>	<u>10</u>	<u>16</u>	<u>5</u>	<u>2 10</u>	<u>16</u>	<u>1,095</u>	<u>434</u>	<b>上</b> (2)
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unknown	5 756		TCGA-PAAD		Female	<u>56</u>	<u>4</u>	<u>5</u>				<u>17</u>	<u>1,062</u>		<b>上</b> (3)

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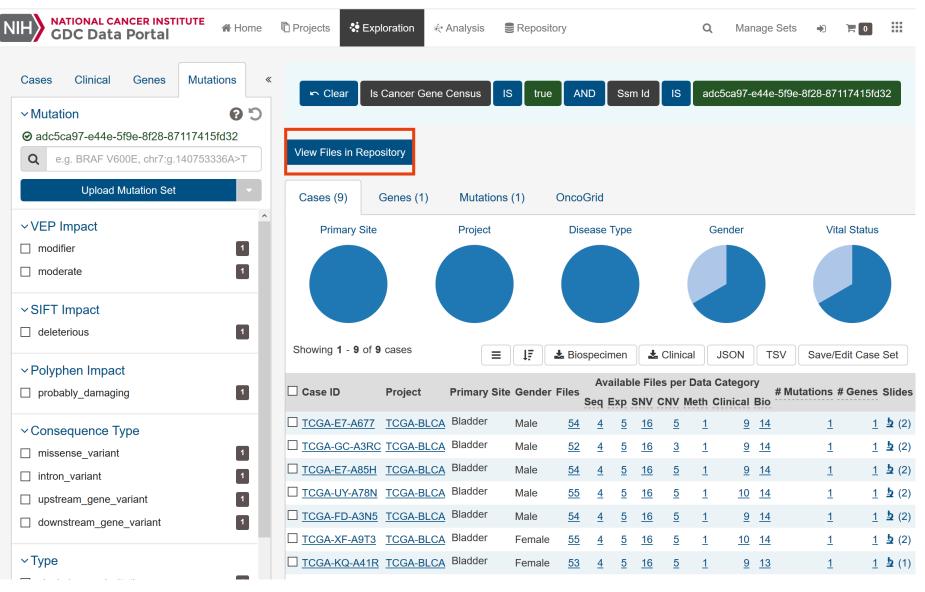
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#### Genomic Data Commons – download WXS BAM files for all ERCC2 N238S mutants



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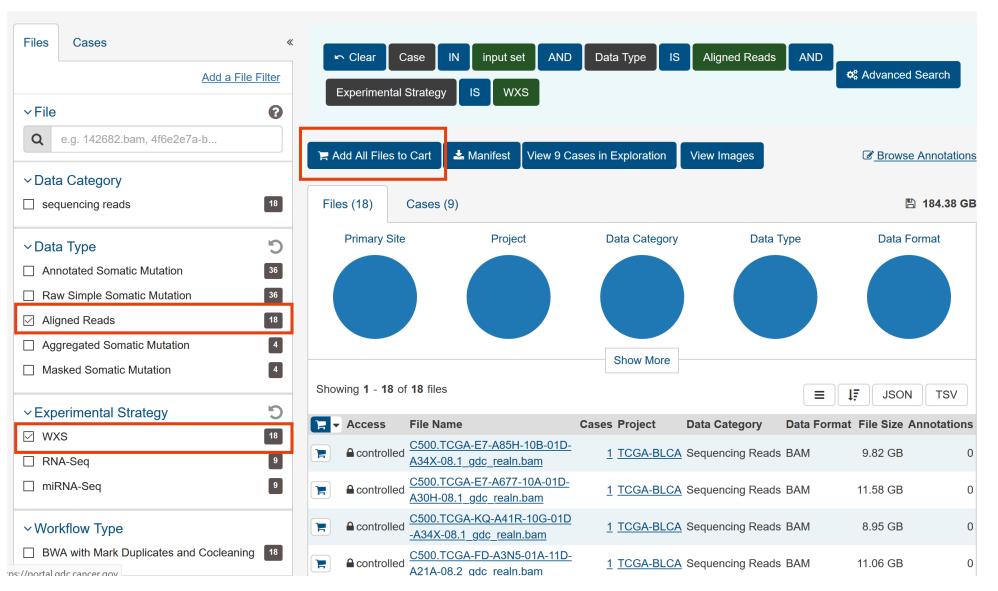






SIUM IN BIOINFORMATIC

# Genomic Data Commons – download WXS BAM files for all ERCC2 N238S mutants



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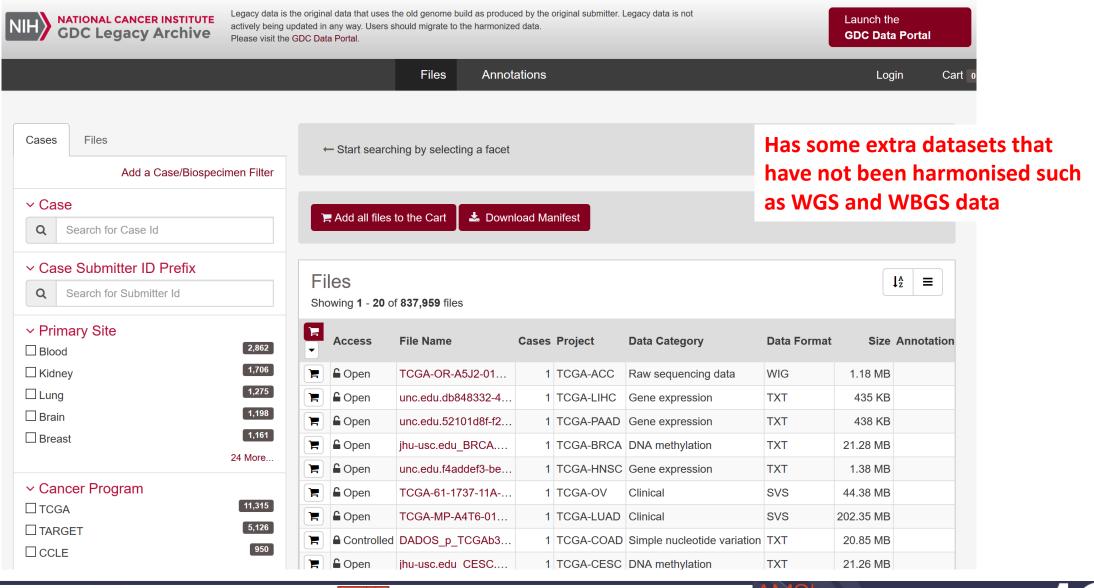




