Package 'cosmicsig'

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Title Mutational Signatures Data from COSMIC (the Catalogue of Somatic Mutations in Cancer)

Version 1.0.1

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Description Mutational signatures data from COSMIC
(the Catalogue Of Somatic Mutations In Cancer)
<https://cancer.sanger.ac.uk/signatures/>.
There are also some helper functions for handling COSMIC signatures.

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URL https://github.com/Rozen-Lab/cosmicsig

BugReports https://github.com/Rozen-Lab/cosmicsig/issues

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ICAMS,
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R topics documented:

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cosmicsig — *cosmicsig: A package of mutational signatures data from COSMIC (the Catalogue Of Somatic Mutations In Cancer)* https://cancer.sanger.ac.uk/signatures/

Description

This is a data package with 2 main package variables: signature and etiology.

Details

There are also two functions for handling COSMIC signatures:

- get_etiology
- SBS96_ID_to_SBS192_ID

Source

https://cancer.sanger.ac.uk/signatures/.

COSMIC_v3.0 — *Mutational signatures data from COSMIC, the Catalogue Of Somatic Mutations In Cancer, (v3.0 - May 2019)*

Description

Mutational signatures data from COSMIC, the Catalogue Of Somatic Mutations In Cancer, (v3.0 - May 2019)

Usage

```
COSMIC_v3.0
```

Format

A list with one element signature.

- signature is a list with the elements:
  - SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
  - DBS78: Strand-agnostic doublet-base substitutions.
  - ID: Strand-agnostic indels.

Remark

SBS96 and DBS78 signatures are from the latest **Human GRCh38** reference genome.
Because COSMIC only has the GRCh37 version of indel signatures, so ID signatures are from **Human GRCh37** reference genome.
See examples below for transforming genome signatures to exome signatures.
See CatalogRowOrder in package ICAMS for the classification of mutation types.

Source

https://cancer.sanger.ac.uk/signatures/.

Examples

```
## Not run:
# As the abundances of the source sequence of the mutations vary between genome
# and exome and between species, users can use package ICAMS to do the transformations.
if (!requireNamespace("ICAMS", quietly = TRUE)) {
  install.packages("ICAMS")
}
library(ICAMS)
SBS96_sigs_GRCh38_genome <- signature$SBS96

# Transform SBS96 GRCh38 genome signatures to GRCh38 exome signatures
SBS96_sigs_GRCh38_exome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                            target.ref.genome = "GRCh38",
                                            target.region = "exome")

# Transform SBS96 GRCh38 genome signatures to GRCh37 genome signatures
SBS96_sigs_GRCh37_genome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                             target.ref.genome = "GRCh37",
                                             target.region = "genome")

# Transform SBS96 GRCh38 genome signatures to mm10 genome signatures
SBS96_sigs_mm10_genome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                           target.ref.genome = "mm10",
                                           target.region = "genome")

## End(Not run)
```

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 one element `signature`.

- `signature` is 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.

Remark

SBS96 and DBS78 signatures are from the latest **Human GRCh38** reference genome.
Because COSMIC only has the GRCh37 version of SBS transcriptional strand bias and indel signatures, so SBS192 and ID signatures are from **Human GRCh37** reference genome.
See examples below for transforming genome signatures to exome signatures.
See CatalogRowOrder in package ICAMS for the classification of mutation types.

Source

https://cancer.sanger.ac.uk/signatures/.

Examples

```
## Not run:
# As the abundances of the source sequence of the mutations vary between genome
# and exome and between species, users can use package ICAMS to do the transformations.
if (!requireNamespace("ICAMS", quietly = TRUE)) {
  install.packages("ICAMS")
}
library(ICAMS)
SBS96_sigs_GRCh38_genome <- signature$SBS96

# Transform SBS96 GRCh38 genome signatures to GRCh38 exome signatures
SBS96_sigs_GRCh38_exome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                            target.ref.genome = "GRCh38",
                                            target.region = "exome")

# Transform SBS96 GRCh38 genome signatures to GRCh37 genome signatures
SBS96_sigs_GRCh37_genome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                             target.ref.genome = "GRCh37",
                                             target.region = "genome")

# Transform SBS96 GRCh38 genome signatures to mm10 genome signatures
SBS96_sigs_mm10_genome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                           target.ref.genome = "mm10",
                                           target.region = "genome")

## End(Not run)
```

COSMIC_v3.2 *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

```
COSMIC_v3.2
```

Format

A list with two elements, `signature` and `etiology`.

- `signature` is 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.
- `etiology` is a list with elements:
  - `SBS96`
  - `SBS192`
  - `DBS78`
  - `ID`

  Each element in `etiology` 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 `get_etiology`, which handles new signatures without elements in `etiology`.

