# Package 'PCAWG7'

## September 17, 2021

Title Repository of data from COSMIC and 'The repertoire of Mutational Signatures in Human Cancer
<b>Version</b> 0.1.1.9001
Description Contains data from The COSMIC  web site https://cancer.sanger.ac.uk/cosmic/signatures/index.tt  and from the paper by Alexandrov, Kim, Haradhvala, Huang et al.,  'The repertoire of Mutational Signatures in Human Cancer'. Please see ?PCAWG7.  https://doi.org/10.1038/s41586-020-1943-3. The funny name  comes from the fact that this paper was generated by  Working Group 7 of the Pan Cancer Analysis of Whole Genomes  (PCAWG) consortium. The signature profiles were later placed on the COSMIC  web site and have been subsequently updated.
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 ${\tt CancerTypes}$ 

Return a character vector of some common cancer types

## Description

Return a character vector of some common cancer types

## Usage

CancerTypes()

## Examples

CancerTypes()[1:5]

COSMIC.v3.0

PCAWG7 SigProfiler reference signatures.

## Description

PCAWG7 SigProfiler reference signatures.

## Usage

COSMIC.v3.0

COSMIC.v3.1

#### **Format**

A list with one element, signature, which in turn is a list with elements:

**genome** A list with the elements:

SBS96 Strand-agnostic single-base substitutions in trinucleotide context.

SBS192 Transcriptionally stranded single-base substitutions in trinucleotide context.

DBS78 Strand-agnostic doublet-base substitutions.

**ID** Strand-agnostic indels.

exome A list with the elements:

**SBS96** As above, for exome count signatures, which look different than genome count signatures, because of differences in trinucleotide frequencies in exomes versus whole genomes. These were signatures that were extracted from exome data in the PCAWG7 paper, not simple adjustment of the genome signatures for exome trinucleotide abundances.

#### **Source**

Subdirectories of https://www.synapse.org/#!Synapse:syn12009743, 2019 Oct 09, populated by data-raw/sig.profiler.signatures/populate.variable.siganture.R.

#### **Examples**

```
SBS96.sigs <- COSMIC.v3.0$signature$genome$SBS96
```

COSMIC.v3.1

Mutational signatures data from COSMIC, the Catalogue Of Somatic Mutations In Cancer, (v3.1 - June 2020)

## **Description**

Mutational signatures data from COSMIC, the Catalogue Of Somatic Mutations In Cancer,  $(v3.1 - June\ 2020)$ 

## Usage

COSMIC.v3.1

#### Format

A list with a single element, genome, which is a list containing:

SBS96 Strand-agnostic single-base substitutions in trinucleotide context.

SBS192 Transcriptionally stranded single-base substitutions in trinucleotide context.

DBS78 Strand-agnostic doublet-base substitutions.

**ID** Strand-agnostic indels.

#### Remark

The signatures are all from Human GRCh37 reference genome.

4 exposure

#### Source

Files downloaded from https://cancer.sanger.ac.uk/cosmic/signatures/index.tt, 2021 Feb and saved in data-raw/COSMIC.v3.1/data/.

Populated by data-raw/COSMIC.v3.1/code/generate-COSMIC.v3.1-genome-sigs.R.

#### **Examples**

SBS96.sigs <- COSMIC.v3.1\$signature\$genome\$SBS96

exposure

PCAWG7 SigProfiler signature assignments (numbers of mutations due to each signature in each tumor).

### **Description**

PCAWG7 SigProfiler signature assignments (numbers of mutations due to each signature in each tumor).

### Usage

exposure

#### **Format**

A list with the elements:

**PCAWG** A list with the elements:

SBS96 Strand-agnostic single-base substitutions in trinucleotide context.

**DBS78** Strand-agnostic doublet-base substitutions.

ID Strand-agnostic indels. These are signature assignments for the PCAWG platinum genomes.

**TCGA** A list with the elements:

SBS96 As above.

**ID** As above. These are signature assignments for the TCGA exomes.

other.genome A list with the element:

SBS96 As above. This contains signature assignments for non-TCGA genomes.

**other.exome** A list with the element:

SBS96 As above. This contains signature assignments for non-TCGA exomes.

