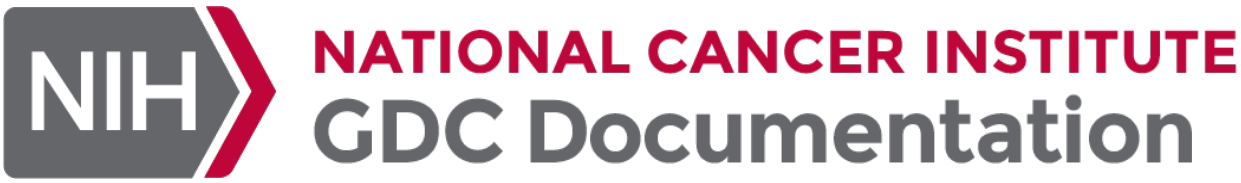


GDC Data User's Guide

NCI Genomic Data Commons (GDC)

NCI GDC

Table of contents

1. GDC Data

1.1 Introduction

The Genomic Data Commons receives, processes, and distributes genomic, clinical, and biospecimen data from cancer research programs. General information about data in the GDC can be found on the GDC website.

This document provides details about data included in the Genomic Data Commons, including information about the GDC data model, data formats, data processing, data security, and data releases.

1.2 GDC Data Model

1.2.1 Introduction

The GDC Data Model is the central method of organization of all data artifacts in the GDC. An overview of the data model, including a visual representation of its components, is provided on the GDC website. This section provides technical details about its implementation for data users, submitters, and developers.

1.2.2 Entities, Properties, and Links

Although the GDC Data Model may contain some cyclic elements, it can be helpful to think of it as a Directed Acyclic Graph (DAG) composed of interconnected **entities**. Each entity in the GDC has a set of properties and links.

- **Properties** are key-value pairs associated with an entity. Properties cannot be nested, which means that the value must be numerical, boolean, or a string, and cannot be another key-value set. Properties can be either required or optional. The following properties are of particular importance in constructing the GDC Data Model:
- **Type** is a required property for all entities. Entity types include `project`, `case`, `demographic`, `sample`, `read_group` and others.
- **System properties** are properties used in GDC system operation and maintenance. They cannot be modified except under special circumstances.
- **Unique keys** are properties, or combinations of properties, that can be used to uniquely identify the entity in the GDC. For example, the tuple (combination) of `[ project_id, submitter_id ]` is a unique key for most entities, which means that although `submitter_id` does not need to be unique in GDC, it must be unique within a project. See GDC Identifiers below for details.
- **Links** define relationships between entities, and the multiplicity of those relationships (e.g. one-to-one, one-to-many, many-to-many).

The GDC Data Dictionary determines which properties and links an entity can have according to entity `type`.

Functionally similar entity types are grouped under the same **category**. For example, entity types `slide_image` and `submitted_unaligned_reads` belong to `data_file` category, which comprises entities that represent downloadable files.

1.2.3 GDC Identifiers

UUIDs

When an entity is created, it is assigned a unique identifier in the form of a version 4 universally unique identifier (UUID). The UUID uniquely identifies the entity in the GDC, and is stored in the entity's `id` property.

Program Name, Project Code, and Project ID

Programs are the highest level of organization of GDC datasets. Each program is assigned a unique `program.name` property. Datasets within a program are organized into projects, and each project is assigned a `project.code` property.

The `project_id` property is associated with most entities in the GDC data model and is generated by appending `project.code` to `program.name` as follows:

```
program.name-project.code
(e.g. TCGA-LAML)
```

Note that `program.name` never contains hyphens.

Submitter ID

In addition to UUIDs stored in the `id` property, many entities also have a `submitter_id` property. This property can contain any string that the submitter wishes to use to identify the entity (e.g. a "barcode"). This can be used to identify a corresponding entry in the submitter's records. The GDC requires that `submitter_id` be unique for each entity within a project: the tuple (combination) of `[ project_id, submitter_id ]` is a unique key.

Note: The `submitter_id` of a `case` entity corresponds to the `submitted_subject_id` of the study participant in dbGaP records for the project.

1.2.4 Working with the GDC Data Model

Data Users

Users can access information stored in the GDC Data Model using the GDC Data Portal, the GDC API, and the GDC Data Transfer Tool. For more information see Data Access Processes and Tools.

Data Submitters

Data submitters can create and update submittable entities in the GDC Data Model and upload data files registered in the model using the GDC Data Submission Portal, the GDC API, and the GDC Data Transfer Tool. For more information see Data Submission Processes and Tools.

1.3 Data Security

To protect the privacy of research participants and support data integrity, the GDC requires user authorization and authentication for:

- downloading controlled-access data
- submitting data to the GDC

To perform these functions, GDC users must first obtain appropriate authorization via dbGaP and then authenticate via eRA Commons. The GDC sets user permissions at the project level according to dbGaP authorizations.

See Data Access Processes and Tools to learn more about the difference between open-access and controlled-access data.

1.3.1 Authorization via dbGaP

Instructions for obtaining authorization via dbGaP are provided in Obtaining Access to Controlled Data and Obtaining Access to Submit Data.

1.3.2 Authentication via eRA Commons

The following authentication methods are supported by the GDC:

GDC Tool	Authentication Method
GDC Data Portal	Log in using eRA Commons account
GDC Data Submission Portal	Log in using eRA Commons account
GDC Data Transfer Tool	Authentication Token
GDC API	Authentication Token

Authentication Tokens

The GDC Data Transfer Tool and the GDC API use tokens for authentication. GDC authentication tokens are alphanumeric strings of characters like this one:

```
ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-
ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-
ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-
ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-
ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-
ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-
ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-
ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-
ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-ALPHANUMERICTOKEN-01234567890+AlPhAnUmErIcToKeN=0123456789-ALPHANUMERICTO
```

1.3.3 Logging into the GDC

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

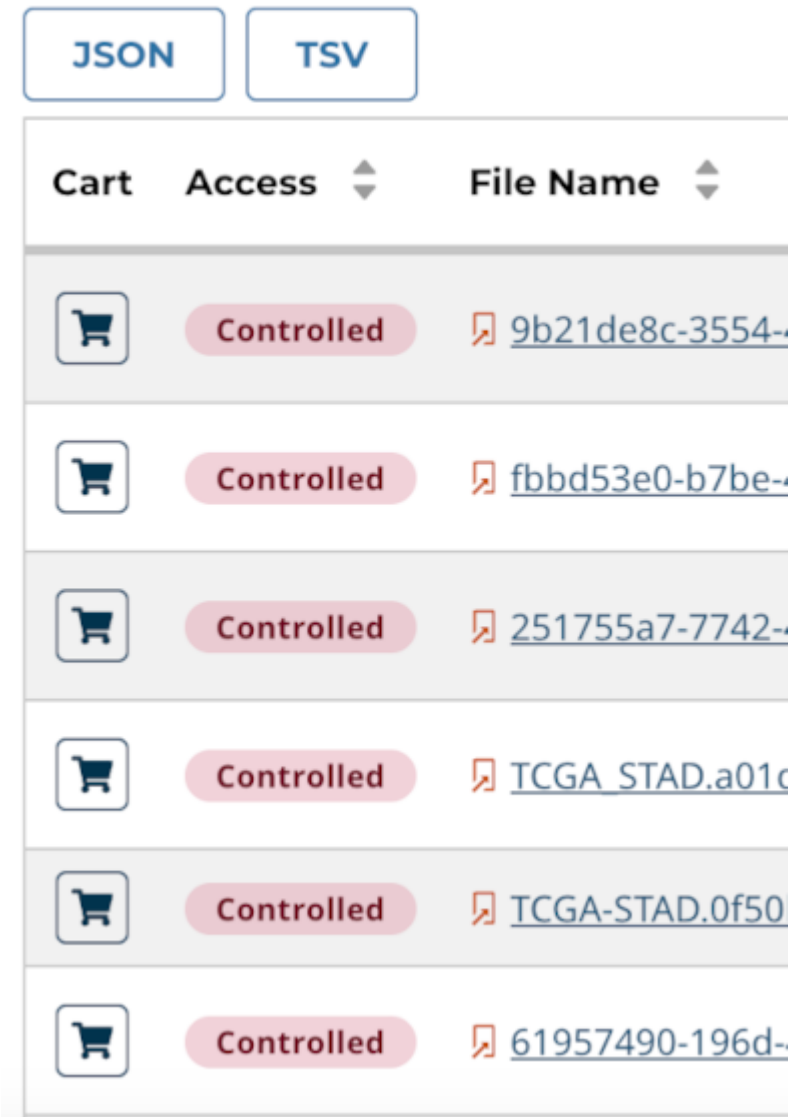
Login

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 can access.
- 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.
- See controlled-access mutation data they can access.

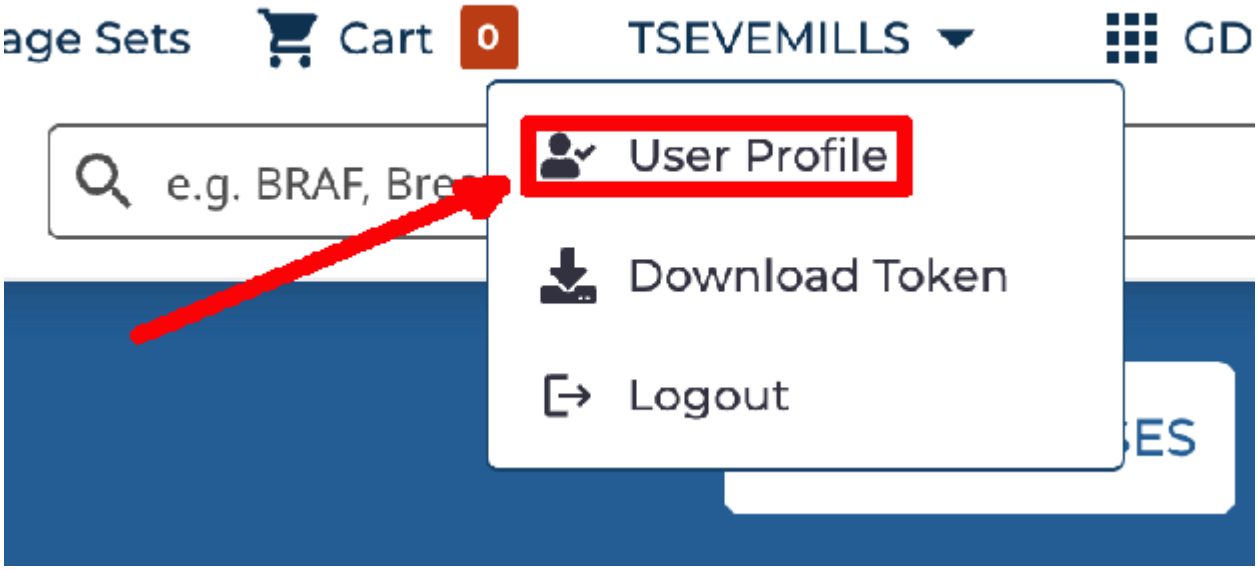
Controlled-access files are identified via their status in the Access Column:



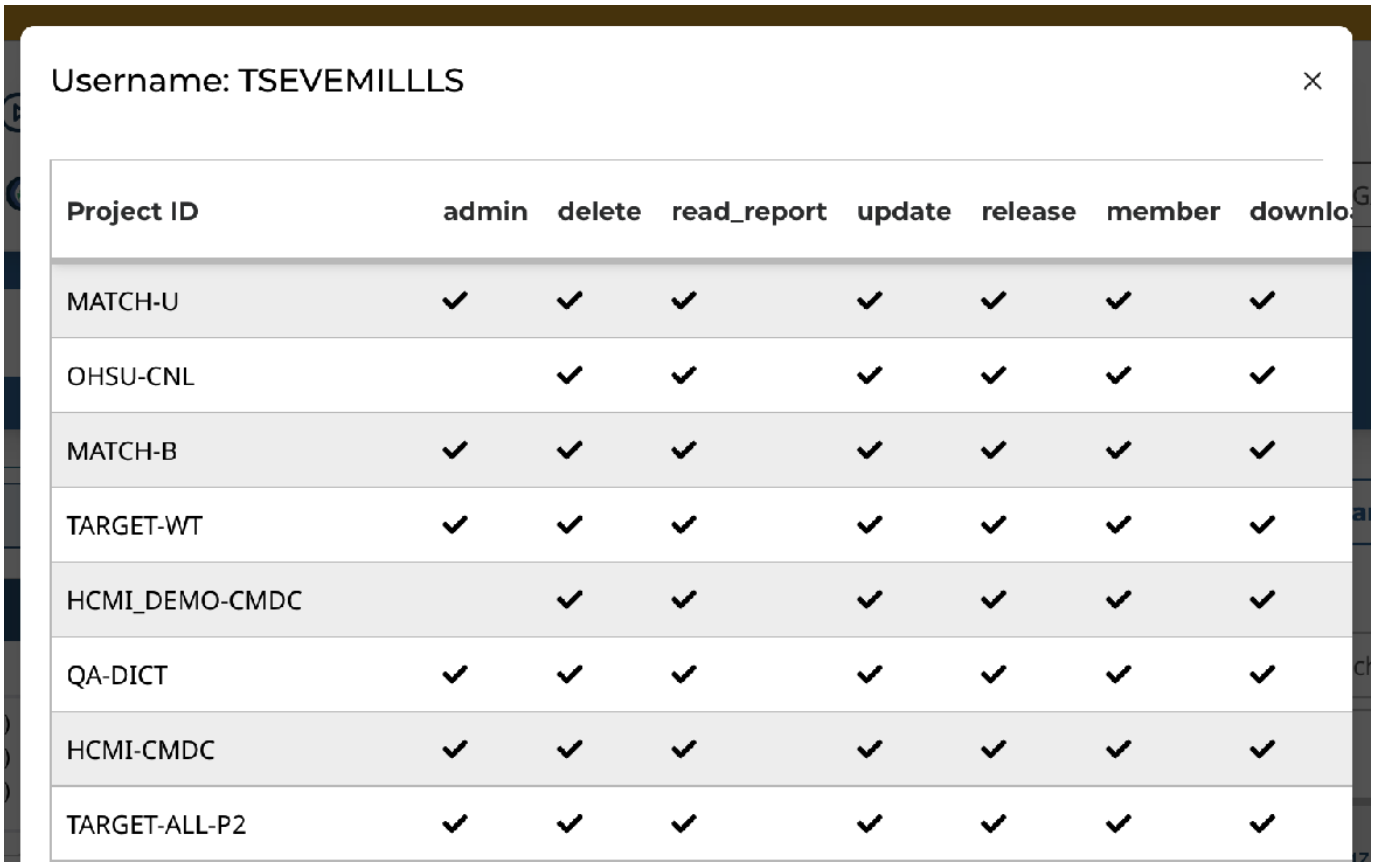
The rest of this section describes controlled data access features of the GDC Data Portal available to authorized users. For more information about open and controlled-access data, and about obtaining access to controlled data, see Data Access Processes and Tools.

1.3.4 User Profile

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



Clicking this button shows the list of projects.



1.3.5 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.

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.

1.3.6 Logging Out

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

Logout link

OBTAINING A TOKEN

Users can obtain authentication tokens from the GDC Data Portal and the GDC Data Submission Portal. See the GDC Data Submission Portal User's Guide for instructions.

TOKEN EXPIRATION

Tokens are valid for 30 days from the time of issue. Any request to the GDC API that uses an expired token will result in an error.

Tokens can be replaced at any time by downloading a new token, which will be valid for another 30 days.

1.3.7 Checking User Permissions

Users can view the permissions granted to them by the GDC system as follows:

1. Log into the GDC Data Portal or the GDC Data Submission Portal using your eRA Commons account.
2. Open the URL `https://portal.gdc.cancer.gov/auth/user` to see a JSON object that describes user permissions.

1.4 GDC MAF Format v.1.0.0

1.4.1 Introduction

Mutation Annotation Format (MAF) is a tab-delimited text file with aggregated mutation information from VCF Files and are generated on a project-level. MAF files are produced through the Somatic Aggregation Workflow. The GDC produces MAF files at two permission levels: **protected** and **somatic** (or open-access). One MAF file is produced per variant calling pipeline per GDC project. MAFs are produced by aggregating the GDC annotated VCF files generated from one pipeline for one project.

Annotated VCF files often have variants reported on multiple transcripts whereas the MAF files generated from the VCFs (*protected.maf) only report the most critically affected one. Somatic MAFs (*somatic.maf), which are also known as Masked Somatic Mutation files, are further processed to remove lower quality and potential germline variants. For tumor samples that contain variants from multiple combinations of tumor-normal aliquot pairs, only one pair is selected in the Somatic MAF based on their sample type. Somatic MAFs are publicly available and can be freely distributed within the boundaries of the GDC Data Access Policies.

The GDC MAF file format is based on the TCGA Mutation Annotation Format specifications, with additional columns included.

Note: The criteria for allowing mutations into open-access are purposefully implemented to overcompensate and filter out germline variants. If omission of true-positive somatic mutations is a concern, the GDC recommends using protected MAFs.

1.4.2 Somatic MAF File Generation

The process for modifying a protected MAF into a somatic MAF is as follows:

- Aliquot Selection: only one tumor-normal pair are selected for each tumor sample based on the plate number, sample type, analyte type and other features extracted from tumor TCGA aliquot barcode.
- Low quality variant filtering and germline masking:

a. Variants with `Mutation_Status != 'Somatic'` or `GDC_FILTER = 'Gapfiller', 'ContEst', 'multiallelic', 'nonselectedaliquot', 'BCR_Duplicate'` or `'BadSeq'` are **removed**.

b. Remaining variants with `GDC_Valid_Somatic = True` are **included** in the Somatic MAF.

c. Remaining variants with `FILTER != 'panel_of_normals'` or `PASS` are **removed**. Note that the `FILTER != panel_of_normals` value is only relevant for the variants generated from the MuTect2 pipeline.

d. Remaining variants with `MC3_Overlap = True` are **included** in the Somatic MAF.

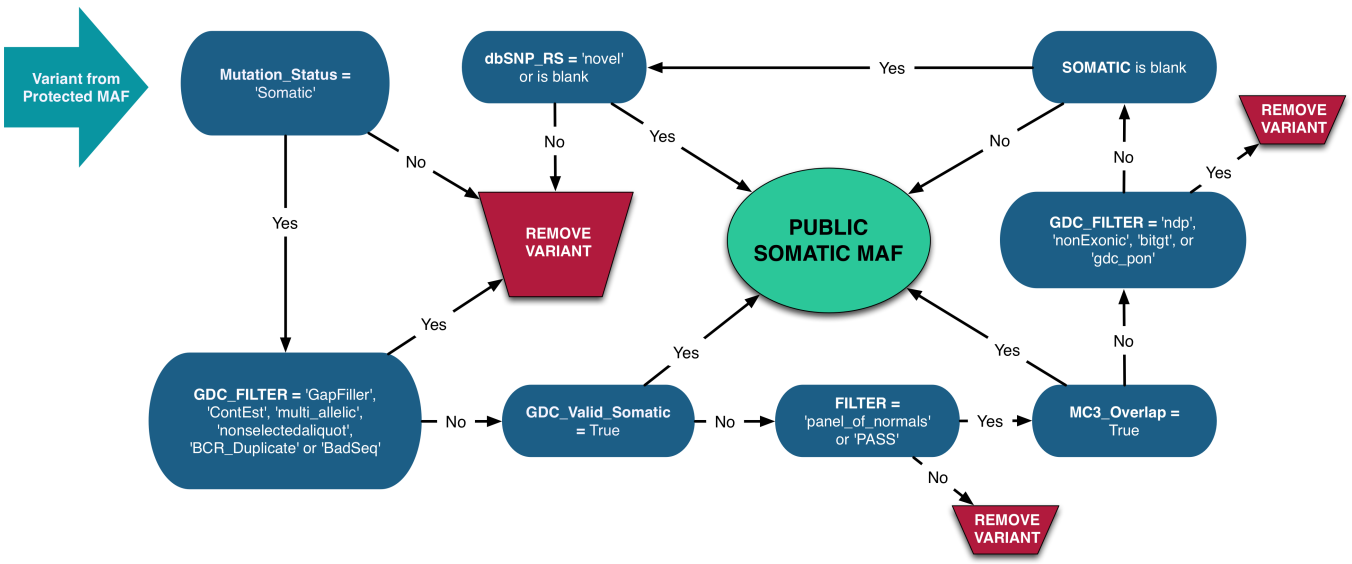
e. Remaining variants with `GDC_FILTER = 'ndp', 'NonExonic', 'bitgt', 'gdc_pon'` are **removed**.

f. Remaining variants with `SOMATIC != null` are **included** in the Somatic MAF.

g. Remaining variants with `dbSNP_RS = 'novel'` or `null` are **included** in the Somatic MAF.

h. Remaining variants are **removed**.

- Removal of the following columns:
- vcf_region
- vcf_info
- vcf_format
- vcf_tumor_gt
- vcf_normal_gt
- GDC_Valid_Somatic
- Set values to be blank in the following columns that may contain information about germline genotypes:
- Match_Norm_Seq_Allele1
- Match_Norm_Seq_Allele2
- Match_Norm_Validation_Allele1
- Match_Norm_Validation_Allele2
- n_ref_count
- n_alt_count


Variant from Protected MAF
Mutation_Status = 'Somatic'
dbSNP_RS = 'novel' or is blank
SOMATIC is blank
REMOVE VARIANT
PUBLIC SOMATIC MAF
GDC_FILTER = 'ndp', 'nonExonic', 'bitgt', or 'gdc_pon'
GDC_FILTER = 'GapFiller', 'ContEst', 'multi_allelic', 'nonselectedaliquot', 'BCR_Duplicate' or 'BadSeq'
GDC_Valid_Somatic = True
FILTER = 'panel_of_normals' or 'PASS'
MC3_Overlap = True
Yes
No

1.4.3 Protected MAF File Structure

The table below describes the columns in a protected MAF and their definitions. Note that the somatic (open-access) MAF structure is the same except for having the last six columns removed.