Remark

`SBS96` and `DBS78` signatures are from the latest **Human GRCh38** reference genome.
Because COSMIC only has the GRCh37 version of SBS transcriptional strand bias and indel signatures, so `SBS192` and `ID` signatures are from **Human GRCh37** reference genome.
See examples below for transforming genome signatures to exome signatures.
See CatalogRowOrder in package ICAMS for the classification of mutation types.

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

https://cancer.sanger.ac.uk/signatures/.

Examples

```
## Not run:
# As the abundances of the source sequence of the mutations vary between genome
# and exome and between species, users can use package ICAMS to do the transformations.
if (!requireNamespace("ICAMS", quietly = TRUE)) {
  install.packages("ICAMS")
}
library(ICAMS)
SBS96_sigs_GRCh38_genome <- signature$SBS96

# Transform SBS96 GRCh38 genome signatures to GRCh38 exome signatures
SBS96_sigs_GRCh38_exome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                            target.ref.genome = "GRCh38",
                                            target.region = "exome")
```

```
# Transform SBS96 GRCh38 genome signatures to GRCh37 genome signatures
SBS96_sigs_GRCh37_genome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                             target.ref.genome = "GRCh37",
                                             target.region = "genome")

# Transform SBS96 GRCh38 genome signatures to mm10 genome signatures
SBS96_sigs_mm10_genome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                           target.ref.genome = "mm10",
                                           target.region = "genome")

## End(Not run)
```

`etiology` *List of mutational signature's proposed etiology summarized from COSMIC, the Catalogue Of Somatic Mutations In Cancer (v3.2 - March 2021)*

Description

List of mutational signature's proposed etiology summarized from COSMIC, the Catalogue Of Somatic Mutations In Cancer (v3.2 - March 2021)

Usage

```
etiology
```

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 `get_etiology`, which handles new signatures do not have an element in `etiology`.

Source

https://cancer.sanger.ac.uk/signatures/.

Examples

```
SBS96_etiology <- etiology$SBS96
```

get_etiology *Get the proposed etiology of COSMIC signature*

Description

The level of evidence supporting the proposed etiologies varies. In addition, some proposed etiologies are more akin to associations than specific, mechanistic causes.

Usage

```
get_etiology(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","SBS5").

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.

Note

The etiologies information is not versionized in COSMIC website compared to signatures.

Examples

```
get_etiology(mutation_type = "ID", sig_id = c("ID1", "foo", "ID3"))
```

SBS96_ID_to_SBS192_ID *Translate SBS96 signature IDs to SBS192 signature IDs by adding "-E" if necessary*

Description

"-E" added to the name of a transcriptional strand bias signature indicates that it was extracted only from exome sequencing data, and thus reflects transcriptional strand bias in the exome rather than in the entire transcript, including introns.

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.

Examples

```
SBS96_ids <- c("SBS1", "SBS23", "SBS25")
SBS192_ids <- SBS96_ID_to_SBS192_ID(SBS96_ids)
```

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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 the following 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.

Remark

`SBS96` and `DBS78` signatures are from the latest **Human GRCh38** reference genome.
Because COSMIC only has the GRCh37 version of SBS transcriptional strand bias and indel signatures, so `SBS192` and `ID` signatures are from **Human GRCh37** reference genome.
See examples below for transforming genome signatures to exome signatures.
See CatalogRowOrder in package ICAMS for the classification of mutation types.

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

https://cancer.sanger.ac.uk/signatures/.

Examples

```
## Not run:
# As the abundances of the source sequence of the mutations vary between genome
# and exome and between species, users can use package ICAMS to do the transformations.
if (!requireNamespace("ICAMS", quietly = TRUE)) {
  install.packages("ICAMS")
}
library(ICAMS)
SBS96_sigs_GRCh38_genome <- signature$SBS96

# Transform SBS96 GRCh38 genome signatures to GRCh38 exome signatures
SBS96_sigs_GRCh38_exome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                            target.ref.genome = "GRCh38",
                                            target.region = "exome")

# Transform SBS96 GRCh38 genome signatures to GRCh37 genome signatures
SBS96_sigs_GRCh37_genome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                             target.ref.genome = "GRCh37",
                                             target.region = "genome")

# Transform SBS96 GRCh38 genome signatures to mm10 genome signatures
SBS96_sigs_mm10_genome <- TransformCatalog(catalog = SBS96_sigs_GRCh38_genome,
                                           target.ref.genome = "mm10",
                                           target.region = "genome")

## End(Not run)
```

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