## Source

Files of https://www.synapse.org/#!Synapse:syn12009743, 2019 Oct 09, populated by data-raw/sig.profiler.

#### **Examples**

SBS96.exposure <- exposure\$PCAWG\$SBS96

exposure.stats 5

exposure.stats

Exposure statistics from the PCAWG7 paper

### **Description**

Exposure statistics from the PCAWG7 paper

### Usage

```
exposure.stats
```

#### **Format**

A list with one element, PCAWG, which has the sub-elements SBS96, DBS78, ID with statistics for the corresponding mutation types by cancer type. I.e. each element has a sub-element for each cancer type, and this element is a data.frame with one row for each signature and columns mean.of.those.present (the mean number of mutations for those tumors that have the mutation) and proportion.present (the proportion of tumors in which the signature is present).

#### **Source**

Computed from other package variables using GatherPCAWG7ExposureStatsSBS96.

#### **Examples**

```
exposure.stats$PCAWG$SBS96$`Biliary-AdenoCA`[1:3, ]
```

GetEtiology

Get the proposed etiology of a signature

## Description

Get the proposed etiology of a signature

#### Usage

```
GetEtiology(mutation.type, sig.id)
```

## Arguments

```
mutation.type character string, one of SBS96, SBS192, DBS78, ID sig.id character vector with signature ids, e.g. c("SBS3", "foo").
```

### Value

A character vector of the same length as sig.id, each element of which is the etiology of the corresponding signature, if known, or else the empty string.

```
GetEtiology(mutation.type = "ID", sig.id = c("ID1", "foo", "ID3"))
```

## **Description**

Translate aliquot IDs (e.g. e0fccaf5-925a-41f9-b87c-cd5ee4aecb59) to "SP" IDs (e.g. SP1682)

#### Usage

```
map_aliquot_ID_to_SP_ID(aliquot.ids)
```

#### **Arguments**

aliquot.ids Character vector of aliquot IDs.

### Value

Character vector of corresponding "SP" IDs. If a corresponding aliquot ID cannot be found, then return an empty string.

#### **Examples**

```
map_SP_ID_to_tumor_type
```

Given PCAWG "SP" IDs (e.g. SP123958) return either the "full" IDs (Kidney-ChRCC::SP123958) or just the tumor type (Kidney-ChRCC)

## Description

Given PCAWG "SP" IDs (e.g. SP123958) return either the "full" IDs (Kidney-ChRCC::SP123958) or just the tumor type (Kidney-ChRCC)

### Usage

```
map_SP_ID_to_tumor_type(SP.IDs, merge = TRUE)
```

## **Arguments**

SP. IDs A character vector of PCAWG "SP" IDs.

merge If TRUE return a parallel vector of <tumor\_type>::<SP\_ID>; otherwise just <tu-

mor\_type>.

## Details

Fails with an "subscript out of bounds" error if any of the elements of SP. IDs is unknown.

PCAWG.sample.id 7

#### **Examples**

```
map_SP_ID_to_tumor_type(c("SP123958", "SP43633"))
map_SP_ID_to_tumor_type(c("SP123958", "SP43633"), merge = FALSE)
```

PCAWG.sample.id

Vectors of the PCAWG tumor\_wgs\_icgc\_specimen\_ids.

## Description

Note that the PCAWG7 spectra catalogs have 2 sample ids that were blacklisted after the mutational signature analysis was underway. The blacklisted samples are SP116419 and SP116883, which are in PCAWG. sample.id\$black.

#### Usage

```
PCAWG.sample.id
```

#### **Format**

A list with the elements:

white Whitelisted IDsgrey Greylisted IDsblack Blacklisted IDs

## Source

 $https://dcc.icgc.org/api/v1/download?fn=/PCAWG/data\_releases/latest/release\_may2016. v1.4.with\_consensus\_calls.tsv, 2019 Oct 09$ 

PCAWG.sample.sheet

PCAWG sample sheet which contains various sample information

## Description

PCAWG sample sheet which contains various sample information

## Usage

```
PCAWG.sample.sheet
```

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#### **Format**

A data table with the following columns:

