

GDC Data Portal User's Guide

NCI Genomic Data Commons (GDC)

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Chapter 1

Getting Started

Getting Started

The GDC Data Portal: An Overview

The Genomic Data Commons (GDC) Data Portal provides users with web-based access to data from cancer genomics studies. Key GDC Data Portal features include:

- Open, granular access to information about all datasets available in the GDC
- Advanced search and visualization-assisted filtering of data files
- Data visualization tools to support the analysis and exploration of data (including on a gene and mutation level from Open-Access MAF files)
- Cart for collecting data files of interest
- Authentication using eRA Commons credentials for access to controlled data files
- Secure data download directly from the cart or using the [GDC Data Transfer Tool](#)

For more information about available datasets, see the [GDC Website](#).

Accessing the GDC Data Portal

The GDC Data Portal is accessible using a web browser such as Chrome, Internet Explorer, and Firefox at the following URL:

<https://portal.gdc.cancer.gov>

The front page displays a summary of all available datasets:

NIH NATIONAL CANCER INSTITUTE GDC Data Portal Home Projects Exploration Analysis Repository Quick Search Manage Sets Login Cart 0 GDC Apps

Harmonized Cancer Datasets Genomic Data Commons Data Portal

Get Started by Exploring:

Projects Exploration Analysis Repository

Q e.g. BRAF, Breast, TCGA-BLCA, TCGA-A5-A0G2

Data Portal Summary Data Release 8.0 - August 22, 2017

PROJECTS	PRIMARY SITES	CASES
40	60	32,555
FILES	GENES	MUTATIONS
310,858	22,144	3,115,606

Cases by Major Primary Site

Primary Site	Cases
Adrenal Gland	~100
Bile Duct	~100
Bladder	~100
Blood	~100
Bone	~100
Bone Marrow	~100
Brain	~100
Breast	~3,500
Cervix	~100
Colorectal	~2,500
Esophagus	~100
Eye	~100
Head and Neck	~100
Kidney	~100
Liver	~100
Lung	~4,000
Lymph Nodes	~100
Nervous System	~100
Ovary	~100
Pancreas	~100
Pleura	~100
Prostate	~1,500
Skin	~100
Soft Tissue	~100
Stomach	~100
Testis	~100
Thymus	~100
Thyroid	~100
Uterus	~100

GDC Applications

The GDC Data Portal is a robust data-driven platform that allows cancer researchers and bioinformaticians to search and download cancer data for analysis. The GDC applications include:

- Data Portal
- Website
- Data Transfer Tool
- API
- Data Submission Portal
- Documentation
- Legacy Archive

Navigation

Views

The GDC Data Portal provides five navigation options (*Views*) for browsing available harmonized datasets:

Harmonized Cancer Datasets Genomic Data Commons Data Portal

Get Started by Exploring:

Projects Exploration Analysis Repository

Q e.g. BRAF, Breast, TCGA-BLCA, TCGA-A5-A0G2

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Colorectal	~2,500
Esophagus	~100
Eye	~100
Head and Neck	~100
Kidney	~100
Liver	~100
Lung	~4,000
Lymph Nodes	~100
Nervous System	~100
Ovary	~100
Pancreas	~100
Pleura	~100
Prostate	~1,500
Skin	~100
Soft Tissue	~100
Stomach	~100
Testis	~100
Thymus	~100
Thyroid	~100
Uterus	~100

- **Projects:** The Projects link directs users to the Projects Page, which gives an overall summary of project-level information, including the available data for each project.
- **Exploration:** The Exploration link takes users to the Exploration Page, which allows users to explore data by utilizing various case, genes and mutation filters.

- **Analysis:** The Analysis link directs users to the Analysis Page. This page has features available for users to compare different cohorts. These cohorts can either be generated with existing filters (e.g. males with lung cancer) or through custom selection.
- **Repository:** The Repository link directs users to the Repository Page. Here users can see the data files available for download at the GDC and apply file/case filters to narrow down their search.
- **Human Outline:** The home page displays a human anatomical outline that can be used to refine their search. Choosing an associated organ will direct the user to a listing of all projects associated with that primary site. For example, clicking on the human brain will show only cases and projects associated with brain cancer (TCGA-GBM and TCGA-LGG). The number of cases associated with each primary site is also displayed here and separated by project.

Each view provides a distinct representation of the same underlying set of GDC data and metadata. The GDC also provides access to certain unharmonized data files generated by GDC-hosted projects. These files and their associated metadata are not represented in the views above; instead they can be found in the GDC Legacy Archive.

The Projects, Exploration, Analysis and Repository pages can be accessed from the GDC Data Portal front page and from the toolbar (see below). The annotations view is accessible from Repository view. A link to the GDC Legacy Archive is available on the GDC Data Portal front page and in the GDC Apps menu (see below).

Toolbar

The toolbar available at the top of all pages in the GDC Data Portal provides convenient navigation links and access to authentication and quick search.

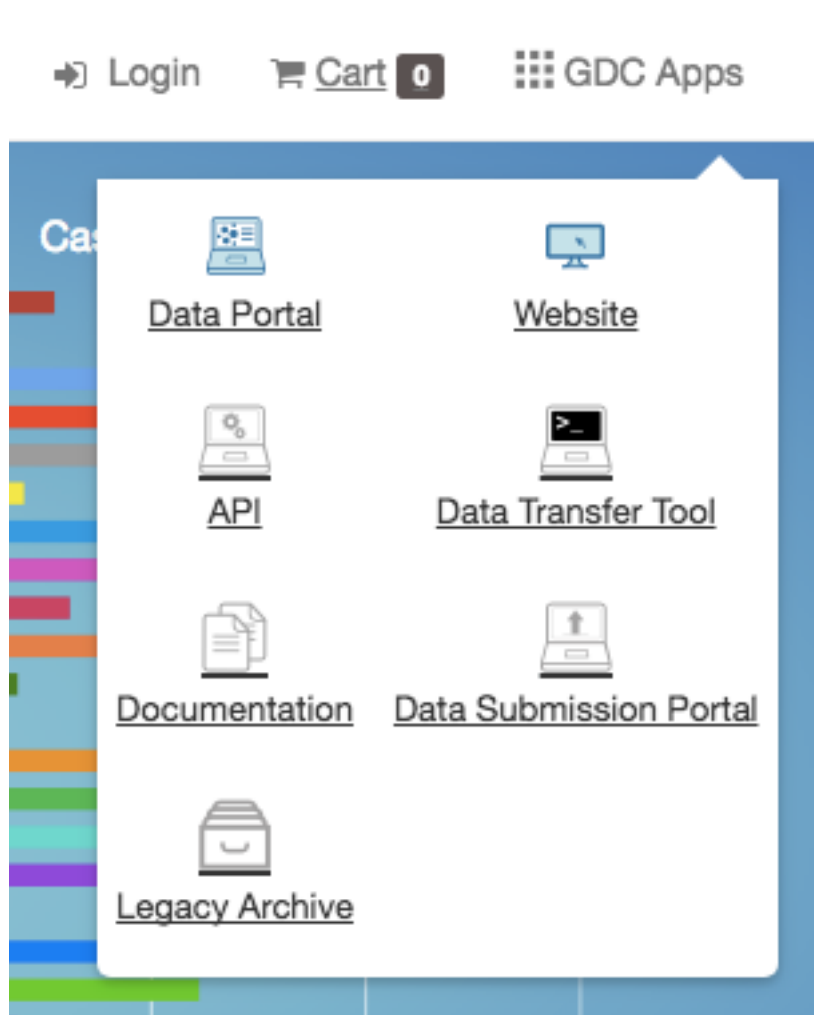
The left portion of this toolbar provides access to the Home Page, **Projects Page**, **Exploration Page**, **Analysis Page**, and a link to **Repository Page**:



The right portion of this toolbar provides access to quick search, the cart, and the GDC Apps menu:



The GDC Apps menu provides links to all resources provided by the GDC, including the GDC Legacy Archive.



Tables

Tabular listings are the primary method of representing available data in the GDC Data Portal. Tables are available in all views and in the file cart. Users can customize each table by specifying columns, size, and sorting.

Table Sort

The *sort table* button is available in the top right corner of each table. To sort by a column, place a checkmark next to it and select the preferred sort direction. If multiple columns are selected for sorting, data is sorted column-by-column in the order that columns appear in the sort menu: the topmost selected column becomes the primary sorting parameter; the selected column below it is used for secondary sort, etc.

Cart Items

Showing 1 - 8 of 8 files

Access	File Name	Cases	Project	Data Category	Data Format	Actions
controlled	1267c52a-607b-4d96-9d1e-49e18abe059d_gdc_realn_rehead.bam	1	TCGA-PAAD	Raw Sequencing Data	BAM	
controlled	130046.bam	1	TCGA-PAAD	Raw Sequencing Data	BAM	
controlled	143558.bam	1	TCGA-PAAD	Raw Sequencing Data	BAM	
controlled	37c5acdc-7406-4ea3-a7d0-ac572b738730_gdc_realn_rehead.bam	1	TCGA-PAAD	Raw Sequencing Data	BAM	
controlled	C546.TCGA-3A-A91B-01A-21D-A397-08.2_gdc_realn.bam	1	TCGA-PAAD	Raw Sequencing Data	BAM	
controlled	C546.TCGA-HZ-A49G-01A-11D-A261-08.4_gdc_realn.bam	1	TCGA-PAAD	Raw Sequencing Data	BAM	
controlled	C546.TCGA-1B-7654-10A-01D-2154-08.3_gdc_realn.bam	1	TCGA-PAAD	Raw Sequencing Data	BAM	
controlled	a0bbcf1d-3a05-41c6-9316-ae454e184205_gdc_realn_rehead.bam	1	TCGA-PAAD	Raw Sequencing Data	BAM	

Show 20 entries

File UUID, File Submitter ID, Access, File Name, Project, Data Category, Data Format, Size

Table Arrangement

The *arrange columns* button allows users to adjust the order of columns in the table and select which columns are displayed.

Table Size

Table size can be adjusted using the menu in the bottom left corner of the table. The menu sets the maximum number of rows to display. If the number of entries to be displayed exceeds the maximum number of rows, then the table will be paginated, and navigation buttons will be provided in the bottom right corner of the table to navigate between pages.

Table Export

In the Repository, Projects, and Annotations views, tables can be exported in either a JSON or TSV format. The JSON button will export the entire table's contents into a JSON file. The TSV button will export the current view of the table into a TSV file.

JSON TSV

Cases per Data Category					Files
W	CNV	Meth	Clinical	Bio	
6	0	0	7	1,127	2,806
4	1.096	1.095	1.097	1.098	27.207

Filtering and Searching

The GDC Data Portal offers three different means of searching and filtering the available data: facet filters, quick search, and advanced search.

Facet Filters

Facets on the left of each view (Projects, Exploration, and Repository) represent properties of the data that can be used for filtering. Some of the available facets are project name, disease type, patient gender and age at diagnosis, and various data

			≡	↓	TSV
Data Category	Q	Filter Columns	Annotations		
Raw Sequencing Data		Restore Defaults			0
Raw Sequencing Data	<input type="checkbox"/>	File UUID	≡		0
Raw Sequencing Data	<input type="checkbox"/>	File Submitter ID	≡		0
Raw Sequencing Data	<input checked="" type="checkbox"/>	Access	≡		0
Raw Sequencing Data	<input checked="" type="checkbox"/>	File Name	≡		0
Raw Sequencing Data	<input checked="" type="checkbox"/>	Cases	≡		0
Raw Sequencing Data	<input checked="" type="checkbox"/>	Project	≡		1
Raw Sequencing Data	<input checked="" type="checkbox"/>	Data Category	≡		0
	<input checked="" type="checkbox"/>	Data Format	≡		
	<input checked="" type="checkbox"/>	Size	≡	1	,
	<input checked="" type="checkbox"/>	Annotations	≡		»
	<input type="checkbox"/>	Data Type	≡		
	<input type="checkbox"/>	Experimental Strategy	≡		
	<input type="checkbox"/>	Platform	≡		

Figure 1.1: Selecting table columns

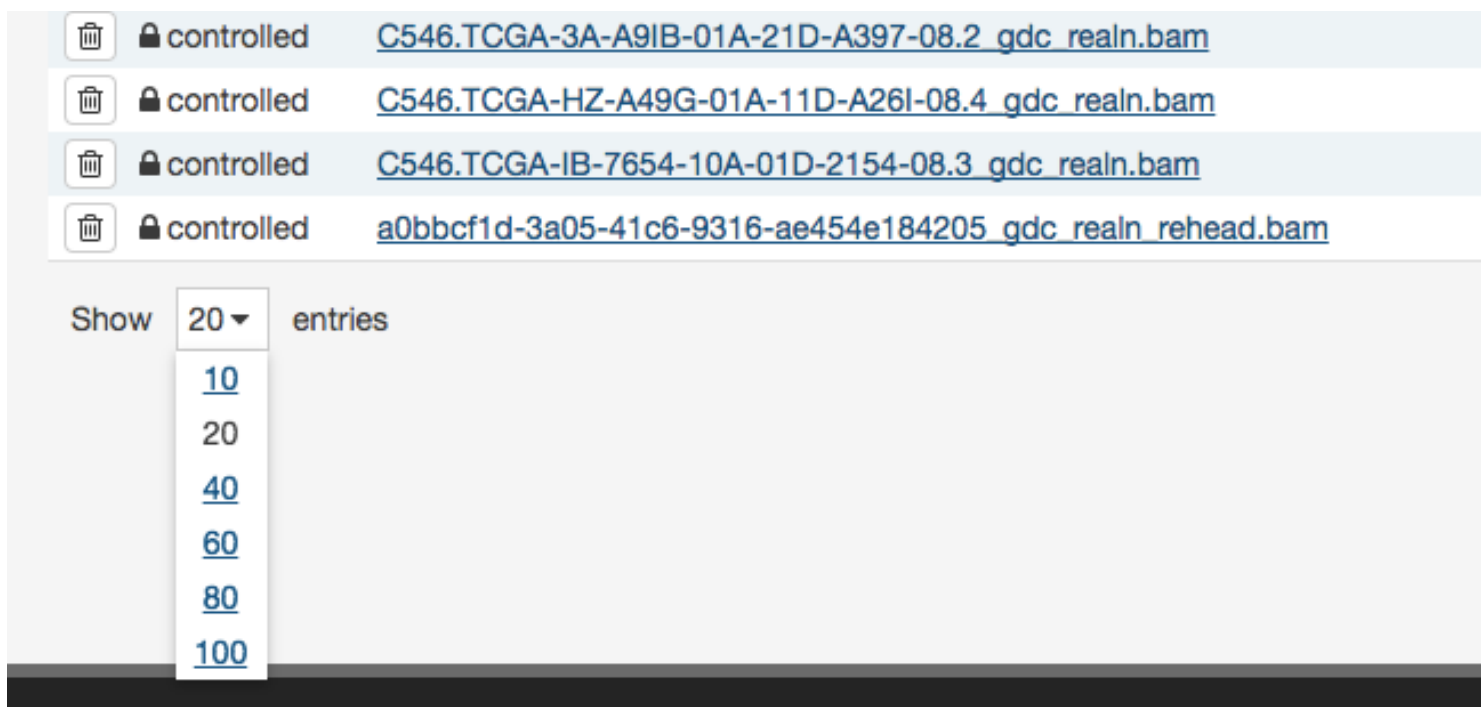


Figure 1.2: Specifying table size

formats and categories. Each facet displays the name of the data property, the available values, and numbers of matching entities for each value (files, cases, mutations, genes, annotations, or projects, depending on the context).

Below are two file facets available in the Repository view. A *Data Type* facet filter is applied, filtering for “Aligned Reads” files. Multiple selections within a facet are treated as an “OR” query: e.g. “Aligned Reads” OR “Annotated Somatic Mutation”. Selections in different facets are treated as “AND” queries: e.g. Data Type: “Aligned Reads” AND Experimental Strategy: “RNA-Seq”.

The information displayed in each facet reflects this: in the example above, marking the “Aligned Reads” checkbox does not change the numbers or the available values in the *Data Type* facet where the checkbox is found, but it does change the values available in the *Experimental Strategy* facet. The *Experimental Strategy* facet now displays only values from files of *Data Type* “Aligned Reads”.

Custom facet filters can be added in Repository View to expand the GDC Data Portal’s filtering capabilities.

Quick Search

The quick search feature allows users to find cases, files, mutations, or genes using a search query (i.e. UUID, filename, gene name, DNA Change, project name, id, disease type or primary site). Quick search is available by clicking on the magnifier in the right section of the toolbar (which appears on every page) or by using the search bar on the Home Page.

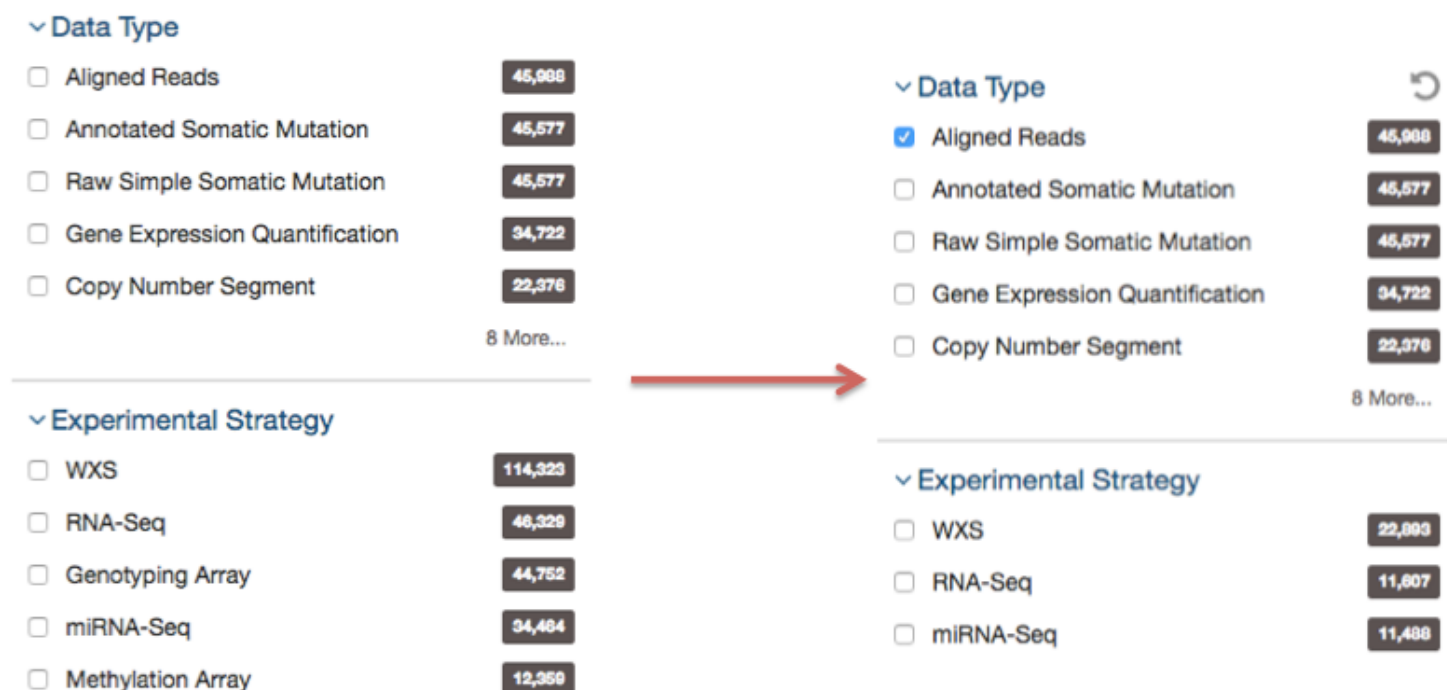
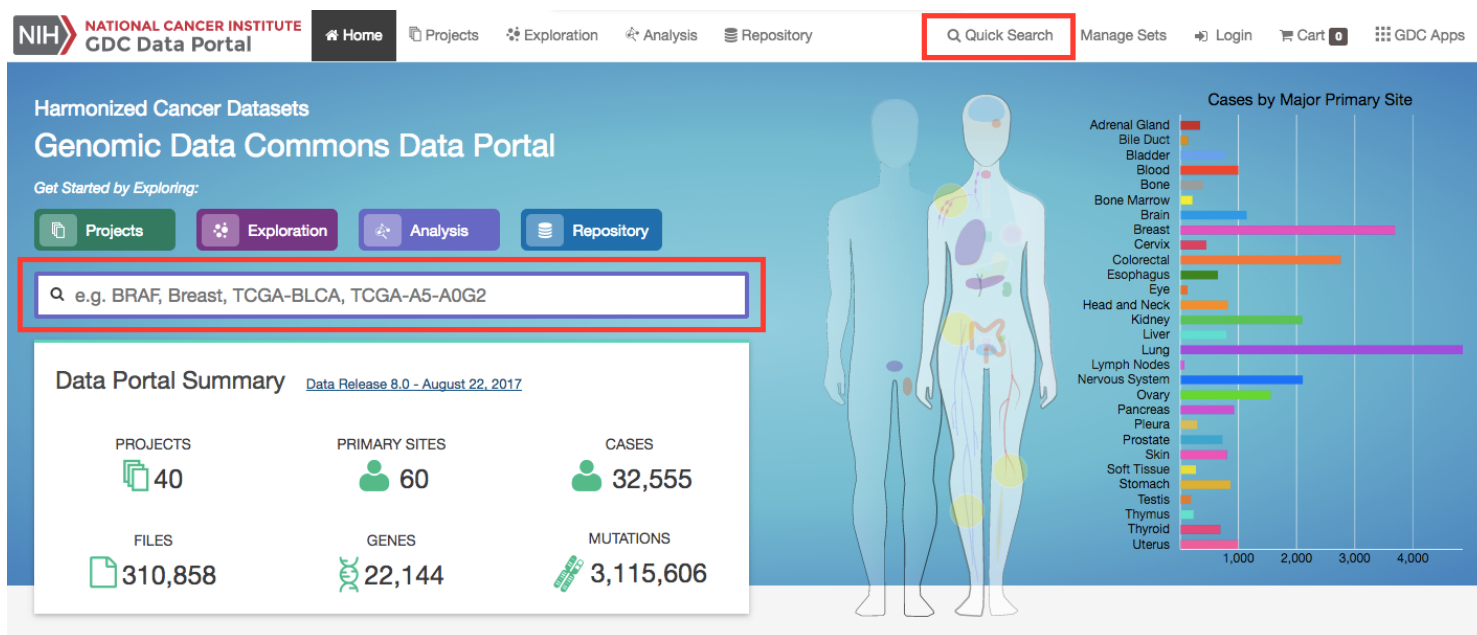


Figure 1.3: Facets (no filter applied)



Search results are displayed as the user is typing, with labels indicating the type of each search result in the list (project, case, or file). Users will see a brief description of the search results, which may include the UUID, submitter ID, or file name. Clicking on a selected result or pressing enter will open a detail page with additional information.

Home Page Quick Search:

Harmonized Cancer Datasets

Genomic Data Commons Data Portal

Get Started by Exploring:



Projects



Exploration



Analysis



Repository

Q TCGA-44-6147

CA	889aec8e-14ba-48d9-8fe1-f2416e82b333 TCGA-44-6147
FL	46f09f9b-6e3b-45f3-bb17-afbfdf5c5ecf TCGA-44-6147-01A-11D-A271-08_TCGA-44-6147-10A-01D-A271-08_mutect_annotated
FL	1662fb41-db13-4a4b-afff-e5b58160cda5 TCGA-44-6147-01A-21D-A27T-08_TCGA-44-6147-10A-01D-A271-08_varscan_annotated
FL	f797b2f9-0c16-419a-9eb1-e4da30bbefa9 TCGA-44-6147-01A-11D-1753-08_TCGA-44-6147-10A-01D-1753-08_muse_annotated
FL	22942df1-fa8b-45f6-87df-037d6e37abba TCGA-44-6147-01A-31D-A27T-08_TCGA-44-6147-10A-01D-1753-08_varscan_annotated

Toolbar Quick Search:



TCGA-BRCA

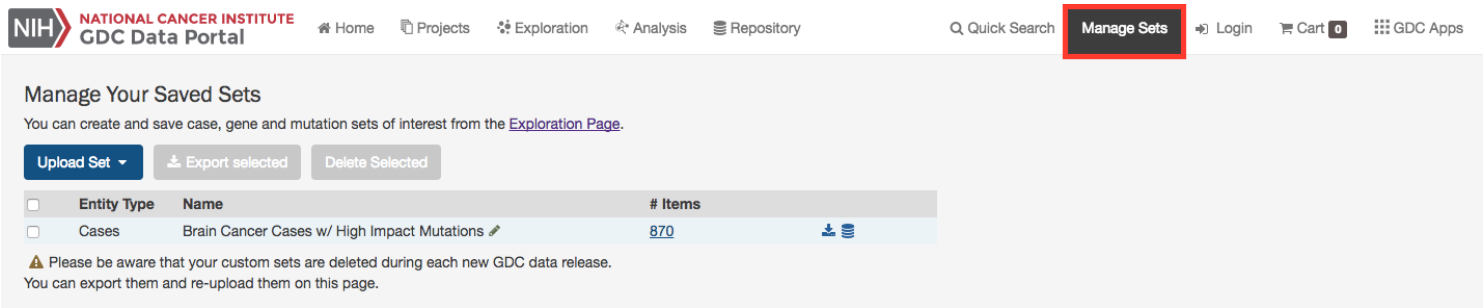
PR	TCGA-BRCA TCGA-BRCA
FL	6fcfff20-4993-4789-b27b-69d165130466 TCGA-BRCA-muse-public
FL	71f2cbda-32f4-481f-ad7e-faa0e1b5bc53 TCGA-BRCA-mutect-protected
FL	489ce525-6eb7-45e5-8acd-11fc1f5bc4ea TCGA-BRCA-somaticsniper-protected
FL	96983226-d92a-449d-8890-e1b210cee0fe TCGA-BRCA-mutect-public

Advanced Search

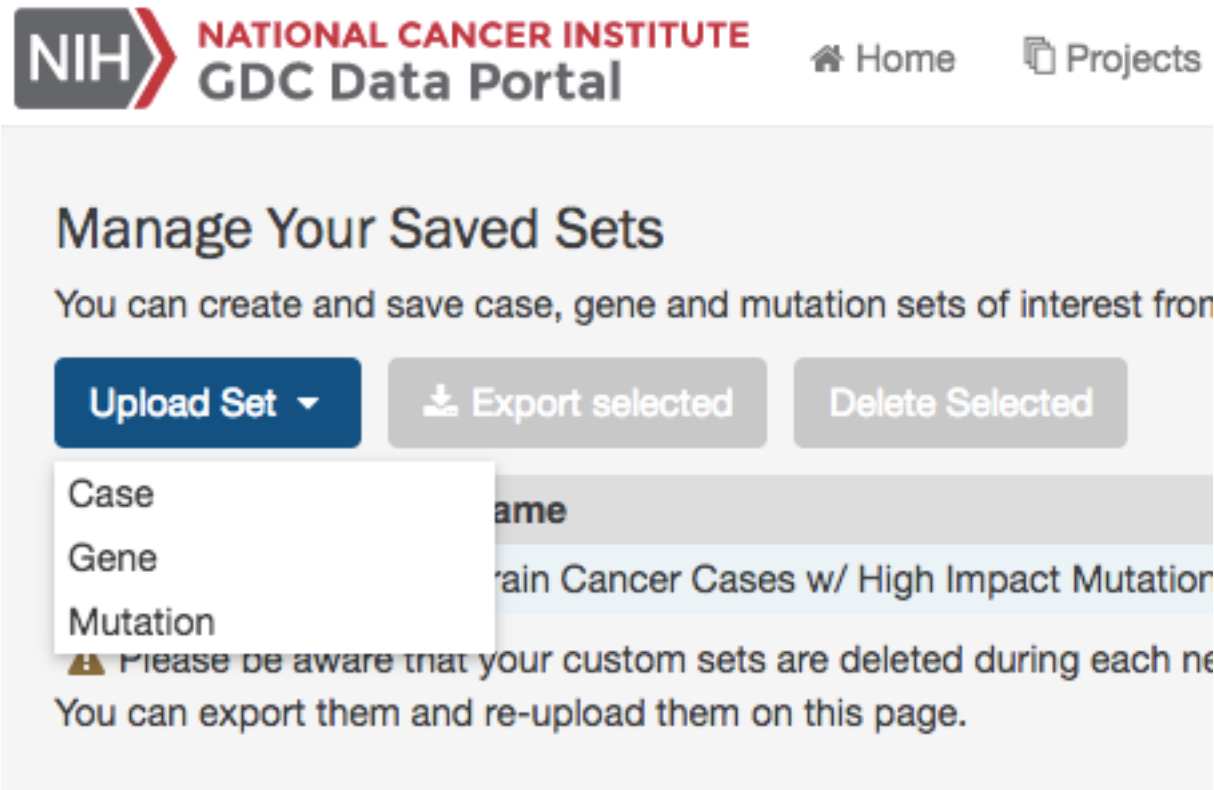
Advanced Search is available in Repository View. It allows users to construct complex queries with a custom query language and auto-complete suggestions. See Advanced Search for details.

Manage Sets

The **Manage Sets** button at the top of the GDC Portal stores sets of cases, genes, or mutations of interest. On this page, users can review the sets that have been saved as well as upload new sets and delete existing sets.



Upload Sets Clicking the Upload Set button shows options for creating Case, Gene, or Mutation sets.



Upon clicking one of the menu items, users are shown a dialog where they can enter unique identifiers (i.e. UUIDs, TCGA Barcodes, gene symbols, mutation UUIDs, etc.) that describe the set.

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GDC Data Portal

[Home](#)
[Projects](#)
[Exploration](#)
[Analysis](#)
[Repository](#)

[Quick Search](#)
[Manage Sets](#)
[Login](#)
[Cart 0](#)

Your Saved Sets

Create and save case, gene

Set

Export selected

Entity Type

Name

Cases

Brain Cancer

be aware that your custom

port them and re-upload

Upload Gene Set

Name:

Top Genes

Type or copy-and-paste a list of identifiers

IDH1,TP53,ATRX,PTEN,EGFR,CIC,PIK3CA,NF1

Or choose a file to upload

Browse

Summary Table (8 matched, 0 unmatched)

Matched (8)

Unmatched (0)

8 submitted gene identifiers mapped to 8 unique GDC genes

TSV

Submitted Gene Identifier		Mapped To	
Symbol	GDC Gene ID	Symbol	
IDH1	ENSG00000138413	IDH1	
TP53	ENSG00000141510	TP53	
ATRX	ENSG00000085224	ATRX	
PTEN	ENSG00000171862	PTEN	
EGFR	ENSG00000146648	EGFR	
CIC	ENSG00000079432	CIC	
PIK3CA	ENSG00000121879	PIK3CA	
NF1	ENSG00000196712	NF1	

Cancel

Clear

Submit

Clicking the Submit button will add the set of items to the list of sets on the Manage Sets page.

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[Projects](#)
[Exploration](#)
[Analysis](#)
[Repository](#)

Manage Your Saved Sets

You can create and save case, gene and mutation sets of interest from the [Exploration Page](#).

Upload Set

Export selected

Delete Selected

<input type="checkbox"/>	Entity Type	Name	# Items	
<input type="checkbox"/>	Cases	Brain Cancer Cases w/ High Impact Mutations	870	Download
<input type="checkbox"/>	Genes	Top Genes	8	Download

⚠ Please be aware that your custom sets are deleted during each new GDC data release.

You can export them and re-upload them on this page.

Export Sets Users can export selected sets on this page by first clicking the checkboxes next to each set, then clicking the Export selected button at the top of the table.

17

Manage Your Saved Sets

You can create and save case, gene and mutation sets of interest from the [Exploration Page](#).

<input type="checkbox"/>	Entity Type	Name	# Items	
<input checked="" type="checkbox"/>	Cases	Brain Cancer Cases w/ High Impact Mutations	870	
<input type="checkbox"/>	Genes	Top Genes	8	

⚠ Please be aware that your custom sets are deleted during each new GDC data release. You can export them and re-upload them on this page.

A text file containing the UUID of each case, gene or mutation is downloaded after clicking this button.

Review Sets There are a few buttons in the list of sets that allows a user to get further information about each one.

- ___ Items___: Clicking the link under the Items column navigates the user to the Exploration page using the set as a filter.
- Download/View:** To the right of the Items column are buttons that will download the list as a tsv or open the cases in the Repository page.

Creating Sets from GDC Portal Filters Many pages on the GDC Portal have an option called **Save Sets** that allows users to save a group of cases, mutations, or genes for further analysis. After using the filtering options on the **Exploration** page as an example, users can click the **Save Case/Gene/Mutation Set** button to save this set.

Cases Genes Mutations

~ Mutation

Q e.g. BRAF V600E, chr7:g.140753336A>T

Upload Mutation Set

~ Impact (VEP)

☐ MODERATE 9,943

☐ LOW 1,199

☒ HIGH 1,144

☐ MODIFIER 533

~ Consequence Type

☐ frameshift_variant 552

☐ stop_gained 427

☐ splice_acceptor_variant 84

☐ splice_donor_variant 88

☐ start_lost 8

1 More...

Clear Primary Site IS Brain AND Blototype IS protein_coding AND

Is Cancer Gene Census IS true AND Impact IS HIGH

View Files in Repository

Cases (582) Genes (287) Mutations (1,144) OncoGrid

Primary Site Project Disease Type Gender Vital Status

Showing 1 - 20 of 582 cases

Save/Edit Case Set

Case ID	Project	Primary Site	Gender	Files	Available Files per Data Category							# Mutations	# Genes
					Seq	Exp	SNV	CNV	Meth	Clinical	Bio		
TCGA-DU-6392	TCGA-LGG	Brain	Female	32	4	5	16	4	1	1	1	49	47
TCGA-06-5416	TCGA-GBM	Brain	Female	29	3	3	16	4	1	1	1	47	45

Chapter 2

Projects

Projects

Summary

At a high level, data in the Genomic Data Commons is organized by project. Typically, a project is a specific effort to look at particular type(s) of cancer undertaken as part of a larger cancer research program. The GDC Data Portal allows users to access aggregate project-level information via the Projects Page and Project Summary pages.

Projects Page

The Projects Page provides an overview of all harmonized data available in the Genomic Data Commons, organized by project. It also provides filtering, navigation, and advanced visualization features that allow users to identify and browse projects of interest. Users can access Projects Page from the GDC Data Portal Home page, from the Data Portal toolbar, or directly at <https://portal.gdc.cancer.gov/projects>.

