GDC Data Portal User's Guide

NCI Genomic Data Commons (GDC)

Contents

1 Getting Started **6**

Getting Started . . . 6
The GDC Data Portal: An Overview . . . 6
Accessing the GDC Data Portal . . . 6
Navigation . . . 7
Views . . . 7
Toolbar . . . 8
Tables . . . 9
Table Sort . . . 9
Table Arrangement . . . 10
Table Size . . . 10
Table Export . . . 10
Filtering and Searching . . . 10
Facet Filters . . . 10
Quick Search . . . 12
Advanced Search . . . 15

2 Projects **16**

Projects . . . 16
Summary . . . 16
Projects Page . . . 16
Visualizations . . . 18
Top Mutated Cancer Genes in Selected Projects . . . 18
Case Distribution per Project . . . 18
Projects Table . . . 18
Projects Graph . . . 19
Facets Panel . . . 20
Project Summary Page . . . 22
Most Frequently Mutated Genes . . . 22
Survival Analysis . . . 24
Most Frequent Mutations . . . 25
Most Affected Cases . . . 27

3 Exploration **28**

Exploration . . . 28
Filters / Facets . . . 28
Case Filters . . . 29
Gene Filters . . . 31
Upload Gene Set . . . 33
Mutation Filters . . . 33
Results . . . 35
Cases . . . 35
Genes . . . 37
Mutations . . . 38
OncoGrid . . . 40
OncoGrid Options . . . 42
File Navigation . . . 42

4 Repository **43**

Repository . . . 43
Summary . . . 43
Filters / Facets . . . 43
Facets Panel . . . 44
Adding Custom Facets . . . 46
Files List . . . 47
Cases List . . . 49
Navigation . . . 50
Case Summary Page . . . 51
Clinical and Biospecimen Information . . . 52
Biospecimen Search . . . 52
Most Frequent Somatic Mutations . . . 53
File Summary Page . . . 54
BAM Slicing . . . 55

5 Genes and Mutations **56**

Gene and Mutation Summary Pages . . . 56
Gene Summary Page . . . 56
Summary . . . 56
External References . . . 57
Cancer Distribution . . . 57
Protein Viewer . . . 58
Most Frequent Mutations . . . 58
Mutation Summary Page . . . 59

Summary . . . 59
External References . . . 60
Consequences . . . 60
Cancer Distribution . . . 60
Protein Viewer . . . 62

6 Annotations **63**
Annotations . . . 63
Annotations View . . . 63
Facets Panel . . . 64
Annotation Categories and Classification . . . 64
Annotation Detail Page . . . 64

7 Advanced Search **66**
Advanced Search . . . 66
Overview: GQL . . . 66
Switching between Advanced Search and Facet Filters . . . 67
Using the Advanced Search . . . 68
Auto-complete . . . 68
Field Auto-complete . . . 68
Value Auto-complete . . . 68
Setting Precedence of Operators . . . 69
Keywords . . . 69
AND Keyword . . . 69
OR Keyword . . . 70
Operators . . . 70
List of Operators and Query format . . . 70
“=” operator - EQUAL . . . 70
“!=” operator - NOT EQUAL . . . 70
“>” operator - GREATER THAN . . . 71
“>=” operator - GREATER THAN OR EQUALS . . . 71
“<” operator - LESS THAN . . . 71
“<=” operator - LESS THAN OR EQUALS . . . 71
“IN” Operator . . . 71
“EXCLUDE” Operator . . . 72
“IS MISSING” Operator . . . 72
“NOT MISSING” Operator . . . 72
Special Cases . . . 72
Date format . . . 72
Using Quotes . . . 73

Age at Diagnosis - Unit in Days . . . 73
Fields Reference . . . 73
Files . . . 73
Cases . . . 74

8 Authentication **76**
Authentication . . . 76
Overview . . . 76
Logging into the GDC . . . 76
GDC Authentication Tokens . . . 78
Logging Out . . . 78

9 File Cart **79**
Cart and File Download . . . 79
Overview . . . 79
GDC Cart . . . 79
Cart Summary . . . 79
Cart Items . . . 80
Download Options . . . 80
GDC Data Transfer Tool . . . 81
Individual Files Download . . . 81
Controlled Files . . . 81

10 Legacy Archive **83**
Legacy Archive . . . 83
Overview . . . 83
File Page . . . 84
Archive . . . 85
Metadata files . . . 85
File Cart . . . 85

11 Release Notes **86**
Data Portal Release Notes . . . 86
Release 1.9.0 . . . 86
New Features and Changes . . . 86
Bugs Fixed Since Last Release . . . 86
Known Issues and Workarounds . . . 86
Release 1.8.0 . . . 87
New Features and Changes . . . 87
Bugs Fixed Since Last Release . . . 87
Known Issues and Workarounds . . . 88

Release 1.6.0 . . . 88
New Features and Changes . . . 88
Bugs Fixed Since Last Release . . . 89
Known Issues and Workarounds . . . 89
Release 1.5.2 . . . 90
New Features and Changes . . . 90
Bugs Fixed Since Last Release . . . 90
Known Issues and Workarounds . . . 90
Release 1.4.1 . . . 91
New Features and Changes . . . 91
Bugs Fixed Since Last Release . . . 91
Known Issues and Workarounds . . . 91
Release 1.3.0 . . . 92
New Features and Changes . . . 92
Bugs Fixed Since Last Release . . . 92
Known Issues and Workarounds . . . 92
Release 1.2.0 . . . 93
New Features and Changes . . . 93
Bugs Fixed Since Last Release . . . 93
Release 1.1.0 . . . 94
New Features and Changes . . . 94
Bugs Fixed Since Last Release . . . 94
Known Issues and Workarounds . . . 95
Release 1.0.1 . . . 95
New Features and Changes . . . 96
Bugs Fixed Since Last Release . . . 96
Known Issues and Workarounds . . . 96

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

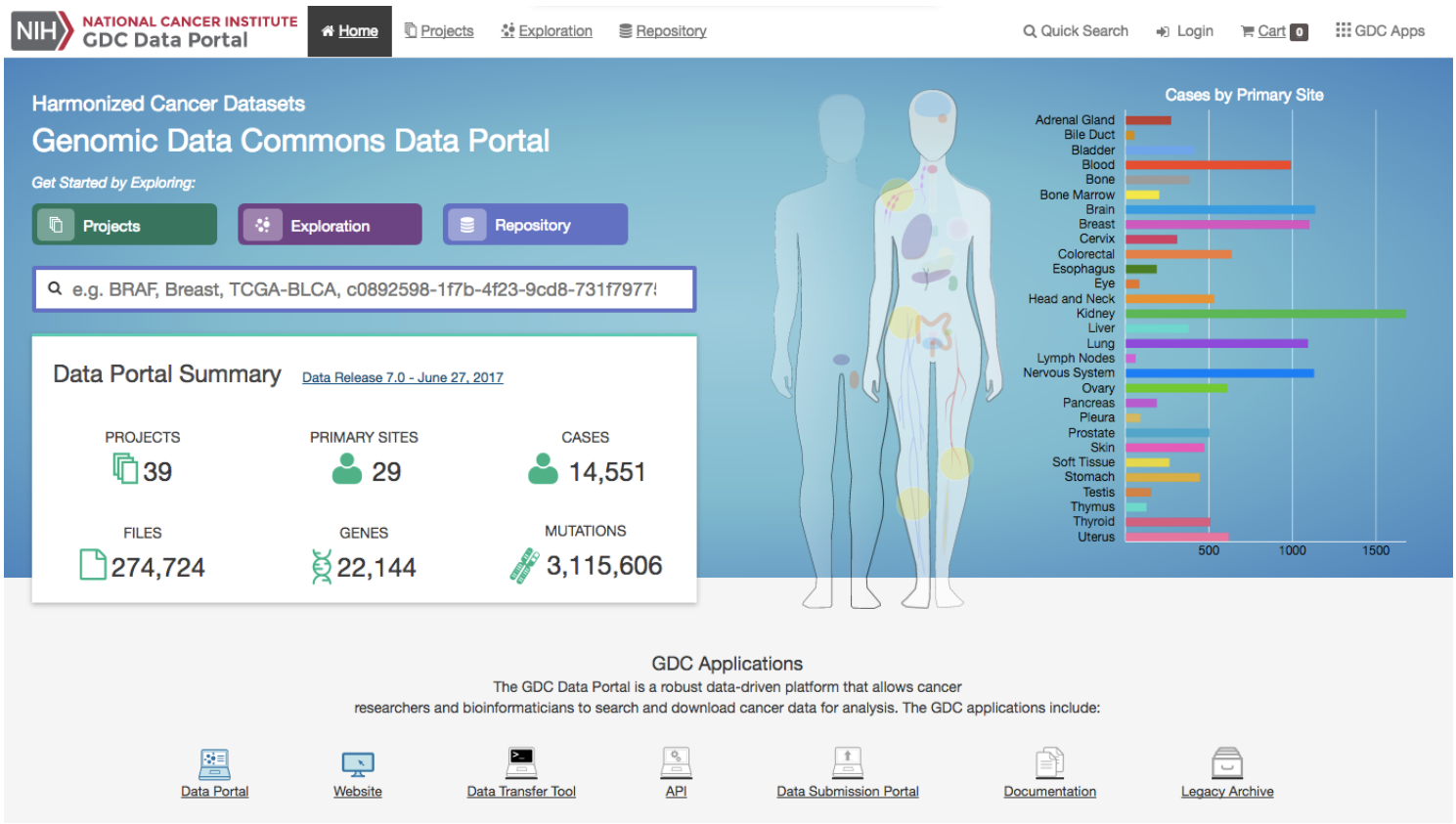
For more information about available datasets, see the GDC Website.

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

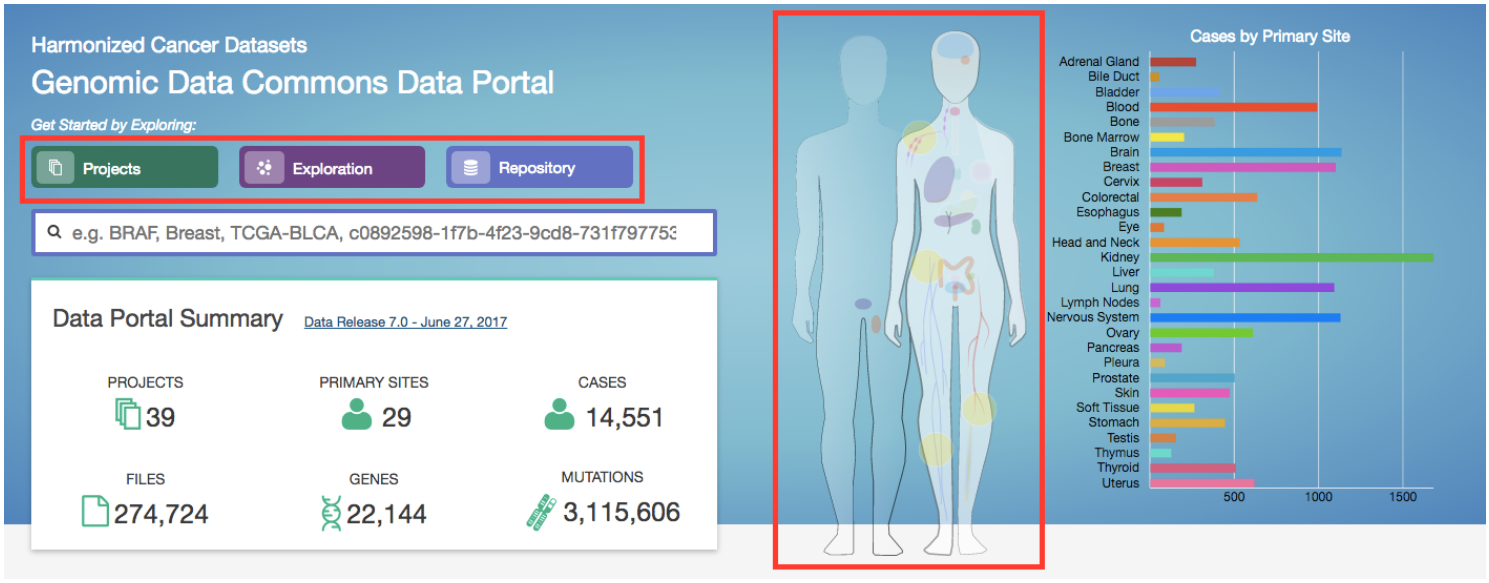
The front page displays an overview of all available datasets:



Navigation

Views

The GDC Data Portal provides four interfaces (*Views*) for browsing available harmonized datasets:



- **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.

- **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.

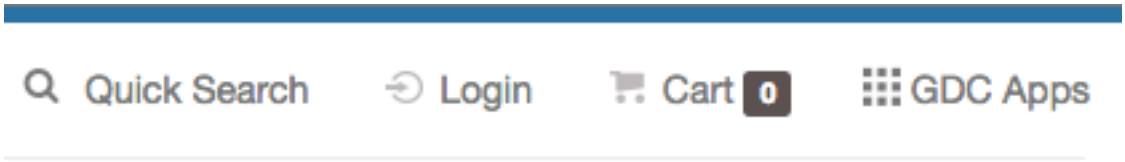
The Projects, Exploration, and Repository pages can be accessed from the GDC Data Portal front page and from the toolbar (see below). The annotations view is accessible from Repository view. A link to the GDC Legacy Archive is available on the GDC Data Portal front page and in the GDC Apps menu (see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

Toolbar

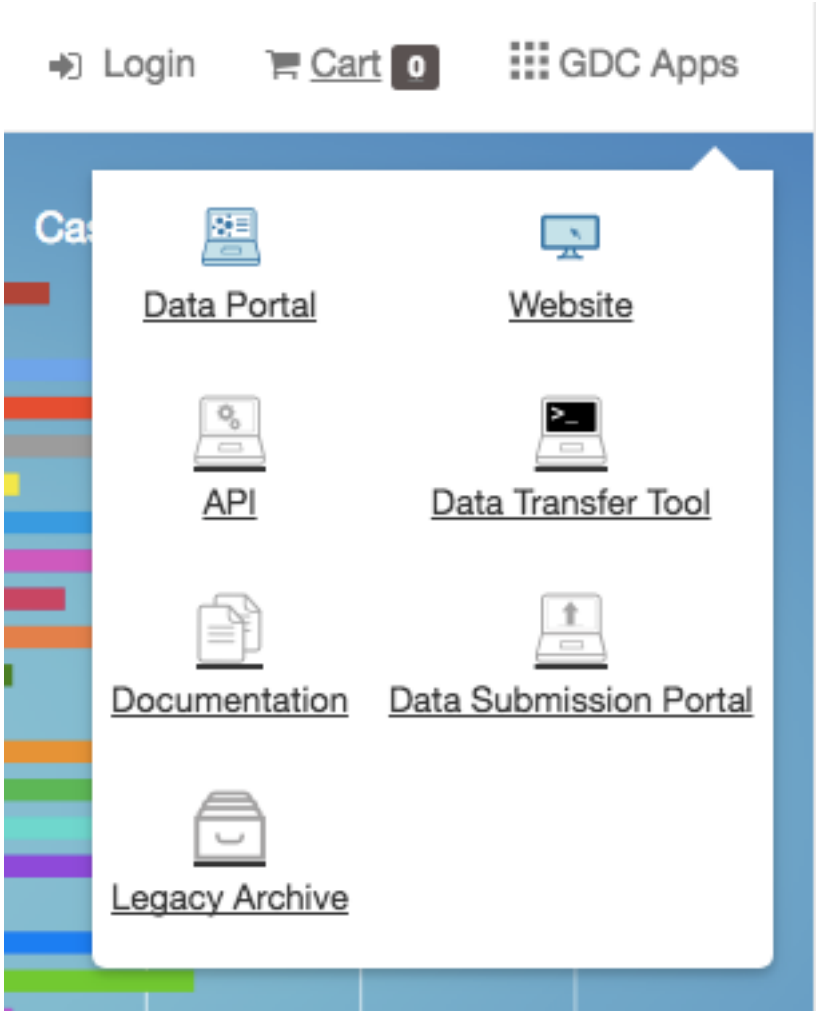
The toolbar available at the top of all pages in the GDC Data Portal provides convenient navigation links and access to authentication and quick search.

The left portion of this toolbar provides access to the Home Page, **Projects Page**, **Exploration Page**, 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 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.

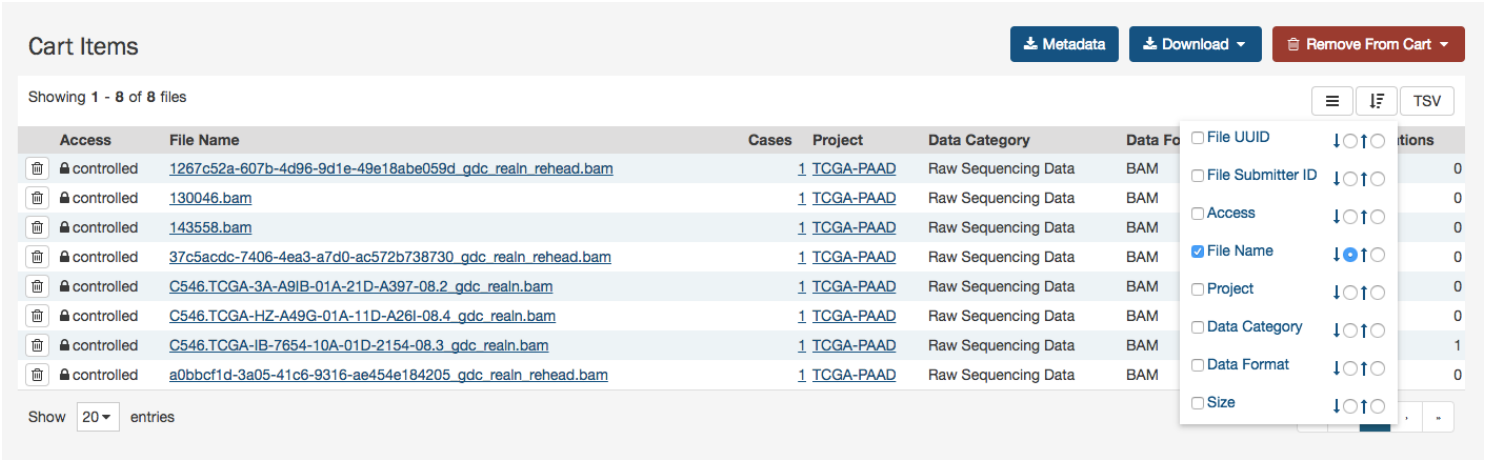


Table Arrangement

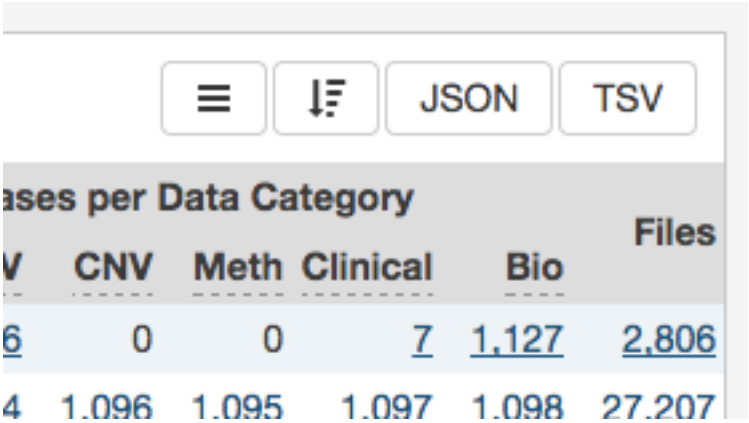
The *arrange columns* button allows users to adjust the order of columns in the table and select which columns are displayed.

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.



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

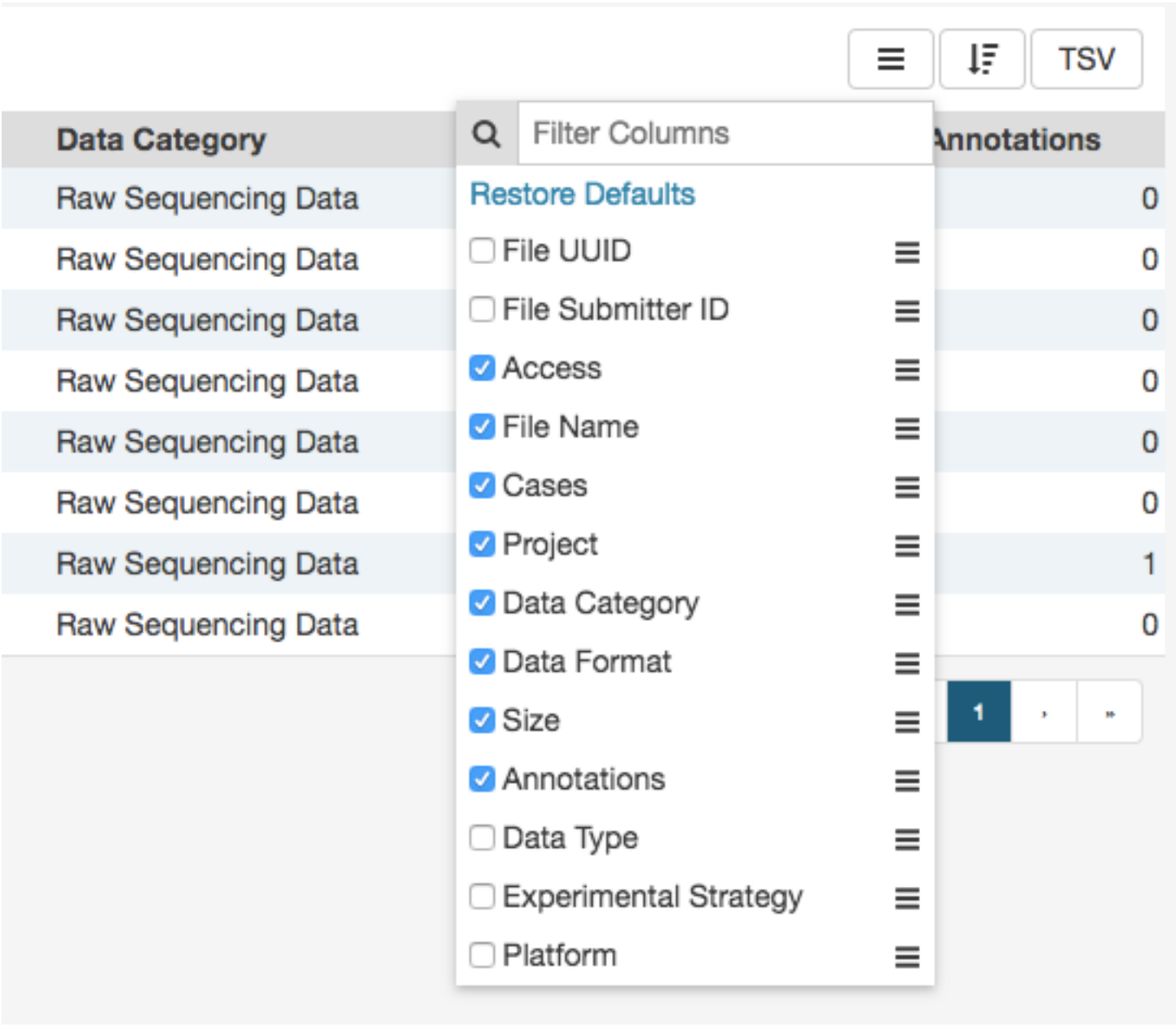


Figure 1.1: Selecting table columns

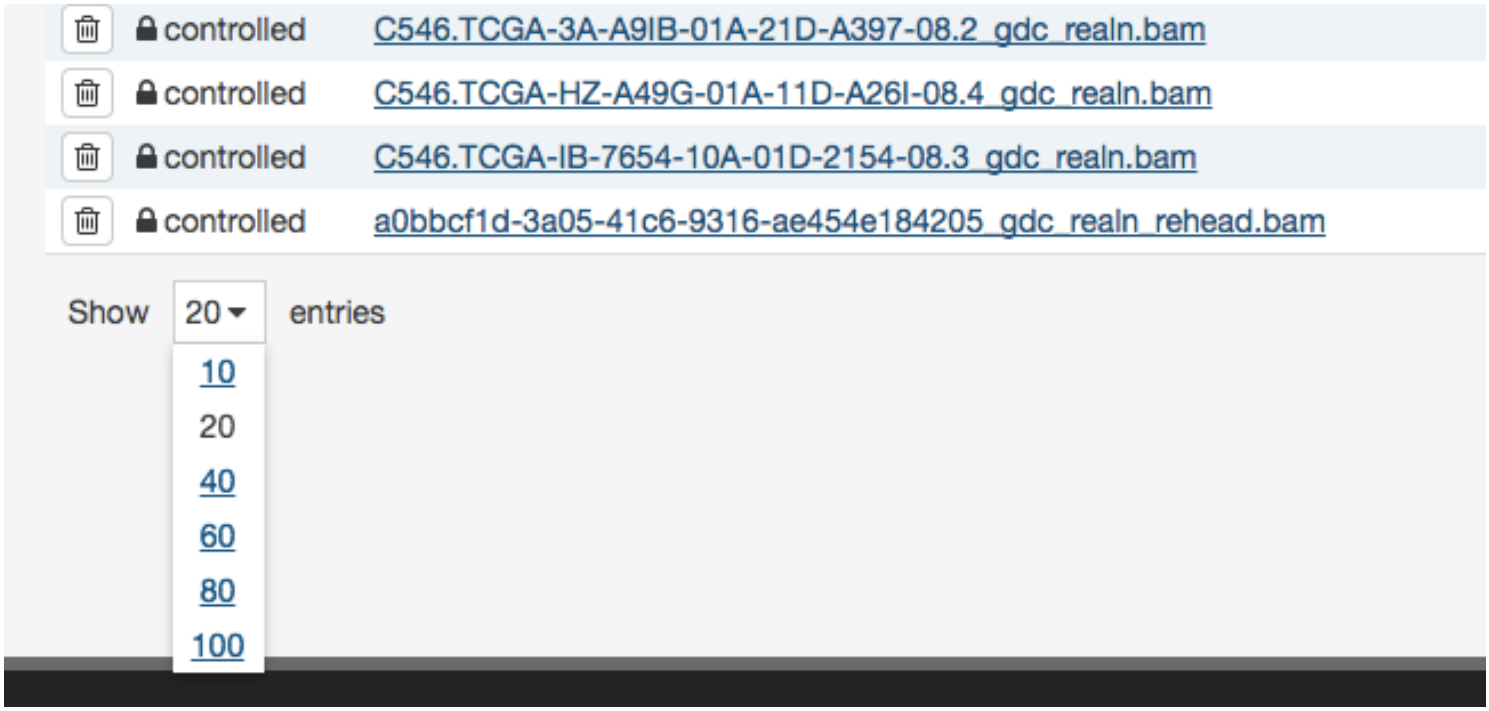


Figure 1.2: Specifying table size

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.

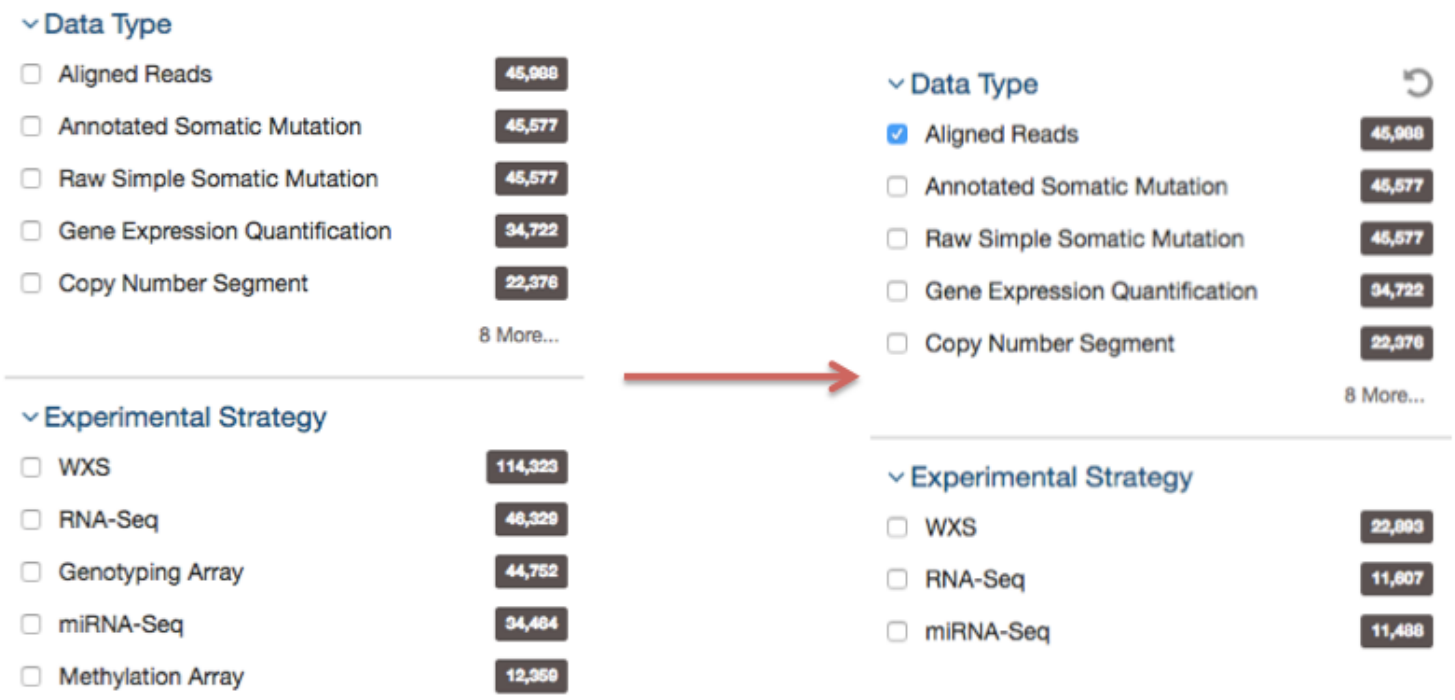
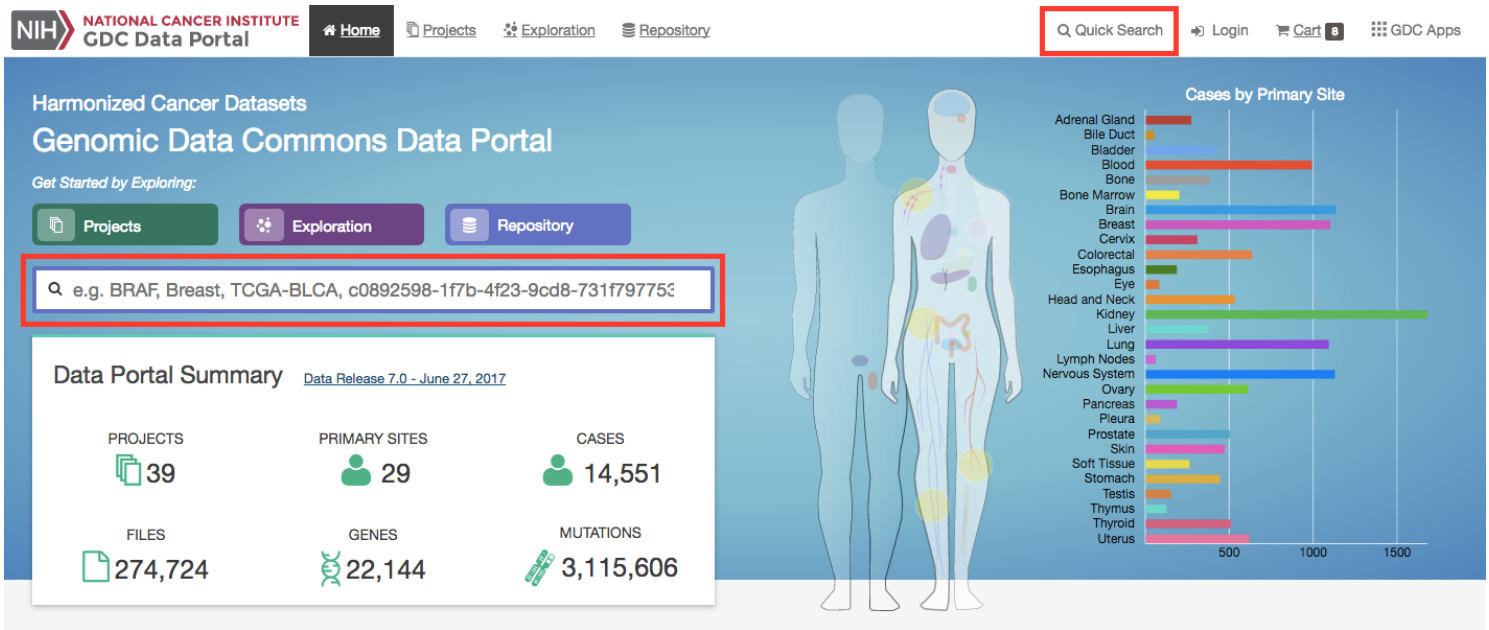
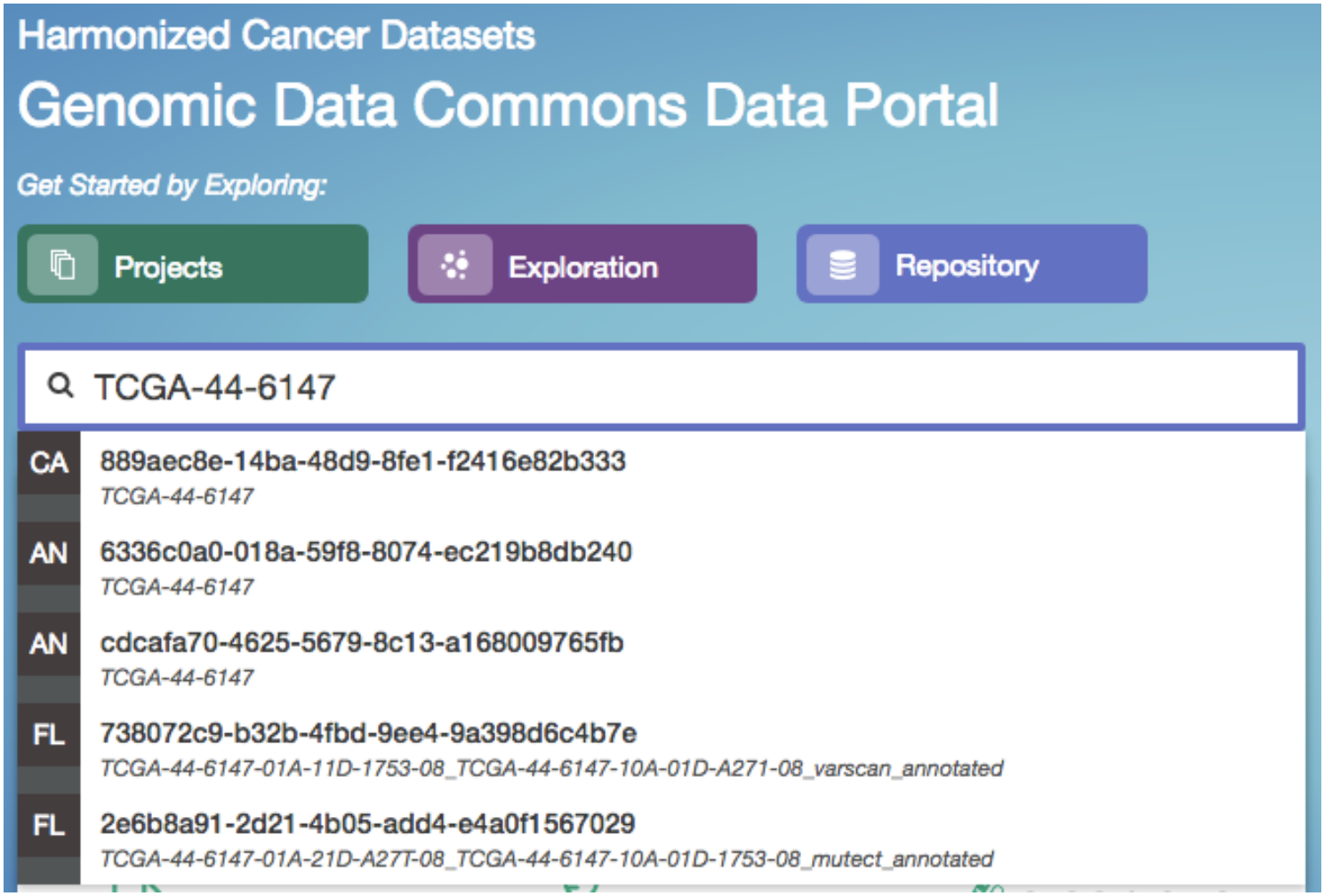