- · donor\_unique\_id
- donor\_wgs\_exclusion\_white\_gray
- · submitter\_donor\_id
- · icgc\_donor\_id
- dcc\_project\_code
- aliquot\_id
- submitter\_specimen\_id
- icgc\_sample\_id
- dcc\_specimen\_type
- library\_strategy

#### Source

 $https://dcc.icgc.org/api/v1/download?fn=/PCAWG/data\_releases/latest/pcawg\_sample\_sheet.v1.4.2016-09-14.tsv, 2019 Oct 15$ 

PCAWG7

PCAWG7: A package of data from the COSMIC web site https://cancer.sanger.ac.uk/signatures/ and paper 'Repertoire of Mutational Signatures in Human Cancer'

### **Description**

This is a data package with 3 main package variables: exposure, signature, and spectra.

## Details

There are also PDF plots of the signatures in data-raw/plots/.

There are also several functions for handling PCAWG identifiers:

```
* map_SP_ID_to_tumor_type
```

- \* map\_aliquot\_ID\_to\_SP\_ID
- \* SampleIDToCancerType
- \* SplitPCAWGMatrixByTumorType
- \* SplitMatrixBySampleType

The reference for the data is

Alexandrov, L.B., Kim, J., Haradhvala, N.J. et al. The repertoire of mutational signatures in human cancer. Nature 578, 94-101 (2020). https://doi.org/10.1038/s41586-020-1943-3.

SampleIDToCancerType Split out the cancer type from the sample ID for PCAWG IDs

### **Description**

Split out the cancer type from the sample ID for PCAWG IDs

### Usage

```
SampleIDToCancerType(PCAWGID)
```

### **Arguments**

**PCAWGID** 

A character vector of PCAWG IDs of the form <cancer.type>::<sample.id>.

#### Value

A character vector parallel to PCAWGID containing only the <cancer.type> strings.

#### **Examples**

```
cancer.type <- SampleIDToCancerType("Biliary-AdenoCA::SP117655")</pre>
```

## **Description**

Translate SBS96 signature IDs to SBS192 signature IDs by adding "-E" if necessary.

### Usage

```
SBS96_ID_to_SBS192_ID(sig.ids)
```

## **Arguments**

sig.ids

Character vector of SBS96 signature IDs.

## Value

Character vector of corresponding SBS192 signature IDs; some have "-E" (for exome) post-pended.

```
SBS96.ids <- c("SBS1", "SBS23", "SBS25")
SBS192.ids <- SBS96_ID_to_SBS192_ID(SBS96.ids)
```

10 signature

signature	Mutational signatures data from COSMIC, the Catalogue Of Somatic Mutations In Cancer, (v3.2 - March 2021)

### **Description**

Mutational signatures data from COSMIC, the Catalogue Of Somatic Mutations In Cancer, (v3.2 - March 2021)

## Usage

signature

### **Format**

A list with a single element, genome, which is a list containing:

SBS96 Strand-agnostic single-base substitutions in trinucleotide context.

SBS192 Transcriptionally stranded single-base substitutions in trinucleotide context.

**DBS78** Strand-agnostic doublet-base substitutions.

**ID** Strand-agnostic indels.

## Remark

The signatures are all from Human GRCh37 reference genome.

#### Note

```
SBS10c, SBS10d, SBS91, SBS92, SBS93, SBS94 (total 6) new SBS signatures were added in COSMIC v3.2. See the news from COSMIC release for more details https://cosmic-blog.sanger.ac.uk/cosmic-mutational-signatures-release-v3-2/
```

#### **Source**

```
Files downloaded from \verb|https://cancer.sanger.ac.uk/signatures/downloads/, 2021 Sep and saved in data-raw/COSMIC.v3.2/data/.
```

Populated by data-raw/COSMIC.v3.2/code/generate-COSMIC.v3.2-genome-sigs.R.

```
SBS96.sigs <- signature$genome$SBS96
```

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	t of proposed etiologies from PCAWG7 paper, some manually abviated and a few summarized from the COSMIC web site.
--	-------------------------------------------------------------------------------------------------------------------

### **Description**

List of proposed etiologies from PCAWG7 paper, some manually abbreviated and a few summarized from the COSMIC web site.