On the left, a panel of facets allow users to apply filters to find projects of interest. When facet filters are applied, the table and visualizations on the right are updated to display only the matching projects. When no filters are applied, all projects are displayed.

The right side of this page displays a few visualizations of the data (Top Mutated Genes in Selected Projects and Case Distribution per Project). Below these graphs is a table that contains a list of projects and select details about each project, such as the number of cases and data files. The Graph tab provides a visual representation of this information.

Project

e.g. TCGA-GBM, Brain

Primary Site

- Kidney
- Adrenal Gland
- Brain
- Colorectal
- Lung

24 More...

Program

- TCGA
- TARGET

Disease Type

- Acute Myeloid Leukemia
- Adrenocortical Carcinoma
- Bladder Urothelial Carcinoma
- Brain Lower Grade Glioma
- Breast Invasive Carcinoma

33 More...

Data Category

- Biospecimen
- Clinical
- Raw Sequencing Data
- Transcriptome Profiling
- Simple Nucleotide Variation

2 More...

Experimental Strategies

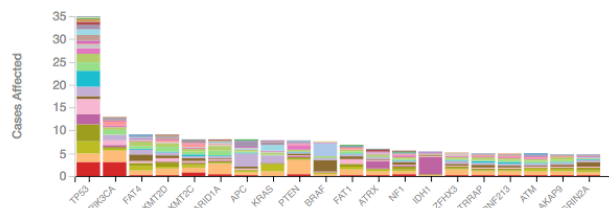
- RNA-Seq
- WXS
- miRNA-Seq

Start searching by selecting a facet

Top Mutated Cancer Genes in Selected Projects

10,188 Unique Cases with Somatic Mutation Data

% of Cases Affected # of Cases Affected



Case Distribution per Project

14,551 Cases across 39 Projects

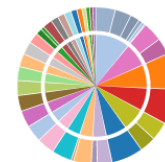


Table Graph

39 Projects

JSON TSV

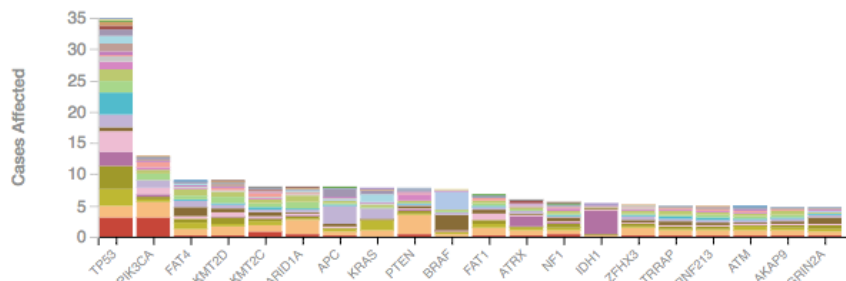
Project	Disease Type	Primary Site	Program	Cases	Available Cases per Data Category							Files
					Seq	Exp	SNV	CNV	Meth	Clinical	Bio	
TARGET-NBL	Neuroblastoma	Nervous System	TARGET	1,127	270	151	216	0	0	7	1,127	2,806
TCGA-BRCA	Breast Invasive Carcinoma	Breast	TCGA	1,098	1,098	1,097	1,044	1,096	1,095	1,097	1,098	27,207
TARGET-AML	Acute Myeloid Leukemia	Blood	TARGET	988	299	272	8	0	0	935	988	1,873
TARGET-WT	High-Risk Wilms Tumor	Kidney	TARGET	652	128	128	34	0	0	652	652	1,324
TCGA-GBM	Glioblastoma Multiforme	Brain	TCGA	617	406	166	396	593	423	596	617	9,657
TCGA-OV	Ovarian Serous Cystadenocarcinoma	Ovary	TCGA	608	575	492	443	573	602	587	608	13,054
TCGA-LUAD	Lung Adenocarcinoma	Lung	TCGA	585	582	519	569	518	579	522	585	14,804
TCGA-UCEC	Uterine Corpus Endometrial Carcinoma	Uterus	TCGA	560	559	559	542	547	559	548	560	13,604
TCGA-KIRC	Kidney Renal Clear Cell Carcinoma	Kidney	TCGA	537	535	534	339	532	533	537	537	12,272
TCGA-HNSC	Head and Neck Squamous Cell Carcinoma	Head and Neck	TCGA	528	528	528	510	521	528	528	528	12,895
TCGA-LGG	Brain Lower Grade Glioma	Brain	TCGA	516	516	516	513	514	516	515	516	12,603
TCGA-THCA	Thyroid Carcinoma	Thyroid	TCGA	507	507	507	496	505	507	507	507	12,703
TCGA-LUSC	Lung Squamous Cell Carcinoma	Lung	TCGA	504	504	504	497	504	503	504	504	13,124
TCGA-PRAD	Prostate Adenocarcinoma	Prostate	TCGA	500	498	498	498	498	498	500	500	12,568
TCGA-SKCM	Skin Cutaneous Melanoma	Skin	TCGA	470	470	469	470	470	470	470	470	11,265
TCGA-COAD	Colon Adenocarcinoma	Colorectal	TCGA	461	460	459	433	458	458	459	461	11,824
TCGA-STAD	Stomach Adenocarcinoma	Stomach	TCGA	443	443	439	441	443	443	443	443	10,731
TCGA-BLCA	Bladder Urothelial Carcinoma	Bladder	TCGA	412	412	412	412	412	412	412	412	10,193
TARGET-OS	Osteosarcoma	Bone	TARGET	381	0	0	0	0	0	282	381	4

Visualizations

Top Mutated Cancer Genes in Selected Projects

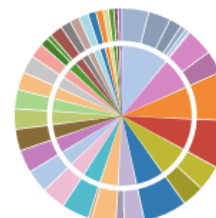
10,188 Unique Cases with Somatic Mutation Data

% of Cases Affected # of Cases Affected



Case Distribution per Project

14,551 Cases across 39 Projects



Top Mutated Cancer Genes in Selected Projects

This dynamically generated bar graph shows the 20 genes with the most mutations across all projects. The genes are filtered by those that are part of the Cancer Gene Census and that have the following types of mutations: **missense_variant**, **frameshift_variant**, **start_lost**, **stop_lost**, **initiator_codon_variant**, and **stop_gained**. The bars represent the frequency of each mutation and is broken down into different colored segments by project and disease type. The graphic is updated as filters are applied for projects, programs, disease types, and data categories available in the project. Note, that due to these filters the number of cases displayed here will be less than the total number of cases per project.

Hovering the cursor over each bar will display information about the number of cases affected by the disease type and clicking on each bar will launch the Gene Summary Page for the gene associated with the mutation.

Users can toggle the Y-Axis of this bar graph between a percentage or raw number of cases affected.

Case Distribution per Project

A pie chart displays the relative number of cases for each primary site (inner circle), which is further divided by project (outer circle). Hovering the cursor over each portion of the graph will display the primary site or project with the number of associated cases. Filtering projects at the left panel will update the pie chart.

Projects Table

The **Table** tab lists projects by Project ID and provides additional information about each project. If no facet filters have been applied, the table will display all available projects; otherwise it will display only those projects that match the selected criteria.

Table

Graph

39 Projects

JSON

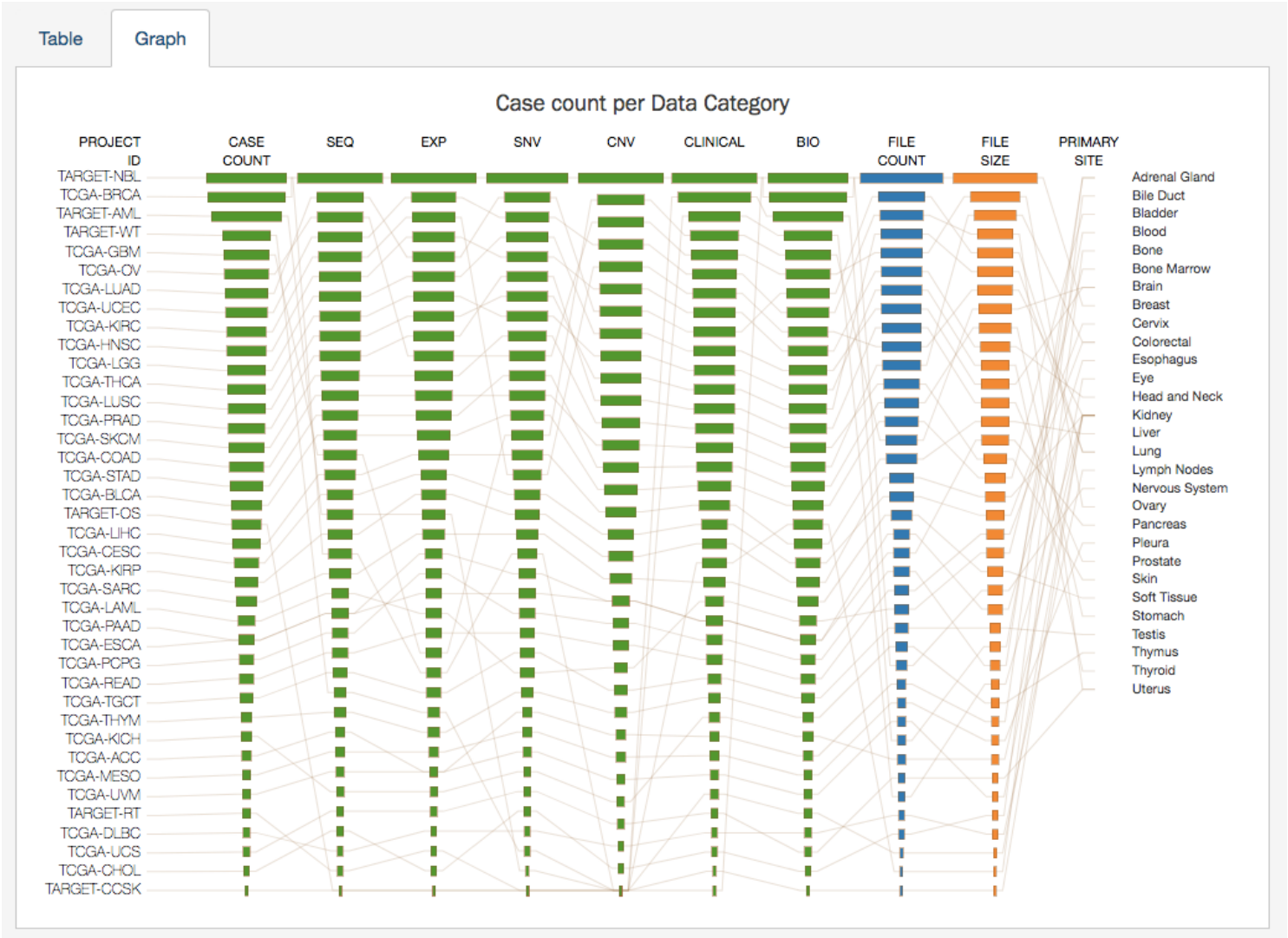
TSV

Project ID	Disease Type	Primary Site	Program	Cases	Available Cases per Data Category							Files
					Seq	Exp	SNV	CNV	Meth	Clinical	Bio	
TARGET-NBL	Neuroblastoma	Nervous System	TARGET	1,127	270	151	216	0	0	7	1,127	2,806
TCGA-BRCA	Breast Invasive Carcinoma	Breast	TCGA	1,098	1,098	1,097	1,044	1,096	1,095	1,097	1,098	27,207
TARGET-AML	Acute Myeloid Leukemia	Blood	TARGET	988	299	272	8	0	0	935	988	1,873
TARGET-WT	High-Risk Wilms Tumor	Kidney	TARGET	652	128	128	34	0	0	652	652	1,324
TCGA-GBM	Glioblastoma Multiforme	Brain	TCGA	617	406	166	396	593	423	596	617	9,657
TCGA-OV	Ovarian Serous Cystadenocarcinoma	Ovary	TCGA	608	575	492	443	573	602	587	608	13,054
TCGA-LUAD	Lung Adenocarcinoma	Lung	TCGA	585	582	519	569	518	579	522	585	14,804
TCGA-UCEC	Uterine Corpus Endometrial Carcinoma	Uterus	TCGA	560	559	559	542	547	559	548	560	13,604
TCGA-KIRC	Kidney Renal Clear Cell Carcinoma	Kidney	TCGA	537	535	534	339	532	533	537	537	12,272
TCGA-HNSC	Head and Neck Squamous Cell Carcinoma	Head and Neck	TCGA	528	528	528	510	521	528	528	528	12,895
TCGA-LGG	Brain Lower Grade Glioma	Brain	TCGA	516	516	516	513	514	516	515	516	12,603
TCGA-THCA	Thyroid Carcinoma	Thyroid	TCGA	507	507	507	496	505	507	507	507	12,703
TCGA-LUSC	Lung Squamous Cell Carcinoma	Lung	TCGA	504	504	504	497	504	503	504	504	13,124
TCGA-PRAD	Prostate Adenocarcinoma	Prostate	TCGA	500	498	498	498	498	498	500	500	12,568
TCGA-SKCM	Skin Cutaneous Melanoma	Skin	TCGA	470	470	469	470	470	470	470	470	11,265
TCGA-COAD	Colon Adenocarcinoma	Colorectal	TCGA	461	460	459	433	458	458	459	461	11,824
TCGA-STAD	Stomach Adenocarcinoma	Stomach	TCGA	443	443	439	441	443	443	443	443	10,731
TCGA-BLCA	Bladder Urothelial Carcinoma	Bladder	TCGA	412	412	412	412	412	412	412	412	10,193
TARGET-OS	Osteosarcoma	Bone	TARGET	381	0	0	0	0	0	282	381	4
TCGA-LIHC	Liver Hepatocellular Carcinoma	Liver	TCGA	377	377	376	375	376	377	377	377	9,511
TCGA-CESC	Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma	Cervix	TCGA	307	307	307	305	302	307	307	307	7,349

The table provides links to Project Summary pages in the Project ID column. Columns with file and case counts include links to open the corresponding files or cases in Repository Page.

Projects Graph

The **Graph** tab contains an interactive view of information in the Table tab. The numerical values in Case Count, File Count, and File Size columns are represented by bars of varying length according to size. These columns are sorted independently in descending order. Mousing over an element of the graph connects it to associated elements in other columns, including Project ID and Primary Site



Most elements in the graph are clickable, allowing the user to open the associated cases or files in Repository Page. Like the projects table, the graph will reflect any applied facet filters.

Facets Panel

Facets represent properties of the data that can be used for filtering. The facets panel on the left allows users to filter the projects presented in the Table and Graph tabs as well as visualizations.

NATIONAL CANCER INSTITUTE
GDC Data Portal

[Home](#)
[Projects](#)
[Exploration](#)
[Analysis](#)
[Repository](#)

[Quick Search](#)
[Manage Sets](#)
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[Cart](#)
[GDC Apps](#)

Project

Primary Site

☐ Kidney
 ☐ Adrenal Gland
 ☐ Brain
 ☐ Colorectal
 ☐ Lung

24 More...

Program

☐ TCGA
 ☐ TARGET

93 6

Disease Type

☐ Acute Myeloid Leukemia
 ☐ Adrenocortical Carcinoma
 ☐ Bladder Urothelial Carcinoma
 ☐ Brain Lower Grade Glioma
 ☐ Breast Invasive Carcinoma

33 More...

Data Category

☐ Biospecimen
 ☐ Clinical
 ☐ Raw Sequencing Data
 ☐ Transcriptome Profiling
 ☐ Simple Nucleotide Variation

2 More...

Experimental Strategies

☐ RNA-Seq
 ☐ WXS
 ☐ miRNA-Seq
 ☐ Genotyping Array
 ☐ Methylation Array

97 96 96 93 93

Start searching by selecting a facet

Top Mutated Cancer Genes in Selected Projects

10,188 Unique Cases with Somatic Mutation Data

☒ % of Cases Affected
 ☐ # of Cases Affected

Case Distribution per Project

14,551 Cases across 39 Projects

Table

Graph

39 Projects

≡

⌵

JSON

TSV

Project	Disease Type	Primary Site	Program	Cases	Available Cases per Data Category							Files
					Seq	Exp	SNV	CNV	Meth	Clinical	Bio	
TARGET-NBL	Neuroblastoma	Nervous System	TARGET	1,127	270	151	216	0	0	7	1,127	2,806
TCGA-BRCA	Breast Invasive Carcinoma	Breast	TCGA	1,098	1,098	1,097	1,044	1,096	1,095	1,097	1,098	27,207
TARGET-AML	Acute Myeloid Leukemia	Blood	TARGET	988	299	272	8	0	0	935	988	1,873
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TCGA-GBM	Glioblastoma Multiforme	Brain	TCGA	617	406	166	396	593	423	596	617	9,657
TCGA-OV	Ovarian Serous Cystadenocarcinoma	Ovary	TCGA	608	575	492	443	573	602	587	608	13,054
TCGA-LUAD	Lung Adenocarcinoma	Lung	TCGA	585	582	519	569	518	579	522	585	14,804
TCGA-UCEC	Uterine Corpus Endometrial Carcinoma	Uterus	TCGA	560	559	559	542	547	559	548	560	13,604
TCGA-KIRC	Kidney Renal Clear Cell Carcinoma	Kidney	TCGA	537	535	534	339	532	533	537	537	12,272
TCGA-HNSC	Head and Neck Squamous Cell Carcinoma	Head and Neck	TCGA	528	528	528	510	521	528	528	528	12,895
TCGA-LGG	Brain Lower Grade Glioma	Brain	TCGA	516	516	516	513	514	516	515	516	12,603
TCGA-THCA	Thyroid Carcinoma	Thyroid	TCGA	507	507	507	496	505	507	507	507	12,703
TCGA-LUSC	Lung Squamous Cell Carcinoma	Lung	TCGA	504	504	504	497	504	503	504	504	13,124
TCGA-PRAD	Prostate Adenocarcinoma	Prostate	TCGA	500	498	498	498	498	498	500	500	12,568
TCGA-SKCM	Skin Cutaneous Melanoma	Skin	TCGA	470	470	469	470	470	470	470	470	11,265
TCGA-COAD	Colon Adenocarcinoma	Colorectal	TCGA	461	460	459	433	458	458	459	461	11,824
TCGA-STAD	Stomach Adenocarcinoma	Stomach	TCGA	443	443	439	441	443	443	443	443	10,731
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TCGA-LIHC	Liver Hepatocellular Carcinoma	Liver	TCGA	377	377	376	375	376	377	377	377	9,511
TCGA-CESC	Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma	Cervix	TCGA	307	307	307	305	302	307	307	307	7,349
TCGA-KIRP	Kidney Renal Papillary Cell Carcinoma	Kidney	TCGA	291	291	291	288	290	291	291	291	7,368

Users can filter by the following facets:

- **Project:** Individual project ID
- **Primary Site:** Anatomical site of the cancer under investigation or review
- **Program:** Research program that the project is part of
- **Disease Type:** Type of cancer studied
- **Data Category:** Type of data available in the project
- **Experimental Strategy:** Experimental strategies used for molecular characterization of the cancer

Filters can be applied by selecting values of interest in the available facets, for example “WXS” and “RNA-Seq” in the “Experimental Strategy” facet and “Brain” in the “Primary Site” facet. When facet filters are applied, the Table and Graph tabs are updated to display matching projects, and the banner above the tabs summarizes the applied filters. The banner allows the user to click on filter elements to remove the associated filters, and includes a link to view the matching cases and files.

23

Clear

Primary Site

IS

Brain

AND

Experimental Strategy

IN

(

RNA-Seq

WXS

)

Open Query In Data Page

For information on how to use facet filters, see [Getting Started](#).

Project Summary Page

Each project has a summary page that provides an overview of all available cases, files, and annotations available. Clicking on the numbers in the summary table will display the corresponding data.

PR TCGA-BRCA

Download Biospecimen

Download Clinical

Download Manifest

Summary

Project ID	TCGA-BRCA
Project Name	Breast Invasive Carcinoma
Disease Type	Breast Invasive Carcinoma
Primary Site	Breast
Program	TCGA

CASES

1,098

FILES

27,207

ANNOTATIONS

78

Cases and File Counts by Experimental Strategy

Experimental Strategy	Cases	Files
Genotyping Array	1,096	4,446
Methylation Array	1,095	1,234
WXS	1,050	10,823
RNA-Seq	1,092	4,888
miRNA-Seq	1,079	3,621

Cases and File Counts by Data Category

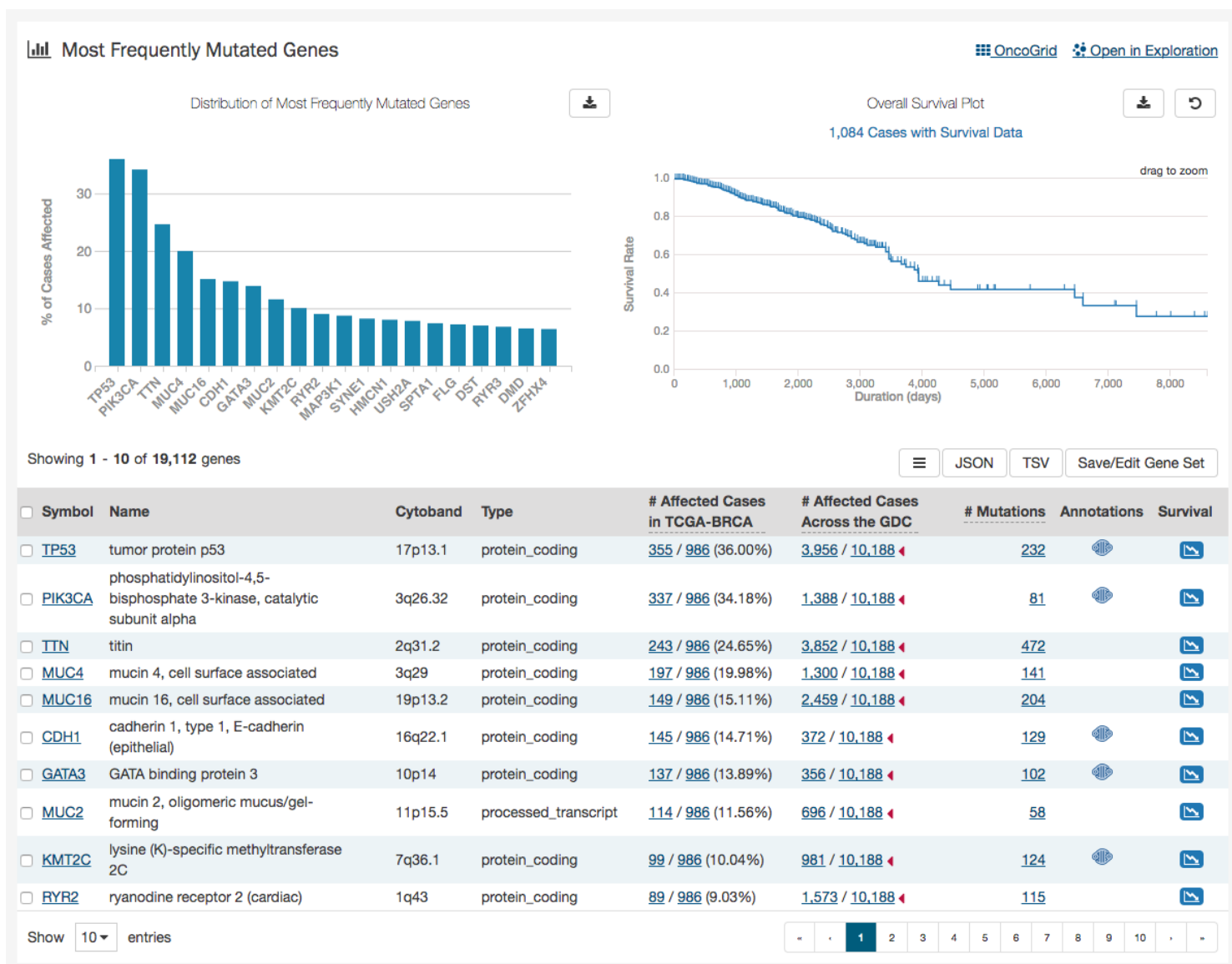
Data Category	Cases	Files
Raw Sequencing Data	1,098	4,604
Transcriptome Profiling	1,097	6,080
Simple Nucleotide Variation	1,044	8,648
Copy Number Variation	1,096	4,446
DNA Methylation	1,095	1,234
Clinical	1,097	1,097
Biospecimen	1,098	1,098

Three download buttons in the top right corner of the screen allow the user to download the entire project dataset, along with the associated project metadata:

- Download Biospecimen:** Downloads biospecimen metadata associated with all cases in the project.
- Download Clinical:** Downloads clinical metadata about all cases in the project.
- Download Manifest:** Downloads a manifest for all data files available in the project. The manifest can be used with the GDC Data Transfer Tool to download the files.

Most Frequently Mutated Genes

The Project Summary page also reports the genes that have somatic mutations in the greatest numbers of cases in a graphical and tabular format.



The top of this section contains a bar graph of the most frequently mutated genes as well as a survival plot of all the cases within the specified project. Hovering over each bar in the plot will display information about the number of cases affected. Users may choose to download the underlying data in JSON or TSV format or an image of the graph in SVG or PNG format by clicking the download icon at the top of each graph.

Also at the top of this section are two links: **OncoGrid** and **Open in Exploration**. The **OncoGrid** button will take the user to the OncoGrid. **Open in Exploration** will take the user to the Exploration page with this filters applied for the current project selected.

Below these graphs is a tabular view of the genes affected, which includes the following information:

- **Symbol:** The gene symbol, which links to the Gene Summary Page
- **Name:** Full name of the gene
- **Cytoband:** The location of the mutation on the chromosome in terms of Giemsa-stained samples.
- **___ Affected Cases in Project:**___ The number of cases within the project that contain a mutation on this gene, which links to the Cases tab in the Exploration Page
- **___ Affected Cases Across the GDC:**___ The number of cases within all the projects in the GDC that contain a mutation on this gene. Clicking the red arrow will display the cases broken down by project
- **___ Mutations:**___ The number of SSMs (simple somatic mutations) detected in that gene, which links to the Mutation tab in the Exploration Page
- **Annotations:** Includes a COSMIC symbol if the gene belongs to [The Cancer Gene Census](#)

- **Survival Analysis:** An icon that, when clicked, will plot the survival rate between cases in the project with mutated and non-mutated forms of the gene

Survival Analysis

Survival analysis is used to analyze the occurrence of event data over time. In the GDC, survival analysis is performed on the mortality of the cases. Survival analysis requires:

- Data on the time to a particular event (days to death or last follow up)
 - Fields: **diagnoses.days_to_death** and **diagnoses.days_to_last_follow_up**
- Information on whether the event has occurred (alive/deceased)
 - Fields: **diagnoses.vital_status**
- Data split into different categories or groups (i.e. gender, etc.)
 - Fields: **demographic.gender**

The survival analysis in the GDC uses a Kaplan-Meier estimator:

$$S(t_i) = \prod_{t_j \leq t} \left(1 - \frac{d_j}{n_j}\right)$$

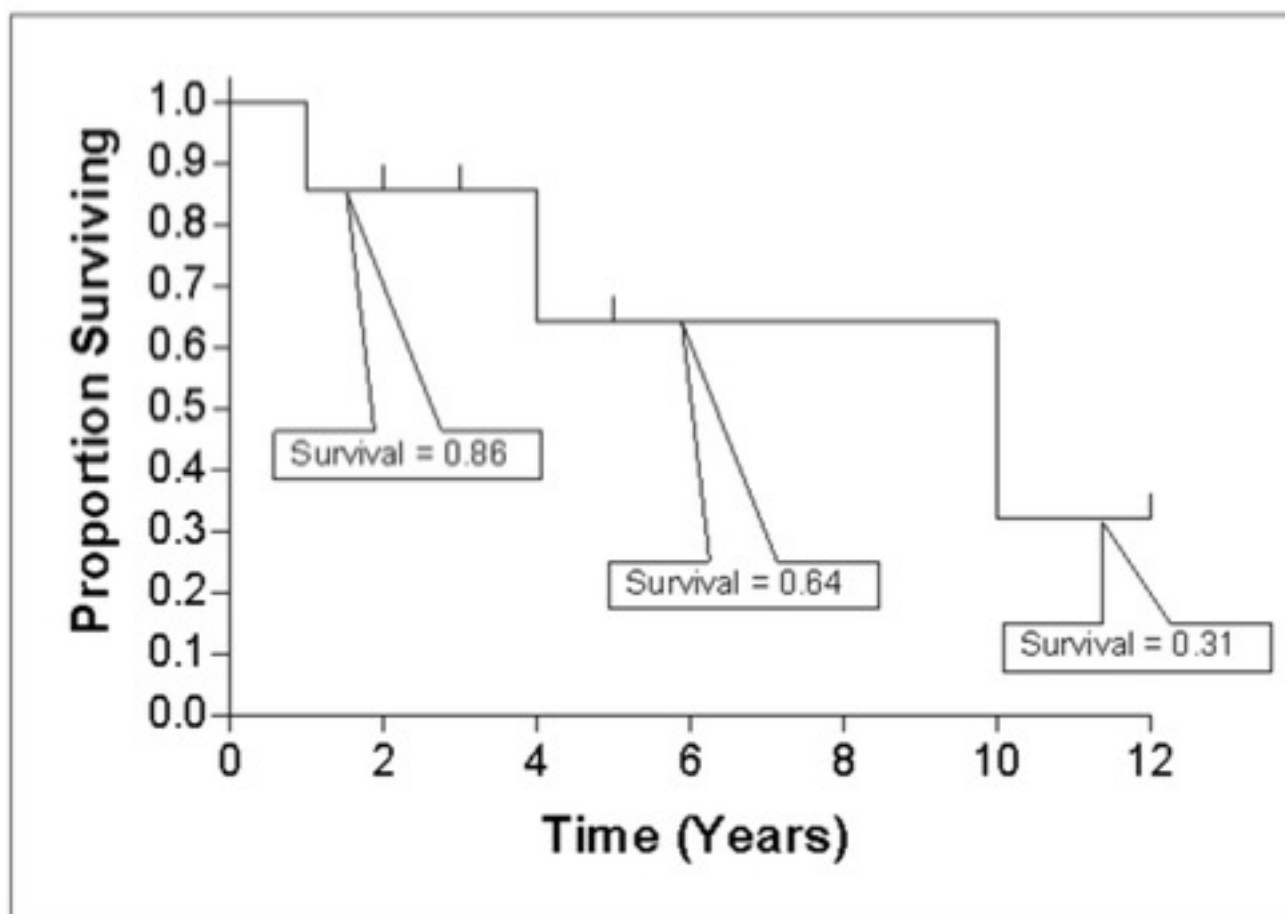
Where:

- S(t_i) is the estimated survival probability for any particular one of the t time periods
- n_i is the number of subjects at risk at the beginning of time period t_i
- and d_i is the number of subjects who die during time period t_i

The table below is an example data set to calculate survival for a set of seven cases:

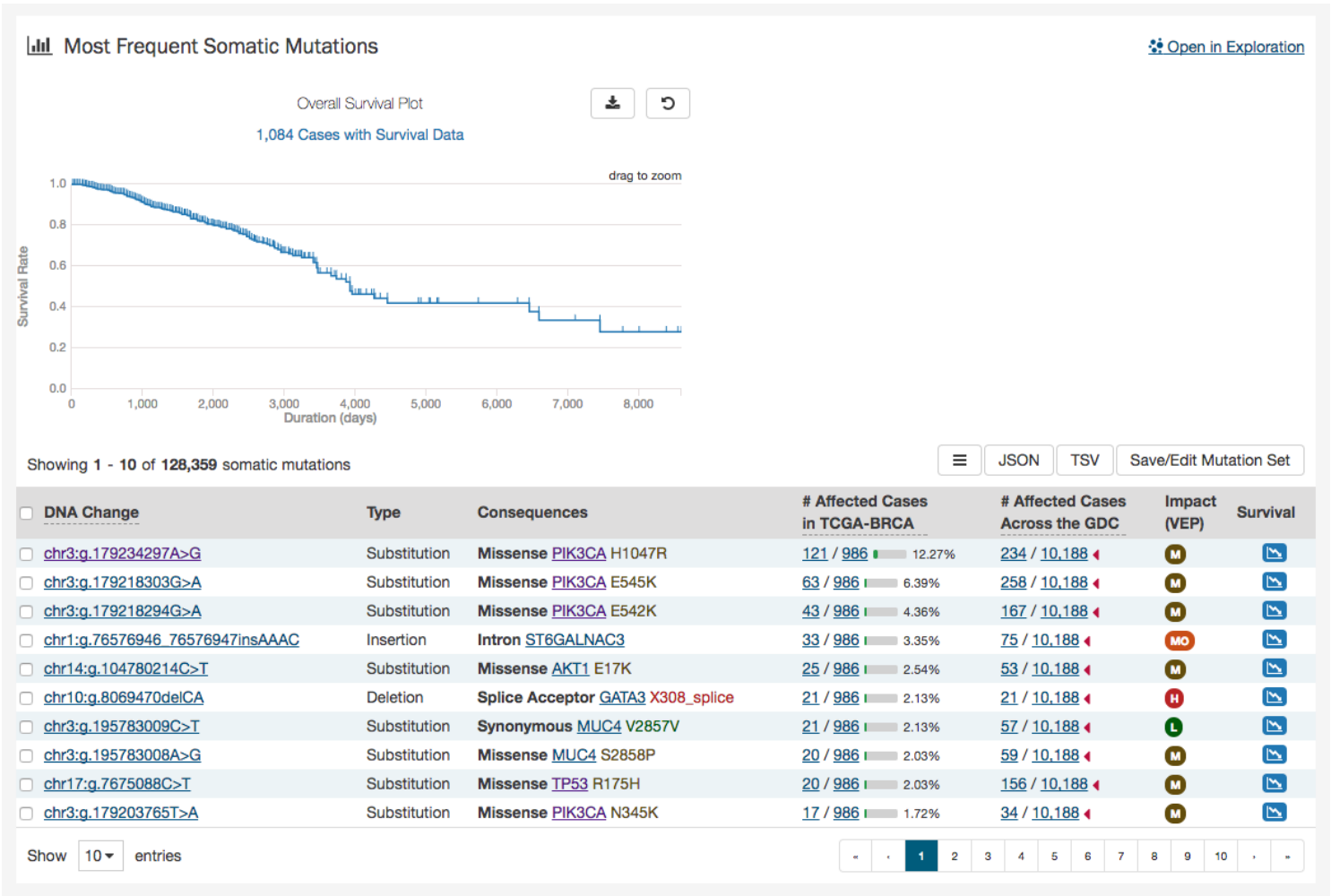
overall_survival_time (Years)	interval		# of donors at risk at start of interval (r)	# of censored donors during interval (c)	# of donors at risk at end of interval (n=r-c)	# of donors died at end of interval (d)	estimated interval survival probability ((n-d)/n)	estimated cumulative survival probability at end of interval (S)
	start	end						
0	0							1
1	0	1	7	0	7	1	(7-1)/7 = 0.86	1 * 0.86 = 0.86
4	1	4	6	2	4	1	(4-1)/4 = 0.75	0.86 * 0.75 = 0.64
10	4	10	3	1	2	1	(2-1)/2 = 0.5	0.86 * 0.75 * 0.5 = 0.31
>12	10	12	1	0	1	0	(1-0)/1 = 1.0	0.86 * 0.75 * 0.5 * 1.0 = 0.31

The calculated cumulated survival probability can be plotted against the interval to obtain a survival plot like the one shown below.