Figure 1.3: Facets (no filter applied)

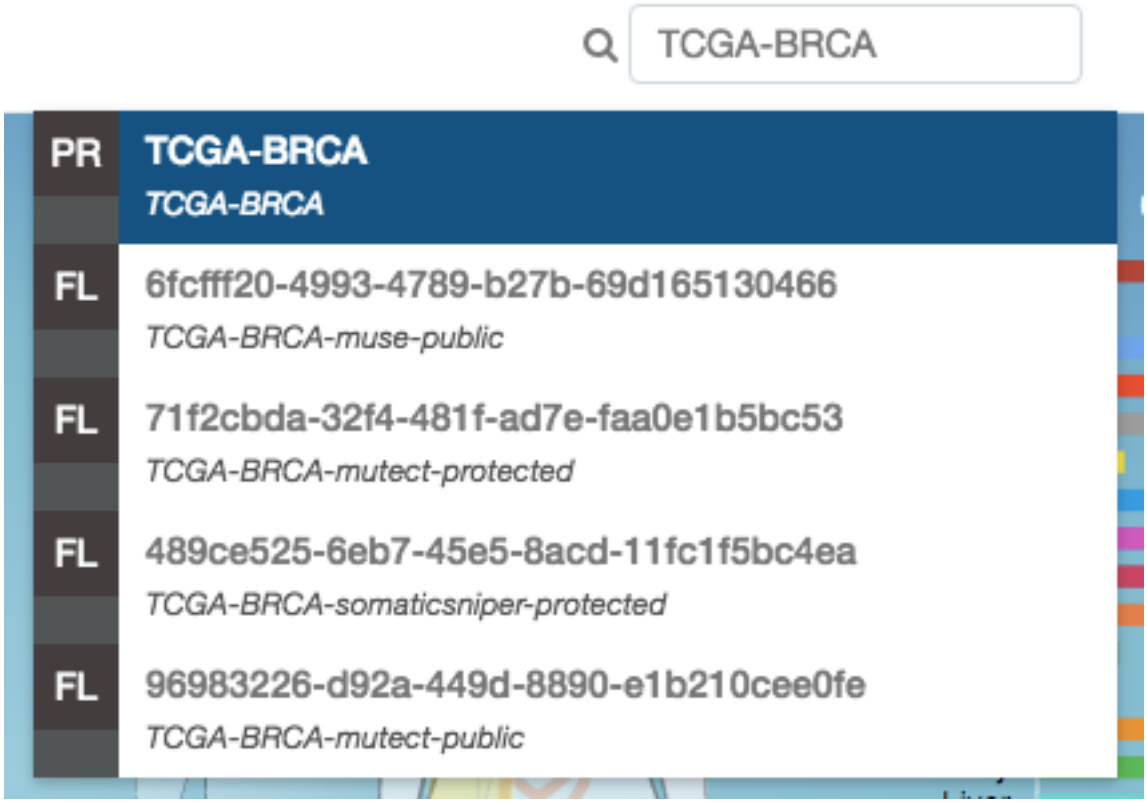


Search results are displayed as the user is typing, with labels indicating the type of each search result in the list (project, case, or file). Users users will see a brief description of the search results, which may include the UUID, submitter ID, or file name. Clicking on a selected result or pressing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

Home Page Quick Search:



Toolbar Quick Search:



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.

Chapter 2

Projects

Projects

Summary

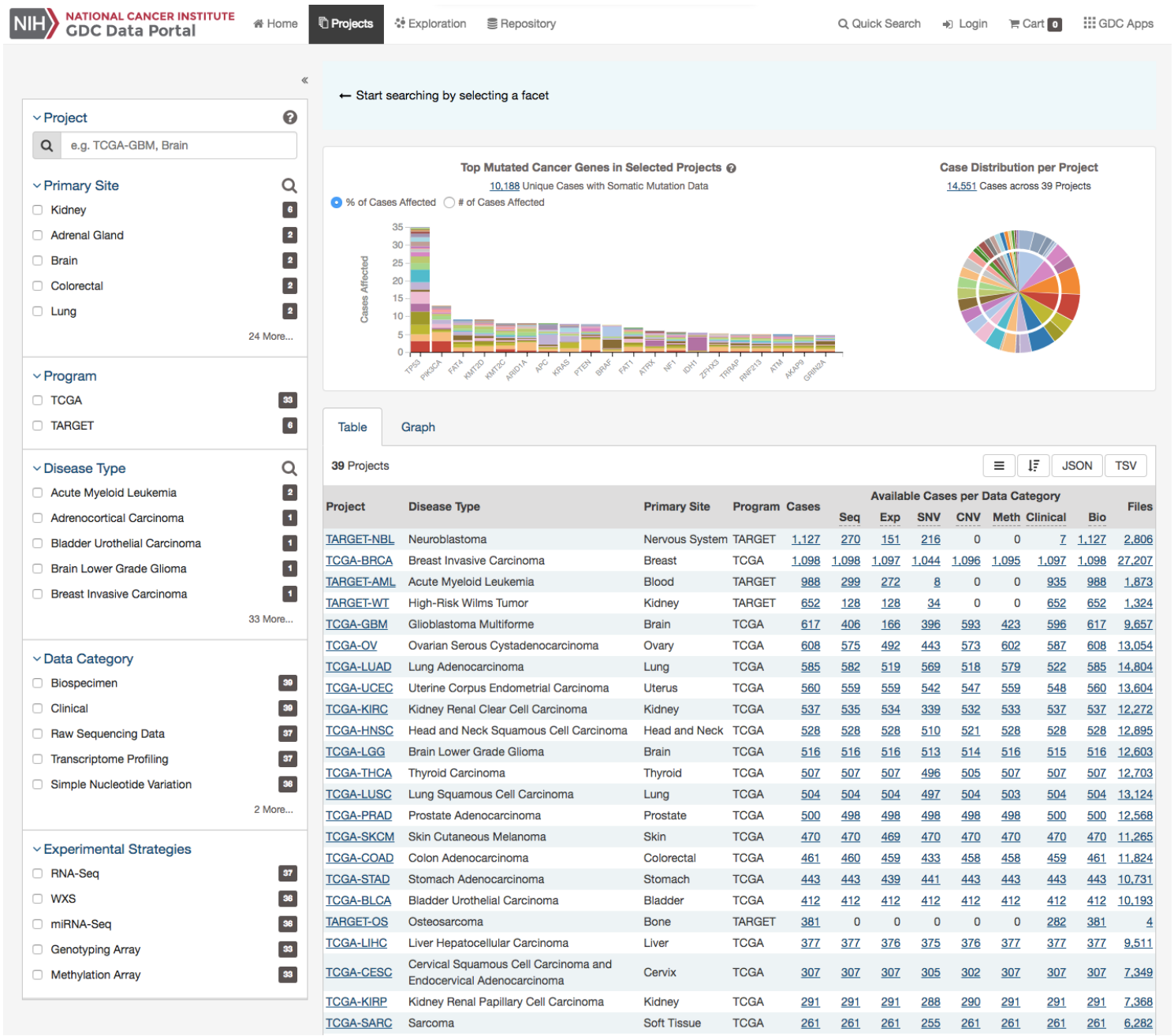
At a high level, data in the Genomic Data Commons is organized by project. Typically, a project is a specific effort to look at particular type(s) of cancer undertaken as part of a larger cancer research program. The GDC Data Portal allows users to access aggregate project-level information via the Projects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

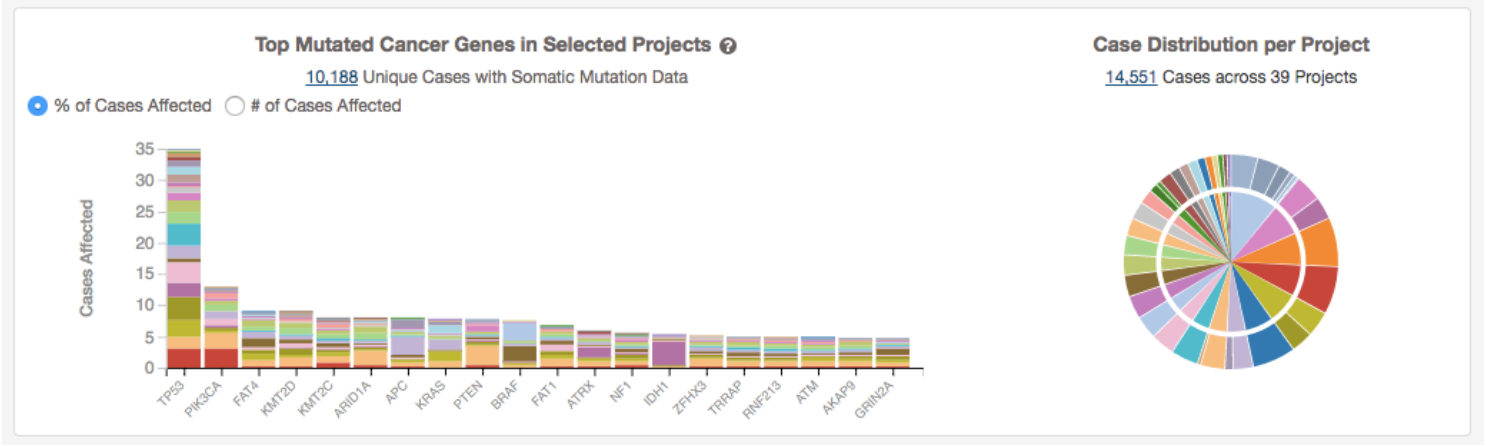
On the left, a panel of facets allow users to apply filters to find projects of interest. When facet filters are applied, the table and visualizations on the right are updated to display only the matching projects. When no filters are applied, all projects are displayed.

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	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
TCGA-KIRP	Kidney Renal Papillary Cell Carcinoma	Kidney	TCGA	291	291	291	288	290	291	291	291	7,368
TCGA-SARC	Sarcoma	Soft Tissue	TCGA	261	261	261	255	261	261	261	261	6,282

Visualizations



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.

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 page for the gene associated with the mutation.

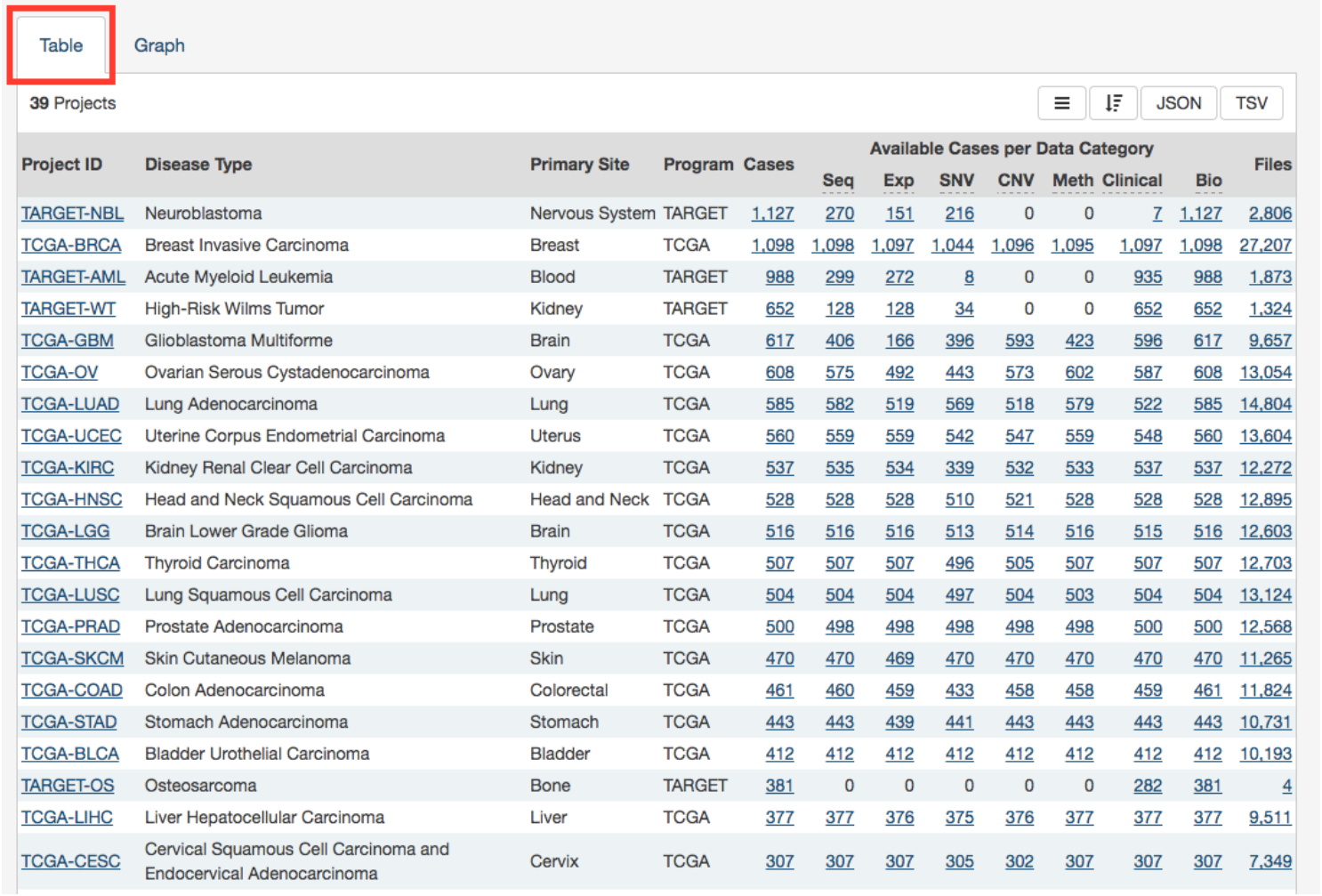
Users can toggle the Y-Axis of this bar graph between a percentage or raw number of cases affected.

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.

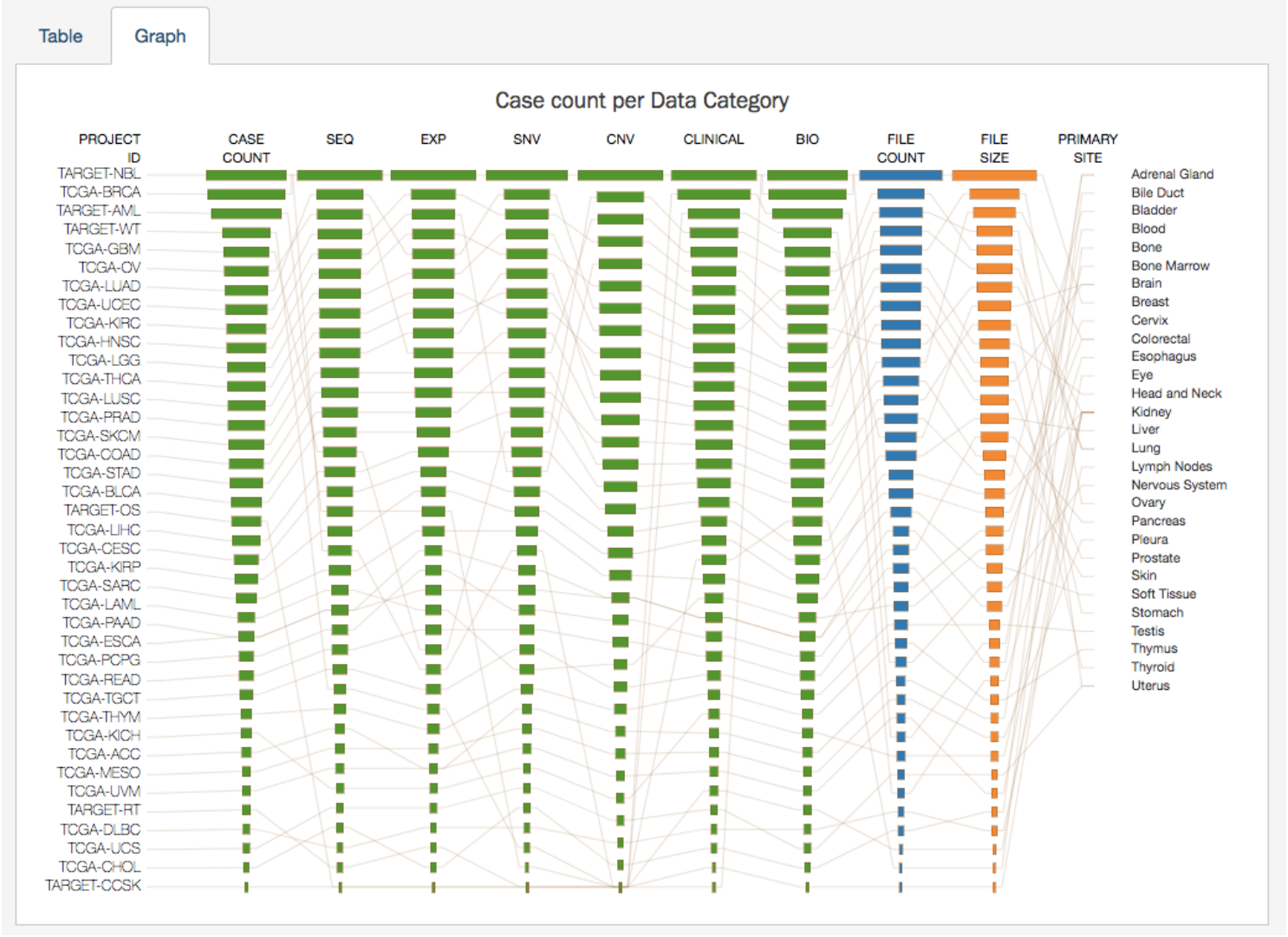


Project ID	Disease Type	Primary Site	Program	Cases	Available Cases per Data Category							Files
					Seq	Exp	SNV	CNV	Meth	Clinical	Bio	
TARGET-NBL	Neuroblastoma	Nervous System	TARGET	1,127	270	151	216	0	0	7	1,127	2,806
TCGA-BRCA	Breast Invasive Carcinoma	Breast	TCGA	1,098	1,098	1,097	1,044	1,096	1,095	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

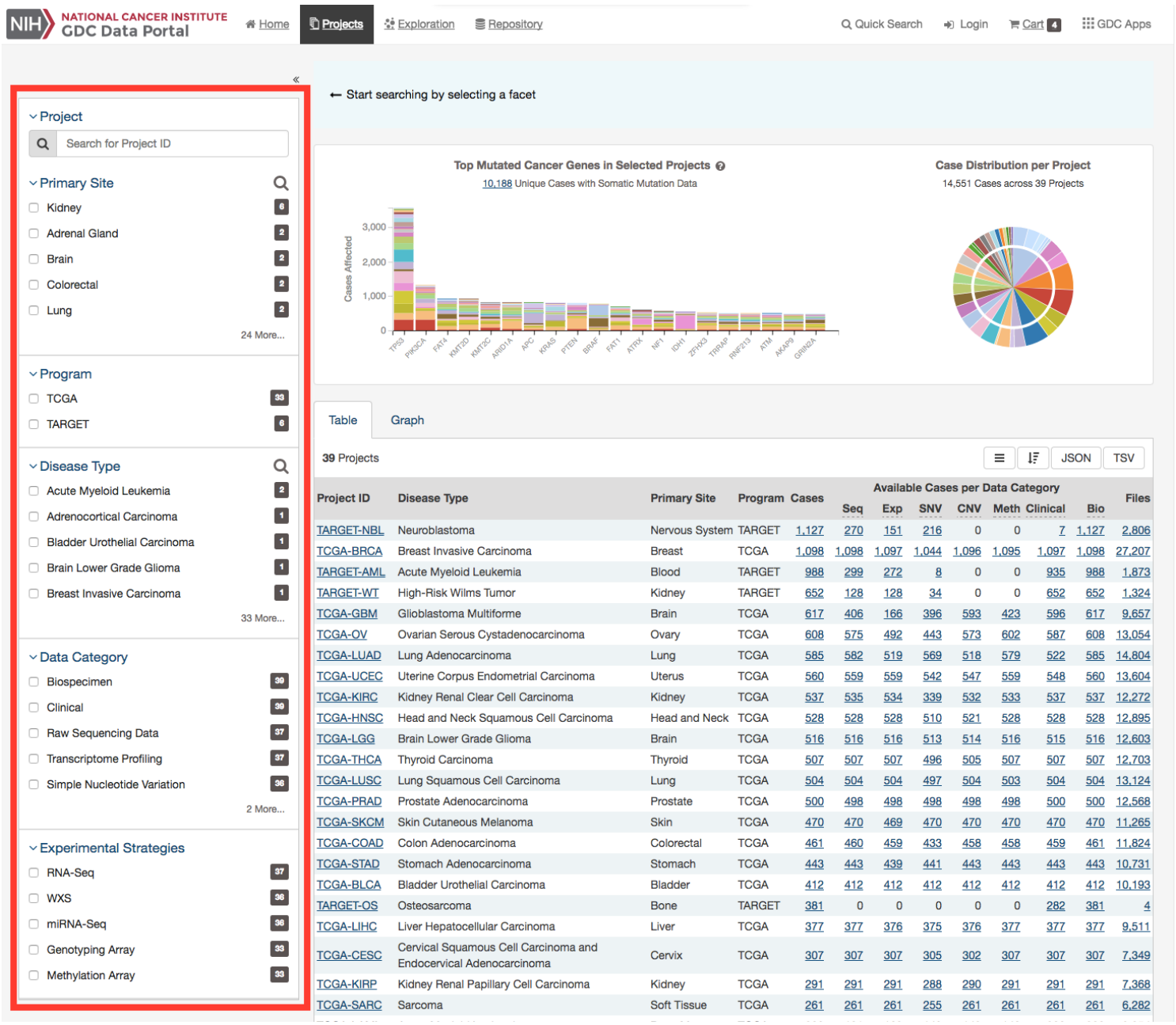


Most elements in the graph are clickable, allowing the user to open the associated cases or files in Repository Page.

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.

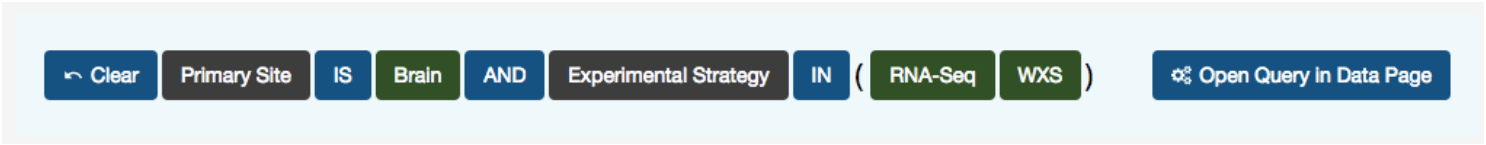


Project ID	Disease Type	Primary Site	Program	Cases	Available Cases per Data Category							Files
					Seq	Exp	SNV	CNV	Meth	Clinical	Bio	
TARGET-NBL	Neuroblastoma	Nervous System	TARGET	1,127	270	151	216	0	0	7	1,127	2,806
TCGA-BRCA	Breast Invasive Carcinoma	Breast	TCGA	1,098	1,098	1,097	1,044	1,096	1,095	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
TCGA-KIRP	Kidney Renal Papillary Cell Carcinoma	Kidney	TCGA	291	291	291	288	290	291	291	291	7,368
TCGA-SARC	Sarcoma	Soft Tissue	TCGA	261	261	261	255	261	261	261	261	6,282

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

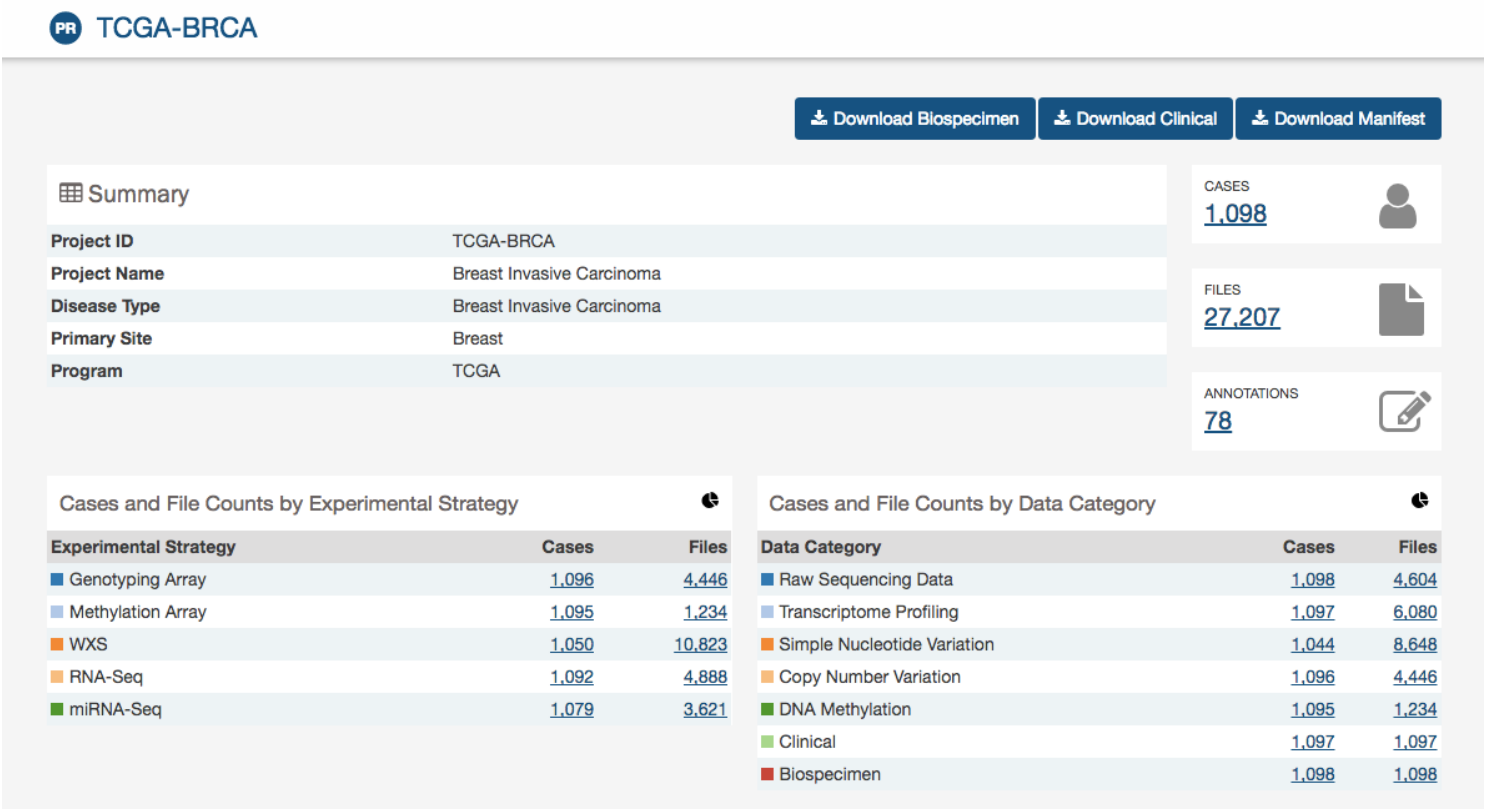
Filters can be applied by selecting values of interest in the available facets, for example "WXS" and "RNA-Seq" in the "Experimental Strategy" facet and "Brain" in the "Primary Site" facet. When facet filters are applied, the Table and Graph tabs are updated to display matching projects, and the banner 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.



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.