## Usage

sigs.etiologies

#### **Format**

A list with the elements:

**SBS96** 

**SBS192** 

**DBS78** 

ID

Each list element is a single column matrix with rownames being the signature IDs and values being a short character string description of the proposed etiology.

In general use GetEtiology, which handles new signatures without elements in sigs.etiologies.

spectra

PCAWG7 mutational spectra (catalogs).

### **Description**

PCAWG7 mutational spectra (catalogs).

#### Usage

spectra

### **Format**

A list with the elements:

SBS96 Deprecated.

DBS78 Deprecated.

**PCAWG** A list with the elements:

SBS96 Strand-agnostic single-base substitutions in trinucleotide context.

SBS192 Single-base substitutions in transcripts based on the sense strand.

SBS1536 Strand-agnostic single-base substitutions in pentanucleotide context.

**DBS78** Strand-agnostic doublet-base substitutions.

**ID** Strand-agnostic indels.

**TCGA** A list with the same elements as the PCAWG element.

**other.genome** A list with the same elements as the PCAWG element but with ID omitted.

other.exome A list with the same elements as the PCAWG element but with ID omitted.

#### Source

Files below https://www.synapse.org/#!Synapse:syn11801889, 2019 Oct 09. Populated by data-raw/spectra/load.package.variable.specra.R.

## **Examples**

```
SBS96.spectra <- spectra$PCAWG$SBS96
```

```
SplitMatrixBySampleType
```

Split an exposure matrix or spectrum matrix into a list of matrices, each for a single sample type.

### **Description**

Split an exposure matrix or spectrum matrix into a list of matrices, each for a single sample type.

#### Usage

```
SplitMatrixBySampleType(M, sample.type)
```

### **Arguments**

M A numerical matrix or data frame or ICAMS catalog in which columns are sam-

ples (e.g. tumors) and rows are either mutational signatures (for exposures) or mutation types (for spectra), and, each element is the number of mutations due

to a given mutational signature or mutation type in a single sample

sample.type A character or numeric vector, each element of which indicates a particular sam-

ple type.

#### Value

Invisibly, the list of exposure or spectrum matrices created by splitting M by sample.type.

```
ff <- matrix(1, nrow=3, ncol = 2)
colnames(ff) <- c("sample1", "sample2")
xx <- SplitMatrixBySampleType(ff, c("sample.type.x", "sample.type.y"))
xx</pre>
```

SplitPCAWGMatrixByTumorType

Extract tumor type from column names and return the input matrix split by tumor type based on the PCAWG <tumor\_type>::<sample\_id> convention.

### **Description**

Extract tumor type from column names and return the input matrix split by tumor type based on the PCAWG <tumor\_type>::<sample\_id> convention.

## Usage

SplitPCAWGMatrixByTumorType(M)

#### **Arguments**

Μ

A numerical matrix or data frame or ICAMS catalog in which columns are samples (e.g. tumors) and rows are either mutational signatures (for exposures) or mutation types (for spectra), and each element is the number of mutations due to a given mutational signature or mutation type in a single sample. The column names must be of the the form <cancer.type>::<sample.ID>.

#### Value

Invisibly, the list of exposure matrices or ICAMS catalogs created by splitting matrix by the tumor type encoded in the column names.

### **Examples**

```
mm <- SplitPCAWGMatrixByTumorType(spectra$PCAWG$DBS78)
mm[[3]][1:4, 1:5]</pre>
```

TCGA\_ID\_to\_ICGC\_ID Translate TCGA(The Cancer Genome Atlas) IDs to ICGC(International Cancer Genome Consortium) IDs

## **Description**

Translate TCGA(The Cancer Genome Atlas) IDs to ICGC(International Cancer Genome Consortium) IDs

#### Usage

```
TCGA_ID_to_ICGC_ID(tcga.ids)
```

#### **Arguments**

tcga.ids Character vector of TCGA IDs.

## Value

Character vector of corresponding ICGC IDs. If a corresponding ICGC ID cannot be found, then return an empty string.

```
tcga.ids <- c("TCGA-AA-A01V", "foo", "TCGA-CA-6717", "bar")
icgc.ids <- TCGA_ID_to_ICGC_ID(tcga.ids)
icgc.ids <- icgc.ids[nzchar(icgc.ids)]</pre>
```

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