Most Frequent Mutations

At the top of this section is a survival plot of all the cases within the specified exploration page filters.

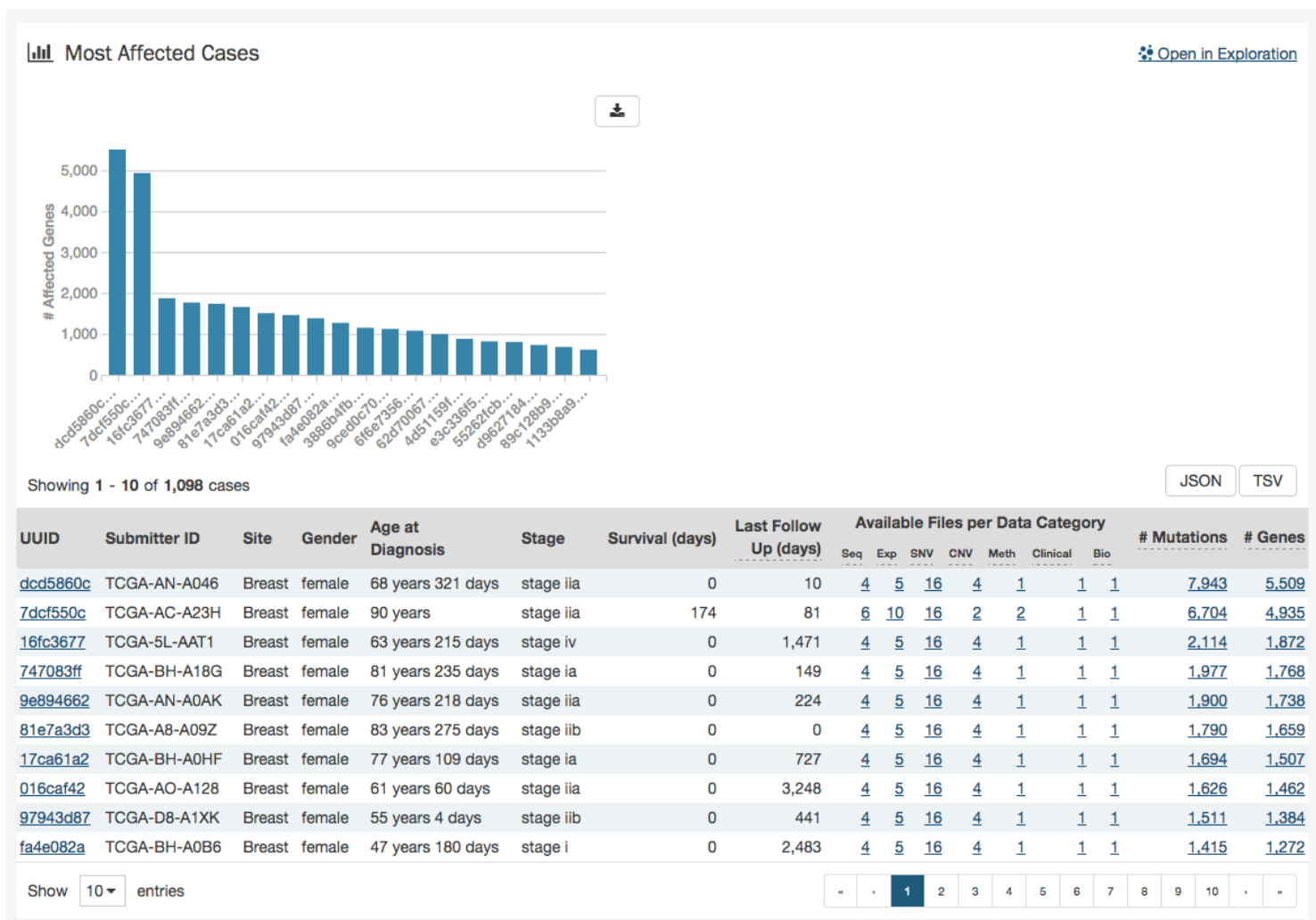


A table is displayed below that lists information about each mutation:

- **Mutation ID:** A UUID for the mutation assigned by the GDC, when clicked will bring a user to the Mutation Summary Page
- **DNA Change:** The chromosome and starting coordinates of the mutation are displayed along with the nucleotide differences between the reference and tumor allele
- **Type:** A general classification of the mutation
- **Consequences:** The effects the mutation has on the gene coding for a protein (i.e. synonymous, missense, non-coding transcript). A link to the Gene Summary Page for the gene affected by the mutation is included
- **___ Affected Cases in Project:**___ The number of affected cases in the project expressed as a fraction and percentage
- **___ Affected Cases in Across the GDC:**___ The number of affected cases, expressed as number across all projects. Choosing the arrow next to the percentage will display a breakdown of each affected project
- **Impact:** A subjective classification of the severity of the variant consequence. This determined using [Ensembl VEP](#), [PolyPhen](#), and [SIFT](#). The categories are outlined [here](#).
- **Survival Analysis:** An icon that when clicked, will plot the survival rate between the gene's mutated and non-mutated cases

Most Affected Cases

The final section of the Project Summary page is a display of the top 20 cases in a specified project, with the greatest number of affected genes.



Below the bar graph is a table contains information about these cases:

- **UUID:** The UUID of the case, which links to the Case Summary Page
- **Submitter ID:** The Submitter ID of the case (i.e. the TCGA Barcode)
- **Site:** The anatomical location of the site affected
- **Gender:** Text designations that identify gender. Gender is described as the assemblage of properties that distinguish people on the basis of their societal roles
- **Age at Diagnosis:** Age at the time of diagnosis expressed in number of days since birth
- **Stage:** The extent of a cancer in the body. Staging is usually based on the size of the tumor, whether lymph nodes contain cancer, and whether the cancer has spread from the original site to other parts of the body. The accepted values for tumor_stage depend on the tumor site, type, and accepted staging system
- **Survival (days):** The number of days until death
- **Last Follow Up (days):** Time interval from the date of last follow up to the date of initial pathologic diagnosis, represented as a calculated number of days
- **Available Files per Data Category:** Five columns displaying the number of files available in each of the five data categories. These link to the files for the specific case.
- ___ Mutations:___ The number of mutations for the case
- ___ Genes:___ The number of genes affected by mutations for the case

Chapter 3

Exploration

Exploration

The Exploration page allows users to explore data in the GDC using advanced filters/facets, which includes those on a gene and mutation level. Users choose filters on specific **Cases**, **Genes**, and/or **Mutations** on the left of this page and then can visualize these results on the right. The Gene/Mutation data for these visualizations comes from the Open-Access MAF files on the GDC Portal.

NIH NATIONAL CANCER INSTITUTE GDC Data Portal

Home Projects Exploration Analysis Repository

Quick Search Manage Sets Login Cart 0 GDC Apps

Cases Genes Mutations

Add a Case Filter

Case

Q e.g. TCGA-A5-A0G2, 432fe4a9-2...

Upload Case Set

Primary Site

Kidney 1,861

Brain 1,193

Nervous System 1,127

Breast 1,090

Lung 1,089

24 More...

Program

TCGA 11,515

TARGET 3,536

Project

TARGET-NBL 1,127

TCGA-BRCA 1,098

TARGET-AML 988

TARGET-WT 862

TCGA-GBM 817

34 More...

Start searching by selecting a facet

View Files in Repository

Cases (14,551) Genes (22,144) Mutations (3,115,606) OncoGrid

Primary Site Project Disease Type Gender Vital Status

Showing 1 - 10 of 14,551 cases

JSON TSV Save/Edit Case Set

Case ID	Project	Primary Site	Gender	Files	Available Files per Data Category							# Mutations	# Genes
					Seq	Exp	SNV	CNV	Meth	Clinical	Bio		
<input type="checkbox"/> TCGA-A5-A0G2	TCGA-UCEC	Uterus	Female	32	4	5	16	4	1	1	1	41,966	14,347
<input type="checkbox"/> TCGA-E0-A22U	TCGA-UCEC	Uterus	Female	32	4	5	16	4	1	1	1	26,792	12,600
<input type="checkbox"/> TCGA-FI-A2D5	TCGA-UCEC	Uterus	Female	32	4	5	16	4	1	1	1	26,064	12,467
<input type="checkbox"/> TCGA-AX-A2HC	TCGA-UCEC	Uterus	Female	40	6	10	16	4	2	1	1	24,665	12,161
<input type="checkbox"/> TCGA-E0-A22R	TCGA-UCEC	Uterus	Female	33	4	5	16	4	2	1	1	24,186	11,910
<input type="checkbox"/> TCGA-B5-A3FC	TCGA-UCEC	Uterus	Female	32	4	5	16	4	1	1	1	23,984	11,793
<input type="checkbox"/> TCGA-IB-7651	TCGA-PAAD	Pancreas	Female	32	4	5	16	4	1	1	1	23,071	11,450
<input type="checkbox"/> TCGA-AP-A1DV	TCGA-UCEC	Uterus	Female	30	4	5	16	2	1	1	1	22,619	11,420
<input type="checkbox"/> TCGA-E6-A1LX	TCGA-UCEC	Uterus	Female	32	4	5	16	4	1	1	1	23,473	11,384
<input type="checkbox"/> TCGA-AP-A0LM	TCGA-UCEC	Uterus	Female	32	4	5	16	4	1	1	1	22,479	11,297

Show 10 entries

1 2 3 4 5 6 7 8 9 10

Filters / Facets

On the left of this page, users can create advanced filters to narrow down results to create synthetic cohorts.

30

Case Filters

The first tab of filters is for cases in the GDC.

Cases

Genes

Mutations



[Add a Case Filter](#)

▼ Case



e.g. TCGA-A5-A0G2, 432fe4a9-2...

Upload Case Set



▼ Primary Site



☐ Kidney

1,681

☐ Brain

1,133

☐ Nervous System

1,127

☐ Breast

1,098

☐ Lung

1,089

24 More...

▼ Program

☐ TCGA

11,315

☐ TARGET

3,236

▼ Project



☐ TARGET-NBL

1,127

☐ TCGA-BRCA

1,098

☐ TARGET-AML

988

☐ TARGET-WT

652

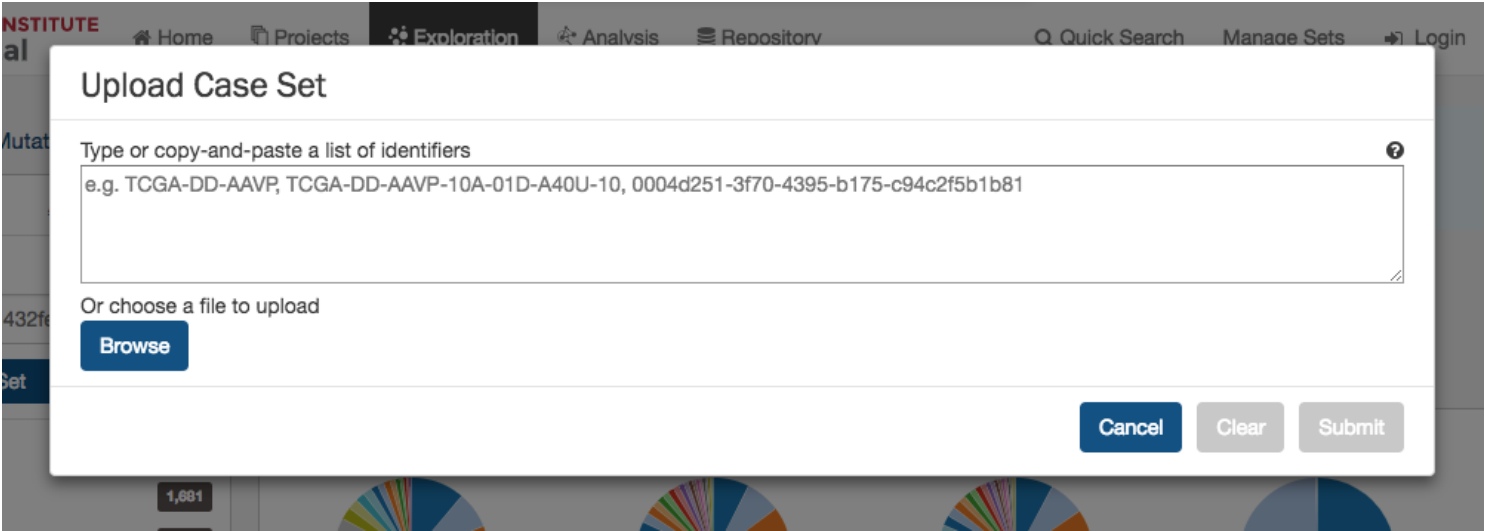
These criteria limit the results only to specific cases within the GDC. The default filters available are:

- **Case:** Specify individual cases using submitter ID (barcode), UUID, or list of Cases ('Case Set')
- **Case Submitter ID:** Search for cases using a part (prefix) of the submitter ID (barcode).
- **Primary Site:** Anatomical site of the cancer under investigation or review.
- **Program:** A cancer research program, typically consisting of multiple focused projects.
- **Project:** A cancer research project, typically part of a larger cancer research program.
- **Disease Type:** Type of cancer studied.
- **Gender:** Gender of the patient.
- **Age at Diagnosis:** Patient age at the time of diagnosis.
- **Vital Status:** Indicator of whether the patient was living or deceased at the date of last contact.
- **Days to Death:** Number of days from date of diagnosis to death of the patient.
- **Race:** Race of the patient.
- **Ethnicity:** Ethnicity of the patient.

In addition to the defaults, users can add additional case filters by clicking on the link titled 'Add a Case Filter'

Upload Case Set

In the **Cases** filters panel, instead of supplying cases one-by-one, users can supply a list of cases. Clicking on the **Upload Case Set** button will launch a dialog as shown below, where users can supply a list of cases or upload a comma-separated text file of cases.



After supplying a list of cases, a table below will appear which indicates whether the case was found.

STITUTE

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Upload Case Set

Type or copy-and-paste a list of identifiers

TCGA-A5-A0G2 TCGA-AX-A2HC TCGA-IB-7651 e5bc45ce-8a14-40b5-b9b5-ce45609fef3a

Or choose a file to upload

Browse

Summary Table (4 matched, 0 unmatched) ▼

Matched (4)

Unmatched (0)

4 submitted case identifiers mapped to 4 unique GDC cases TSV

Submitted Case Identifier		Mapped To	
Case UUID	Case ID	Case ID	Project
--	TCGA-A5-A0G2	TCGA-A5-A0G2	TCGA-UCEC
--	TCGA-AX-A2HC	TCGA-AX-A2HC	TCGA-UCEC
--	TCGA-IB-7651	TCGA-IB-7651	TCGA-PAAD
e5bc45ce-8a14-40b5-b9b5-ce45609fef3a	--	TCGA-FW-A3R5	TCGA-SKCM

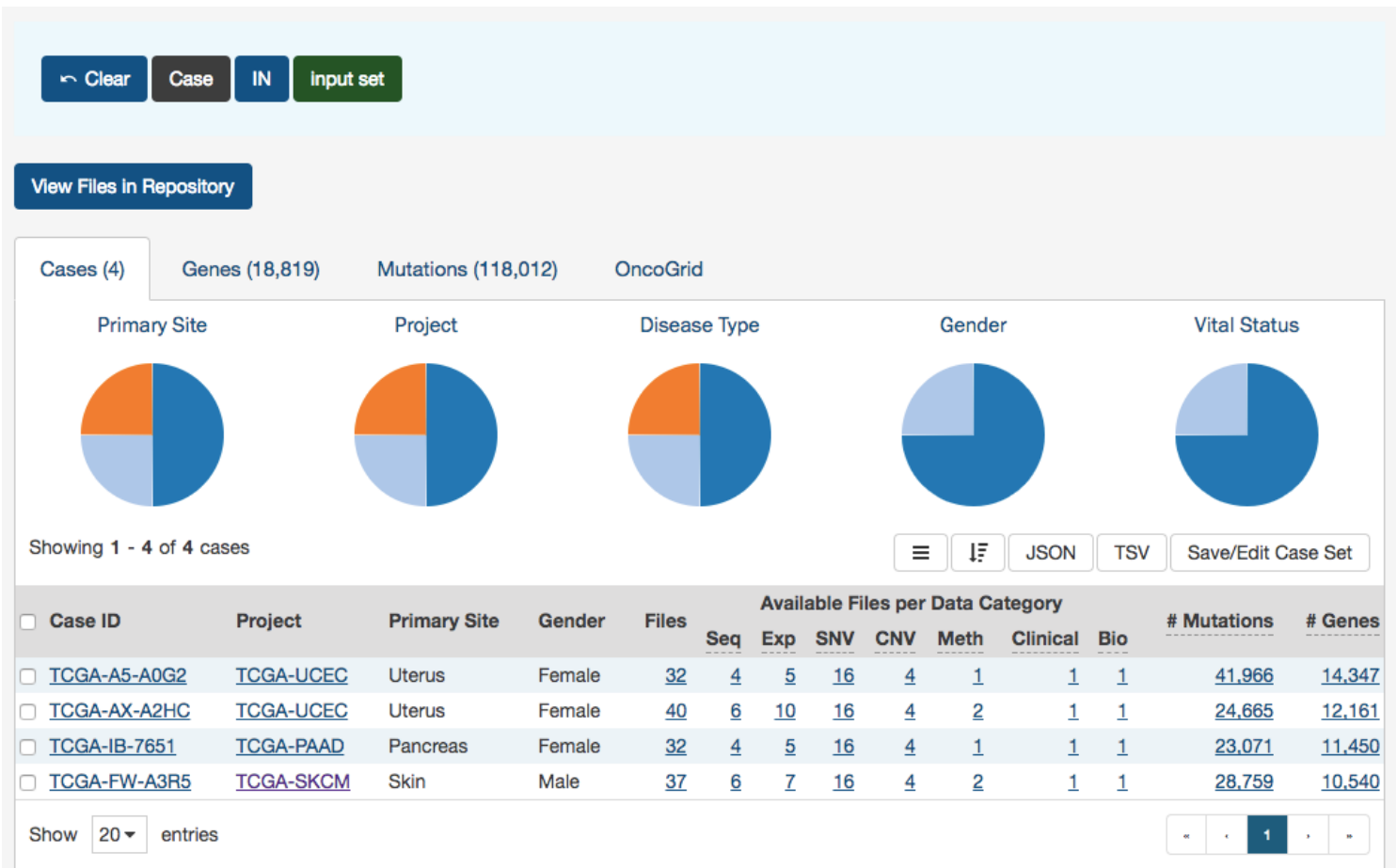
Cancel

Clear

Submit

☐ 53e8e07e-18f9-4c9b-9783-a5b67ceddcf7 [TCGA-AX-A2HC](#) [TCGA-UCEC](#) Uterus Female 40 6 10 16 4

Clicking on **Submit** will filter the results in the Exploration Page by those cases.



Gene Filters

The second tab of filters is for genes affected by mutations in the GDC.

Cases
Genes
Mutations
«

Gene
?

Q
e.g. BRAF, ENSG00000157764

Upload Gene Set

Biotype

☐ protein_coding 19,031
☐ lincRNA 731
☐ miRNA 407
☐ transcribed_unprocessed_pseudogene 363
☐ processed_pseudogene 326
27 More...

Is Cancer Gene Census

☐ true 575

The second tab of filters are for specific genes. Users can filter by:

- **Gene** - Entering in a specific Gene Symbol, ID, or list of Genes ('Gene Set')
- **Biotype** - Classification of the type of gene according to Ensembl. The biotypes can be grouped into protein coding, pseudogene, long noncoding and short noncoding. Examples of biotypes in each group are as follows:
 - **Protein coding:** IGC gene, IGD gene, IG gene, IGJ gene, IGLV gene, IGM gene, IGV gene, IGZ gene, nonsense mediated decay, nontranslating CDS, non stop decay, polymorphic pseudogene, TRC gene, TRD gene, TRJ gene.
 - **Pseudogene:** disrupted domain, IGC pseudogene, IGJ pseudogene, IG pseudogene, IGV pseudogene, processed pseudogene, transcribed processed pseudogene, transcribed unitary pseudogene, transcribed unprocessed pseudogene, translated processed pseudogene, TRJ pseudogene, unprocessed pseudogene
 - **Long noncoding:** 3prime overlapping ncRNA, ambiguous orf, antisense, antisense RNA, lincRNA, ncRNA host, processed transcript, sense intronic, sense overlapping
 - **Short noncoding:** miRNA, miRNA_pseudogene, miscRNA, miscRNA pseudogene, Mt rRNA, Mt tRNA, rRNA, scRNA, snlRNA, snoRNA, snRNA, tRNA, tRNA_pseudogene
- **Is Cancer Gene Census** - Whether or not a gene is part of [The Cancer Gene Census](#)

Upload Gene Set

In the **Genes** filters panel, instead of supplying genes one-by-one, users can supply a list of genes. Clicking on the **Upload Gene Set** button will launch a dialog as shown below, where users can supply a list of genes or upload a comma-separated text file of genes.

Upload Gene Set

Type or copy-and-paste a list of gene identifiers ?

e.g. ENSG00000155657, TTN, 7273, HGNC:12403, 188840, Q8WZ42

Or choose a file to upload

Browse

Cancel Clear Submit

After supplying a list of genes, a table below will appear which indicates whether the gene was found.

Upload Gene Set

Type or copy-and-paste a list of gene identifiers ?

TP53,ENSG00000155657,Q8WZ44

Or choose a file to upload

Browse

Summary Table (2 matched, 1 unmatched) ▼

Matched (2) Unmatched (1)

2 submitted gene identifiers mapped to 2 unique GDC genes TSV

Symbol	Submitted Gene Identifier	GDC Gene ID	Mapped To
	Ensembl		Symbol
TP53	--	ENSG00000141510	TP53
--	ENSG00000155657	ENSG00000155657	TTN

Cancel Clear Submit

Clicking on **Submit** will filter the results in the Exploration Page by those genes.

Mutation Filters

The final tab of filters is for specific mutations.

Cases

Genes

Mutations



▼ Mutation



e.g. BRAF V600E, chr7:g.140753336A>T

Upload Mutation Set



▼ Impact (VEP)

☐ MODERATE

1,618,053

☐ LOW

655,702

☐ MODIFIER

573,080

☐ HIGH

268,771

▼ Consequence Type



☐ missense_variant

1,648,415

☐ non_coding_transcript_exon_variant

1,110,869

☐ downstream_gene_variant

1,056,982

☐ upstream_gene_variant

681,344

☐ 3_prime_UTR_variant

678,835

17 More...

▼ Type

☐ Single base substitution

2,948,802

☐ Small deletion

98,040

☐ Small insertion

68,764

Users can filter by:

- **Mutation** - Unique ID for that mutation. Users can use the following:
 - UUID - c7c0aeaa-29ed-5a30-a9b6-395ba4133c63
 - DNA Change - chr12:g.121804752delC
 - COSMIC ID - COSM202522
 - List of any mutation UUIDs or DNA Change id's ('Mutation Set')
- **Consequence Type** - Consequence type of this variation; [sequence ontology](#) terms
- **Impact** - A subjective classification of the severity of the variant consequence. This information comes from the [Ensembl VEP](#).
- **Type** - A general classification of the mutation
- **Variant Caller** - The variant caller used to identify the mutation
- **COSMIC ID** - The identifier of the gene or mutation maintained in COSMIC, the Catalogue Of Somatic Mutations In Cancer
- **dbSNP rs ID** - The reference SNP identifier maintained in dbSNP

Upload Mutation Set

In the Mutations filters panel, instead of supplying mutation id's one-by-one, users can supply a list of mutations. Clicking on the Upload Mutation Set button will launch a dialog as shown below, where users can supply a list of mutations or upload a comma-separated text file of mutations.

Upload Mutation Set

Type or copy-and-paste a list of mutation identifiers

e.g. chr3:g.179234297A>G, 92b75ae1-8d4d-52c2-8658-9c981eef0e57

Or choose a file to upload

Browse

Cancel Clear Submit

After supplying a list of mutations, a table below will appear which indicates whether the mutation was found.

Home

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Upload Mutation Set

Type or copy-and-paste a list of mutation identifiers

84aef48f-31e6-52e4-8e05-7d5b9ab15087 fa9713e8-ce92-5413-aacc-ed3d95ab7906 chr12:g.25245350C>T

Or choose a file to upload

Browse

Summary Table (3 matched, 0 unmatched) ▾

Matched (3)

Unmatched (0)

3 submitted mutation identifiers mapped to 3 unique GDC mutations

TSV

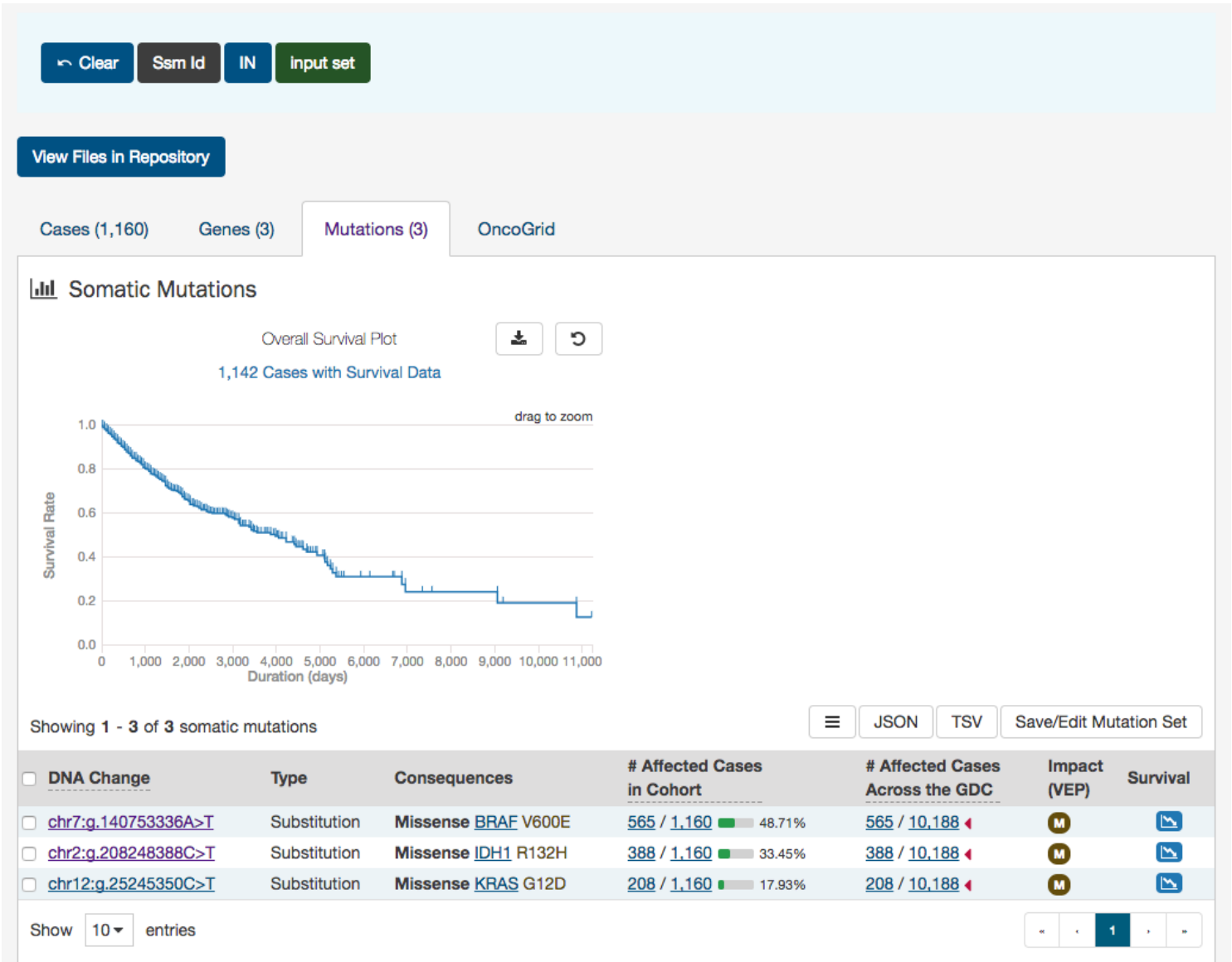
Submitted Mutation Identifier		Mapped To
Mutation UUID	DNA Change	GDC Mutation ID
84aef48f-31e6-52e4-8e05-7d5b9ab15087	--	84aef48f-31e6-52e4-8e05-7d5b9ab15087
fa9713e8-ce92-5413-aacc-ed3d95ab7906	--	fa9713e8-ce92-5413-aacc-ed3d95ab7906
--	chr12:g.25245350C>T	edd1ae2c-3ca9-52bd-a124-b09ed304fcc2

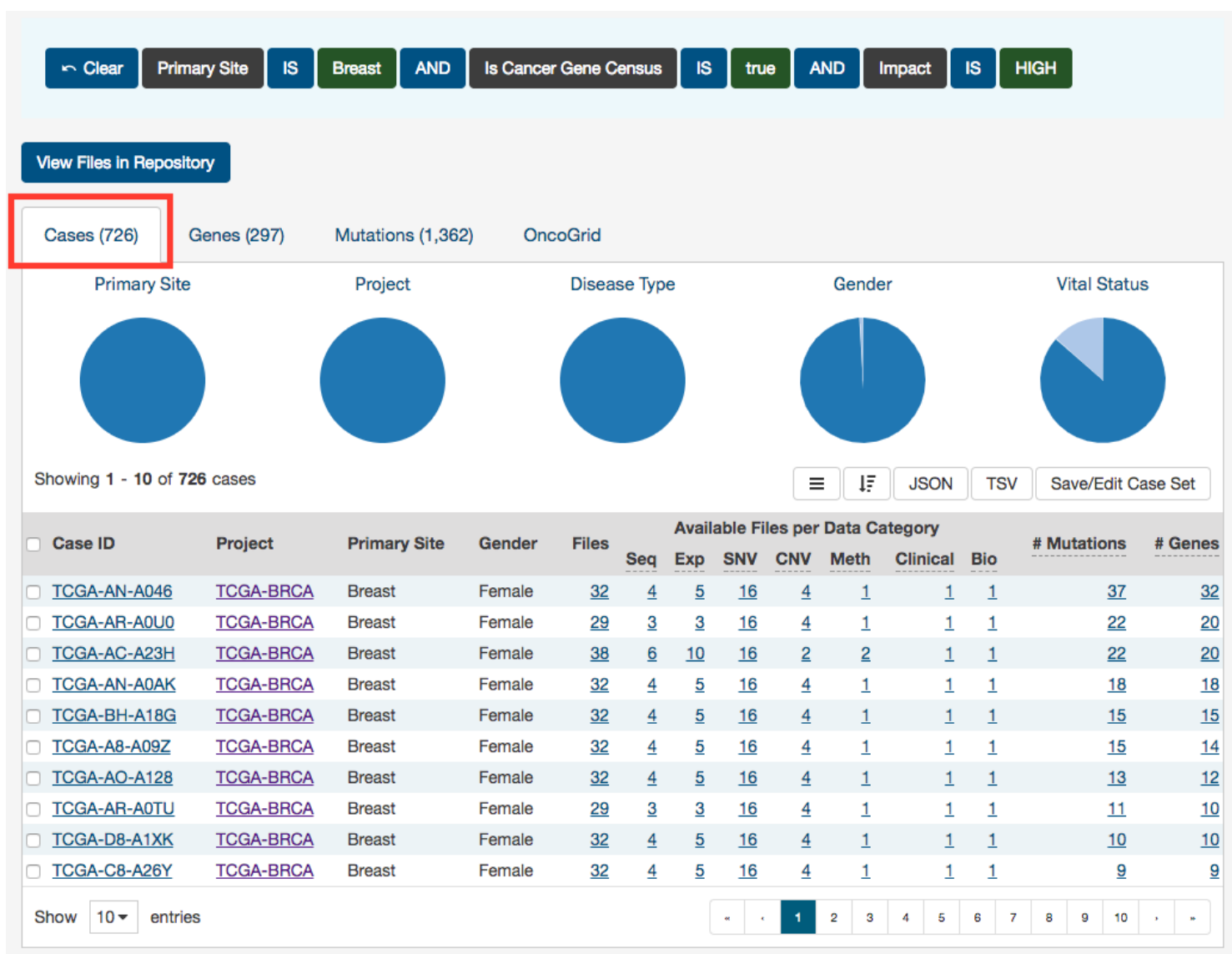
Cancel

Clear

Submit

Clicking on **Submit** will filter the results in the Exploration Page by those mutations.





The top of this section contains a few pie graphs with categorical information regarding the Primary Site, Project, Disease Type, Gender, and Vital Status.