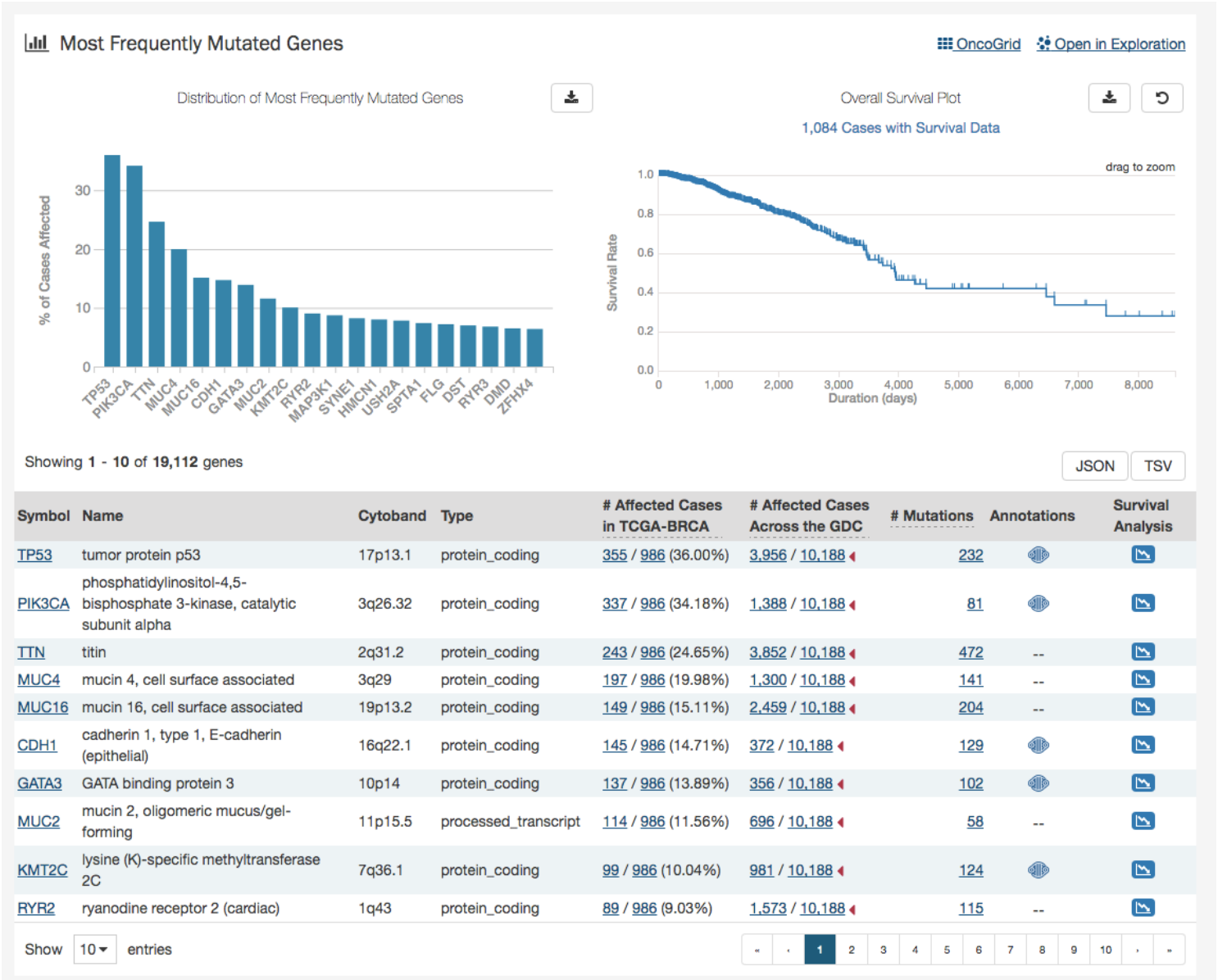


Three download buttons in the top right corner of the screen allow the user to download the entire project dataset, along with the associated project metadata:

- **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.



Symbol	Name	Cytoband	Type	# Affected Cases in TCGA-BRCA	# Affected Cases Across the GDC	# Mutations	Annotations	Survival Analysis
TP53	tumor protein p53	17p13.1	protein_coding	355 / 986 (36.00%)	3,956 / 10,188	232		
PIK3CA	phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit alpha	3q26.32	protein_coding	337 / 986 (34.18%)	1,388 / 10,188	81		
TTN	titin	2q31.2	protein_coding	243 / 986 (24.65%)	3,852 / 10,188	472	--	
MUC4	mucin 4, cell surface associated	3q29	protein_coding	197 / 986 (19.98%)	1,300 / 10,188	141	--	
MUC16	mucin 16, cell surface associated	19p13.2	protein_coding	149 / 986 (15.11%)	2,459 / 10,188	204	--	
CDH1	cadherin 1, type 1, E-cadherin (epithelial)	16q22.1	protein_coding	145 / 986 (14.71%)	372 / 10,188	129		
GATA3	GATA binding protein 3	10p14	protein_coding	137 / 986 (13.89%)	356 / 10,188	102		
MUC2	mucin 2, oligomeric mucus/gel-forming	11p15.5	processed_transcript	114 / 986 (11.56%)	696 / 10,188	58	--	
KMT2C	lysine (K)-specific methyltransferase 2C	7q36.1	protein_coding	99 / 986 (10.04%)	981 / 10,188	124		
RYR2	ryanodine receptor 2 (cardiac)	1q43	protein_coding	89 / 986 (9.03%)	1,573 / 10,188	115	--	

Show 10 entries

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:

$$S(t_i) = \prod_{t_i \leq t} \left(1 - \frac{d_i}{n_i}\right)$$

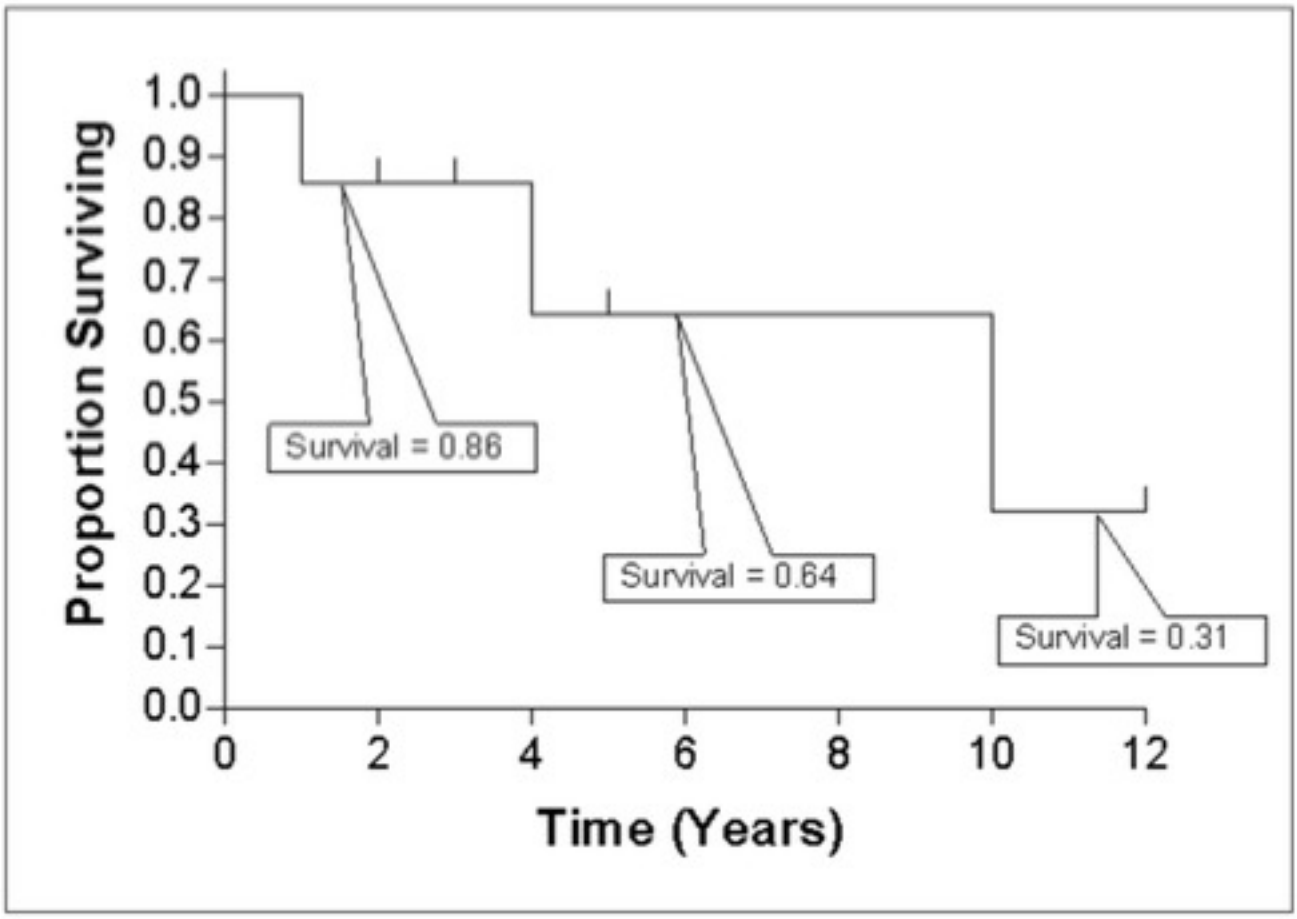
Where:

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

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

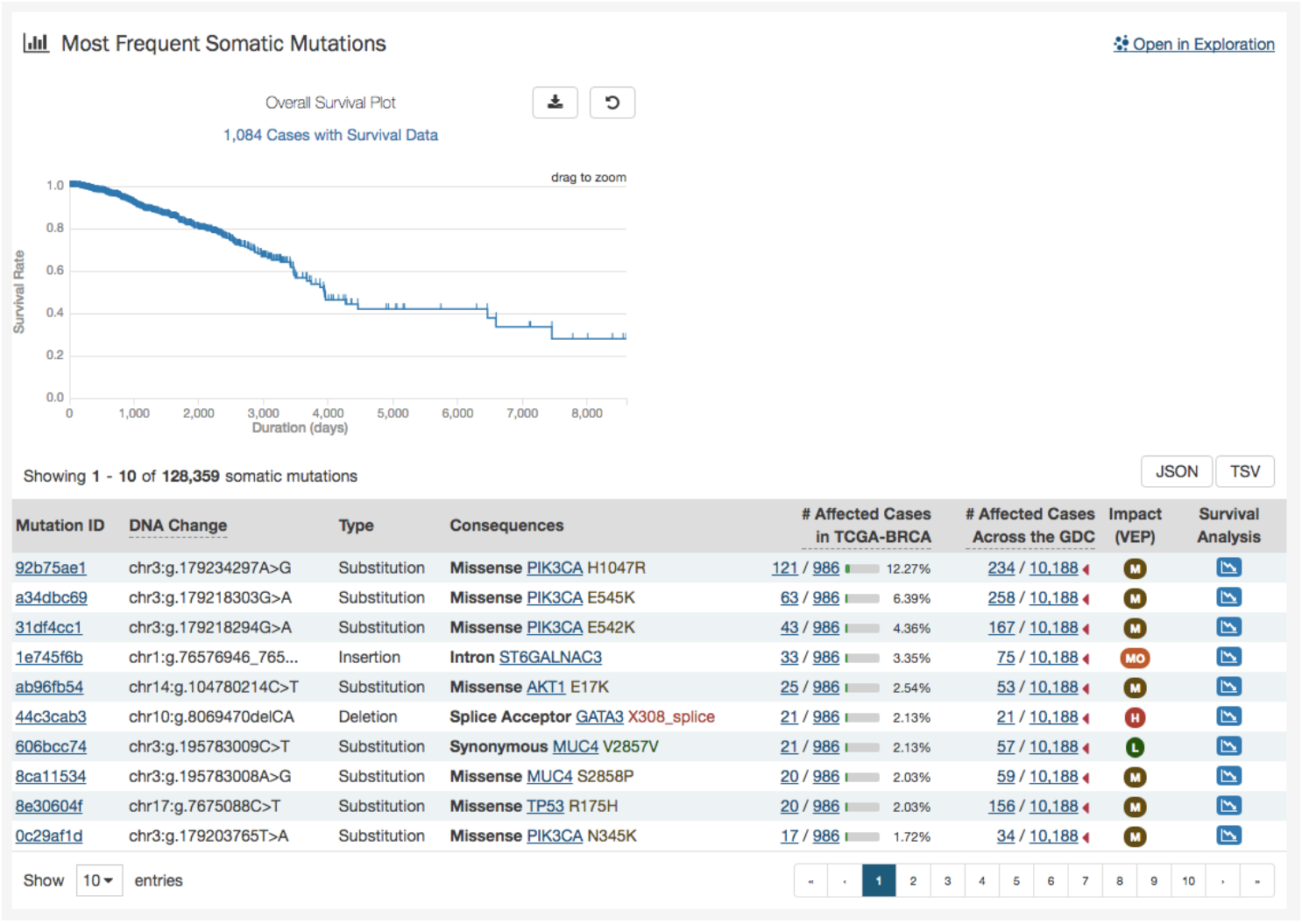
overall_survial_time (Years)	interval		# of donors at risk at start of interval (r)	# of censored donors during interval (c)	# of donors at risk at end of interval (n=r-c)	# of donors died at end of interval (d)	estimated interval survival probability ((n-d)/n)	estimated cumulative survival probability 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.



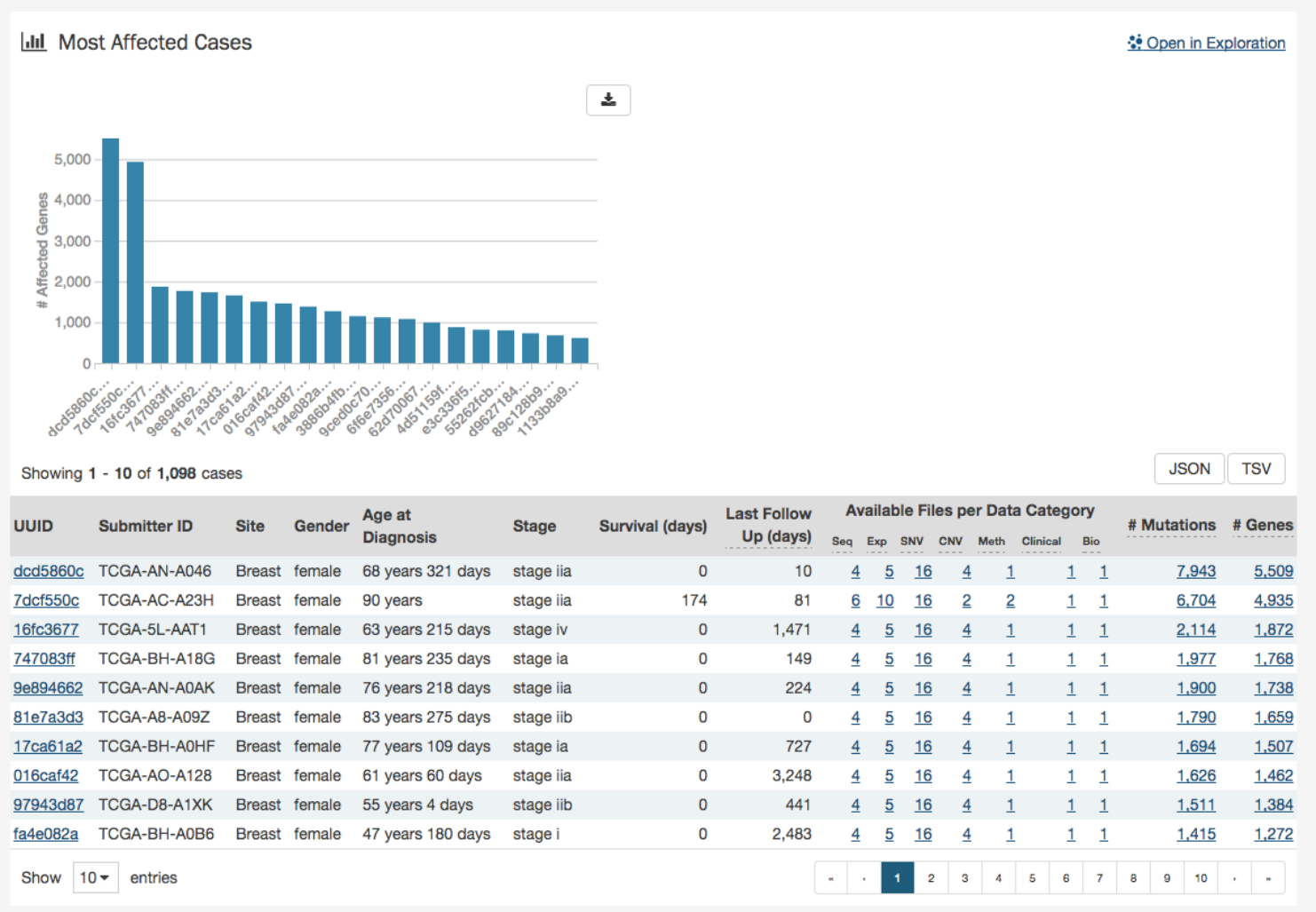
Mutation ID	DNA Change	Type	Consequences	# Affected Cases in TCGA-BRCA	# Affected Cases Across the GDC	Impact (VEP)	Survival Analysis
92b75ae1	chr3:g.179234297A>G	Substitution	**Missense** PIK3CA H1047R	121 / 986 12.27%	234 / 10,188	M	
a34dbc69	chr3:g.179218303G>A	Substitution	**Missense** PIK3CA E545K	63 / 986 6.39%	258 / 10,188	M	
31df4cc1	chr3:g.179218294G>A	Substitution	**Missense** PIK3CA E542K	43 / 986 4.36%	167 / 10,188	M	
1e745f6b	chr1:g.76576946_765...	Insertion	**Intron** ST6GALNAC3	33 / 986 3.35%	75 / 10,188	MO	
ab96fb54	chr14:g.104780214C>T	Substitution	**Missense** AKT1 E17K	25 / 986 2.54%	53 / 10,188	M	
44c3cab3	chr10:g.8069470delCA	Deletion	**Splice Acceptor** GATA3 X308_splice	21 / 986 2.13%	21 / 10,188	H	
606bcc74	chr3:g.195783009C>T	Substitution	**Synonymous** MUC4 V2857V	21 / 986 2.13%	57 / 10,188	L	
8ca11534	chr3:g.195783008A>G	Substitution	**Missense** MUC4 S2858P	20 / 986 2.03%	59 / 10,188	M	
8e30604f	chr17:g.7675088C>T	Substitution	**Missense** TP53 R175H	20 / 986 2.03%	156 / 10,188	M	
0c29af1d	chr3:g.179203765T>A	Substitution	**Missense** PIK3CA N345K	17 / 986 1.72%	34 / 10,188	M	

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 (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
- **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.



UUID	Submitter ID	Site	Gender	Age at Diagnosis	Stage	Survival (days)	Last Follow Up (days)	Available Files per Data Category							# Mutations	# Genes
								Seq	Exp	SNV	CNV	Meth	Clinical	Bio		
dcd5860c	TCGA-AN-A046	Breast	female	68 years 321 days	stage iia	0	10	4	5	16	4	1	1	1	7,943	5,509
7dcf550c	TCGA-AC-A23H	Breast	female	90 years	stage iia	174	81	6	10	16	2	2	1	1	6,704	4,935
16fc3677	TCGA-5L-AAT1	Breast	female	63 years 215 days	stage iv	0	1,471	4	5	16	4	1	1	1	2,114	1,872
747083ff	TCGA-BH-A18G	Breast	female	81 years 235 days	stage ia	0	149	4	5	16	4	1	1	1	1,977	1,768
9e894662	TCGA-AN-A0AK	Breast	female	76 years 218 days	stage iia	0	224	4	5	16	4	1	1	1	1,900	1,738
81e7a3d3	TCGA-A8-A09Z	Breast	female	83 years 275 days	stage iib	0	0	4	5	16	4	1	1	1	1,790	1,659
17ca61a2	TCGA-BH-A0HF	Breast	female	77 years 109 days	stage ia	0	727	4	5	16	4	1	1	1	1,694	1,507
016caf42	TCGA-AO-A128	Breast	female	61 years 60 days	stage iia	0	3,248	4	5	16	4	1	1	1	1,626	1,462
97943d87	TCGA-D8-A1XK	Breast	female	55 years 4 days	stage iib	0	441	4	5	16	4	1	1	1	1,511	1,384
fa4e082a	TCGA-BH-A0B6	Breast	female	47 years 180 days	stage i	0	2,483	4	5	16	4	1	1	1	1,415	1,272

Show 10 entries

« ‹ 1 2 3 4 5 6 7 8 9 10 › »

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

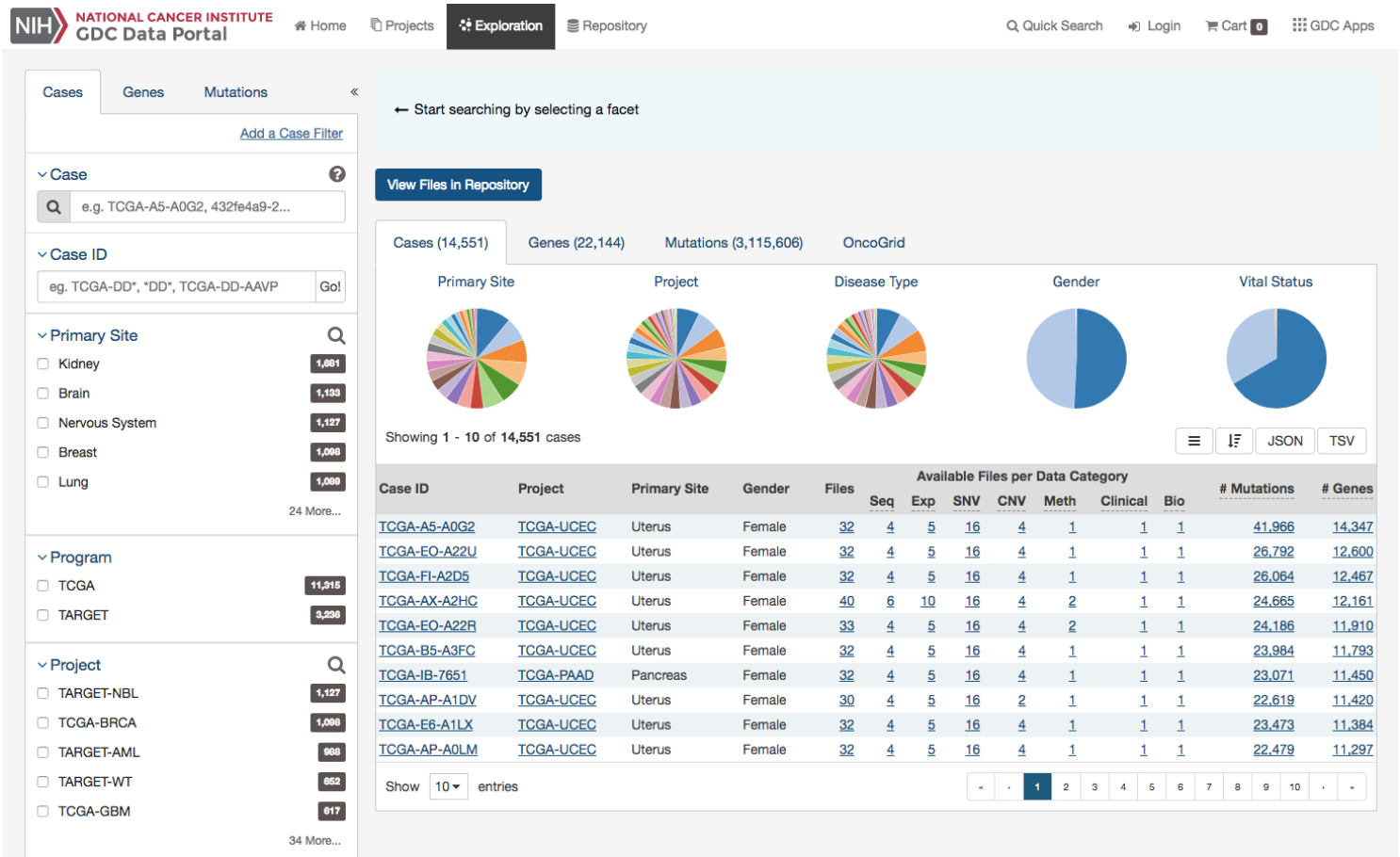
- **UUID:** The UUID of the case, which links to the Case Summary Page
- **Submitter ID:** The Submitter ID of the case (i.e. the TCGA Barcode)
- **Site:** The anatomical location of the site affected
- **Gender:** Text designations that identify gender. Gender is described as the assemblage of properties 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.

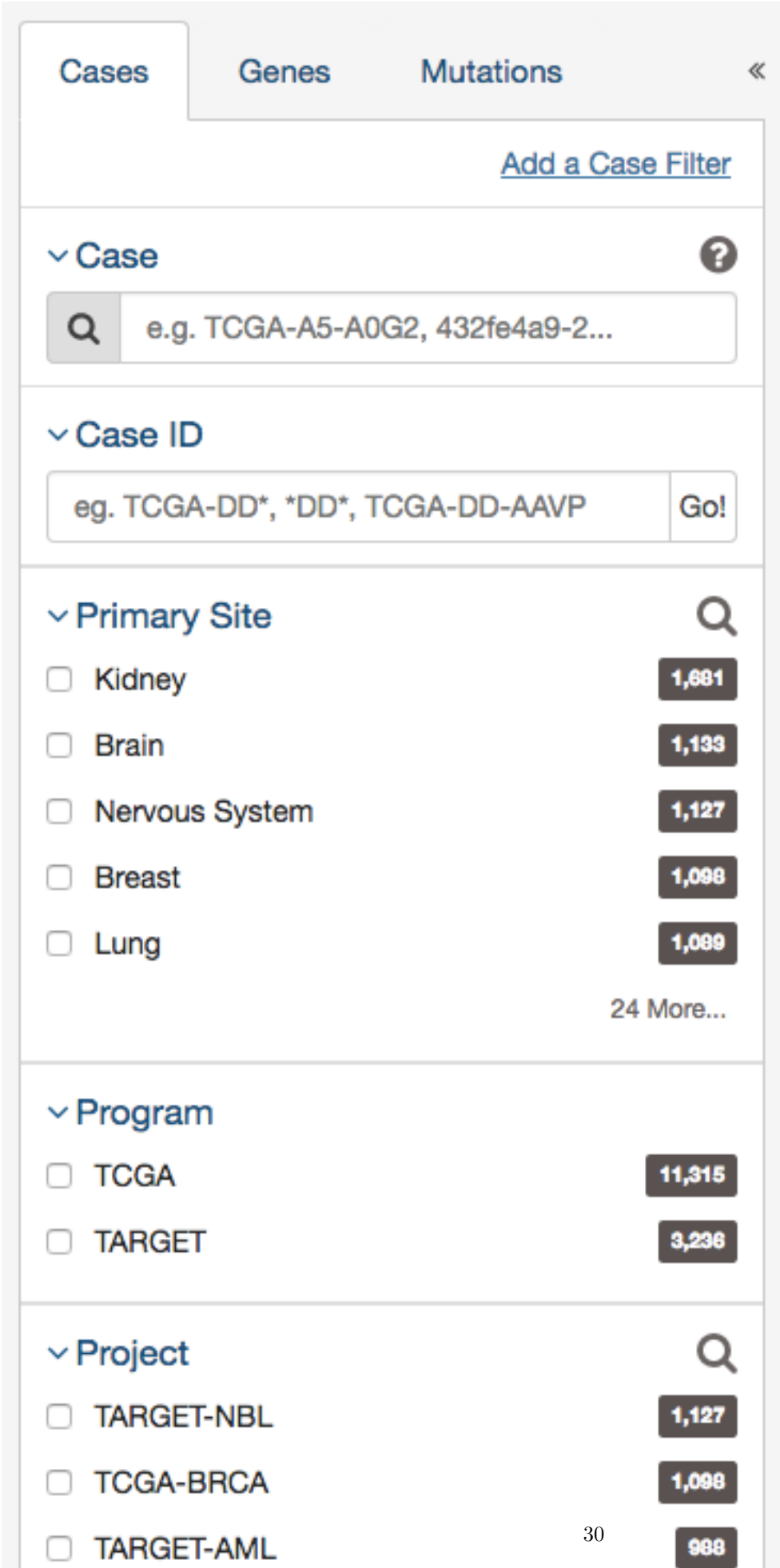


Filters / Facets

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

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...
Case ID
eg. TCGA-DD*, *DD*, TCGA-DD-AAVP
Go!
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

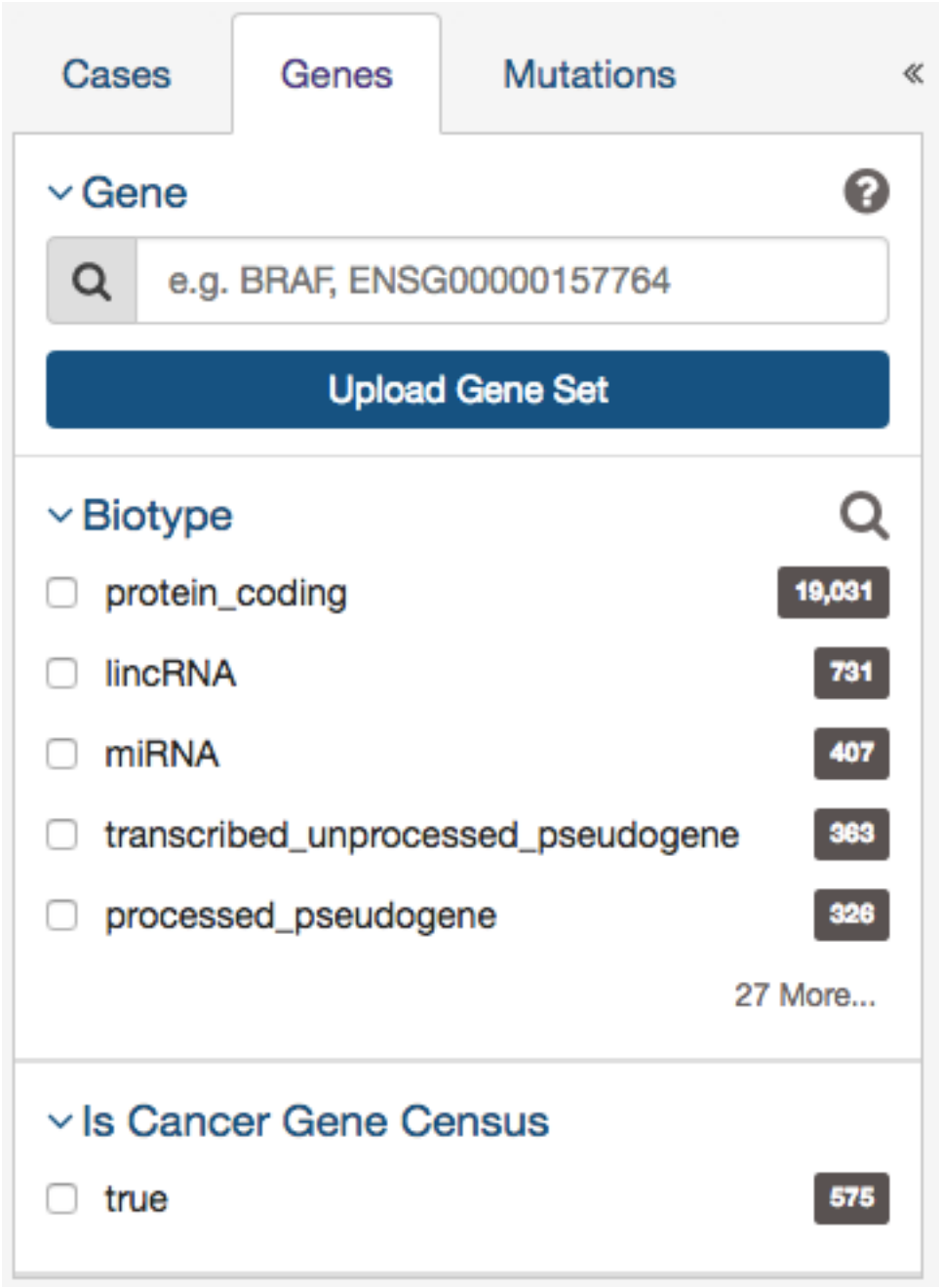
These criteria limit the results only to specific cases within the GDC. The default filters available are:

- **Case**: Specify individual cases using submitter ID (barcode) or UUID.
- **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'