Column	Description
1 - Hugo_Symbol	HUGO symbol for the gene (HUGO symbols are always in all caps). "Unknown" is used for regions that d not correspond to a gene
2 - Entrez_Gene_Id	Entrez gene ID (an integer). "0" is used for regions that do not correspond to a gene region or Ensembl
3 - Center	One or more genome sequencing center reporting the variant
4 - NCBI_Build	The reference genome used for the alignment (GRCh38)
5 - Chromosome	The affected chromosome (chr1)
6 - Start_Position	Lowest numeric position of the reported variant on the genomic reference sequence. Mutation start coordinate
7 - End_Position	Highest numeric genomic position of the reported variant on the genomic reference sequence. Mutation end coordinate
8 - Strand	Genomic strand of the reported allele. Currently, all variants will report the positive strand: '+'
9 - Variant_Classification	Translational effect of variant allele
10 - Variant_Type	Type of mutation. TNP (tri-nucleotide polymorphism) is analogous to DNP (di-nucleotide polymorphism) for three consecutive nucleotides. ONP (oligo-nucleotide polymorphism) is analogous to TNP but for consecutive runs of four or more (SNP, DNP, TNP, ONP, INS, DEL, or Consolidated)
11 - Reference_Allele	The plus strand reference allele at this position. Includes the deleted sequence for a deletion or "-" for a insertion
12 - Tumor_Seq_Allele1	Primary data genotype for tumor sequencing (discovery) allele 1. A "-" symbol for a deletion represents variant. A "-" symbol for an insertion represents wild-type allele. Novel inserted sequence for insertion d not include flanking reference bases
13 - Tumor_Seq_Allele2	Tumor sequencing (discovery) allele 2
14 - dbSNP_RS	The rs-IDs from the dbSNP database, "novel" if not found in any database used, or null if there is no dbS record, but it is found in other databases
15 - dbSNP_Val_Status	The dbSNP validation status is reported as a semicolon-separated list of statuses. The union of all rs-ID taken when there are multiple
16 - Tumor_Sample_Barcode	Aliquot barcode for the tumor sample
17 - Matched_Norm_Sample_Barcode	Aliquot barcode for the matched normal sample
18 - Match_Norm_Seq_Allele1	Primary data genotype. Matched normal sequencing allele 1. A "-" symbol for a deletion represents a variant. A "-" symbol for an insertion represents wild-type allele. Novel inserted sequence for insertion d not include flanking reference bases (cleared in somatic MAF)
19 - Match_Norm_Seq_Allele2	Matched normal sequencing allele 2
20 - Tumor_Validation_Allele1	Secondary data from orthogonal technology. Tumor genotyping (validation) for allele 1. A "-" symbol for deletion represents a variant. A "-" symbol for an insertion represents wild-type allele. Novel inserted sequence for insertion does not include flanking reference bases
21 - Tumor_Validation_Allele2	Secondary data from orthogonal technology. Tumor genotyping (validation) for allele 2
22 - Match_Norm_Validation_Allele1	Secondary data from orthogonal technology. Matched normal genotyping (validation) for allele 1. A "-" symbol for a deletion represents a variant. A "-" symbol for an insertion represents wild-type allele. Nov inserted sequence for insertion does not include flanking reference bases (cleared in somatic MAF)
23 - Match_Norm_Validation_Allele2	Secondary data from orthogonal technology. Matched normal genotyping (validation) for allele 2 (cleare somatic MAF)
24 - Verification_Status	Second pass results from independent attempt using same methods as primary data source. Generally reserved for 3730 Sanger Sequencing

Column	Description
25 - Validation_Status	Second pass results from orthogonal technology
26 - Mutation_Status	An assessment of the mutation as somatic, germline, LOH, post transcriptional modification, unknown, c none. The values allowed in this field are constrained by the value in the Validation_Status field
27 - Sequencing_Phase	TCGA sequencing phase (if applicable). Phase should change under any circumstance that the targets ur consideration change
28 - Sequence_Source	Molecular assay type used to produce the analytes used for sequencing. Allowed values are a subset of t SRA 1.5 library_strategy field values. This subset matches those used at CGHub
29 - Validation_Method	The assay platforms used for the validation call
30 - Score	Not in use
31 - BAM_File	Not in use
32 - Sequencer	Instrument used to produce primary sequence data
33 - Tumor_Sample_UUID	GDC aliquot UUID for tumor sample
34 - Matched_Norm_Sample_UUID	GDC aliquot UUID for matched normal sample
35 - HGVSc	The coding sequence of the variant in HGVS recommended format
36 - HGVSp	The protein sequence of the variant in HGVS recommended format. "p.=" signifies no change in the prot
37 - HGVSp_Short	Same as the HGVSp column, but using 1-letter amino-acid codes
38 - Transcript_ID	Ensembl ID of the transcript affected by the variant
39 - Exon_Number	The exon number (out of total number)
40 - t_depth	Read depth across this locus in tumor BAM
41 - t_ref_count	Read depth supporting the reference allele in tumor BAM
42 - t_alt_count	Read depth supporting the variant allele in tumor BAM
43 - n_depth	Read depth across this locus in normal BAM
44 - n_ref_count	Read depth supporting the reference allele in normal BAM (cleared in somatic MAF)
45 - n_alt_count	Read depth supporting the variant allele in normal BAM (cleared in somatic MAF)
46 - all_effects	A semicolon delimited list of all possible variant effects, sorted by priority ([Symbol,Consequence,HGVSp_Short,Transcript_ID,RefSeq,HGVSc,Impact,Canonical,Sift,PolyPhen,Stra
47 - Allele	The variant allele used to calculate the consequence
48 - Gene	Stable Ensembl ID of affected gene
49 - Feature	Stable Ensembl ID of feature (transcript, regulatory, motif)
50 - Feature_type	Type of feature. Currently one of Transcript, RegulatoryFeature, MotifFeature (or blank)
51 - One_Consequence	The single consequence of the canonical transcript in sequence ontology terms
52 - Consequence	Consequence type of this variant; sequence ontology terms
53 - cDNA_position	Relative position of base pair in the cDNA sequence as a fraction. A "-" symbol is displayed as the numerator if the variant does not appear in cDNA
54 - CDS_position	Relative position of base pair in coding sequence. A "-" symbol is displayed as the numerator if the varia does not appear in coding sequence

Column	Description
55 - Protein_position	Relative position of affected amino acid in protein. A "-" symbol is displayed as the numerator if the varia does not appear in coding sequence
56 - Amino_acids	Only given if the variation affects the protein-coding sequence
57 - Codons	The alternative codons with the variant base in upper case
58 - Existing_variation	Known identifier of existing variation
59 - ALLELE_NUM	Allele number from input; 0 is reference, 1 is first alternate etc.
60 - DISTANCE	Shortest distance from the variant to transcript
61 - TRANSCRIPT_STRAND	The DNA strand (1 or -1) on which the transcript/feature lies
62 - SYMBOL	The gene symbol
63 - SYMBOL_SOURCE	The source of the gene symbol
64 - HGNC_ID	Gene identifier from the HUGO Gene Nomenclature Committee if applicable
65 - BIOTYPE	Biotype of transcript
66 - CANONICAL	A flag (YES) indicating that the VEP-based canonical transcript, the longest translation, was used for thi gene. If not, the value is null
67 - CCDS	The CCDS identifier for this transcript, where applicable
68 - ENSP	The Ensembl protein identifier of the affected transcript
69 - SWISSPROT	UniProtKB/Swiss-Prot accession
70 - TREMBL	UniProtKB/TrEMBL identifier of protein product
71 - UNIPARC	UniParc identifier of protein product
72 - RefSeq	RefSeq identifier for this transcript
73 - SIFT	The SIFT prediction and/or score, with both given as prediction (score)
74 - PolyPhen	The PolyPhen prediction and/or score
75 - EXON	The exon number (out of total number)
76 - INTRON	The intron number (out of total number)
77 - DOMAINS	The source and identifier of any overlapping protein domains
78 - GMAF	Non-reference allele and frequency of existing variant in 1000 Genomes
79 - AFR_MAF	Non-reference allele and frequency of existing variant in 1000 Genomes combined African population
80 - AMR_MAF	Non-reference allele and frequency of existing variant in 1000 Genomes combined American population
81 - ASN_MAF	Non-reference allele and frequency of existing variant in 1000 Genomes combined Asian population
82 - EAS_MAF	Non-reference allele and frequency of existing variant in 1000 Genomes combined East Asian populatio
83 - EUR_MAF	Non-reference allele and frequency of existing variant in 1000 Genomes combined European population
84 - SAS_MAF	Non-reference allele and frequency of existing variant in 1000 Genomes combined South Asian populati
85 - AA_MAF	Non-reference allele and frequency of existing variant in NHLBI-ESP African American population
86 - EA_MAF	Non-reference allele and frequency of existing variant in NHLBI-ESP European American population
87 - CLIN_SIG	Clinical significance of variant from dbSNP as annotated in ClinVar
88 - SOMATIC	Somatic status of each ID reported under Existing_variation (0, 1, or null)

Column	Description
89 - PUBMED	Pubmed ID(s) of publications that cite existing variant
90 - MOTIF_NAME	The source and identifier of a transcription factor binding profile aligned at this position
91 - MOTIF_POS	The relative position of the variation in the aligned TFBP
92 - HIGH_INF_POS	A flag indicating if the variant falls in a high information position of a transcription factor binding profile (TFBP) (Y, N, or null)
93 - MOTIF_SCORE_CHANGE	The difference in motif score of the reference and variant sequences for the TFBP
94 - IMPACT	The impact modifier for the consequence type
95 - PICK	Indicates if this block of consequence data was picked by VEP's pick feature (1 or null)
96 - VARIANT_CLASS	Sequence Ontology variant class
97 - TSL	Transcript support level, which is based on independent RNA analyses
98 - HGVS_OFFSET	Indicates by how many bases the HGVS notations for this variant have been shifted
99 - PHENO	Indicates if existing variant is associated with a phenotype, disease or trait (0, 1, or null)
100 - MINIMISED	Alleles in this variant have been converted to minimal representation before consequence calculation (1 null)
101 - ExAC_AF	Global Allele Frequency from ExAC
102 - ExAC_AF_Adj	Adjusted Global Allele Frequency from ExAC
103 - ExAC_AF_AFR	African/African American Allele Frequency from ExAC
104 - ExAC_AF_AMR	American Allele Frequency from ExAC
105 - ExAC_AF_EAS	East Asian Allele Frequency from ExAC
106 - ExAC_AF_FIN	Finnish Allele Frequency from ExAC
107 - ExAC_AF_NFE	Non-Finnish European Allele Frequency from ExAC
108 - ExAC_AF_OTH	Other Allele Frequency from ExAC
109 - ExAC_AF_SAS	South Asian Allele Frequency from ExAC
110 - GENE_PHENO	Indicates if gene that the variant maps to is associated with a phenotype, disease or trait (0, 1, or null)
111 - FILTER	Copied from input VCF. This includes filters implemented directly by the variant caller and other externa software used in the DNA-Seq pipeline. See below for additional details.
112 - CONTEXT	The reference allele per VCF specs, and its five flanking base pairs
113 - src_vcf_id	GDC UUID for the input VCF file
114 - tumor_bam_uuid	GDC UUID for the tumor bam file
115 - normal_bam_uuid	GDC UUID for the normal bam file
116 - case_id	GDC UUID for the case
117 - GDC_FILTER	GDC filters applied universally across all MAFs
118 - COSMIC	Overlapping COSMIC variants
119 - MC3_Overlap	Indicates whether this region overlaps with an MC3 variant for the same sample pair
120 - GDC_Validation_Status	GDC implementation of validation checks. See notes section (#5) below for details
121 - GDC_Valid_Somatic	True or False (not in somatic MAF)

Column	Description
122 - vcf_region	Colon separated string containing the CHROM, POS, ID, REF, and ALT columns from the VCF file (e.g., chrZ:20:rs1234:A:T) (not in somatic MAF)
123 - vcf_info	INFO column from VCF (not in somatic MAF)
124 - vcf_format	FORMAT column from VCF (not in somatic MAF)
125 - vcf_tumor_gt	Tumor sample genotype column from VCF (not in somatic MAF)
126 - vcf_normal_gt	Normal sample genotype column from VCF (not in somatic MAF)

Notes About GDC MAF Implementation

1. Column #4: **NCBI_Build** is GRCh38 by default
2. Column #32: **Sequencer** includes the sequencers used. If different sequencers were used to generate normal and tumor data, the normal sequencer is listed first.
3. Column #61: VEP name "STRAND" is changed to **TRANSCRIPT_STRAND** to avoid confusion with Column#8 "Strand"
4. Column #94: **IMPACT** categories are defined by the VEP software and do not necessarily reflect the relative biological influence of each mutation.
5. Column #122-125: **vcf_info, vcf_format, vcf_tumor_gt, and vcf_normal_gt** are the corresponding columns from the VCF files. Including them facilitates parsing specific variant information.
6. Column #120: **GDC_Validation_Status**: GDC also collects TCGA validation sequences. It compares these with variants derived from Next-Generation Sequencing data from the same sample and populates the comparison result in "GDC_Validation_Status".

- "Valid", if the alternative allele(s) in the tumor validation sequence is(are) the same as GDC variant call
- "Invalid", if none of the alternative allele(s) in the tumor validation sequence is the same as GDC variant call
- "Inconclusive" if two alternative allele exists, and one matches while the other does not
- "Unknown" if no validation sequence exists

7. Column #121: **GDC_Valid_Somatic** is TRUE if GDC_Validation_Status is "Valid" and the variant is "Somatic" in validation calls. It is FALSE if these criteria are not met

FILTER Value Definitions (column 111)

- **oxog :** Signifies that this variant was determined to be an OxoG artifact. This was calculated with D-ToxoG.
- **bPcr :** Signifies that this variant was determined to be an artifact of bias on the PCR template strand. This was calculated with the DKFZ Bias Filter.
- **bSeq :** Signifies that this variant was determined to be an artifact of bias on the forward/reverse strand. This was also calculated with the DKFZ Bias Filter.

1.4.4 Impact Categories

VEP

- **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

PolyPhen

- **probably damaging (PR)**: It is with high confidence supposed to affect protein function or structure
- **possibly damaging (PO)**: It is supposed to affect protein function or structure
- **benign (BE)**: Most likely lacking any phenotypic effect
- **unknown (UN)**: When in some rare cases, the lack of data does not allow PolyPhen to make a prediction

SIFT

- **tolerated**: Not likely to have a phenotypic effect
- **tolerated_low_confidence**: More likely to have a phenotypic effect than 'tolerated'
- **deleterious**: Likely to have a phenotypic effect
- **deleterious_low_confidence**: Less likely to have a phenotypic effect than 'deleterious'

1.5 GDC VCF Format

1.5.1 Introduction

The GDC DNA-Seq somatic variant-calling pipeline compares a set of matched tumor/normal alignments and produces a VCF file. VCF files report the somatic variants that were detected by each of the four variant callers. Four raw VCFs (Data Type: Raw Simple Somatic Mutation) are produced for each tumor/normal pair of BAMs. Four additional annotated VCFs (Data Type: Annotated Somatic Mutation) are produced by adding biologically relevant information about each variant.

The GDC VCF file format follows standards of the Variant Call Format (VCF) Version 4.1 Specification. Raw Simple Somatic Mutation VCF files are unannotated, whereas Annotated Somatic Mutation VCF files include extensive, consistent, and pipeline-agnostic annotation of somatic variants.

1.5.2 VCF file structure

Metadata header

A VCF file starts with lines of metadata that begin with `##`. Some key components of this section include:

- **gdcWorkflow:** Information on the pipelines that were used by the GDC to generate the VCF file. Annotated VCF files contain two *gdcWorkflow* lines, one that reports the variant calling process and one that reports the variant annotation process.
- **INDIVIDUAL:** information about the study participant (`case`), including:
- *NAME:* Submitter ID (barcode) associated with the participant
- *ID:* GDC case UUID
- **SAMPLE:** sample information, including:
- *ID:* NORMAL or TUMOR
- *NAME:* Submitter ID (barcode) of the aliquot
- *ALIQUOT_ID:* GDC aliquot UUID
- *BAM_ID:* The UUID for the BAM file used to produce the VCF
- **INFO:** Format of *additional information* fields
- NOTE: GDC Annotated VCFs may contain multiple INFO lines. The last INFO line contains information about annotation fields generated by the Somatic Annotation Workflow (see GDC INFO Fields below).
- **FILTER:** Description of filters that have been applied to the variants
- **FORMAT:** Description of genotype fields
- **reference:** The reference genome used to generate the VCF file (GRCh38.d1.vd1.fa)
- **contig:** A list of IDs for the contiguous DNA sequences that appear in the reference genome used to produce VCF files
- NOTE: Annotated VCFs include contig information for autosomes, sex chromosomes, and mitochondrial DNA. Unplaced, unlocalized, human decoy, and viral genome sequences are not included.
- **VEP:** the VEP command used by the Somatic Annotation Workflow to generate the annotated VCF file.

Column Header Line

Each variant is represented by a row in the VCF file. Below each of the columns are described:

1. **CHROM:** The chromosome on which the variant is located
2. **POS:** The position of the variant on the chromosome. Refers to the first position if the variant includes more than one base
3. **ID:** A unique identifier for the variant; usually a dbSNP rs number if applicable
4. **REF:** The base(s) exhibited by the reference genome at the variant's position
5. **ALT:** The alternate allele(s), comma-separated if there are more than one
6. **QUAL:** Not populated
7. **FILTER:** The names of the filters that have flagged this variant. The types of filters used will depend on the variant caller used.
8. **INFO:** Additional information about the variant. This includes the annotation applied by the VEP.
9. **FORMAT:** The format of the sample genotype data in the next two columns. This includes descriptions of the colon-separated values.
10. **NORMAL:** Colon-separated values that describe the normal sample
11. **TUMOR:** Colon-separated values that describe the tumor sample

See Variant Call Format (VCF) Version 4.1 Specification for details.

1.5.3 GDC INFO fields

The following variant annotation fields are currently included in Annotated Somatic Mutation VCF files. Please refer to the DNA-Seq Analysis Pipeline documentation for details on how this information is generated. VEP Documentation provides additional information about some of these fields.

Field	Description
Allele	The variant allele used to calculate the consequence
Consequence	Consequence type of this variant
IMPACT	The impact modifier for the consequence type
SYMBOL	The HUGO gene symbol
Gene	Ensembl stable ID of the affected gene
Feature_type	Type of feature. Currently one of Transcript, RegulatoryFeature, MotifFeature.
Feature	Ensembl stable ID of the feature
BIOTYPE	The type of transcript or regulatory feature (e.g. protein_coding)
EXON	Exon number (out of total exons)
INTRON	Intron number (out of total introns)
HGVSc	The HGVS coding sequence name
HGVSp	The HGVS protein sequence name
cDNA_position	Relative position of base pair in cDNA sequence
CDS_position	Relative position of base pair in coding sequence
Protein_position	Relative position of the affected amino acid in protein
Amino_acids	Change in amino acids (only given if the variant affects the protein-coding sequence)
Codon	The affected codons with the variant base in upper case
Existing_variation	Known identifier of existing variant; usually a dbSNP rs number if applicable
ALLELE_NUM	Allele number from input; 0 is reference, 1 is first alternate, etc.
DISTANCE	Shortest distance from variant to transcript
STRAND	The DNA strand (1 or -1) on which the transcript/feature lies
FLAGS	Transcript quality flags
VARIANT_CLASS	Sequence Ontology variant class
SYMBOL_SOURCE	The source of the gene symbol
HGNC_ID	HGNC gene ID
CANONICAL	A flag indicating if the transcript is denoted as the canonical transcript for this gene
TSL	Transcript support level
APPRIS	APPRIS isoform annotation
CCDS	The CCDS identifer for this transcript, where applicable
ENSP	The Ensembl protein identifier of the affected transcript
SWISSPROT	UniProtKB/Swiss-Prot identifier of protein product
TREMBL	UniProtKB/TrEMBL identifier of protein product
UNIPARC	UniParc identifier of protein product
RefSeq	RefSeq gene ID
GENE_PHENO	Indicates if the gene is associated with a phenotype, disease or trait

Field	Description
SIFT	The SIFT prediction and/or score, with both given as prediction (score)
PolyPhen	The PolyPhen prediction and/or score
DOMAINS	The source and identifier of any overlapping protein domains
HGVS_OFFSET	Indicates by how many bases the HGVS notations for this variant have been shifted
GMAF	Non-reference allele and frequency of existing variant in 1000 Genomes
AFR_MAF	Non-reference allele and frequency of existing variant in 1000 Genomes combined African population
AMR_MAF	Non-reference allele and frequency of existing variant in 1000 Genomes combined American population
EAS_MAF	Non-reference allele and frequency of existing variant in 1000 Genomes combined East Asian population
EUR_MAF	Non-reference allele and frequency of existing variant in 1000 Genomes combined European population
SAS_MAF	Non-reference allele and frequency of existing variant in 1000 Genomes combined South Asian population
AA_MAF	Non-reference allele and frequency of existing variant in NHLBI-ESP African American population
EA_MAF	Non-reference allele and frequency of existing variant in NHLBI-ESP European American population
ExAC_MAF	Frequency of existing variant in ExAC combined population
ExAC_Adj_MAF	Adjusted frequency of existing variant in ExAC combined population
ExAC_AFR_MAF	Frequency of existing variant in ExAC African/American population
ExAC_AMR_MAF	Frequency of existing variant in ExAC American population
ExAC_EAS_MAF	Frequency of existing variant in ExAC East Asian population
ExAC_FIN_MAF	Frequency of existing variant in ExAC Finnish population
ExAC_NFE_MAF	Frequency of existing variant in ExAC Non-Finnish European population
ExAC_OTH_MAF	Frequency of existing variant in ExAC combined other combined populations
ExAC_SAS_MAF	Frequency of existing variant in ExAC South Asian population
CLIN_SIG	Clinical significance of variant from dbSNP
SOMATIC	Somatic status of existing variant(s)
PHENO	Indicates if existing variant is associated with a phenotype, disease or trait
PUBMED	Pubmed ID(s) of publications that cite existing variant
MOTIF_NAME	The source and identifier of a transcription factor binding profile aligned at this position
MOTIF_POS	The relative position of the variation in the aligned TFBP
HIGH_INF_POS	A flag indicating if the variant falls in a high information position of a transcription factor binding profile (TFBP)
MOTIF_SCORE_CHANGE	The difference in motif score of the reference and variant sequences for the TFBP
ENTREZ	Entrez ID

Field	Description
EVIDENCE	Evidence that the variant exists

1.6 DNA-Seq Analysis Pipeline

1.6.1 Introduction

The GDC DNA-Seq analysis pipeline identifies somatic variants within whole exome sequencing (WXS) and whole genome sequencing (WGS) data. Somatic variants are identified by comparing allele frequencies in normal and tumor sample alignments, annotating each mutation, and aggregating mutations from multiple cases into one project file.

The first pipeline starts with a reference alignment step followed by co-cleaning to increase the alignment quality. Four different variant calling pipelines are then implemented separately to identify somatic mutations. Somatic-caller-identified variants are then annotated. An aggregation pipeline incorporates variants from all cases in one project into a MAF file for each pipeline.

DNA-Seq analysis is implemented across six main procedures:

- Genome Alignment
- Alignment Co-Cleaning
- Somatic Variant Calling
- Variant Annotation
- Mutation Aggregation
- Aggregated Mutation Masking

1.6.2 Data Processing Steps

Pre-Alignment

Prior to alignment, BAM files that were submitted to the GDC are split by read groups and converted to FASTQ format. Reads that failed the Illumina chastity test are removed. Note that this filtering step is distinct from trimming reads using base quality scores.

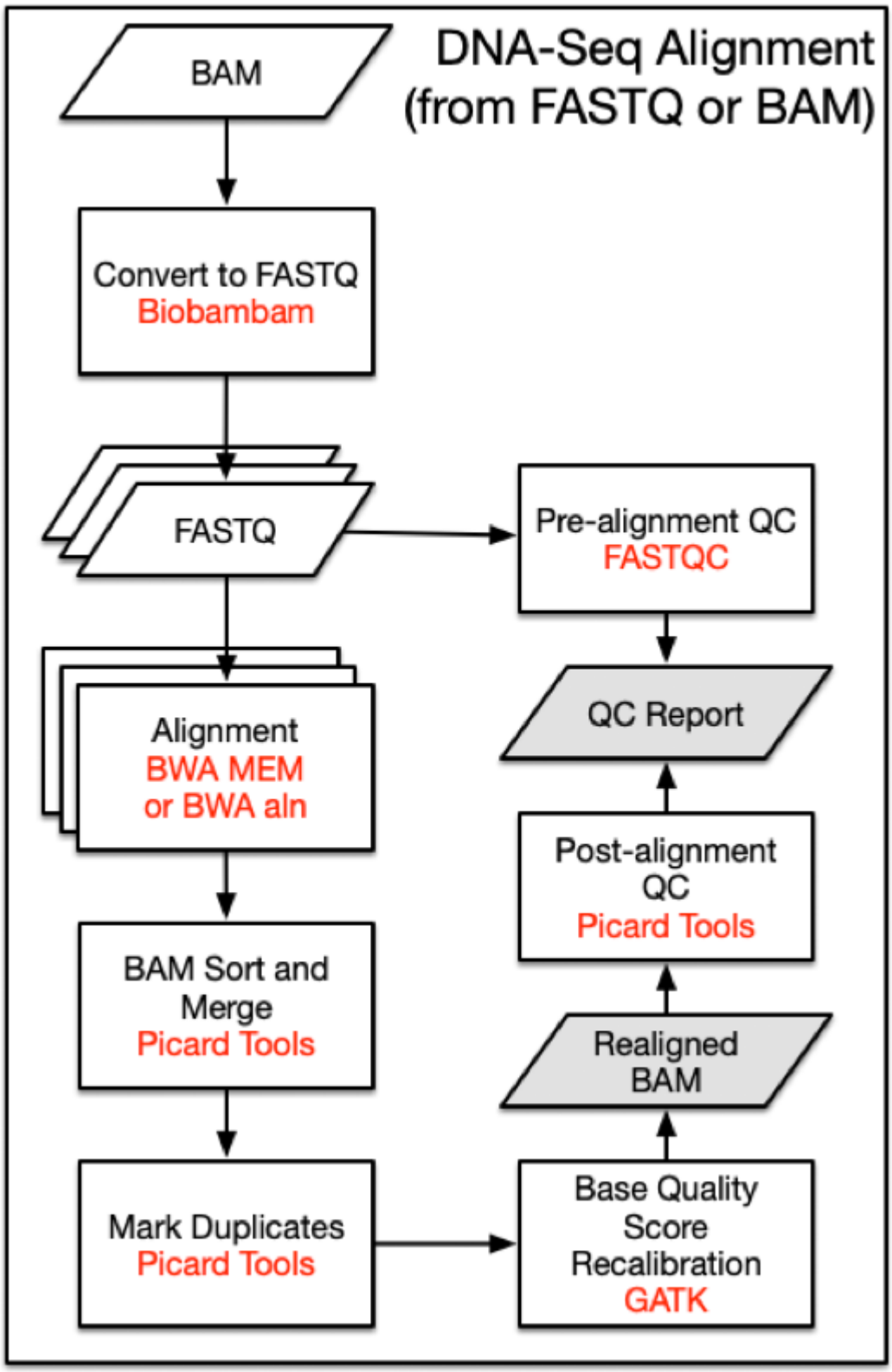
Alignment Workflow

DNA-Seq analysis begins with the Alignment Workflow. Read groups are aligned to the reference genome using one of two BWA algorithms [1]. BWA-MEM is used if mean read length is greater than or equal to 70 bp. Otherwise BWA-aln is used. Each read group is aligned to the reference genome separately and all read group alignments that belong to a single aliquot are merged using Picard Tools SortSam and MergeSamFiles. Duplicate reads, which may persist as PCR artifacts, are then flagged to prevent downstream variant call errors.

