Package 'supersigs'

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Title Supervised mutational signatures

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Depends R (>= 4.1)

Imports assertthat, caret, dplyr, tidyr, rsample, methods, rlang, utils, Biostrings, stats, SummarizedExperiment

Suggests BSgenome.Hsapiens.UCSC.hg19, BSgenome.Hsapiens.UCSC.hg38, knitr, rmarkdown, ggplot2, testthat, VariantAnnotation

Description Generate SuperSigs (supervised mutational signatures) from single nucleotide variants in the cancer genome. Functions included in the package allow the user to learn supervised mutational signatures from their data and apply them to new data. The methodology is based on the one described in Afsari (2021, ELife).

biocViews FeatureExtraction, Classification, Regression, Sequencing, WholeGenome, SomaticMutation

BugReports https://github.com/TomasettiLab/supersigs/issues

URL https://tomasettilab.github.io/supersigs/

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LazyDataCompression gzip

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Author Albert Kuo [aut, cre] (ORCID: <https://orcid.org/0000-0001-5155-0748>), Yifan Zhang [aut], Bahman Afsari [aut], Cristian Tomasetti [aut]

Maintainer Albert Kuo <albertkuo@jhu.edu>

Contents

`example_dt` *Example dataset of mutations*

Description

A dataset containing a list of mutations and other necessary attributes

Usage

```
example_dt
```

Format

A data frame with 10 rows and 5 columns:

sample_id ID of the patient

age age of the patient

chromosome chromosomal position of the mutation

position position of the mutation

ref original nucleotide

alt mutated nucleotide

`get_signature` *Function to obtain a SuperSig*

Description

Generate a tissue-specific SuperSig for a given dataset of mutations and exposure factor. Returns the SuperSig and a classification model trained with the SuperSig.

Usage

```
get_signature(data, factor, wgs = FALSE)
```

Arguments

data	a data frame of mutations containing columns for sample_id, age, IndVar, and the 96 trinucleotide mutations (see vignette for details)
factor	the factor/exposure (e.g. "age", "smoking"). If the factor = "age", the SuperSig is computed using counts. Otherwise, rates (counts/age) are used.
wgs	logical value indicating whether sequencing data is whole-genome (wgs = TRUE) or whole-exome (wgs = FALSE)

Value

get_signature returns an object of class SuperSig

Examples

```
head(example_dt) # use example data from package
input_dt <- make_matrix(example_dt) # convert to correct format
input_dt$IndVar <- c(1, 1, 1, 0, 0) # add IndVar column
get_signature(data = input_dt, factor = "Age") # get SuperSig
```

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make_matrix *Function to transform mutations into "matrix" format*

Description

Transform a data frame of mutations in long format into a data frame of trinucleotide mutations with flanking bases in a wide matrix format.

Usage

```
make_matrix(data, genome = "hg19")
```

Arguments

data	a data frame of mutations in VCF format (see vignette for details)
genome	the reference genome used ("hg19" or "hg38")

Value

make_matrix returns a data frame of mutations, one row per sample

Examples

```
head(example_dt) # use example data from package
input_dt <- make_matrix(example_dt) # convert to correct format
head(input_dt)
```

partial_signature *Function to remove the contribution of a SuperSig*

Description

Remove the contribution of a SuperSig from the data and return the data.

Usage

```
partial_signature(data, object)
```

Arguments

data	a data frame of mutations containing columns for sample_id, age, IndVar, and the 96 trinucleotide mutations (see vignette for details)
object	an object of class SuperSig

Value

predict_signature returns the original data frame with the contribution of a supervised signature removed

Examples

```
head(example_dt) # use example data from package
input_dt <- make_matrix(example_dt) # convert to correct format
input_dt$IndVar <- c(1, 1, 1, 0, 0) # add IndVar column
supersig <- get_signature(data = input_dt, factor = "Age") # get SuperSig
partial_signature(data = input_dt, object = supersig)
```

predict_signature *Function to predict using SuperSig object*

Description

Using a generated SuperSig, predict on a new dataset and return predicted probabilities for each observation.

Usage

```
predict_signature(object, newdata, factor)
```

Arguments

object	an object of class SuperSig
newdata	a data frame of mutations containing columns for sample_id, age, IndVar, and the 96 trinucleotide mutations (see vignette for details)
factor	the factor/exposure (e.g. "age", "smoking")

Value

predict_signature returns the original data frame with additional columns for the feature counts and classification score

Examples

```
head(example_dt) # use example data from package
input_dt <- make_matrix(example_dt) # convert to correct format
input_dt$IndVar <- c(1, 1, 1, 0, 0) # add IndVar column
out <- get_signature(data = input_dt, factor = "Age") # get SuperSig
newdata <- predict_signature(out, newdata = input_dt, factor = "age")
suppressPackageStartupMessages({library(dplyr)})
head(newdata %>% select(score))
```

---

process_vcf *Function to transform VCF object into "matrix" format*

Description

Transform a VCF object into a data frame of trinucleotide mutations with flanking bases in a wide matrix format. The function assumes that the VCF object contains only one sample and that each row in rowRanges represents an observed mutation in the sample.

Usage

```
process_vcf(vcf)
```

Arguments

vcf	a VCF object (from VariantAnnotation package)

Value

process_vcf returns a data frame of mutations, one row per mutation

Examples

```
# Use example vcf from VariantAnnotation
suppressPackageStartupMessages({library(VariantAnnotation)})
fl <- system.file("extdata", "chr22.vcf.gz", package="VariantAnnotation")
vcf <- VariantAnnotation::readVcf(fl, "hg19")

# Subset to first sample
vcf <- vcf[, 1]
# Subset to row positions with homozygous or heterozygous alt
positions <- geno(vcf)$GT != "0|0"
vcf <- vcf[positions[, 1],]
colData(vcf)$age <- 50        # Add patient age to colData (optional)

# Run function
dt <- process_vcf(vcf)
head(dt)
```

simplify_signature *Function to simplify signature representation into interpretable labels for visualization purposes*

Description

Take a signature representation from SuperSig and group trinucleotides within each feature into interpretable labels, with optional IUPAC labeling from IUPAC_CODE_MAP in the Biostrings package

Usage

```
simplify_signature(object, iupac)
```

Arguments

object	an object of class SuperSig
iupac	logical value indicating whether to use IUPAC labels (iupac = TRUE) or not (iupac = FALSE)

Value

simplify_signature returns a vector of simplified features and their difference in mean mean rates between exposed and unexposed (or average rate if the factor is "age")

Examples

```
head(example_dt) # use example data from package
input_dt <- make_matrix(example_dt) # convert to correct format
input_dt$IndVar <- c(1, 1, 1, 0, 0) # add IndVar column
supersig <- get_signature(data = input_dt, factor = "Smoking")
simplify_signature(object = supersig, iupac = FALSE)
simplify_signature(object = supersig, iupac = TRUE)
```

SuperSig-class *An S4 class for SuperSig*

Description

An S4 class for SuperSig

Slots

Signature data frame of features and their difference in mean rates between exposed and unexposed (or the average rate if the factor is "age")

Features list of features that comprise the signature and their representation in terms of the fundamental (trinucleotide) mutations

AUC length-one numeric vector of the apparent AUC (i.e. not cross-validated)

Model list of a glm class for trained logistic regression model

supersig_ls *Trained SuperSigs from TCGA*

Description

A list containing 67 SuperSigs

Usage

```
supersig_ls
```

Format

A named list with 67 elements, each of which is a 'SuperSig'

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