Gene Filters

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

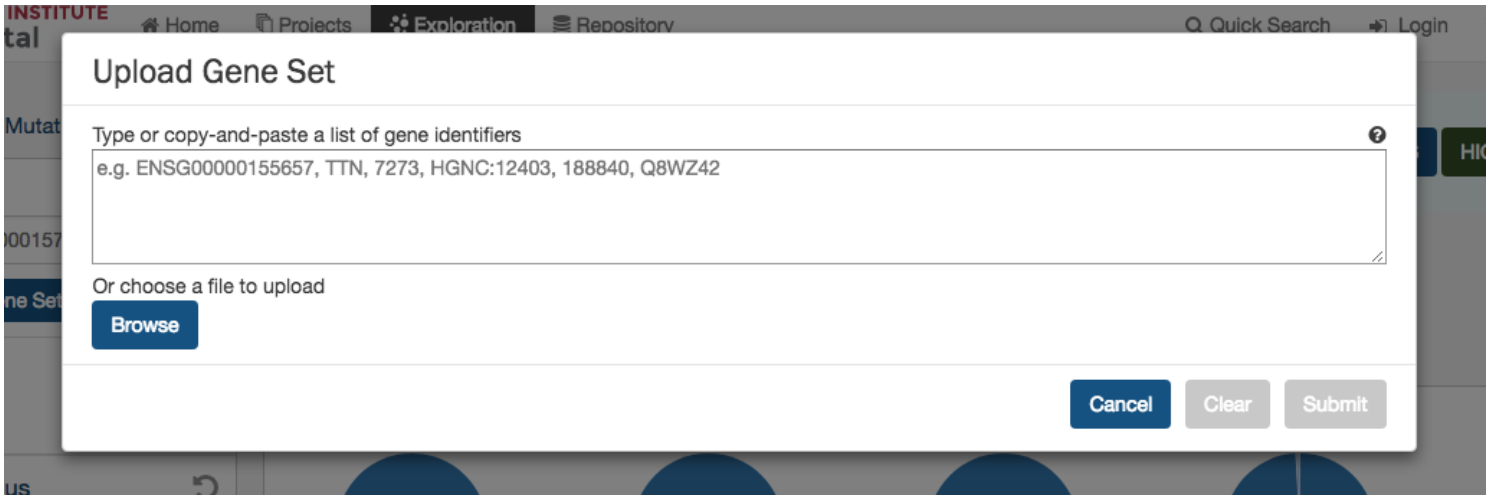


The second tab of filters are for specific genes. Users can filter by:

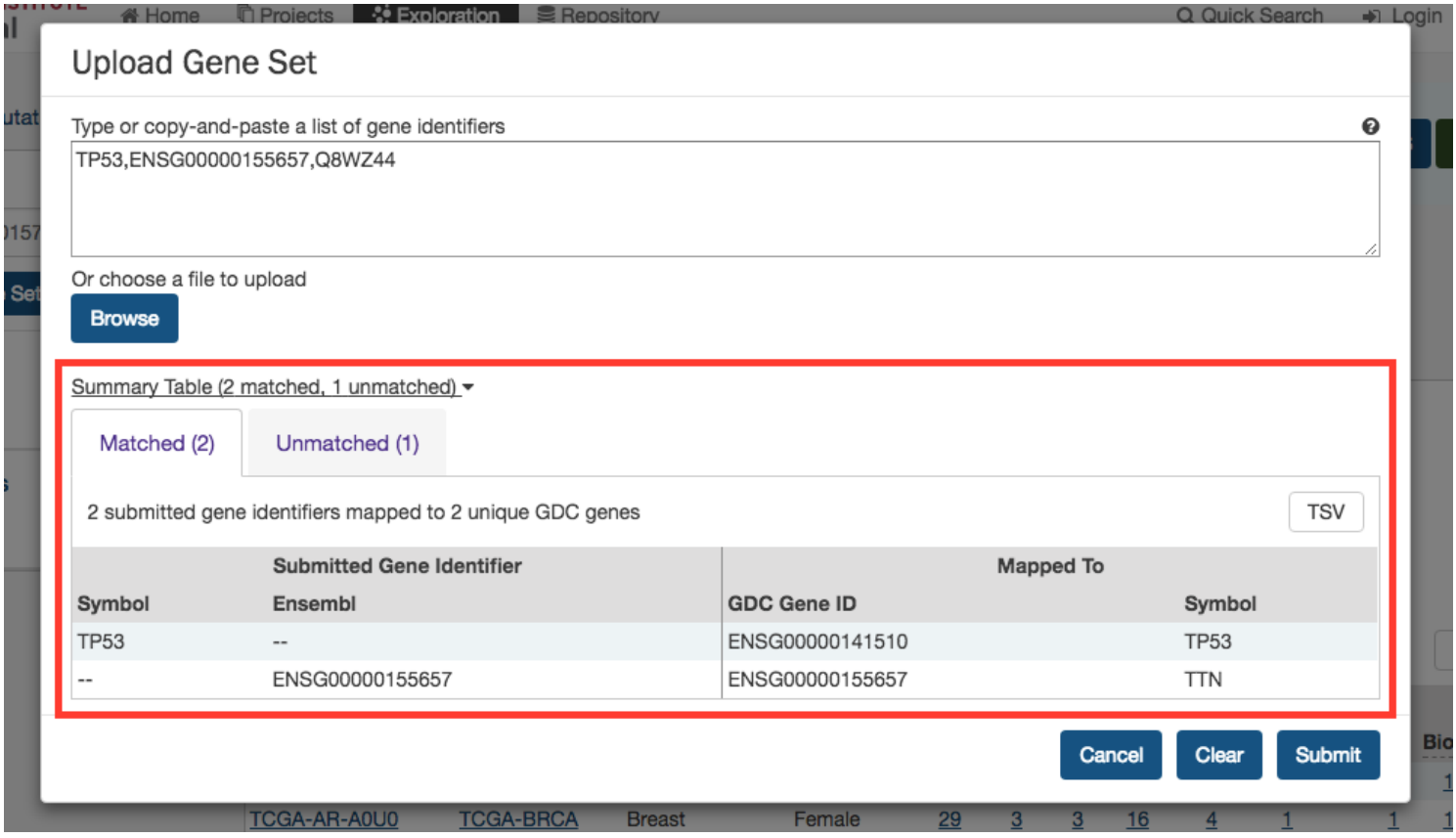
- **Gene** - Entering in a specific Gene Symbol, ID, or List of Genes ('Gene Set')
- **Biotype** - Classification of the type of gene according to Ensembl. The biotypes can be grouped into protein coding, pseudogene, long noncoding and short noncoding. Examples of biotypes in each group are as follows:
  - **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.



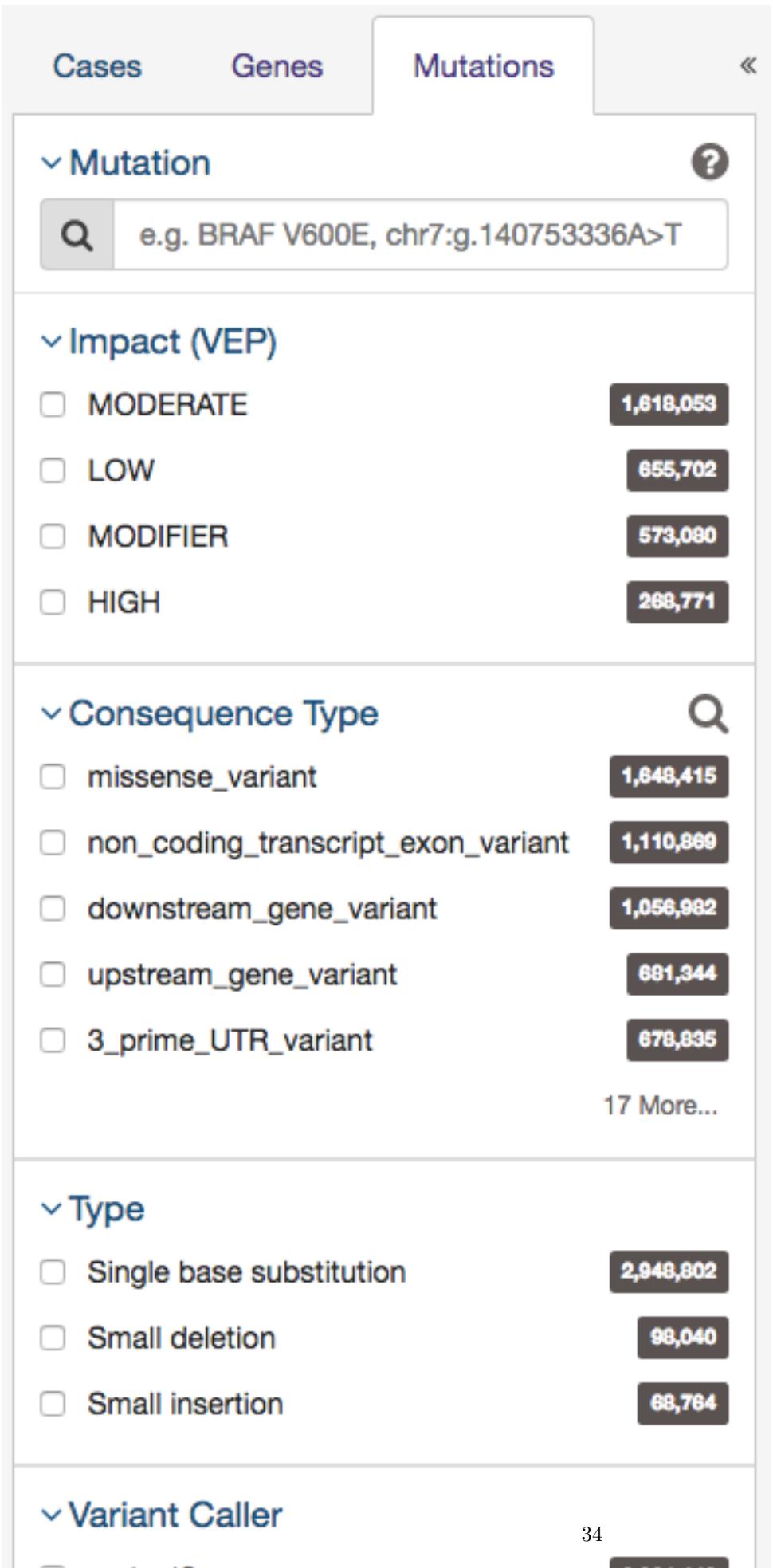
After supplying a list of genes, a table below will appear which indicates whether the gene was found.



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
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
Variant Caller

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
- **Consequence Type** - Consequence type of this variation; sequence ontology terms
- **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
- **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

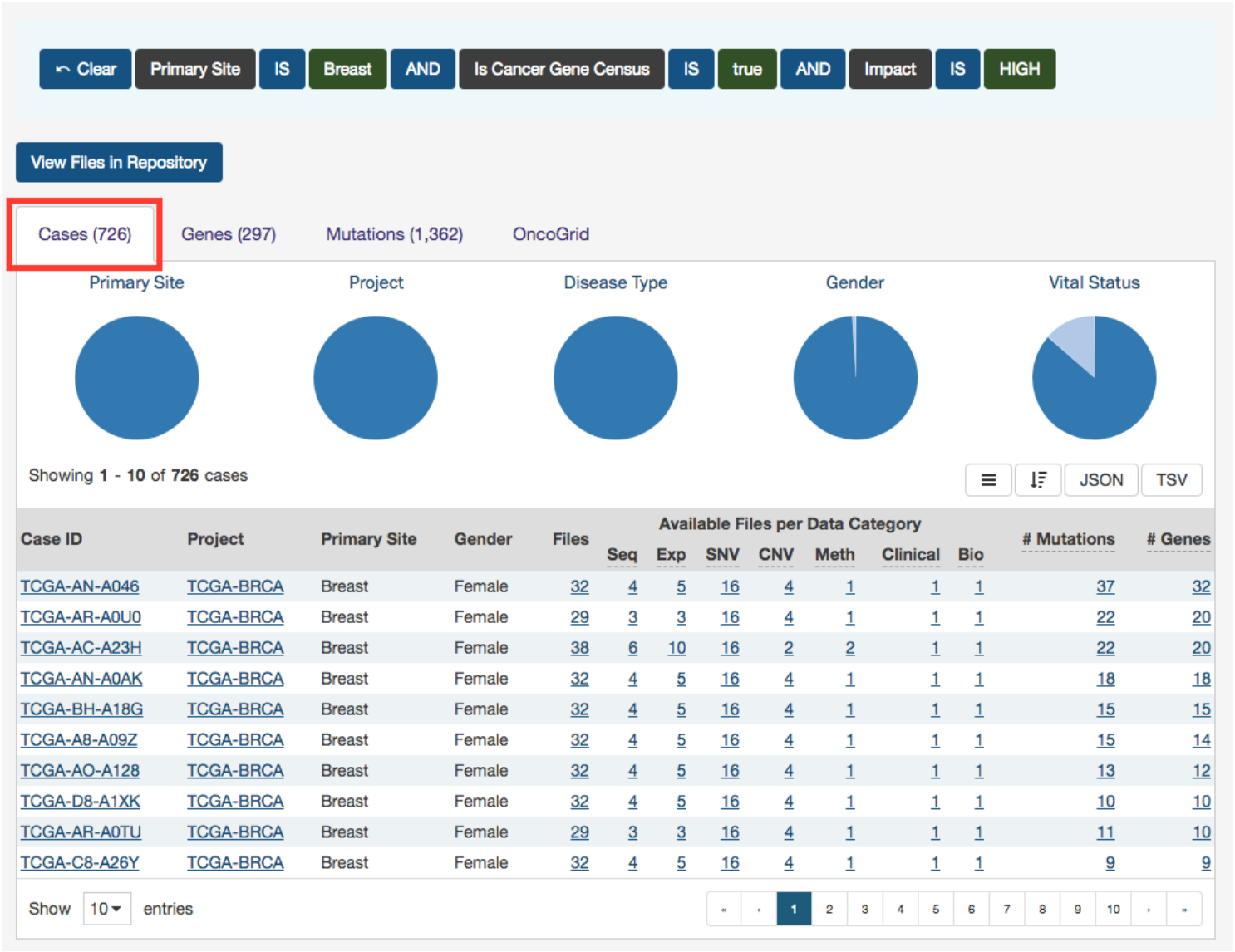
Results

As users add filters to the data on the Exploration Page, the Results section will automatically be updated. Results are divided into different tabs: `Cases`, `Genes`, `Mutations`, and `OncoGrid`.

To illustrate these tabs, Case, Gene, and Mutation filters have been chosen (Genes in the Cancer Gene Census, that have HIGH Impact for the TCGA-BRCA project) and a description of what each tab displays follows.

Cases

The `Cases` tab will give an overview of all the cases/patients who correspond to the filters chosen (Cohort).



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

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

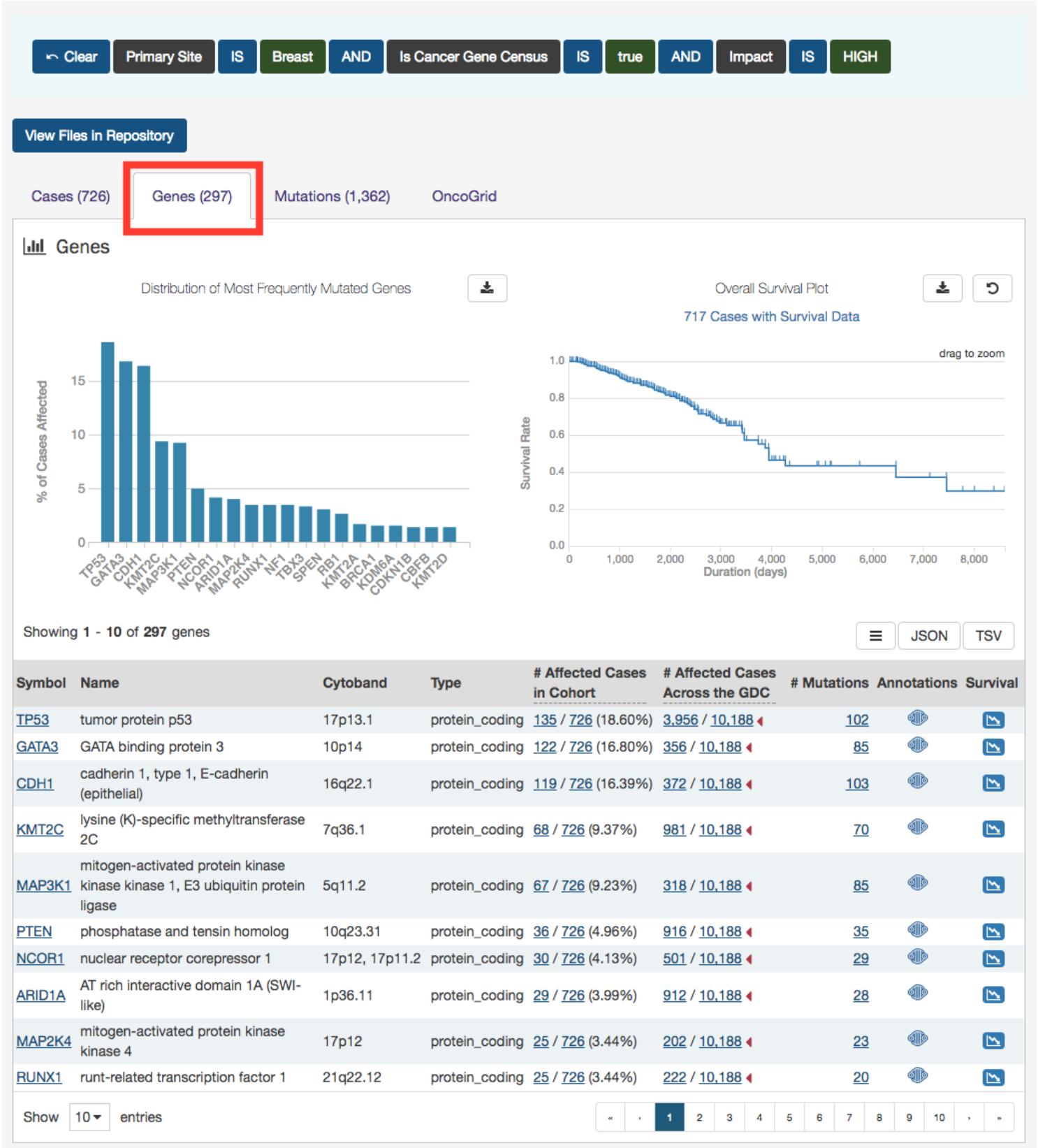
Below these pie charts is a tabular view (which can be exported and sorted using the buttons on the right) of the cases affected, which includes the following information:

- **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 detected 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).



Symbol	Name	Cytoband	Type	# Affected Cases in Cohort	# Affected Cases Across the GDC	# Mutations	Annotations	Survival
TP53	tumor protein p53	17p13.1	protein_coding	135 / 726 (18.60%)	3,956 / 10,188	102		
GATA3	GATA binding protein 3	10p14	protein_coding	122 / 726 (16.80%)	356 / 10,188	85		
CDH1	cadherin 1, type 1, E-cadherin (epithelial)	16q22.1	protein_coding	119 / 726 (16.39%)	372 / 10,188	103		
KMT2C	lysine (K)-specific methyltransferase 2C	7q36.1	protein_coding	68 / 726 (9.37%)	981 / 10,188	70		
MAP3K1	mitogen-activated protein kinase kinase kinase 1, E3 ubiquitin protein ligase	5q11.2	protein_coding	67 / 726 (9.23%)	318 / 10,188	85		
PTEN	phosphatase and tensin homolog	10q23.31	protein_coding	36 / 726 (4.96%)	916 / 10,188	35		
NCOR1	nuclear receptor corepressor 1	17p12, 17p11.2	protein_coding	30 / 726 (4.13%)	501 / 10,188	29		
ARID1A	AT rich interactive domain 1A (SWI-like)	1p36.11	protein_coding	29 / 726 (3.99%)	912 / 10,188	28		
MAP2K4	mitogen-activated protein kinase kinase 4	17p12	protein_coding	25 / 726 (3.44%)	202 / 10,188	23		
RUNX1	runt-related transcription factor 1	21q22.12	protein_coding	25 / 726 (3.44%)	222 / 10,188	20		

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

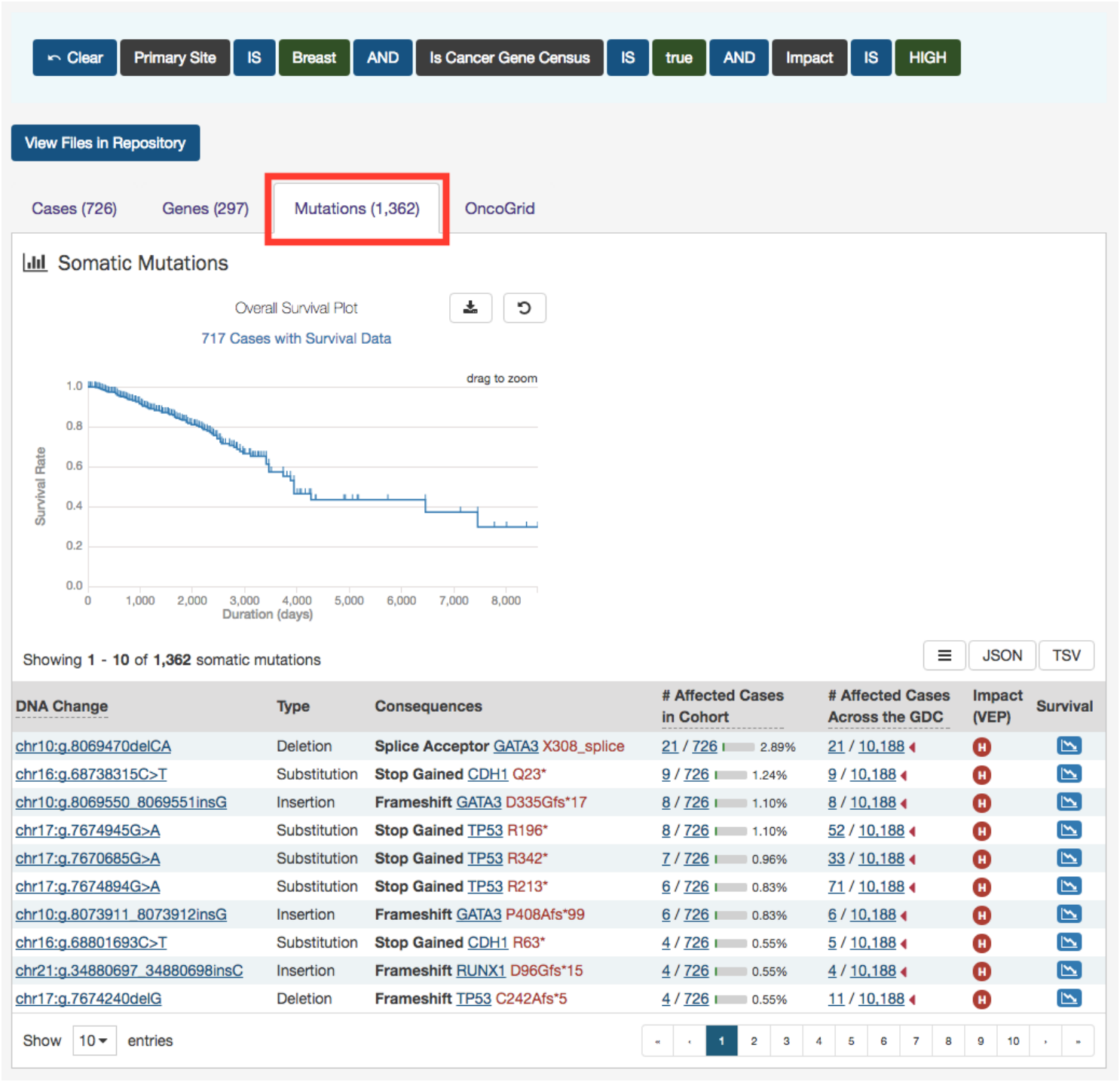
cases affected. Users may choose to download the underlying data in JSON or TSV format or an image of the graph in SVG or PNG format by clicking the `download` icon at the top of each graph.

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

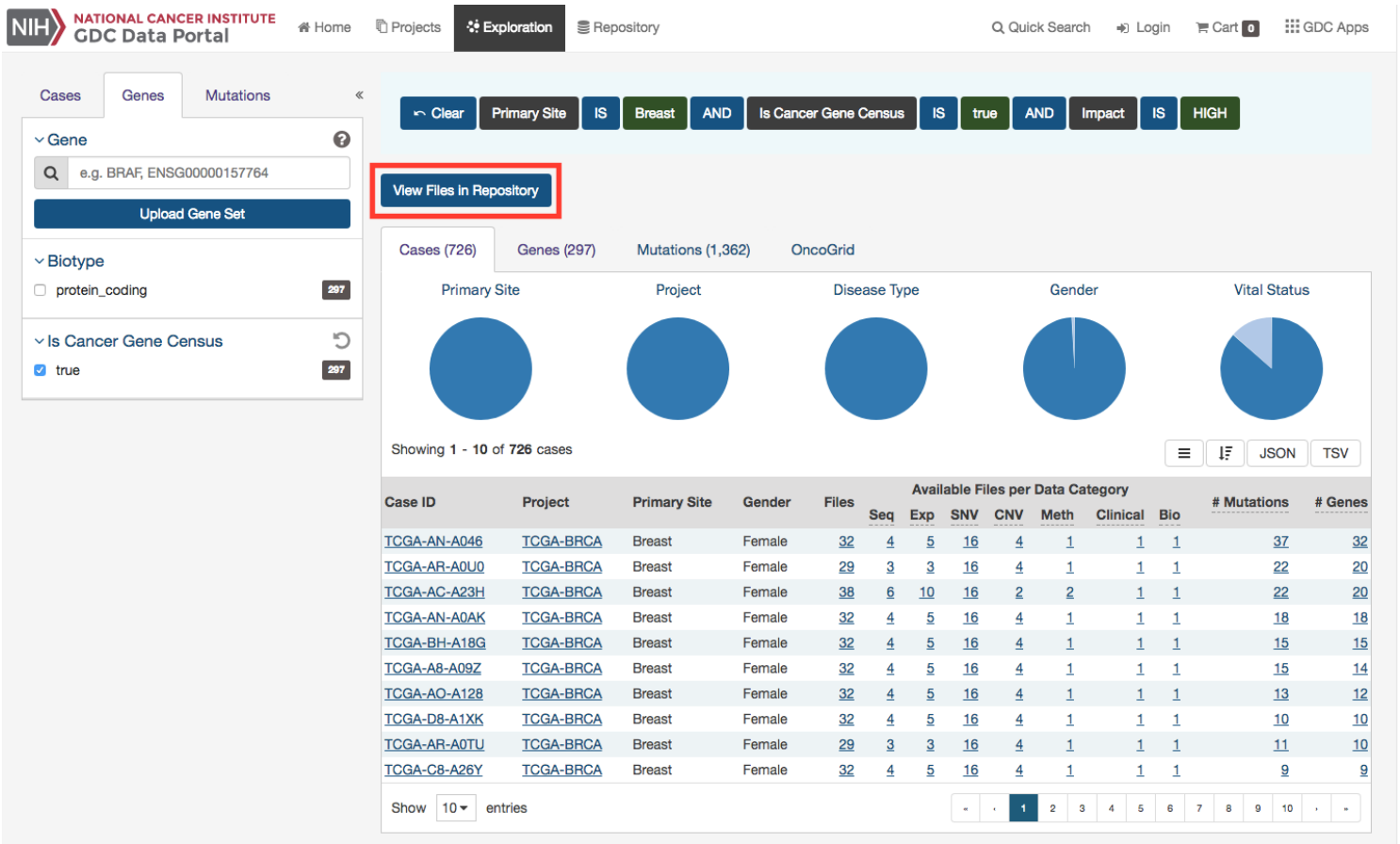
To facilitate readability and comparisons, drag-and-drop can be used to reorder the gene rows. Double clicking a row in the " Cases Affected" bar at the right side of the graphic launches the respective Gene Summary Page page. Hovering over a cell will display information about the mutation such as its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



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

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

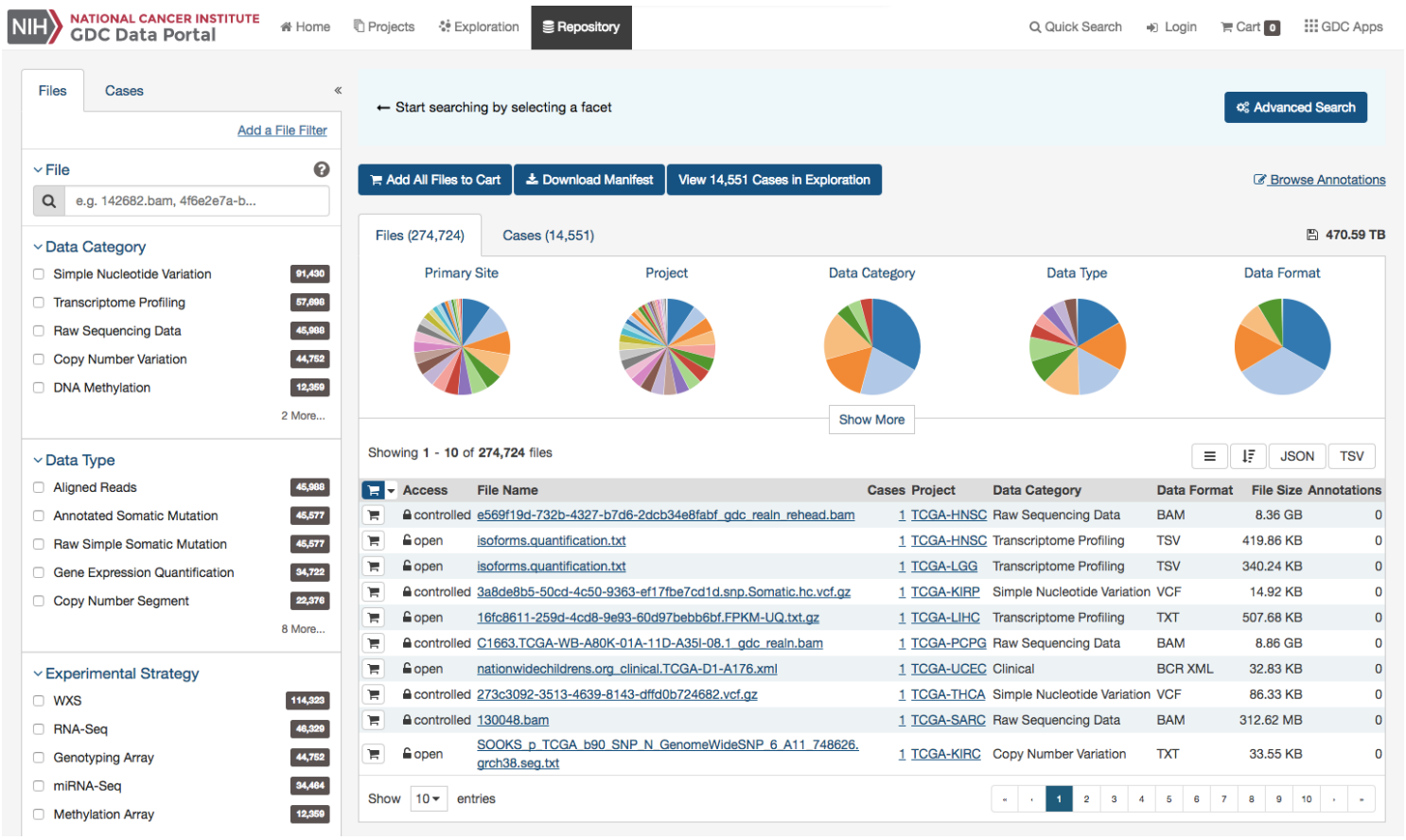
On the left, a panel of data facets allows users to filter cases and files using a variety of criteria. If facet filters are applied, the tabs on the right will display information about matching cases and files. If no filters are applied, the tabs on the right will display information about all available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

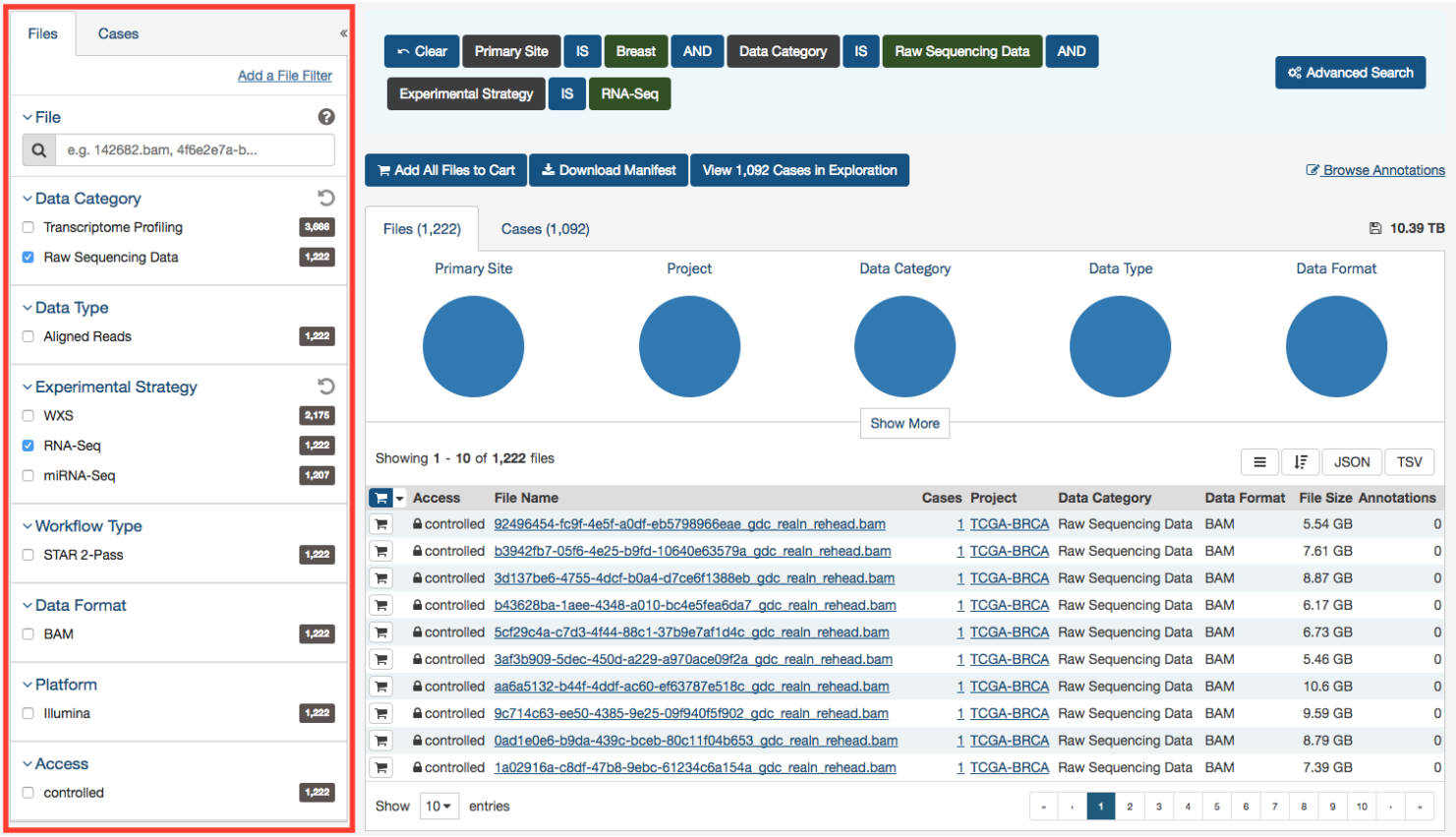


Facets Panel

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 default set of facets is listed below.