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## **Genomic Data Commons – Legacy archive**

## https://portal.gdc.cancer.gov/legacy-archive/search/f



CHILDREN'S MEDICAL RESEARCH INSTITUTE Jeans for Genes





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# **UCSC Xena Browser**

Downloading tidy processed data













Overview	Analysis	Tutorials	What's New	Cite Us	Subscribe	

# **UCSC Xena**

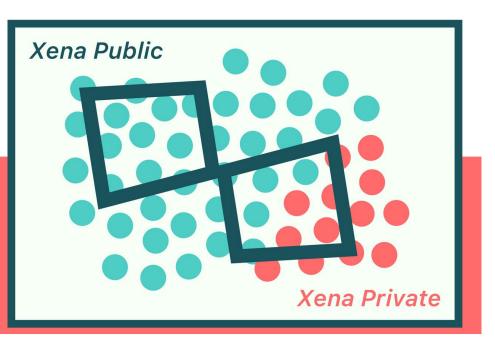
## See the bigger picture

An online exploration tool for public and private, multi-omic and clinical/phenotype data

Launch Xena

## **Tutorials and walkthroughs**

Don't know where to start? Jump in with one of our tutorials or "How do I ..." walkthroughs



Totals



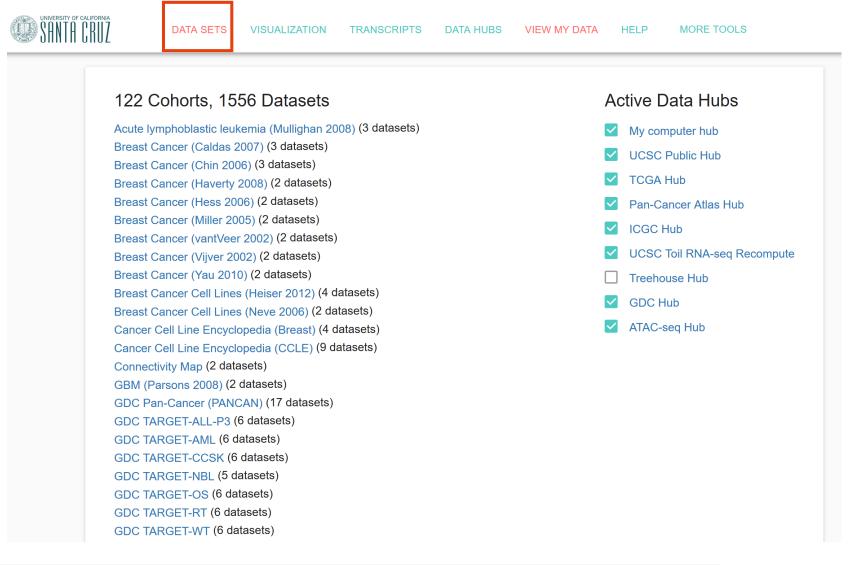








## https://xenabrowser.net/datapages/



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## cohort: GDC TCGA Lung Adenocarcinoma (LUAD)

### VISUALIZE

## copy number

Copy Number Segment (n=532) GDC Hub More information on the GDC pipeline used to generate this data: https://docs.gdc.cancer.gov/Data/Bioinformatics Pipelines/CNV Pipeline/

Masked Copy Number Segment (n=532) GDC Hub More information on the GDC pipeline used to generate this data: https://docs.gdc.cancer.gov/Data/Bioinformatics Pipelines/CNV Pipeline/

### copy number (gene-level)

### GISTIC - focal score by gene (n=531) GDC Hub

The GDC GISTIC copy number dataset is derived from focal copy number estimates. Larger chromosomal-level deletions may not be not captured in this dataset. More information on the GDC pipeline used to generate this data. https://docs.gdc.cancer.gov/Data/Bioinformatics Pipelines/CNV Pipeline/

### DNA methylation

Illumina Human Methylation 27 (n=150) GDC Hub More information on the GDC pipeline used to generate this data: https://docs.gdc.cancer.gov/Data/Bioinformatics Pipelines/Methylation LO Pipeline/

Illumina Human Methylation 450 (n=503) GDC Hub More information on the GDC pipeline used to generate this data: https://docs.gdc.cancer.gov/Data/Bioinformatics\_Pipelines/Methylation\_LO\_Pipeline/

### gene expression RNAseq

HTSeg - Counts (n=585) GDC Hub More information on the GDC pipeline used to generate this data: https://docs.gdc.cancer.gov/Data/Bioinformatics\_Pipelines/Expression\_mRNA\_Pipeline/