REFERENCE GENOME

All alignments are performed using the human reference genome GRCh38.d1.vd1. Decoy viral sequences are included in the reference genome to prevent reads from aligning erroneously and attract reads from viruses known to be present in human samples. Ten types of human viral genomes are included: human cytomegalovirus (CMV), Epstein-Barr virus (EBV), hepatitis B (HBV), hepatitis C (HCV), human immunodeficiency virus (HIV), human herpes virus 8 (HHV-8), human T-lymphotropic virus 1 (HTLV-1), Merkel cell polyomavirus (MCV), Simian vacuolating virus 40 (SV40), and human papillomavirus (HPV). Reference sequences used by the GDC can be downloaded here.

I/O	Entity	Format
Input	Submitted Unaligned Reads or Submitted Aligned Reads	FASTQ or BAM
Output	Aligned Reads	BAM



DNA-Seq Alignment Command Line Parameters

Note that version numbers may vary in files downloaded from the GDC Portal due to ongoing pipeline development and improvement.

STEP 1: CONVERTING BAMS TO FASTQS WITH BIOBAMBAM - BIOBAMBAM2

Shell

```
bamtofastq \
collate=1 \
exclude=QCFAIL,SECONDARY,SUPPLEMENTARY \
filename= <input.bam> \
gz=1 \
inputformat=bam \
level=5 \
outputdir= <output_path> \
outputperreadgroup=1 \
outputperreadgroupsuffixF=_1.fq.gz \
outputperreadgroupsuffixF2=_2.fq.gz \
outputperreadgroupsuffixO=_o1.fq.gz \
outputperreadgroupsuffixO2=_o2.fq.gz \
outputperreadgroupsuffixS=_s.fq.gz \
tryoq=1 \
```

STEP 2: BWA ALIGNMENT - BWA - SAMTOOLS

If mean read length is greater than or equal to 70bp:

Shell

```
bwa mem \
-t 8 \
-T 0 \
-R <read_group> \
<reference> \
<fastq_1.fq.gz> \
<fastq_2.fq.gz> |
samtools view \
-Shb
-o <output.bam> -
```

If mean read length is less than 70bp:

Shell

```
bwa aln -t 8 <reference> <fastq_1.fq.gz> > <sai_1.sai> &&
bwa aln -t 8 <reference> <fastq_2.fq.gz> > <sai_2.sai> &&
bwa sampe -r <read_group> <reference> <sai_1.sai> <sai_2.sai> <fastq_1.fq.gz> <fastq_2.fq.gz> | samtools view -Shb -o <output.bam> -
```

If the quality scores are encoded as Illumina 1.3 or 1.5, use BWA aln with the "-l" flag.

STEP 3: BAM SORT - PICARD 2

Shell

```
java -jar picard.jar SortSam \
CREATE_INDEX=true \
INPUT=<input.bam> \
OUTPUT=<output.bam> \
SORT_ORDER=coordinate \
VALIDATION_STRINGENCY=STRICT
```

STEP 4: BAM MERGE - PICARD 2

Shell

```
java -jar picard.jar MergeSamFiles \
ASSUME_SORTED=false \
CREATE_INDEX=true \
[INPUT= <input.bam>]  \
MERGE_SEQUENCE_DICTIONARIES=false \
OUTPUT= <output_path> \
SORT_ORDER=coordinate \
USE_THREADING=true \
VALIDATION_STRINGENCY=STRICT
```

STEP 5: MARK DUPLICATES - PICARD 2

Shell

```
java -jar picard.jar MarkDuplicates \
CREATE_INDEX=true \
INPUT=<input.bam> \
VALIDATION_STRINGENCY=STRICT
```

Co-cleaning Workflow

The alignment quality is further improved by the Co-cleaning workflow. Co-cleaning is performed as a separate pipeline as it uses multiple BAM files (i.e. the tumor BAM and normal tissue BAM) associated with the same patient. Both steps of this process are implemented using GATK.

INDEL LOCAL REALIGNMENT

Local realignment of insertions and deletions is performed using IndelRealigner. This step locates regions that contain misalignments across BAM files, which can often be caused by insertion-deletion (indel) mutations with respect to the reference genome. Misalignment of indel mutations, which can often be erroneously scored as substitutions, reduces the accuracy of downstream variant calling steps.

BASE QUALITY SCORE RECALIBRATION

A base quality score recalibration (BQSR) step is then performed using BaseRecalibrator. This step adjusts base quality scores based on detectable and systematic errors. This step also increases the accuracy of downstream variant calling algorithms. Note that the original quality scores are kept in the OQ field of co-cleaned BAM files. These scores should be used if conversion of BAM files to FASTQ format is desired.

I/O	Entity	Format
Input	Aligned Reads	BAM
Output	Harmonized Aligned Reads	BAM

DNA-Seq Co-Cleaning Command Line Parameters

STEP 1: REALIGNTARGETCREATOR

Shell

```
java -jar GenomeAnalysisTK.jar \
-T RealignerTargetCreator \
-R <reference>
-known <known_indels.vcf>
[ -I <input.bam> ]
-o <realign_target.intervals>
```

STEP 2: INDELREALIGNER

Shell

```
java -jar GenomeAnalysisTK.jar \
-T IndelRealigner \
-R <reference> \
-known <known_indels.vcf> \
-targetIntervals <realign_target.intervals> \
--noOriginalAlignmentTags \
[ -I <input.bam> ] \
-nWayOut <output.map>
```

STEP 3: BASERECALIBRATOR; DBSNP V.144

Shell

```
java -jar GenomeAnalysisTK.jar \
-T BaseRecalibrator \
-R <reference> \
-I <input.bam> \
-knownSites <dbsnp.vcf>
-o <bqsr.grp>
```

STEP 4: PRINTREADS

Shell

```
java -jar GenomeAnalysisTK.jar \
-T PrintReads \
-R <reference> \
-I <input.bam> \
--BQSR <bqsr.grp> \
-o <output.bam>
```

Somatic Variant Calling Workflow

Aligned and co-cleaned BAM files are processed through the Somatic Mutation Calling Workflow as tumor-normal pairs. Variant calling is performed using four separate pipelines:

- MuSE [2]
- MuTect2 [3]
- VarScan2 [4]
- Pindel

Note that SomaticSniper [5] was used and available on the GDC Data Portal prior to GDC Data Release 35.

Variant calls are reported by each pipeline in a VCF formatted file. See the GDC VCF Format documentation for details on each available field. At this point in the DNA-Seq pipeline, all downstream analyses are branched into four separate paths that correspond to their respective variant calling pipeline.

PIPELINE DESCRIPTIONS

Four separate variant calling pipelines are implemented for GDC data harmonization. There is currently no scientific consensus on the best variant calling pipeline so the investigator is responsible for choosing the pipeline(s) most appropriate for the data. Some details about the pipelines are indicated below.

The MuTect2 pipeline employs a "Panel of Normals" to identify additional germline mutations. This panel is generated using TCGA blood normal genomes from thousands of individuals that were curated and confidently assessed to be cancer-free. This method allows for a higher level of confidence to be assigned to somatic variants that were called by the MuTect2 pipeline.

Basic outlines for the other two pipelines can be found here:

- VarScan2 pipeline
- MuSE pipeline

INDELS

Indel mutations that were generated with the MuTect2, Pindel, and VarScan pipelines are detected and reported in GDC VCF files.

GERMLINE VARIANTS

At this time, germline variants are deliberately excluded as harmonized data. The GDC does not recommend using germline variants that were previously detected and stored in the Legacy Archive as they do not meet the GDC criteria for high-quality data.

I/O	Entity	Format
Input	Aligned Reads	BAM
Output	Raw Simple Somatic Mutation	VCF

Variant Call Command-Line Parameters

MUSE

MuSEv1.0; dbSNP v.144

Step 1: MuSE call

Shell

```
MuSE call \
-f <reference> \
-r <region> \
<tumor.bam> \
<normal.bam> \
-O <intermediate_muse_call.txt>
```

Step 2: MuSE sump

Shell

```
MuSE sump \
-I <intermediate_muse_call.txt> \
-E \
-D <dbsnp_known_snp_sites.vcf> \
-O <muse_variants.vcf>
```

Note: -E is used for WXS data and -G can be used for WGS data.

MUTECT2

GATK; dbSNP v.144

Shell

```
java -jar GenomeAnalysisTK.jar \
-T MuTect2 \
-R <reference> \
-L <region> \
-I:tumor <tumor.bam> \
-I:normal <normal.bam> \
--normal_panel <pon.vcf> \
--cosmic <cosmic.vcf> \
--dbsnp <dbsnp.vcf> \
--contamination_fraction_to_filter 0.02 \
-o <mutect_variants.vcf> \
--output_mode EMIT_VARIANTS_ONLY \
--disable_auto_index_creation_and_locking_when_reading_rods
```

VARSCAN

Step 1: Mpileup; Samtools

Shell

```
samtools mpileup \
-f <reference> \
-q 1 \
-B \
<normal.bam> \
<tumor.bam> >
<intermediate_mpileup.pileup>
```

Step 2: Varscan Somatic; Varscan.v2

Shell

```
java -jar VarScan.jar somatic \
<intermediate_mpileup.pileup> \
<output_path> \
--mpileup      1 \
--min-coverage 8 \
--min-coverage-normal 8 \
--min-coverage-tumor 6 \
--min-var-freq 0.10 \
--min-freq-for-hom 0.75 \
--normal-purity 1.0 \
--tumor-purity 1.00 \
--p-value 0.99 \
--somatic-p-value 0.05 \
--strand-filter 0 \
--output-vcf
```

Step 3: Varscan ProcessSomatic; Varscan.v2

Shell

```
java -jar VarScan.jar processSomatic \
<intermediate_varscan_somatic.vcf> \
--min-tumor-freq 0.10 \
--max-normal-freq 0.05 \
--p-value 0.07
```

PINDEL

Step 1: Filter Reads

Filter BAM reads that are not unmapped or duplicate or secondary_alignment or failed_quality_control or supplementary for both tumor and normal BAM files

Tool: sambamba

Shell

```
Sambamba view $(input.bam) --filter "not (unmapped or duplicate or secondary_alignment or failed_quality_control or supplementary)" --format bam --nthreads 1 --
output-filename $(output.bam)
```

Step 2: Pindel

Pindel Repo

Step 2a.: Calculate mean insert size

Python

```
cmd = "samtools view -f66 %s | head -n 1000000" % (bam)
output = do_shell_command(cmd)
lines = output.decode('utf-8').split('\n')
b_sum = 0
b_count = 0
numlines = 0
for line in lines:
    numlines += 1
    tmp = line.split("\t")
    if len(tmp) < 9:
        break
    if abs(int(tmp[8])) < 10000:
        b_sum += abs(int(tmp[8]))
        b_count += 1
try:
    mean = b_sum / b_count
```

Step 2b.: Write it to a config file

Python

```
for inputBamFile, meanInsertSize, tag in zip(inputBamFiles, meanInsertSizes, tags):
        fil.write("%s\t%s\t%s\n" %(inputBamFile, meanInsertSize, tag))
    fil.close()
```

Step 2c.: Run pindel

Shell

```
pindel \
-f GRCh38.d1.vd1.fa \
-i config_file \
-o $(output_prefix) \
--exclude GRCh38.d1.vd1.centromeres.telomeres.bed
```

Step 2d.: Merge DI and SI OUTPUT

Python

```
with open(os.path.join(args.workdir, "pindel_somatic"), "w") as handle:
        for p in pindel_files:
            if p.endswith("_D"):
            with open(p) as ihandle:
                for line in ihandle:
                    if re.search("ChrID", line):
                        handle.write(line)
        for p in pindel_files:
            if p.endswith("_SI"):
            with open(p) as ihandle:
                for line in ihandle:
                if re.search("ChrID", line):
                    handle.write(line)
```

Step 2e.: Create a config for pindel somatic filter

Python

```
indel.filter.input = $(merged.pindel.output)
indel.filter.vaf = 0.08
indel.filter.cov = 20
indel.filter.hom = 6
indel.filter.pindel2vcf = "/path/to/pindel/pindel2vcf4tcga"
indel.filter.reference =  "GRCh38.d1.vd1.fa"
indel.filter.referencename = "GRCh38"
indel.filter.referencedate = datetime.datetime.now().strftime("%Y%m%d")
indel.filter.output = $(output.file.name.vcf)
```

Step 2f.: Apply somatic filter on pindel output Tool: pindel2vcf4tcga

Perl

```
perl pindel/somatic_filter/somatic_indelfilter.pl $(somatic.indel.filter.config)
```

Step 3: Pindel Tool: Picard.jar 2

Shell

```
java \
-d64 \
-XX: +UseSerialGC \
-Xmx16G \
-jar picard.jar \
SortVcf \
CREATE_INDEX=true \
SEQUENCE_DICTIONARY=GRCh38.d1.vd1.dict \
I=$(pindel.somatic.vcf) \
OUTPUT=$(output.vcf.gz)
```

Step 5: Vt Normalization Tool: GenomeAnalysisTK.jar nightly-2016-02-25-gf39d340

Shell

```
java \
-Xmx4G \
-jar \
/bin/GenomeAnalysisTK.jar \
-T VariantFiltration \
--disable_auto_index_creation_and_locking_when_reading_rods \
--variant $(vt.normal.output.vcf.gz) \
-R GRCh38.d1.vd1.fa \
--filterExpression vc.isBiallelic() && vc.getGenotype(\"TUMOR\").getAD().1 < 3" \
--filterName TALTDP \
-o $(output.vcf.gz)
```

Variant Call Annotation Workflow

Raw VCF files are then annotated in the Somatic Annotation Workflow with the Variant Effect Predictor (VEP) v84 [6] along with VEP GDC plugins.

The VEP uses the coordinates and alleles in the VCF file to infer biological context for each variant including the location of each mutation, its biological consequence (frameshift/ silent mutation), and the affected genes. See the documentation on the GDC VCF Format for more details. Variants in the VCF files are also matched to known variants from external mutation databases. The following databases are used for VCF annotation:

- GENCODE v.22
- sift v.5.2.2
- ESP v.20141103
- polyphen v.2.2.2
- dbSNP v.146
- Ensembl genebuild v.2014-07
- Ensembl regbuild v.13.0
- HGMD public v.20154
- ClinVar v.201601

Due to licensing constraints COSMIC is not utilized for annotation in the GDC VEP workflow.

In addition to annotation, False Positive Filter is used to label low quality variants in VarScan.

I/O	Entity	Format
Input	Simple Somatic Mutation	VCF
Output	Annotated Somatic Mutation	VCF

Tumor-Only Variant Calling Workflow

Tumor only variant calling is performed on a tumor sample with no paired normal at the request of the research group. This method takes advantage of the normal cell contamination that is present in most tumor samples. These calls are made using the version of MuTect2 included in GATK4. Tumor-only variant call files can be found in the GDC Portal by filtering for "Workflow Type: GATK4 MuTect2".

Tumor-Only Variant Call Command-Line Parameters

```
GATK4 v4

## 1. Generate OXOG metrics:

java -d64 -XX:+UseSerialGC -Xmx3G -jar /gatk/gatk.jar \
CollectSequencingArtifactMetrics \
-I Tumor_Sample_Alignment.bam \
-O <job_identifier> \
--FILE_EXTENSION .txt \
-R GRCh38.d1.vd1.fa  ## Only chr1-22 + XYM

## 2. Generate pileup summaries on tumor sample:

java -d64 -XX:+UseSerialGC -Xmx3G -jar /gatk/gatk.jar \
GetPileupSummaries
-I Tumor_Sample_Alignment.bam \
-O <job_identifier>.targeted_sequencing.table \
-V af-only-gnomad-common-biallelic.grch38.main.vcf.gz \ # Germline reference from gnomad
-L intervals.bed \ ## Only chr1-22 + XYM
-R GRCh38.d1.vd1.fa

## 3. Calculate contamination on tumor sample

java -d64 -XX:+UseSerialGC -Xmx3G -jar /gatk/gatk.jar \
CalculateContamination \
-I <job_identifier>.targeted_sequencing.table \ # From step 2
-O <job_identifier>.targeted_sequencing.contamination.table

## 4. Find tumor sample name from BAM

java -d64 -XX:+UseSerialGC -Xmx3G -jar /gatk/gatk.jar \
GetSampleName \
-I Tumor_Sample_Alignment.bam \
-O <job_identifier>.targeted_sequencing.sample_name

## 5. Run MuTect2 using only tumor sample on chromosome level (25 commands with different intervals)

java -Djava.io.tmpdir=/tmp/job_tmp_3 -d64 -jar -Xmx3G -XX:+UseSerialGC \
/bin/gatk-4.2.4.0/gatk-package-4.2.4.0-local.jar \
Mutect2 \
-R GRCh38.d1.vd1.fa \
-L chr4:1-190214555 \ # Specify chromosome
-I Tumor_Sample_Alignment.bam \
-O 3.mt2.vcf \
-tumor <tumor_sample_name> \ # From step 4
--af-of-alleles-not-in-resource 2.5e-06 \
--germline-resource af-only-gnomad.hg38.vcf.gz \ # Germline reference from gnomad
-pon gatk4_mutect2_4136_pon.vcf.gz # New panel of normal created by 4136 TCGA curated normal samples, using GATK4

## After this step, all chromosome level VCFs are merged into one.

## 6. Sort VCF with Picard

java -d64 -XX:+UseSerialGC -Xmx16G -jar /usr/local/bin/picard.jar \
SortVcf \
SEQUENCE_DICTIONARY=GRCh38.d1.vd1.dict \
OUTPUT=<job_identifier>.targeted_sequencing.mutect2.tumor_only.sorted.vcf.gz \
I=merged_multi_gatk4_mutect2_tumor_only_calling.vcf \ # From step 5
CREATE_INDEX=true

## 7. Filter variant calls from MuTect
java -d64 -XX:+UseSerialGC -Xmx3G -jar /gatk/gatk.jar \
FilterMutectCalls \
-O <job_identifier>.targeted_sequencing.mutect2.tumor_only.contFiltered.vcf.gz \
-V <job_identifier>.targeted_sequencing.mutect2.tumor_only.sorted.vcf.gz \ # From step 6
--contamination-table <job_identifier>.targeted_sequencing.contamination.table \ # From step 3
```

```
-L intervals.bed

## 8. Filter variants by orientation bias
java -d64 -XX:+UseSerialGC -Xmx3G -jar /gatk/gatk.jar \
FilterByOrientationBias \
-O <job_identifier>.targeted_sequencing.tumor_only.gatk4_mutect2.raw_somatic_mutation.vcf.gz \ # final output
-P <job_identifier>.pre_adapter_detail_metrics.txt \ # From step 1
-V <job_identifier>.targeted_sequencing.mutect2.tumor_only.contFiltered.vcf.gz \ # From step 7
-L intervals.bed \
-R GRCh38.d1.vd1.fa \
-AM G/T \
-AM C/T
```

Tumor-Only Variant Annotation Workflow

After single-tumor variant calling is performed with MuTect2, a series of filters are applied to minimize the release of germline variants in downloadable VCFs. In all cases, the GDC applies a set of custom filters based on allele frequency, mapping quality, somatic/germline probability, and copy number. In some cases an additional variant classification step is applied before the GDC filters.

The PureCN R-package [7] [8] is used to classify the variants by somatic/germline status and clonality based on tumor purity, ploidy, contamination, copy number, and loss of heterozygosity. The following steps are performed with this package:

- **Interval Capture** : Generates an interval file using a FASTA and BED file coordinates.
- **GC-Normalization** : Calculates GC-normalized tumor/normal coverage data.
- **Normal DB Creation** : Generates a normal database using the normalized coverage file and panel-of-normals VCF
- **Somatic Variant Calling** : Classifies each of the previously called variants

Note that PureCN will not be performed if there is insufficient data to produce a target capture kit specific normal database. In rare occasions, PureCN may not find a numeric solution. If PureCN is not performed or does not find a solution, this is indicated in the VCF header. VCF files that were annotated with these pipelines can be found in the GDC Portal by filtering for "Workflow Type: GATK4 MuTect2 Annotation".

Somatic Aggregation Workflow

The Somatic Aggregation Workflow generates one MAF file from multiple VCF files; see the GDC MAF Format guide for details on file structure. In this step, one MAF file is generated per variant calling pipeline for each project and contains all available cases within this project.

I/O	Entity	Format
Input	Multiple Annotated Somatic Mutation	VCF
Output	Aggregated Somatic Mutation	MAF

Masked Somatic Aggregation Workflow

The MAF files generated by Somatic Aggregation Workflow are controlled-access due to the presence of germline mutations. Open-access MAF files are modified for public release by removing columns and variants that could potentially contain germline mutation information. See the GDC MAF Format for details about the criteria used to remove variants.

While these criteria cause the pipeline to over-filter some of the true positive somatic variants in open-access MAF files, they prevent personally identifiable germline mutation information from becoming publicly available. The GDC recommends that investigators explore both controlled and open-access MAF files if omission of certain somatic mutations is a concern.

I/O	Entity	Format
Input	Aggregated Somatic Mutation	Protected MAF
Output	Masked Somatic Mutation	Somatic MAF

Whole Genome Sequencing Variant Calling

Variant calls are generated from WGS data using a different pipeline than WXS and Targeted Sequencing samples. This pipeline, based on a workflow generated by the Sanger Institute, generates multiple downstream data types using the following software packages:

- **CaVEMan:** Single nucleotide variants, which are available in VCF format.
- **Pindel:** Small indel variants, which are available in VCF format.
- **BRASS:** Structural variants, which are available in *BEDPE format*.
- **AscatNGS:** Copy number variants, which are available as copy number estimates or copy number segment files, data may be available in *tab separated values (.TSV) or plain text file (.TXT)*

BEDPE FILE FORMAT

BEDPE file format, (**b**rowser **e**xtensible **d**ata **p**aired-**e**nd) is designed to concisely describe disjoint genome features, such as structural variations or paired-end sequence alignments. It's an enhanced version of the BED format, as BED does not allow inter-chromosomal feature definitions. In addition, BED only has one strand field, which is insufficient for paired-end sequence alignments, especially when studying structural variation. The BEDPE format is described below.

- **chr*x* (required):** The name of the chromosome on which the *x*th end of the feature exists. (x is 1 or 2). Any string can be used. For example, "chr1", "III", "myChrom", "contig1112.23" (use "." for unknown).
- **start*x* (required):** The zero-based starting position of the **first** end of the feature on chr*x*. The first base in a chromosome is numbered 0. The start position in each BEDPE feature is therefore interpreted to be 1 greater than the start position listed in the feature (use -1 for unknown).
- **end*x* (required):** The one-based ending position of the first end of the feature on chr*x*. The end position in each BEDPE feature is one-based (use -1 for unknown).
- **name (optional):** Defines the name of the BEDPE feature. Any string can be used.
- **score (optional):** A score between 0 and 1000. If the track line *useScore* attribute is set to 1 for this annotation data set, the score value will determine the level of gray in which this feature is displayed (higher numbers = darker gray). Any string can be used.
- **strand*x* (optional):** Defines the strand for the *x*th end of the feature. Either "." (unknown), "+", or "-".

In addition to the above fields, bedtools allows for the addition of user-defined fields to the normal, 10-column BEDPE format as necessary. These columns are merely "passed through" pairToBed and pairToPair and are not part of any analysis. One would use these additional columns to add extra information (e.g., edit distance for each end of an alignment, or "deletion", "inversion", etc.) to each BEDPE feature.

CNV FROM WGS FILE FORMAT

AscatNGS, originally developed by Raine *et al* (2016) (GitHub page), indicates the DNA copy number changes affecting a tumor genome when comparing to a matched normal sample. See below for a description of the copy number segment and copy number estimation files produced by AscatNGS:

- **GDC Aliquot:** The GDC ID for the aliquot collected from the sample (copy number segment files only).
- **Gene ID:** The gene ENSMBL ID (copy number variant only).
- **Gene Name:** The gene symbol (copy number variant only).
- **Chromosome:** The name of the chromosome on which the copy number change exists.
- **Start:** The starting position of the copy.
- **End:** The ending position of the copy.
- **Copy Number:** The weighted median of the strand copy numbers [9].
- **Major Copy Number:** The greater strand copy number of the two strands of the DNA (copy number segment files only).
- **Minor Copy number:** The smaller strand copy number of the two strands of the DNA (copy number segment files only).
- **Max. Copy number:** The highest copy number for overlapped segment (copy number variant only).
- **Min. Copy number:** The lowest copy number for overlapped segment (copy number variant only).