Files facets tab:

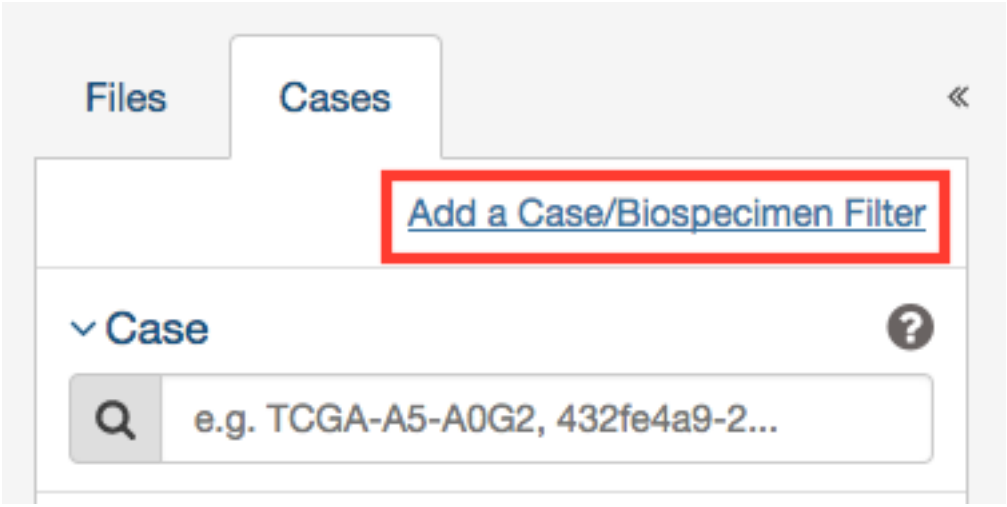
- **File**: Specify individual files using filename or UUID.
- **Data Category**: A high-level data file category, such as "Raw Sequencing Data" or "Transcriptome Profiling".
- **Data Type**: Data file type, such as "Aligned Reads" or "Gene Expression Quantification". Data Type is more granular than 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:

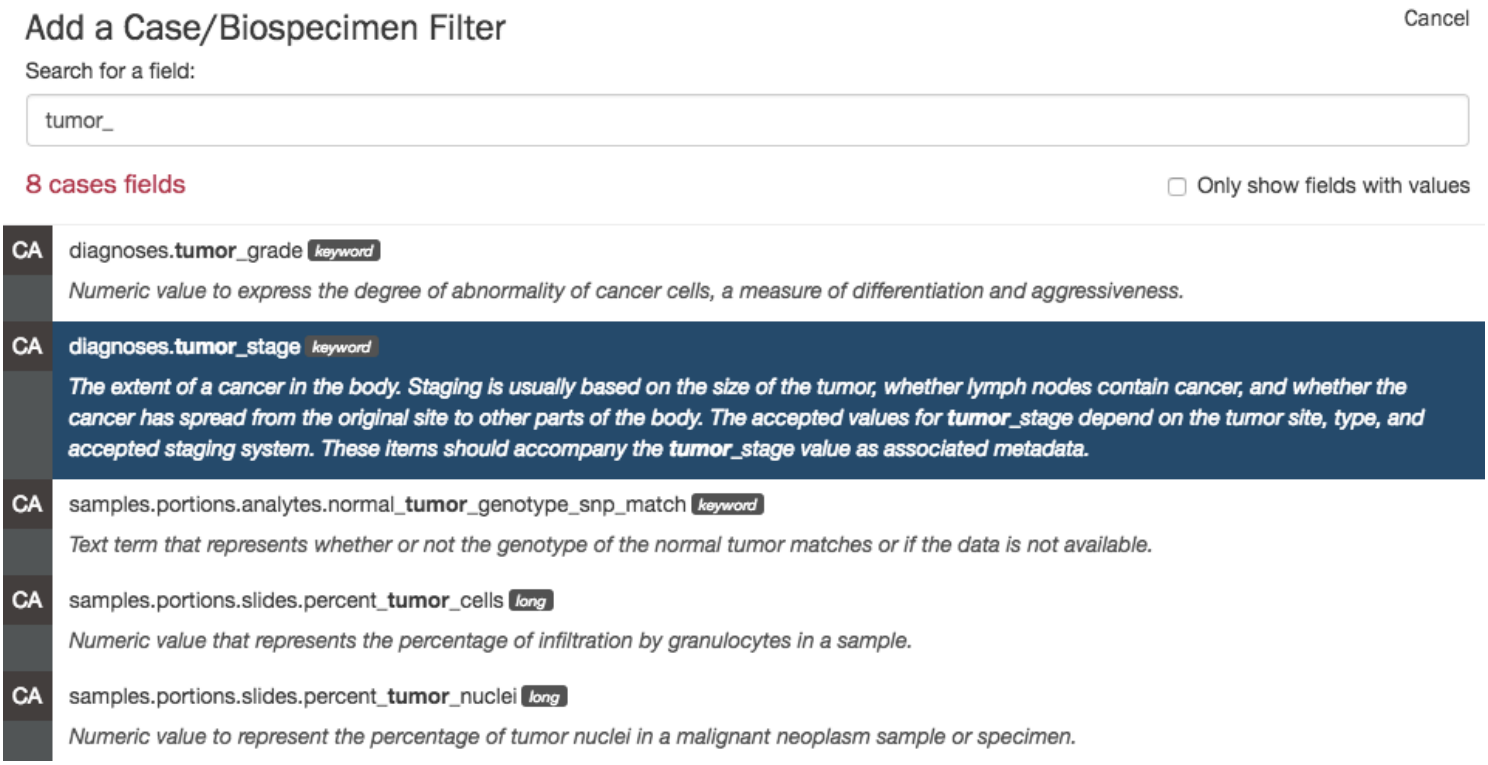
- **Case**: Specify individual cases using submitter ID (barcode) or UUID.
- **Case Submitter ID Prefix**: Search for cases using a part (prefix) of the submitter ID (barcode).
- **Primary Site**: Anatomical site of the cancer under investigation or review.
- **Cancer Program**: A cancer research program, 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

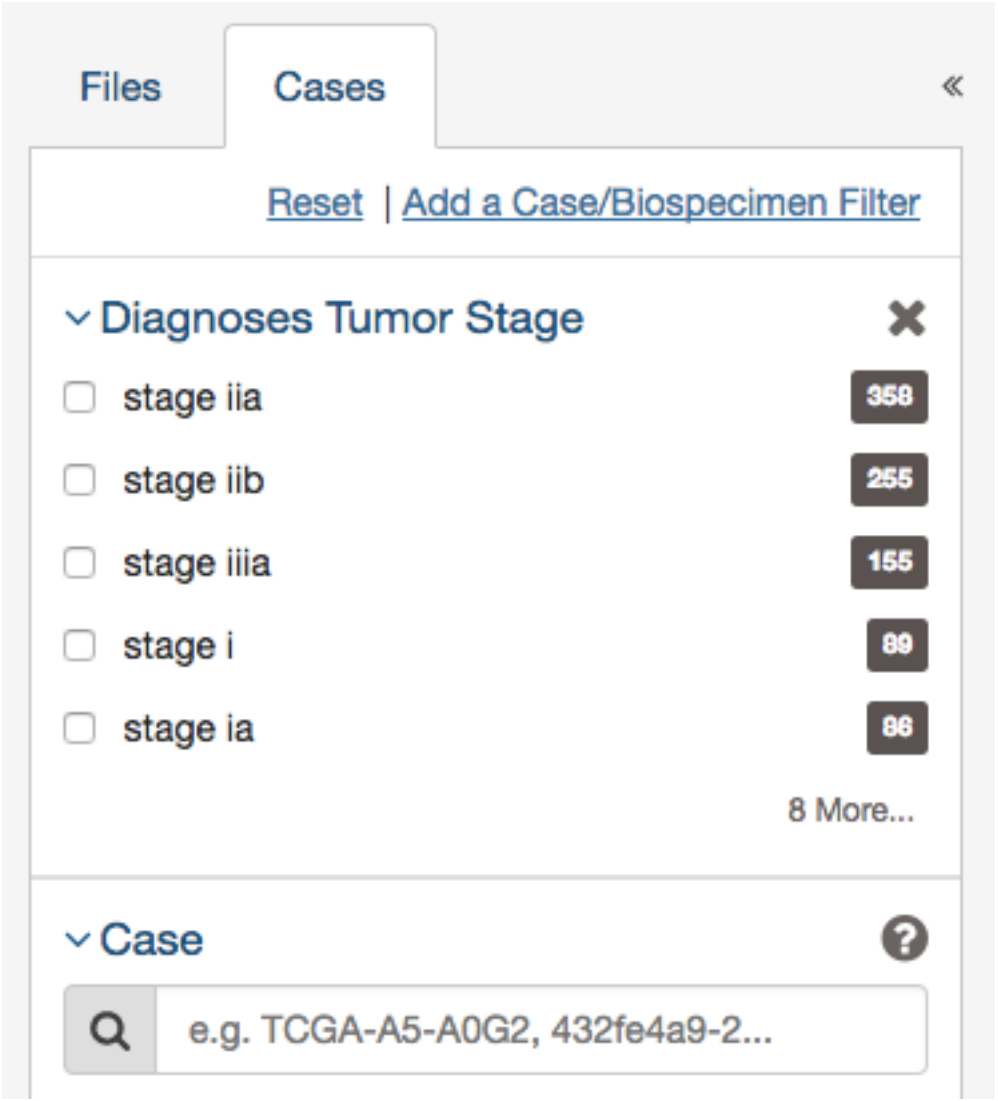
The Repository Page provides access to additional data facets beyond those listed above. Facets corresponding to additional properties listed in the GDC Data Dictionary can be added using the "add a filter" links available at the top of the Cases and Files facet tabs:



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.



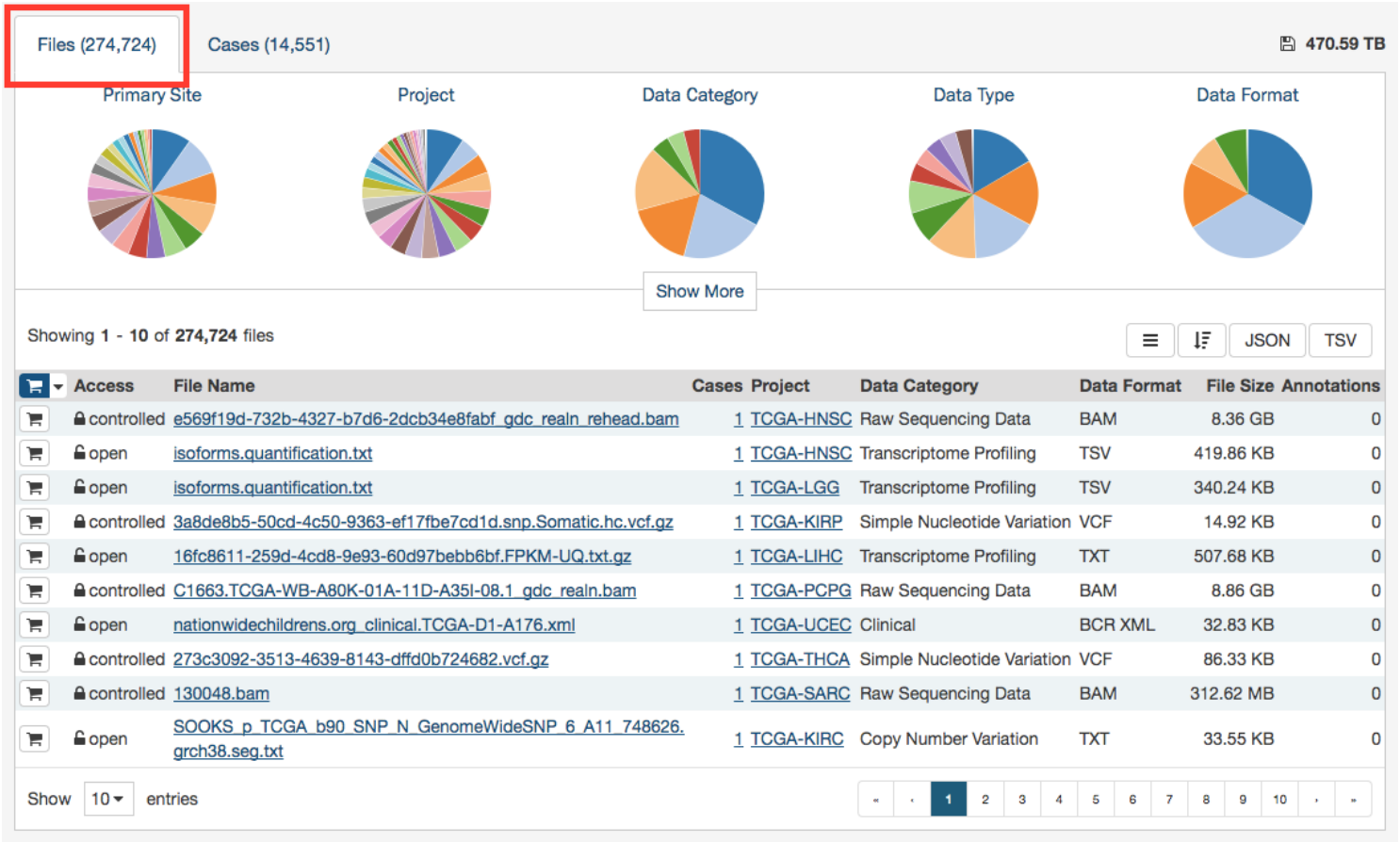
Newly added facets will show up at the top of the facets panel and can be removed individually by clicking on the red cross to the right of the facet name. The default set of facets can be restored by clicking "Reset".



Results

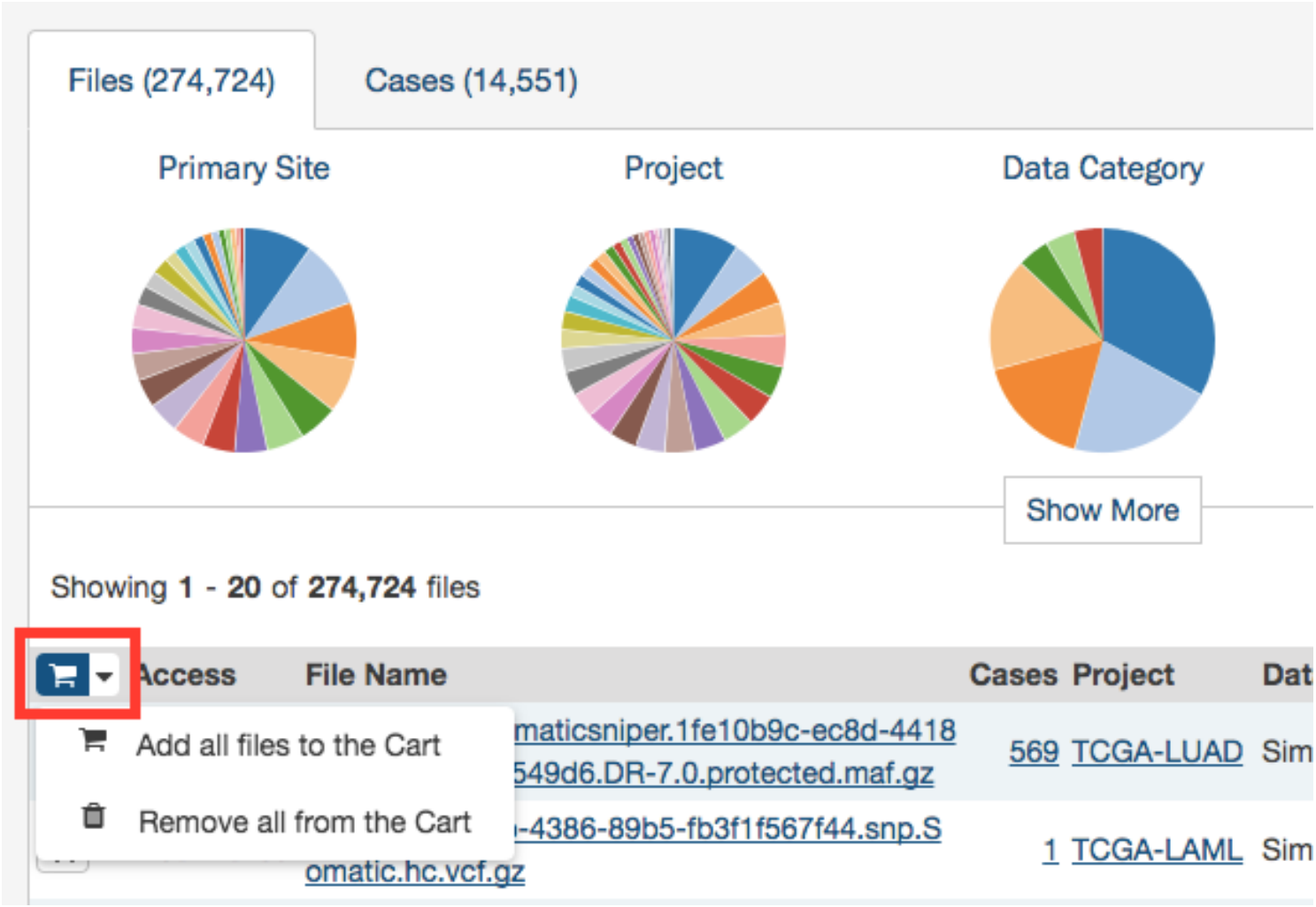
Files List

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.



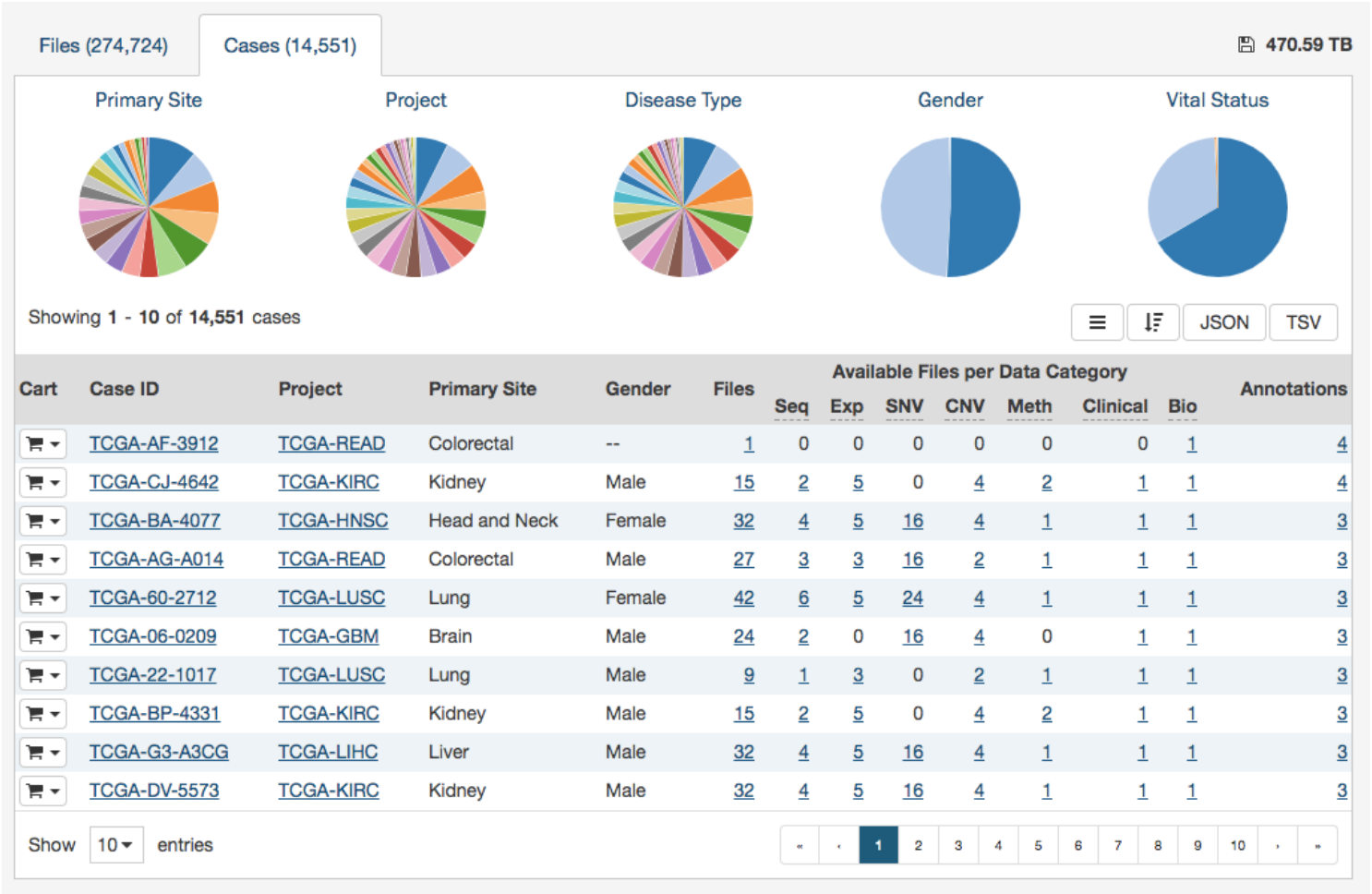
The *File Name* column includes links to file detail pages where the user can learn more about each file.

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:



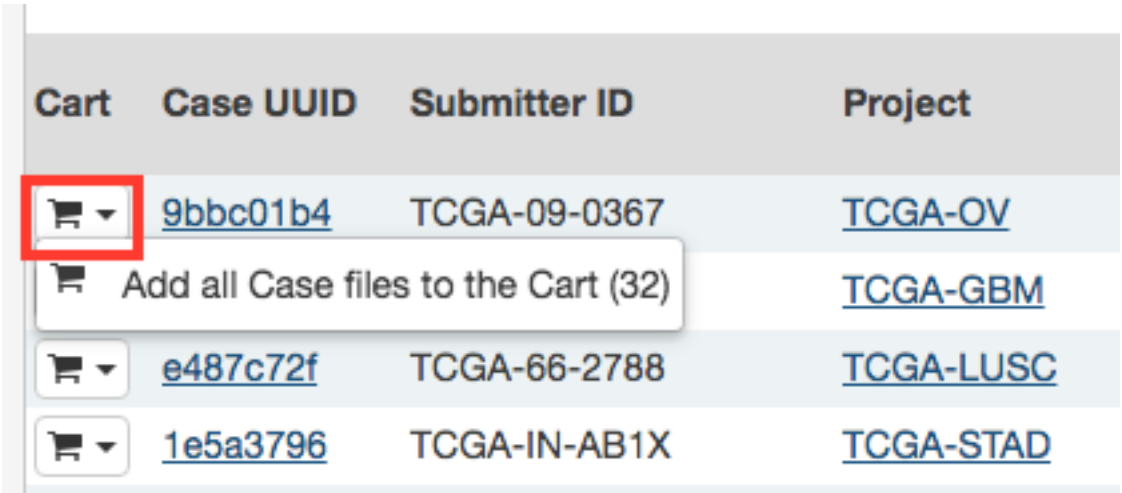
Cases List

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.



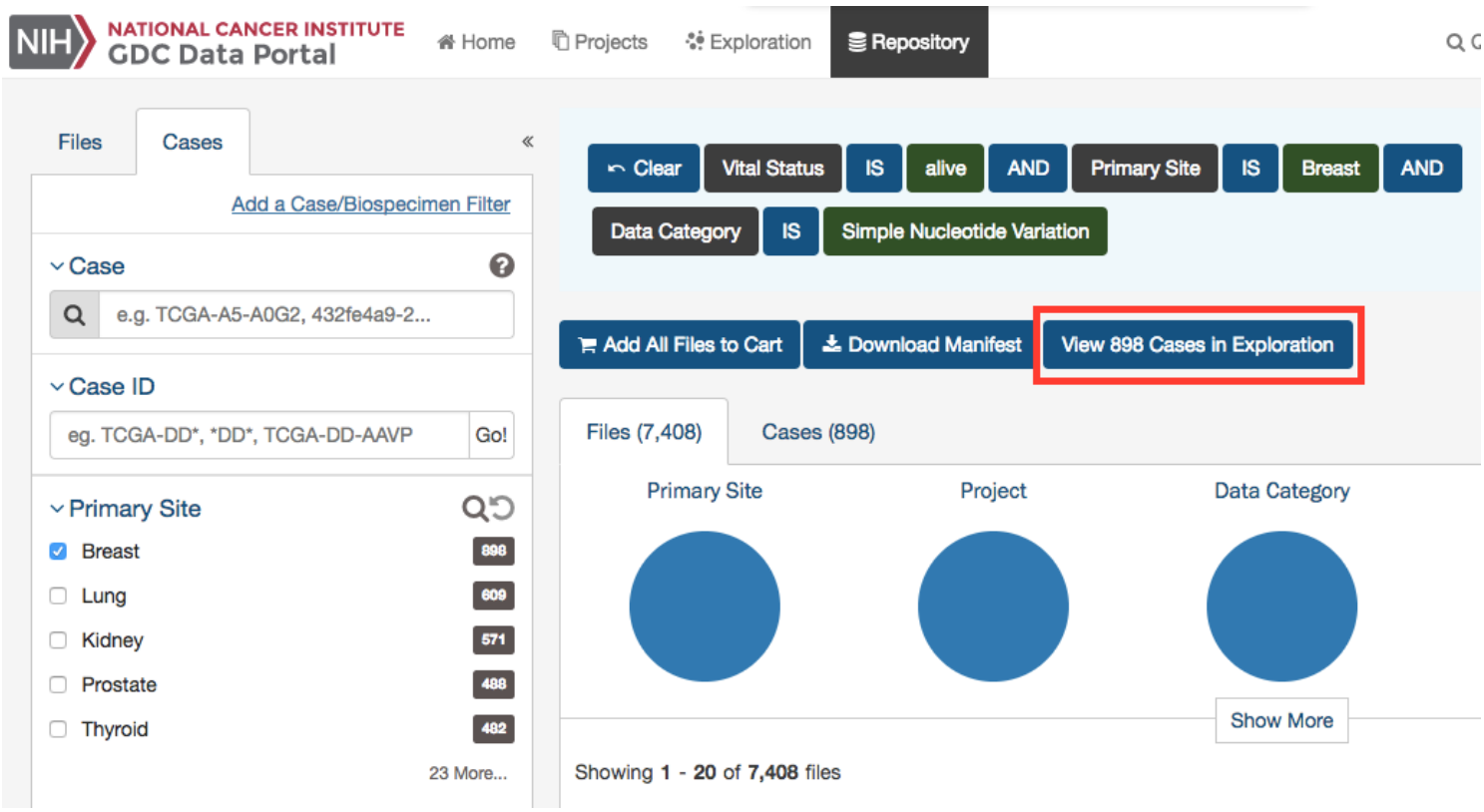
The list includes links to case summary pages in the *Case UUID* column, the Submitter ID (i.e. TCGA Barcode), and counts of the available file types for each case. Clicking on a count will apply facet filters to display the corresponding files.

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:



Navigation

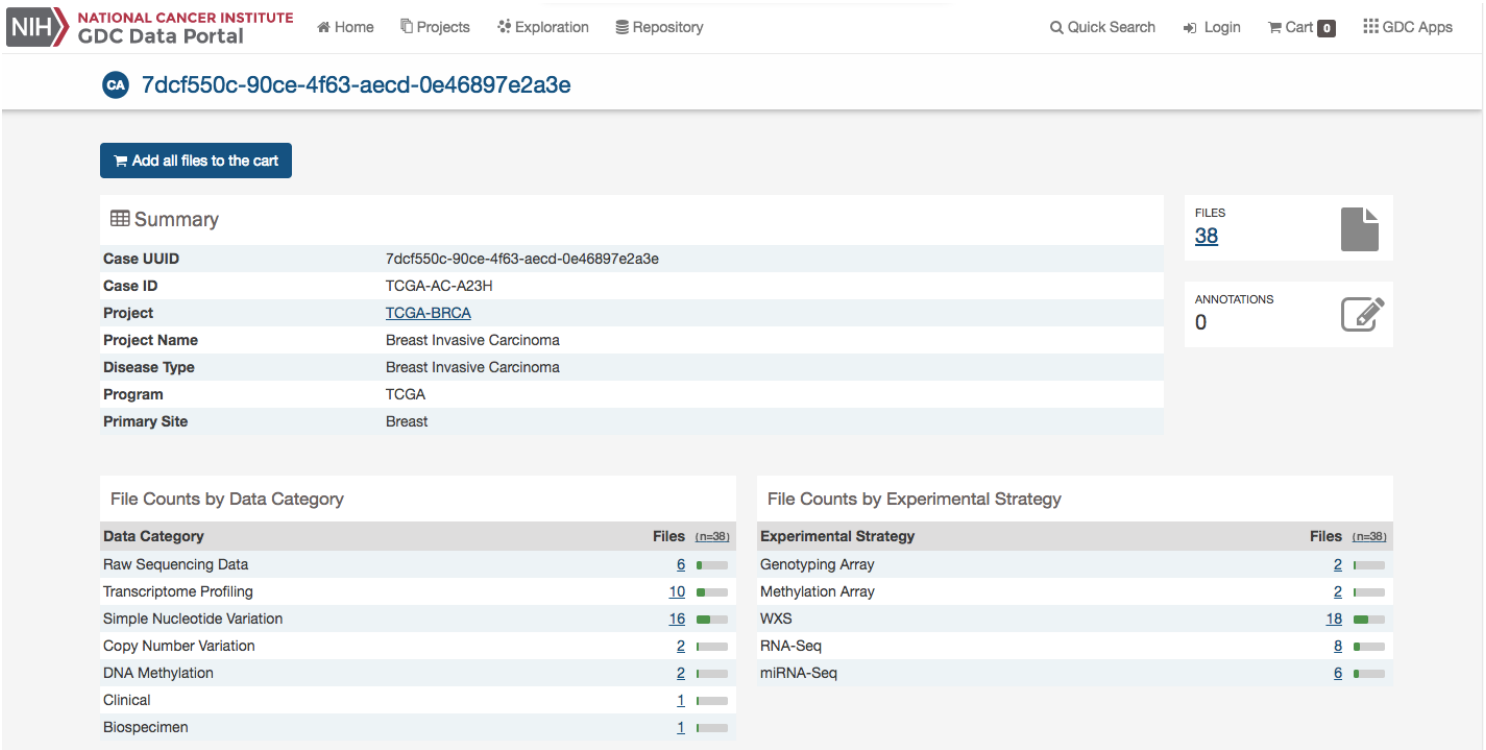
After utilizing the Repository Page to narrow down a specific set of cases, users can continue to explore the mutations and genes affected by these cases by clicking the `View Files in Repository` button as shown in the image below.