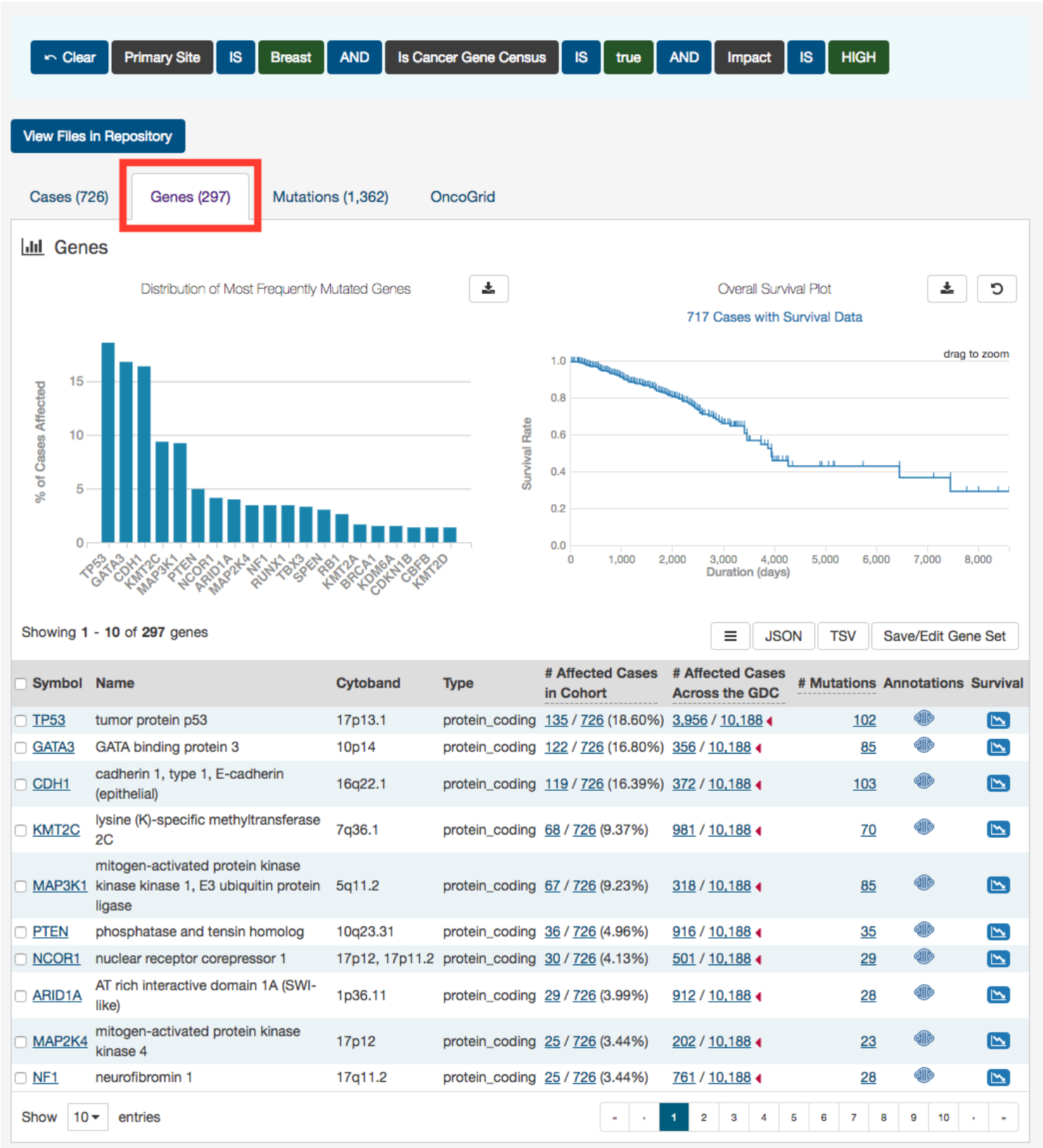
Below these pie charts is a tabular view of cases (which can be exported, sorted and saved using the buttons on the right), that includes the following information:

- **Case ID (Submitter ID):** The Case ID / submitter ID of that case/patient (i.e. TCGA Barcode)
- **Project:** The study name for the project for which the case belongs
- **Primary Site:** The primary site of the cancer/project
- **Gender:** The gender of the case
- **Files:** The total number of files available for that case
- **Available Files per Data Category:** Five columns displaying the number of files available in each of the five data categories. These link to the files for the specific case.
- **___ Mutations:___** The number of SSMS (simple somatic mutations) detected in that case
- **___ Genes:___** The number of genes affected by mutations in that case

Note: By default, the Case UUID is not displayed. You can display the UUID of the case, but clicking on the icon with 3 parallel lines, and choose to display the Case UUID

Genes

The Genes tab will give an overview of all the genes that match the criteria of the filters (Cohort).



The top of this section contains a survival plot of all the cases within the specified Exploration page search, in addition to a bar graph of the most frequently mutated genes. Hovering over each bar in the plot will display information about the percentage of

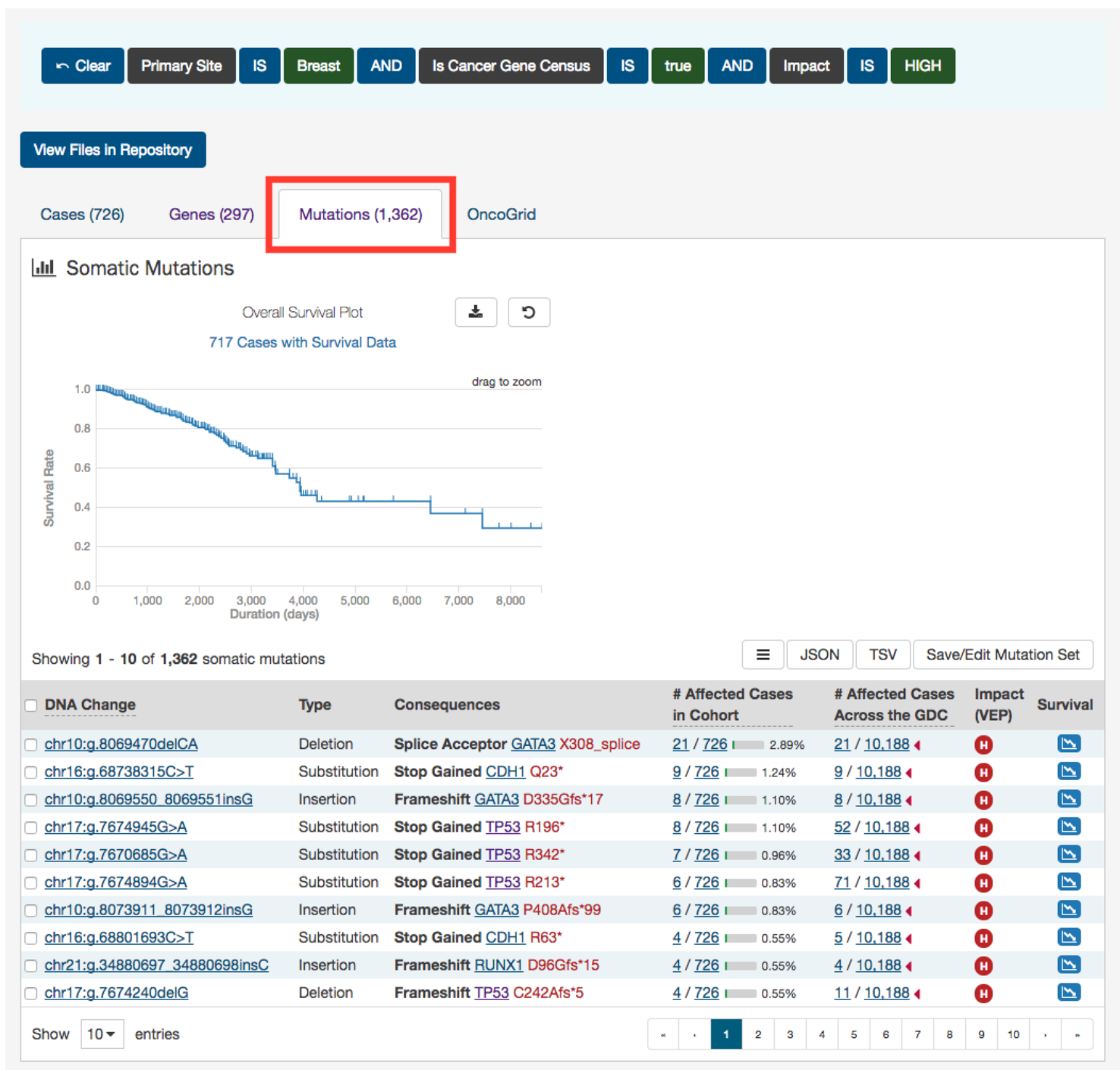
cases affected. Users may choose to download the underlying data in JSON or TSV format or an image of the graph in SVG or PNG format by clicking the **download** icon at the top of each graph.

Below these graphs is a tabular view of the genes affected, which includes the following information:

- **Symbol:** The gene symbol, which links to the Gene Summary Page
- **Name:** Full name of the gene
- **Cytoband:** The location of the mutation on the chromosome in terms of Giemsa-stained samples.
- **Type:** The type of gene
- ___ Affected Cases in Cohort:___ The number of cases affected in the Cohort
- ___ Affected Cases Across all Projects:___ The number of cases within all the projects in the GDC that contain a mutation on this gene. Clicking the red arrow will display the cases broken down by project
- ___ Mutations:___ The number of SSMs (simple somatic mutations) detected in that gene
- **Annotations:** Includes a COSMIC symbol if the gene belongs to [The Cancer Gene Census](#)
- **Survival Analysis:** An icon that, when clicked, will plot the survival rate between cases in the project with mutated and non-mutated forms of the gene

Mutations

The **Mutations** tab will give an overview of all the mutations who match the criteria of the filters (Cohort).



At the top of this tab is a survival plot of all the cases within the specified exploration page filters.

A table is displayed below that lists information about each mutation:

- **DNA Change:** The chromosome and starting coordinates of the mutation are displayed along with the nucleotide differences between the reference and tumor allele
- **Type:** A general classification of the mutation
- **Consequences:** The effects the mutation has on the gene coding for a protein (i.e. synonymous, missense, non-coding transcript). A link to the Gene Summary Page for the gene affected by the mutation is included
- **___ Affected Cases in Cohort:**___ The number of affected cases in the Cohort as a fraction and as a percentage
- **___ Affected Cases in Across all Projects:**___ The number of affected cases, expressed as number across all projects. This information comes from the [Ensembl VEP](#). Choosing the arrow next to the percentage will display a breakdown of each affected project

- **Impact (VEP):** A subjective classification of the severity of the variant consequence. The categories are:
 - **HIGH (H):** The variant is assumed to have high (disruptive) impact in the protein, probably causing protein truncation, loss of function, or triggering nonsense mediated decay
 - **MODERATE (M):** A non-disruptive variant that might change protein effectiveness
 - **LOW (L):** Assumed to be mostly harmless or unlikely to change protein behavior
 - **MODIFIER (MO):** Usually non-coding variants or variants affecting non-coding genes, where predictions are difficult or there is no evidence of impact
- **Survival Analysis:** An icon that when clicked, will plot the survival rate between the gene's mutated and non-mutated cases

Note: By default, the Mutation UUID is not displayed. You can display the UUID of the case, but clicking on the icon with 3 parallel lines, and choose to display the Mutation UUID

OncoGrid

The Exploration page includes an OncoGrid plot of the cases with the most mutations, for the top 50 mutated genes affected by high impact mutations. Genes displayed on the left of the grid (Y-axis) correspond to individual cases on the bottom of the grid (X-axis).



The grid is color-coded with a legend at the top left which describes what type of mutation consequence is observed for each gene/case combination. Clinical information and the available data for each case are available at the bottom of the grid.

The right side of the grid displays additional information about the genes:

- **Gene Sets:** Describes whether a gene is part of [The Cancer Gene Census](#). (The Cancer Gene Census is an ongoing effort to catalogue those genes for which mutations have been causally implicated in cancer)
- **GDC:** Identifies all cases in the GDC affected with a mutation in this gene

OncoGrid Options

To facilitate readability and comparisons, drag-and-drop can be used to reorder the gene rows. Double clicking a row in the "Cases Affected" bar at the right side of the graphic launches the respective Gene Summary Page page. Hovering over a cell will display information about the mutation such as its ID, affected case, and biological consequence. Clicking on the cell will bring the user to the respective Mutation Summary page.

A tool bar at the top right of the graphic allows the user to export the data as a JSON object, PNG image, or SVG image. Seven buttons are available in this toolbar:

- **Download:** Users can choose to export the contents either to a static image file (PNG or SVG format) or the underlying data in JSON format
- **Reload Grid:** Sets all OncoGrid rows, columns, and zoom levels back to their initial positions
- **Cluster Data:** Clusters the rows and columns to place mutated genes with the same cases and cases with the same mutated genes together
- **Toggle Heatmap:** The view can be toggled between cells representing mutation consequences or number of mutations in each gene
- **Toggle Gridlines:** Turn the gridlines on and off
- **Toggle Crosshairs:** Turns crosshairs on, so that users can zoom into specific sections of the OncoGrid
- **Fullscreen:** Turns Fullscreen mode on/off

File Navigation

After utilizing the Exploration Page to narrow down a specific cohort, users can find the specific files that relate to this group by clicking on the **View Files in Repository** button as shown in the image below.

The screenshot shows the NIH GDC Data Portal Exploration page. The top navigation bar includes links for Home, Projects, Exploration (active), Analysis, Repository, Quick Search, Manage Sets, Login, Cart, and GDC Apps. The left sidebar has tabs for Cases, Genes, and Mutations. Under the Cases tab, there is a search bar with the text "e.g. BRAF, ENSG00000157764" and an "Upload Gene Set" button. Below the search bar, there are filters for Biotype (protein_coding, 297) and Is Cancer Gene Census (true, 297). The main content area shows a toolbar with buttons for Clear, Primary Site, IS, Breast, AND, Is Cancer Gene Census, IS, true, AND, Impact, IS, and HIGH. A red box highlights the "View Files in Repository" button. Below the toolbar, there are five pie charts representing different data categories: Primary Site, Project, Disease Type, Gender, and Vital Status. The text "Showing 1 - 10 of 726 cases" is displayed. Below the charts, there is a table with columns for Case ID, Project, Primary Site, Gender, Files, and Available Files per Data Category (Seq, Exp, SNV, CNV, Meth, Clinical, Bio). The table lists 10 cases, each with a checkbox, Case ID, Project, Primary Site, Gender, and a row of counts for the available files. The bottom of the page shows a "Show 10 entries" dropdown and a pagination bar.

Case ID	Project	Primary Site	Gender	Files	Available Files per Data Category							# Mutations	# Genes
					Seq	Exp	SNV	CNV	Meth	Clinical	Bio		
<input type="checkbox"/> TCGA-AN-A046	TCGA-BRCA	Breast	Female	32	4	5	16	4	1	1	1	37	32
<input type="checkbox"/> TCGA-AR-A0U0	TCGA-BRCA	Breast	Female	29	3	3	16	4	1	1	1	22	20
<input type="checkbox"/> TCGA-AC-A23H	TCGA-BRCA	Breast	Female	38	6	10	16	2	2	1	1	22	20
<input type="checkbox"/> TCGA-AN-A0AK	TCGA-BRCA	Breast	Female	32	4	5	16	4	1	1	1	18	18
<input type="checkbox"/> TCGA-BH-A18G	TCGA-BRCA	Breast	Female	32	4	5	16	4	1	1	1	15	15
<input type="checkbox"/> TCGA-A8-A09Z	TCGA-BRCA	Breast	Female	32	4	5	16	4	1	1	1	15	14
<input type="checkbox"/> TCGA-AO-A128	TCGA-BRCA	Breast	Female	32	4	5	16	4	1	1	1	13	12
<input type="checkbox"/> TCGA-AR-A0TU	TCGA-BRCA	Breast	Female	29	3	3	16	4	1	1	1	11	10
<input type="checkbox"/> TCGA-D8-A1XK	TCGA-BRCA	Breast	Female	32	4	5	16	4	1	1	1	10	10
<input type="checkbox"/> TCGA-C8-A26Y	TCGA-BRCA	Breast	Female	32	4	5	16	4	1	1	1	9	9

Clicking this button will navigate the users to the Repository Page, filtered by the cases within the cohort.

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Files

Cases

[Add a File Filter](#)

File

Data Category

☐ Simple Nucleotide Variation **127,480**
☐ Transcriptome Profiling **57,898**
☐ Raw Sequencing Data **45,998**
☐ Copy Number Variation **44,752**
☐ DNA Methylation **12,359**

2 More...

Clear

Case

IN

Input set

[Advanced Search](#)

[Add All Files to Cart](#)
[Download Manifest](#)
[View 726 Cases in Exploration](#)

[Browse Annotations](#)

Files (18,353)

Cases (726)

37.48 TB

Primary Site

Project

Data Category

Data Type

Data Format

Show More

The filters chosen on the Exploration page are displayed as an **input set** on the Repository page. Additional filters may be added on top of this **input set**, but the original set cannot be modified and instead must be created from scratch again.

Chapter 4

Repository

Repository

Summary

The Repository Page is the primary method of accessing data in the GDC Data Portal. It provides an overview of all cases and files available in the GDC and offers users a variety of filters for identifying and browsing cases and files of interest. Users can access the Repository Page from the GDC Data Portal front page, from the Data Portal toolbar, or directly at <https://portal.gdc.cancer.gov/repository>.

Filters / Facets

On the left, a panel of data facets allows users to filter cases and files using a variety of criteria. If facet filters are applied, the tabs on the right will display information about matching cases and files. If no filters are applied, the tabs on the right will display information about all available data.

On the right, two tabs contain information about available data:

- *Files tab* provides a list of files, select information about each file, and links to individual file detail pages.
- *Cases tab* provides a list of cases, select information about each case, and links to individual case summary pages

The banner above the tabs on the right displays any active facet filters and provides access to advanced search.

The top of the Repository Page contains a few summary pie charts for Primary Sites, Projects, Disease Type, Gender, and Vital Status. These reflect all available data or, if facet filters are applied, only the data that matches the filters. Clicking on a specific slice in a pie chart, or on a number in a table, applies corresponding facet filters.

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Files

Cases

[Add a File Filter](#)

File

Data Category

☐ Simple Nucleotide Variation **91,430**
☐ Transcriptome Profiling **57,090**
☐ Raw Sequencing Data **45,900**
☐ Copy Number Variation **44,752**
☐ DNA Methylation **12,359**
2 More...

Data Type

☐ Aligned Reads **45,900**
☐ Annotated Somatic Mutation **45,577**
☐ Raw Simple Somatic Mutation **45,577**
☐ Gene Expression Quantification **34,722**
☐ Copy Number Segment **22,576**
8 More...

Experimental Strategy

☐ WXS **114,323**
☐ RNA-Seq **46,329**
☐ Genotyping Array **44,752**
☐ miRNA-Seq **34,494**
☐ Methylation Array **12,359**

← Start searching by selecting a facet

[Advanced Search](#)

[Add All Files to Cart](#)
[Download Manifest](#)
[View 14,551 Cases in Exploration](#)

[Browse Annotations](#)

Files (274,724) Cases (14,551) 470.59 TB

Primary Site

Project

Data Category

Data Type

Data Format

Show More

Showing 1 - 10 of 274,724 files

Access

File Name

Cases

Project

Data Category

Data Format

File Size

Annotations

open	KIWIS_p_TCGASNP_225_226_N_GenomeWideSNP_6_C07_1151838.grch38.seg.txt	1	TCGA-KICH	Copy Number Variation	TXT	66.56 KB	0
controlled	9f61d40b-6e2e-4c3e-8ab7-0a85e35c8208.vsp.vcf.gz	1	TCGA-PRAD	Simple Nucleotide Variation	VCF	961.32 KB	0
open	jhu-usc.edu_SARC.HumanMethylation450.12.lvl-3.TCGA-X2-A95T-01A-11D-A37D-05.gdc_hg38.txt	1	TCGA-SARC	DNA Methylation	TXT	141.28 MB	0
open	ODDLY_p_TCGASNP_165_184_N_GenomeWideSNP_6_A01_1051570.noconv.grch38.seg.txt	1	TCGA-PRAD	Copy Number Variation	TXT	9.93 KB	0
open	cac14e64-7df9-43ce-8999-7fc1a04b395c.mirbase21.mrnas.quantification.txt	1	TCGA-LUAD	Transcriptome Profiling	TSV	50.26 KB	0
controlled	15c246a5-5e01-43d8-8425-a5fe008d319a.vcf.gz	1	TCGA-LIHC	Simple Nucleotide Variation	VCF	272.71 KB	0
controlled	635512ba-8678-4a7f-bc80-c0ed8c2e3432.snp.Somatic.hc.vcf.gz	1	TCGA-BRCA	Simple Nucleotide Variation	VCF	27.16 KB	0
controlled	d98c0c50-77c5-41a5-823a-1ba3fb1281ef.vcf.gz	1	TCGA-LAML	Simple Nucleotide Variation	VCF	3.11 MB	1
controlled	C495.TCGA-CN-A63Y-10A-01D-A30H-08.2_gdc_realn.bam	1	TCGA-HNSC	Raw Sequencing Data	BAM	12.67 GB	0
controlled	133401.bam	1	TCGA-LUSC	Raw Sequencing Data	BAM	213.34 MB	0

Show 10 entries

1

2

3

4

5

6

7

8

9

10

Facets Panel

Facets represent properties of the data that can be used for filtering. The facets panel on the left allows users to filter the cases and files presented in the tabs on the right.

The facets panel is divided into two tabs, with the Files tab containing facets pertaining to data files and experimental strategies, while the Cases tab containing facets pertaining to the cases and biospecimen information. Users can apply filters in both tabs simultaneously. The applied filters will be displayed in the banner above the tabs on the right, with the option to open the filter in Advanced Search to further refine the query.

The [Getting Started](#) section provides instructions on using facet filters. In the following example, a filter from the Cases tab (“primary site”) and filters from the Files tab (“data category”, “experimental strategy”) are both applied:

The screenshot displays the GDC search interface. On the left, the 'Files' facet is expanded, showing filters for File (e.g., 142682.bam), Data Category (Transcriptome Profiling, Raw Sequencing Data), Data Type (Aligned Reads), Experimental Strategy (WXS, RNA-Seq, miRNA-Seq), Workflow Type (STAR 2-Pass), Data Format (BAM), Platform (Illumina), and Access (controlled). The main panel features search filters (Clear, Primary Site, IS, Breast, AND, Data Category, IS, Raw Sequencing Data, AND, Experimental Strategy, IS, RNA-Seq) and buttons for 'Add All Files to Cart', 'Download Manifest', and 'View 1,092 Cases in Exploration'. Below these are five circular visualizations for Primary Site, Project, Data Category, Data Type, and Data Format. A table shows the first 10 of 1,222 files, with columns for Access, File Name, Cases, Project, Data Category, Data Format, File Size, and Annotations.

The default set of facets is listed below.

Files facets tab:

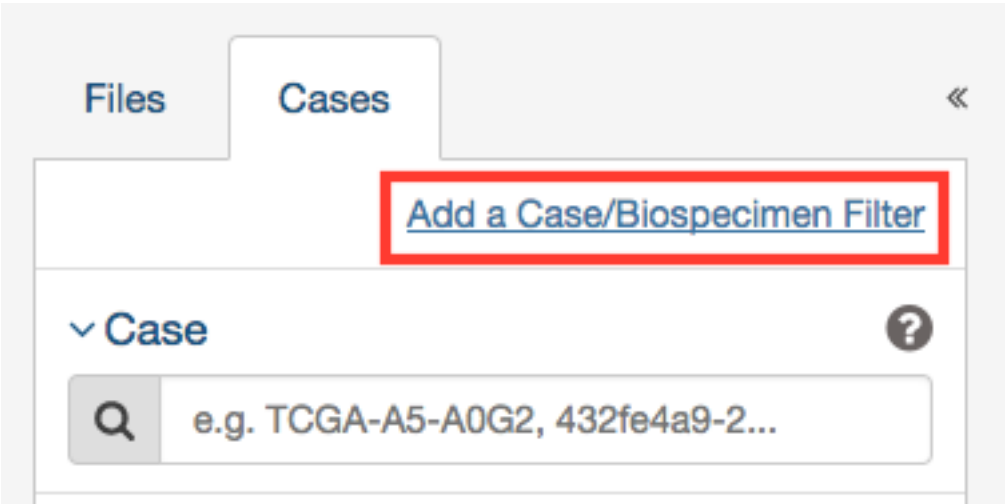
- **File:** Specify individual files using filename or UUID.
- **Data Category:** A high-level data file category, such as “Raw Sequencing Data” or “Transcriptome Profiling”.
- **Data Type:** Data file type, such as “Aligned Reads” or “Gene Expression Quantification”. Data Type is more granular than Data Category.
- **Experimental Strategy:** Experimental strategies used for molecular characterization of the cancer.
- **Workflow Type:** Bioinformatics workflow used to generate or harmonize the data file.
- **Data Format:** Format of the data file.
- **Platform:** Technological platform on which experimental data was produced.
- **Access Level:** Indicator of whether access to the data file is open or controlled.

Cases facets tab:

- **Case:** Specify individual cases using submitter ID (barcode) or UUID.
- **Case Submitter ID Prefix:** Search for cases using a part (prefix) of the submitter ID (barcode).
- **Primary Site:** Anatomical site of the cancer under investigation or review.
- **Cancer Program:** A cancer research program, typically consisting of multiple focused projects.
- **Project:** A cancer research project, typically part of a larger cancer research program.
- **Disease Type:** Type of cancer studied.
- **Gender:** Gender of the patient.
- **Age at Diagnosis:** Patient age at the time of diagnosis.
- **Vital Status:** Indicator of whether the patient was living or deceased at the date of last contact.
- **Days to Death:** Number of days from date of diagnosis to death of the patient.
- **Race:** Race of the patient.
- **Ethnicity:** Ethnicity of the patient.

Adding Custom Facets

The Repository Page provides access to additional data facets beyond those listed above. Facets corresponding to additional properties listed in the GDC Data Dictionary can be added using the “add a filter” links available at the top of the Cases and Files facet tabs:



The links open a search window that allows the user to find an additional facet by name or description. Not all facets have values available for filtering; checking the “Only show fields with values” checkbox will limit the search results to only those that do. Selecting a facet from the list of search results below the search box will add it to the facets panel.

Add a Case/Biospecimen Filter

Cancel

Search for a field:

tumor_

8 cases fields

☐ Only show fields with values

CA	diagnoses.tumor_grade keyword
	Numeric value to express the degree of abnormality of cancer cells, a measure of differentiation and aggressiveness.
CA	diagnoses.tumor_stage keyword
	The extent of a cancer in the body. Staging is usually based on the size of the tumor, whether lymph nodes contain cancer, and whether the cancer has spread from the original site to other parts of the body. The accepted values for tumor_stage depend on the tumor site, type, and accepted staging system. These items should accompany the tumor_stage value as associated metadata.
CA	samples.portions.analytes.normal_tumor_genotype_snp_match keyword
	Text term that represents whether or not the genotype of the normal tumor matches or if the data is not available.
CA	samples.portions.slides.percent_tumor_cells long
	Numeric value that represents the percentage of infiltration by granulocytes in a sample.
CA	samples.portions.slides.percent_tumor_nuclei long
	Numeric value to represent the percentage of tumor nuclei in a malignant neoplasm sample or specimen.

Newly added facets will show up at the top of the facets panel and can be removed individually by clicking on the red cross to the right of the facet name. The default set of facets can be restored by clicking “Reset”.

Files

Cases

«

[Reset](#) | [Add a Case/Biospecimen Filter](#)

▼

Diagnoses Tumor Stage

×

☐

stage iia

358

☐

stage iib

255

☐

stage iia

155

☐

stage i

89

☐

stage ia

86

8 More...

▼

Case

?

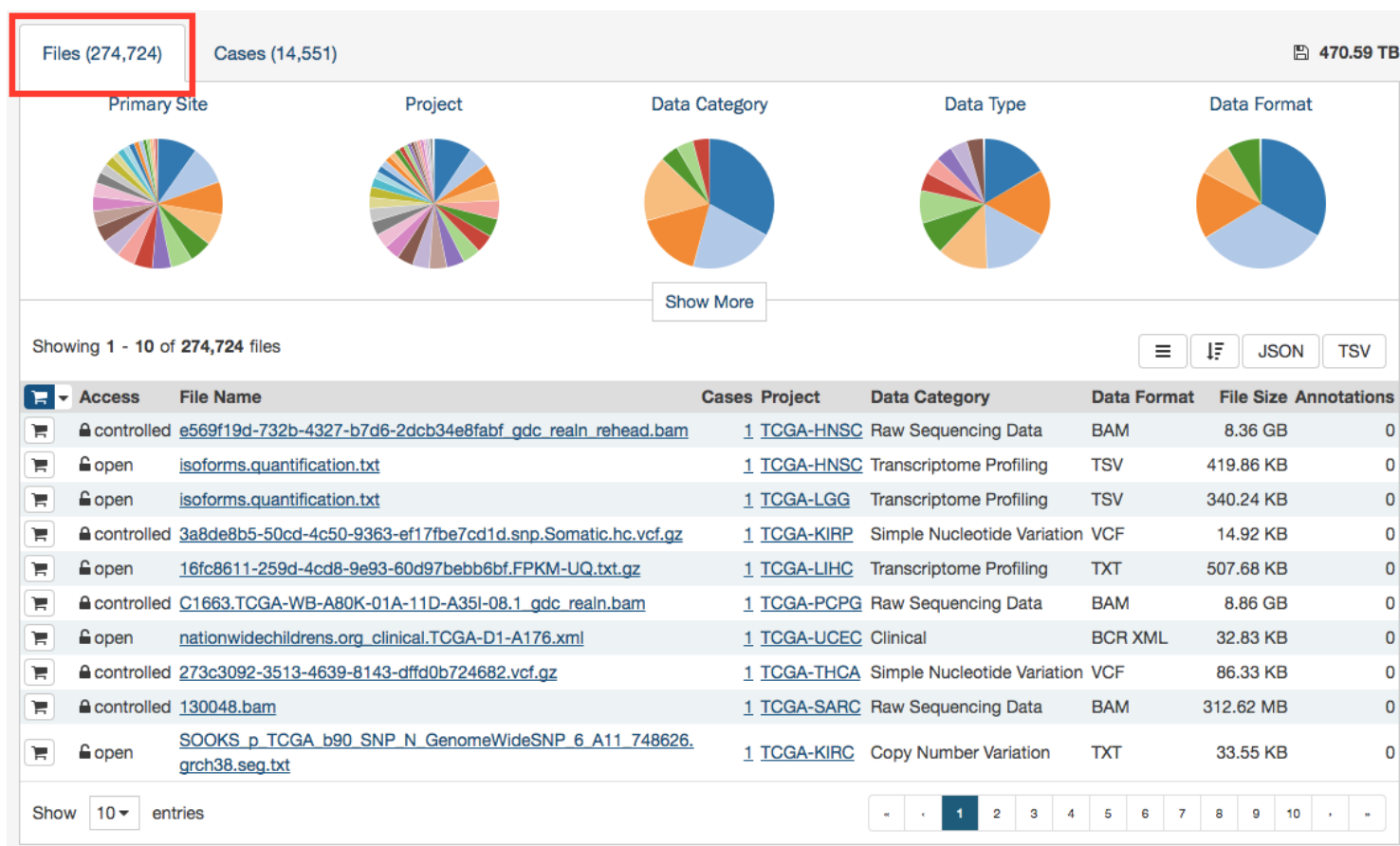
Q

e.g. TCGA-A5-A0G2, 432fe4a9-2...

Results

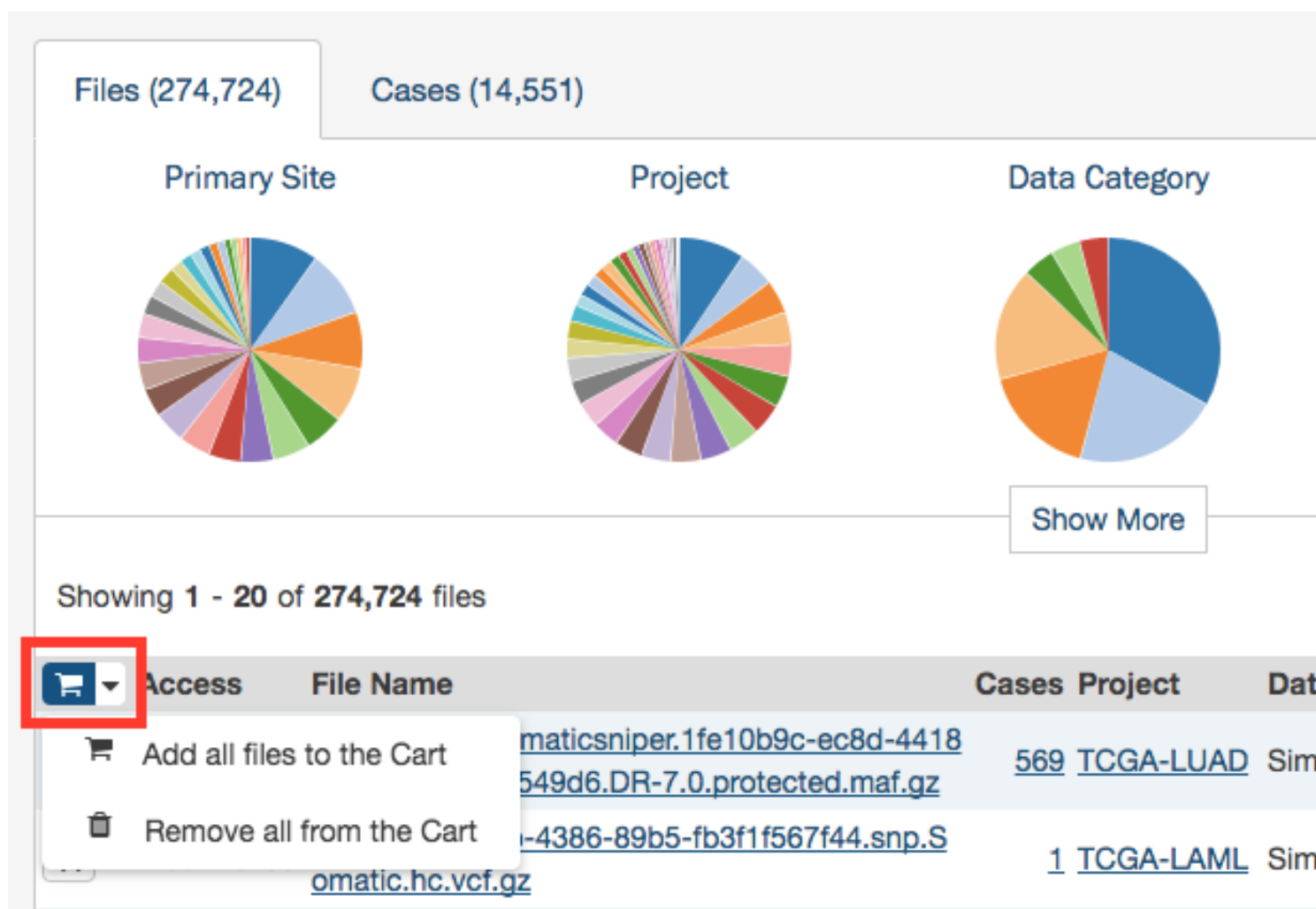
Files List

The Files tab on the right provides a list of available files and select information about each file. If facet filters are applied, the list includes only matching files. Otherwise, the list includes all data files available in the GDC Data Portal.