HTSeg - FPKM (n=585) GDC Hub More information on the GDC pipeline used to generate this data: https://docs.gdc.cancer.gov/Data/Bioinformatics Pipelines/Expression mRNA Pipeline/

HTSeg - FPKM-UQ\* (n=585) GDC Hub More information on the GDC pipeline used to generate this data: https://docs.gdc.cancer.gov/Data/Bioinformatics\_Pipelines/Expression\_mRNA\_Pipeline/

### phenotype

Phenotype (n=877) GDC Hub

survival data (n=738) GDC Hub

somatic mutation (SNPs and small INDELs)

MuSE Variant Aggregation and Masking (n=563) GDC Hub More information on the GDC pipeline used to generate this data: https://docs.gdc.cancer.gov/Data/Bioinformatics\_Pipelines/DNA\_Seq\_Variant\_Calling\_Pipeline/#somatic-variant-calling-workflow

MuTect2 Variant Aggregation and Masking<sup>\*</sup> (n=567) GDC Hub

More information on the GDC pipeline used to generate this data: https://docs.gdc.cancer.gov/Data/Bioinformatics\_Pipelines/DNA\_Seq\_Variant\_Calling\_Pipeline/#somatic-variant-calling-workflow

#### SomaticSniper Variant Aggregation and Masking (n=557) GDC Hub

More information on the GDC pipeline used to generate this data: https://docs.gdc.cancer.gov/Data/Bioinformatics\_Pipelines/DNA\_Seq\_Variant\_Calling\_Pipeline/#somatic-variant-calling-workflow











### VISUALIZE dataset: somatic mutation (SNPs and small INDELs) - MuTect2 Variant

## Aggregation and Masking

hub: https://gdc.xenahubs.net

More information on the GDC pipeline used to generate this data: https://docs.gdc.cancer.gov/Data/Bioinformatics\_Pipelines /DNA\_Seq\_Variant\_Calling\_Pipeline/#somatic-variant-calling-workflow

cohort	GDC TCGA Lung Adenocarcinoma (LUAD)
dataset ID	TCGA-LUAD.mutect2_snv.tsv
download	https://gdc.xenahubs.net/download/TCGA-LUAD.mutect2_snv.tsv.gz; Full metadata
samples	567
version	07-20-2019
type of data	somatic mutation (SNPs and small INDELs)
assembly	hg38
platform	Illumina
author	Genomic Data Commons
raw data	https://docs.gdc.cancer.gov/Data/Release_Notes/Data_Release_Notes/#data-release-180
raw data	https://api.gdc.cancer.gov/data/0458c57f-316c-4a7c-9294-ccd11c97c2f9
wrangling	

Data from different samples is combined into mutation/Vector; "Hugo Symbol", "Chromosome", "Start Position", "End Position", "Reference Allele", "Tumor Seq Allele2", "Tumor Sample Barcode", "HGVSp Short" and "Consequence" data are renamed accordingly and presented; "dna vaf" data is added and is calculated by "t\_alt\_count"/"t\_depth".

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Variant by Position (i.e. mutationVector) input data format

#### All Identifiers All Samples

	alt	amino-acid	chrom	chromstart	chromend	effect	ref	sampleID
TCGA-17- Z056-01A	A	p.W142*	chr1	69516	69516	stop_gained	G	TCGA-17- Z056-01A
TCGA-53- A4EZ-01A	G	p.I218M	chr1	69744	69744	missense_variant	С	TCGA-53- A4EZ-01A
TCGA-55-8505-0 1A	CGA	p.H236_I237insR	chr1	69798	69799	inframe_insertion	-	TCGA-55-8505-0 1A
TCGA-L9- A444-01A	CGAATGG	p.I237Rfs*28	chr1	69798	69799	frameshift_variant	-	TCGA-L9- A444-01A
						10 C C C C C C C C C C C C C C C C C C C		

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# **GDC and Xena Browser**

Exercise...

How many BAM files are present for TCGA-OR-A5J2?

2 x WXS, 1 x ATAC-seq, 1 x RNA-seq and 1 x miRNA-seq

Challenge...

What is the Bcell\_receptors\_score for TCGA-02-2485-01? (Hint: look for Pancancer analysis data in Xena browser. B-Cells are part of the immune system)

-0.4925 TCGA Pan-Cancer (PANCAN) → Signatures → Immune Signature Scores











Accessing and analyzing data in the cloud











Projects - Data - Public Apps Public projects	<ul> <li>Developer</li> </ul>	🌲 👻 jasonwhwong 👻
Projects Search	Q	Getting started
CONTROLLED LAML-WGS		Your account has been successfully created. Check your account settings.
Created by jasonwhwong · Nov. 2, 2019 22:26		Walk through the QuickStart.
Created by jasonwhwong · Sept. 24, 2019 15:37		Create a project.
CONTROLLED POLE Created by jasonwhwong · Sept. 24, 2019 13:17		Invite a collaborator.
CONTROLLED TE Created by jasonwhwong · June 3, 2019 12:36		Browse cases using the Case Explorer.
CONTROLLED Xiaoqiang Created by xiaoqiangzhu · June 3, 2019 12:33		Search datasets using the Data Browser.
		Upload your private data to analyze it along with public datasets.
Create a project     View all projects	< >	Bring your tools using the SDK.
Public Data and Apps		Build a workflow using the workflow editor.
		Get your authentication token and follow the API Quickstart.
Analyze Use som		Join our growing user community on the CGC forum.
1.142.997 41		Hide this forever