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

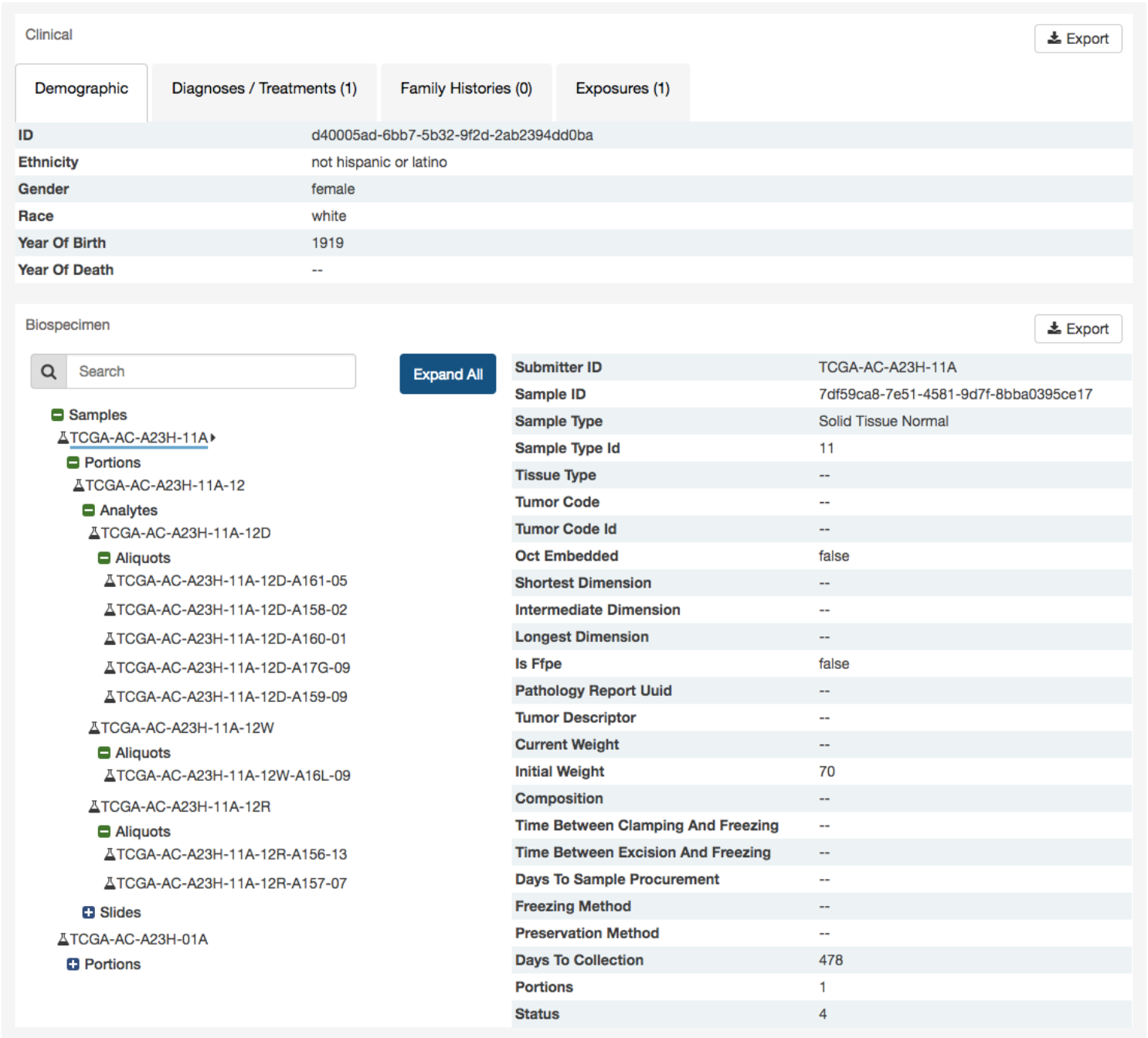
Case Summary Page

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.



Clinical and Biospecimen Information

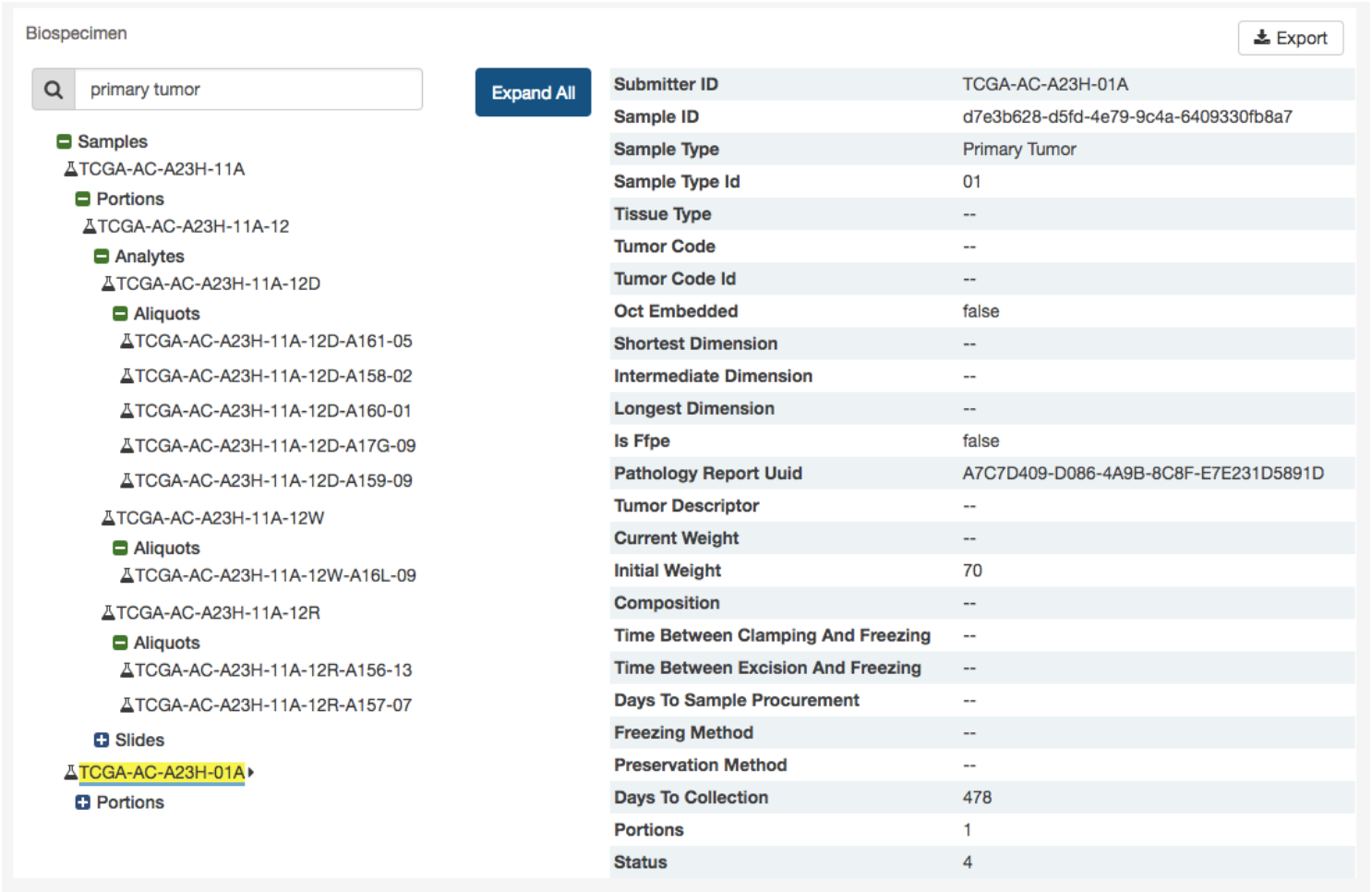
The page also provides clinical and biospecimen information about that case. Links to export clinical and biospecimen information in JSON format are provided.



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.

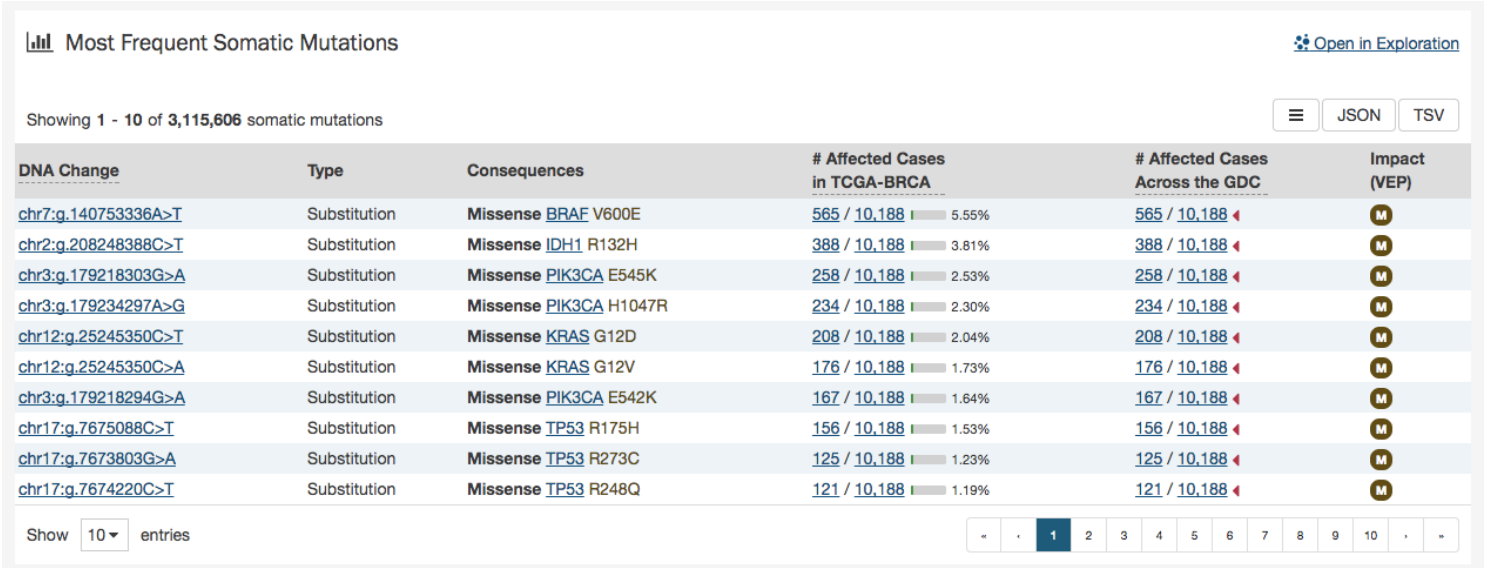
Biospecimen Search

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.



Most Frequent Somatic Mutations

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



The table lists the following information for 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

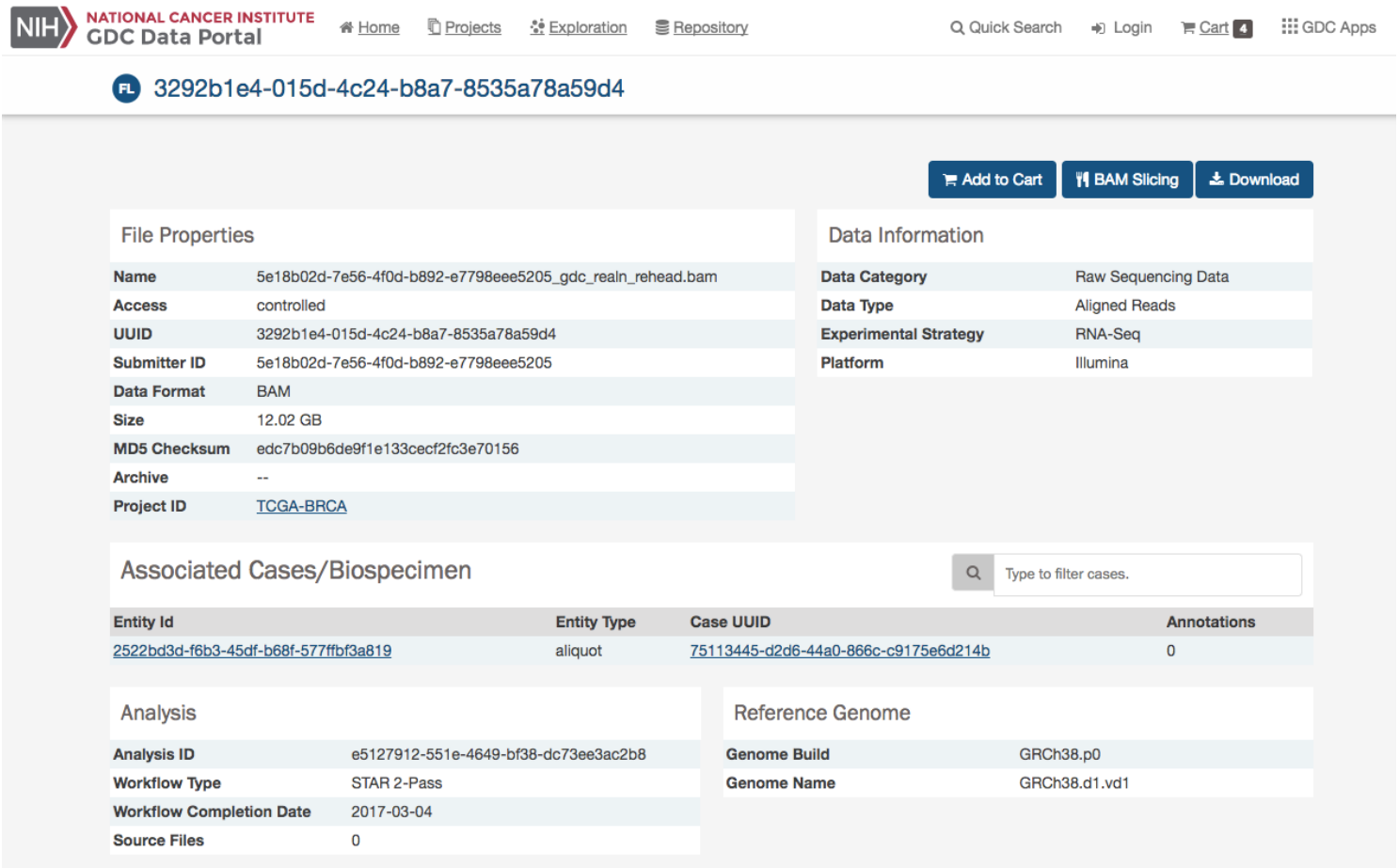
- **Type:** A general classification of the mutation
- **Consequences:** The effects the mutation has on the gene coding for a protein (i.e. synonymous, missense, non-coding transcript)
- ___ Affected Cases in Project:___ The number of affected cases, expressed as number across all mutations within the 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

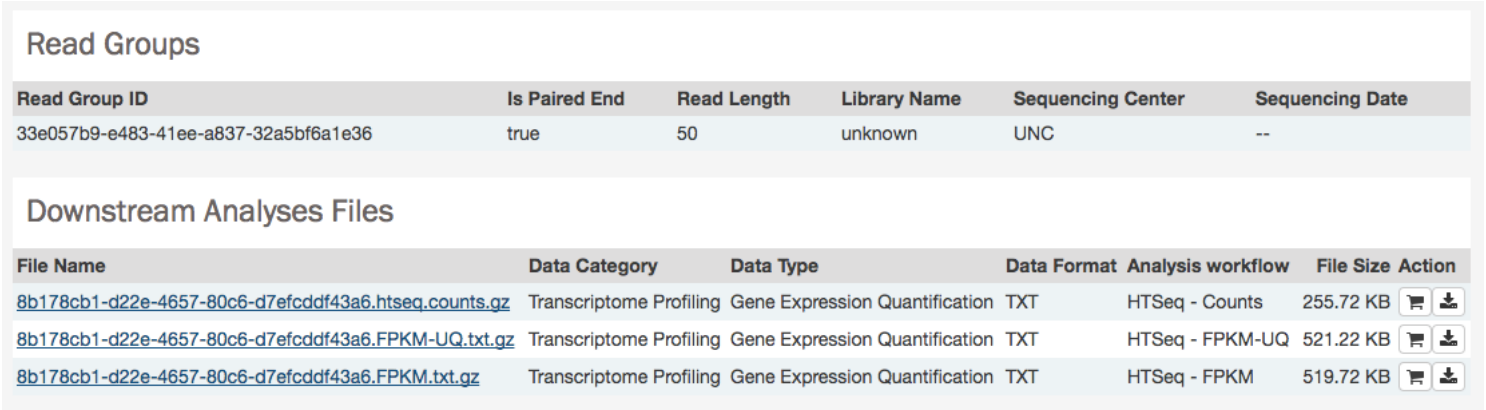
The File Summary page provides information a data file, including file properties like size, md5 checksum, and data format; information on the type of data included; links to the associated case and biospecimen; and information about how the data file was generated or processed.

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



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	--

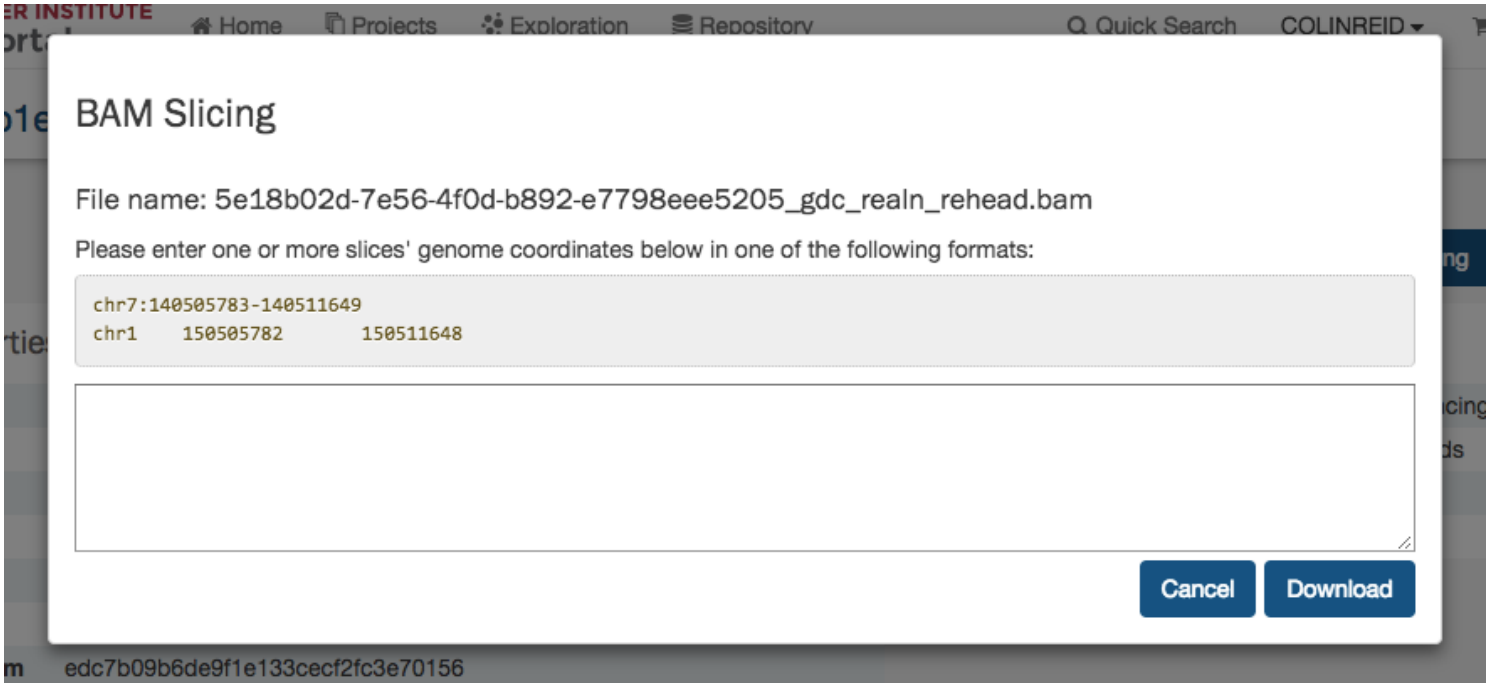
Downstream Analyses Files

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:



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

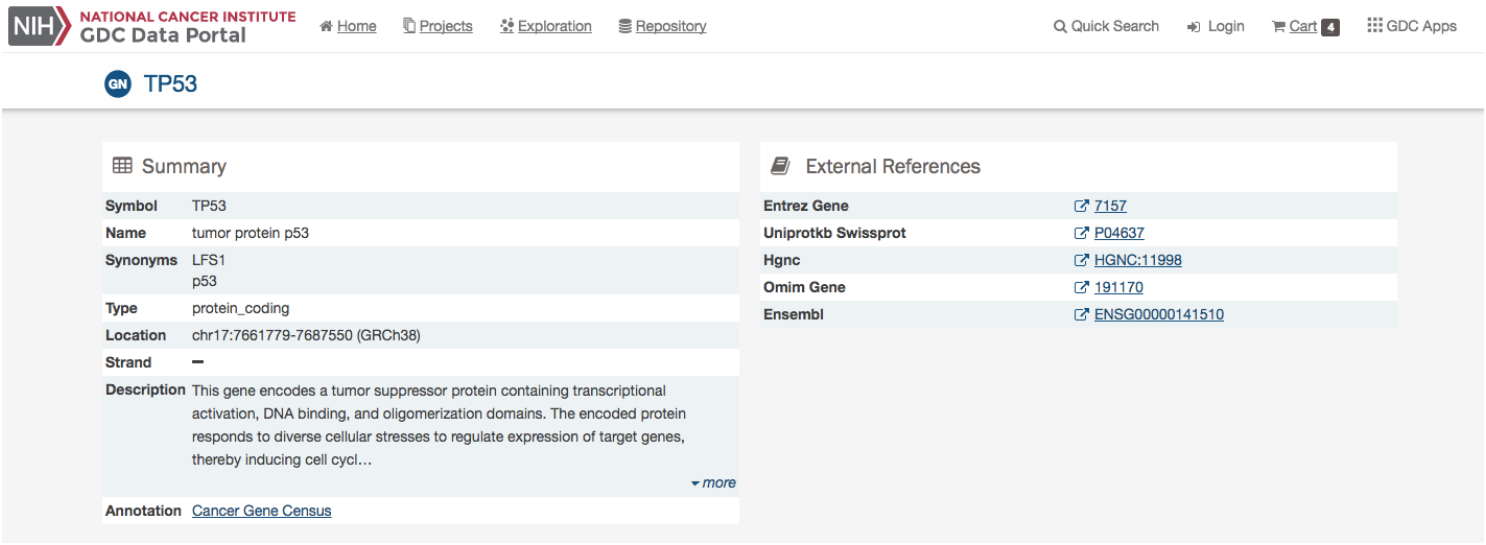
Many parts of the GDC website contain links to Gene and Mutation summary pages. These pages display information about specific genes and mutations, along with visualizations and data showcasing the relationship between themselves and the projects and cases within the GDC. The gene and mutation data that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

Gene Summary Page

The Gene Summary Page describes each gene with mutation data featured at the GDC 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:



- **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

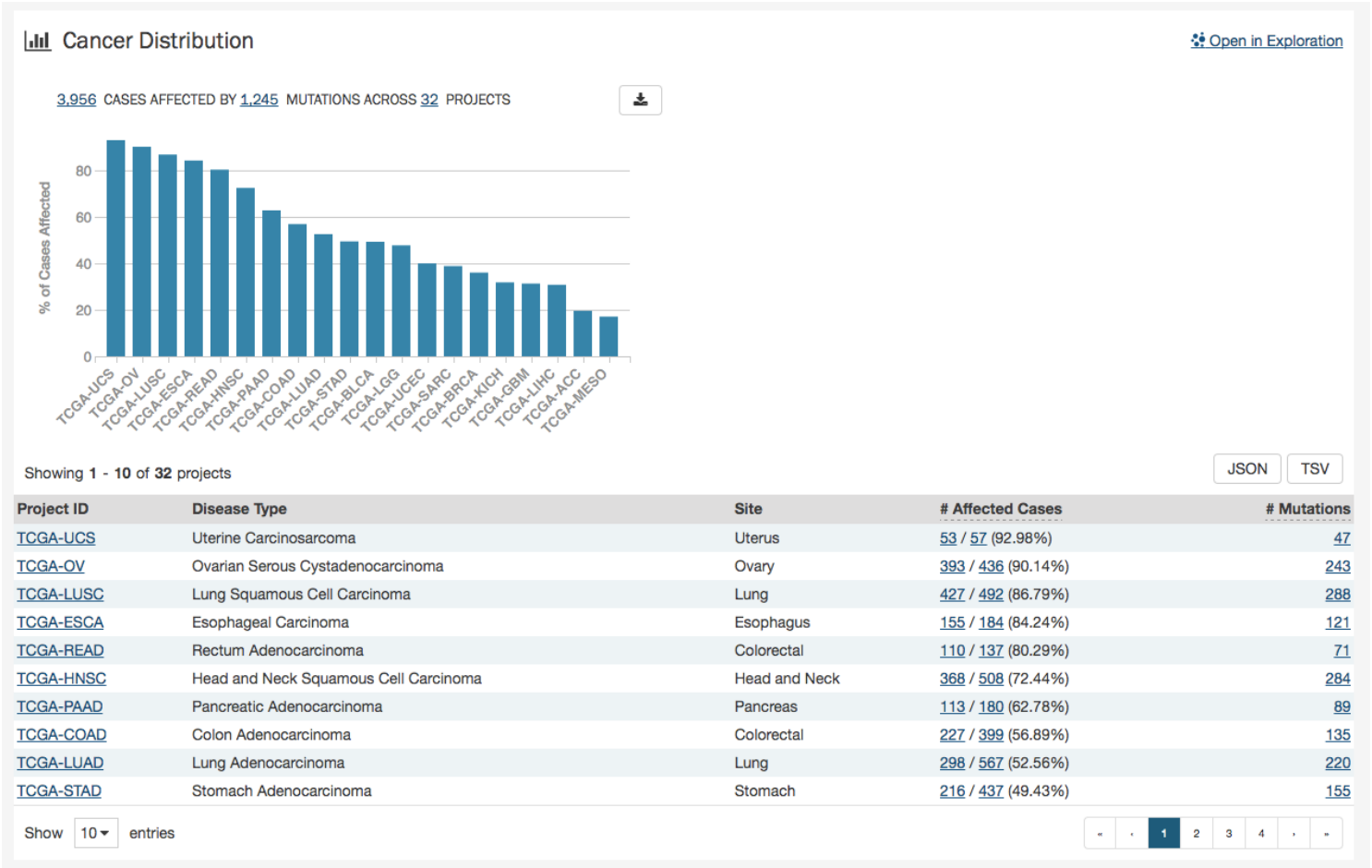
- **Location:** The chromosome on which the gene is located and its coordinates
- **Strand:** If the gene is located on the forward (+) or reverse (-) strand
- **Description:** A description of gene function and downstream consequences of gene alteration
- **Annotation:** A notation/link that states whether the gene is part of The Cancer Gene Census

External References

A list with links that lead to external databases with additional information about each gene is displayed here. These external databases include: Entrez, Uniprot, Hugo Gene Nomenclature Committee, Online Mendelian Inheritance in Man, and Ensembl.

Cancer Distribution

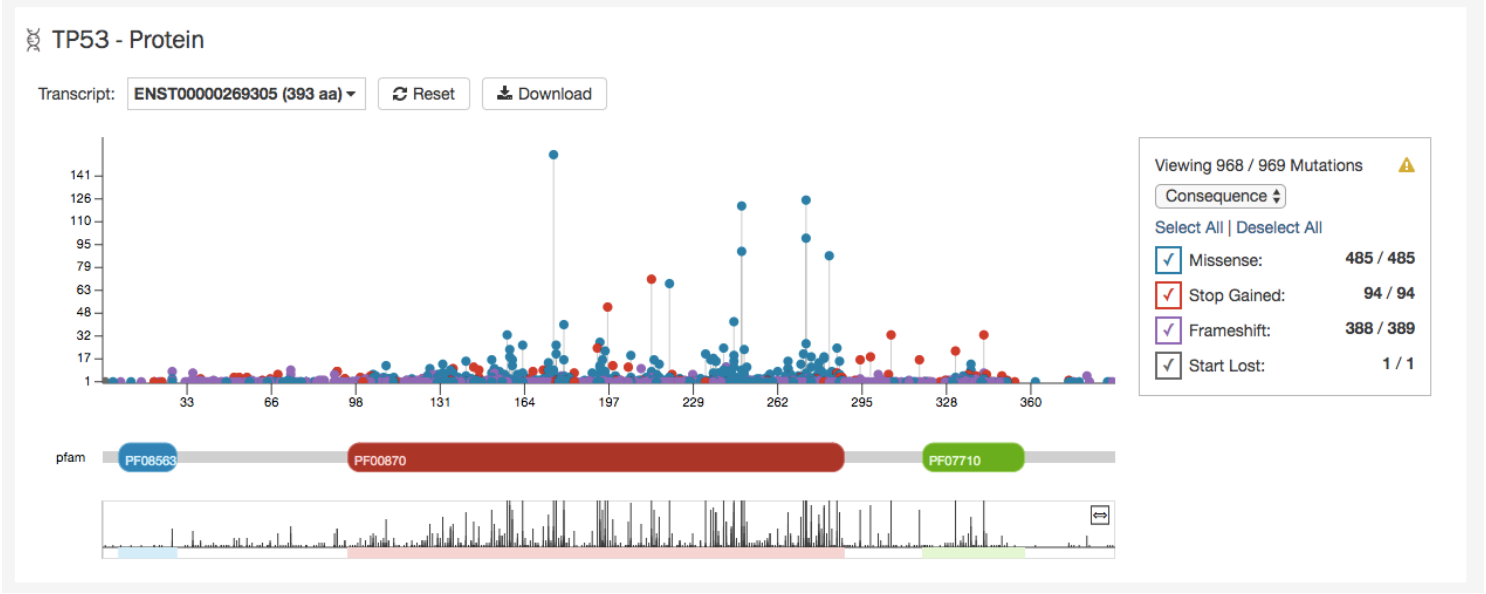
A table and bar graph shows how many cases are affected by mutations within the gene as a ratio and percentage. Each row/bar represents the number of cases for each project. The final column in the table lists the number of unique mutations observed on the gene for each project.



Project ID	Disease Type	Site	# Affected Cases	# Mutations
TCGA-UCS	Uterine Carcinosarcoma	Uterus	53 / 57 (92.98%)	47
TCGA-OV	Ovarian Serous Cystadenocarcinoma	Ovary	393 / 436 (90.14%)	243
TCGA-LUSC	Lung Squamous Cell Carcinoma	Lung	427 / 492 (86.79%)	288
TCGA-ESCA	Esophageal Carcinoma	Esophagus	155 / 184 (84.24%)	121
TCGA-READ	Rectum Adenocarcinoma	Colorectal	110 / 137 (80.29%)	71
TCGA-HNSC	Head and Neck Squamous Cell Carcinoma	Head and Neck	368 / 508 (72.44%)	284
TCGA-PAAD	Pancreatic Adenocarcinoma	Pancreas	113 / 180 (62.78%)	89
TCGA-COAD	Colon Adenocarcinoma	Colorectal	227 / 399 (56.89%)	135
TCGA-LUAD	Lung Adenocarcinoma	Lung	298 / 567 (52.56%)	220
TCGA-STAD	Stomach Adenocarcinoma	Stomach	216 / 437 (49.43%)	155

Show 10 entries

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.