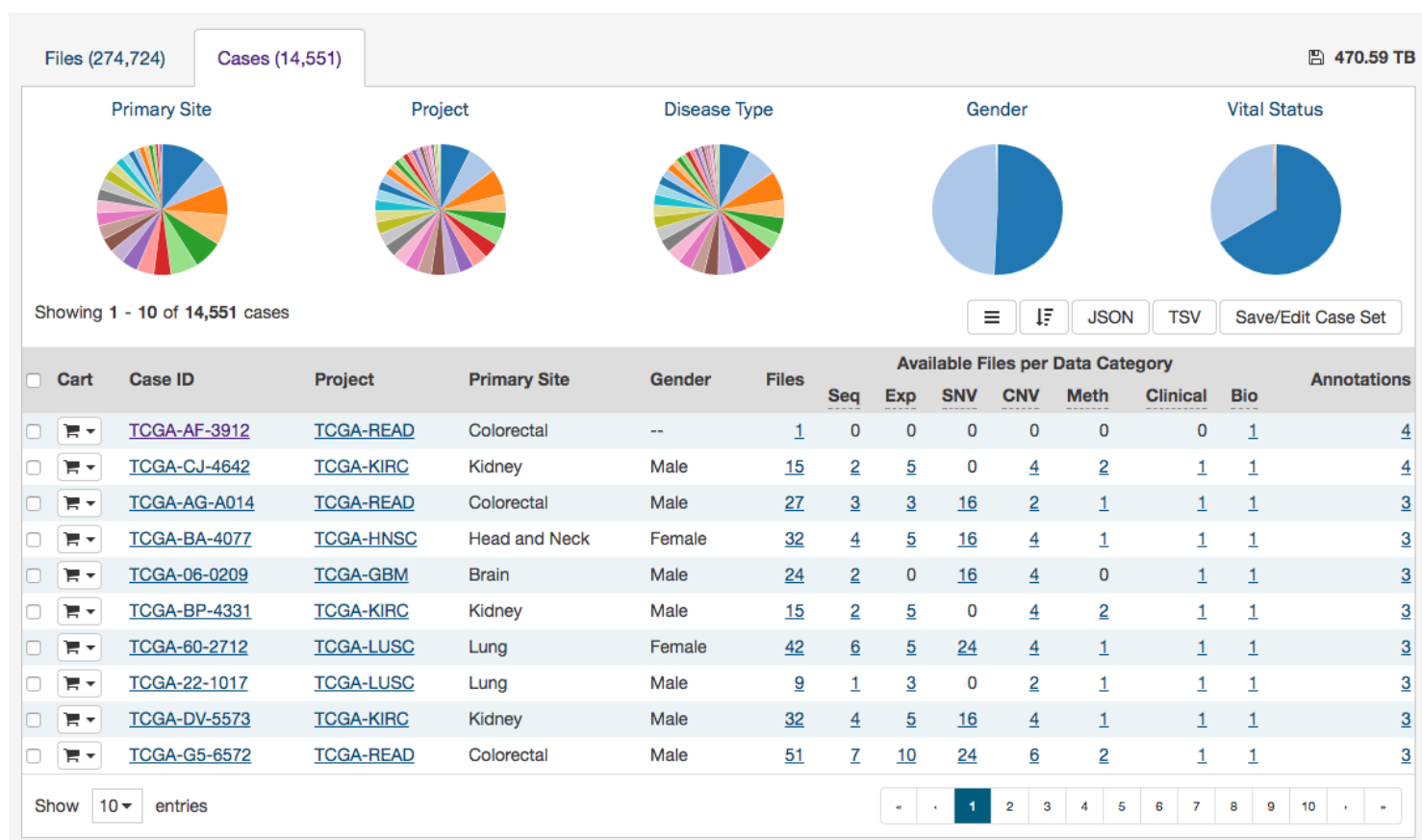
The *File Name* column includes links to file detail pages where the user can learn more about each file.

Users can add individual file(s) to the file cart using the cart button next to each file. Alternatively, all files that match the current facet filters can be added to the cart using the menu in the top left corner of the table:



Cases List

The Cases tab on the right provides a list of available cases and select information about each case. If facet filters are applied, the list includes only matching cases. Otherwise, the list includes all cases available in the GDC Data Portal.



The list includes links to [case summary pages](#) in the *Case UUID* column, the Submitter ID (i.e. TCGA Barcode), and counts of the available file types for each case. Clicking on a count will apply facet filters to display the corresponding files.

The list also includes a shopping cart button, allowing the user to add all files associated with a case to the file cart for downloading at a later time:

Cart	Case UUID	Submitter ID	Project
<input type="checkbox"/>	9bbc01b4	TCGA-09-0367	TCGA-OV
<input type="checkbox"/>	Add all Case files to the Cart (32)		TCGA-GBM
<input type="checkbox"/>	e487c72f	TCGA-66-2788	TCGA-LUSC
<input type="checkbox"/>	1e5a3796	TCGA-IN-AB1X	TCGA-STAD

Navigation

After utilizing the Repository Page to narrow down a specific set of cases, users can continue to explore the mutations and genes affected by these cases by clicking the **View Files in Repository** button as shown in the image below.

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Repository

Q

[Manage Sets](#)

🔊

🛒 0

☰

Files

Cases

[Add a Case/Biospecimen Filter](#)

Case

Upload Case Set

Case ID

Go!

Primary Site

☒ Breast

898

☐ Lung

609

☐ Kidney

571

☐ Prostate

488

☐ Thyroid

482

23 More...

Program

☐ TCGA

898

Clear

Vital Status

IS

alive

AND

Primary Site

IS

Breast

AND

Data Category

IS

Simple Nucleotide Variation

Advanced Search

Add All Files to Cart

Download Manifest

View 898 Cases in Exploration

[Browse Annotations](#)

Files (7,408)

Cases (898)

8.12 GB

Primary Site

Project

Disease Type

Gender

Vital Status

Showing 1 - 10 of 898 cases

☰

⌵

JSON

TSV

Save/Edit Case Set

Cart	Case ID	Project	Primary Site	Gender	Files	Available Files per Data Category							Annotations
						Seq	Exp	SNV	CNV	Meth	Clinical	Bio	
<input type="checkbox"/>	TCGA-A8-A08S	TCGA-BRCA	Breast	Female	32	4	5	16	4	1	1	1	2
<input type="checkbox"/>	TCGA-A8-A08F	TCGA-BRCA	Breast	Female	29	3	3	16	4	1	1	1	2
<input type="checkbox"/>	TCGA-A8-A09E	TCGA-BRCA	Breast	Female	32	4	5	16	4	1	1	1	2

Clicking this button will navigate the users to the Exploration Page, filtered by the cases within the cohort.

Case Summary Page

The Case Summary page displays case details including the project and disease information, data files that are available for that case, and the experimental strategies employed. A button in the top-right corner of the page allows the user to add all files associated with the case to the file cart.

58

CA 7dcf550c-90ce-4f63-aecd-0e46897e2a3e

🛒 Add all files to the cart

📄 Summary

Case UUID	7dcf550c-90ce-4f63-aecd-0e46897e2a3e
Case ID	TCGA-AC-A23H
Project	TCGA-BRCA
Project Name	Breast Invasive Carcinoma
Disease Type	Breast Invasive Carcinoma
Program	TCGA
Primary Site	Breast

FILES

[38](#)



ANNOTATIONS

0



File Counts by Data Category

Data Category	Files	(n=38)
Raw Sequencing Data	6	<div><div></div></div>
Transcriptome Profiling	10	<div><div></div></div>
Simple Nucleotide Variation	16	<div><div></div></div>
Copy Number Variation	2	<div><div></div></div>
DNA Methylation	2	<div><div></div></div>
Clinical	1	<div><div></div></div>
Biospecimen	1	<div><div></div></div>

File Counts by Experimental Strategy

Experimental Strategy	Files	(n=38)
Genotyping Array	2	<div><div></div></div>
Methylation Array	2	<div><div></div></div>
WXS	18	<div><div></div></div>
RNA-Seq	8	<div><div></div></div>
miRNA-Seq	6	<div><div></div></div>

Clinical and Biospecimen Information

The page also provides clinical and biospecimen information about that case. Links to export clinical and biospecimen information in JSON format are provided.

Clinical

Demographic

Diagnoses / Treatments (1)

Family Histories (0)

Exposures (1)

ID

d40005ad-6bb7-5b32-9f2d-2ab2394dd0ba

Ethnicity

not hispanic or latino

Gender

female

Race

white

Year Of Birth

1919

Year Of Death

--

Export

Biospecimen

Search

Expand All

Submitter ID

TCGA-AC-A23H-11A

Sample ID

7df59ca8-7e51-4581-9d7f-8bba0395ce17

Sample Type

Solid Tissue Normal

Sample Type Id

11

Tissue Type

--

Tumor Code

--

Tumor Code Id

--

Oct Embedded

false

Shortest Dimension

--

Intermediate Dimension

--

Longest Dimension

--

Is Ffpe

false

Pathology Report Uuid

--

Tumor Descriptor

--

Current Weight

--

Initial Weight

70

Composition

--

Time Between Clamping And Freezing

--

Time Between Excision And Freezing

--

Days To Sample Procurement

--

Freezing Method

--

Preservation Method

--

Days To Collection

478

Portions

1

Status

4

Samples

TCGA-AC-A23H-11A

Portions

TCGA-AC-A23H-11A-12

Analytes

TCGA-AC-A23H-11A-12D

Aliquots

TCGA-AC-A23H-11A-12D-A161-05

TCGA-AC-A23H-11A-12D-A158-02

TCGA-AC-A23H-11A-12D-A160-01

TCGA-AC-A23H-11A-12D-A17G-09

TCGA-AC-A23H-11A-12D-A159-09

TCGA-AC-A23H-11A-12W

Aliquots

TCGA-AC-A23H-11A-12W-A16L-09

TCGA-AC-A23H-11A-12R

Aliquots

TCGA-AC-A23H-11A-12R-A156-13

TCGA-AC-A23H-11A-12R-A157-07

Slides

TCGA-AC-A23H-01A

Portions

For clinical records that support multiple records of the same type (Diagnoses, Family Histories, or Exposures), a UUID of the record is provided on the left hand side of the corresponding tab, allowing the user to select the entry of interest.

Biospecimen Search

A search filter just below the biospecimen section can be used to find and filter biospecimen data. The wildcard search will highlight entities in the tree that match the characters typed. This will search both the case submitter ID, as well as the additional metadata for each entity. For example, searching ‘Primary Tumor’ will highlight samples that match that type.

Biospecimen

primary tumor

Samples

TCGA-AC-A23H-11A

Portions

TCGA-AC-A23H-11A-12

Analytes

TCGA-AC-A23H-11A-12D

Aliquots

TCGA-AC-A23H-11A-12D-A161-05

TCGA-AC-A23H-11A-12D-A158-02

TCGA-AC-A23H-11A-12D-A160-01

TCGA-AC-A23H-11A-12D-A17G-09

TCGA-AC-A23H-11A-12D-A159-09

TCGA-AC-A23H-11A-12W

Aliquots

TCGA-AC-A23H-11A-12W-A16L-09

TCGA-AC-A23H-11A-12R

Aliquots

TCGA-AC-A23H-11A-12R-A156-13

TCGA-AC-A23H-11A-12R-A157-07

Slides

TCGA-AC-A23H-01A

Portions

Expand All

Submitter ID

TCGA-AC-A23H-01A

Sample ID

d7e3b628-d5fd-4e79-9c4a-6409330fb8a7

Sample Type

Primary Tumor

Sample Type Id

01

Tissue Type

--

Tumor Code

--

Tumor Code Id

--

Oct Embedded

false

Shortest Dimension

--

Intermediate Dimension

--

Longest Dimension

--

Is Ffpe

false

Pathology Report Uuid

A7C7D409-D086-4A9B-8C8F-E7E231D5891D

Tumor Descriptor

--

Current Weight

--

Initial Weight

70

Composition

--

Time Between Clamping And Freezing

--

Time Between Excision And Freezing

--

Days To Sample Procurement

--

Freezing Method

--

Preservation Method

--

Days To Collection

478

Portions

1

Status

4

Export

Most Frequent Somatic Mutations

The case entity page also lists the mutations found in that particular case.

Most Frequent Somatic Mutations

Open in Exploration

Showing 1 - 10 of 6,704 somatic mutations

≡

JSON

TSV

Save/Edit Mutation Set

<input type="checkbox"/> DNA Change	Type	Consequences	# Affected Cases in TCGA-BRCA	# Affected Cases Across the GDC	Impact (VEP)
<input type="checkbox"/> chr2:g.159332656C>T	Substitution	Missense BAZ2B E1943K	1 / 986 0.10%	1 / 10,188	M
<input type="checkbox"/> chr18:g.34766028G>C	Substitution	Missense DTNA Q45H	1 / 986 0.10%	1 / 10,188	M
<input type="checkbox"/> chr7:g.123464853C>T	Substitution	Missense IQUB E580K	1 / 986 0.10%	1 / 10,188	M
<input type="checkbox"/> chr19:g.38489348G>T	Substitution	Stop Gained RYR1 E1907*	1 / 986 0.10%	1 / 10,188	H
<input type="checkbox"/> chr15:g.65059369C>T	Substitution	Synonymous RASL12 L70L	1 / 986 0.10%	1 / 10,188	L
<input type="checkbox"/> chrX:g.53087790G>A	Substitution	Missense TSPYL2 E645K	1 / 986 0.10%	1 / 10,188	M
<input type="checkbox"/> chr17:g.40164065C>T	Substitution	Missense CASC3 S457F	1 / 986 0.10%	1 / 10,188	M
<input type="checkbox"/> chr8:g.94664727C>T	Substitution	Synonymous ESRP1 T225T	1 / 986 0.10%	1 / 10,188	L
<input type="checkbox"/> chr2:g.138288526C>G	Substitution	Non Coding Transcript Exon YWHAEP5	1 / 986 0.10%	1 / 10,188	MO
<input type="checkbox"/> chr1:g.38045855C>G	Substitution	Missense POU3F1 E297Q	1 / 986 0.10%	1 / 10,188	M

Show 10 entries

1

2

3

4

5

6

7

8

9

10

The table lists the following information for each mutation

61


- **DNA Change:** The chromosome and starting coordinates of the mutation are displayed along with the nucleotide differences between the reference and tumor allele
- **Type:** A general classification of the mutation
- **Consequences:** The effects the mutation has on the gene coding for a protein (i.e. synonymous, missense, non-coding transcript)
- **___ Affected Cases in Project:**___ The number of affected cases, expressed as number across all mutations within the Project
- **___ Affected Cases Across GDC:**___ The number of affected cases, expressed as number across all projects. Choosing the arrow next to the percentage will expand the selection with a breakdown of each affected project
- **Impact (VEP):** A subjective classification of the severity of the variant consequence. This information comes from the [Ensembl VEP](#). The categories are:
 - **HIGH (H):** The variant is assumed to have high (disruptive) impact in the protein, probably causing protein truncation, loss of function or triggering nonsense mediated decay
 - **MODERATE (M):** A non-disruptive variant that might change protein effectiveness
 - **LOW (L):** Assumed to be mostly harmless or unlikely to change protein behavior
 - **MODIFIER (MO):** Usually non-coding variants or variants affecting non-coding genes, where predictions are difficult or there is no evidence of impact

Clicking on the **Open in Exploration** button at the top right of this section will navigate the user to the Exploration page, filtered on this case.



File Summary Page

The File Summary page provides information a data file, including file properties like size, md5 checksum, and data format; information on the type of data included; links to the associated case and biospecimen; and information about how the data file was generated or processed.

The page also includes buttons to download the file, add it to the file cart, or (for BAM files) utilize the BAM slicing function.


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FL [5e18b02d-7e56-4f0d-b892-e7798eee5205_gdc_realn_rehead.bam](#)

[Add to Cart](#)
[BAM Slicing](#)
[Download](#)

File Properties

Name	5e18b02d-7e56-4f0d-b892-e7798eee5205_gdc_realn_rehead.bam
Access	controlled
UUID	3292b1e4-015d-4c24-b8a7-8535a78a59d4
Data Format	BAM
Size	12.02 GB
MD5 Checksum	edc7b09b6de9f1e133cecf2fc3e70156
Archive	--
Project	TCGA-BRCA

Data Information

Data Category	Raw Sequencing Data
Data Type	Aligned Reads
Experimental Strategy	RNA-Seq
Platform	Illumina

Associated Cases/Biospecimen

Entity ID	Entity Type	Case UUID	Annotations
TCGA-A2-A0D4-01A-11R-A00Z-07	aliquot	75113445-d2d6-44a0-866c-c9175e6d214b	0

Analysis

Analysis ID	e5127912-551e-4649-bf38-dc73ee3ac2b8
Workflow Type	STAR 2-Pass
Workflow Completion Date	2017-03-04
Source Files	0







Reference Genome

Genome Build	GRCh38.p0
Genome Name	GRCh38.d1.vd1

In the lower section of the screen, the following tables provide more details about the file and its characteristics:

- **Associated Cases / Biospecimen:** List of Cases or biospecimen the file is directly attached to.
- **Analysis and Reference Genome:** Information on the workflow and reference genome used for file generation.
- **Read Groups:** Information on the read groups associated with the file.
- **Metadata Files:** Experiment metadata, run metadata and analysis metadata associated with the file
- **Downstream Analysis Files:** List of downstream analysis files generated by the file

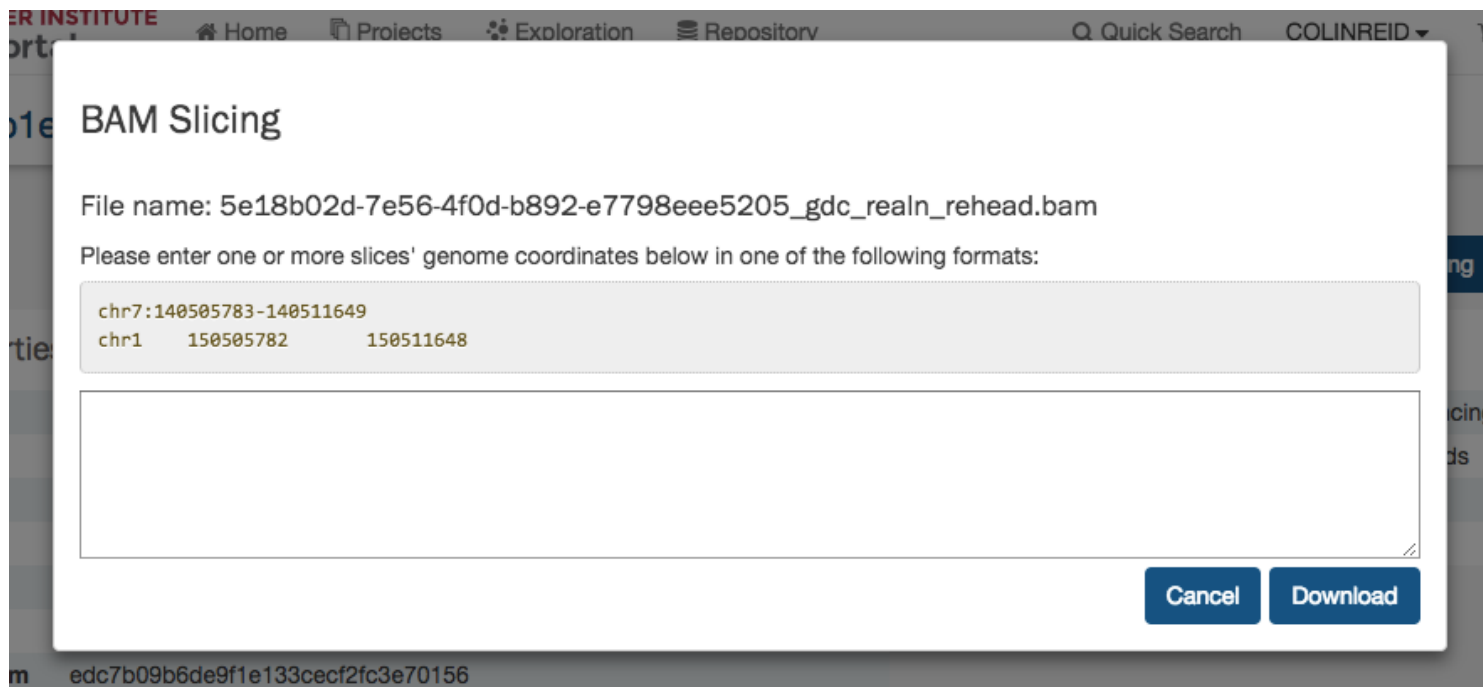
Read Groups					
Read Group ID	Is Paired End	Read Length	Library Name	Sequencing Center	Sequencing Date
33e057b9-e483-41ee-a837-32a5bf6a1e36	true	50	unknown	UNC	--

Downstream Analyses Files						
File Name	Data Category	Data Type	Data Format	Analysis workflow	File Size	Action
8b178cb1-d22e-4657-80c6-d7efcddf43a6.htseq.counts.gz	Transcriptome Profiling	Gene Expression Quantification	TXT	HTSeq - Counts	255.72 KB	 
8b178cb1-d22e-4657-80c6-d7efcddf43a6.FPKM-UQ.txt.gz	Transcriptome Profiling	Gene Expression Quantification	TXT	HTSeq - FPKM-UQ	521.22 KB	 
8b178cb1-d22e-4657-80c6-d7efcddf43a6.FPKM.txt.gz	Transcriptome Profiling	Gene Expression Quantification	TXT	HTSeq - FPKM	519.72 KB	 

Note: *The Legacy Archive* will not display “Workflow, Reference Genome and Read Groups” sections (these sections are applicable to the GDC harmonization pipeline only). However it may provide information on Archives and metadata files like MAGE-TABs and SRA XMLs. For more information, please refer to the section Legacy Archive.

BAM Slicing

BAM file detail pages have a “BAM Slicing” button. This function allows the user to specify a region of a BAM file for download. Clicking on it will open the BAM slicing window:



BAM Slicing

File name: 5e18b02d-7e56-4f0d-b892-e7798eee5205_gdc_realn_rehead.bam

Please enter one or more slices' genome coordinates below in one of the following formats:

chr7:140505783-140511649
chr1 150505782 150511648

Cancel Download

During preparation of the slice, the icon on the BAM Slicing button will be spinning, and the file will be offered for download to the user as soon as ready.

Chapter 5

Genes and Mutations

Gene and Mutation Summary Pages

Many parts of the GDC website contain links to Gene and Mutation summary pages. These pages display information about specific genes and mutations, along with visualizations and data showcasing the relationship between themselves, the projects, and cases within the GDC. The gene and mutation data that is visualized on these pages are produced from the Open-Access MAF files available for download on the GDC Portal.

Gene Summary Page

Gene Summary Pages describe each gene with mutation data and provides results related to the analyses that are performed on these genes.

Summary

The summary section of the gene page contains the following information:

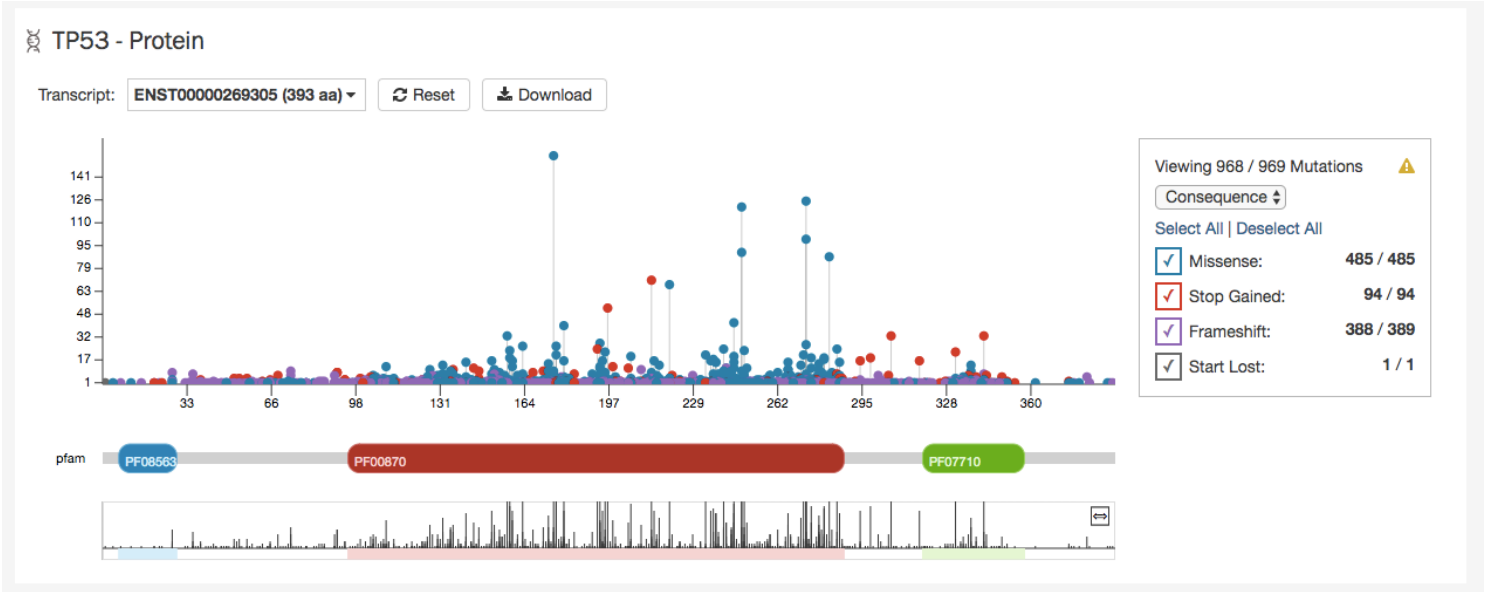
The screenshot shows the GDC Data Portal interface. At the top is a navigation bar with the NIH logo, 'NATIONAL CANCER INSTITUTE GDC Data Portal', and links for Home, Projects, Exploration, Analysis, and Repository. On the right of the navigation bar are links for Quick Search, Manage Sets, Login, Cart (0), and GDC Apps. Below the navigation bar is a header for the gene 'TP53'. The main content area is divided into two columns. The left column, titled 'Summary', contains a table with the following information: Symbol (TP53), Name (tumor protein p53), Synonyms (LFS1, p53), Type (protein_coding), Location (chr17:7661779-7687550 (GRCh38)), Strand (-), and Description (This gene encodes a tumor suppressor protein containing transcriptional activation, DNA binding, and oligomerization domains. The encoded protein responds to diverse cellular stresses to regulate expression of target genes, thereby inducing cell cycl...). There is a 'more' link at the bottom of the description. Below the description is an 'Annotation' link to 'Cancer Gene Census'. The right column, titled 'External References', contains a table with links to Entrez Gene (7157), Uniprotkb Swissprot (P04637), Hgnc (HGNC:11998), Omim Gene (191170), and Ensembl (ENSG00000141510).

Summary	
Symbol	TP53
Name	tumor protein p53
Synonyms	LFS1 p53
Type	protein_coding
Location	chr17:7661779-7687550 (GRCh38)
Strand	-
Description	This gene encodes a tumor suppressor protein containing transcriptional activation, DNA binding, and oligomerization domains. The encoded protein responds to diverse cellular stresses to regulate expression of target genes, thereby inducing cell cycl... more
Annotation	Cancer Gene Census

External References	
Entrez Gene	7157
Uniprotkb Swissprot	P04637
Hgnc	HGNC:11998
Omim Gene	191170
Ensembl	ENSG00000141510

- **Symbol:** The gene symbol
- **Name:** Full name of the gene
- **Synonyms:** Synonyms of the gene name or symbol, if available
- **Type:** A broad classification of the gene

Protein Viewer



Mutations and their frequency across cases are mapped to a graphical visualization of protein-coding regions with a lollipop plot. Pfam domains are highlighted along the x-axis to assign functionality to specific protein-coding regions. The bottom track represents a view of the full gene length. Different transcripts can be selected by using the drop-down menu above the plot.

The panel to the right of the plot allows the plot to be filtered by mutation consequences or impact. The plot will dynamically change as filters are applied. Mutation consequence and impact is denoted in the plot by color.

Note: The impact filter on this panel will not display the annotations for alternate transcripts.

The plot can be viewed at different zoom levels by clicking and dragging across the x-axis, clicking and dragging across the bottom track, or double clicking the pfam domain IDs. The **Reset** button can be used to bring the zoom level back to its original position. The plot can also be exported as a PNG image, SVG image or as JSON formatted text by choosing the **Download** button above the plot.

Most Frequent Mutations

The 20 most frequent mutations in the gene are displayed as a bar graph that indicates the number of cases that share each mutation.

Most Frequent Somatic Mutations

Showing 1 - 10 of 1,245 somatic mutations

[JSON] [TSV] [Save/Edit Mutation Set]

DNA Change	Type	Consequences	# Affected Cases in TP53	# Affected Cases Across the GDC	Impact (VEP)
chr17:g.7675088C>T	Substitution	Missense TP53 R175H	156 / 3,956 3.94%	156 / 10,188	M
chr17:g.7673803G>A	Substitution	Missense TP53 R273C	125 / 3,956 3.16%	125 / 10,188	M
chr17:g.7674220C>T	Substitution	Missense TP53 R248Q	121 / 3,956 3.06%	121 / 10,188	M
chr17:g.7673802C>T	Substitution	Missense TP53 R273H	99 / 3,956 2.50%	99 / 10,188	M
chr17:g.7674221G>A	Substitution	Missense TP53 R248W	90 / 3,956 2.28%	90 / 10,188	M
chr17:g.7673776G>A	Substitution	Missense TP53 R282W	87 / 3,956 2.20%	87 / 10,188	M
chr17:g.7674894G>A	Substitution	Stop Gained TP53 R213*	71 / 3,956 1.79%	71 / 10,188	H
chr17:g.7674872T>C	Substitution	Missense TP53 Y220C	68 / 3,956 1.72%	68 / 10,188	M
chr17:g.7674945G>A	Substitution	Stop Gained TP53 R196*	52 / 3,956 1.31%	52 / 10,188	H
chr17:g.7674230C>T	Substitution	Missense TP53 G245S	42 / 3,956 1.06%	42 / 10,188	M

Show 10 entries

A table is displayed below that lists information about each mutation including:

- **DNA Change:** The chromosome and starting coordinates of the mutation are displayed along with the nucleotide differences between the reference and tumor allele
- **Type:** A general classification of the mutation
- **Consequences:** The effects the mutation has on the gene coding for a protein (i.e. synonymous, missense, non-coding transcript)
- ___ Affected Cases in Gene:___ The number of affected cases, expressed as number across all mutations within the Gene
- ___ Affected Cases Across GDC:___ The number of affected cases, expressed as number across all projects. Choosing the arrow next to the percentage will expand the selection with a breakdown of each affected project
- **Impact:** A subjective classification of the severity of the variant consequence. This determined using [Ensembl VEP](#), [PolyPhen](#), and [SIFT](#). The categories are outlined [here](#).

Note: The Mutation UUID can be displayed in this table by selecting it from the drop-down represented by three parallel lines

Clicking the **Open in Exploration** button will navigate the user to the Exploration page, showing the same results in the table (mutations filtered by the gene).

Mutation Summary Page

The Mutation Summary Page contains information about one somatic mutation and how it affects the associated gene. Each mutation is identified by its chromosomal position and nucleotide-level change.

Summary

The screenshot shows the NIH GDC Data Portal interface. At the top, there's a navigation bar with links to Home, Projects, Exploration, Analysis, and Repository. Below this, the URL bar shows 'chr17:g.7675088C>T'. The main content area is divided into two panels. The left panel, titled 'Summary', contains a table with the following data:

UUID	8e30604f-3a45-5533-bdd7-0a4353700318
DNA Change	chr17:g.7675088C>T
Type	Single base substitution
Reference Genome Assembly	GRCh38
Allele in The Reference Assembly	C
Functional Impact (VEP)	Moderate ENST00000269305

The right panel, titled 'External References', lists links to dbSNP ([rs28934578](#)) and COSMIC ([COSM10648](#) and [COSM1640851](#)), with a link to '5 more' references.

- **ID:** A unique identifier (UUID) for this mutation
- **DNA Change:** Denotes the chromosome number, position, and nucleotide change of the mutation
- **Type:** A broad categorization of the mutation
- **Reference Genome Assembly:** The reference genome in which the chromosomal position refers to
- **Allele in the Reference Assembly:** The nucleotide(s) that compose the site in the reference assembly
- **Functional Impact:** A subjective classification of the severity of the variant consequence.

External References

A separate panel contains links to databases that contain information about the specific mutation. These include [dbSNP](#) and [COSMIC](#).

Consequences

The consequences of the mutation are displayed in a table. The set of consequence terms, defined by the [Sequence Ontology](#).