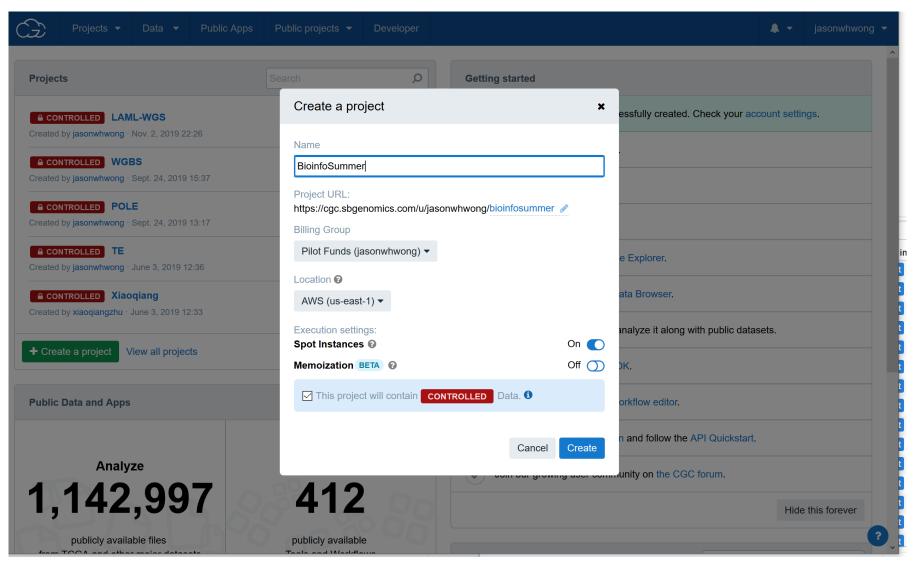






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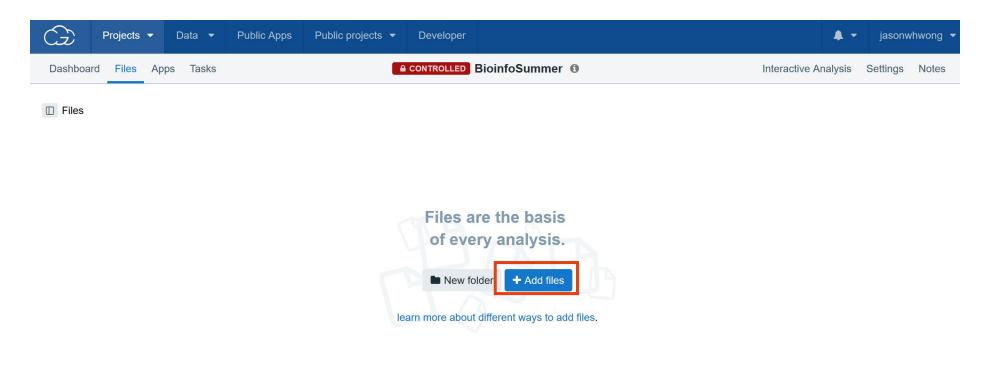






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# **Cancer genomics cloud – Get WXS BAM files for ERCC2 mutant bladder cancers**





 Add files to "BioinfoSummer"

 Public Files
 Case Explorer and Data Browser
 Projects
 FTP / HTTP
 Data Tools
 Volumes
 Import from PDC

Case Explorer Explore processed TCGA data.

Go to Case Explorer

Data Browser Query TCGA metadata and add files to your projects.

Go to Data Browser



lect Investigation 👻 BL	CA and gene	ERCC2				Top mutated	genes in BLCA		
40 - - 35 -						Gene	Cases with mutation		
8						TTN	202		^
85 -						TP53	173		
						KMT2D	93		
80 -						SYNE1	86		
•						PIK3CA	85		
25 -						MACF1	76		
©						TBC1D12	75		
20 <b>P</b>						KDM6A	74		
•						HMCN1	74		
5.0						ARID1A	73		
59						RYR2	73		
<b>*</b>						OBSCN	66		
0-						DST	64		~
15- © 8	0					Partial Data (	(not shown due to mise	sing values)	
0						Case ID		Expression	CNV
- 8							-4cf4-a638-312786f547c2		0.0751
5 0 5	10	15	20	25	30 35		-49ab-ba78-9636f1989fca		-0.0342
					Gene Expression (normalized)		-4736-97f6-2dc4ee30e943		0.2932
					C		-4ea8-8e51-1262b8d6ff90		0.1865
Mutation Variant Class	ification	Gender							
	ense variant (0) gained (0) variant (0)	Male (13) Female (4)							
						Oontinue To			







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TCGA GRCh38 New query Edited	S 68 out of 68 file(s) copi	ied to BioinfoSummer 🗶	Create new query Queries -	Q Search by ID
				Choose the target project
	File 🖓 🙃 x — Add entity	^		
Case Find Specific	File 🖓 🛈 🛪 Add entity			CONTROLLED BioinfoSummer
4ff4d9fe-eab8-4ab3	BAM Sample		-	CONTROLLED LAML-WGS
ed36d054-53c4-46 edc290bf-9731-4f0	Experimental strategy () × WXS Portion		-	
01c815ba-7bda-4f7 76400a3d-45c4-47f	Add property ×		-	
cd20b968-b348-44 661967e5-94b3-4f9	Properties and values <b>O</b>			
6b2d63f2-f390-460 bd7f2c90-863e-438	Find specific			CONTROLLED Xiaoqiang
d2f329b3-5b44-414 🗸	Access level			Files marked with a lock a can be
	Created datetime  Analysis Data category			used inside <b>CONTROLLED</b> project
	Data type 🕕			
_	File size			
Case 17 File 34 >				Export - Details
	Details for C500.TCGA-GC-A3RC-01A-1	11D-A22Z-08.7_gdc_realn.bam	Connections	
C500.TCGA-GC-A3RC-01A-11D-A22Z-08.7_gdc_realn.bam	TCGA GRCh38	^	Inbound: Case 1	
C500.TCGA-DK-AA77-10A-01D-A394-08.1_gdc_realn.bam	Access level () Co	ontrolled	bd7f2c90-863e-4380-ad31-8755b	ucb2f975
C500.TCGA-XF-A9SM-01A-11D-A42E-08.2_gdc_realn.bam		116-05-04T04:46:55.814Z	Outhound	
C500.TCGA-MV-A51V-01A-11D-A26M-08.5_gdc_realn.bam		AM	Outbound:	
C500.TCGA-XF-AAMZ-01A-11D-A42E-08.2_gdc_realn.bam	Data type 🖲 Ali	igned Reads		tbound connections