1.6.3 Microsatellite Instability Detection

The GDC adopts MSIsensor2 to derive Microsatellite Instability (MSI) information from tumor DNA-Seq data. The MSIsensor2 software uses only the tumor BAM as input, and calculates the numeric MSI score (number of msi sites / all valid sites). The MSI status of MSI (Microsatellite Instable) or MSS (Microsatellite Stable) is then determined using a MSI score cutoff value of 20%.

The output `msi_score` and `msi_status` values are stored directly as properties of the `aligned_reads` (BAM files), and can be accessible via API. In addition, the portal/API can be filtered using these proprties by choosing "Add a Custom Filter" in the Repository Page and selecting `msi_score` or `msi_status`.

Please note:

1. MSI status generated from DNA-Seq by the GDC is considered bioinformatics-derived information, and is not considered clinical data. If performed by the clinical lab, the clinical MSI test result would be stored as a `laboratory_test` in the molecular_test entity.
2. MSIsensor2 can theoretically be applied to WGS, WXS, or Targeted Sequencing data. Given the number of MSI sites available in some Targeted Sequencing data, please consider the results carefully.
3. It is possible that multiple MSI statuses exist within the same sample/case if more than one DNA-Seq BAM was generated. It is the users' responsibility to check for their consistency, especially when the MSI scores are close to 20%.

1.6.4 File Access and Availability

Files from the GDC DNA-Seq analysis pipeline are available in the GDC Data Portal in BAM, VCF, and MAF formats. Descriptions are listed below for all available data types and their respective file formats.

Data Type	Description	File Format
Aligned Reads	Reads that have been aligned to the GRCh38 reference and co-cleaned. Unaligned reads and reads that map to decoy sequences are also included in the BAM files.	BAM
Raw Simple Somatic Mutation	A tab-delimited file with genotypic information related to genomic positions. Genomic variants are first identified here.	VCF
Annotated Somatic Mutation	An annotated version of a raw simple somatic mutation file. Annotated files include biological context about each observed mutation.	VCF
Aggregated Somatic Mutation	A tab-delimited file derived from multiple VCF files. Contains information from all available cases in a project.	MAF
Masked Somatic Mutation	A modified version of the Aggregated Somatic Mutation MAF file with sensitive or potentially erroneous data removed.	MAF

[1]. Li, Heng, and Richard Durbin. "Fast and accurate short read alignment with Burrows-Wheeler transform." Bioinformatics 25, no. 14 (2009): 1754-1760.

[2]. Fan, Yu, Liu Xi, Daniel ST Hughes, Jianjun Zhang, Jianhua Zhang, P. Andrew Futreal, David A. Wheeler, and Wenyi Wang. "Accounting for tumor heterogeneity using a sample-specific error model improves sensitivity and specificity in mutation calling for sequencing data." bioRxiv (2016): 055467.

[3]. Cibulskis, Kristian, Michael S. Lawrence, Scott L. Carter, Andrey Sivachenko, David Jaffe, Carrie Sougnez, Stacey Gabriel, Matthew Meyerson, Eric S. Lander, and Gad Getz. "Sensitive detection of somatic point mutations in impure and heterogeneous cancer samples." Nature biotechnology 31, no. 3 (2013): 213-219.

[4]. Koboldt, Daniel C., Qunyuan Zhang, David E. Larson, Dong Shen, Michael D. McLellan, Ling Lin, Christopher A. Miller, Elaine R. Mardis, Li Ding, and Richard K. Wilson. "VarScan 2: somatic mutation and copy number alteration discovery in cancer by exome sequencing." Genome research 22, no. 3 (2012): 568-576.

[5]. Larson, David E., Christopher C. Harris, Ken Chen, Daniel C. Koboldt, Travis E. Abbott, David J. Dooling, Timothy J. Ley, Elaine R. Mardis, Richard K. Wilson, and Li Ding. "SomaticSniper: identification of somatic point mutations in whole genome sequencing data." Bioinformatics 28, no. 3 (2012): 311-317.

[6]. McLaren, William, Bethan Pritchard, Daniel Rios, Yuan Chen, Paul Flicek, and Fiona Cunningham. "Deriving the consequences of genomic variants with the Ensembl API and SNP Effect Predictor." Bioinformatics 26, no. 16 (2010): 2069-2070.

[7]. Riester, Markus, Angad P. Singh, A. Rose Brannon, Kun Yu, Catarina D. Campbell, Derek Y. Chiang, and Michael P. Morrissey. "PureCN: copy number calling and SNV classification using targeted short read sequencing." Source code for biology and medicine 11, no. 1 (2016): 13.

[8]. Oh, Sehyun, Ludwig Geistlinger, Marcel Ramos, Martin Morgan, Levi Waldron, and Markus Riester. "Reliable analysis of clinical tumor-only whole exome sequencing data" bioRxiv 552711 (2019);

[9]. Gene-level copy number data is generated by intersection of copy number segment and gene ranges. It is possible for one gene to overlap with multiple segments, and in this case, copy_number, min_copy_number and max_copy_number could take different values. In particular, the copy_number value is calculated as the median, weighted on length of overlapped bases, of segment copy numbers from all overlapped segments.

1.7 mRNA Analysis Pipeline

1.7.1 Introduction

The GDC mRNA quantification analysis pipeline measures gene level expression with STAR as raw read counts. Subsequently the counts are augmented with several transformations including Fragments per Kilobase of transcript per Million mapped reads (FPKM), upper quartile normalized FPKM (FPKM-UQ), and Transcripts per Million (TPM). These values are additionally annotated with the gene symbol and gene bio-type. These data are generated through this pipeline by first aligning reads to the GRCh38 reference genome and then by quantifying the mapped reads. To facilitate harmonization across samples, all RNA-Seq reads are treated as unstranded during analyses.

1.7.2 Data Processing Steps

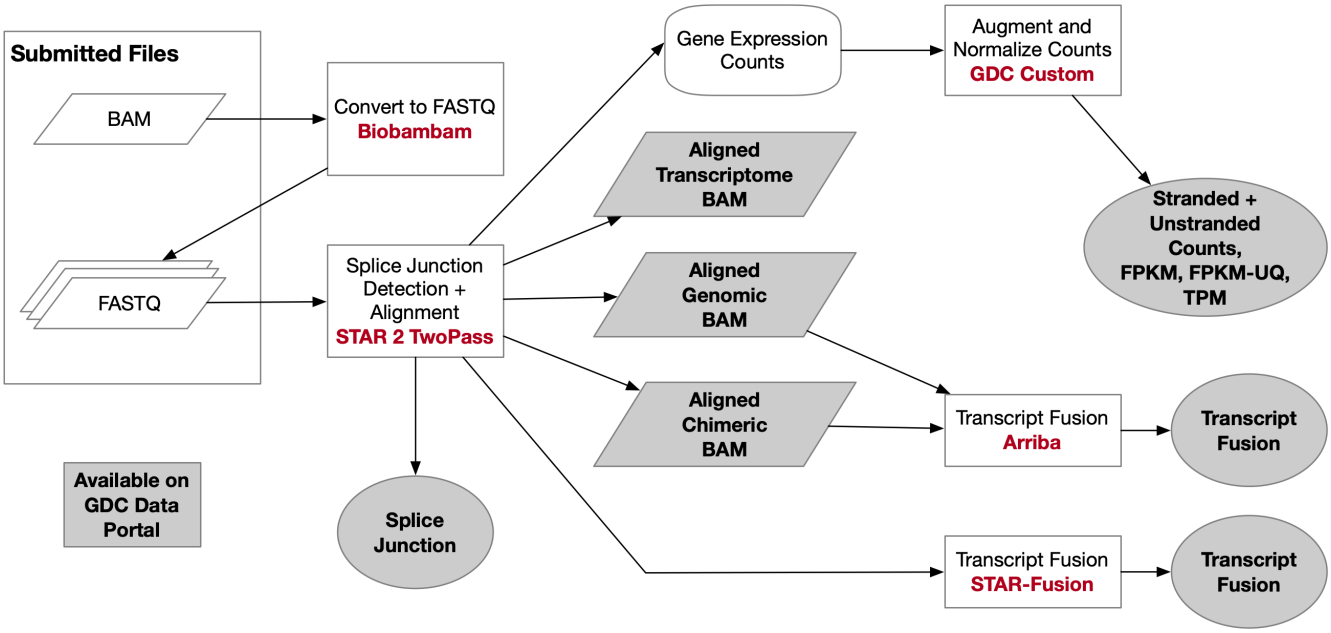
RNA-Seq Alignment Workflow

The mRNA Analysis pipeline begins with the Alignment Workflow, which is performed using a two-pass method with STAR. STAR aligns each read group separately and then merges the resulting alignments into one. Following the methods used by the International Cancer Genome Consortium ICGC (github), the two-pass method includes a splice junction detection step, which is used to generate the final alignment. This workflow outputs a genomic BAM file, which contains both aligned and unaligned reads. Quality assessment is performed pre-alignment with FASTQC and post-alignment with Picard Tools.

Files that were processed after Data Release 14 have associated transcriptomic and chimeric alignments in addition to the genomic alignment detailed above. This only applies to aliquots with at least one set of paired-end reads. The chimeric BAM file contains reads that were mapped to different chromosomes or strands (fusion alignments). The genomic alignment files contain chimeric and unaligned reads to facilitate the retrieval of all original reads. The transcriptomic alignment reports aligned reads with transcript coordinates rather than genomic coordinates. The transcriptomic alignment is also sorted differently to facilitate downstream analyses. BAM index file pairing is not supported by this method of sorting, which does not allow for BAM slicing on these alignments. The splice-junction file for these alignments are also available.

Files that were processed after Data Release 25 will have associated gene fusion files.

As of Data Release 32 the reference annotation will be updated to GENCODE v36 and HT-Seq will no longer be used.



I/O	Entity	Format
Input	Submitted Unaligned Reads or Submitted Aligned Reads	FASTQ or BAM
Output	Aligned Reads	BAM

RNA-Seq Alignment Command Line Parameters

Note that version numbers may vary in files downloaded from the GDC Data Portal due to ongoing pipeline development and improvement.

Original **DR15Plus** **DR32**

```
# STAR-2

### For users with access to the ICGC pipeline:

python star_align.py \
--genomeDir <star_index_path> \
--FastqFileIn <input_fastq_path> \
--workDir <work_dir> \
--out <output_bam> \
--genomeFastaFiles <reference> \
--runThreadN 8 \
--outFilterMultimapScoreRange 1 \
--outFilterMultimapNmax 20 \
--outFilterMismatchNmax 10 \
--alignIntronMax 500000 \
--alignMatesGapMax 1000000 \
--sjdbScore 2 \
--limitBAMsortRAM 0 \
--alignSJDBoverhangMin 1 \
--genomeLoad NoSharedMemory \
--outFilterMatchNminOverLread 0.33 \
--outFilterScoreMinOverLread 0.33 \
--twopass1readsN -1 \
--sjdbOverhang 100 \
--outSAMstrandField intronMotif \
--outSAMunmapped Within

### For users without access to the ICGC pipeline:

### Step 1: Building the STAR index.*

STAR
--runMode genomeGenerate
--genomeDir <star_index_path>
--genomeFastaFiles <reference>
--sjdbOverhang 100
--sjdbGTFfile <gencode.v36.annotation.gtf>
--runThreadN 8

### Step 2: Alignment 1st Pass.

STAR
--genomeDir <star_index_path>
--readFilesIn <fastq_left_1>,<fastq_left2>,... <fastq_right_1>,<fastq_right_2>,...
--runThreadN <runThreadN>
--outFilterMultimapScoreRange 1
--outFilterMultimapNmax 20
--outFilterMismatchNmax 10
--alignIntronMax 500000
--alignMatesGapMax 1000000
--sjdbScore 2
--alignSJDBoverhangMin 1
--genomeLoad NoSharedMemory
--readFilesCommand <bzcat|cat|zcat>
--outFilterMatchNminOverLread 0.33
--outFilterScoreMinOverLread 0.33
--sjdbOverhang 100
--outSAMstrandField intronMotif
--outSAMtype None
--outSAMmode None

### Step 3: Intermediate Index Generation.

STAR
--runMode genomeGenerate
--genomeDir <output_path>
--genomeFastaFiles <reference>
--sjdbOverhang 100
--runThreadN <runThreadN>
--sjdbFileChrStartEnd <SJ.out.tab from previous step>

### Step 4: Alignment 2nd Pass.

STAR
--genomeDir <output_path from previous step>
--readFilesIn <fastq_left_1>,<fastq_left2>,... <fastq_right_1>,<fastq_right_2>,...
--runThreadN <runThreadN>
--outFilterMultimapScoreRange 1
--outFilterMultimapNmax 20
--outFilterMismatchNmax 10
--alignIntronMax 500000
--alignMatesGapMax 1000000
--sjdbScore 2
--alignSJDBoverhangMin 1
--genomeLoad NoSharedMemory
--limitBAMsortRAM 0
--readFilesCommand <bzcat|cat|zcat>
--outFilterMatchNminOverLread 0.33
--outFilterScoreMinOverLread 0.33
--sjdbOverhang 100
--outSAMstrandField intronMotif
--outSAMattributes NH HI NM MD AS XS
--outSAMunmapped Within
--outSAMtype BAM SortedByCoordinate
--outSAMheaderHD @HD VN:1.4
--outSAMattrRGline <formatted RG line provided by wrapper>
```



```
# STAR-2

STAR \
--readFilesIn <fastq_files> \
```

*These indices are available for download at the GDC Website and do not need to be built again.

mRNA Expression Workflow

The primary counting data is generated by STAR and includes a gene ID, unstranded, and stranded counts data. Following alignment, the raw counts files produced by STAR are augmented with commonly used counts transformations (FPKM, FPKM-UQ, and TPM) along with basic annotations as part of the RNA Expression Workflow. These data are provided in a tab-delimited format. GENCODE v36 was used for gene annotation.

Note that the STAR counting results will not count reads that are mapped to more than one different gene. Below are two files that list genes that are completely encompassed by other genes and will likely display a value of zero.

- Overlapped Genes (stranded)
- Overlapped Genes (unstranded)

I/O	Entity	Format
Input	Aligned Reads	BAM
Output	Gene Expression	TXT

mRNA Quantification Command Line Parameters

HTSeq

Current **Original** **DR15-31**

```
Counts are produced by STAR concurrent with alignment.

htseq-count \
-m intersection-nonempty \
-i gene_id \
-r pos \
-s no \
- gencode.v22.annotation.gtf

htseq-count \
-f bam \
-r name \
-s no \
-a 10 \
-t exon \
-i gene_id \
-m intersection-nonempty \
<input_bam> \
<gtf_file> > <counts_file>
```

1.7.3 mRNA Expression Transformation

RNA-Seq expression level read counts produced by the workflow are normalized using three commonly used methods: FPKM, FPKM-UQ, and TPM. Normalized values should be used only within the context of the entire gene set. Users are encouraged to normalize raw read count values if a subset of genes is investigated.

FPKM

The fragments per kilobase of transcript per million mapped reads (FPKM) calculation aims to control for transcript length and overall sequencing quantity.

Upper Quartile FPKM

The upper quartile FPKM (FPKM-UQ) is a modified FPKM calculation in which the protein coding gene in the 75th percentile position is substituted for the sequencing quantity. This is thought to provide a more stable value than including the noisier genes at the extremes.

TPM

The transcripts per million calculation is similar to FPKM, but the difference is that all transcripts are normalized for length first. Then, instead of using the total overall read count as a normalization for size, the sum of the length-normalized transcript values are used as an indicator of size.

Calculations

$$\text{FPKM} = \frac{C_g * 1e^9}{\left(\sum_{i=1}^{N} C_i\right) L_g}$$

$$\text{FPKM-UQ} = \frac{C_g * 1e^9}{C_{qtl(0.75)} * G * L_g}$$

$$\text{TPM} = \frac{\left(C_g * 1e^3 / L_g\right) * 1e^6}{\sum_{g=1}^{N} \left(C_g * 1e^3 / L_g\right)}$$

N = number of protein coding genes
C_g = count of reads aligned to gene g
Lg = union length of exons of gene g
G = number of protein coding genes on autosomes
$C_{qtl(0.75)}$ = count of reads aligned to gene at quantile 0.75

Note: The read count is multiplied by a scalar (10^9) during normalization to account for the kilobase and 'million mapped reads' units.

Examples

Sample 1: Gene A

- Gene length: 3,000 bp
- 1,000 reads mapped to Gene A
- 1,000,000 reads mapped to all protein-coding regions
- Read count in Sample 1 for 75th percentile gene: 2,000
- Number of protein coding genes on autosomes: 19,029
- Sum of length-normalized transcript counts: 9,000,000

FPKM for Gene A = 1,000 * 10^9 / (3,000 * 50,000,000) = **6.67**

FPKM-UQ for Gene A = 1,000) * 10^9 / (3,000 * 2,000 * 19,029) = **8.76**

TPM for Gene A = (1,000 * 1,000 / 3,000) * 1,000,000 / (9,000,000) = **37.04**

1.7.4 Fusion Pipelines

The GDC uses two pipelines for the detection of gene fusions.

STAR-Fusion Pipeline

The GDC gene fusion pipeline uses the STAR-Fusion v1.6 algorithm to generate gene fusion data. STAR-Fusion pipeline processes the output generated by STAR aligner to map junction reads and spanning reads to a junction annotation set. It utilizes a chimeric junction file from running the STAR aligner and produces a tab-limited gene fusion prediction file. The prediction file provides fused gene names, junction read count and breakpoint information.

Arriba Fusion Pipeline

The Arriba gene fusion pipeline uses Arriba v1.1.0 to detect gene fusions from the RNA-Seq data of tumor samples.

1.7.5 scRNA-Seq Pipeline (single-nuclei)

The GDC processes single-cell RNA-Seq (scRNA-Seq) data using the Cell Ranger pipeline to calculate gene expression followed by Seurat for secondary expression analysis.

scRNA Gene Expression Pipeline

The gene expression pipeline, which uses Cell Ranger, generates three files:

- Aligned reads file (BAM)
- Raw counts matrix - contains all barcodes in Market Exchange Format (MEX)
- Filtered counts matrix - contains only detected cellular barcodes (MEX)

scRNA Analysis Pipeline

The analysis pipeline, which uses the Seurat software, generates three files from an input of Filtered counts matrix:

- Analysis - PCA, UMAP, tSNE values, and graph-based clustering results with associated metadata (TSV).
- Differential gene expression - DEG information comparing cells from one cluster to the rest of the cells (TSV).
- Full Seurat analysis log as a loom object in HDF5 format.

When the input RNA was extracted from nuclei instead of cytoplasm, a slightly modified quantification method is implemented to include introns. Currently, these single-nuclei RNA-Seq (snRNA-Seq) analyses share the same experimental strategy (scRNA-Seq) in the Data Portal, and can be filtered by querying for aliquot.analyte_type = "Nuclei RNA".

1.7.6 File Access and Availability

To facilitate the use of harmonized data in user-created pipelines, RNA-Seq gene expression is accessible in the GDC Data Portal at several intermediate steps in the pipeline. Below is a description of each type of file available for download in the GDC Data Portal.

Type	Description	Format
RNA-Seq Alignment	RNA-Seq reads that have been aligned to the GRCh38 build. Reads that were not aligned are included to facilitate the availability of raw read sets.	BAM
STAR Read Counts	The number of reads aligned to each gene, calculated by STAR, along with values using common normalization methods.	TSV

1.8 miRNA Analysis Pipeline

1.8.1 Introduction

The GDC miRNA quantification analysis makes use of a modified version of the profiling pipeline that the British Columbia Genome Sciences Centre developed. The pipeline generates TCGA-formatted miRNAseq data. The first step is read alignment. The tool then compares the individual reads to sequence feature annotations in miRBase v21 and UCSC. Of note, however, the tool only annotates those reads that have an exact match with known miRNAs in miRBase and should therefore not be considered for novel miRNA identification or mismatched alignments.

For more information see BCGSC's GitHub or the original publication.

1.8.2 Data Processing Steps

Alignment Workflow

The miRNA pipeline begins with the Alignment Workflow, which in the case of miRNA uses BWA-aln. This outputs one BAM file for each read group in the input.

I/O	Entity	Format
Input	Submitted Unaligned Reads or Submitted Aligned Reads	FASTQ or BAM
Output	Aligned Reads	BAM

miRNA Expression Workflow

Following alignment, BAM files are processed through the miRNA Expression Workflow.

The outputs of the miRNA profiling pipeline report raw read counts and counts normalized to reads per million mapped reads (RPM) in two separate files mirnas.quantification.txt and isoforms.quantification.txt. The former contains summed expression for all reads aligned to known miRNAs in the miRBase reference. If there are multiple alignments to different miRNAs or different regions of the same miRNA, the read is flagged as cross-mapped and every miRNA annotation is preserved. The latter contains observed isoforms.

I/O	Entity	Format
Input	Aligned Reads	BAM
Output	miRNA Expression	TXT

1.8.3 File Access and Availability

Type	Description	Format
Aligned Reads	miRNA-Seq reads that have been aligned to the GRCh38 build. Reads that were not aligned are included to facilitate the availability of raw read sets.	BAM
miRNA Expression Quantification	A table that associates miRNA IDs with read count and a normalized count in reads-per-million-miRNA-mapped.	TXT
Isoform Expression Quantification	A table with the same information as the miRNA Expression Quantification files with the addition of isoform information such as the coordinates of the isoform and the type of region it constitutes within the full miRNA transcript.	TXT

1.9 Copy Number Variation Analysis Pipeline

1.9.1 Introduction

The copy number variation (CNV) pipeline uses either NGS or Affymetrix SNP 6.0 (SNP6) array data to identify genomic regions that are repeated and infer the copy number of these repeats. Three sets of pipelines have been used for CNV inferences.

- ASCAT
- ABSOLUTE
- DNAcopy

The first set of CNV pipelines are built upon the ASCAT [1] algorithm for both WGS and SNP6 data. ASCAT is able to generate Allele-specific Copy Number Segment data with integer copy number values, and the derived integer Gene-Level Copy Number. 1.) The WGS copy number analysis pipeline, ascatNGS, is described in detail here. 2.) The SNP6 copy number analysis pipeline, ASCAT2, is adopted from the example ASCAT analysis. 3.) The SNP6 copy number analysis pipeline, ASCAT3, is an updated version of ASCAT2. The ASCAT3 analysis in TCGA was done by the Vanloo lab, and the GDC released a reformatted version of these calls. Both ASCAT2 and ASCAT3 generates data similar to ascatNGS.

The second CNV pipeline, ABSOLUTE, also uses Affymetrix SNP 6.0 (SNP6) array data as input. The hg19 version of the segments were published as one of the TCGA PanCancer analysis papers and the data is available in the GDC publication page. These calls have been manually curated and thus are considered of good quality. The GDC performed segment liftover and generated gene-level copy numbers. Note that the intermediate output of GRCh38 segments contain liftover artifacts and were not released in the GDC. Users can also obtain corresponding purity and ploidy measurements from the GDC publication page mentioned above.

The third set of CNV pipelines are built onto the existing TCGA level 2 SNP6 data generated by Birdsuite and uses the DNAcopy R-package to perform a circular binary segmentation (CBS) analysis [2]. CBS translates noisy intensity measurements into chromosomal regions of equal copy number. The final output files are segmented into genomic regions with the estimated copy number for each region. The GDC further transforms these copy number values into segment mean values, which are equal to log2(copy-number/ 2). Diploid regions will have a segment mean of zero, amplified regions will have positive values, and deletions will have negative values.

1.9.2 ASCAT Pipelines

Data Processing Steps

COPY NUMBER SEGMENTATION

The Somatic Copy Number Workflow uses a tumor-normal pair of either SNP6 raw CEL data, or WGS data as input. The ASCAT algorithm derives allele-specific copy number segments while estimating and adjusting for tumor purity and ploidy [1]. Because there are two parental strands, the resulting Copy Number Segment or Allele-Specific Copy Number Segment files contain 3 different copy number integer values: Major_Copy_Number refers to the larger strand copy number, Minor_Copy_Number refers to the smaller strand copy number, Copy_Number is the sum of Major_Copy_Number and Minor_Copy_Number, and thus equals to the total copy number at the locus.

