

Package ‘cosmicsig’

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

Version 1.0.3

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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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cosmicsig	<i>cosmicsig: A package of mutational signatures data from COSMIC (the Catalogue Of Somatic Mutations In Cancer)</i> https://cancer.sanger.ac.uk/signatures/
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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	<i>Mutational signatures data from COSMIC, the Catalogue Of Somatic Mutations In Cancer, (v3.0 - May 2019)</i>
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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:
 - GRCh37: Homo sapiens (human) genome assembly GRCh37.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.
 - * ID: Strand-agnostic indels.
 - GRCh38: Homo sapiens (human) genome assembly GRCh38.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.
 - mm9: Mus musculus (house mouse) genome assembly mm9.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.

- mm10: Mus musculus (house mouse) genome assembly mm10.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.
- rn6: Rattus norvegicus (Norway rat) genome assembly rn6.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.

Remark

The signatures are all genome signatures. 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, users can use package ICAMS to do the transformations.
if (!requireNamespace("ICAMS", quietly = TRUE)) {
  install.packages("ICAMS")
}
library(ICAMS)
SBS96_sig_GRCh37_genome <- COSMIC_v3.0$signature$GRCh37$SBS96

# Transform SBS96 GRCh37 genome signatures to GRCh37 exome signatures
SBS96_sig_GRCh37_exome <- TransformCatalog(
  catalog = SBS96_sig_GRCh37_genome,
  target.ref.genome = "GRCh37",
  target.region = "exome"
)

## 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:
 - GRCh37: Homo sapiens (human) genome assembly GRCh37.
 - * 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.
 - GRCh38: Homo sapiens (human) genome assembly GRCh38.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.
 - mm9: Mus musculus (house mouse) genome assembly mm9.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.
 - mm10: Mus musculus (house mouse) genome assembly mm10.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.
 - rn6: Rattus norvegicus (Norway rat) genome assembly rn6.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.

Remark

The signatures are all genome signatures. 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, users can use package ICAMS to do the transformations.
if (!requireNamespace("ICAMS", quietly = TRUE)) {
  install.packages("ICAMS")
}
library(ICAMS)
SBS96_sig_GRCh37_genome <- COSMIC_v3.1$signature$GRCh37$SBS96

# Transform SBS96 GRCh37 genome signatures to GRCh37 exome signatures
SBS96_sig_GRCh37_exome <- TransformCatalog(
  catalog = SBS96_sig_GRCh37_genome,
  target.ref.genome = "GRCh37",
  target.region = "exome"
)

## End(Not run)
```

COSMIC_v3.2	<i>Mutational signatures data from COSMIC, the Catalogue Of Somatic Mutations In Cancer, (v3.2 - March 2021)</i>
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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:
 - GRCh37: Homo sapiens (human) genome assembly GRCh37.
 - * 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.
 - GRCh38: Homo sapiens (human) genome assembly GRCh38.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.
 - mm9: Mus musculus (house mouse) genome assembly mm9.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.
 - mm10: Mus musculus (house mouse) genome assembly mm10.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.
 - rn6: Rattus norvegicus (Norway rat) genome assembly rn6.
 - * SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - * DBS78: Strand-agnostic doublet-base substitutions.
- 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.

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/>

Remark

The signatures are all genome signatures. 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, users can use package ICAMS to do the transformations.
if (!requireNamespace("ICAMS", quietly = TRUE)) {
  install.packages("ICAMS")
}
library(ICAMS)
SBS96_sig_GRCh37_genome <- COSMIC_v3.2$signature$GRCh37$SBS96

# Transform SBS96 GRCh37 genome signatures to GRCh37 exome signatures
SBS96_sig_GRCh37_exome <- TransformCatalog(
  catalog = SBS96_sig_GRCh37_genome,
  target.ref.genome = "GRCh37",
  target.region = "exome"
)

## 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	<i>Get the proposed etiology of COSMIC signature</i>
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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"))
```

possible_artifacts	<i>Return a character vector of the IDs of possible SBS96 signature artifacts</i>
--------------------	---

Description

Return a character vector of the IDs of possible SBS96 signature artifacts

Usage

```
possible_artifacts()
```

Examples

```
artifact_sigs <- possible_artifacts()
```

rare_signatures	<i>Return a character vector of the IDs of rare SBS96 signatures</i>
-----------------	--

Description

Return a character vector of the IDs of rare SBS96 signatures

Usage

```
rare_signatures()
```

Examples

```
rare_sigs <- rare_signatures()
```

SBS96_ID_to_SBS192_ID	<i>Translate SBS96 signature IDs to SBS192 signature IDs by adding "-E" if necessary</i>
-----------------------	--

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)
```

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

- GRCh37: Homo sapiens (human) genome assembly GRCh37.
 - 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.
- GRCh38: Homo sapiens (human) genome assembly GRCh38.
 - SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - DBS78: Strand-agnostic doublet-base substitutions.
- mm9: Mus musculus (house mouse) genome assembly mm9.
 - SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - DBS78: Strand-agnostic doublet-base substitutions.
- mm10: Mus musculus (house mouse) genome assembly mm10.
 - SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - DBS78: Strand-agnostic doublet-base substitutions.
- rn6: Rattus norvegicus (Norway rat) genome assembly rn6.
 - SBS96: Strand-agnostic single-base substitutions in trinucleotide context.
 - DBS78: Strand-agnostic doublet-base substitutions.

Remark

The signatures are all genome signatures. 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, users can use package ICAMS to do the transformations.
if (!requireNamespace("ICAMS", quietly = TRUE)) {
  install.packages("ICAMS")
}
library(ICAMS)
SBS96_sig_GRCh37_genome <- signature$GRCh37$SBS96

# Transform SBS96 GRCh37 genome signatures to GRCh37 exome signatures
SBS96_sig_GRCh37_exome <- TransformCatalog(
  catalog = SBS96_sig_GRCh37_genome,
  target.ref.genome = "GRCh37",
  target.region = "exome"
)

## End(Not run)
```

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