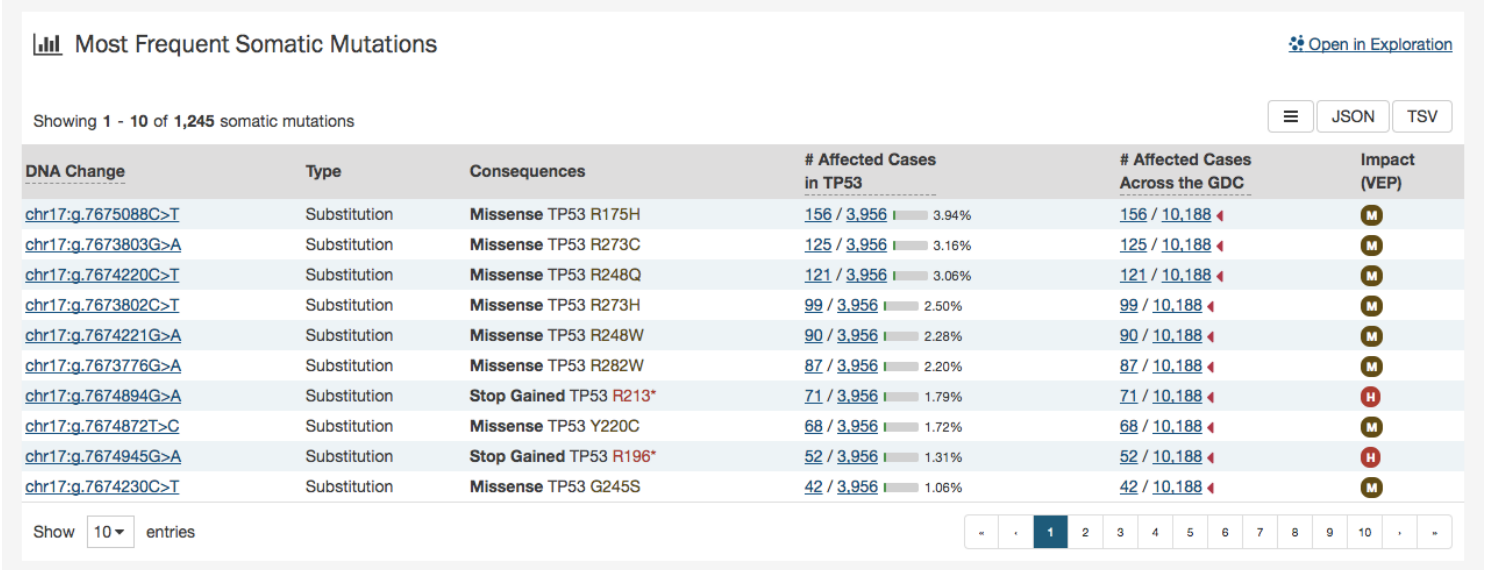
The panel to the right of the plot allows the plot to be filtered by mutation consequences or impact. The plot will dynamically change as filters are applied. Mutation consequence and impact is denoted in the plot by color.

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.



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

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 (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

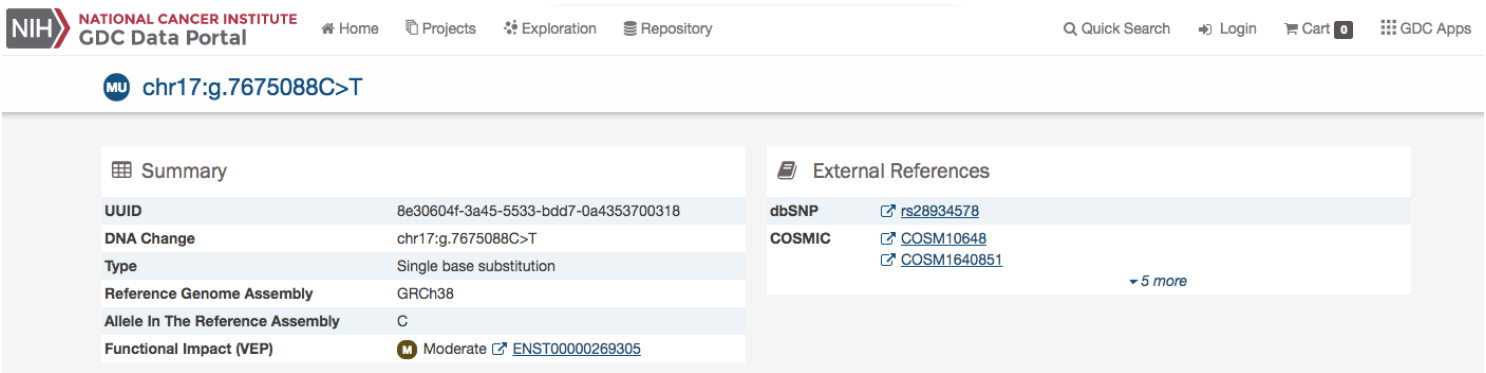
Note: The Mutation UUID can be displayed in this table by selecting it from the drop-down represented by three parallel lines

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



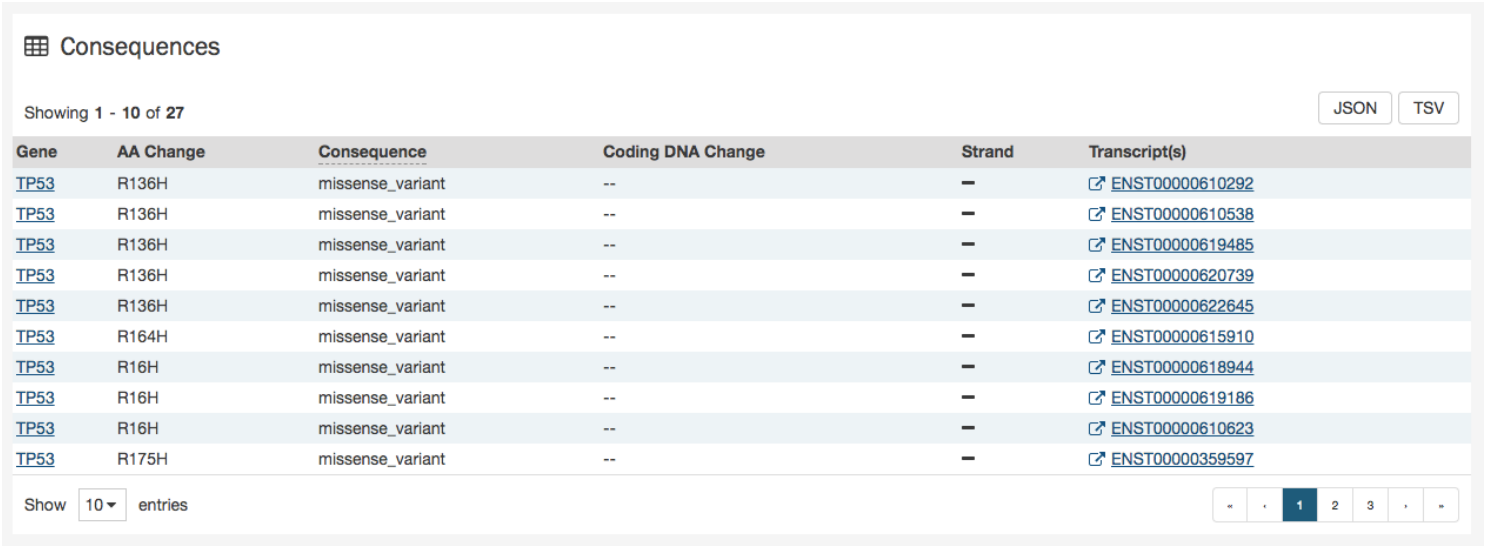
- **ID:** A unique identifier (UUID) for this mutation
- **DNA Change:** Denotes the chromosome number, position, and nucleotide change of the mutation
- **Type:** A broad categorization of the mutation
- **Reference Genome Assembly:** The reference genome in which the chromosomal position refers to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

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.

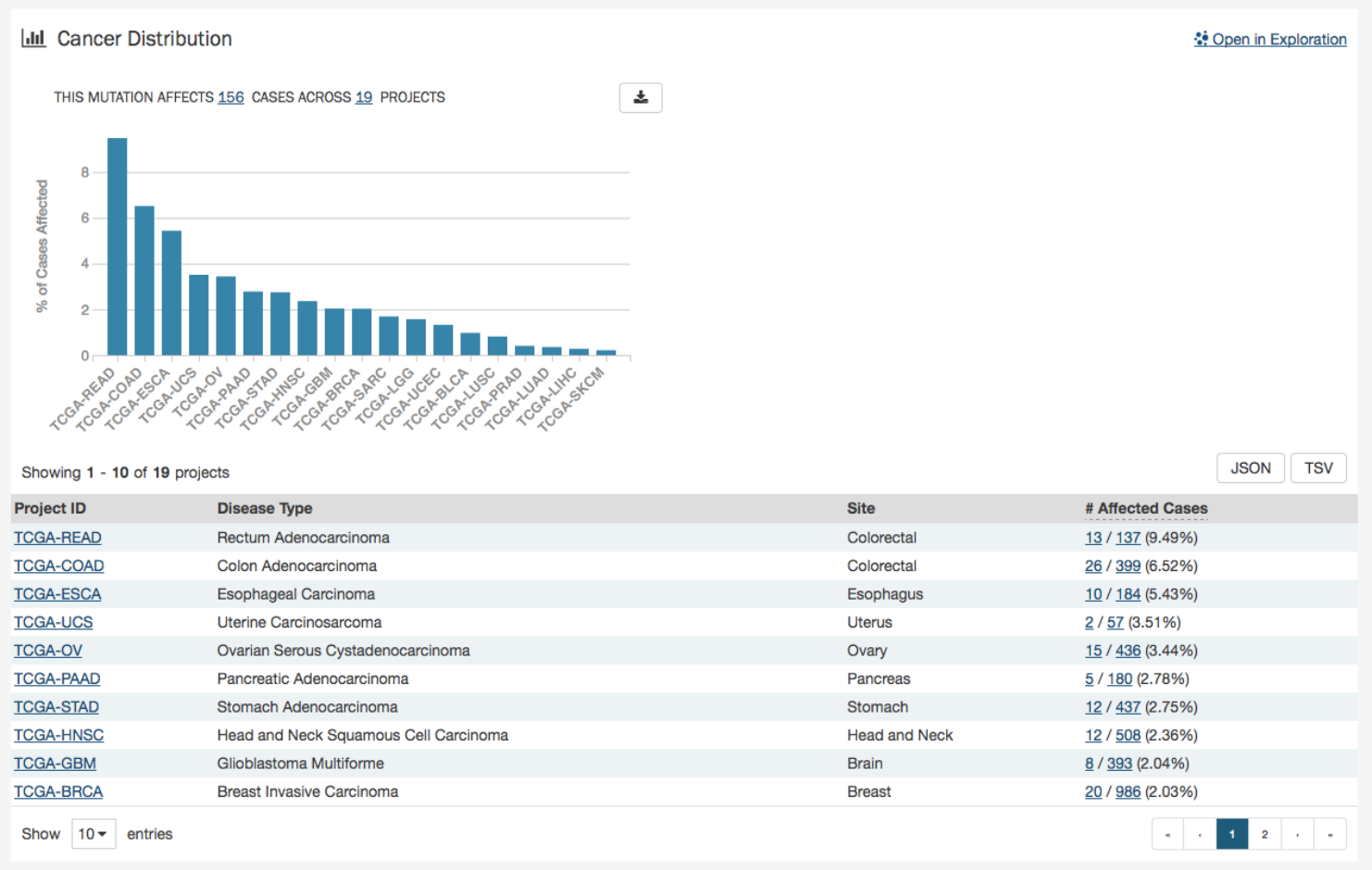


The fields that describe each consequence are listed below:

- **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

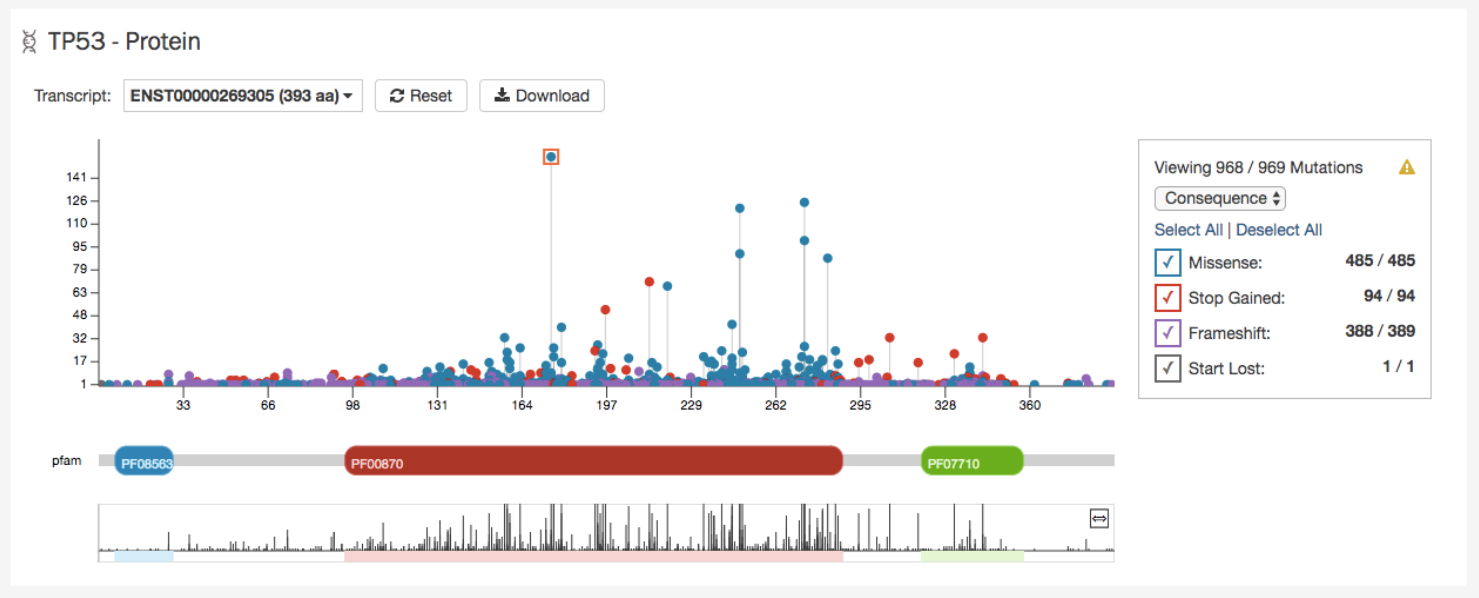
A table and bar graph shows how many cases are affected by the particular mutation. Each row/bar represents the number of cases for each project.



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 associated with the mutation. 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

Annotations

Annotations

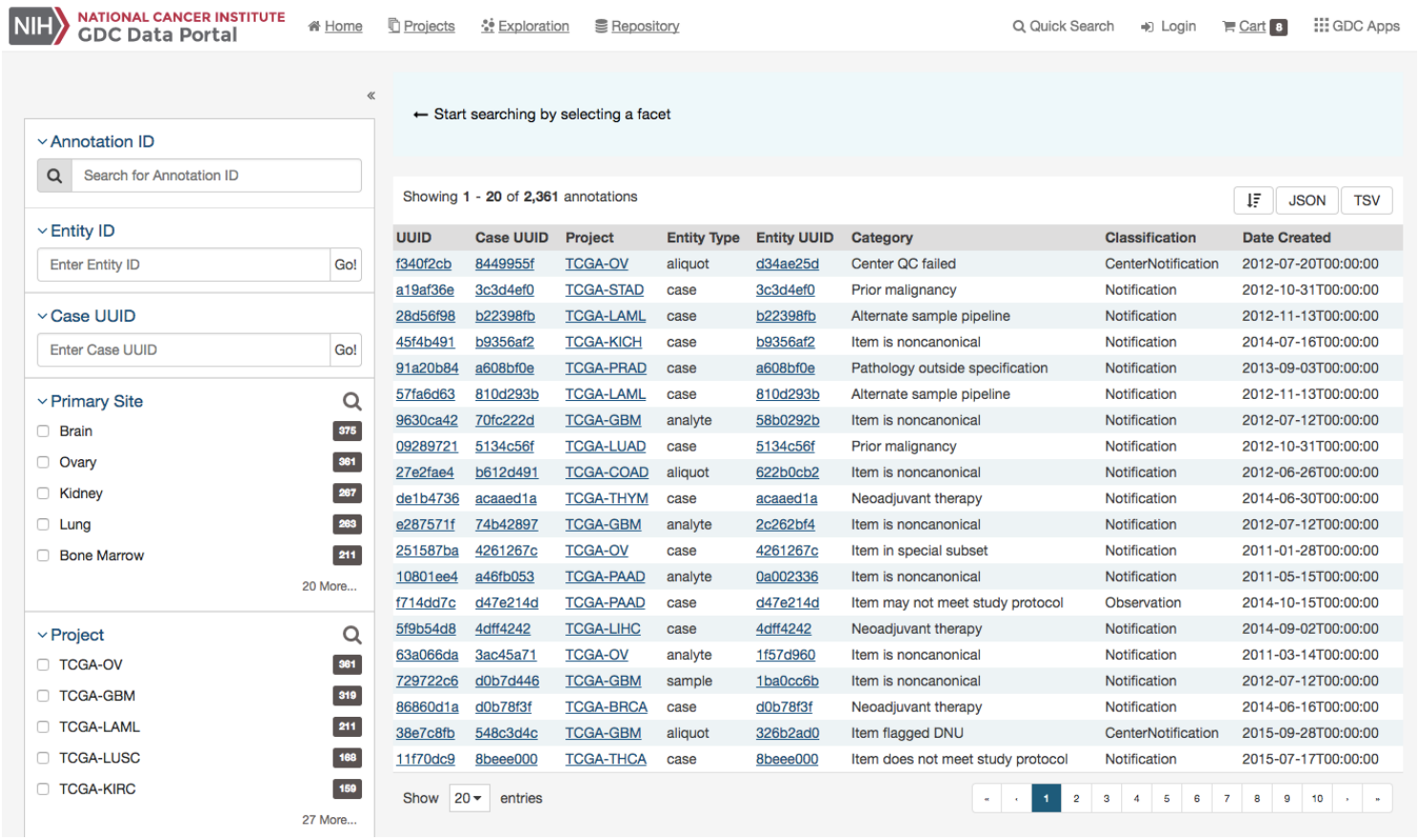
Annotations are notes added to individual cases, samples or files.

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.



Facets Panel

The following facets are available to search for annotations:

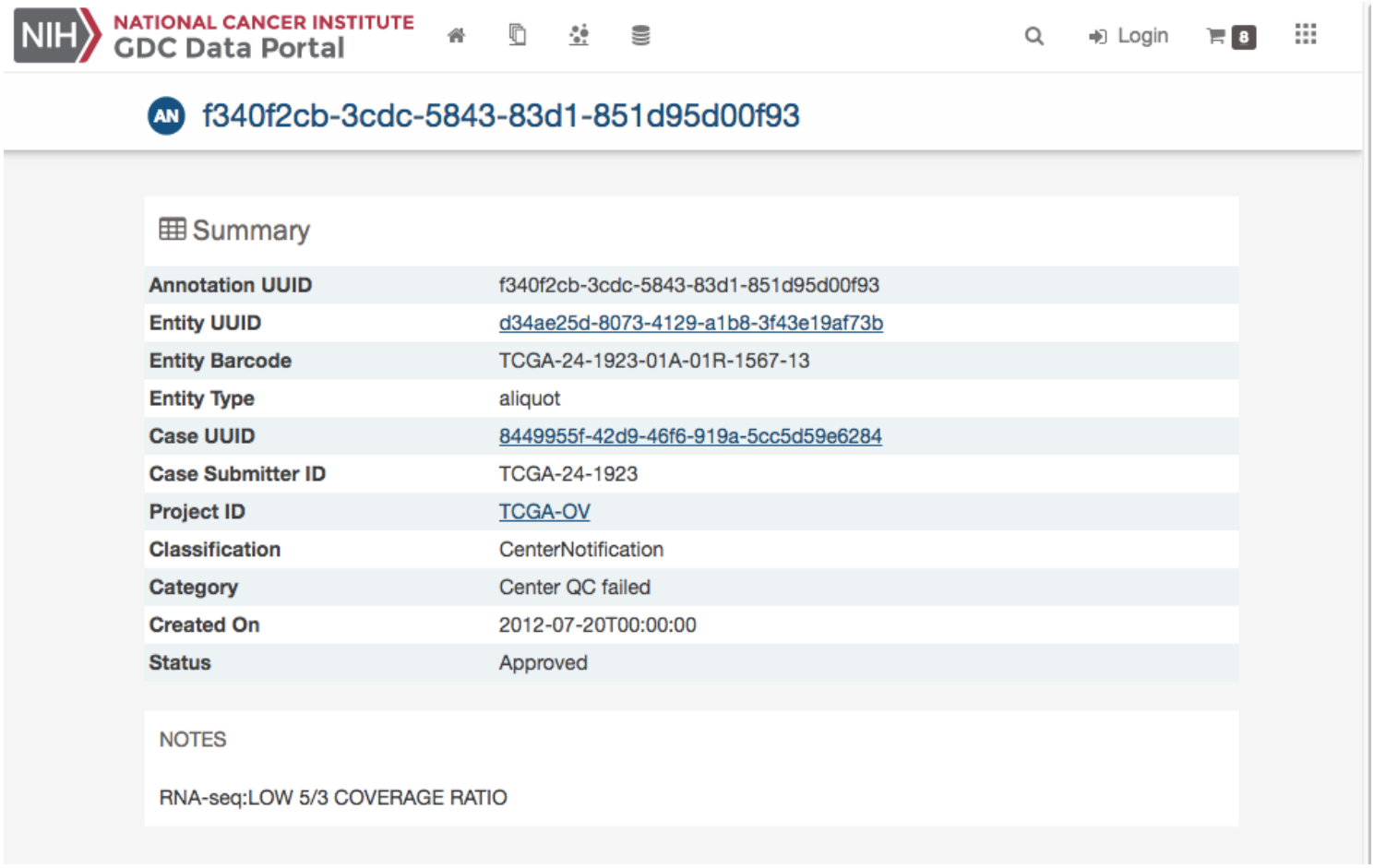
- **Annotation ID**: Seach using annotation ID
- **Entity ID**: Seach using entity ID
- **Case UUID**: Seach using case UUID
- **Primary Site**: Anatomical site of the cancer
- **Project**: A cancer research project, typically part of a larger cancer research program
- **Entity Type**: The type of entity 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.

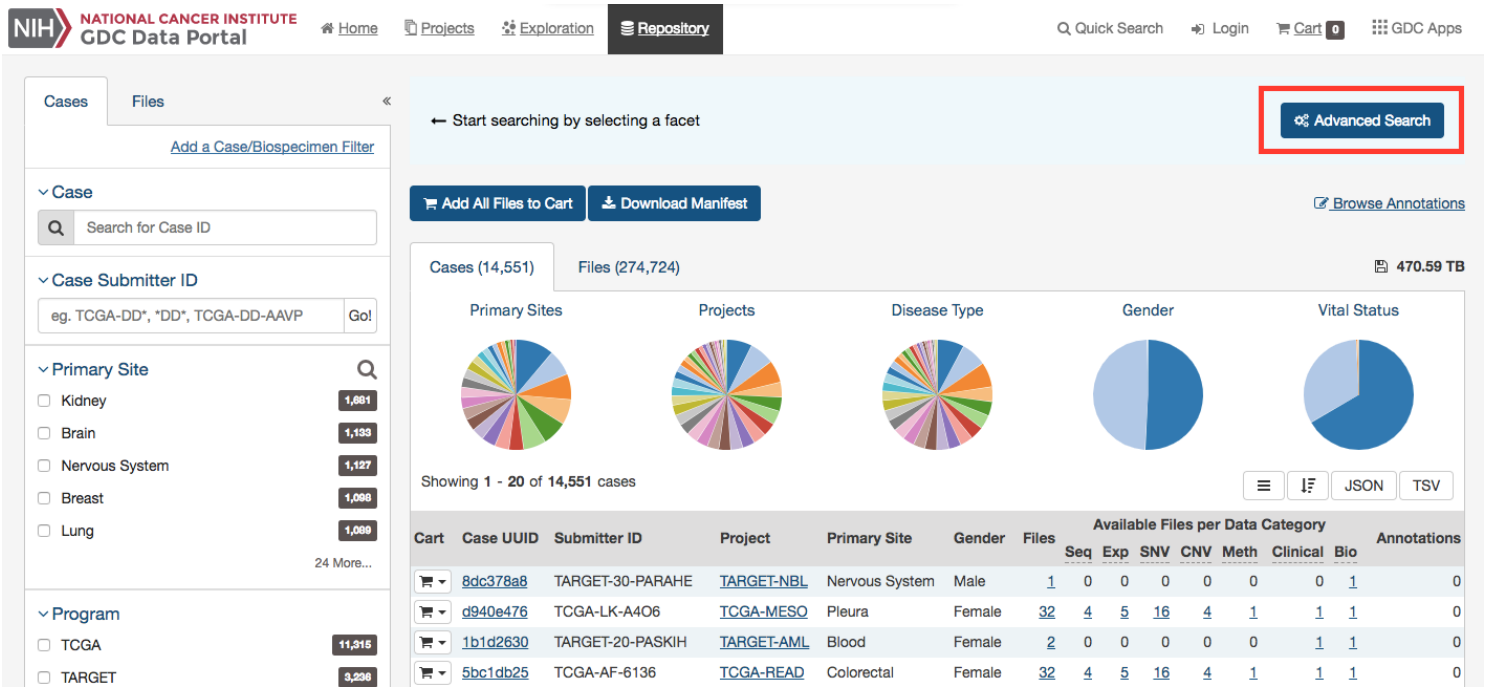

NIH
NATIONAL CANCER INSTITUTE
GDC Data Portal
Login
8
AN
f340f2cb-3cdc-5843-83d1-851d95d00f93
Summary
Annotation UUID
f340f2cb-3cdc-5843-83d1-851d95d00f93
Entity UUID
d34ae25d-8073-4129-a1b8-3f43e19af73b
Entity Barcode
TCGA-24-1923-01A-01R-1567-13
Entity Type
aliquot
Case UUID
8449955f-42d9-46f6-919a-5cc5d59e6284
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 7

Advanced Search

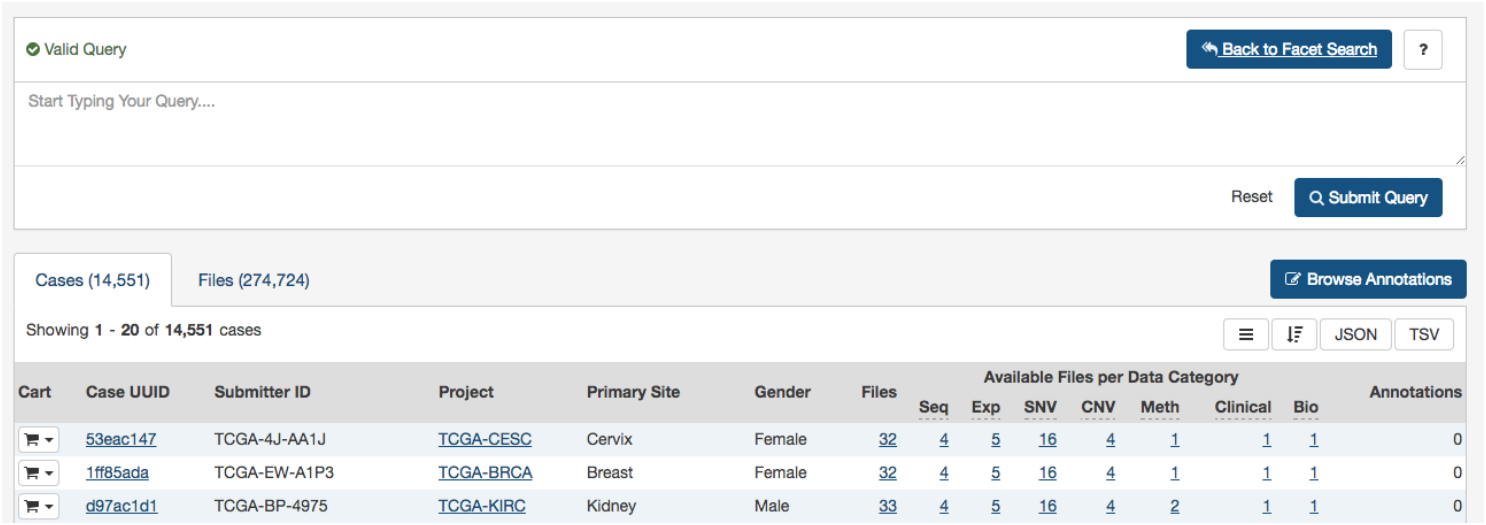
Advanced Search

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

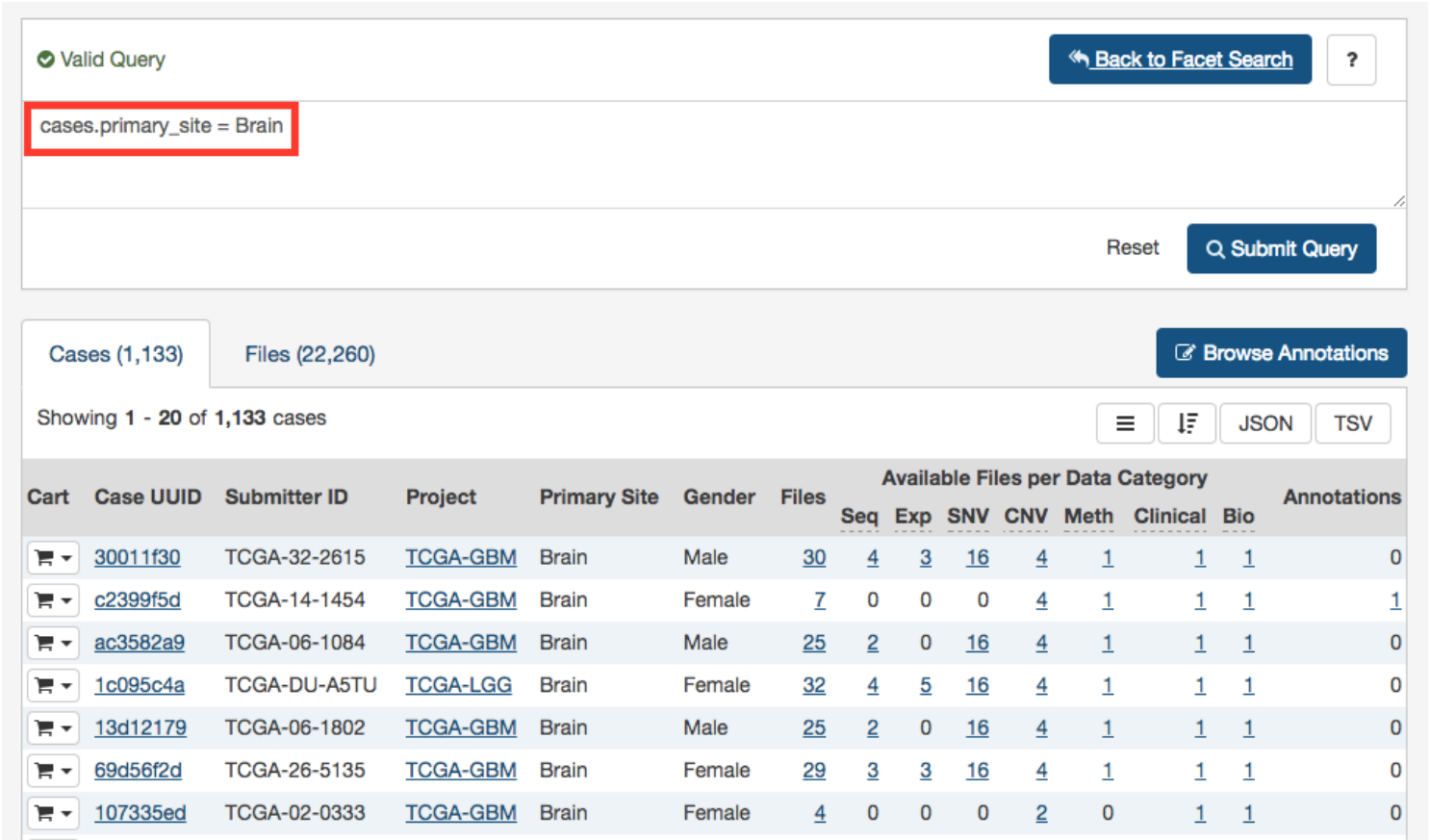


Overview: GQL

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



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:



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".