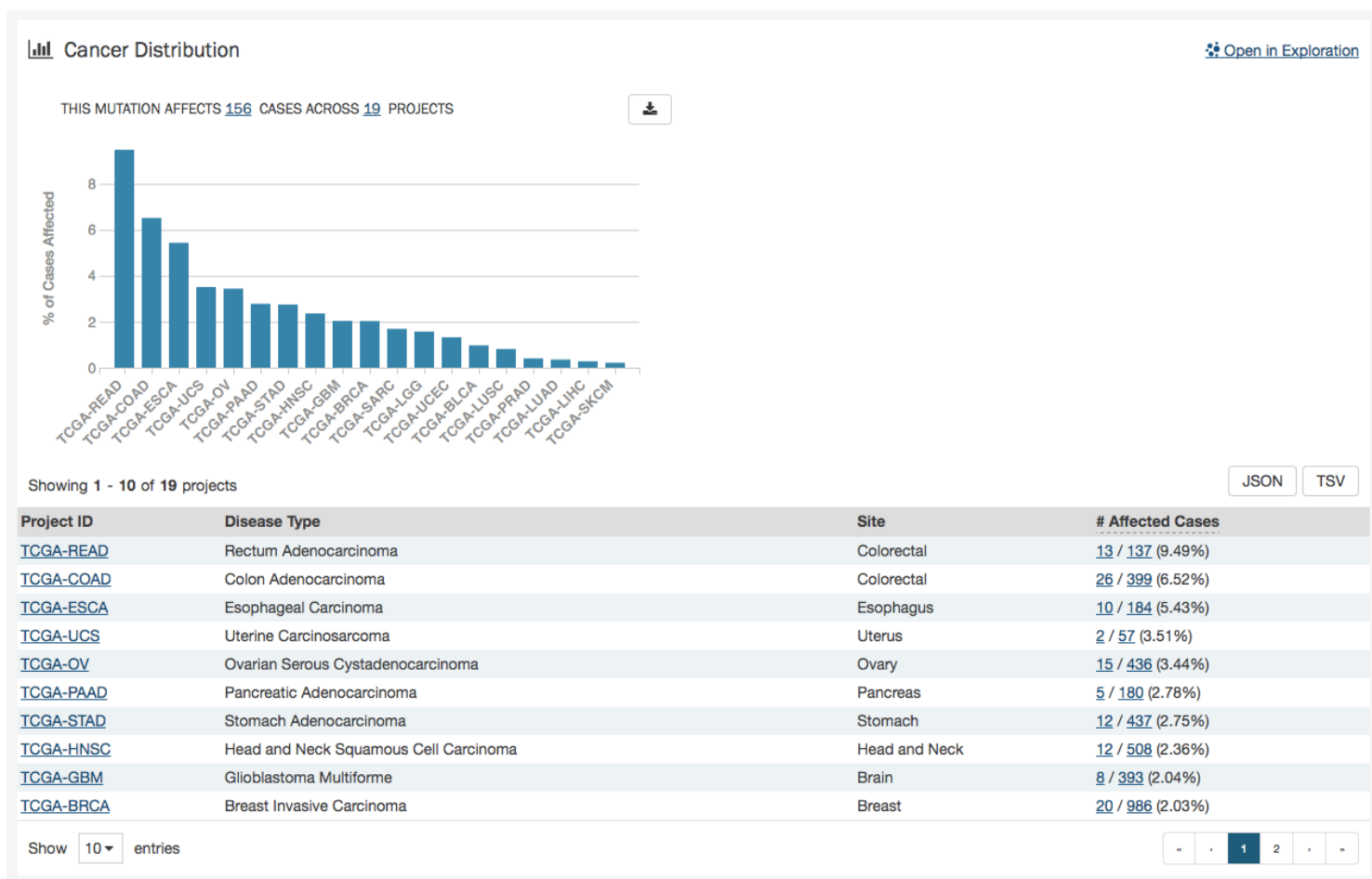
Consequences					
Showing 1 - 10 of 27					
JSON TSV					
Gene	AA Change	Consequence	Coding DNA Change	Strand	Transcript(s)
TP53	R136H	missense_variant	--	—	ENST00000610292
TP53	R136H	missense_variant	--	—	ENST00000610538
TP53	R136H	missense_variant	--	—	ENST00000619485
TP53	R136H	missense_variant	--	—	ENST00000620739
TP53	R136H	missense_variant	--	—	ENST00000622645
TP53	R164H	missense_variant	--	—	ENST00000615910
TP53	R16H	missense_variant	--	—	ENST00000618944
TP53	R16H	missense_variant	--	—	ENST00000619186
TP53	R16H	missense_variant	--	—	ENST00000610623
TP53	R175H	missense_variant	--	—	ENST00000359597
Show 10 entries					
<div> <div> <div></div> <div></div> <div>1</div> <div>2</div> <div>3</div> <div></div> <div></div> </div> </div>					

The fields that describe each consequence are listed below:

- **Gene:** The symbol for the affected gene
- **AA Change:** Details on the amino acid change, including compounds and position, if applicable
- **Consequence:** The biological consequence of each mutation
- **Coding DNA Change:** The specific nucleotide change and position of the mutation within the gene
- **Strand:** If the gene is located on the forward (+) or reverse (-) strand
- **Transcript(s):** The transcript(s) affected by the mutation. Each contains a link to the [Ensembl](#) entry for the transcript

Cancer Distribution

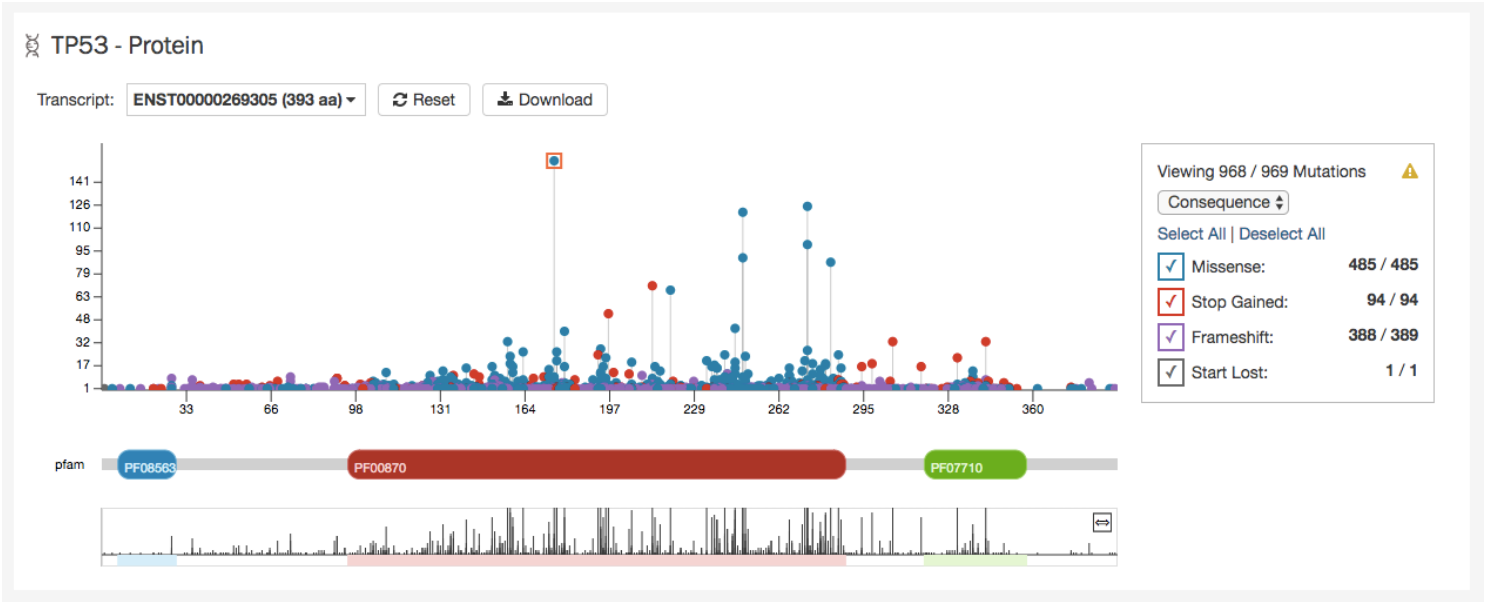
A table and bar graph shows how many cases are affected by the particular mutation. Each row/bar represents the number of cases for each project.



The table contains the following fields:

- **Project ID:** The ID for a specific project
- **Disease Type:** The disease associated with the project
- **Site:** The anatomical site affected by the disease
- **___ Affected Cases___:** The number of affected cases and total number of cases displayed as a fraction and percentage

Protein Viewer



The protein viewer displays a plot representing the position of mutations along the polypeptide chain. The y-axis represents the number of cases that exhibit each mutation, whereas the x-axis represents the polypeptide chain sequence. [Pfam domains](#) that were identified along the polypeptide chain are identified with colored rectangles labeled with pfam IDs. See the Gene Summary Page for additional details about the protein viewer.

The panel to the right of the plot allows the plot to be filtered by mutation consequences or impact. The plot will dynamically change as filters are applied. Mutation consequence and impact is denoted in the plot by color.

Note: The impact filter on this panel will not display the annotations for alternate transcripts.

The plot can be viewed at different zoom levels by clicking and dragging across the x-axis, clicking and dragging across the bottom track, or double clicking the pfam domain IDs. The **Reset** button can be used to bring the zoom level back to its original position. The plot can also be exported as a PNG image, SVG image or as JSON formatted text by choosing the **Download** button above the plot.

Chapter 6

Custom Set Analysis

Custom Set Analysis

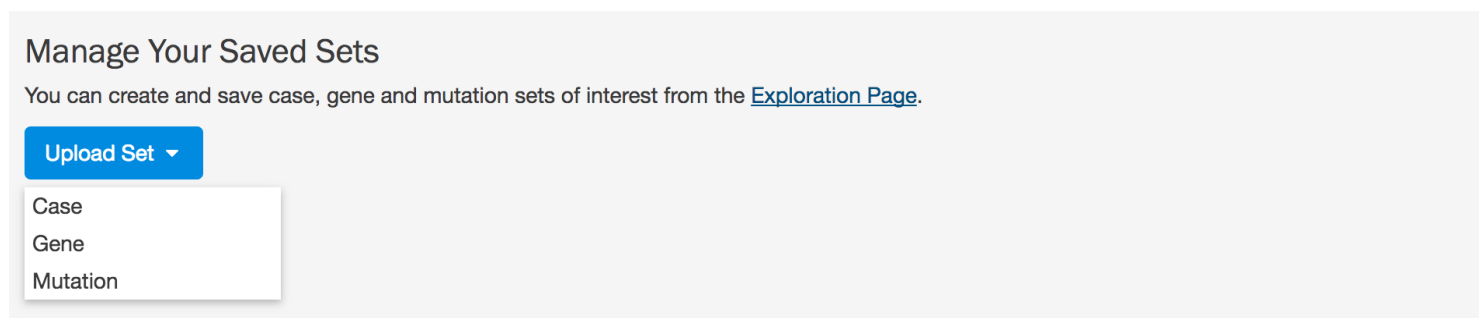
In addition to the Exploration page, the GDC Data Portal also has features used to save and compare sets of cases, genes, and mutations. These sets can either be generated with existing filters (e.g. males with lung cancer) or through custom selection (e.g. a user-generated list of case IDs).

Note that saving a set only saves the type of entity included in the set. For example, a saved case set will not include filters that were applied to genes or mutations. Please be aware that your custom sets are deleted during each new GDC data release. You can export them and re-upload them in the “Manage Sets” link at the top right of the Portal.

Generating a Cohort for Analysis

Cohort sets are completely customizable and can be generated for cases, genes, or mutations using the following methods:

Upload ID Set: This feature is available in the “Manage Sets” link at the top right of the Portal. Choose “Upload Set” and then select whether the set comprises cases, genes, or mutations. A set of IDs (IDs* or UUIDs) can then be uploaded in a text file or copied and pasted into the list of identifiers field along with a name identifying the set. Once the list of identifiers is uploaded, they are validated and grouped according to whether the identifier matched an existing GDC ID or did not match (“Unmatched”).

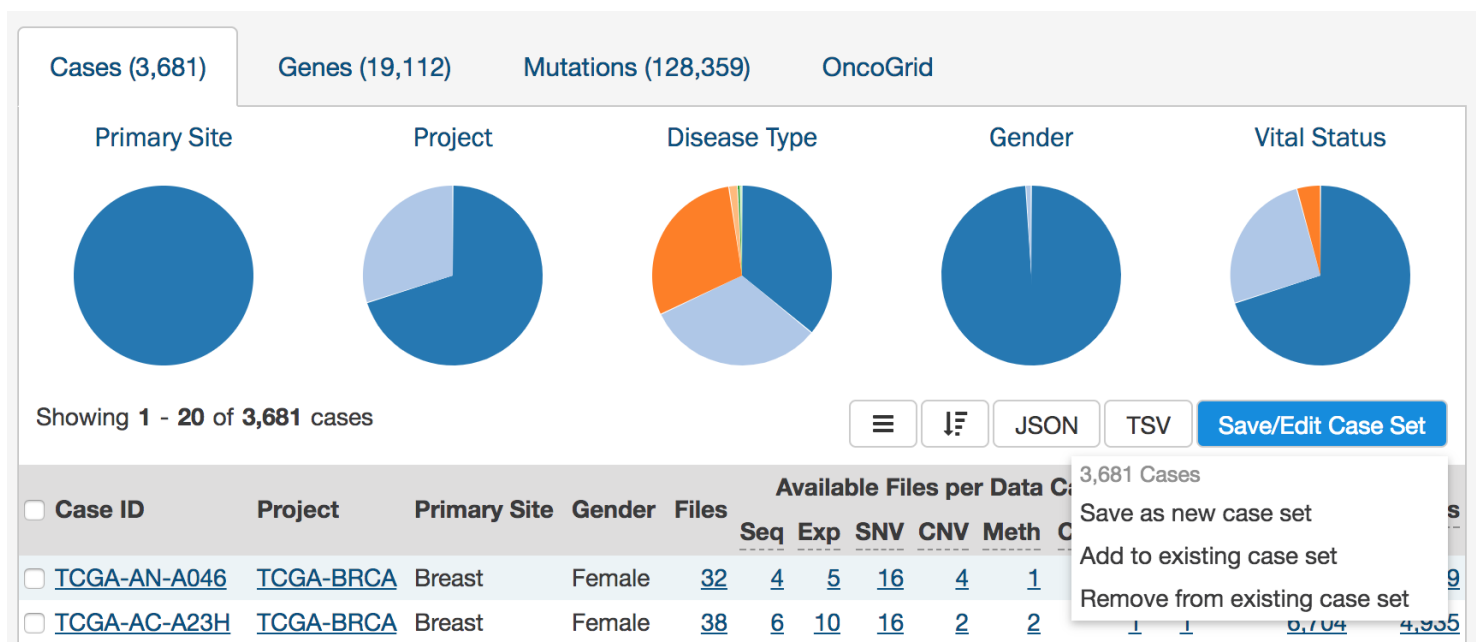


* This is referred to as a `submitter_id` in the GDC API, which is a non-UUID identifier such as a TCGA barcode.

Apply Filters in Exploration: Sets can be assembled using the existing filters in the Exploration page. They can be saved by choosing the “Save/Edit Case Set” button under the pie charts for case sets. This will prompt a decision to:

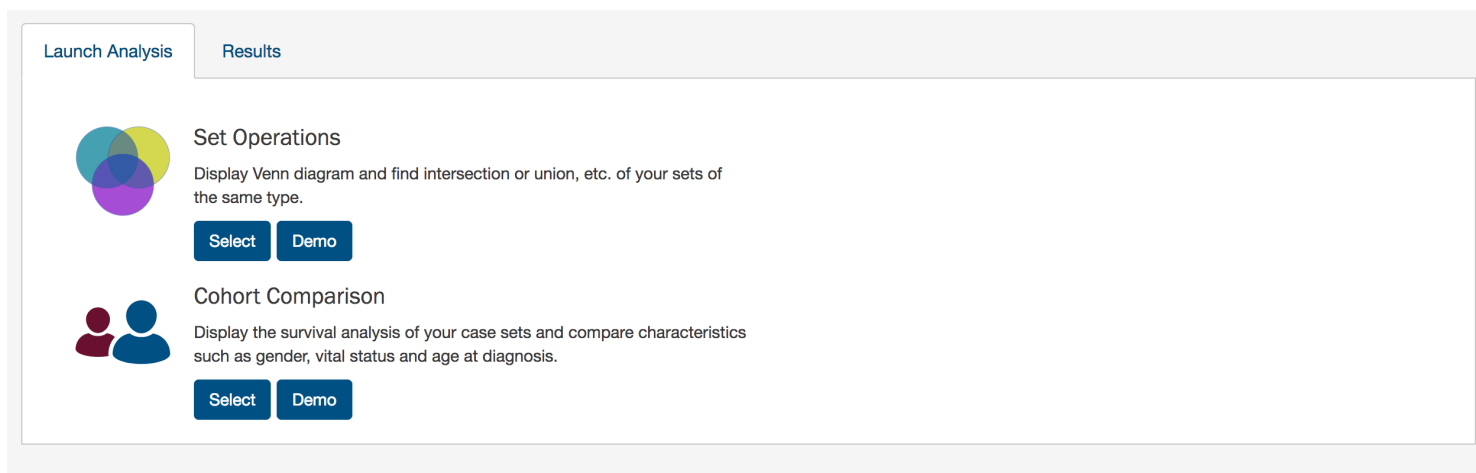
- Save as new case set
- Add to existing case set
- Remove from existing case set

Similarly, gene and mutation filters can be applied and saved in the Exploration page in the Genes and Mutations tab, respectively.



Analysis Page

Clicking on the **Analysis** button in the top toolbar will launch the Analysis Page which displays the various options available for comparing saved sets.

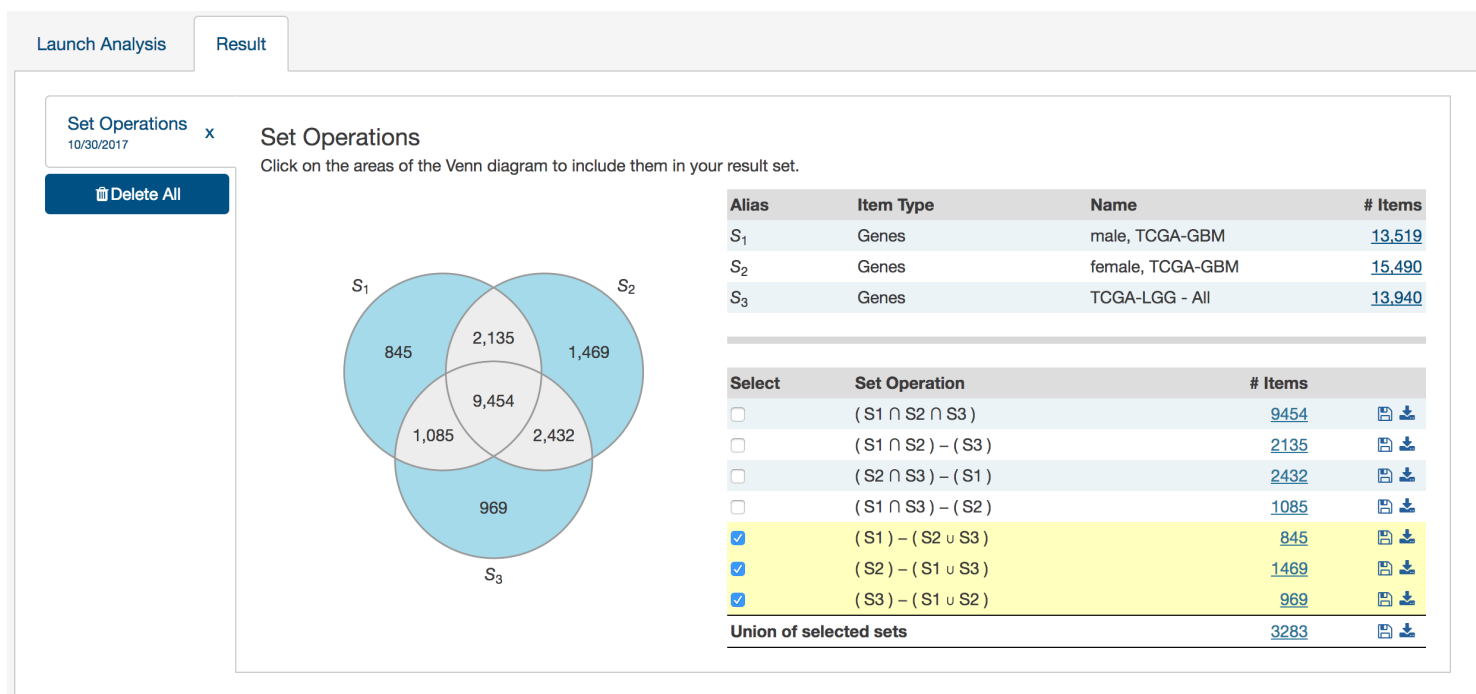


There are two tabs on this page:

- **Launch Analysis:** Where users can select either to do **Set Operations** or **Cohort Comparison**
- **Results:** Where users can view the results of current or previous set analyses

Analysis Page: Set Operations

Up to three sets of the same set type can be compared and exported based on complex overlapping subsets. The features of this page include:



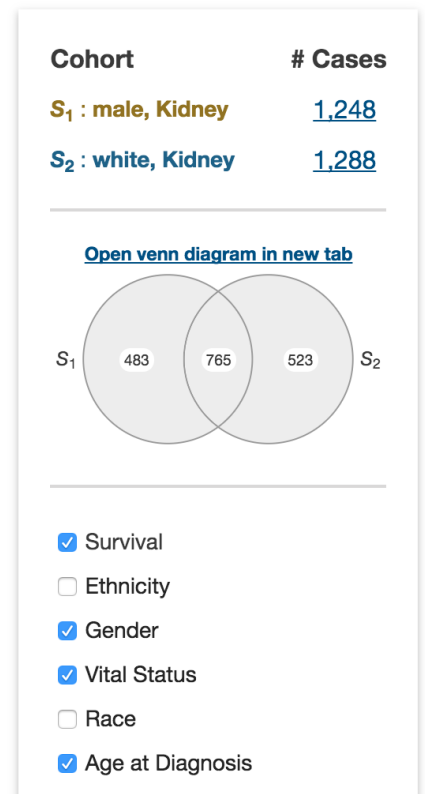
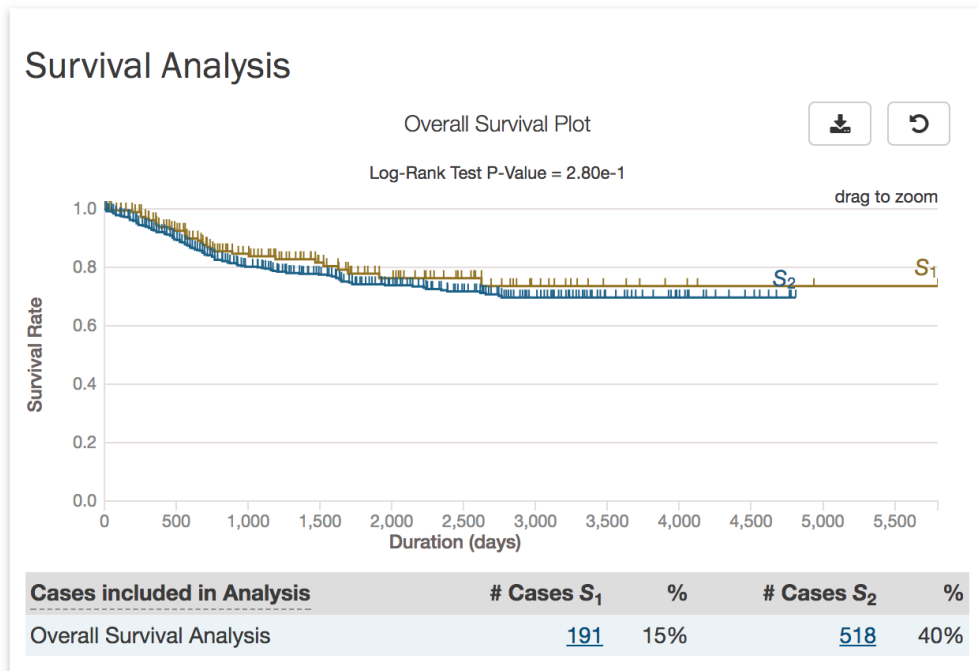
- **Venn Diagram:** Visually displays the overlapping items included within the three sets. Subsets based on overlap can be selected by clicking one or many sections of the Venn diagram. As sections of the Venn Diagram become highlighted in blue, their corresponding row in the overlap table becomes highlighted.
- **Summary Table:** Displays the alias, item type, and name for each set included in this analysis
- **Overlap Table:** Displays the number of overlapping items with set operations rather than a visual diagram. Subsets can be selected by checking boxes in the “Select” column, which will highlight the corresponding section of the Venn Diagram. As rows are selected, the “Union of selected sets” row is populated. Each row has an option to save the subset as a new set, export the set as a TSV, or view files in the repository. The links that correspond to the number of items in each row will open the cohort in the Exploration page.

Analysis Tab: Cohort Comparison

The “Cohort Comparison” analysis displays a series of graphs and tables that demonstrate the similarities and differences between two case sets. The following features are displayed for each two sets:

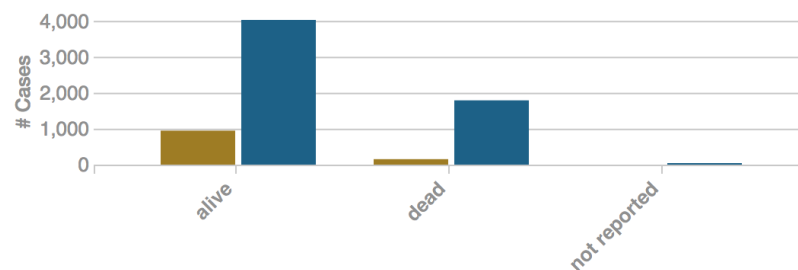
- A key detailing the number of cases in each cohort and the color that represents each (blue/gold)
- A Venn diagram, which shows the overlap between the two cohorts. The Venn diagram can be opened in a ‘Set Operations’ tab by choosing “Open venn diagram in new tab”
- A selectable [survival plot](#) that compares both sets with information about the percentage of represented cases

Cohort Comparison



- A breakdown of each cohort by selectable clinical facets with a bar graph and table. Facets include `vital_status`, `gender`, `race`, `ethnicity`, and `age_at_diagnosis`. A p-value (if it can be calculated from the data) that demonstrates whether the statuses are proportionally represented is displayed for the `vital_status`, `gender`, and `ethnicity` facets.

Vital Status



TSV

Vital Status	# Cases S ₁	%	# Cases S ₂	%
alive	945	86%	4025	69%
dead	152	14%	1788	31%
not reported	--	0%	2	0%

P-Value = 3.08e-30

Analysis Page: Results

The results of the previous analyses are displayed on this page.

Launch Analysis

Results

Set Operations

11/15/2017

x

Cohort Comparison

11/15/2017

x

Delete All

Cohort Comparison

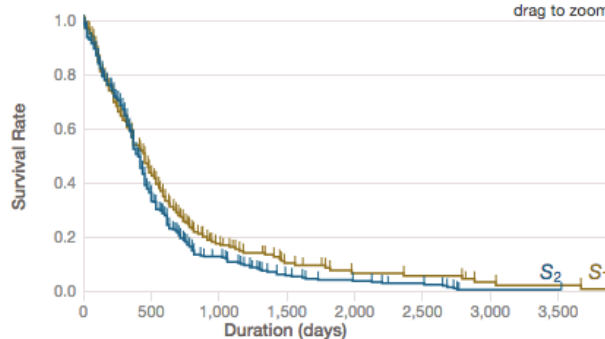
Survival Analysis

Overall Survival Plot

Log-Rank Test P-Value = 4.11e-2



drag to zoom



Cases included in Analysis	# Cases S ₁	%	# Cases S ₂	%
Overall Survival Analysis	230	100%	365	100%

Cohort

Cases

S₁ : female, TCGA-GBM 230

S₂ : male, TCGA-GBM 366

[Open venn diagram in new tab](#)



- ☒ Survival
- ☐ Ethnicity
- ☒ Gender
- ☒ Vital Status

Each tab at the left side of the page is labeled according to the analysis type and the date that the analysis was performed and can be reviewed as long as it is present. The “Delete All” button will remove all of the previous analyses.

Chapter 7

Annotations

Annotations

Annotations are notes added to individual cases, samples or files.

Annotations View

The Annotations View provides an overview of the available annotations and allows users to browse and filter the annotations based on a number of annotation properties (facets), such as the type of entity the annotation is attached to or the annotation category.

The view presents a list of annotations in tabular format on the right, and a facet panel on the left that allows users to filter the annotations displayed in the table. If facet filters are applied, the tabs on the right will display only the matching annotations. If no filters are applied, the tabs on the right will display information about all available data.

Clicking on an annotation ID in the annotations list will take the user to the [Annotation Detail Page](#).

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[Repository](#)

[Quick Search](#)
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[GDC Apps](#)

Annotation ID

Entity ID

Case UUID

Primary Site

☐ Brain
 ☐ Ovary
 ☐ Kidney
 ☐ Lung
 ☐ Bone Marrow

Project

☐ TCGA-OV
 ☐ TCGA-GBM
 ☐ TCGA-LAML
 ☐ TCGA-LUSC
 ☐ TCGA-KIRC

← Start searching by selecting a facet

Showing 1 - 20 of 2,361 annotations

UUID	Case UUID	Project	Entity Type	Entity UUID	Category	Classification	Date Created
f340f2cb	8449955f	TCGA-OV	aliquot	d34ae25d	Center QC failed	CenterNotification	2012-07-20T00:00:00
a19af36e	3c3d4ef0	TCGA-STAD	case	3c3d4ef0	Prior malignancy	Notification	2012-10-31T00:00:00
28d56f98	b22398fb	TCGA-LAML	case	b22398fb	Alternate sample pipeline	Notification	2012-11-13T00:00:00
45f4b491	b9356af2	TCGA-KICH	case	b9356af2	Item is noncanonical	Notification	2014-07-16T00:00:00
91a20b84	a608bf0e	TCGA-PRAD	case	a608bf0e	Pathology outside specification	Notification	2013-09-03T00:00:00
57fa6d63	810d293b	TCGA-LAML	case	810d293b	Alternate sample pipeline	Notification	2012-11-13T00:00:00
9630ca42	70fc222d	TCGA-GBM	analyte	58b0292b	Item is noncanonical	Notification	2012-07-12T00:00:00
09289721	5134c56f	TCGA-LUAD	case	5134c56f	Prior malignancy	Notification	2012-10-31T00:00:00
27e2fae4	b612d491	TCGA-COAD	aliquot	622b0cb2	Item is noncanonical	Notification	2012-06-26T00:00:00
de1b4736	acaaed1a	TCGA-THYM	case	acaaed1a	Neoadjuvant therapy	Notification	2014-06-30T00:00:00
e287571f	74b42897	TCGA-GBM	analyte	2c262bf4	Item is noncanonical	Notification	2012-07-12T00:00:00
251587ba	4261267c	TCGA-OV	case	4261267c	Item in special subset	Notification	2011-01-28T00:00:00
10801ee4	a46fb053	TCGA-PAAD	analyte	0a002336	Item is noncanonical	Notification	2011-05-15T00:00:00
f714dd7c	d47e214d	TCGA-PAAD	case	d47e214d	Item may not meet study protocol	Observation	2014-10-15T00:00:00
59b54d8	4dff4242	TCGA-LIHC	case	4dff4242	Neoadjuvant therapy	Notification	2014-09-02T00:00:00
63a066da	3ac45a71	TCGA-OV	analyte	1f57d960	Item is noncanonical	Notification	2011-03-14T00:00:00
729722c6	d0b7d446	TCGA-GBM	sample	1ba0cc6b	Item is noncanonical	Notification	2012-07-12T00:00:00
86860d1a	d0b78f3f	TCGA-BRCA	case	d0b78f3f	Neoadjuvant therapy	Notification	2014-06-16T00:00:00
38e7c8fb	548c3d4c	TCGA-GBM	aliquot	326b2ad0	Item flagged DNU	CenterNotification	2015-09-28T00:00:00
11f70dc9	8beee000	TCGA-THCA	case	8beee000	Item does not meet study protocol	Notification	2015-07-17T00:00:00

Show 20 entries

1

2

3

4

5

6

7

8

9

10

Facets Panel

The following facets are available to search for annotations:

- **Annotation ID:** Search using annotation ID
- **Entity ID:** Search using entity ID
- **Case UUID:** Search using case UUID
- **Primary Site:** Anatomical site of the cancer
- **Project:** A cancer research project, typically part of a larger cancer research program
- **Entity Type:** The type of entity the annotation is associated with: Patient, Sample, Portion, Slide, Analyte, Aliquot
- **Annotation Category:** Search by annotation category.
- **Annotation Created:** Search for annotations by date of creation.
- **Annotation Classification:** Search by annotation classification.

Annotation Categories and Classification

For more details about categories and classifications please refer to the [TCGA Annotations page on NCI Wiki](#).

Annotation Detail Page

The annotation entity page provides more details about a specific annotation. It is available by clicking on an annotation ID in Annotations View.

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AN f340f2cb-3cdc-5843-83d1-851d95d00f93

Summary

Annotation UUID	f340f2cb-3cdc-5843-83d1-851d95d00f93
Entity UUID	d34ae25d-8073-4129-a1b8-3f43e19af73b
Entity Barcode	TCGA-24-1923-01A-01R-1567-13
Entity Type	aliquot
Case UUID	8449955f-42d9-46f6-919a-5cc5d59e6284
Case Submitter ID	TCGA-24-1923
Project ID	TCGA-OV
Classification	CenterNotification
Category	Center QC failed
Created On	2012-07-20T00:00:00
Status	Approved

NOTES

RNA-seq:LOW 5/3 COVERAGE RATIO

Chapter 8

Advanced Search

Advanced Search

Only available in the Repository view, the Advanced Search page offers complex query building capabilities to identify specific set of cases and files.

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GDC Data Portal

Home

Projects

Exploration

Repository

Quick Search

Login

Cart 0

GDC Apps

CasesFiles

Add a Case/Biospecimen Filter

Case

Search for Case ID

Case Submitter ID

eg. TCGA-DD*, *DD*, TCGA-DD-AAVPGo!

Primary Site

Kidney1,001

Brain1,133

Nervous System1,127

Breast1,090

Lung1,009

24 More...

Program

TCGA11,015

TARGET3,000

Start searching by selecting a facet

Add All Files to CartDownload Manifest

Browse Annotations

Cases (14,551)Files (274,724)

470.59 TB

Primary SitesProjectsDisease TypeGenderVital Status

Showing 1 - 20 of 14,551 cases

JSONTSV

Cart	Case UUID	Submitter ID	Project	Primary Site	Gender	Files	Available Files per Data Category							Annotations
							Seq	Exp	SNV	CNV	Meth	Clinical	Bio	
<div>8dc378a8</div>	TARGET-30-PARAHE	TARGET-NBL	Nervous System	Male	1	0	0	0	0	0	0	1	0	
<div>d940e476</div>	TCGA-LK-A4O6	TCGA-MESO	Pleura	Female	32	4	5	16	4	1	1	1	0	
<div>1b1d2630</div>	TARGET-20-PASKIH	TARGET-AML	Blood	Female	2	0	0	0	0	0	1	1	0	
<div>5bc1db25</div>	TCGA-AF-6136	TCGA-READ	Colorectal	Female	32	4	5	16	4	1	1	1	0	

Overview: GQL

Advanced search allows, via Genomic Query Language (GQL), to use structured queries to search for files and cases.