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Projects - Data - Public Apps Public projects - Developer				🌲 👻 jaso	onwhwong 🔻
Dashboard Files Apps Tasks	ner 0			Interactive Analysis Setting	gs Notes
Files				New folder + Add	l files
□ ▼ <sub>▼</sub> Name	Experimental strategy	Туре	Size	Sample ID	
C500.TCGA-YF-AA3L-10A-01D-A38J-08.2_gdc_realn.bam	WXS	BAM	7.4 GiB	TCGA-YF-AA3L-10A	^
C500.TCGA-YF-AA3L-10A-01D-A38J-08.2_gdc_realn.bai	WXS	BAI	5.9 MiB	TCGA-YF-AA3L-10A	
TCCA GRCH38 C500.TCGA-YF-AA3L-01A-11D-A38G-08.2_gdc_realn.bam	WXS	BAM	8.6 GiB	TCGA-YF-AA3L-01A	
C500.TCGA-YF-AA3L-01A-11D-A38G-08.2_gdc_realn.bai	WXS	BAI	5.9 MiB	TCGA-YF-AA3L-01A	
C500.TCGA-XF-AAMZ-10A-01D-A42H-08.2_gdc_realn.bam	WXS	BAM	5.8 GiB	TCGA-XF-AAMZ-10A	
C500.TCGA-XF-AAMZ-10A-01D-A42H-08.2_gdc_realn.bai	WXS	BAI	5.9 MiB	TCGA-XF-AAMZ-10A	
C500.TCGA-XF-AAMZ-01A-11D-A42E-08.2_gdc_realn.bam	WXS	BAM	7.7 GiB	TCGA-XF-AAMZ-01A	
C500.TCGA-XF-AAMZ-01A-11D-A42E-08.2_gdc_realn.bai	WXS	BAI	5.9 MiB	TCGA-XF-AAMZ-01A	
C500.TCGA-XF-AAMX-10A-01D-A42H-08.2_gdc_realn.bam	WXS	BAM	7.5 GiB	TCGA-XF-AAMX-10A	
C500.TCGA-XF-AAMX-10A-01D-A42H-08.2_gdc_realn.bai	WXS	BAI	5.9 MiB	TCGA-XF-AAMX-10A	
C500.TCGA-XF-AAMX-01A-11D-A42E-08.2_gdc_realn.bam	WXS	BAM	7.9 GiB	TCGA-XF-AAMX-01A	
C500.TCGA-XF-AAMX-01A-11D-A42E-08.2_gdc_realn.bai	WXS	BAI	5.9 MiB	TCGA-XF-AAMX-01A	
C500.TCGA-XF-A9SM-10A-01D-A42H-08.2_gdc_realn.bam	WXS	BAM	6.1 GiB	TCGA-XF-A9SM-10A	
C500.TCGA-XF-A9SM-10A-01D-A42H-08.2_gdc_realn.bai	WXS	BAI	5.9 MiB	TCGA-XF-A9SM-10A	~
C Refresh				Showing 1-68	of 68 < >

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CHILDREN'S MEDICAL C RESEARCH INSTITUTE Jeans for Genes







Dashboard Files Apps Tasks

## CONTROLLED BioinfoSummer ①

Interactive Analysis

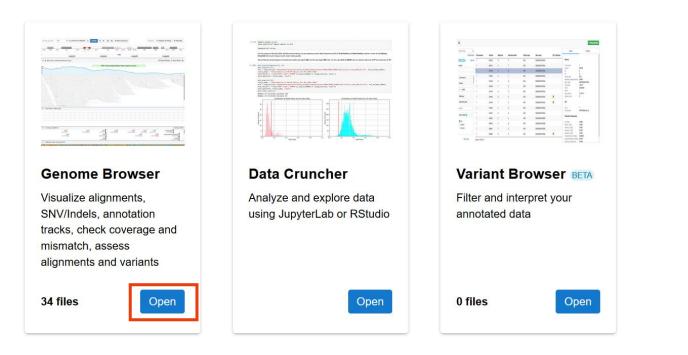
## Explore genomics data

Understand complex genomics data with interactive analysis tools.