I/O	Entity	Format
Input	Submitted Genotype_Array	CEL
Output	Copy Number Segment or Allele-Specific Copy Number Segment	TXT

I/O	Entity	Format
Input	Aligned Reads	BAM
Output	Copy Number Segment or Allele-Specific Copy Number Segment	TXT

GENE-LEVEL COPY NUMBER

Gene-level Copy Number is generated by inheriting the Copy_Number value of the residing segment in the Copy Number Segment file generated from ASCAT2, ASCAT3, or ascatNGS workflows.

In some occasions, one gene may overlap with more than one segment. In this case, min_copy_number is the minimum value of all segments it overlaps, max_copy_number is the maximum value of all segments it overlaps, and copy_number is calculated as the weighted (on length of overlapped regions) median of copy number values from all overlapped segments. When there is a tie (very rare), the smaller number is used. If a gene overlaps with only one segment, copy_number = min_copy_number = max_copy_number. If a gene overlaps with no segments, the gene gets empty value "" in copy_number, min_copy_number and max_copy_number.

I/O	Entity	Format
Input	Copy Number Segment or Allele-Specific Copy Number Segment	TXT
Output	Copy Number Estimate	TXT

File Access and Availability

Type	Description	Format
Copy Number Segment	A table that associates contiguous chromosomal segments with genomic coordinates, and integer copy numbers.	TXT
Allele-Specific Copy Number Segment	A table that associates contiguous chromosomal segments with genomic coordinates, and integer copy numbers.	TXT
Copy Number Estimate	A Gene-level Copy Number file that displays integer copy number on a gene level. Generated from Copy Number Segment or Allele-Specific Copy Number Segment files.	TXT

1.9.3 ABSOLUTE Copy Number

Data Processing Steps

The source data were generated by external groups. Please check the corresponding publication for details.

File Access and Availability

File Access and Availability is similar to that from the ASCAT pipelines, except that only gene-level copy numbers are available, but not segmentation calls.

1.9.4 DNACopy Pipeline

Data Processing Steps

The GRCh38 SNP6 probe-set was produced by mapping probe sequences to the GRCh38 reference genome and can be downloaded at the GDC Reference File Website.

COPY NUMBER SEGMENTATION

The Copy Number Liftover Workflow uses TCGA level 2 tangent.copynumber files. These files were generated by first normalizing array intensity values, estimating raw copy number, and performing tangent normalization, which subtracts variation that is found in a set of normal samples.

The Copy Number Liftover Workflow performs CBS analysis using the DNACopy R-package to process tangent normalized data into Copy Number Segment files, which associate contiguous chromosome regions with log2 ratio segment means in a tab-delimited format. The number of probes with intensity values associated with each chromosome region is also reported (probes with no intensity values are not included in this count). During copy number segmentation probe sets from Pseudo-Autosomal Regions (PARs) were removed from males and Y chromosome segments were removed from females.

Masked copy number segments are generated using the same method except that a filtering step is performed that removes the Y chromosome and probe sets that were previously indicated to be associated with frequent germline copy-number variation.

I/O	Entity	Format
Input	Submitted Tangent Copy Number	TXT
Output	Copy Number Segment or Masked Copy Number Segment	TXT

[1] Van Loo, P., Nordgard, S. H., Lingjaerde, O. C., Russnes, H. G., Rye, I. H., Sun, W. et al. "Allele-specific copy number analysis of tumors." Proceedings of the National Academy of Sciences, 107.39 (2010): 16910-16915.

[2] Olshen, Adam B., E. S. Venkatraman, Robert Lucito, and Michael Wigler. "Circular binary segmentation for the analysis of array-based DNA copy number data." Biostatistics 5, no. 4 (2004): 557-572.

1.10 Methylation Analysis

1.10.1 Methylation Array Harmonization Workflow

Introduction

The Methylation Array Harmonization Workflow uses raw methylation array data from multiple generations of Illumina Infinium DNA methylation arrays, namely Human Methylation 27 (HM27), HumanMethylation 450 (HM450) and EPIC platforms, to measure the level of methylation at known CpG sites as beta values, calculated from array intensities (Level 2 data) as Beta = M/(M+U). This differs from the Methylation Liftover Pipeline in that the raw methylation array data is used instead of submitted methylation beta values, and the data is processed through the software package SeSAMe[1]. Additionally, the analysis results from the Methylation Array Harmonization Workflow are expected to be of higher quality than results from the Methylation Liftover Pipeline.

SeSAMe offers correction to detection failures that occur in other DNA methylation array software commonly due to germline and somatic deletions by utilizing a novel way to calculate the significance of detected signals in methylation arrays. By correcting for these artifacts as well as other improvements to DNA methylation data processing, SeSAMe improves upon detection calling and quality control of processed DNA methylation data. SeSAMe output files include: two Masked Methylation Array IDAT files, one for each color channel, that contains channel data from a raw methylation array after masking potential genotyping information; and a subsequent Methylation Beta Value TXT file derived from the two Masked Methylation Array IDAT files, that displays the calculated methylation beta value for CpG sites.

SeSAMe Methylation Beta Values File Format

Descriptions for fields present in GDC Harmonized Methylation Beta Values File are detailed below:

Field	Definition
Composite Element	A unique ID for the array probe associated with a CpG site
Beta Value	Represents the ratio between the methylated array intensity and total array intensity, falls between 0 (lower levels of methylation) and 1 (higher levels of methylation)

I/O	Entity	Format
Input	Raw Methylation Array	IDAT
Output	Masked Methylation Array	IDAT
Output	Methylation Beta Values	TXT

1.10.2 Methylation Liftover Pipeline

Note: as of Data Release 32, Methylation Liftover files are no longer supported and do not appear in the GDC Data Portal.

Introduction

The DNA Methylation Liftover Pipeline uses data from the Illumina Infinium Human Methylation 27 (HM27) and HumanMethylation450 (HM450) arrays to measure the level of methylation at known CpG sites as beta values, calculated from array intensities (Level 2 data) as Beta = M/(M+U).

Using probe sequence information provided in the manufacturer's manifest, HM27 and HM450 probes were remapped to the GRCh38 reference genome [2]. Type II probes with a mapping quality of <10, or Type I probes for which the methylated and unmethylated probes map to different locations in the genome, and/or had a mapping quality of <10, had an entry of '*' for the 'chr' field, and '-1' for coordinates. These coordinates were then used to identify the associated transcripts from GENCODE v22, the associated CpG island (CGI), and the CpG sites' distance from each of these features. Multiple transcripts overlapping the

target CpG were separated with semicolons. Beta values were inherited from existing TCGA Level 3 DNA methylation data (hg19-based) based on Probe IDs.

Methylation Liftover Pipeline Table Format

Field	Definition
Composite Element	A unique ID for the array probe associated with a CpG site
Beta Value	Represents the ratio between the methylated array intensity and total array intensity, falls between 0 (lower levels of methylation) and 1 (higher levels of methylation)
Chromosome	The chromosome in which the probe binding site is located
Start	The start of the CpG site on the chromosome
End	The end of the CpG site on the chromosome
Gene Symbol	The symbol for genes associated with the CpG site. Genes that fall within 1,500 bp upstream of the transcription start site (TSS) to the end of the gene body are used.
Gene Type	A general classification for each gene (e.g. protein coding, miRNA, pseudogene)
Transcript ID	Ensembl transcript IDs for each transcript associated with the genes detailed above
Position to TSS	Distance in base pairs from the CpG site to each associated transcript's start site
CGI Coordinate	The start and end coordinates of the CpG island associated with the CpG site
Feature Type	The position of the CpG site in reference to the island: Island, N_Shore or S_Shore (0-2 kb upstream or downstream from CGI), or N_Shelf or S_Shelf (2-4 kbp upstream or downstream from CGI)

I/O	Entity	Format
Input	Submitted Methylation Beta Values	TXT
Output	Methylation Beta Values or Masked Methylation Array	TXT/IDAT

1.10.3 File Access and Availability

Type	Description	Format
Methylation Beta Value	A table that associates array probes with CpG sites and associated metadata.	TXT
Masked Methylation Array	A data file that contains channel data from a raw methylation array after masking of potential genotyping information.	IDAT

[1]. Zhou, Wanding, Triche Timothy J., Laird Peter W. and Shen Hui. "SeSAMe: Reducing artifactual detection of DNA methylation by Infinium BeadChips in genomic deletions." Nucleic Acids Research. (2018): doi: 10.1093/nar/gky691

[2]. Zhou, Wanding, Laird Peter L., and Hui Shen. "Comprehensive characterization, annotation and innovative use of Infinium DNA methylation BeadChip probes." Nucleic Acids Research. (2016): doi: 10.1093/nar/gkw967

1.11 Protein Expression - RPPA

1.11.1 Introduction

Reverse **P**hase **P**rotein **A**rray (RPPA) is a high-throughput antibody-based technique with a procedure similar to that of Western blots. In the procedure carried by MD Anderson Cancer Center, hundreds to thousands of different cell lysates are immobilized on a nitrocellulose-coated slide as many individual spots, followed by incubations with one protein-specific antibody, and detection. A group (often several hundreds) of antibodies form a *set*, which are used for each assay. Occasionally, antibodies may be added to or removed from the set depending on feasibility/functionality, which forms a new set.

To quantify protein expression, a "standard curve" is constructed from spots on each slide (one slide probed for one antibody). These spots include serial dilutions of each sample plus QC spots of standard lysates at different concentrations.

The technique is capable of the following types of analyses:

- Patient tumor classification
- DNA, RNA, and Protein correlation
- Prognosis
- Response prediction for targeted therapies
- Pharmacodynamics and biologically relevant dose
- Determination of appropriate handling procedures for clinical samples (based on antigen stability analysis)

1.11.2 RPPA Data in the GDC

The antigens used for RPPA are available at ref [5], with the following information available:

- `AGID` : The antigen unique ID
- `peptide_target` : The unique ID for the target site that the antigen binds to
- `gene_symbol` : The unique gene name abbreviation that codes the peptide
- `antibody_origin` : The species that the antibody originated from
- `source` : The antibody vendor company
- `catalog_number` : Antibody vendor's catalog number
- `validation_status` : Indicating how trustworthy those antibodies are, based on QC tests of antibody quality by the MD Anderson.

The GDC protein expression quantification data set is available in TSV format and contains `AGID`, `catalog_number`, and `peptide_target` from the reference file, plus the following fields:

- `lab_id` : The unique antibody ID
- `set_id` : The ID for a set, ie list of antibodies (eg refs [3] & [4]).
- `protein_expression` : Relative levels of protein expression - interpolation of each dilution curve to the "standard curve" (supercurve) of the slide (antibody).

1.11.3 References

[1]. https://bioinformatics.mdanderson.org/public-software/tcpa/

[2]. https://www.mdanderson.org/research/research-resources/core-facilities/functional-proteomics-rppa-core/rppa-process.html

[3]. https://www.mdanderson.org/content/dam/mdanderson/documents/core-facilities/Functional%20Proteomics%20RPPA%20Core%20Facility/RPPA_Expanded_Ab_List_Updated.xlsx

[4]. https://www.mdanderson.org/content/dam/mdanderson/documents/core-facilities/Functional%20Proteomics%20RPPA%20Core%20Facility/RPPA_Standard_Ab_List_Updated.xlsx

[5]. https://gdc.cancer.gov/about-data/gdc-data-processing/gdc-reference-files

1.12 Aligned Reads Summary Metrics

Various summary metrics are added to the aligned reads entity for query by the user. These are generated by such tools as SAMtools, Picard, and GATK4. These may be helpful to determine underlying quality or summary information regarding the submitted data. Examples are included below:

- average_base_quality
- average_insert_size
- average_read_length
- contamination
- contamination_error
- mean_coverage
- msi_score
- msi_status
- pairs_on_diff_chr
- proportion_base_mismatch
- proportion_coverage_10x
- proportion_coverage_30x
- proportion_reads_duplicated
- proportion_reads_mapped
- proportion_targets_no_coverage
- total_reads

For a complete list of the summary metrics as well as the tools used to generate them please visit the Data Dictionary Viewer.

1.13 Data Release Notes

Version	Date
v41.0	August 28, 2024
v40.0	March 29, 2024
v39.0	December 4, 2023
v38.0	August 31, 2023
v37.0	March 29, 2023
v36.0	December 12, 2022
v35.0	September 28, 2022
v34.0	July 27, 2022
v33.1	May 31, 2022
v33.0	May 3, 2022
v32.0	March 29, 2022
v31.0	October 29, 2021
v30.0	September 23, 2021
v29.0	March 31, 2021
v28.0	February 2, 2021
v27.0-fix	November 9, 2020
v27.0	October 29, 2020
v26.0	September 8, 2020
v25.0	July 22, 2020
v24.0	May 7, 2020
v23.0	April 7, 2020
v22.0	January 16, 2020
v21.0	December 10, 2019
v20.0	November 11, 2019
v19.1	November 6, 2019
v19.0	September 17, 2019
v18.0	July 8, 2019
v17.1	June 12, 2019
v17.0	June 5, 2019
v16.0	March 26, 2019
v15.0	February 20, 2019
v14.0	December 18, 2018
v13.0	September 27, 2018
v12.0	June 13, 2018
v11.0	May 21, 2018

Version	Date
v10.1	February 15, 2018
v10.0	December 21, 2017
v9.0	October 24, 2017
v8.0	August 22, 2017
v7.0	June 29, 2017
v6.0	May 9, 2017
v5.0	March 16, 2017
v4.0	October 31, 2016
v3.0	September 16, 2016
v2.0	August 9, 2016
v1.0	June 6, 2016

1.13.1 Data Release 41.0

- **GDC Product**: Data
- **Release Date**: August 28, 2024

New Updates

- New Projects
- MATCH-C1
- 11 cases
- WXS, RNA-Seq
- MATCH-P
- 28 cases
- WXS, RNA-Seq
- MATCH-Z1B
- 29 cases
- WXS, RNA-Seq
- New Cases from Existing Projects
- CPTAC-3 - 31 cases

- New Data Sets
- TARGET-AML Tumor-Only Targeted Sequencing - 163 variant call sets
- TCGA U133 Submitted Expression Arrays
- TCGA-GBM - 560 aliquots
- TCGA-LAML - 183 aliquots
- TCGA-LUSC - 135 aliquots
- TCGA-OV - 548 aliquots
- TCGA-LUAD Methylation Data - 53 aliquots
- CDDP_EAGLE-1 Slide Images - 49 cases
- HCMI-CMDC
- Tumor-Only WGS Data - 2 aliquot BAMs, 2 variant call sets
- Tumor-Only WXS Data - 3 aliquot BAMs, 3 variant call sets
- Updated clinical supplements
- BEATAML1.0-COHORT scRNA-Seq Data - 8 aliquots
- Data Updates
- Indexing of ABSOLUTE Liftover copy number variation data
- Release of data for Other Clinical Attribute clinical entities
- `platform` field populated for harmonized data files, can be used as a filter in `Repository`

A complete list of files included in the GDC Data Portal can be found below:

- gdc_manifest_20240826_data_release_41.0_active.tsv.gz
- DR41 Project Level Manifests
- DR41 New Files Manifest

Bugs Fixed Since Last Release

- Fixed 4 TARGET-NBL gene expression sets that pointed to multiple cases/aliquots
- Fixed multiple expression files per aliquot for several TARGET-AML RNA-Seq aliquots

Known Issues and Workarounds

- The slide image viewer does not display for any non-TCGA slides. At this time, these slides will need to be downloaded and viewed locally. Additionally, the slide image viewer does not display properly for 14 TCGA slides, which are identified here.
- Pathology reports do not have any associated case/biospecimen information in the portal. This information can be found in the reports themselves.
- 397 alignments from the TCGA program were found to have contamination values over 0.04 (alignment list). The ensemble MAFs produced by these alignments were removed from the Data Portal.
- One methylation aliquot from the TCGA-COAD project, TCGA-D5-6930-01A-11D-1926-05, was not added to the portal and will be added in a future release.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.

- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- No data from TARGET-MDLS is available.
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Data Portal. These annotations can be found here.
- Tumor_grade property is not populated
- Progression_or_recurrence property is not populated

1.13.2 Data Release 40.0

- **GDC Product**: Data
- **Release Date**: March 29, 2024

New Updates

- New Projects
- MATCH-R - Genomic Characterization CS-MATCH-0007 Arm R - phs002029
- 28 cases
- WXS, RNA-Seq
- MATCH-S1 - Genomic Characterization CS-MATCH-0007 Arm S1 - phs002153
- 41 cases
- WXS, RNA-Seq
- MATCH-S2 - Genomic Characterization CS-MATCH-0007 Arm S2 - phs002178
- 3 cases
- WXS, RNA-Seq
- MATCH-Z1I - Genomic Characterization CS-MATCH-0007 Arm Z1I - phs002058
- 26 cases
- WXS, RNA-Seq
- New Cases from Existing Projects
- CPTAC-3 - 79 cases
- REBC-THYR - 9 cases

- New Data Sets
- Targeted Sequencing
- TARGET-AML - 1,596 aliquot BAMs, 769 variant calls
- TARGET-NBL - 998 aliquot BAMs, 476 variant calls
- TARGET-OS - 233 aliquot BAMs, 65 variant calls
- TCGA WGS
- 57 alignments
- 486 variant call aliquot pairs
- REBC-THYR
- WGS - 90 aliquot BAMs, 69 variant calls
- miRNA-Seq - 177 aliquots
- RNA-Seq - 78 aliquots
- RNA-Seq - Addition of STAR-Fusion data to existing aliquots
- HCMI-CMDC
- Slide images for released cases
- Updated clinical supplements
- TCGA-GBM
- miRNA-Seq - 8 aliquots
- RNA-Seq - 1 aliquot

A complete list of files included in the GDC Data Portal can be found below:

- gdc_manifest_27Mar2024_data_release_40.0_active.tsv.gz
- DR40 Project Level Manifests
- DR40 New Files Manifest

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- The slide image viewer does not display for any non-TCGA slides. At this time, these slides will need to be downloaded and viewed locally. Additionally, the slide image viewer does not display properly for 14 TCGA slides, which are identified here.
- Pathology reports do not have any associated case/biospecimen information in the portal. This information can be found in the reports themselves.
- 397 alignments from the TCGA program were found to have contamination values over 0.04 (alignment list). The ensemble MAFs produced by these alignments were removed from the Data Portal.
- One methylation aliquot from the TCGA-COAD project, TCGA-D5-6930-01A-11D-1926-05, was not added to the portal and will be added in a future release.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.

- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- No data from TARGET-MDLS is available.
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Data Portal. These annotations can be found here.
- Tumor_grade property is not populated
- Progression_or_recurrence property is not populated

1.13.3 Data Release 39.0

- **GDC Product**: Data
- **Release Date**: December 4, 2023

New Updates

- New Projects
- MATCH-H - Genomic Characterization CS-MATCH-0007 Arm H - phs001888
- 21 cases
- WXS, RNA-Seq
- MATCH-I - Genomic Characterization CS-MATCH-0007 Arm I - phs002181
- 60 cases
- WXS, RNA-Seq
- MATCH-U - Genomic Characterization CS-MATCH-0007 Arm U - phs002179
- 23 cases
- WXS, RNA-Seq
- MATCH-W - Genomic Characterization CS-MATCH-0007 Arm W - phs001948
- 45 cases
- WXS, RNA-Seq
- MATCH-Z1A - Genomic Characterization CS-MATCH-0007 Arm Z1A - phs001973
- 45 cases
- WXS, RNA-Seq
- New Cases from Existing Projects
- HCMI-CMDC - 19 cases

- New Data Sets
- 6,957 WGS alignments from the TCGA program
- 1,002 sets of WGS variants from TCGA
- MP2PRT-ALL: WXS and RNA-Seq data
- Tumor-only data produced with a new pipeline. This includes raw and annotated VCFs and MAFs for the following projects. Note that all tumor-only variants are controlled-access:
- BEATAML1.0-COHORT
- BEATAML1.0-CRENOLANIB
- CGCI-BLGSP
- CPTAC-3
- HCMI-CMDC
- MATCH-B
- MATCH-H
- MATCH-I
- MATCH-N
- MATCH-Q
- MATCH-U
- MATCH-W
- MATCH-Y
- MATCH-Z1A
- MATCH-Z1D
- OHSU-CNL
- ORGANOID-PANCREATIC
- TARGET-ALL-P3
- TARGET-WT
- VAREPOP-APOLLO
- New Metadata
- Sample type refactoring:
- Four fields (tissue_type, specimen_type, preservation_method, tumor_descriptor) have been populated to contain the information that was previously populated in the sample_type field
- The new field, specimen_type, is now available in the API to accommodate information about the biological makeup of the sample
- The follow up data for CPTAC-3 has been updated

- Other Updates
- CNV mutations are now available on the exploration page for projects that only had ASCAT CNV data from WGS files. This includes CNV mutations for the following projects:
- APOLLO-LUAD
- CDDP_EAGLE-1
- CGCI-BLGSP
- CGCI-HTMCP-CC
- CGCI-HTMCP-DLBCL
- CGCI-HTMCP-LC
- CPTAC-3
- HCMI-CMDC
- MP2PRT-ALL
- REBC-THYR
- The GENIE program was removed from the GDC Portal because it was not representative of the latest version of GENIE
- GENIE data can be accessed from the AACR Repositories

A complete list of files included in the GDC Data Portal can be found below:

- gdc_manifest_20231204_data_release_39.0_active.tsv.gz
- DR39 Project Level Manifests

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- The slide image viewer does not display for any non-TCGA slides. At this time, these slides will need to be downloaded and viewed locally. Additionally, the slide image viewer does not display properly for 14 TCGA slides, which are identified here.
- Pathology reports do not have any associated case/biospecimen information in the portal. This information can be found in the reports themselves.
- 397 alignments from the TCGA program were found to have contamination values over 0.04 (alignment list). The ensemble MAFs produced by these alignments were removed from the Data Portal.
- One methylation aliquot from the TCGA-COAD project, TCGA-D5-6930-01A-11D-1926-05, was not added to the portal and will be added in a future release.
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.

- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated
- TARGET projects
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the NCI's webpage on Using TARGET Data. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

1.13.4 Data Release 38.0

- **GDC Product**: Data
- **Release Date**: August 31, 2023

New Updates

- New Projects
- MP2PRT-ALL - Molecular Profiling to Predict Response to Treatment for Acute Lymphoblastic Leukemia - phs002005
- 1,507 cases
- WGS
- CGCI-HTMCP-DLBCL - HIV+ Tumor Molecular Characterization Project - Diffuse Large B-Cell Lymphoma - phs000235
- 70 cases
- WGS, RNA-Seq, miRNA-Seq, Tissue Slide Images
- MATCH-B - Genomic Characterization CS-MATCH-0007 Arm B - phs002028
- 33 cases
- WXS, RNA-Seq
- MATCH-N - Genomic Characterization CS-MATCH-0007 Arm N - phs002151
- 21 cases
- WXS, RNA-Seq
- New Cases from Existing Projects
- CPTAC-3 - GBM and Kidney cohorts - 50 cases
- HCMI-CMDC - 31 cases
- CGCI-BLGSP - 204 cases
- TCGA-TGCT - 113 cases
- New Data Sets
- 9,368 WGS alignments from the TCGA program
- 4,676 Cases
- 9,368 Aliquots
- All methylation files that were produced with the SeSAMe pipeline was replaced with a new version.
- TCGA SNP6 data processed with the ASCAT3 and ABSOLUTE pipelines
- 172 CEL and birdseed files from TCGA SNP6
- Release of remaining data for CGCI projects CGCI-BGLSP and CGCI-HTMCP-CC
- New Metadata
- The `wgs_coverage` field is now populated for most BAMs and will allow for WGS BAMs to be queried by coverage range category.
- The QC metrics for applicable BAMs are now queryable through the GDC Data Portal and API.
- The `msi_status` and `msi_score` fields, which were produced using MSISensor2, are now queryable through the GDC Data Portal and API

A complete list of files included in the GDC Data Portal can be found below:

- gdc_manifest_20230830_data_release_38.0_active.tsv.gz
- DR38 Project Level Manifests

Bugs Fixed Since Last Release

- The files produced with the SeSAMe pipeline had unfiltered methylation beta values that should be set as N/A for quality reasons. These files were replaced.
- A bug in which certain files were shown to be associated with more aliquots than usual has been fixed.