79

Valid Query
Back to Facet Search
?

Start Typing Your Query....
Reset
Submit Query

Cases (14,551)
Files (274,724)
Browse Annotations

Showing 1 - 20 of 14,551 cases
JSON
TSV

Cart	Case UUID	Submitter ID	Project	Primary Site	Gender	Files	Available Files per Data Category								Annotations
							Seq	Exp	SNV	CNV	Meth	Clinical	Bio		
	53eac147	TCGA-4J-AA1J	TCGA-CESC	Cervix	Female	32	4	5	16	4	1	1	1	0	
	1ff85ada	TCGA-EW-A1P3	TCGA-BRCA	Breast	Female	32	4	5	16	4	1	1	1	0	
	d97ac1d1	TCGA-BP-4975	TCGA-KIRC	Kidney	Male	33	4	5	16	4	2	1	1	0	

A simple query in GQL (also known as a ‘clause’) consists of a **field**, followed by an **operator**, followed by one or more **values**. For example, the simple query `cases.primary_site = Brain` will find all cases for projects in which the primary site is Brain:

Valid Query
Back to Facet Search
?

`cases.primary_site = Brain`
Reset
Submit Query

Cases (1,133)
Files (22,260)
Browse Annotations

Showing 1 - 20 of 1,133 cases
JSON
TSV

Cart	Case UUID	Submitter ID	Project	Primary Site	Gender	Files	Available Files per Data Category								Annotations
							Seq	Exp	SNV	CNV	Meth	Clinical	Bio		
	30011f30	TCGA-32-2615	TCGA-GBM	Brain	Male	30	4	3	16	4	1	1	1	0	
	c2399f5d	TCGA-14-1454	TCGA-GBM	Brain	Female	7	0	0	0	4	1	1	1	1	
	ac3582a9	TCGA-06-1084	TCGA-GBM	Brain	Male	25	2	0	16	4	1	1	1	0	
	1c095c4a	TCGA-DU-A5TU	TCGA-LGG	Brain	Female	32	4	5	16	4	1	1	1	0	
	13d12179	TCGA-06-1802	TCGA-GBM	Brain	Male	25	2	0	16	4	1	1	1	0	
	69d56f2d	TCGA-26-5135	TCGA-GBM	Brain	Female	29	3	3	16	4	1	1	1	0	
	107335ed	TCGA-02-0333	TCGA-GBM	Brain	Female	4	0	0	0	2	0	1	1	0	

Note that it is not possible to compare two fields (e.g. `disease_type = project.name`).

Note: GQL is not a database query language. For example, GQL does not have a “SELECT” statement.

Switching between Advanced Search and Facet Filters

When accessing Advanced Search from Repository View, a query created using facet filters in Repository View will be automatically translated to an Advanced Search GQL Query.

A query created in Advanced Search is not translated back to facet filters. Clicking on “Back to Facet Search” will return the user to Data View and reset the filters.

Using the Advanced Search

When opening the advanced search page (via the Repository view), the search field will be automatically populated with facets filters already applied (if any).

This default query can be removed by pressing “Reset”.

Once the query has been entered and is identified as a “Valid Query”, click on “Search” to run your query.

Auto-complete

As a query is being written, the GDC Data Portal will analyze the context and offer a list of auto-complete suggestions. Auto-complete suggests both fields and values as described below.

Field Auto-complete

The list of auto-complete suggestions includes **all** available fields matching the user text input. The user has to scroll down to see more fields in the dropdown:

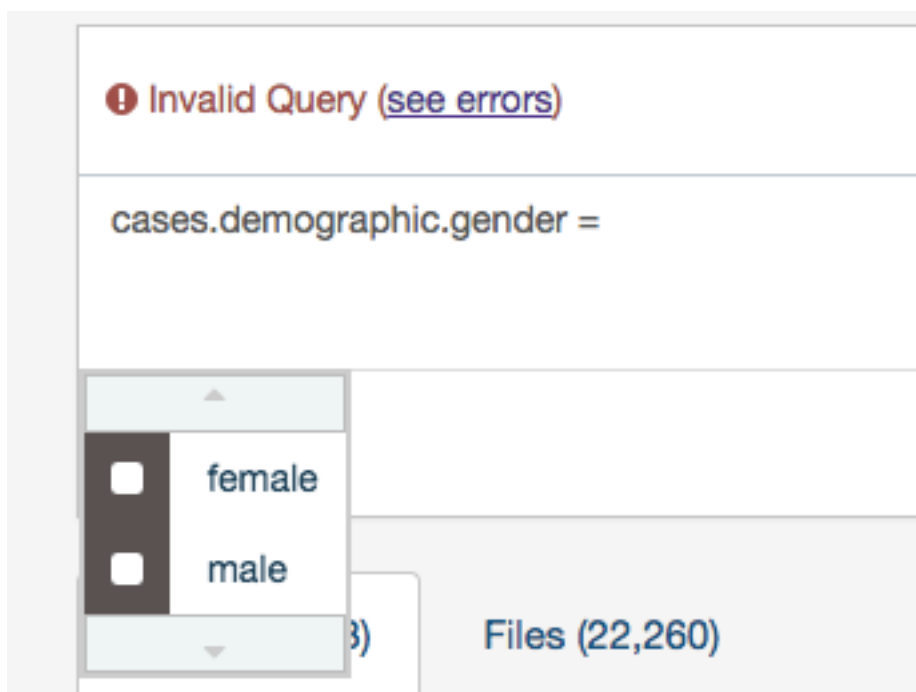
The screenshot shows the GDC Data Portal search interface. At the top, there is a message: "Invalid Query (see errors)" with a red icon. To the right of this message is a button labeled "Back to Facet Search" and a question mark icon. Below the message, the search input field contains the text "gen". A dropdown menu is open, displaying a list of field suggestions. Each suggestion consists of a small person icon, the field name, and a description. The suggestions are:

- demographic.gender** keyword
*Text designations that identify **gender**. Gender is described as the assemblage of properties that distinguish people on the basis of their societal roles. [Explanatory Comment 1: Identification of gender is based upon self-report and may come from a form, questionnaire, interview, etc.]*
- diagnoses.hpv_status** keyword
*The findings of the **oncogenic** HPV.*
- diagnoses.ldh_level_at_diagnosis** long
*The 2 decimal place numeric laboratory value measured, assigned or computed related to the assessment of **lactate dehydrogenase** in a specimen.*
- diagnoses.ldh_normal_range_upper** long
*The top value of the range of statistical characteristics that are supposed to represent accepted standard, non-pathological pattern for **lactate dehydrogenase** (units not specified).*
- diagnoses.prior_treatment** keyword
*A yes/no/unknown/not applicable indicator related to the administration of therapeutic **agents** received before the body specimen was collected.*
- diagnoses.treatments.therapeutic_agents** keyword

Value Auto-complete

The list of auto-complete suggestions includes top 100 values that match the user text input. The user has to scroll down to see more values in the dropdown.

The value auto-complete is not aware of the general context of the query, the system will display all available values in GDC for the selected field. It means the query could return 0 results depending of other filters.



Note: Quotes are automatically added to the value if it contains spaces.

Setting Precedence of Operators

You can use parentheses in complex GQL statements to enforce the precedence of operators.

For example, if you want to find all the open files in TCGA program as well as the files in TARGET program, you can use parentheses to enforce the precedence of the boolean operators in your query, i.e.:

```
1 (files.access = open and cases.project.program.name = TCGA) or cases.project.program.name = TARGET
```

Note: Without parentheses, the statement will be evaluated left-to-right.

Keywords

A GQL keyword is a word that joins two or more clauses together to form a complex GQL query.

List of Keywords:

- AND
- OR

Note: parentheses can be used to control the order in which clauses are executed.

AND Keyword

Used to combine multiple clauses, allowing you to refine your search.

Examples:

- Find all open files in breast cancer
cases.project.primary_site = Breast and files.access = open
- Find all open files in breast cancer and data type is copy number variation
cases.project.primary_site = Breast and files.access = open and files.data_type = "Copy number variation"

OR Keyword

Used to combine multiple clauses, allowing you to expand your search.

Note: **IN** keyword can be an alternative to OR and result in simplified queries.

Examples:

- Find all files that are raw sequencing data or raw microarray data:
`files.data_type = "Raw microarray data" or files.data_type = "Raw sequencing data"`
- Find all files where donors are male or vital status is alive:
`cases.demographic.gender = male or cases.diagnoses.vital_status = alive`

Operators

An operator in GQL is one or more symbols or words comparing the value of a field on its left with one or more values on its right, such that only true results are retrieved by the clause.

List of Operators and Query format

Operator	Description
=	Field EQUAL Value (String or Number)
!=	Field NOT EQUAL Value (String or Number)
<	Field LOWER THAN Value (Number or Date)
<=	Field LOWER THAN OR EQUAL Value (Number or Date)
>	Field GREATER THAN Value (Number or Date)
>=	Field GREATER THAN OR EQUAL Value (Number or Date)
IN	Field IN [Value 1, Value 2]
EXCLUDE	Field EXCLUDE [Value 1, Value 2]
IS MISSING	Field IS MISSING
NOT MISSING	Field NOT MISSING

“=” operator - EQUAL

The “=” operator is used to search for files where the value of the specified field exactly matches the specified value.

Examples:

- Find all files that are gene expression:
`files.data_type = "Gene expression"`
- Find all cases whose gender is female:
`cases.demographic.gender = female`

“!=” operator - NOT EQUAL

The “!=” operator is used to search for files where the value of the specified field does not match the specified value.

The “!=” operator will not match a field that has no value (i.e. a field that is empty). For example, ‘gender != male’ will only match cases who have a gender and the gender is not male. To find cases other than male or with no gender populated, you would need to type gender != male or gender is missing.

Example:

- Find all files with an experimental different from genotyping array:
files.experimental_strategy != “Genotyping array”

“>” operator - GREATER THAN

The “>” operator is used to search for files where the value of the specified field is greater than the specified value.

Example:

- Find all cases whose number of days to death is greater than 60:
cases.diagnoses.days_to_death > 60

“>=” operator - GREATER THAN OR EQUALS

The “>=” operator is used to search for files where the value of the specified field is greater than or equal to the specified value.

Example:

- Find all cases whose number of days to death is equal or greater than 60:
cases.diagnoses.days_to_death >= 60

“<” operator - LESS THAN

The “<” operator is used to search for files where the value of the specified field is less than the specified value.

Example:

- Find all cases whose age at diagnosis is less than 400 days:
cases.diagnoses.age_at_diagnosis < 400

“<=” operator - LESS THAN OR EQUALS

The “<=” operator is used to search for files where the value of the specified field is less than or equal to the specified value.

Example:

- Find all cases with a number of days to death less than or equal to 20:
cases.diagnoses.days_to_death <= 20

“IN” Operator

The “IN” operator is used to search for files where the value of the specified field is one of multiple specified values. The values are specified as a comma-delimited list, surrounded by brackets [].

Using “IN” is equivalent to using multiple ‘EQUALS (=)’ statements, but is shorter and more convenient. That is, typing ‘project IN [ProjectA, ProjectB, ProjectC]’ is the same as typing ‘project = “ProjectA” OR project = “ProjectB” OR project = “ProjectC”’.

Examples:

- Find all files in breast, breast and lung and cancer:
cases.project.primary_site IN [Brain, Breast,Lung]
- Find all files tagged with exon or junction or hg19:
files.data_type IN ["Aligned reads", "Unaligned reads"]

“EXCLUDE” Operator

The “EXCLUDE” operator is used to search for files where the value of the specified field is not one of multiple specified values.

Using “EXCLUDE” is equivalent to using multiple ‘NOT_EQUALS (!=)’ statements, but is shorter and more convenient. That is, typing ‘project EXCLUDE [ProjectA, ProjectB, ProjectC]’ is the same as typing ‘project != “ProjectA” OR project != “ProjectB” OR project != “ProjectC”’

The “EXCLUDE” operator will not match a field that has no value (i.e. a field that is empty). For example, ‘experimental strategy EXCLUDE [“WGS”,“WXS”]’ will only match files that have an experimental strategy **and** the experimental strategy is not “WGS” or “WXS”. To find files with an experimental strategy different from than “WGS” or “WXS” **or is not assigned**, you would need to type: files.experimental_strategy in [“WXS”,“WGS”] or files.experimental_strategy is missing.

Examples:

- Find all files where experimental strategy is not WXS, WGS, Genotyping array:
files.experimental_strategy EXCLUDE [WXS, WGS, “Genotyping array”]

“IS MISSING” Operator

The “IS” operator can only be used with “MISSING”. That is, it is used to search for files where the specified field has no value.

Examples:

- Find all cases where gender is missing:
cases.demographic.gender is MISSING

“NOT MISSING” Operator

The “NOT” operator can only be used with “MISSING”. That is, it is used to search for files where the specified field has a value.

Examples:

- Find all cases where race is not missing:
cases.demographic.race NOT MISSING

Special Cases

Date format

The date format should be the following: **YYYY-MM-DD** (without quotes).

Example:

```
1 files.updated_datetime > 2015-12-31
```

Using Quotes

A value must be quoted if it contains a space. Otherwise the advanced search will not be able to interpret the value.

Quotes are not necessary if the value consists of one single word.

- Example: Find all cases with primary site is brain and data type is copy number variation:
`cases.project.primary_site = Brain and files.data_type = "Copy number variation"`

Age at Diagnosis - Unit in Days

The unit for age at diagnosis is in **days**. The user has to convert the number of years to number of days.

The **conversion factor** is 1 year = 365.25 days

- Example: Find all cases whose age at diagnosis > 40 years old ($40 * 365.25$)
`cases.diagnoses.age_at_diagnosis > 14610`

Fields Reference

The full list of fields available on the GDC Data Portal can be found through the GDC API using the following endpoint:

https://api.gdc.cancer.gov/gql/_mapping

Alternatively, a static list of fields is available below (not exhaustive).

Files

- files.access
- files.acl
- files.archive.archive_id
- files.archive.revision
- files.archive.submitter_id
- files.center.center_id
- files.center.center_type
- files.center.code
- files.center.name
- files.center.namespace
- files.center.short_name
- files.data_format
- files.data_subtype
- files.data_type
- files.experimental_strategy
- files.file_id
- files.file_name
- files.file_size
- files.md5sum
- files.origin
- files.platform
- files.related_files.file_id
- files.related_files.file_name
- files.related_files.md5sum
- files.related_files.type

- files.state
- files.state_comment
- files.submitter_id
- files.tags

Cases

- cases.case_id
- cases.submitter_id
- cases.diagnoses.age_at_diagnosis
- cases.diagnoses.days_to_death
- cases.demographic.ethnicity
- cases.demographic.gender
- cases.demographic.race
- cases.diagnoses.vital_status
- cases.project.disease_type
- cases.project.name
- cases.project.program.name
- cases.project.program.program_id
- cases.project.project_id
- cases.project.state
- cases.samples.sample_id
- cases.samples.submitter_id
- cases.samples.sample_type
- cases.samples.sample_type_id
- cases.samples.shortest_dimension
- cases.samples.time_between_clamping_and_freezing
- cases.samples.time_between_excision_and_freezing
- cases.samples.tumor_code
- cases.samples.tumor_code_id
- cases.samples.current_weight
- cases.samples.days_to_collection
- cases.samples.days_to_sample_procurement
- cases.samples.freezing_method
- cases.samples.initial_weight
- cases.samples.intermediate_dimension
- cases.samples.is_ffpe
- cases.samples.longest_dimension
- cases.samples.oct_embedded
- cases.samples.pathology_report_uuid
- cases.samples.portions.analytes.a260_a280_ratio
- cases.samples.portions.analytes.aliquots.aliquot_id
- cases.samples.portions.analytes.aliquots.amount
- cases.samples.portions.analytes.aliquots.center.center_id
- cases.samples.portions.analytes.aliquots.center.center_type
- cases.samples.portions.analytes.aliquots.center.code
- cases.samples.portions.analytes.aliquots.center.name
- cases.samples.portions.analytes.aliquots.center.namespace
- cases.samples.portions.analytes.aliquots.center.short_name
- cases.samples.portions.analytes.aliquots.concentration
- cases.samples.portions.analytes.aliquots.source_center
- cases.samples.portions.analytes.aliquots.submitter_id
- cases.samples.portions.analytes.amount

- cases.samples.portions.analytes.analyte_id
- cases.samples.portions.analytes.analyte_type
- cases.samples.portions.analytes.concentration
- cases.samples.portions.analytes.spectrophotometer_method
- cases.samples.portions.analytes.submitter_id
- cases.samples.portions.analytes.well_number
- cases.samples.portions.center.center_id
- cases.samples.portions.center.center_type
- cases.samples.portions.center.code
- cases.samples.portions.center.name
- cases.samples.portions.center.namespace
- cases.samples.portions.center.short_name
- cases.samples.portions.is_ffpe
- cases.samples.portions.portion_id
- cases.samples.portions.portion_number
- cases.samples.portions.slides.number_proliferating_cells
- cases.samples.portions.slides.percent_eosinophil_infiltration
- cases.samples.portions.slides.percent_granulocyte_infiltration
- cases.samples.portions.slides.percent_inflam_infiltration
- cases.samples.portions.slides.percent_lymphocyte_infiltration
- cases.samples.portions.slides.percent_monocyte_infiltration
- cases.samples.portions.slides.percent_necrosis
- cases.samples.portions.slides.percent_neutrophil_infiltration
- cases.samples.portions.slides.percent_normal_cells
- cases.samples.portions.slides.percent_stromal_cells
- cases.samples.portions.slides.percent_tumor_cells
- cases.samples.portions.slides.percent_tumor_nuclei
- cases.samples.portions.slides.section_location
- cases.samples.portions.slides.slide_id
- cases.samples.portions.slides.submitter_id
- cases.samples.portions.submitter_id
- cases.samples.portions.weight

Chapter 9

Authentication

Authentication

Overview

The GDC Data Portal provides granular metadata for all datasets available in the GDC. Any user can see a listing of all available data files, including controlled-access files. The GDC Data Portal also allows users to download open-access files without logging in. However, downloading of controlled-access files is restricted to authorized users and requires authentication.

Logging into the GDC

To login to the GDC, users must click on the **Login** button on the top right of the GDC website.

After clicking Login, users authenticate themselves using their eRA Commons login and password. If authentication is successful, the eRA Commons username will be displayed in the upper right corner of the screen, in place of the “Login” button.

Upon successful authentication, GDC Data Portal users can:

- see which controlled-access files they have access to;
- download controlled-access files directly from the GDC Data Portal;
- download an authentication token for use with the GDC Data Transfer Tool or the GDC API.

Controlled-access files are identified using a “lock” icon:

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GDC Data Portal

[Home](#)
[Projects](#)
[Exploration](#)
[Repository](#)

[Login](#)
[Cart](#)
[GDC Apps](#)

Harmonized Cancer Datasets

Genomic Data Commons Data Portal

Get Started by Exploring:

[Projects](#)
[Exploration](#)
[Repository](#)

Data Portal Summary

[Data Release 7.0 - June 27, 2017](#)

PROJECTS
39

FILES
274,724

PRIMARY SITES
29

GENES
22,144

CASES
14,551

MUTATIONS
3,115,606

Cases by Primary Site

Primary Site	Cases
Adrenal Gland	~100
Bile Duct	~100
Bladder	~100
Blood	~100
Bone	~100
Bone Marrow	~100
Brain	~100
Breast	~100
Cervix	~100
Colorectal	~100
Esophagus	~100
Eye	~100
Head and Neck	~100
Kidney	~100
Liver	~100
Lung	~100
Lymph Nodes	~100
Nervous System	~100
Ovary	~100
Pancreas	~100
Pleura	~100
Prostate	~100
Skin	~100
Soft Tissue	~100
Stomach	~100
Testis	~100
Thymus	~100
Thyroid	~100
Uterus	~100

GDC Applications

The GDC Data Portal is a robust data-driven platform that allows cancer researchers and bioinformaticians to search and download cancer data for analysis. The GDC applications include:

[Data Portal](#)
[Website](#)
[Data Transfer Tool](#)
[API](#)
[Data Submission Portal](#)
[Documentation](#)
[Legacy Archive](#)

Figure 9.1: Login

Cases (11,736)
Files (45,988)

Primary Site

Project ID

Data Category

[Show More](#)

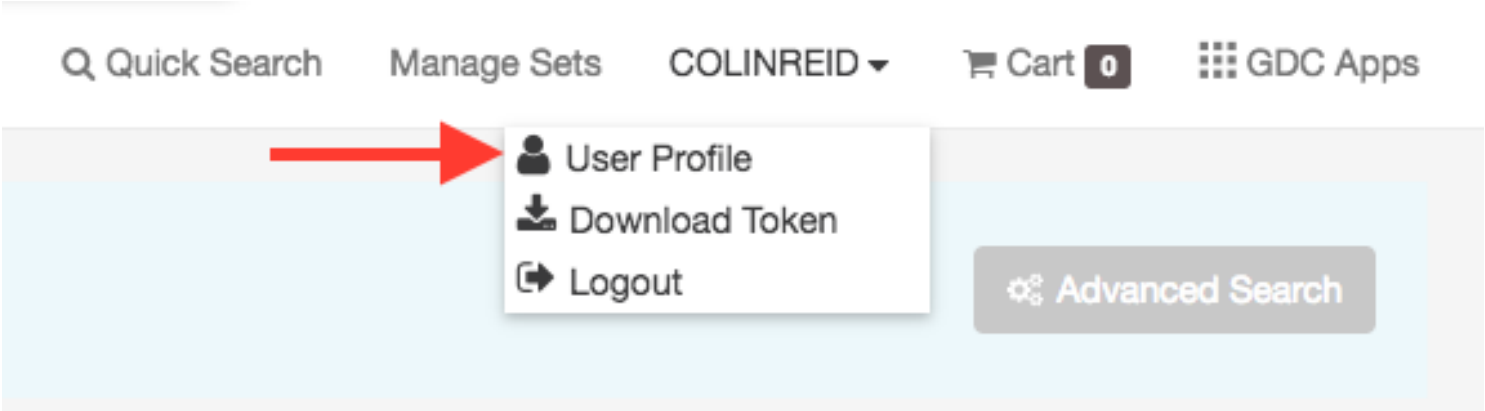
Showing 1 - 20 of 45,988 files

Access	File Name	Cases	Project
controlled	TCGA-AR-A0U2-01A-11D-A10G-09_IlluminaGA-DNASeq_exome_HOLD_QC_PENDING_gdc_realn.bam	1	TCGA-BRCA
controlled	584124a0-58b8-45bd-bc2c-f5b7eb4b6bc4_gdc_realn_reheald.bam	1	TCGA-UCEC
controlled	C494.TCGA-HT-8104-01A-11D-2395-08.1_gdc_realn.bam	1	TCGA-LGG
controlled	04a3d925-2d80-44b2-aea0-d0d46924f120_gdc_realn_rehea	1	TCGA-SKCM

The rest of this section describes controlled data access features of the GDC Data Portal available to authorized users. For more information about open and controlled-access data, and about obtaining access to controlled data, see [Data Access Processes and Tools](#).

User Profile

After logging into the GDC Portal, users can view which projects they have access to by clicking the **User Profile** section in the dropdown menu in the top corner of the screen.



Clicking this button shows the list of projects.

The image shows the 'User Profile' page for user COLINREID. It displays a table of projects with columns for Project ID, read, create, update, release, download, _member_, read_report, and delete. The table lists several projects, all of which the user has access to (indicated by checkmarks in the 'read', 'create', 'update', 'release', 'download', and '_member_' columns).

Project ID	read	create	update	release	download	_member_	read_report	delete
phs001179	✓	✓	✓	✓	✓	✓	✓	✓
phs000345	✓	✓	✓	✓	✓	✓	✓	✓
CCLEV2	✓	✓	✓	✓	✓	✓	✓	✓
phs001449	✓	✓	✓	✓	✓	✓	✓	✓
phs001134	✓	✓	✓	✓	✓	✓	✓	✓
phs001444	✓	✓	✓	✓	✓	✓	✓	✓

GDC Authentication Tokens

The GDC Data Portal provides authentication tokens for use with the GDC Data Transfer Tool or the GDC API. To download a token:

1. Log into the GDC using your eRA Commons credentials
2. Click the username in the top right corner of the screen
3. Select the “Download token” option

A new token is generated each time the **Download Token** button is clicked.

For more information about authentication tokens, see [Data Security](#).

NOTE: The authentication token should be kept in a secure location, as it allows access to all data accessible by the associated user account.

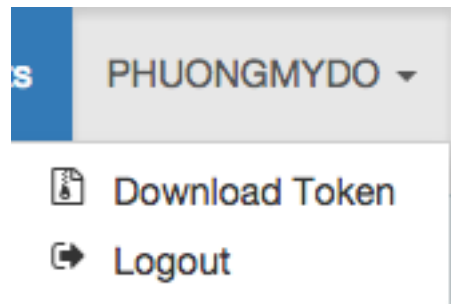


Figure 9.2: Token Download Button

Logging Out

To log out of the GDC, click the username in the top right corner of the screen, and select the Logout option.

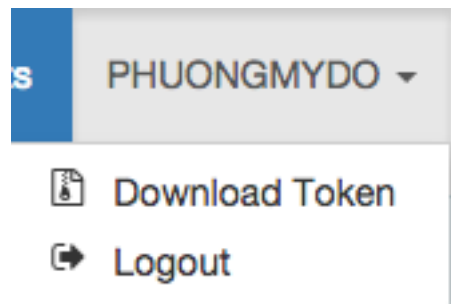


Figure 9.3: Logout link

Chapter 10

File Cart

Cart and File Download

Overview

While browsing the GDC Data Portal, files can either be downloaded individually from [file detail pages](#) or collected in the file cart to be downloaded as a bundle. Clicking on the shopping cart icon that is next to any item in the GDC will add the item to your cart.

GDC Cart

FILES
4

CASES
3

FILE SIZE
22.87 GB

File Counts by Project

Project	Cases (n=3)	Files (n=4)	File Size (Σ=22.87 GB)
TCGA-GBM	3	4	22.87 GB

File Counts by Authorization Level

Level	Files (n=4)	File Size (Σ=22.87 GB)
Authorized	2	44.77 KB
Unauthorized	2	22.87 GB

How to download files in my Cart?

Download Manifest:
Download a manifest for use with the [GDC Data Transfer Tool](#). The GDC Data Transfer Tool is recommended for transferring large volumes of data.

Download Cart:
Download Files in your Cart directly from the Web Browser.

Sample Sheet

Metadata

Download

Remove From Cart

Cart Items

Showing 1 - 4 of 4 files

Access

File Name

Cases

Project

Data Category

Data Format

File Size

Annotations

[C484.TCGA-19-5956-11A-01D-1696-08.3_gdc_realn.bam](#)

[SEXES_p_TCGA_b111_SNP_N_GenomeWideSNP_6_D12_780496.nocnv_grch38.seg.txt](#)

[SEXES_p_TCGA_b111_SNP_N_GenomeWideSNP_6_F01_780458.grch38.seg.txt](#)

[C484.TCGA-19-5956-01A-11D-1696-08.3_gdc_realn.bam](#)

1

1

1

1

TCGA-GBM

TCGA-GBM

TCGA-GBM

TCGA-GBM

Raw Sequencing Data

Copy Number Variation

Copy Number Variation

Raw Sequencing Data

BAM

TXT

TXT

BAM

11.81 GB

5.46 KB

39.32 KB

11.06 GB

0

0

0

0

Show

20

entries

-

.

1

.

-

Cart Summary

The cart page shows a summary of all files currently in the cart:

- Number of files
- Number of cases associated with the files
- Total file size

The Cart page also displays two tables:

- **File count by project:** Breaks down the files and cases by each project
- **File count by authorization level:** Breaks down the files in the cart by authorization level. A user must be logged into the GDC in order to download 'Controlled-Access files'

The cart also directs users how to download files in the cart. For large data files, it is recommended that the GDC Data Transfer Tool be used.

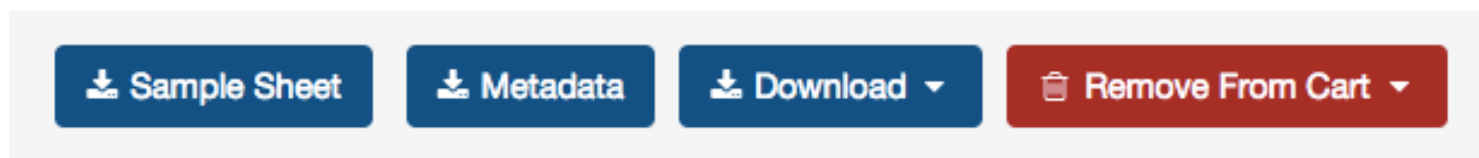
Cart Items

Access	File Name	Cases	Project	Data Category	Data Format	File Size	Annotations
controlled	C484.TCGA-19-5956-11A-01D-1696-08.3_gdc_realn.bam	1	TCGA-GBM	Raw Sequencing Data	BAM	11.81 GB	0
open	SEXES_p_TCGA_b111_SNP_N_GenomeWideSNP_6_D12_780496.nocnv_grch38.seg.txt	1	TCGA-GBM	Copy Number Variation	TXT	5.46 KB	0
open	SEXES_p_TCGA_b111_SNP_N_GenomeWideSNP_6_F01_780458.grch38.seg.txt	1	TCGA-GBM	Copy Number Variation	TXT	39.32 KB	0
controlled	C484.TCGA-19-5956-01A-11D-1696-08.3_gdc_realn.bam	1	TCGA-GBM	Raw Sequencing Data	BAM	11.06 GB	0

The Cart Items table shows the list of all the files that were added to the Cart. The table gives the following information for each file in the cart:

- **Access:** Displays whether the file is open or controlled access. Users must login to the GDC Portal and have the appropriate credentials to access these files.
- **File Name:** Name of the file. Clicking the link will bring the user to the file summary page.
- **Cases:** How many cases does the file contain. Clicking the link will bring the user to the case summary page.
- **Project:** The Project that the file belongs to. Clicking the link will bring the user to the Project summary page.
- **Category:** Type of data
- **Format:** The file format
- **Size:** The size of the file
- **Annotations:** Whether there are any annotations

Download Options



There are a few buttons on the Cart page that allow users to download files. The following download options are available:

- **Sample Sheet:** Downloads a tab-separated file which contains the associated case/sample IDs and sample type for each file in the cart.
- **Metadata:** GDC harmonized clinical, biospecimen, and file metadata associated with the files in the cart.

- **Download Manifest:** Download a manifest file for use with the GDC Data Transfer Tool to download files. A manifest file contains a list of the UUIDs that correspond to the files in the cart.
- **Download Cart:** Download the files in the Cart directly through the browser. Users have to be cautious of the amount of data in the cart since this option will not optimize bandwidth and will not provide resume capabilities.
- **SRA XML, MAGE-TAB:** This option is available in the GDC Legacy Archive only. It is used to download metadata files associated with the files in the cart.

The cart allows users to download up to 5 GB of data directly through the web browser. This is not recommended for downloading large volumes of data, in particular due to the absence of a retry/resume mechanism. For downloads over 5 GB we recommend using the GDC Data Transfer Tool.

Note: when downloading multiple files from the cart, they are automatically bundled into one single Gzipped (.tar.gz) file.

GDC Data Transfer Tool

The **Download Manifest** button will download a manifest file that can be imported into the GDC Data Transfer Tool. Below is an example of the contents of a manifest file used for download:

id	filename	md5	size	state		
2	4ea9c657-8f85-44d0-9a77-ad59cced8973				mdanderson.org_ESCA.MDA_RPPA_Core.mage-tab.1.1.0.tar.gz	
	2516051	live				
3	b8342cd5-330e-440b-b53a-1112341d87db				mdanderson.org_SARC.MDA_RPPA_Core.mage-tab.1.1.0.tar.gz	
	4523632	live				
4	c57673ac-998a-4a50-a12b-4cac5dc3b72e				mdanderson.org_KIRP.MDA_RPPA_Core.mage-tab.1.2.0.tar.gz	
	4195746	live				
5	3f22dd8d-59c8-43a4-89cf-3b595f2e5a06				14-3-3_beta-R-V_GBL1112940.tif	56df0e4b4fc092fc3643bd2e316ac05b
	6257840	live				
6	7ce05059-9197-4d38-830f-04356f5f851a				14-3-3_beta-R-V_GBL11066140.tif	6abfee483974bc2e61a37b5499ae9a07
	6261580	live				
7	8e00d22a-ca6f-4da8-a1c3-f23144cb21b7				14-3-3_beta-R-V_GBL1112940.tif	56df0e4b4fc092fc3643bd2e316ac05b
	6257840	live				
8	96487cd7-8fa8-4bee-9863-17004a70b2e9				14-3-3_beta-R-V_GBL1112940.tif	56df0e4b4fc092fc3643bd2e316ac05b
	6257840	live				

The Manifest contains a list of the file UUIDs in the cart and can be used together with the GDC Data Transfer Tool to download all files.

Information on the GDC Data Transfer Tool is available in the [GDC Data Transfer Tool User's Guide](#).

Individual Files Download

Similar to the files page, each row contains a download button to download a particular file individually.

Controlled Files

If a user tries to download a cart containing controlled files and without being authenticated, a pop-up will be displayed to offer the user either to download only open access files or to login into the GDC Data Portal through eRA Commons. See Authentication for details.

Access Error

You are attempting to download files that you are not authorized to access.

2 files that you are authorized to download.

2 files that you are not authorized to download.