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shboard Files Apps	Tasks		BioinfoSummer			Interactive Analysis	s Settings
Genome Brow							+ Add Bam files
Genome Brow							
uman hg38 🔹 chr19	<ul> <li>✓ 45,365,042-45,370,528</li> </ul>	> Q Q III					Marker
oband Track							
p13.3	p13.2 p13.13 p13.12 p13.11	p12 p11 q11	q12 q13.11	q13.12 q13.2	q13.31 q13.32	q13.33 q13.41	q13.42 g
			5.49 Kbp				
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	3X1-01A-12D-A22Z-08.7_gdc_realn.bam File id: 5de	1 I I I					

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Dashboard Files Apps Tasks

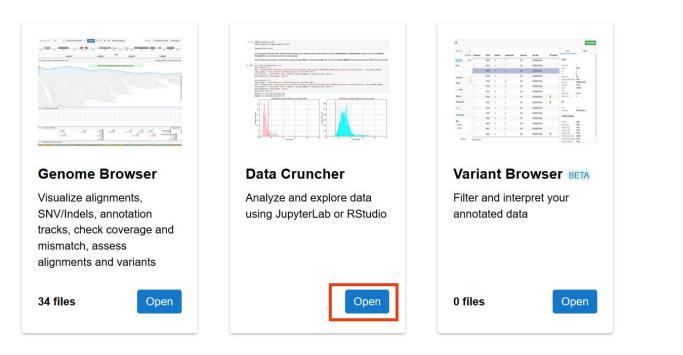
## CONTROLLED BioinfoSummer ①

Interactive Analysis

## Explore genomics data

Understand complex genomics data with interactive analysis tools.

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Dashboard Files Apps Tasks

Interactive Analysis S

Create new analysis	ډ		Create new analysis
Basic information	Compute requirements		Basic information Compute requirements
Analysis name Test Environment		Internet	Select an instance type with adequate CPU, memory and storage allocation for your analysis. This can be changed between analysis runs, but not while the analysis is running.
JupyterLab Web-based UI for Project Jupyter	RStudio BETA IDE for R	ting across multiple pro	Instance type       Suspend time I       On       On         m4.large (1024GB EBS, 2vCPUs, 8GB RA)       30       ₩ Minutes
Skip wizard	Previous	Create your first ana	Instance types m4.16xlarge (1024GB EBS, 64vCPUs, 256GB RAM)
Skip wizaru	Previous Next		m4.2xlarge (1024GB EBS, 8vCPUs, 32GB RAM) m4.4xlarge (1024GB EBS, 16vCPUs, 64GB RAM)
			m4.large (1024GB EBS, 2vCPUs, 8GB RAM) m4.xlarge (1024GB EBS, 4vCPUs, 16GB RAM)

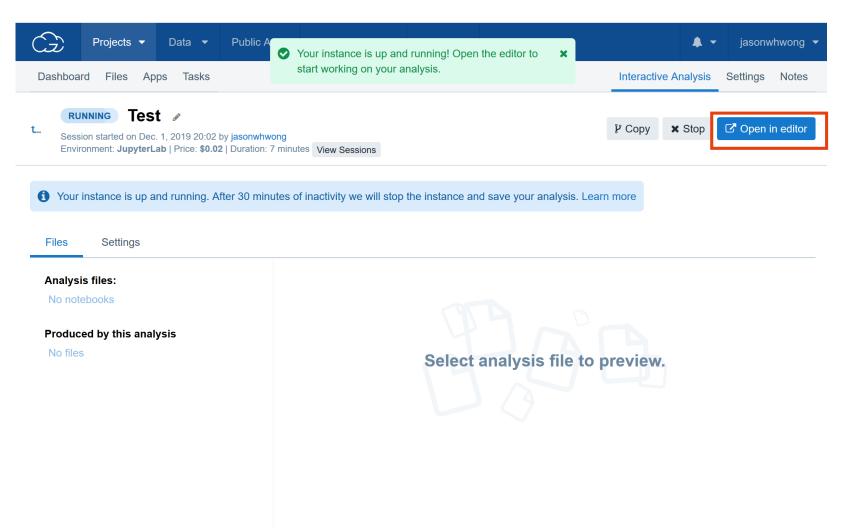












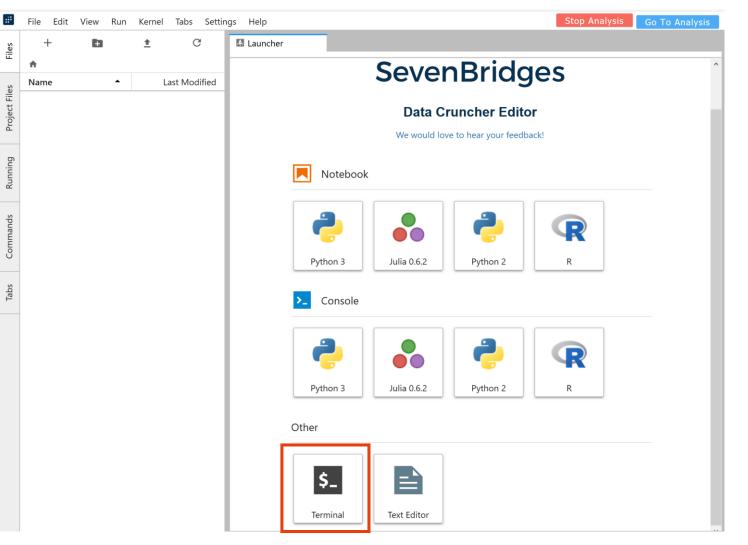












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Project files are in "/sbgenomics/project-files" Write files to "/sbgenomics/workspace"