Known Issues and Workarounds

- The slide image viewer does not display for any non-TCGA slides. At this time, these slides will need to be downloaded and viewed locally. Additionally, the slide image viewer does not display properly for 14 TCGA slides, which are identified here.
- Pathology reports do not have any associated case/biospecimen information in the portal. This information can be found in the reports themselves.
- 397 alignments from the TCGA program were found to have contamination values over 0.04 (alignment list). The ensemble MAFs produced by these alignments were removed from the Data Portal.
- One methylation aliquot from the TCGA-COAD project, TCGA-D5-6930-01A-11D-1926-05, was not added to the portal and will be added in a future release.
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the NCI's webpage on Using TARGET Data. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

1.13.5 Data Release 37.0

- **GDC Product**: Data
- **Release Date**: March 29, 2023

New Updates

- New Projects
- APOLLO-LUAD - Proteogenomic characterization of lung adenocarcinoma - phs003011
- 87 cases
- WGS, RNA-Seq
- CGCI-HTMCP-LC - HIV+ Tumor Molecular Characterization Project - Lung Cancer - phs000530
- 39 cases
- WGS, RNA-Seq, miRNA-Seq, Slide Images
- MATCH-Q - Genomic Characterization CS-MATCH-0007 Arm Q - phs001926
- 35 cases
- WXS, RNA-Seq
- MATCH-Y - Genomic Characterization CS-MATCH-0007 Arm Y - phs001904
- 31 cases
- WXS, RNA-Seq
- New Data from Existing Projects
- CPTAC-3 - 139 new cases and two new snRNA-Seq samples
- HCMI-CMDC - 118 new cases
- TCGA-THCA - 941 new WGS alignments
- TARGET-OS and TARGET-ALL-P2 - Masked Somatic Mutation MAFs are now open access and their mutations now appear in the exploration portal.

- Data Migrated from the Legacy Archive to Active Portal
- Birdseed files that were generated from Affymetrix SNP6 arrays
- Additional WGS Alignments are now available for TCGA projects
- Additional samples from RNA-Seq and WXS are now available for TCGA projects

A complete list of files included in the GDC Data Portal can be found below:

- gdc_manifest_20230329_data_release_37.0_active.tsv.gz
- DR37 Project Level Manifests

Unavailable Files

- 56 CPTAC-3 snRNA-Seq files are currently unavailable for download. A list of the affected files can be found here. These files will be restored for download by the next data release.

Bugs Fixed Since Last Release

- Outcome data for the CPTAC program has been updated.
- The `age_at_index` field was incorrectly reported in days in the GENIE program. These values have been removed as it contained the same information as the `days_to_birth` field.

Known Issues and Workarounds

- The current files produced with the SeSAMe pipeline have unfiltered methylation beta values that should be set as N/A for quality reasons. These files will be replaced in a future release.
- Pathology reports do not have any associated case/biospecimen information in the portal. This information can be found in the reports themselves.
- 397 alignments from the TCGA program were found to have contamination values over 0.04 (alignment list). The ensemble MAFs produced by these alignments were removed from the Data Portal.
- One methylation aliquot from the TCGA-COAD project, TCGA-D5-6930-01A-11D-1926-05, was not added to the portal and will be added in a future release.
- The clinical supplement for TARGET-ALL-P1 is not currently available. It will be made available in a future release.
- The slide image viewer does not display properly for 14 slides, which are identified here. The full slide image can be downloaded as an SVS file.
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.

- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated
- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.6 Data Release 36.0

- **GDC Product**: Data
- **Release Date**: December 12, 2022

New Updates

- New Projects
- MATCH-Z1D - Genomic Characterization CS-MATCH-0007 Arm Z1D - phs001859
- 36 cases
- WXS, RNA-Seq
- CDDP_EAGLE-1 - CDDP Integrative Analysis of Lung Adenocarcinoma (Phase 2) - phs001239
- 50 cases
- WXS, WGS, RNA-Seq
- New Data from Existing Projects
- CMI-MPC - new RNA-Seq and WXS data
- Data Migrated from the Legacy Archive to Active Portal
- WGS Alignments are now available for 25 TCGA Projects
- Pathology reports from TCGA
- Affymetrix SNP6 Genotyping Array CEL files
- A set of WXS and RNA-Seq samples from TCGA and TARGET that failed harmonization at launch have been rerun and are now available in the active portal.
- TCGA Bisulfite-Seq files can be downloaded using the following manifests:
- TARGET-RT
- TCGA-BLCA
- TCGA-BRCA
- TCGA-COAD
- TCGA-GBM
- TCGA-LUAD
- TCGA-LUSC
- TCGA-READ
- TCGA-STAD
- TCGA-UCEC

A complete list of files included in the GDC Data Portal can be found below:

- gdc_manifest_20221212_data_release_36.0_active.tsv.gz

Unavailable Files

- None

Bugs Fixed Since Last Release

- The copy number variation data is now available on the GDC Exploration portal.
- The mutations on GDC Exploration were re-built with the correct gene model.

Known Issues and Workarounds

- Outcome data for the CPTAC program is not up-to-date. Please visit the Proteomic Data Commons for updated outcome data for CPTAC.
- Pathology reports do not have any associated case/biospecimen information in the portal. This information can be found in the reports themselves.
- 397 alignments from the TCGA program were found to have contamination values over 0.04 (alignment list). The ensemble MAFs produced by these alignments were removed from the Data Portal.
- One methylation aliquot from the TCGA-COAD project, TCGA-D5-6930-01A-11D-1926-05, was not added to the portal and will be added in a future release.
- The clinical supplement for TARGET-ALL-P1 is not currently available. It will be made available in a future release.
- The slide image viewer does not display properly for 14 slides, which are identified here. The full slide image can be downloaded as an SVS file.
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg

- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated
- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.7 Data Release 35.0

- **GDC Product**: Data
- **Release Date**: September 28, 2022

New Updates

- The SomaticSniper variant calling pipeline was deprecated. To support this, the following changes were made:
- All SomaticSniper files no longer appear in the portal, but still can be downloaded using the Data Transfer Tool or API using the original UUID.
- The aggregated somatic mutation and masked somatic mutation files (multi-caller MAFs) have been replaced to reflect the absence of variants from the SomaticSniper pipeline.
- The mutations on the exploration portal reflect the above-mentioned masked somatic mutation files.
- 10 snRNA-Seq samples were released from the CPTAC-3 project.
- Additional RNA-Seq samples from 2,082 additional cases are now available for the TARGET-AML project.
- Demographic data has been added for 94 cases in TARGET-ALL-P2 and TARGET-ALL-P3 projects. A list of the updated cases can be found here.

A complete list of files included in the GDC Data Portal can be found below:

- gdc_manifest_20220928_data_release_35.0_active.tsv.gz

Unavailable Files

- None

Bugs Fixed Since Last Release

- Data from two HCMI-CMDC aliquots (HCM-BROD-0100-C15-85A-01D-A786-36 and HCM-BROD-0679-C43-85M-01D-A80U-36) were incorrectly selected for inclusion into the Exploration Page in Data Release 32 and has been replaced with the correct aliquots (HCM-BROD-0100-C15-01A-11D-A786-36 and HCM-BROD-0679-C43-06A-11D-A80U-36).

Known Issues and Workarounds

- The mutations on GDC Exploration were built with an incorrect gene model.
- The mutations are still correct in terms of the gene affected, coordinates, DNA changes, amino acid changes, and impact.
- Mutations associated with genes that were present in GENCODE v36 and not GENCODE v22 are not displayed. This affects less than 1% of mutations.
- Files downloaded from the the GDC Repository are not affected by this issue. This only affects mutations that are downloaded from GDC Exploration.
- Pathology reports do not have any associated case/biospecimen information in the portal. This information can be found in the reports themselves.

- 397 alignments from the TCGA program were found to have contamination values over 0.04 (alignment list). The ensemble MAFs produced by these alignments were removed from the Data Portal.
- One methylation aliquot from the TCGA-COAD project, TCGA-D5-6930-01A-11D-1926-05, was not added to the portal and will be added in a future release.
- The clinical supplement for TARGET-ALL-P1 is not currently available. It will be made available in a future release.
- Copy number variations currently do not appear in the Exploration page. This will be restored in a future release.
- Mutations from SomaticSniper were erroneously labelled as LOH (loss of heterozygosity). This affects the VCF files, MAF files, and may cause SomaticSniper mutations to be absent from ensemble MAFs.
- The slide image viewer does not display properly for 14 slides, which are identified here. The full slide image can be downloaded as an SVS file.
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.
- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.8 Data Release 34.0

- **GDC Product**: Data
- **Release Date**: July 27, 2022

New updates

- 251 cases from the CPTAC-3 project were added to the portal. This includes all files associated with these cases.

- 243 cases from the BEATAML1.0-COHORT project were added to the portal. This includes most of the files associated with these cases.
- The raw tumor-only VCFs from BEATAML1.0-COHORT are downloadable from the BEATAML1.0-COHORT (2022) publication page here and will be added to the Data Portal in a future release.
- WXS mutations from the BEATAML1.0-COHORT project are now available in the Exploration portal.
- Transcript fusion files are now available for the following projects:
- BEATAML1.0-COHORT
- CMI-ASC
- CMI-MBC
- CPTAC-2
- CTSP-DLBCL1
- MMRF-COMMPASS
- NCICCR-DLBCL
- OHSU-CNL
- ORGANOID-PANCREATIC
- WCDT-MCRPC

A complete list of files included in the GDC Data Portal can be found below:

- gdc_manifest_20220727_data_release_34.0_active.tsv.gz

Unavailable Files

- None

Bugs Fixed Since Last Release

- Data from two HCMI-CMDC aliquots (HCM-BROD-0100-C15-85A-01D-A786-36 and HCM-BROD-0679-C43-85M-01D-A80U-36) were incorrectly selected for inclusion into the Exploration Page in Data Release 32 and has been replaced with the correct aliquots (HCM-BROD-0100-C15-01A-11D-A786-36 and HCM-BROD-0679-C43-06A-11D-A80U-36).

Known Issues and Workarounds

- Pathology reports do not have any associated case/biospecimen information in the portal. This information can be found in the reports themselves.
- 397 alignments from the TCGA program were found to have contamination values over 0.04 (alignment list). The ensemble MAFs produced by these alignments were removed from the Data Portal.
- One methylation aliquot from the TCGA-COAD project, TCGA-D5-6930-01A-11D-1926-05, was not added to the portal and will be added in a future release.
- The clinical supplement for TARGET-ALL-P1 is not currently available. It will be made available in a future release.
- Copy number variations currently do not appear in the Exploration page. This will be restored in a future release.
- Mutations from SomaticSniper were erroneously labelled as LOH (loss of heterozygosity). This affects the VCF files, MAF files, and may cause SomaticSniper mutations to be absent from ensemble MAFs.
- The slide image viewer does not display properly for 14 slides, which are identified here. The full slide image can be downloaded as an SVS file.
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"

- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.
- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.9 Data Release 33.1

- **GDC Product**: Data
- **Release Date**: May 31, 2022

New updates

- None, see "Bugs Fixed Since Last Release" section below.

A complete list of files included in the GDC Data Portal can be found below:

- gdc_manifest_20220531_data_release_33.1_active.tsv.gz

Unavailable Files

- None

Bugs Fixed Since Last Release

- 32 cases from the EXCEPTIONAL_RESPONDERS-ER project were released as they were missing from the previous release.
- All mutations from EXCEPTIONAL_RESPONDERS-ER in the exploration portal come from WXS data, whereas they were previously a mixture of WXS and Targeted Sequencing.

Known Issues and Workarounds

- Pathology reports do not have any associated case/biospecimen information in the portal. This information can be found in the reports themselves.
- 397 alignments from the TCGA program were found to have contamination values over 0.04 (alignment list). The ensemble MAFs produced by these alignments were removed from the Data Portal.
- One methylation aliquot from the TCGA-COAD project, TCGA-D5-6930-01A-11D-1926-05, was not added to the portal and will be added in a future release.
- The clinical supplement for TARGET-ALL-P1 is not currently available. It will be made available in a future release.
- Copy number variations currently do not appear in the Exploration page. This will be restored in a future release.
- Mutations from SomaticSniper were erroneously labelled as LOH (loss of heterozygosity). This affects the VCF files, MAF files, and may cause SomaticSniper mutations to be absent from ensemble MAFs.
- The slide image viewer does not display properly for 14 slides, which are identified here. The full slide image can be downloaded as an SVS file.
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg

- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated
- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.10 Data Release 33.0

- **GDC Product**: Data
- **Release Date**: May 3, 2022

New updates

1. New Project: NCI Exceptional Responders Initiative (EXCEPTIONAL_RESPONDERS-ER, phs001145)
- RNA-Seq - 45 Cases
- WXS - 50 Cases
- Targeted Sequencing - 41 Cases
- Mutations from WXS and Targeted Sequencing are present in the exploration page.
2. New Project: Molecular Profiling to Predict Response to Treatment - Wilms Tumor (MP2PRT-WT, phs001965)
- WGS - 52 Cases
- RNA-Seq - 52 Cases
- miRNA-Seq - 52 Cases
3. Methylation files from the SeSAMe pipeline are now available for CGCI-HTMCP-CC and the TARGET projects.

A complete list of files for this release are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20220503_data_release_33.0_active.tsv.gz
- gdc_manifest_20220503_data_release_33.0_legacy.tsv.gz

Unavailable Files

- The Arriba pipeline failed for one aliquot from EXCEPTIONAL-RESPONDERS-ER and is documented here.

Bugs Fixed Since Last Release

- Gene-level copy number files from TCGA-THCA and TCGA-UCEC were set as controlled-access files. These have been corrected to be available as open-access files.
- Due to a problem with the columns generated by the pipeline, all scRNA-Seq files have been replaced with a new version.

Known Issues and Workarounds

- 397 alignments from the TCGA program were found to have contamination values over 0.04 (alignment list). The ensemble MAFs produced by these alignments were removed from the Data Portal.
- One methylation aliquot from the TCGA-COAD project, TCGA-D5-6930-01A-11D-1926-05, was not added to the portal and will be added in a future release.
- The clinical supplement for TARGET-ALL-P1 is not currently available. It will be made available in a future release.

- Copy number variations currently do not appear in the Exploration page. This will be restored in a future release.
- Mutations from SomaticSniper were erroneously labelled as LOH (loss of heterozygosity). This affects the VCF files, MAF files, and may cause SomaticSniper mutations to be absent from ensemble MAFs.
- The slide image viewer does not display properly for 14 slides, which are identified here. The full slide image can be downloaded as an SVS file.
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
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- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
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- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.
- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.11 Data Release 32.0

- **GDC Product**: Data - GENCODE v36 Release
- **Release Date**: March 29, 2022

New updates

NEW DATA FILES

1. The following data types have been replaced with new GENCODE v36 versions
   - RNA-Seq: all files, including alignments, gene expression files, and transcript fusion files.
   - WXS and Targeted Sequencing: annotated VCFs, single-caller MAFs, Ensemble MAFs.
   - WGS: BEDPE-format structural variants and gene-level copy number variants.
   - GENIE Targeted Sequencing files.
   - FM-AD Targeted Sequencing files.
   - The primary-site-level FM-AD MAF files have been replaced with aliquot-level MAF files.
2. RNA-Seq STAR-Counts files now contain additional normalized counts such as FPKM, FPKM-UQ, and TPM.
3. All WXS files for TCGA have been replaced with new versions. Alignments will contain QC metrics and variants were produced using the same pipelines as all other GDC projects.
4. TCGA RNA-Seq has been changed to contain three alignments (genomic, transcriptome, and chimeric), STAR-counts files, and transcript fusion files for each aliquot.
5. The project-level MAFs in TCGA and FM-AD have been replaced with aliquot-level MAFs.
6. GENCODE v22 derived files (not BAM) that no longer appear in the portal will be downloadable as previous versions of v36 files.
7. Methylation data produced from the SeSAMe pipeline is now available for all TCGA projects.
8. Note that miRNA-Seq data remains unchanged. The miRNA-Seq pipeline uses the miRBase database, which is not affected by the GENCODE version change.
9. A set of manifests were generated at the project-level that map each v22 file to its corresponding v36 file. These can be used to help users transition from v22 to v36 and can be downloaded here.

REMOVED DATA FILES AND PIPELINES

1. Files from the HTSeq pipeline are no longer supported and will no longer appear in the portal. Normalized counts can now be found in the STAR-Counts files.
2. Files that originated from the methylation liftover pipeline are no longer supported and will no longer appear in the portal.
3. GENCODE v22 BAM files that no longer appear in the portal will be available for six months past this release. They may not be available after that.
4. New variant calling tumor-normal pairing was implemented in TCGA, which results in certain aliquots no longer being available as a v36 version (see the aliquots labeled "Unpaired Aliquots" here).
5. Some aliquots failed harmonization when the new v36 gene model was used, which results in some new versions no longer being available (see the aliquots labeled "Failed Harmonization" here).
6. Some aliquots were found to contain a cross-patient contamination level of over 0.04 as measured by GATK4 CalculateContamination (see the aliquots labeled "Contamination" here).

DATA PORTAL EXPLORATION DATA

1. The Data Portal Exploration Page is now populated based on open-access mutations from analyses that used GENCODE v36.
2. Mutations from SomaticSniper will not appear on the Exploration page.
3. Due to the copy number variation pipeline transition from GISTIC to ASCAT, the CNV data was not included in the GDC Exploration page. This will be replaced in a future release once visualization of the new pipeline is fully assessed.

4. The TCGA program mutations have been processed using the same pipeline as all other projects, which resulted in a 26% reduction in the number of open-access mutations. Some points on this change are listed below with TCGA-BRCA as the benchmark project:

- 97% of the previously released open-access mutations are still discoverable in the new GDC controlled-access MAFs. This number increases to 99.95% when focusing only on mutations that were also called by MC3.
- Somatic mutations will now be removed from the Data Portal Exploration Page unless they are detected by more than one variant calling software. This accounts for 40% of the total reduction.
- Somatic mutations will now be removed from the Data Portal Exploration Page if they are detected outside of the target capture region, while previously out-of-target mutations detected from the TCGA Gene Annotation File (GAF) regions were allowed. This accounts for 36% of the total reduction.
- Some TCGA-specific variant-rescue steps have been removed in favor of a more robust and uniform filtering pipeline.
- Some other minor changes due to updates in the gene model or other databases (e.g., the ExAC germline variant database was replaced with gnomAD in DR32).

A complete list of files for this release are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20220316_data_release_32.0_active.tsv.gz
- gdc_manifest_20220316_data_release_32.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- 397 alignments from the TCGA program were found to have contamination values over 0.04 (alignment list). The ensemble MAFs produced by these alignments were removed from the Data Portal.
- One methylation aliquot from the TCGA-COAD project, TCGA-D5-6930-01A-11D-1926-05, was not added to the portal and will be added in a future release.
- The clinical supplement for TARGET-ALL-P1 is not currently available. It will be made available in a future release.
- Copy number variations currently do not appear in the Exploration page. This will be restored in a future release.
- Mutations from SomaticSniper were erroneously labelled as LOH (loss of heterozygosity). This affects the VCF files, MAF files, and may cause SomaticSniper mutations to be absent from ensemble MAFs.
- The slide image viewer does not display properly for 14 slides, which are identified here. The full slide image can be downloaded as an SVS file.
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
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- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.

- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated
- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.12 Data Release 31.0

- **GDC Product**: Data
- **Release Date**: October 29, 2021

New updates

1. TCGA Slide Images:
- All TCGA slide images that were removed earlier this year have been restored.
- Note that the UUIDs for most TCGA slide images have changed. Older manifest files may not work when downloading slide images.
2. CPTAC-3 clinical data has been refreshed and includes new follow up entities.
3. REBC-THYR
- The clinical and biospecimen XML files were removed as they were not intended for release in DR 30.
- The case REBC-ADL5 was added, which includes one WGS pair.

A complete list of files for this release are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20211029_data_release_31.0_active.tsv.gz
- gdc_manifest_20211029_data_release_31.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- One file from a previous version of the methylation pipeline appeared in the data portal (bd2f864a-3f00-47b5-815d-bd01ca21ef61; CPTAC-3). This file should no longer appear in the data portal.

Known Issues and Workarounds

- The slide image viewer does not display properly for 14 slides, which are identified here. The full slide image can be downloaded as an SVS file.
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.

- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
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- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated
- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
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- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
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- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
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- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.13 Data Release 30.0

- **GDC Product**: Data
- **Release Date**: September 23, 2021

New updates

1. New Projects:
- TRIO-CRU (phs001163) - Ukrainian National Research Center for Radiation Medicine Trio Study
- WGS Alignments
- REBC-THYR (phs001134) - Comprehensive genomic characterization of radiation-related papillary thyroid cancer in the Ukraine
- miRNA-Seq
- RNA-Seq
- WGS
2. CPTAC Program
- CPTAC-3 methylation data produced from the SeSAMe pipeline is now available.
- CPTAC-2 miRNA-Seq files have been replaced with better quality data.
3. HCMI-CMDC
- 31 New cases have been released to the GDC Data Portal.
- Methylation data produced from the SeSAMe pipeline is now available.
4. TCGA
- Protein expression data (RPPA) is now available for 32 projects.
- RNA-Seq data for TCGA-TGCT was replaced with files from an updated pipeline.
5. TARGET-AML - New RNA-Seq and miRNA-Seq aliquots have been released.

A complete list of files for this release are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20210923_data_release_30.0_active.tsv.gz
- gdc_manifest_20210923_data_release_30.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- One file from a previous version of the methylation pipeline appears in the data portal (bd2f864a-3f00-47b5-815d-bd01ca21ef61; CPTAC-3). This file cannot be downloaded, but may cause bulk downloads to fail. Remove this file from any manifest or cart you plan on downloading.

- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
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- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
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- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
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- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.
- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.14 Data Release 29.0

- **GDC Product**: Data
- **Release Date**: March 31, 2021

New updates

1. Count Me In Program

- Aliquot-level MAFs are now available for projects CMI-ASC, CMI-MBC, and CMI-MPC.
- Somatic mutation are now explorable for projects CMI-ASC, CMI-MBC, and CMI-MPC

2. CPTAC Program

- CPTAC-2 open-access somatic mutations are now browsable through the GDC Exploration Portal.
- MSI data is now browsable through the faceted search for CPTAC-2 and CPTAC-3.

3. HCMI-CMDC - Data files and explorable mutations for 18 new cases are now available.

A complete list of files for this release are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20210331_data_release_29.0_active.tsv.gz
- gdc_manifest_20210331_data_release_29.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- The aggregated and masked MAF files that were missing for seven pancreatic cases in CPTAC-3 have been restored to the data portal.
- The missing RNA-Seq data files for the seven normal pancreatic cases in CPTAC-3 have been restored to the data portal.

Known Issues and Workarounds

- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
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- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.
- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.15 Data Release 28.0

- **GDC Product**: Data
- **Release Date**: February 2, 2021

New updates

a. New Project: CMI-MPC - Count Me In - The Metastatic Prostate Cancer Project

- WXS alignments and variant calls (VCFs) are available.

b. New Data Type: Single nuclei (snRNA-Seq) data is now available for 18 CPTAC-3 cases. See the RNA-Seq documentation for details.

c. CPTAC-3

- Data files for 147 new cases from the pancreatic cohort are now available.
- CPTAC-3 open-access somatic mutations are now browsable through the GDC Exploration Portal.
- RNA-Seq transcript fusion files are now available.
- Targeted Sequencing alignments and raw tumor-only variant calls (VCF) are now available.

d. HCMI-CMDC

- Data files for 22 new cases are now available.
- The HCMI-CMDC open-access somatic mutations have been refreshed on the GDC Exploration Portal to reflect all newly released cases.

A complete list of files for this release are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20210202_data_release_28.0_active.tsv.gz
- gdc_manifest_20210202_data_release_28.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- The aggregated and masked MAF files for seven pancreatic cases in CPTAC-3 do not appear in the Data Portal. See below for download instructions.
- This manifest can be used to download the files.
- To download the raw aggregated MAF files, dbGaP access to CPTAC-3 (phs001287) is required. The masked MAF files are open-access.
- The seven cases are as follows: C3L-04027, C3L-04080, C3N-02585, C3N-02768, C3N-02971, C3N-03754, and C3N-03839. The case the each file is associated with is denoted in the manifest.
- The RNA-Seq data files for the seven normal pancreatic cases in CPTAC-3 do not appear in the Data Portal. See below for download instructions.
- This manifest can be used to download the files.
- To download the alignments or splice-junction files, dbGaP access to CPTAC-3 (phs001287) is required. The other gene expression files are open-access.
- The seven cases are as follows: C3L-03513, C3L-07032, C3L-07033, C3L-07034, C3L-07035, C3L-07036, C3L-07037. The case the each file is associated with is denoted in the manifest.
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
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- TARGET-10-PAPZST-09A-01D
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- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
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- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.16 Data Release 27.0 Bug Fix

- **GDC Product**: Data
- **Release Date**: November 9, 2020

New updates

1. None, see bug fix section below.

A complete list of files for this release are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20201109_data_release_27.0_active.tsv.gz
- gdc_manifest_20201109_data_release_27.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- Some files in projects CGCI-BLGSP, CGCI-HTMCP-CC, and HCMI-CMDC were marked on the portal as controlled-access, when they were supposed to be open-access. These are now downloadable as open-access files.