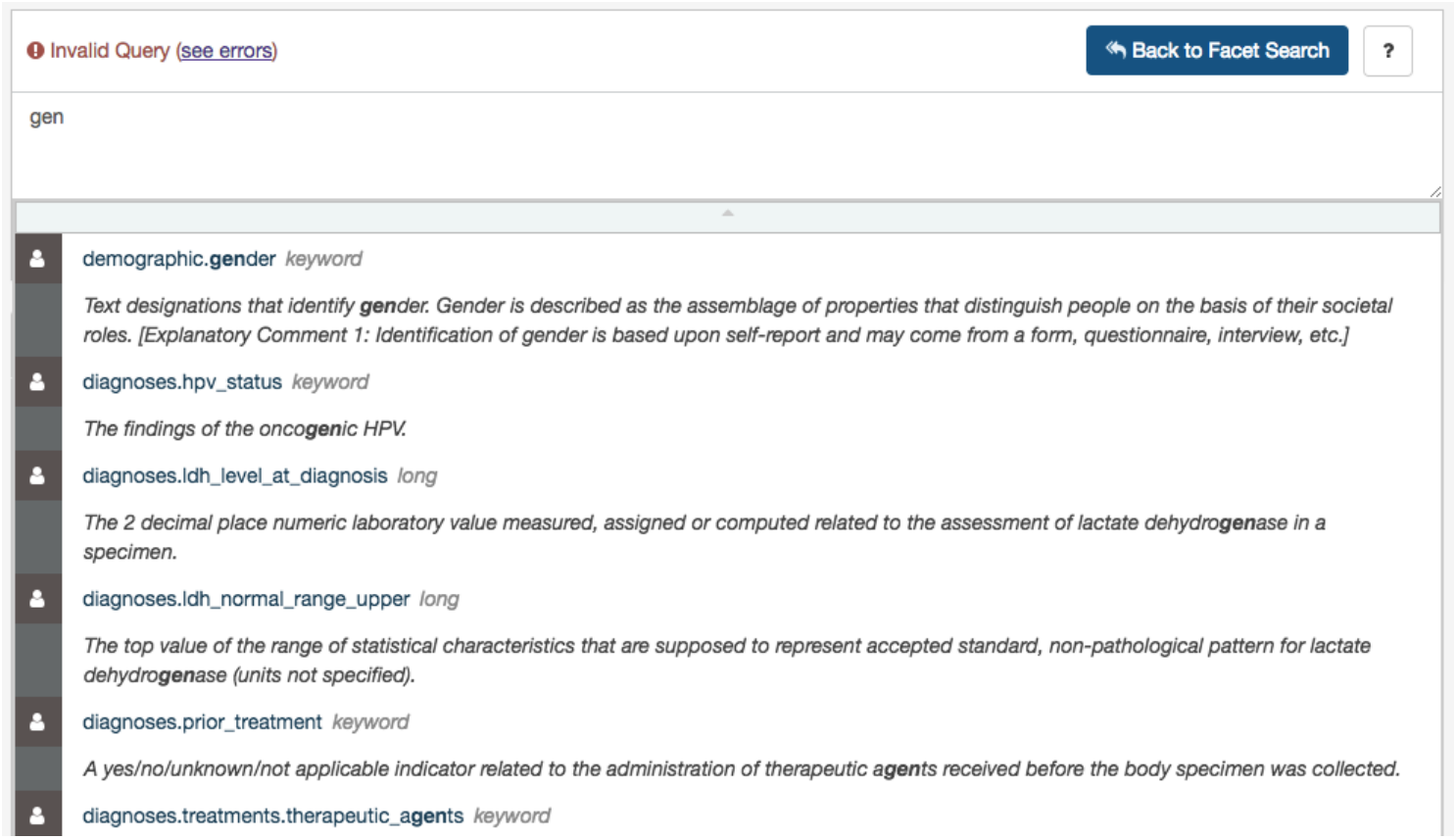
Once the query has been entered and is identified as a "Valid Query", click on "Search" to run your query.

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

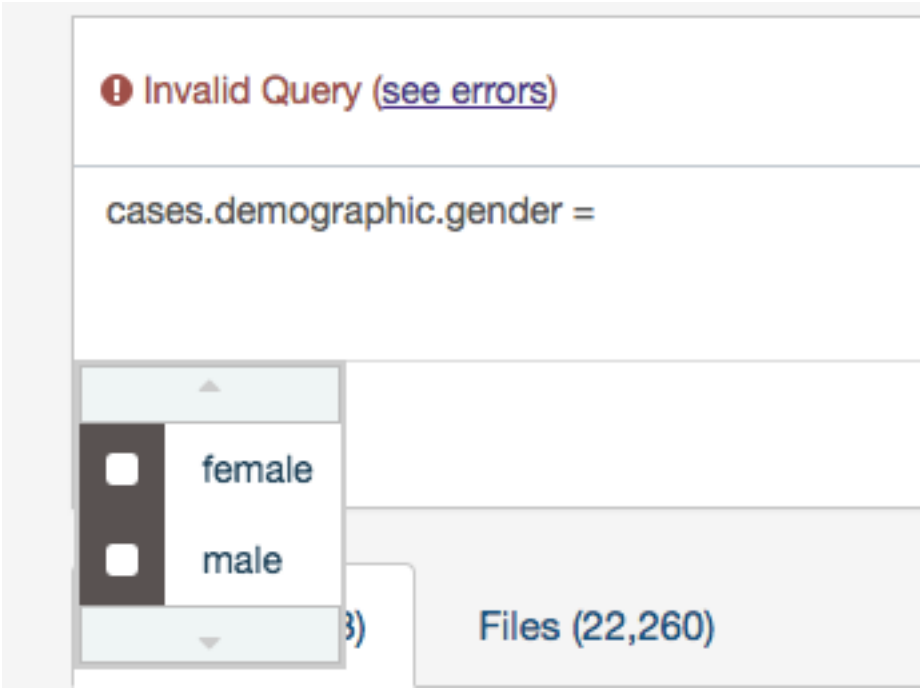
The list of auto-complete suggestions includes **all** available fields matching the user text input. The user has to scroll down to see more fields in the dropdown:



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.:

```
(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 8

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:

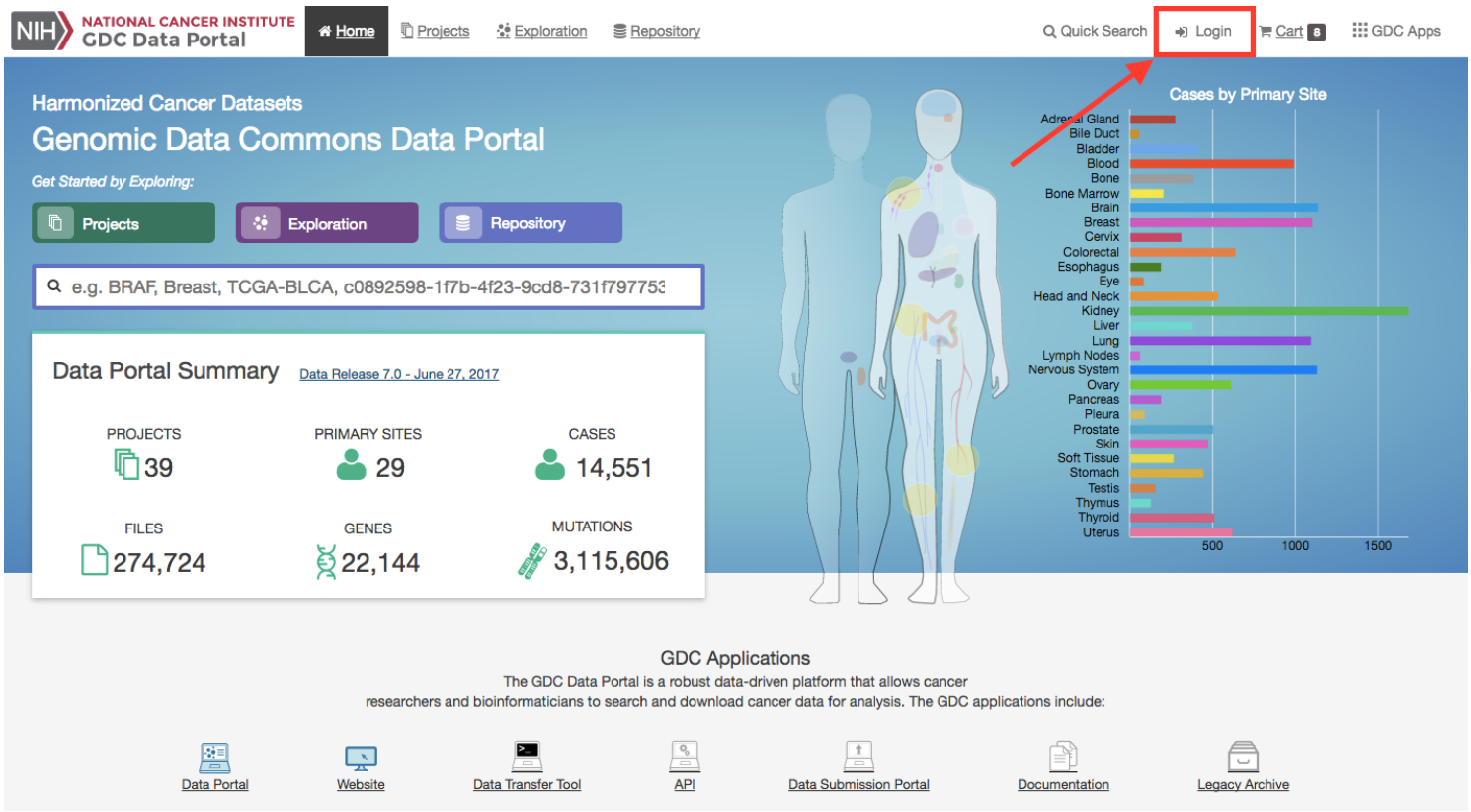
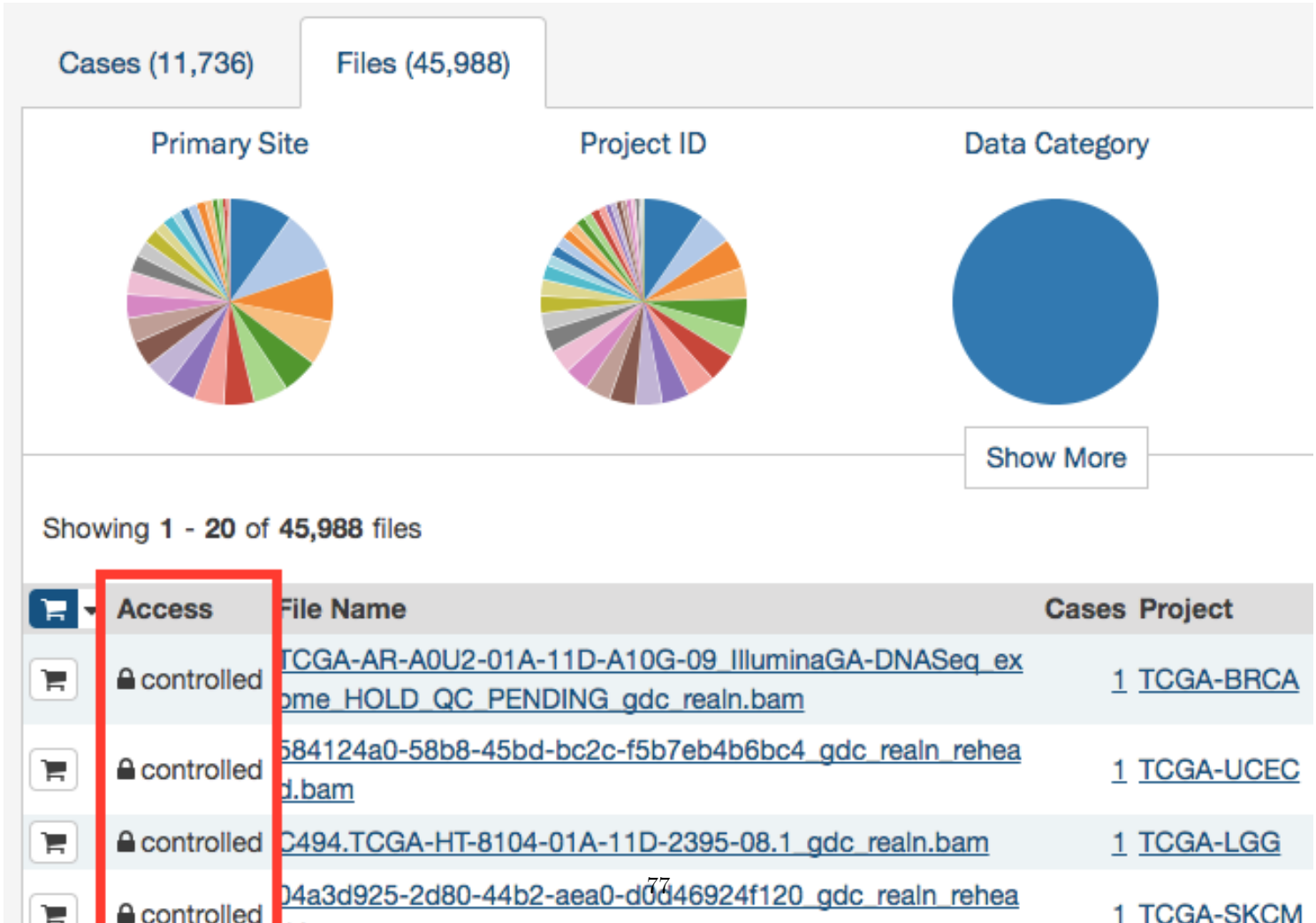


Figure 8.1: Login



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.

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

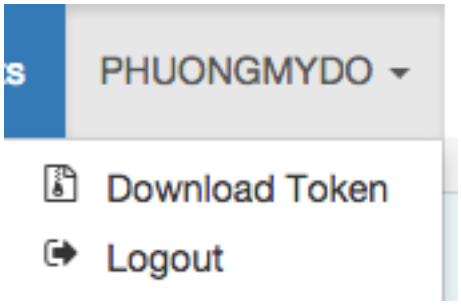


Figure 8.2: Token Download Button

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.

Logging Out

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

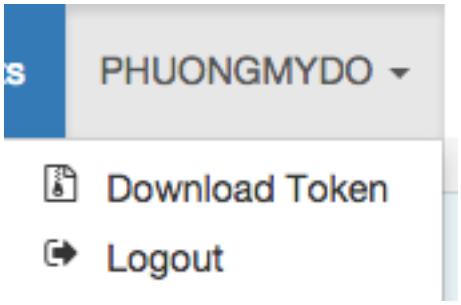


Figure 8.3: Logout link

Chapter 9

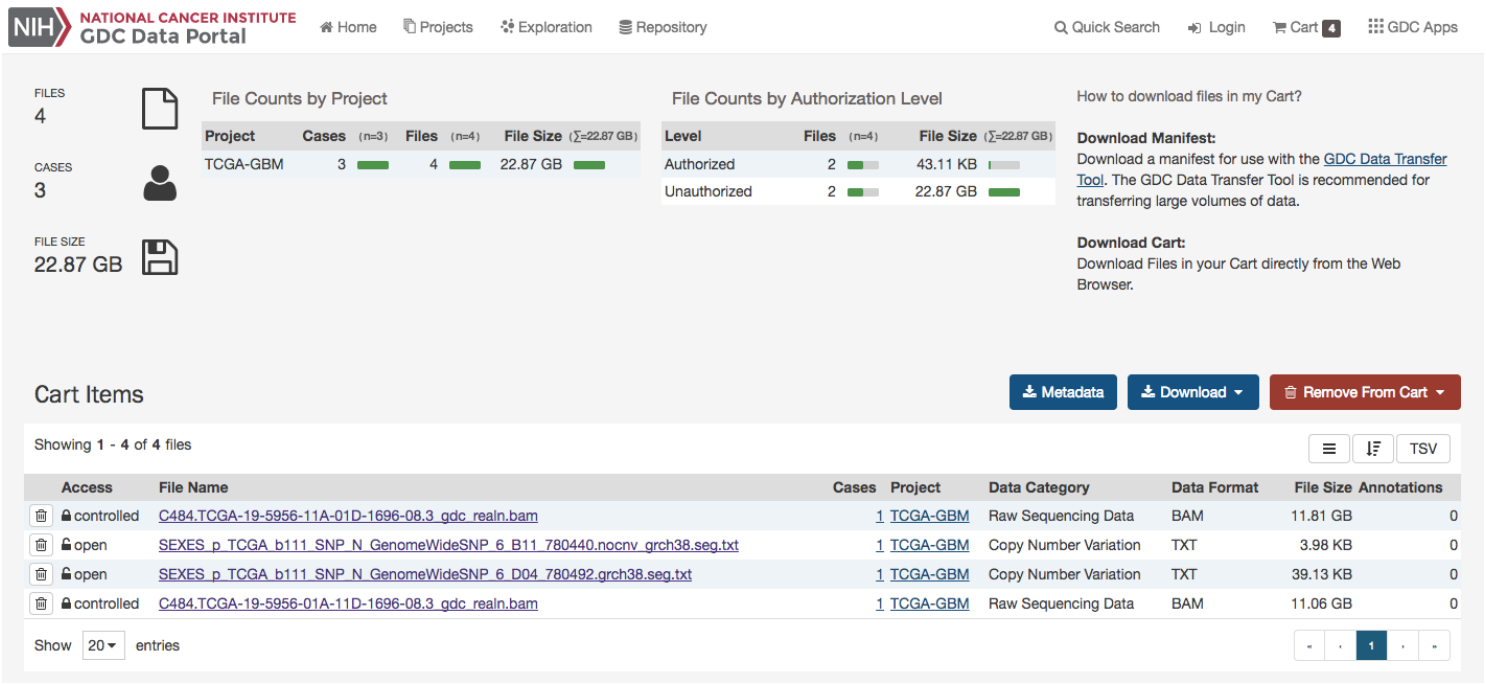
File Cart

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



Cart Summary

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

- Number of files

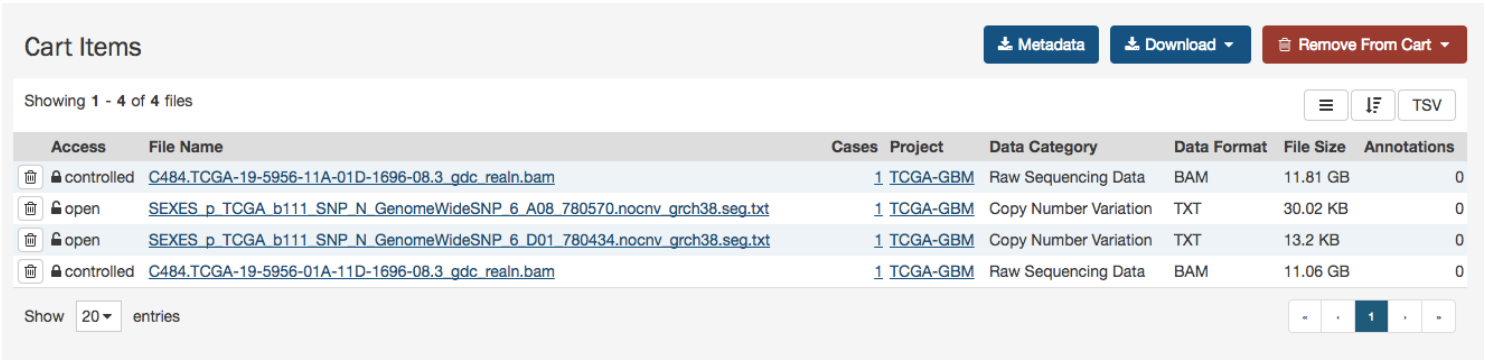
- Number of cases associated with the files
- Total file size

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.

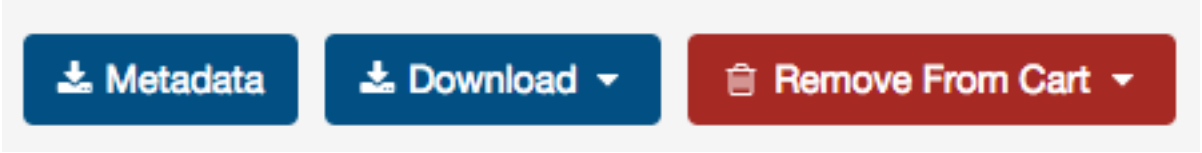
Cart Items



The Cart Items table shows the list of all the files that were added to the Cart. The table gives the folowing 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:

- **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:

```
1 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.

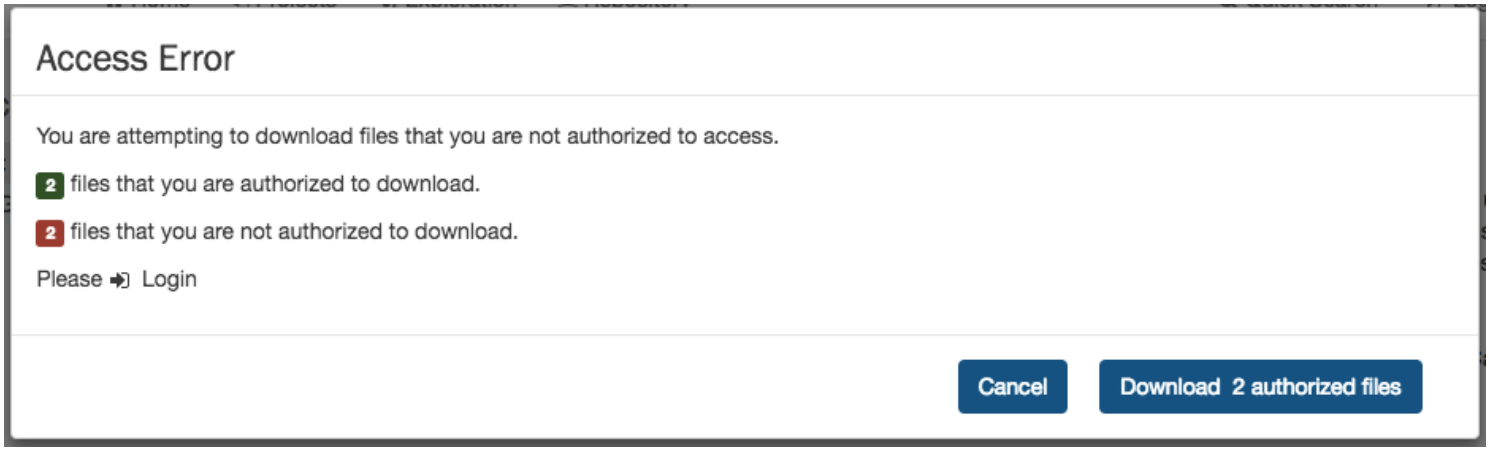
Information on the GDC Data Transfer Tool is available in the GDC Data Transfer Tool User's Guide.

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

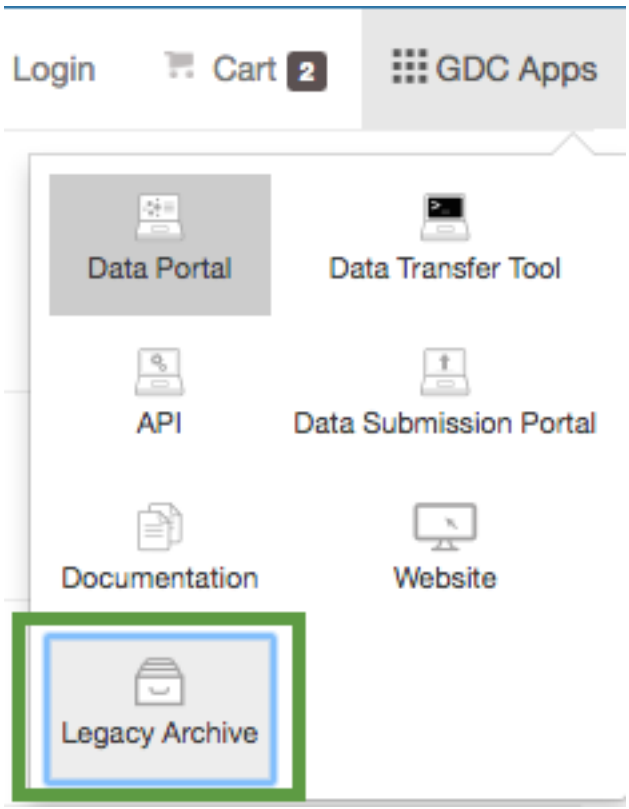
Chapter 10

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.

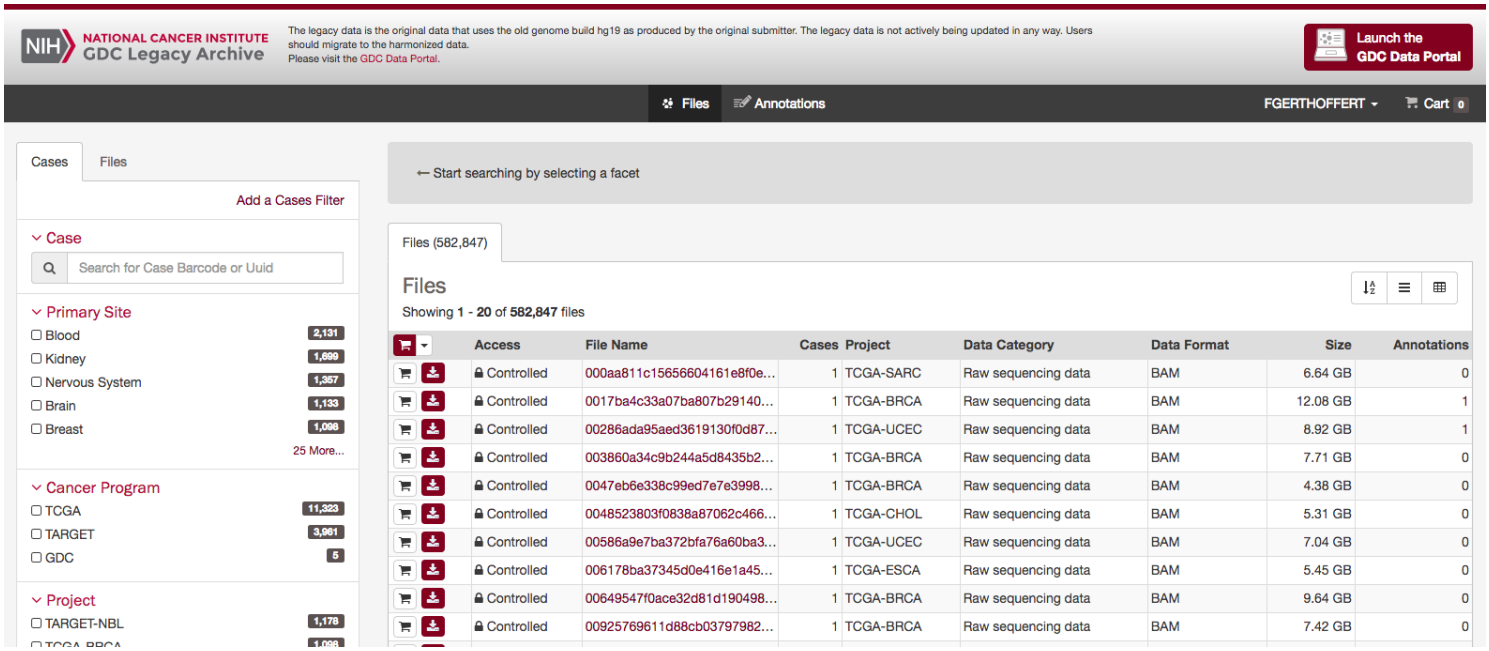


Overview

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

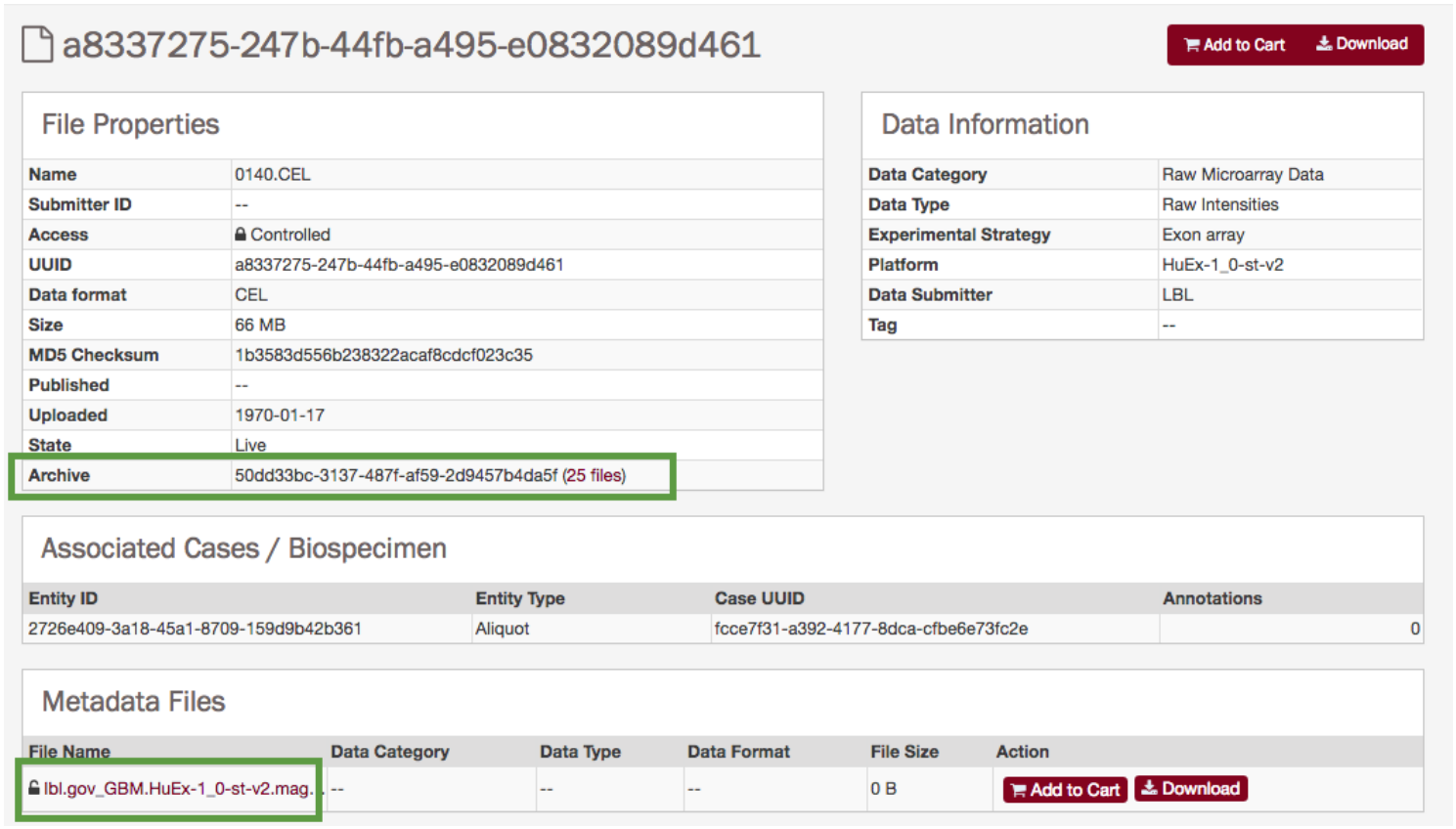
- **Facet search**: Ability to look for legacy files or legacy annotations based on case, file and annotation facets.
- **File and Annotation tables**: List of all the legacy files and list of all the legacy annotations.
- **File and Annotation detail pages**: Information page for each legacy file and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

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



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.



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 11

Release Notes

Data Portal Release Notes

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.fi
```

- 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

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

- 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

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

- 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 looses 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 compatibilty 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 looses 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 compatibilty 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 clic 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.

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