	+ 🗈 ±	C	⊑ jovyan@6b036; ×	
	A		/sbgenomics/workspace\$ ll /sbgenomics/project-files/	
	Name		total 325891758	
	Name L	ast Modified	-rw-rr- 1 1001 1002 6185888 Dec 1 08:54 C500.TCGA-DK-A3X1-01A-12D-A22Z-08.7_gdc_realn.bai	
			-rw-rr- 1 1001 1002 10705997580 Dec 1 08:54 C500.TCGA-DK-A3X1-01A-12D-A22Z-08.7_gdc_realn.bam	
5			-rw-rr- 1 1001 1002 6243376 Dec 1 08:54 C500.TCGA-DK-A3X1-10A-01D-A22Z-08.9_gdc_realn.bai	
110000			-rw-rr- 1 1001 1002 12245607442 Dec 1 08:54 C500.TCGA-DK-A3X1-10A-01D-A22Z-08.9_gdc_realn.bam	
•			-rw-rr-1 1001 1002 6234200 Dec 1 08:54 C500.TCGA-DK-A6AW-01A-11D-A30E-08.1_gdc_realn.bai	
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			-rw-rr- 1 1001 1002 6226784 Dec 1 08:54 C500.TCGA-DK-AA77-10A-01D-A394-08.1_gdc_realn.bai	
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			-rw-rr- 1 1001 1002 12784067246 Dec 1 08:54 C500.TCGA-E7-A7XN-01A-11D-A34U-08.1_gdc_realn.bam	
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3			-rw-rr- 1 1001 1002 10036280024 Dec 1 08:54 C500.TCGA-E7-A7XN-10A-01D-A34X-08.1_gdc_realn.bam	
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			-rw-rr 1 1001 1002 6283792 Dec 1 08:54 C500.TCGA-FJ-A3Z7-01A-12D-A23M-08.3_gdc_realn.bai	
			-rw-rr 1 1001 1002 13982022480 Dec 1 08:54 C500.TCGA-FJ-A3Z7-01A-12D-A23M-08.3_gdc_realn.bam	
			-rw-rr 1 1001 1002 6204624 Dec 1 08:54 C500.TCGA-FJ-A3Z7-10A-01D-A23K-08.3_gdc_realn.bai	
			-rw-rr 1 1001 1002 11157407292 Dec 1 08:54 C500.TCGA-FJ-A3Z7-10A-01D-A23K-08.3_gdc_realn.bam	
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			-rw-rr 1 1001 1002 6159448 Dec 1 08:54 C500.TCGA-GC-A3RC-10B-01D-A22Z-08.9_gdc_realn.bai	
			-rw-rr 1 1001 1002 9651371746 Dec 1 08:54 C500.TCGA-GC-A3RC-10B-01D-A22Z-08.9_gdc_realn.bam	
			-rw-rr 1 1001 1002 6215448 Dec 1 08:54 C500.TCGA-GU-AATO-01A-11D-A391-08.1_gdc_realn.bai	
			-rw-rr 1 1001 1002 10145211195 Dec 1 08:54 C500.TCGA-GU-AATO-01A-11D-A391-08.1_gdc_realn.bam	
			-rw-rr 1 1001 1002 6273616 Dec 1 08:54 C500.TCGA-GU-AATO-10A-01D-A394-08.1_gdc_realn.bai	
			-rw-rr 1 1001 1002 7439992923 Dec 1 08:54 C500.TCGA-GU-AATO-10A-01D-A394-08.1_gdc_realn.bam	
			-rw-rr- 1 1001 1002 6199584 Dec 1 08:54 C500.TCGA-GU-AATP-01A-11D-A391-08.1_gdc_realn.bai	
			-rw-rr- 1 1001 1002 9805654032 Dec 1 08:54 C500.TCGA-GU-AATP-01A-11D-A391-08.1_gdc_realn.bam	

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## Upload your precompiled Ubuntu tools

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Files	<b>↑</b>		/sbgenomics/workspace\$ ll	
	Name 🔺	Last Modified	total 2980 -rw-rr 1 jovyan users 3050748 Dec  1 09:19 Tools_for_linux.tar.gz	
Project Files	Tools	a year ago	/sbgenomics/workspace\$ tar -zxvf Tools_for_linux.tar.gz	
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			Tools/maskFastaFromBed	
			Tools/bamToBed	
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ڒ			Tools/fastaFromBed	
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			Tools/lofreq2_call_pparallel.py	
			Tools/slopBed	
			Tools/intersectBed	
			Tools/randomBed	
			Tools/complementBed	
			Tools/shuffleBed	
			Tools/mergeBed	
			Tools/closestBed	
			Tools/sortBed	
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			Tools/lofreq	
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			/sugenomites/workspaces	