Known Issues and Workarounds

- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg

- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated
- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.17 Data Release 27.0

- **GDC Product**: Data
- **Release Date**: October 29, 2020

New updates

1. Initial release for the WGS variant calling pipeline. See the documentation on WGS variant calling for more details on the available files. This includes data from the following projects:
   - CGCI-BLGSP
   - CGCI-HTMCP-CC
   - HCMI-CMDC
2. RNA-Seq transcript fusion files are available for the following projects:
   - CGCI-BLGSP
   - CGCI-HTMCP-CC
   - HCMI-CMDC
3. Aliquot level MAFs were released for CGCI-HTMCP-CC Targeted Sequencing variants. Open access MAFs are included.
4. 17 new cases were released for the HCMI-CMDC project. This includes WGS, WXS, and RNA-Seq data.
5. WGS alignments were released for 99 TCGA-LUAD cases (196 files).
6. Therapeutic agents (treatment) and tumor stage (diagnosis) properties were migrated to remove deprecated values and better adhere to a standardized set of values.

A complete list of files for DR27.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20201029_data_release_27.0_active.tsv.gz
- gdc_manifest_20201029_data_release_27.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- Some files in projects CGCI-BLGSP, CGCI-HTMCP-CC, and HCMI-CMDC are marked on the portal as controlled-access. These files are publicly downloadable using the Data Transfer Tool or API. All files from the following data types should be open-access within the previously specified projects: Biospecimen Supplement, Clinical Supplement, Gene Expression Quantification, Masked Somatic Mutation
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"

- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.
- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.18 Data Release 26.0

- **GDC Product**: Data
- **Release Date**: September 8, 2020

New updates

1. New program released:

- Count Me In (CMI)
- CMI-ASC - The Angiosarcoma Project
- RNA-Seq
- WXS
- CMI-MBC - The Metastatic Breast Cancer Project
- RNA-Seq
- WXS

2. Somatic mutations are now available on the exploration portal for the following projects:

- MMRF-COMMPASS
- TARGET-ALL-P3
- TARGET-AML
- TARGET-NBL
- TARGET-WT

3. Primary sites and disease types were updated for multiple projects to correspond to GDC Dictionary updates.

A complete list of files for DR26.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20200908_data_release_26.0_active.tsv.gz
- gdc_manifest_20200908_data_release_26.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- The CPTAC-3 head and neck cohort can now be queried by choosing the head and neck anatomic site on the GDC home page.

Known Issues and Workarounds

- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg

- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated
- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.19 Data Release 25.0

- **GDC Product**: Data
- **Release Date**: July 22, 2020

New updates

a. New data types released:

- RNA-Seq Transcript Fusion files were released for the following projects:
- TARGET-ALL-P1
- TARGET-ALL-P2
- TARGET-ALL-P3
- TARGET-CCSK
- TARGET-NBL
- TARGET-OS
- TARGET-RT
- TARGET-WT
- The msi_status and msi_score properties can be queried on the GDC Portal for the CPTAC-3 project.
- To query for these fields: go to the GDC Repository, click on "Add a File Filter" at the top left of the screen, type msi_score or msi_status in the field, and click on "msi_score" or "msi_status". This should bring up the corresponding filters to use on the portal.

b. 108 cases from the CPTAC-3 LSCC Cohort were released. Includes the following data types:

- WXS
- WGS
- RNA-Seq
- miRNA-Seq

c. Aliquot level MAFs were released for MMRF-COMMPASS WXS variants. Open access MAFs are included.

d. HCMI-CMDC open-access somatic mutations were released to the Exploration Portal.

A complete list of files for DR25.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20200722_data_release_25.0_active.tsv.gz
- gdc_manifest_20200722_data_release_25.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- A few supplements from CGCI-BLGSP are now associated with their correct versions.

Known Issues and Workarounds

- Currently the CPTAC-3 HNSCC cohort does not appear when the "Head and Neck" primary site is selected from the GDC home page. This cohort can be queried by clicking here
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
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- Two tissue slide images are unavailable for download from GDC Data Portal
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- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
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- Issues in the Legacy Archive
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- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.20 Data Release 24.0

- **GDC Product**: Data
- **Release Date**: May 7, 2020

New updates

1. New project released: CGCI-HTMCP-CC - HIV+ Tumor Molecular Characterization Project - Cervical Cancer
- RNA-Seq: Alignments and gene expression levels
- miRNA-Seq: Alignments and miRNA expression levels
- WGS: Alignments
- Targeted Sequencing: Alignments
2. 110 new cases were released from the HNSCC cohort of CPTAC-3. This includes WXS, WGS, RNA-Seq and miRNA-Seq data.
3. Aliquot-level WXS MAFs are now available from the following projects:
- CPTAC-2
- CPTAC-3

A complete list of files for DR24.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20200507_data_release_24.0_active.tsv.gz
- gdc_manifest_20200507_data_release_24.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- Currently the CPTAC-3 HNSCC cohort does not appear when the "Head and Neck" primary site is selected from the GDC home page. This cohort can be queried by clicking here
- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
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- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg

- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
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- Tumor grade property is not populated
- Progression_or_recurrence property is not populated
- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
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- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.21 Data Release 23.0

- **GDC Product**: Data
- **Release Date**: April 7, 2020

New updates

1. New data types released:

- Aliquot-level MAFs: MAF Files with mutations derived from one tumor/normal pair
- HCMI-CMDC
- TARGET-ALL-P2
- TARGET-ALL-P3
- TARGET-AML
- TARGET-NBL
- TARGET-OS
- TARGET-WT
- Note: Previously released TARGET project level MAFs can be downloaded with the following manifest: TARGET_Project-Level-MAF_GDC-Manifest.txt
- Copy number segment and estimate files from SNP6 ASCAT
- All TCGA Projects
- TARGET-ALL-P2
- TARGET-AML

2. To accommodate users who prefer to use project-level MAFs, a MAF aggregation tool was developed by the GDC:

- Github Release

3. New RNA-Seq data was released from HCMI-CMDC for nine additional cases.
4. Clinical updates were performed for the following projects

- CGCI-BLGSP
- HCMI-CMDC
- WCDT-MCRPC

A complete list of files for DR23.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20200407_data_release_23.0_active.tsv.gz
- gdc_manifest_20200407_data_release_23.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- The 6 HCMI-CMDC cases without clinical data now have clinical data.

- Most of the "associated_entities" fields in CGCI-BLGSP were not populated correct, this has been resolved.

Known Issues and Workarounds

- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
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- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.
- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.22 Data Release 22.0

- **GDC Product**: Data
- **Release Date**: January 16, 2020

New updates

1. New projects released:

- WCDT-MCRPC - Genomic Characterization of Metastatic Castration Resistant Prostate Cancer (phs001648)
- RNA-Seq; WGS Data

2. New data from HCMI-CMDC
- 16 New Cases
- Includes WXS, WGS, and RNA-Seq data
3. New data from CPTAC-3
- 108 New Cases
- Includes WXS, WGS, and RNA-Seq data
- miRNA-Seq data for currently released cases

A complete list of files for DR22.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20200116_data_release_22.0_active.tsv.gz
- gdc_manifest_20200116_data_release_22.0_legacy.tsv.gz

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- 6 of the HCMI-CMDC cases are missing clinical nodes
- HCM-CSHL-0060-C18
- HCM-CSHL-0089-C25
- HCM-CSHL-0090-C25
- HCM-CSHL-0092-C25
- HCM-CSHL-0091-C25
- HCM-CSHL-0057-C18
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg

- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated
- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.23 Data Release 21.0

- **GDC Product**: Data
- **Release Date**: December 10, 2019

New updates

1. New projects released:

- GENIE - AACR Project Genomics Evidence Neoplasia Information Exchange (phs001337)
- Includes Targeted Sequencing, Transcript Fusion, Copy Number Estimate from GENIE 5.0
- AACR Project GENIE is divided by sequencing center:
- GENIE-MSK
- GENIE-DFCI
- GENIE-MDA
- GENIE-JHU
- GENIE-UHN
- GENIE-VICC
- GENIE-GRCC
- GENIE-NKI

A complete list of files for DR21.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20191210_data_release_21.0_active.txt.gz
- gdc_manifest_20191210_data_release_21.0_legacy.txt.gz

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- The Copy Number Estimate files in GENIE are labeled on the portal as TXT while the files are actually in TSV format.
- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.

- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
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- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.
- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.24 Data Release 20.0

- **GDC Product**: Data
- **Release Date**: November 11, 2019

New updates

1. New projects released:
- CPTAC-2 - CPTAC Proteogenomic Confirmatory Study (phs000892)
- Includes WXS, RNA-Seq, and miRNA-Seq
- OHSU-CNL - Genomic landscape of Neutrophilic Leukemias of Ambiguous Diagnosis (phs001799)
- Includes WXS and RNA-Seq
- No VCF files will be included at this time. They will follow in a later release.
2. New TARGET data released
- TARGET-OS: WGS, WXS
- TARGET-NBL: WGS
- TARGET-AML: miRNA
3. CGCI-BLGSP miRNA-Seq released

A complete list of files for DR20.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20191111_data_release_20.0_active.txt.gz
- gdc_manifest_20191111_data_release_20.0_legacy.txt.gz

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
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- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg

- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
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- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
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- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
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- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
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- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
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- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.25 Data Release 19.1

- **GDC Product**: Data
- **Release Date**: November 6, 2019

New updates

- The following cases are no longer available in the GDC Data Portal. They had no data files associated with them in DR 19 so there are no changes in file availability in this release.
- TARGET-00-NAAENF
- TARGET-00-NAAENG
- TARGET-00-NAAENH
- TARGET-00-NAAENI
- TARGET-00-NAAENJ
- TARGET-00-NAAENK
- TARGET-00-NAAENL
- TARGET-00-NAAENM
- TARGET-00-NAAENN
- TARGET-00-NAAENP
- TARGET-00-NAAENR
- TARGET-00-NAAEPE

A complete list of files for DR19.1 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20190917_data_release_19.0_active.txt.gz
- gdc_manifest_20190917_data_release_19.0_legacy.txt.gz

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release

- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
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- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
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- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.
- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.26 Data Release 19.0

- **GDC Product**: Data
- **Release Date**: September 17, 2019

New updates

1. New projects released:

- BEATAML1.0-COHORT - Functional Genomic Landscape of Acute Myeloid Leukemia (phs001657)
- Includes WXS and RNA-Seq

2. New TARGET data released
- TARGET-ALL-P1 RNA-Seq
- TARGET-ALL-P2 RNA-Seq, WXS, and miRNA-Seq
- TARGET-ALL-P3 miRNA-Seq
- TARGET-AML WXS, WGS, and miRNA-Seq
- TARGET-NBL WXS and RNA-Seq
- TARGET-RT WGS and RNA-Seq
- TARGET-WT WGS, WXS, and RNA-Seq
3. Additional CGCI-BLGSP WGS data released
4. Pindel VCFs released for TARGET-ALL-P2, TARGET-ALL-P3, TARGET-AML, TARGET-NBL, TARGET-WT, MMRF-COMMPASS, HCMI-CMDC, and CPTAC-3
5. Disease-specific staging properties for many projects were released

A complete list of files for DR19.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20190917_data_release_19.0_active.txt.gz
- gdc_manifest_20190917_data_release_19.0_legacy.txt.gz

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg

- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
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- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
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- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
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- TARGET-30-PAPTLY-01A-01D
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- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.* Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.27 Data Release 18.0

- **GDC Product**: Data
- **Release Date**: July 8, 2019

New updates

1. New Projects released
- MMRF-COMMPASS - Multiple Myeloma CoMMpass Study (phs000748)
- Includes WGS, WXS, and RNA-Seq
- ORGANOID-PANCREATIC - Pancreas Cancer Organoid Profiling (phs001611)
- Includes WGS, WXS, and RNA-Seq
- TARGET-ALL-P1 - Acute Lymphoblastic Leukemia - Phase I (phs000218)
- Includes WGS
- TARGET-ALL-P2 - Acute Lymphoblastic Leukemia - Phase II (phs000218)
- Includes WGS
- CGCI-BLGSP - Burkitt Lymphoma Genome Sequencing Project (phs000235)
- Includes WGS and RNA-Seq
2. New versions of RNA-Seq data for TARGET-ALL-P3
3. New RNA-Seq data for TARGET-CCSK
4. New RNA-Seq data for TARGET-OS

A complete list of files for DR18.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20190708_data_release_18.0_active.txt.gz
- gdc_manifest_20190708_data_release_18.0_legacy.txt.gz

Bugs Fixed Since Last Release

- New versions of RNA-Seq data for TARGET-ALL-P3 resolve issue with missing reads from BAM files.

Known Issues and Workarounds

- Some tumor-only annotated VCFs (not raw VCFs) could have a small proportion of variants that appear twice. Tumor-only annotated VCFs can be identified by searching for workflow "GATK4 MuTect2 Annotation"
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release

- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- TCGA Projects
- Incorrect information about treatment may be included for patients within TCGA-HNSC and TCGA-LGG. Please refer to the clinical XML for accurate information on treatment
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- Two tissue slide images are unavailable for download from GDC Data Portal
- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

- TARGET projects
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
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- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- Some TARGET cases are missing `days_to_last_follow_up`
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- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
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- SDF Files are not linked to Project or Case in the Legacy Archive
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- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.28 Data Release 17.1

- **GDC Product**: Data
- **Release Date**: June 12, 2019

New updates

1. Rebuilt indices for NCICCR-DLBCL and CTSP-DLBCL1. Fewer files viewable in GDC Data Portal or API.

A complete list of files for DR17.1 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20190612_data_release_17.1_active.txt.gz
- gdc_manifest_20190612_data_release_17.1_legacy.txt.gz

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
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- Tumor grade property is not populated
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- TARGET projects
- TARGET ALL-P3 RNA-Seq results from DR14 are missing ~18% of reads. Downsampling appears to be completely random and count files have a very high correlation (>99.99%) with complete data. New versions of these files will be created that include the entire set of reads.
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
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- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
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- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.29 Data Release 17.0

- **GDC Product**: Data
- **Release Date**: June 5, 2019

New updates

1. New Projects released
   - HCMI-CMDC - NCI Cancer Model Development for the Human Cancer Model Initiative (HCMI) (phs001486)
   - BEATAML1.0-CRENOLANIB - Clinical Resistance to Crenolanib in Acute Myeloid Leukemia Due to Diverse Molecular Mechanisms (phs001628)
2. RNA-Seq data for NCICCR-DLBCL and CTSP-DLBCL1 are released
3. ATAC-Seq data for TCGA projects are released
4. CPTAC-3 RNA-Seq data are released
5. Clinical data updates for TCGA - to see parser code updates review API v1.20 release notes
6. Clinical data updates for other projects to accommodate migration of vital_status, days_to_birth, and days_to_death from the Diagnosis to the Demographic node

A complete list of files for DR17.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20190605_data_release_17.0_active.txt.gz
- gdc_manifest_20190605_data_release_17.0_legacy.txt.gz.

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

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- TARGET ALL-P3 RNA-Seq results from DR14 are missing ~18% of reads. Downsampling appears to be completely random and count files have a very high correlation (>99.99%) with complete data. New versions of these files will be created that include the entire set of reads.
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- No data from TARGET-MDLS is available.

- Issues in the Legacy Archive
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- TARGET-MDLS cases do not have disease_type or primary_site populated

1.13.30 Data Release 16.0

- **GDC Product**: Data
- **Release Date**: March 26, 2019

New updates

1. The CPTAC-3 project (phs001287) is released with WXS and WGS data. RNA-Seq will be released at a later date. Additional project details can be found at on the CPTAC Data Source page.
2. TARGET-ALL-P3 (phs000218) WGS BAM files are released.
3. VAREPOP-APOLLO (phs001374) VCF files are released.

A complete list of files for DR16.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20190326_data_release_16.0_active.txt.gz
- gdc_manifest_20190326_data_release_16.0_legacy.txt.gz.

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- TARGET ALL-P3 RNA-Seq results from DR14 are missing ~18% of reads. Downsampling appears to be completely random and count files have a very high correlation (>99.99%) with complete data. New versions of these files will be created that include the entire set of reads.
- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
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- TARGET-20-PASMYS-14A-02D
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- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- Two tissue slide images are unavailable for download from GDC Data Portal

- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- TARGET-MDLS cases do not have disease_type or primary_site populated
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- No data from TARGET-MDLS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

1.13.31 Data Release 15.0

- **GDC Product**: Data
- **Release Date**: February 20, 2019

New updates

1. TARGET-ALL-P3 is now available and includes RNA-Seq and WXS data.

2. New RNA-Seq workflow is now being utilized for new projects. More details can be found in the RNA-Seq pipeline documentation.
3. New tumor only variant calling pipeline is now being utilized for new projects. More details can be found in the Tumor only pipeline documentation.

A complete list of files for DR15.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20190220_data_release_15.0_active.txt.gz
- gdc_manifest_20190220_data_release_15.0_legacy.txt.gz.

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- The read alignment end coordinates in the x.isoform.quantification.txt files produced by the miRNA pipeline are exclusive (i.e. offset by 1) for all TCGA miRNA legacy (GRCh37/hg19) and current harmonized (GRCh38/hg38) miRNA data. This error has no impact on miRNA alignment or quantification - only the coordinates reported in the quantification file.
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- Two tissue slide images are unavailable for download from GDC Data Portal

- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- TARGET-MDLS cases do not have disease_type or primary_site populated
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- No data from TARGET-MDLS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

1.13.32 Data Release 14.0

- **GDC Product**: Data
- **Release Date**: December 18, 2018

New updates

1. Copy Number Variation (CNV) data derived from GISTIC2 results are now available for download for TCGA projects

2. New miRNA data available for 181 aliquots for TARGET and TCGA
3. Released two SNP6 files (6cd4ef5e-324a-4ace-8779-7a33bd559c83, dfa89ee9-6ee5-460b-bd58-b5ca0e9cb7ac)
4. New versions of TCGA biospecimen supplements are available
5. Updated primary site for `TCGA-AG-3881` to `Unknown`
6. 8 New Harmonized WGS BAM files for TARGET-WT, TARGET-NBL, TARGET-AML added to the portal

A complete list of files for DR14.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20181218_data_release_14.0_active.txt.gz
- gdc_manifest_20181218_data_release_14.0_legacy.txt.gz.

Bugs Fixed Since Last Release

- FM-AD clinial and biospecimen supplements are now correctly labeled as TSV rather than XLSX

Known Issues and Workarounds

- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- Two tissue slide images are unavailable for download from GDC Data Portal

- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- TARGET-MDLS cases do not have disease_type or primary_site populated
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- No data from TARGET-MDLS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

1.13.33 Data Release 13.0

- **GDC Product**: Data
- **Release Date**: September 27, 2018

New updates

1. Three new projects are released to the GDC (VAREPOP-APOLLO (phs001374), CTSP-DLBCL1 (phs001184), NCICCR-DLBCL (phs001444)
2. TARGET WGS alignments are released. VCFs will be provided in a later release
3. Clinical data was harmonized with ICD-O-3 terminology for TCGA properties case.primary_site, case.disease_type, diagnosis.primary_diagnosis, diagnosis.site_of_resection_or_biopsy, diagnosis.tissue_or_organ_of_origin
4. Redaction annotations applied to 11 aliquots in TCGA-DLBC
5. Redaction annotations applied to incorrectly trimmed miRNA file in the Legacy Achive

A complete list of files for DR13.0 are listed for the GDC Data Portal and the GDC Legacy Archive are found below:

- gdc_manifest_20180927_data_release_13.0_active.txt.gz
- gdc_manifest_20180927_data_release_13.0_legacy.txt.gz.

Bugs Fixed Since Last Release

- 253 files Copy Number Segment and Masked Copy Number Segment files were released. These were skipped in DR 12.0
- 36 Diagnostic TCGA slides were released. They were skipped in DR 12.0

Known Issues and Workarounds

- 506 Copy Number Segment and 36 Slide Image files are designated as controlled-access on the GDC Data Portal. These files are actually open-access and will be downloadable without a token using this manifest.
- 2 Copy Number Segment files from TCGA-TGCT do not appear on the GDC Portal. They can be downloaded using the Data Transfer Tool using the following UUIDs.
- 6cd4ef5e-324a-4ace-8779-7a33bd559c83 - RAMPS_p_TCGA_Batch_430_NSP_GenomeWideSNP_6_E07_1538238.nocnv_grch38.seg.v2.txt
- dfa89ee9-6ee5-460b-bd58-b5ca0e9cb7ac - RAMPS_p_TCGA_Batch_430_NSP_GenomeWideSNP_6_E07_1538238.grch38.seg.v2.txt
- TARGET CGI BAMs in the Legacy Archive for the following aliquots should not be used because they were not repaired and concatenated into their original composite BAM files by CGHub.
- TARGET-20-PASJGZ-04A-02D
- TARGET-30-PAPTLY-01A-01D
- TARGET-20-PAEIKD-09A-01D
- TARGET-20-PASMYS-14A-02D
- TARGET-20-PAMYAS-14A-02D
- TARGET-10-PAPZST-09A-01D
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- Two tissue slide images are unavailable for download from GDC Data Portal

- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.
- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- TARGET-MDLS cases do not have disease_type or primary_site populated
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- FM-AD clinical and biospecimen supplement files have incorrect data format. They are listed as XLSX, but are in fact TSV files.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- No data from TARGET-MDLS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

1.13.34 Data Release 12.0

- **GDC Product**: Data
- **Release Date**: June 13, 2018

New updates

1. Updated clinical and biospecimen XML files for TCGA cases are available in the GDC Data Portal. Equivalent Legacy Archive files may no longer be up to date.
2. All biospecimen and clinical supplement files for TCGA projects formerly only found in the Legacy Archive have been updated and transferred to the GDC Data Portal. Equivalent Legacy Archive files and metadata retrieved from the API may no longer be up to date.
3. Diagnostic slides from TCGA are now available in the GDC Data Portal and Slide Image Viewer. They were formerly only available in the Legacy Archive.
4. Updated Copy Number Segment and Masked Copy Number Segment files are now available. These were generated using an improved mapping of hg38 coordinates for the Affymetrix SNP6.0 probe set.
5. VCF files containing SNVs produced from TARGET WGS CGI data are available. The variant calls were initially produced by CGI and lifted over to hg38.

Updated files for this release are listed here. A complete list of files for DR12.0 are listed for the GDC Data Portal here and the GDC Legacy Archive here.

Bugs Fixed Since Last Release

- TARGET NBL RNA-Seq data is now associated with the correct aliquot.

Known Issues and Workarounds

- Some Copy Number Segment and Masked Copy Number Segment were not replaced in DR 12.0. 253 files remain to be swapped in a later release
- Some miRNA files with QC failed reads were not swapped in DR11.0. 361 aliquots remain to be swapped in a later release
- 74 Diagnostic TCGA slides are attached to a portion rather than a sample like the rest of the diagnostic slides. The reflects how these original samples were handled.
- 36 Diagnostic TCGA slides are not yet available in the active GDC Portal. They are still available in the GDC Legacy Archive.
- 11 BAM files for TARGET-NBL RNA-Seq are not available in the GDC Data portal
- Two tissue slide images are unavailable for download from GDC Data Portal

- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` are not available. These VCFs files will be replaced in a later release.

- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- TARGET-MDLS cases do not have disease_type or primary_site populated
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- FM-AD clinical and biospecimen supplement files have incorrect data format. They are listed as XLSX, but are in fact TSV files.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- There are 11 cases in project TCGA-DLBC that are known to have incorrect WXS data in the GDC Data Portal. Impacted cases are listed below. This affects the BAMs and VCFs associated with these cases in the GDC Data Portal. Corrected BAMs can be found in the GDC Legacy Archive. Variants from affected aliquots appear in the protected MAFs with GDC_FILTER=ContEst to indicate a sample contamination problem, but are removed during the generation of the Somatic MAF file. In a later release we will supply corrected BAM, VCF, and MAF files for these cases. In the mean time, we advise you not to use any of the WXS files associated with these cases in the GDC Data Portal. A list of these files can be found here. Download list of affected files.
- TCGA-FF-8062
- TCGA-FM-8000
- TCGA-G8-6324
- TCGA-G8-6325
- TCGA-G8-6326
- TCGA-G8-6906
- TCGA-G8-6907
- TCGA-G8-6909
- TCGA-G8-6914
- TCGA-GR-7351
- TCGA-GR-7353
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- No data from TARGET-MDLS is available.

- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

1.13.35 Data Release 11.0

- **GDC Product**: Data
- **Release Date**: May 21, 2018

New updates

1. Updated miRNA files to remove QCFail reads. This included all BAM and downstream count files.
2. TCGA Tissue slide images now available in GDC Data Portal. Previously these were found only in the Legacy Archive

Updated files for this release are listed here. A complete list of files for DR11.0 are listed for the GDC Data Portal here and the GDC Legacy Archive here.

Bugs Fixed Since Last Release

- N/A

Known Issues and Workarounds

- Two tissue slide images are unavailable for download from GDC Data Portal
- RNA-Seq files for TARGET-NBL are attached to the incorrect aliquot. The BAM files contain the correct information in their header but the connection in the GDC to read groups and aliquots is incorrect. The linked file below contains a mapping between aliquots where file are currently associated and the aliquot where they should instead be associated (mapping file).

- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` were not replaced in DR10.0 and thus do not contain indels. However, the indels from this aliquot can be found in the MAF files and are displayed in the Exploration section in the Data Portal. These VCFs files will be replaced in a later release.

- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- TARGET-MDLS cases do not have disease_type or primary_site populated
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- miRNA alignments include QC failed reads.
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- FM-AD clinical and biospecimen supplement files have incorrect data format. They are listed as XLSX, but are in fact TSV files.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- There are 11 cases in project TCGA-DLBC that are known to have incorrect WXS data in the GDC Data Portal. Impacted cases are listed below. This affects the BAMs and VCFs associated with these cases in the GDC Data Portal. Corrected BAMs can be found in the GDC Legacy Archive. Variants from affected aliquots appear in the protected MAFs with GDC_FILTER=ContEst to indicate a sample contamination problem, but are removed during the generation of the Somatic MAF file. In a later release we will supply corrected BAM, VCF, and MAF files for these cases. In the mean time, we advise you not to use any of the WXS files associated with these cases in the GDC Data Portal. A list of these files can be found here. Download list of affected files.
- TCGA-FF-8062
- TCGA-FM-8000
- TCGA-G8-6324
- TCGA-G8-6325
- TCGA-G8-6326
- TCGA-G8-6906
- TCGA-G8-6907
- TCGA-G8-6909
- TCGA-G8-6914
- TCGA-GR-7351
- TCGA-GR-7353
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.

- No data from TARGET-MDLS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

1.13.36 Data Release 10.1

- **GDC Product**: Data
- **Release Date**: February 15, 2018

New updates

1. Updated FM-AD clinical data to conform with Data Dictionary release v1.11

Bugs Fixed Since Last Release

None

Known Issues and Workarounds

- RNA-Seq files for TARGET-NBL are attached to the incorrect aliquot. The BAM files contain the correct information in their header but the connection in the GDC to read groups and aliquots is incorrect. The linked file below contains a mapping between aliquots where file are currently associated and the aliquot where they should instead be associated (mapping file).

- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` were not replaced in DR10.0 and thus do not contain indels. However, the indels from this aliquot can be found in the MAF files and are displayed in the Exploration section in the Data Portal. These VCFs files will be replaced in a later release.

- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- TARGET-MDLS cases do not have disease_type or primary_site populated
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- miRNA alignments include QC failed reads.
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- FM-AD clinical and biospecimen supplement files have incorrect data format. They are listed as XLSX, but are in fact TSV files.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- There are 11 cases in project TCGA-DLBC that are known to have incorrect WXS data in the GDC Data Portal. Impacted cases are listed below. This affects the BAMs and VCFs associated with these cases in the GDC Data Portal. Corrected BAMs can be found in the GDC Legacy Archive. Variants from affected aliquots appear in the protected MAFs with GDC_FILTER=ContEst to indicate a sample contamination problem, but are removed during the generation of the Somatic MAF file. In a later release we will supply corrected BAM, VCF, and MAF files for these cases. In the mean time, we advise you not to use any of the WXS files associated with these cases in the GDC Data Portal. A list of these files can be found here. Download list of affected files.
- TCGA-FF-8062
- TCGA-FM-8000
- TCGA-G8-6324
- TCGA-G8-6325
- TCGA-G8-6326
- TCGA-G8-6906
- TCGA-G8-6907
- TCGA-G8-6909
- TCGA-G8-6914
- TCGA-GR-7351
- TCGA-GR-7353
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.

- No data from TARGET-MDLS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

1.13.37 Data Release 10.0

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

New updates

1. New TARGET files for all projects
2. TARGET updates for clinical and biospecimen data
3. Replace corrupted .bai files
4. Update TCGA and TARGET MAF files to include VarScan2 indels and more information in all_effects column
5. Update VarScan VCF files

   Updated files for this release are listed here. A complete list of files for DR10.0 are listed for the GDC Data Portal here and the GDC Legacy Archive here.

Bugs Fixed Since Last Release

None

Known Issues and Workarounds

- The raw and annotated VarScan VCF files for aliquot `TCGA-VR-A8ET-01A-11D-A403-09` were not replaced in DR10.0 and thus do not contain indels. However, the indels from this aliquot can be found in the MAF files and are displayed in the Exploration section in the Data Portal. These VCFs files will be replaced in a later release.

- There are 5051 TARGET files for which `experimental_strategy`, `data_format`, `platform`, and `data_subtype` are blank
- There are two cases with identical submitter_id `TARGET-10-PARUYU`
- TARGET-MDLS cases do not have disease_type or primary_site populated
- Some TARGET cases are missing `days_to_last_follow_up`
- Some TARGET cases are missing `age_at_diagnosis`
- Some TARGET files are not connected to all related aliquots
- miRNA alignments include QC failed reads.
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- FM-AD clinical and biospecimen supplement files have incorrect data format. They are listed as XLSX, but are in fact TSV files.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- There are 11 cases in project TCGA-DLBC that are known to have incorrect WXS data in the GDC Data Portal. Impacted cases are listed below. This affects the BAMs and VCFs associated with these cases in the GDC Data Portal. Corrected BAMs can be found in the GDC Legacy Archive. Variants from affected aliquots appear in the protected MAFs with GDC_FILTER=ContEst to indicate a sample contamination problem, but are removed during the generation of the Somatic MAF file. In a later release we will supply corrected BAM, VCF, and MAF files for these cases. In the mean time, we advise you not to use any of the WXS files associated with these cases in the GDC Data Portal. A list of these files can be found here. Download list of affected files.
- TCGA-FF-8062
- TCGA-FM-8000
- TCGA-G8-6324
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- TCGA-G8-6326
- TCGA-G8-6906
- TCGA-G8-6907
- TCGA-G8-6909
- TCGA-G8-6914
- TCGA-GR-7351
- TCGA-GR-7353
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.

- No data from TARGET-MDLS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

1.13.38 Data Release 9.0

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

New updates

1. Foundation Medicine Data Release
2. This includes controlled-access VCF and MAF files as well as clinical and biospecimen supplements and metadata.
3. Original Foundation Medicine supplied data can be found on the Foundation Medicine Project Page.
4. Updated RNA-Seq data for TARGET NBL
5. Includes new BAM and count files

Updated files for this release are listed here. A complete list of files for DR9.0 are listed here.

Bugs Fixed Since Last Release

None

Known Issues and Workarounds

- miRNA alignments include QC failed reads.
- Samples of TARGET sample_type `Recurrent Blood Derived Cancer - Bone Marrow` are mislabeled as `Recurrent Blood Derived Cancer - Peripheral Blood`. A workaround is to look at the sample barcode, which is -04 for `Recurrent Blood Derived Cancer - Bone Marrow`. (e.g. `TARGET-20-PAMYAS-04A-03R`)
- FM-AD clinical and biospecimen supplement files have incorrect data format. They are listed as XLSX, but are in fact TSV files.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- There are 11 cases in project TCGA-DLBC that are known to have incorrect WXS data in the GDC Data Portal. Impacted cases are listed below. This affects the BAMs and VCFs associated with these cases in the GDC Data Portal. Corrected BAMs can be

found in the GDC Legacy Archive. Variants from affected aliquots appear in the protected MAFs with GDC_FILTER=ContEst to indicate a sample contamination problem, but are removed during the generation of the Somatic MAF file. In a later release we will supply corrected BAM, VCF, and MAF files for these cases. In the mean time, we advise you not to use any of the WXS files associated with these cases in the GDC Data Portal. A list of these files can be found here. Download list of affected files.

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- TCGA-G8-6906
- TCGA-G8-6907
- TCGA-G8-6909
- TCGA-G8-6914
- TCGA-GR-7351
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- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- No data from TARGET-MDLS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

1.13.39 Data Release 8.0

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

New updates

1. Released updated miRNA quantification files to address double counting of some normalized counts described in DR7.0 release notes.

   Updated files for this release are listed here. A Complete list of files for DR8.0 are listed here.

Bugs Fixed Since Last Release

None

Known Issues and Workarounds

- TARGET-NBL RNA-Seq files were run as single ended even though they are derived from paired-end data. These files will be rerun through the GDC RNA-Seq pipelines in a later release. Impacted files can be found here. Downstream count files are also affected. Users may access original FASTQ files in the GDC Legacy Archive, which are not impacted by this issue.
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.
- There are 11 cases in project TCGA-DLBC that are known to have incorrect WXS data in the GDC Data Portal. Impacted cases are listed below. This affects the BAMs and VCFs associated with these cases in the GDC Data Portal. Corrected BAMs can be found in the GDC Legacy Archive. Variants from affected aliquots appear in the protected MAFs with GDC_FILTER=ContEst to indicate a sample contamination problem, but are removed during the generation of the Somatic MAF file. In a later release we will supply corrected BAM, VCF, and MAF files for these cases. In the mean time, we advise you not to use any of the WXS files associated with these cases in the GDC Data Portal. A list of these files can be found here. Download list of affected files.
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- TCGA-G8-6907
- TCGA-G8-6909
- TCGA-G8-6914
- TCGA-GR-7351
- TCGA-GR-7353
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- No data from TARGET-MDLS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive

- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

1.13.40 Data Release 7.0

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

New updates

1. Updated public Mutation Annotation Format (MAF) files are now available. Updates include filtering to remove variants impacted by OxoG artifacts and those impacted by strand bias.
2. Protected MAF files are updated to include flags for OxoG and strand bias.
3. Annotated VCFs are updated to include flags for OxoG artifacts and strand bias.

Updated files for this release are listed here. A Complete list of files for DR7.0 are listed here

Bugs Fixed Since Last Release

None

Known Issues and Workarounds

- TARGET-NBL RNA-Seq files were run as single ended even though they are derived from paired-end data. These files will be rerun through the GDC RNA-Seq pipelines in a later release. Impacted files can be found here. Downstream count files are also affected. Users may access original FASTQ files in the GDC Legacy Archive, which are not impacted by this issue.
- Reads that are mapped to multiple genomic locations are double counted in some of the GDC miRNA results. The GDC will release updated files correcting the issue in an upcoming release. The specific impacts are described further below:
- Isoform Expression Quantification files
- Raw reads counts are accurate
- Normalized counts are proportionally skewed (r^2=1.0)
- miRNA Expression Quantification files
- A small proportion of miRNA counts are overestimated (mean r^2=0.9999)
- Normalized counts are proportionally skewed (mean r^2=0.9999)
- miRNA BAM files
- no impact
- Mutation frequency may be underestimated when using MAF files for genes that overlap other genes. This is because MAF files only record one gene per variant.
- Most intronic mutations are removed for MAF generation. However, validated variants may rescue these in some cases. Therefore intronic mutations in MAF files are not representative of those called by mutation callers.
- The latest TARGET data is not yet available at the GDC. For the complete and latest data, please see the TARGET Data Matrix. Data that is not present or is not the most up to date includes:
- All microarray data and metadata
- All sequencing analyzed data and metadata
- 1180 of 12063 sequencing runs of raw data
- Demographic information for some TARGET patients is incorrect. The correct information can be found in the associated clinical supplement file. Impacted patients are TARGET-50-PAJNUS.

- There are 11 cases in project TCGA-DLBC that are known to have incorrect WXS data in the GDC Data Portal. Impacted cases are listed below. This affects the BAMs and VCFs associated with these cases in the GDC Data Portal. Corrected BAMs can be found in the GDC Legacy Archive. Variants from affected aliquots appear in the protected MAFs with GDC_FILTER=ContEst to indicate a sample contamination problem, but are removed during the generation of the Somatic MAF file. In a later release we will supply corrected BAM, VCF, and MAF files for these cases. In the mean time, we advise you not to use any of the WXS files associated with these cases in the GDC Data Portal. A list of these files can be found here. Download list of affected files.
- TCGA-FF-8062
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- TCGA-G8-6907
- TCGA-G8-6909
- TCGA-G8-6914
- TCGA-GR-7351
- TCGA-GR-7353
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- No data from TARGET-MLDS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

1.13.41 Data Release 6.0

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

New updates

1. GDC updated public Mutation Annotation Format (MAF) files are now available. Updates include leveraging the MC3 variant filtering strategy, which results in more variants being recovered relative to the previous version. A detailed description of the new format can be found here.
2. Protected MAFs are updated to include additional variant annotation information
3. Some MuTect2 VCFs updated to include dbSNP and COSMIC annotations found in other VCFs

   Updated files for this release are listed here.

Bugs Fixed Since Last Release

None

Known Issues and Workarounds

- There are 11 cases in project TCGA-DLBC that are known to have incorrect WXS data in the GDC Data Portal. Impacted cases are listed below. This affects the BAMs and VCFs associated with these cases in the GDC Data Portal. Corrected BAMs can be found in the GDC Legacy Archive. Variants from affected aliquots appear in the protected MAFs with GDC_FILTER=ContEst to indicate a sample contamination problem, but are removed during the generation of the Somatic MAF file. In a later release we will supply corrected BAM, VCF, and MAF files for these cases. In the mean time, we advise you not to use any of the WXS files associated with these cases in the GDC Data Portal. A list of these files can be found here. Download list of affected files.
- TCGA-FF-8062
- TCGA-FM-8000
- TCGA-G8-6324
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- TCGA-G8-6907
- TCGA-G8-6909
- TCGA-G8-6914
- TCGA-GR-7351
- TCGA-GR-7353
- Variants found in VCF and MAF files may contain OxoG artifacts, which are produced during library preparation and may result in the apparent substitutions of C to A or G to T in certain sequence contexts. In the future we will plan to label potential oxoG artifacts in the MAF files.
- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Some validated somatic mutations may not be present in open-access MAF files. Please review the protected MAF files in the GDC Data Portal if you are unable to find your mutation in the open-access files.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- No data from TARGET-MLDS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

Details are provided in Data Release Manifest

1.13.42 Data Release 5.0

- **GDC Product**: Data
- **Release Date**: March 16, 2017

New updates

1. Additional annotations from TCGA DCC are available
- Complete list of updated TCGA files is found here
2. Clinical data added for TARGET ALL P1 and P2
3. Pathology reports now have submitter IDs as assigned by the BCR
4. TARGET Data refresh
- Most recent biospecimen and clinical information from the TARGET DCC. New imported files are listed here
- Updated indexed biospecimen and clinical metadata
- Updated SRA XMLs files
- Does not include updates to TARGET NBL

Bugs Fixed Since Last Release

1. Missing cases from TCGA-LAML were added to Legacy Archive
2. Biotab files are now linked to Projects and Cases in Legacy Archive

Known Issues and Workarounds

- Some TCGA annotations are unavailable in the Legacy Archive or Data Portal. These annotations can be found here.
- Some validated somatic mutations may not be present in open-access MAF files. When creating open-access MAF files from the protected versions we are extremely conservative in removing potential germline variants. Our approach is to remove all mutations that are present in dbSNP. In a subsequent release we will provide updated open-access MAF files, which preserve variants found in MC3 or a TCGA validation study. Please review the protected MAF files in the GDC Data Portal if you are unable to find your mutation in the open-access files.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- MAF Column #109 "FILTER" entries are separated by both commas and semi-colons.
- TARGET-AML is undergoing reorganization. Pending reorganization, cases from this projects may not contain many clinical, biospecimen, or genomic data files.
- No data from TARGET-MLDS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- Two biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg
- Tumor grade property is not populated
- Progression_or_recurrence property is not populated

Details are provided in Data Release Manifest

1.13.43 Data Release 4.0

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

New updates

1. TARGET ALL P1 and P2 biospecimen and molecular data are now available in the Legacy Archive. Clinical data will be available in a later release.
2. Methylation data from 27k/450k Arrays has been lifted over to hg38 and is now available in the GDC Data Portal
3. Public MAF files are now available for VarScan2, MuSE, and SomaticSniper. MuTect2 MAFs were made available in a previous release.
4. Updated VCFs and MAF files are available for MuTect2 pipeline to compensate for WGA-related false positive indels. See additional information on that change here. A listing of replaced files is provided here.
5. Added submitter_id for Pathology Reports in Legacy Archive

Bugs Fixed Since Last Release

- None

Known Issues and Workarounds

- Some validated somatic mutations may not be present in open-access MAF files. When creating open-access MAF files from the protected versions we are extremely conservative in removing potential germline variants. Our approach is to remove all mutations that are present in dbSNP. In a subsequent release we will provide updated open-access MAF files, which preserve variants found in COSMIC or a TCGA validation study. Please review the protected MAF files in the GDC Data Portal if you are unable to find your mutation in the open-access files.
- Public MAF files for different variant calling pipelines but the same project may contain different numbers of samples. Samples are omitted from the public MAF files if they have no PASS variants, which can lead to this apparent discrepancy.
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- MAF Column #109 "FILTER" entries are separated by both commas and semi-colons.
- TARGET-AML is undergoing reorganization. Pending reorganization, cases from this projects may not contain many clinical, biospecimen, or genomic data files.
- No data from TARGET-MLDS is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- There are 200 cases from TCGA-LAML that do not appear in the Legacy Archive
- Biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg

Details are provided in Data Release Manifest

1.13.44 Data Release 3.0

- **GDC Product**: Data
- **Release Date**: September 16, 2016

New updates

1. CCLE data now available (in the Legacy Archive only)
2. BMI calculation is corrected
3. Slide is now categorized as a Biospecimen entity

Bugs Fixed Since Last Release

- BMI calculation is corrected

Known Issues and Workarounds

- Insertions called for tumor samples that underwent whole genome amplification may be of lower quality. Whether a sample underwent this process can be found in the analyte_type property within analyte and aliquot. TCGA analyte type can be also identified in the 20th character of TCGA barcode, at which "W" corresponds to WGA.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Public MAFs (those with germline variants removed) are only available for MuTect2 pipeline. MAFs for other pipelines are forthcoming.
- MAF Column #109 "FILTER" entries are separated by both commas and semi-colons.
- TARGET-AML and TARGET-ALL projects are undergoing reorganization. Pending reorganization, cases from these projects may not contain many clinical, biospecimen, or genomic data files.
- No data from TARGET-PPTP is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- There are 200 cases from TCGA-LAML that do not appear in the Legacy Archive
- Biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg

Details are provided in Data Release Manifest

1.13.45 Data Release 2.0

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

New updates

1. Additional data, previously available via CGHub and the TCGA DCC, is now available in the GDC
2. Better linking between files and their associated projects and cases in the Legacy Archive
3. MAF files are now available in the GDC Data Portal

Known Issues and Workarounds

- Insertions called for tumor samples that underwent whole genome amplification may be of lower quality. These are present in VCF and MAF files produced by the MuTect2 variant calling pipeline. This information can be found in the analyte_type property within analyte and aliquot. TCGA analyte type can be also identified in the 20th character of TCGA barcode, at which "W" corresponds to WGA.
- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- Public MAFs (those with germline variants removed) are only available for MuTect2 pipeline. MAFs for other pipelines are forthcoming.
- MAF Column #109 "FILTER" entries are separated by both commas and semi-colons.
- TARGET-AML and TARGET-ALL projects are undergoing reorganization. Pending reorganization, cases from these projects may not contain many clinical, biospecimen, or genomic data files.
- No data from TARGET-PPTP is available.
- Slide barcodes (`submitter_id` values for Slide entities in the Legacy Archive) are not available
- SDF Files are not linked to Project or Case in the Legacy Archive
- There are 200 cases from TCGA-LAML that do not appear in the Legacy Archive
- Biotab files are not linked to Project or Case in the Legacy Archive
- SDRF files are not linked to Project or Case in the Legacy Archive
- Portion "weight" property is incorrectly described in the Data Dictionary as the weight of the patient in kg, should be described as the weight of the portion in mg

Details are provided in Data Release Manifest

1.13.46 Initial Data Release (1.0)

- **GDC Product**: Data
- **Release Date**: June 6, 2016

Available Program Data

- The Cancer Genome Atlas (TCGA)
- Therapeutically Applicable Research To Generate Effective Treatments (TARGET)

Available Harmonized Data

- WXS
- Co-cleaned BAM files aligned to GRCh38 using BWA
- mRNA-Seq
- BAM files aligned to GRCh38 using STAR 2-pass strategy
- Expression quantification using HTSeq
- miRNA-Seq
- BAM files aligned to GRCh38 using BWA aln
- Expression quantification using BCCA miRNA Profiling Pipeline*
- Genotyping Array
- CNV segmentation data

Known Issues and Workarounds

- BAM files produced by the GDC RNA-Seq Alignment workflow will currently fail validation using the Picard ValidateSamFiles tool. This is caused by STAR2 not recording mate mapping information for unmapped reads, which are retained in our BAM files. Importantly, all affected BAM files are known to behave normally in downstream workflows including expression quantification.
- All legacy files for TCGA are available in the GDC Legacy Archive, but not always linked back to cases depending on available metadata.
- Public MAFs (those with germline variants removed) are only available for MuTect2 pipeline. MAFs for other pipelines are forthcoming.
- TARGET-AML and TARGET-ALL projects are undergoing reorganization. Pending reorganization, cases from these projects may not contain many clinical, biospecimen, or genomic data files.
- No data from TARGET-PPTP is available.
- Legacy data not available in harmonized form:
- Annotated VCF files from TARGET, anticipated in future data release
- TCGA data that failed harmonization or QC or have been newly updated in CGHub: ~1.0% of WXS aliquots, ~1.6% of RNA-Seq aliquots
- TARGET data that failed harmonization or QC, have been newly updated in CGHub, or whose project names are undergoing reorganization: ~76% of WXS aliquots, ~49% of RNA-Seq aliquots, ~57% of miRNA-Seq.
- MAF Column #109 "FILTER" entries are separated by both commas and semi-colons.
- MAFs are not yet available for query or search in the GDC Data Portal or API. You may download these files using the following manifests, which can be passed directly to the Data Transfer Tool. Links for the open-access TCGA MAFs are provided below for downloading individual files.
- Open-access MAFs manifest
- Controlled-access MAFs manifest

Details are provided in Data Release Manifest

Download Open-access MAF files

- Please note that these links no longer point to files and will be updated in the future.

TCGA.ACC.mutect.somatic.maf.gz
TCGA.BLCA.mutect.somatic.maf.gz
TCGA.BRCA.mutect.somatic.maf.gz
TCGA.CESC.mutect.somatic.maf.gz
TCGA.CHOL.mutect.somatic.maf.gz
TCGA.COAD.mutect.somatic.maf.gz
TCGA.DLBC.mutect.somatic.maf.gz
TCGA.ESCA.mutect.somatic.maf.gz
TCGA.GBM.mutect.somatic.maf.gz
TCGA.HNSC.mutect.somatic.maf.gz
TCGA.KICH.mutect.somatic.maf.gz
TCGA.KIRC.mutect.somatic.maf.gz
TCGA.KIRP.mutect.somatic.maf.gz
TCGA.LAML.mutect.somatic.maf.gz
TCGA.LGG.mutect.somatic.maf.gz
TCGA.LIHC.mutect.somatic.maf.gz
TCGA.LUAD.mutect.somatic.maf.gz
TCGA.LUSC.mutect.somatic.maf.gz
TCGA.MESO.mutect.somatic.maf.gz
TCGA.OV.mutect.somatic.maf.gz

TCGA.PAAD.mutect.somatic.maf.gz
TCGA.PCPG.mutect.somatic.maf.gz
TCGA.PRAD.mutect.somatic.maf.gz
TCGA.READ.mutect.somatic.maf.gz
TCGA.SARC.mutect.somatic.maf.gz
TCGA.SKCM.mutect.somatic.maf.gz
TCGA.STAD.mutect.somatic.maf.gz
TCGA.TGCT.mutect.somatic.maf.gz
TCGA.THCA.mutect.somatic.maf.gz
TCGA.THYM.mutect.somatic.maf.gz
TCGA.UCEC.mutect.somatic.maf.gz
TCGA.UCS.mutect.somatic.maf.gz
TCGA.UVM.mutect.somatic.maf.gz