Please [Login](#)

Cancel

Download 2 authorized files

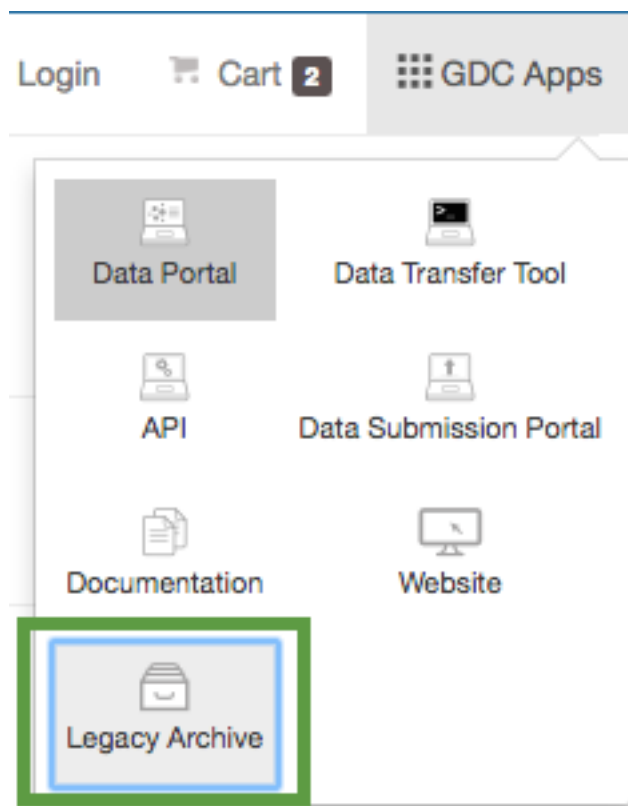
Chapter 11

Legacy Archive

Legacy Archive

The GDC Legacy Archive hosts unharmonized legacy data from repositories that predate the GDC (e.g. CGHub). Legacy data is not actively maintained, processed, or harmonized by the GDC. Legacy users are encouraged to migrate to harmonized datasets.

The GDC Legacy Archive can be accessed from the GDC Data Portal front page as well as from the “GDC Apps” menu.



Overview

The GDC Legacy Archive contains a limited set of features of the GDC Data Portal:

- **Facet search:** Ability to look for legacy files or legacy annotations based on case, file and annotation facets.
- **File and Annotation tables:** List of all the legacy files and list of all the legacy annotations.
- **File and Annotation detail pages:** Information page for each legacy file and annotation.

- **Cart:** The GDC Legacy Archive and the GDC Data Portal are separate systems with separate download carts.

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GDC Legacy Archive

The legacy data is the original data that uses the old genome build hg19 as produced by the original submitter. The legacy data is not actively being updated in any way. Users should migrate to the harmonized data. Please visit the [GDC Data Portal](#).

Files
Annotations
FGERTHOFFERT
Cart 0

Cases
Files

Add a Cases Filter

Case

Search for Case Barcode or Uuid

Primary Site

☐ Blood 2,131
☐ Kidney 1,099
☐ Nervous System 1,367
☐ Brain 1,133
☐ Breast 1,086
25 More...

Cancer Program

☐ TCGA 11,323
☐ TARGET 3,061
☐ GDC 6

Project

☐ TARGET-NBL 1,178
☐ TCGA-BRCA 1,093

Files (582,847)

Start searching by selecting a facet

Files
Showing 1 - 20 of 582,847 files

	Access	File Name	Cases	Project	Data Category	Data Format	Size	Annotations
	Controlled	000aa811c15656604161e8f0e...	1	TCGA-SARC	Raw sequencing data	BAM	6.64 GB	0
	Controlled	0017ba4c33a07ba807b29140...	1	TCGA-BRCA	Raw sequencing data	BAM	12.08 GB	1
	Controlled	00286ada95aed3619130f0d87...	1	TCGA-UCEC	Raw sequencing data	BAM	8.92 GB	1
	Controlled	003860a34c9b244a5d8435b2...	1	TCGA-BRCA	Raw sequencing data	BAM	7.71 GB	0
	Controlled	0047eb6e338c99ed7e7e3998...	1	TCGA-BRCA	Raw sequencing data	BAM	4.38 GB	0
	Controlled	0048523803f0838a87062c466...	1	TCGA-CHOL	Raw sequencing data	BAM	5.31 GB	0
	Controlled	00586a9e7ba372bfa76a60ba3...	1	TCGA-UCEC	Raw sequencing data	BAM	7.04 GB	0
	Controlled	006178ba37345d0e416e1a45...	1	TCGA-ESCA	Raw sequencing data	BAM	5.45 GB	0
	Controlled	00649547f0ace32d81d190498...	1	TCGA-BRCA	Raw sequencing data	BAM	9.64 GB	0
	Controlled	00925769611d88cb03797982...	1	TCGA-BRCA	Raw sequencing data	BAM	7.42 GB	0

File Page

The file page of the GDC Legacy Archive is similar to the [file page of the GDC Data Portal](#). It does not include the Workflow, Reference Genome, and Read Groups sections as these are only applicable to harmonized data available in the GDC Data Portal. The Legacy Archive includes additional archive information as described below.

a8337275-247b-44fb-a495-e0832089d461

Add to Cart
Download

File Properties

Name	0140.CEL
Submitter ID	--
Access	Controlled
UUID	a8337275-247b-44fb-a495-e0832089d461
Data format	CEL
Size	66 MB
MD5 Checksum	1b3583d556b238322acaf8cdc023c35
Published	--
Uploaded	1970-01-17
State	Live
Archive	50dd33bc-3137-487f-af59-2d9457b4da5f (25 files)

Data Information

Data Category	Raw Microarray Data
Data Type	Raw Intensities
Experimental Strategy	Exon array
Platform	HuEx-1_0-st-v2
Data Submitter	LBL
Tag	--

Associated Cases / Biospecimen

Metadata Files

Archive

If a file was originally produced as part of an archive containing other files, the archive information (Archive ID and number of files in the archive) is displayed in the file properties and, if selected, the user will see a list of files containing all other files in that archive.

Metadata files

If a file has any associated MAGE-TAB or SRA XML metadata files, these files will be listed at the bottom of the page. These files will can be downloaded directly from here. Alternatively, metadata files can be downloaded from the file cart.

File Cart

The file cart in the GDC Legacy Archive is analogous to the file cart of the GDC Data Portal. It provides an additional button to download any SRA-XML and MAGE-TAB metadata files associated with the files in the cart.

Chapter 12

Release Notes

Data Portal Release Notes

Release 1.11.0

- **GDC Product:** GDC Data Portal
- **Release Date:** December 21, 2017

New Features and Changes

- Updated UI to support SIFT and Polyphen annotations
- A **Sample Sheet** can now be created which allows easy association between file names and the case and sample submitter_id
- Updated Advanced Search page to include options to **Add All Files to Cart**, **Download Manifest**, and **View X Cases in Exploration**
- Provide clear message rather than blank screen if survival plots cannot be calculated for particular cohort comparison
- Display sample_type on associated entities section on file page
- Allows for special characters in case, gene, and mutation set upload (-, :, >, .)

Bugs Fixed Since Last Release

- Fixed error when trying to download large number of files from the Legacy Archive cart
- Fixed number of annotations displayed in Legacy Archive for particular entities
- Replaced missing bars to indicate proportion of applicable files and cases on project entity page in Cases and File Counts by Data Category table
- Fixed project page display when projects are selected that contain no mutation data in the facet panel
- Fixed error where exporting case sets as TSV included fewer cases than the total
- Fixed error in exploration section when adding custom facets. Previously selecting ‘Only show fields with values’ did not result in the expected behavior
- Fixed error where number of associated entities for a file was showing an incorrect number

Known Issues and Workarounds

- Sample sheet will download with a file name including the date duplicated (e.g. gdc_sample_sheet_YYYY-MM-DD_HH-MM.tsv.YYYY-MM-DD_HH-MM.tsv)
- Custom facet filters
 - Definitions are missing from the property list when adding custom facet file or case filters

- Visualizations
 - Data Portal graphs cannot be exported as PNG images in Internet Explorer. Graphs can be exported in PNG or SVG format from Chrome or Firefox browsers . Internet Explorer does not display chart legend and title when re-opening previously downloaded SVG files, the recommendation is to open downloaded SVG files with another program.
 - In the protein viewer there may be overlapping mutations. In this case mousing over a point will just show a single mutation and the other mutations at this location will not be apparent.
- Entity page
 - On the mutation entity page, in the Consequences Table, the “Coding DNA Change” column is not populated for rows that do not correspond to the canonical mutation.
- Repository and Cart
 - The annotation count in File table of Repository and Cart does not link to the Annotations page anymore. The user can navigate to the annotations through the annotation count in Repository - Case table.
- Legacy Archive
 - Downloading a token in the GDC Legacy Archive does not refresh it. If a user downloads a token in the GDC Data Portal and then attempts to download a token in the GDC Legacy Archive, an old token may be provided. Reloading the Legacy Archive view will allow the user to download the updated token.
 - Exporting the Cart table in JSON will export the GDC Archive file table instead of exporting the files in the Cart only.
- Web Browsers
 - Browsers limit the number of concurrent downloads, it is generally recommended to add files to the cart and download large number of files through the GDC Data Transfer Tool, more details can be found on [GDC Website](#).
 - The GDC Portals are not compatible with Internet Explorer running in compatibility mode. Workaround is to disable compatibility mode.

Release details are maintained in the GDC Data Portal Change Log.

Release 1.10.0

- **GDC Product:** GDC Data Portal
- **Release Date:** November 16, 2017

New Features and Changes

- Support for uploading Case and Mutation sets in Exploration page
- Support for saving, editing, removing Case, Gene and Mutation sets in the Exploration page
- Added a Managed Sets menu where the user can see their saved sets
- Added an Analysis menu with two analyses: Set Operation and Cohort Comparison
- Added a User Profile page that shows all the projects and permissions assigned to the user: available in the username dropdown after the user logs in

Bugs Fixed Since Last Release

- Project page
 - On the project page, the Summary Case Count link should open the case tab on the Repository page - instead it opens the file page

Known Issues and Workarounds

- Custom facet filters
 - Definitions are missing from the property list when adding custom facet file or case filters
 - Selecting ‘Only show fields with values’ will show some fields without values in the Repository section. This works correctly under the Exploration section.
- Visualizations
 - Data Portal graphs cannot be exported as PNG images in Internet Explorer. Graphs can be exported in PNG or SVG format from Chrome or Firefox browsers. Internet Explorer does not display chart legend and title when re-opening previously downloaded SVG files, the recommendation is to open downloaded SVG files with another program.
 - In the protein viewer there may be overlapping mutations. In this case mousing over a point will just show a single mutation and the other mutations at this location will not be apparent.
- Entity page
 - On the mutation entity page, in the Consequences Table, the “Coding DNA Change” column is not populated for rows that do not correspond to the canonical mutation.
- Repository and Cart
 - The annotation count in File table of Repository and Cart does not link to the Annotations page anymore. The user can navigate to the annotations through the annotation count in Repository - Case table.
- Legacy Archive
 - Downloading a token in the GDC Legacy Archive does not refresh it. If a user downloads a token in the GDC Data Portal and then attempts to download a token in the GDC Legacy Archive, an old token may be provided. Reloading the Legacy Archive view will allow the user to download the updated token.
 - Exporting the Cart table in JSON will export the GDC Archive file table instead of exporting the files in the Cart only.
- Web Browsers
 - Browsers limit the number of concurrent downloads, it is generally recommended to add files to the cart and download large number of files through the GDC Data Transfer Tool, more details can be found on [GDC Website](#).
 - The GDC Portals are not compatible with Internet Explorer running in compatibility mode. Workaround is to disable compatibility mode.

Release details are maintained in the GDC Data Portal Change Log.

Release 1.9.0

- **GDC Product:** GDC Data Portal
- **Release Date:** October 24, 2017

New Features and Changes

- Support for projects with multiple primary sites per project
- Support for slides that are linked to **sample** rather than **portion**

Bugs Fixed Since Last Release

None

Known Issues and Workarounds

- Visualizations
 - Data Portal graphs cannot be exported as PNG images in Internet Explorer. Graphs can be exported in PNG or SVG format from Chrome or Firefox browsers. Internet Explorer does not display chart legend and title when re-opening previously downloaded SVG files, the recommendation is to open downloaded SVG files with another program.
 - In the protein viewer there may be overlapping mutations. In this case mousing over a point will just show a single mutation and the other mutations at this location will not be apparent.
- Project page
 - On the project page, the Summary Case Count link should open the case tab on the Repository page - instead it opens the file page
- Entity page
 - On the mutation entity page, in the Consequences Table, the “Coding DNA Change” column is not populated for rows that do not correspond to the canonical mutation.
- Repository and Cart
 - The annotation count in File table of Repository and Cart does not link to the Annotations page anymore. The user can navigate to the annotations through the annotation count in Repository - Case table.
- Legacy Archive
 - Downloading a token in the GDC Legacy Archive does not refresh it. If a user downloads a token in the GDC Data Portal and then attempts to download a token in the GDC Legacy Archive, an old token may be provided. Reloading the Legacy Archive view will allow the user to download the updated token.
 - Exporting the Cart table in JSON will export the GDC Archive file table instead of exporting the files in the Cart only.
- Web Browsers
 - Browsers limit the number of concurrent downloads, it is generally recommended to add files to the cart and download large number of files through the GDC Data Transfer Tool, more details can be found on [GDC Website](#).
 - The GDC Portals are not compatible with Internet Explorer running in compatibility mode. Workaround is to disable compatibility mode.

Release details are maintained in the GDC Data Portal Change Log.

Release 1.8.0

- **GDC Product:** GDC Data Portal
- **Release Date:** August 22, 2017

New Features and Changes

Major features/changes:

- A feature that links the exploration and repository pages was added. For example:
 - In the exploration page, cases with a specific mutation could be selected. This set could then be linked to the repository page to download the data files associated with these cases.
 - In the repository menu, the user can select cases associated with specific files. The set could then be linked to exploration page to view the variants associated with this set of cases.
- Users can now upload a custom gene list to the exploration page and leverage the GDC search and visualization features for cases and variants associated with the gene set.
- Filters added for the gene entity page. For example:

- Clicking on a mutated gene from the project page will display mutations associated with the gene that are present in this project (filtered protein viewer, etc.).
- Clicking on a mutated gene from the exploration page will display the mutations associated with the gene filtered by additional search criteria, such as “primary site is Kidney and mutation impact is high”.
- UUIDs are now hidden from tables and charts to simplify readability. The UUIDs can still be exported and viewed in the tables using the “arrange columns” feature. In the mutation table, UUIDs are automatically exported.
- Mutation entity page - one consequence per transcript is shown (10 rows by default) in the consequence table. The user should display all rows before exporting the table.

Bugs Fixed Since Last Release

- Exploration
 - Combining “Variant Caller” mutation filter with a case filter will display incorrect counts in the mutation facet. The number of mutations in the resulting mutation table is correct.
 - Mutation table: it is difficult to click on the denominator in “#Affected Cases in Cohort” column displayed to the left side of the bar. The user should click at a specific position at the top of the number to be able to go to the corresponding link.

Known Issues and Workarounds

- Visualizations
 - Data Portal graphs cannot be exported as PNG images in Internet Explorer. Graphs can be exported in PNG or SVG format from Chrome or Firefox browsers . Internet Explorer does not display chart legend and title when re-opening previously downloaded SVG files, the recommendation is to open downloaded SVG files with another program.
 - In the protein viewer there may be overlapping mutations. In this case mousing over a point will just show a single mutation and the other mutations at this location will not be apparent.
- Project page
 - On the project page, the Summary Case Count link should open the case tab on the Repository page - instead it opens the file page
- Entity page
 - On the mutation entity page, in the Consequences Table, the “Coding DNA Change” column is not populated for rows that do not correspond to the canonical mutation.
- Repository and Cart
 - The annotation count in File table of Repository and Cart does not link to the Annotations page anymore. The user can navigate to the annotations through the annotation count in Repository - Case table.
- Legacy Archive
 - Downloading a token in the GDC Legacy Archive does not refresh it. If a user downloads a token in the GDC Data Portal and then attempts to download a token in the GDC Legacy Archive, an old token may be provided. Reloading the Legacy Archive view will allow the user to download the updated token.
 - Exporting the Cart table in JSON will export the GDC Archive file table instead of exporting the files in the Cart only.
- Web Browsers
 - Browsers limit the number of concurrent downloads, it is generally recommended to add files to the cart and download large number of files through the GDC Data Transfer Tool, more details can be found on [GDC Website](#).
 - The GDC Portals are not compatible with Internet Explorer running in compatibility mode. Workaround is to disable compatibility mode.

Release details are maintained in the GDC Data Portal Change Log.

Release 1.6.0

- **GDC Product:** GDC Data Portal
- **Release Date:** June 29, 2017

New Features and Changes

There was a major new release of the GDC Data Portal focused on Data Analysis, Visualization, and Exploration (DAVE). Some important new features include the following:

- New visual for the Homepage: a human body provides the number of Cases per Primary Site with a link to an advanced Cancer Projects search
- The Projects menu provides the Top 20 Cancer Genes across the GDC Projects and the Case Distribution per Project
- A new menu “Exploration” is an advanced Cancer Projects search which provides the ability to apply Case, Gene, and Mutation filters to look for:
 - List of Cases with the largest number of Somatic Mutations
 - The most frequently mutated Genes
 - The most frequent Variants
 - Oncogrid view of mutation frequency
- Visualizations are provided across the Project, Case, Gene and Mutation entity pages:
 - List of most frequently mutated genes and most frequent variants
 - Survival plots for patients with or without specific variants
 - Survival plots for patients with or without variants in specific genes
 - Lollipop plots of mutation frequency across protein domains
- Links to external databases (COSMIC, dbSNP, Uniprot, Ensembl, OMIM, HGNC)
- Quick Search for Gene and Mutation entity pages
- The ability to export the current view of a table in TSV
- Retired GDC cBioPortal

For detailed updates please review the [Data Portal User Guide](#).

Bugs Fixed Since Last Release

- BAM Slicing dialog box does not disappear automatically upon executing the BAM slicing function. The box can be closed manually.
- Very long URLs will produce a 400 error. Users may encounter this after clicking on “source files” on a file page where the target file is derived from hundreds of other files such as for MAF files.
- If bam slicing produces an error pop-up message it will be obscured behind the original dialog box.
 - Internet Explorer users are not able to use the “Only show fields with no values” when adding custom facets
 - Exporting large tables in the Data Portal may produce a 500 error. Filtering this list to include fewer cases or files should eliminate the error

Known Issues and Workarounds

- New Visualizations
 - Cannot export Data Portal graphs in PNG in Internet Explorer. Graphs can be exported to PNG or SVG from Chrome or Firefox browsers. Internet would not display chart legend and title when re-opening previously downloaded SVG files, recommendation is to open downloaded SVG files with another software.
 - In the protein viewer there may be overlapping mutations. In this case mousing over a point will just show a single mutation and the other mutations at this location will not be apparent.

- Exploration
 - Combining “Variant Caller” mutation filter with a case filter will display wrong counts in the mutation facet. The number of mutations in the result mutation table is correct.
 - Mutation table: it is difficult to click on the denominator in “#Affected Cases in Cohort” column displayed to the left side of the bar. The user should click at a specific position at the top of the number to be able to go to the corresponding link.
- Entity page
 - On the mutation entity page, in the Consequences Table, the “Coding DNA Change” column is not populated for rows that do not correspond to the canonical mutation.
- Repository and Cart
 - The annotation count in File table of Repository and Cart does not link to the Annotations page anymore. The user can navigate to the annotations through the annotation count in Repository - Case table.
- Legacy Archive
 - Downloading a token in the GDC Legacy Archive does not refresh it. If a user downloads a token in the GDC Data Portal and then attempts to download a token in the GDC Legacy Archive, an old token may be provided. Reloading the Legacy Archive view will allow the user to download the updated token.
 - Exporting the Cart table in JSON will export the GDC Archive file table instead of exporting the files in the Cart only.
- Web Browsers
 - Browsers limit the number of concurrent downloads, it is generally recommended to add files to the cart and download large number of files through the GDC Data Transfer Tool, more details can be found on [GDC Website](#).
 - The GDC Portals are not compatible with Internet Explorer running in compatibility mode. Workaround is to disable compatibility mode.

Release details are maintained in the GDC Data Portal Change Log.

Release 1.5.2

- **GDC Product:** GDC Data Portal
- **Release Date:** May 9, 2017

New Features and Changes

- Removed link to Data Download Statistics Report
- Updated version numbers of API, GDC Data Portal, and Data Release

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- General
 - Exporting large tables in the Data Portal may produce a 500 error. Filtering this list to include fewer cases or files should eliminate the error
 - After successful authentication, the authentication popup does not close for Internet Explorer users running in “Compatibility View”. Workaround is to uncheck “Display Intranet sites in Compatibility View” in Internet Explorer options. Alternatively, refreshing the portal will correctly display authentication status.

- BAM Slicing dialog box does not disappear automatically upon executing the BAM slicing function. The box can be closed manually.
- Due to preceding issue, If bam slicing produces an error pop-up message it will be obscured behind the original dialog box.
- Very long URLs will produce a 400 error. Users may encounter this after clicking on “source files” on a file page where the target file is derived from hundreds of other files such as for MAF files. To produce a list of source files an API call can be used with the search parameter “fields=analysis.input_files.file_name”.
 - * Downloading a token in the GDC Legacy Archive does not refresh it. If a user downloads a token in the GDC Data Portal and then attempts to download a token in the GDC Legacy Archive, an old token may be provided. Reloading the Legacy Archive view will allow the user to download the updated token.

Example

1 https://api.gdc.cancer.gov/files/455e26f7-03f2-46f7-9e7a-9c51ac322461?pretty=true&fields=analysis.input_files.file_name

- Cart
 - Counts displayed in the top right of the screen, next to the Cart icon, may become inconsistent if files are removed from the server.
- Web Browsers
 - Browsers limit the number of concurrent downloads, it is generally recommended to add files to the cart and download large number of files through the GDC Data Transfer Tool, more details can be found on [GDC Website](#).
 - Internet Explorer users are not able to use the “Only show fields with no values” when adding custom facets
 - The GDC Portals are not compatible with Internet Explorer running in compatibility mode. Workaround is to disable compatibility mode.

Release details are maintained in the GDC Data Portal Change Log.

Release 1.4.1

- **GDC Product:** GDC Data Portal
- **Release Date:** October 31, 2016

New Features and Changes

- Added a search feature to help users select values of interest in certain facets that have many values.
- Added support for annotation ID queries in quick search.
- Added a warning when a value greater than 90 is entered in the “Age at Diagnosis” facet.
- Added Sample Type column to file entity page.
- Authentication tokens are refreshed every time they are downloaded from the GDC Data Portal.
- Buttons are inactive when an action is in progress.
- Improved navigation features in the overview chart on portal homepage.
- Removed State/Status from File and Case entity pages
- Removed the “My Projects” feature.
- Removed “Created” and “Updated” dates from clinical and biospecimen entities.

Bugs Fixed Since Last Release

- Advanced search did not accept negative values for integer fields.
- Moving from facet search to advanced search resulted in an incorrect advanced search query.
- Some facets were cut off in Internet Explorer and Firefox.

Known Issues and Workarounds

- General
 - Exporting large tables in the Data Portal may produce a 500 error. Filtering this list to include fewer cases or files should eliminate the error
 - After successful authentication, the authentication popup does not close for Internet Explorer users running in “Compatibility View”. Workaround is to uncheck “Display Intranet sites in Compatibility View” in Internet Explorer options. Alternatively, refreshing the portal will correctly display authentication status.
 - BAM Slicing dialog box does not disappear automatically upon executing the BAM slicing function. The box can be closed manually.
 - Due to preceding issue, If bam slicing produces an error pop-up message it will be obscured behind the original dialog box.
 - Very long URLs will produce a 400 error. Users may encounter this after clicking on “source files” on a file page where the target file is derived from hundreds of other files such as for MAF files. To produce a list of source files an API call can be used with the search parameter “fields=analysis.input_files.file_name”.
 - * Downloading a token in the GDC Legacy Archive does not refresh it. If a user downloads a token in the GDC Data Portal and then attempts to download a token in the GDC Legacy Archive, an old token may be provided. Reloading the Legacy Archive view will allow the user to download the updated token.

Example

```
1 https://api.gdc.cancer.gov/files/455e26f7-03f2-46f7-9e7a-9c51ac322461?pretty=true&fields=analysis.input_files.f
```

- Cart
 - Counts displayed in the top right of the screen, next to the Cart icon, may become inconsistent if files are removed from the server.
- Web Browsers
 - Browsers limit the number of concurrent downloads, it is generally recommended to add files to the cart and download large number of files through the GDC Data Transfer Tool, more details can be found on [GDC Website](#).
 - Internet Explorer users are not able to use the “Only show fields with no values” when adding custom facets
 - The GDC Portals are not compatible with Internet Explorer running in compatibility mode. Workaround is to disable compatibility mode.

Release details are maintained in the GDC Data Portal Change Log.

Release 1.3.0

- **GDC Product:** GDC Data Portal
- **Release Date:** September 7, 2016

New Features and Changes

- A new “Metadata” button on the cart page to download merged clinical, biospecimen, and file metadata in a single consolidated JSON file. **May require clearing browser cache**
- Added a banner on the Data Portal to help users find data
- Added support for “Enter” key on login button
- On the Data page, the browser will remember which facet tab was selected when hitting the “Back” button
- In file entity page, if there is a link to one single file, redirect to this file’s entity page instead of a list page.

Bugs Fixed Since Last Release

- Adding a mix of open and controlled files to the cart from any Case entity pages was creating authorization issues
- Opening multiple browser tabs and adding files in those browser tabs was not refreshing the cart in other tabs.
- When user logs in from the advanced search page, the login popup does not automatically close
- When removing a file from the cart and clicking undo, GDC loses track of permission status of the user towards this file and will ask for the user to log-in again.
- Download File Metadata button produces incomplete JSON output omitting such fields as file_name and submitter_id. The current workaround includes using the API to return file metadata.
- Annotations notes do not wrap to the next line at the beginning or the end of a word, some words might be split in two lines
- Sorting annotations by Case UUID causes error

Known Issues and Workarounds

- General
 - When no filters are engaged in the Legacy Archive or Data Portal, clicking the Download Manifest button may produce a 500 error and the message “We are currently experiencing issues. Please try again later.”. To avoid this error the user can first filter by files or cases to reduce the number files added to the manifest.
 - After successful authentication, the authentication popup does not close for Internet Explorer users running in “Compatibility View”. Workaround is to uncheck “Display Intranet sites in Compatibility View” in Internet Explorer options. Alternatively, refreshing the portal will correctly display authentication status.
 - BAM Slicing dialog box does not disappear automatically upon executing the BAM slicing function. The box can be closed manually.
 - Due to preceding issue, If bam slicing produces an error pop-up message it will be obscured behind the original dialog box.
 - Very long URLs will produce a 400 error. Users may encounter this after clicking on “source files” on a file page where the target file is derived from hundreds of other files such as for MAF files. To produce a list of source files an API call can be used with the search parameter “fields=analysis.input_files.file_name”.
 - On the Legacy Archive, searches for “Case Submitter ID Prefix” containing special characters are not displayed correctly above the result list. The result list is correct, however.

Example

```
1 https://api.gdc.cancer.gov/files/455e26f7-03f2-46f7-9e7a-9c51ac322461?pretty=true&fields=analysis.input_files.f
```

- Cart
 - Counts displayed in the top right of the screen, next to the Cart icon, may become inconsistent if files are removed from the server.
- Web Browsers
 - Browsers limit the number of concurrent downloads, it is generally recommended to add files to the cart and download large number of files through the GDC Data Transfer Tool, more details can be found on [GDC Website](#).
 - Internet Explorer users are not able to use the “Only show fields with no values” when adding custom facets
 - The GDC Portals are not compatible with Internet Explorer running in compatibility mode. Workaround is to disable compatibility mode.

Release details are maintained in the GDC Data Portal Change Log.

Release 1.2.0

- **GDC Product:** GDC Data Portal
- **Release Date:** August 9th, 2016

New Features and Changes

- Added a retry (1x) mechanism for API calls
- Added support for ID fields in custom facets
- Added Case Submitter ID to the Annotation entity page
- Added a link to Biospeciment in the Case entity page

Bugs Fixed Since Last Release

- General.
 - Not possible to use the browser’s back button after hitting a 404 page
 - 404 page missing from Legacy Archive Portal
 - Table widget icon and export JSON icon should be different
 - Download SRA XML files from the legacy archive portal might not be possible in some context
- Data and facets
 - Default values for age at diagnosis is showing 0 to 89 instead of 0 to 90
 - Biospecimen search in the case entity page does not highlight (but does bold and filter) results in yellow when title case is not followed
 - Table sorting icon does not include numbers
 - ‘_’ symbol is missing on empty fields (blank instead), additional missing fields identified since last release. ### Known Issues and Workarounds
- General
 - When no filters are engaged in the Legacy Archive or Data Portal, clicking the Download Manifest button may produce a 500 error and the message “We are currently experiencing issues. Please try again later.”. To avoid this error the user can first filter by files or cases to reduce the number files added to the manifest.
 - After successful authentication, the authentication popup does not close for Internet Explorer users running in “Compatibility View”. This only impact users at the NIH. Workaround is to uncheck “Display Intranet sites in Compatibility View” in Internet Explorer options. Alternatively, refreshing the portal will correctly display authentication status.
 - When user login from the advanced search page, the login popup does not automatically close
- Cart
 - When removing a file from the cart and clicking undo, GDC loses track of permission status of the user towards this file and will ask for the user to log-in again.
 - Counts displayed in the top right of the screen, next to the Cart icon, might get inconsistent if files are removed from the server.
 - Download File Metadata button produces incomplete JSON output omitting such fields as file_name and submitter_id. The current workaround includes using the API to return file metadata.
- Annotations
 - Annotations notes do not wrap to the next line at the beginning or the end of a word, some words might be split in two lines
 - Sorting annotations by Case UUID causes error
- Web Browsers
 - Browsers limit the number of concurrent downloads, it is generally recommended to add files to the cart and download large number of files through the GDC Data Transfer Tool, more details can be found on [GDC Website](#).
 - Internet Explorer users are not able to use the “Only show fields with no values” when adding custom facets
 - The GDC Portals are not compatible with Internet Explorer running in compatibility mode. Workaround is to disable compatibility mode

Release details are maintained in the GDC Data Portal Change Log.

Release 1.1.0

- **GDC Product:** GDC Data Portal
- **Release Date:** June 1st, 2016

New Features and Changes

- This is a bug-fixing release, no new features were added.

Bugs Fixed Since Last Release

- General
 - Fixed 508 compliance issues.
 - Disabled download manifest action on projects without files.
 - Updated the portal to indicate to the user that his session expired when he tries to download the authentication token.
 - Unselected “My project” filter after user logs-in.
 - Fixed missing padding when query includes “My Projects”.
 - Enforced “Add to cart” limitation to 10,000 files everywhere on the Data Portal.
- Tables
 - Improved usability of the “Sort” feature
 - Updated the “Add all files to cart” button to add all files corresponding to the current query (and not only displayed files).
 - Fixed an issue where Platform would show “0” when selected platform is “Affymetrix SNP 6.0”.
- Data
 - Corrected default values populated when adding a custom range facet.
 - Fixed an issue preventing the user to sort by File Submitter ID in data tables.
- File Entity Page
 - Improved “Associated Cases/Biospecimen” table for files associated to a lot of cases.
 - Fixed an error when performing BAM Slicing.

Known Issues and Workarounds

- General.
 - After successful authentication, the authentication popup does not close for Internet Explorer users running in “Compatibility View”. This only impact users at the NIH. Workaround is to uncheck “Display Intranet sites in Compatibility View” in Internet Explorer options. Alternatively, refreshing the portal will correctly display authentication status.
 - Download SRA XML files from the legacy archive portal might not be possible in some context
 - Not possible to use the browser’s back button after hitting a 404 page
 - 404 page missing from Legacy Archive Portal
 - Table widget icon and export JSON icon should be different
- Data and facets
 - Default values for age at diagnosis is showing 0 to 89 instead of 0 to 90
 - Biospecimen search in the case entity page does not highlight (but does bold and filter) results in yellow when title case is not followed
 - Table sorting icon does not include numbers
 - ‘-’ symbol is missing on empty fields (blank instead), additional missing fields identified since last release.
- Cart

- When removing a file from the cart and clicking undo, GDC loses track of permission status of the user towards this file and will ask for the user to log-in again.
- Counts displayed in the top right of the screen, next to the Cart icon, might get inconsistent if files are removed from the server.
- Annotations
 - Annotations notes do not wrap to the next line at the beginning or the end of a word, some words might be split in two lines
- Web Browsers
 - Browsers limit the number of concurrent downloads, it is generally recommended to add files to the cart and download large number of files through the GDC Data Transfer Tool, more details can be found on [GDC Website](#).
 - Internet Explorer users are not able to use the “Only show fields with no values” when adding custom facets
 - The GDC Portals are not compatible with Internet Explorer running in compatibility mode. Workaround is to disable compatibility mode

Release details are maintained in the GDC Data Portal Change Log.

Release 1.0.1

- **GDC Product:** GDC Data Portal
- **Release Date:** May 18, 2016

New Features and Changes

- This is a bug-fixing release, no new features were added.

Bugs Fixed Since Last Release

- Tables and Export
 - Restore default table column arrangement does not restore to the default but it restores to the previous state
- Cart and Download
 - Make the cart limit warning message more explanatory
 - In some situations, adding filtered files to the cart might fail
- Layout, Browser specific and Accessibility
 - When disabling CSS, footer elements are displayed out of order
 - If javascript is disabled html tags are displayed in the warning message
 - Layout issues when using the browser zoom in function on tables
 - Cart download spinner not showing at the proper place
 - Not all facets are expanded by default when loading the app

Known Issues and Workarounds

- General
 - If a user has previously logged into the Portal and left a session without logging out, if the user returns to the Portal after the user’s sessionID expires, it looks as if the user is still authenticated. The user cannot download the token and gets an error message that would not close. The user should clear the cache to properly log out.
 - ‘-’ symbol is missing on empty fields (blank instead)
 - Download manifest button is available for TARGET projects with 0 files, resulting in error if user click on button

- After successful authentication, the authentication popup does not close for Internet Explorer users running in “Compatibility View”. This only impact users at the NIH. Workaround is to uncheck “Display Intranet sites in Compatibility View” in Internet Explorer options. Alternatively, refreshing the portal will correctly display authentication status.
- Data
 - When adding a custom range facet, default values are incorrectly populated
 - The portal might return incorrect match between cases and files when using field `cases.samples.portions.created_datetime` (custom facet or advanced search). Note: this is not a UI issue.
 - Sorting File Submitter ID option on the file tab result in a Data Portal Error
- Tables and Export
 - Table sorting icon does not include numbers
- Browsers limit the number of concurrent downloads, it is generally recommended to add files to the cart and download large number of files through the GDC Data Transfer Tool, more details can be found on [GDC Website](#).

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