https://cloudstor.aarnet.edu.au/plus/s/O7gw6CRJU3H0rIK/download











≤ jovyan@6b036; × A Launcher  $\times$ /sbgenomics/workspace\$ samtools view /sbgenomics/project-files/C500.TCGA-MV-A51V-01A-11D-A26M-08.5\_gdc\_realn. am |more H0WVJADXX130808:1:2113:14065:6145 10064 chr9 13371363 161 chr1 0 46M30S 0 CCTAACCCTAACCCTAACCCTAACCCTAACCCTAACCCATTAACCATCCTCTCATTTTCTCCCCCTTTCC =><@>@ABBC@BAA@C@BBB@B@BBABC? BAAC@BBAAC@BBAABAA@BBABACACBCBBABAACBCBBBB??BA SA:Z:chr9,13371215,+,41S35M,0,0; MC:Z:76M BD:Z: MKMMMMMKMLLLLJLLLLJLLMLMKLMMMMKMMMMMKMMMMMKMNLLMMMMNNMMMMMNNLFFMMMMKKNLFMM MD:Z:46 RG:Z:H0WVJ.1 BI:Z: NM:i:0 MQ:i:60 AS:i:46 XS:i:4 6 10377 chr2 242183414 H0WVKADXX130808:1:1212:19059:9322 113 chr1 0 66M10S 0 CCCTAACCCTAACCCCTAACCCTAACCCTAACCCTAACCCTAACCCTAACCCTAACCCTAAGGAGAAC #66=<>>?>=<>>?@@C?<?BBBA>:>BB >>>BBB>;?BBB>;>BBA>>>BBA>;?BAB>:>AAD@=CACBB=@@ MC:Z:37S31M1I7M BD:Z:MLNMMNNMNMLMMLLNMLMMLNMLMMLNMLMMLNMLMML MLMMLNMLMLLNLKLLKMLKLMLMMLLMMMNMMMM RG:Z:HOWVK.1 MD:Z:0A65 BI:Z:SRSSSSSRSSSSSRRSSSSSRRSSSSSRSSSS NM:i:1 MO:i:0 AS:i:65 XS:i:63 H0WG1ADXX130811:1:1201:16182:18532 65 chr1 11184 0 76M 43450325 43439 42 CGGGCACTACAGGACCCGCTTGCTCACGGTGCTGTCCCAGGGCGCCCCCTGCTGGCGACTAGGGCAACTGCAGGGN @<=@CBCBAB@DA@AAA<BA@ BAAA@<C@CCBC@AABACBBC<A@A>?ABCBAAC=ABB>D??@A>BBD@@A>7# MC:Z:76M BD:Z:MMMLONMNMMMONNNNLMLNKLMMMNLLLLLM /sbgenomics/workspace\$ samtools view /sbgenomics/project-files/C500.TCGA-MV-A51V-01A-11D-A26M-08.5\_gdc\_realn. more am H0WVJADXX130808:1:2113:14065:6145 161 chr1 10064 0 46M30S chr9 13371363 0 CCTAACCCTAACCCTAACCCTAACCCTAACCCTAACCCATTAACCATCCT<u>CTCATTTTCTCCCCTTTCC</u> =><@>@ABBC@BAA@C@BBB@B@BBABC? BAAC@BBAAC@BBAABAA@BBABACACBCBBABAACBCBBBB??BA SA:Z:chr9,13371215,+,41S35M,0,0; MC:Z:76M BD:Z: MD:Z:46 RG:Z:HOWVJ.1 BI:Z: NM:i:0 MQ:i:60 AS:i:46 XS:i:4

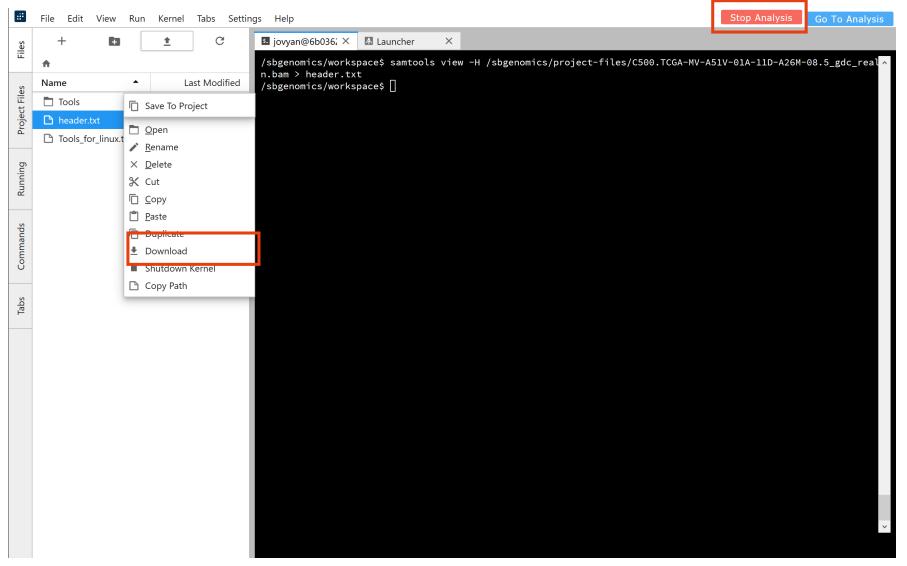
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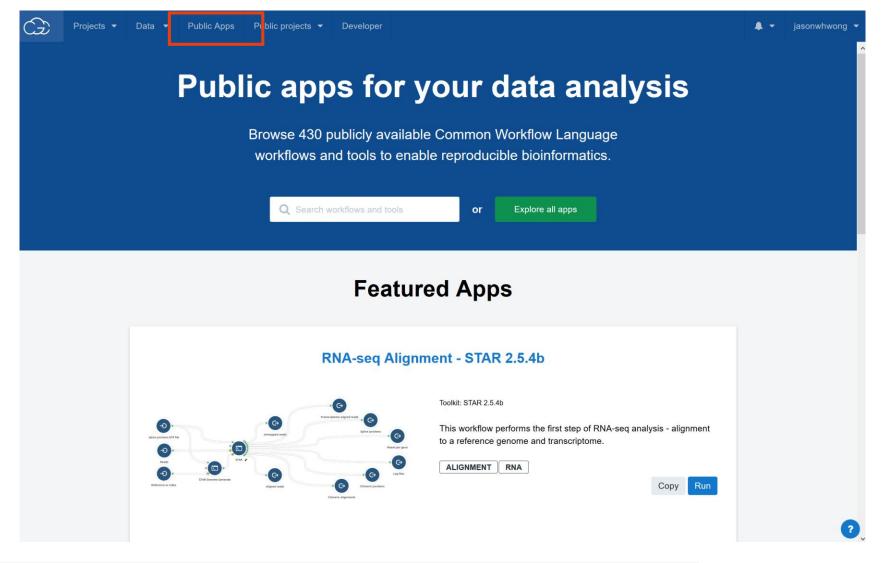
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