

Package ‘tMAE’

September 26, 2019

Title Tests and visualize for Mono-allelic expressed variants

Version 0.0.0.9000

Description What the package does (one paragraph).

License What license it uses

Encoding UTF-8

LazyData true

RoxygenNote 6.1.1

Imports dplyr,
data.table,
magrittr,
tidyr,
GenomicRanges,
SummarizedExperiment,
ggplot2

Depends dplyr,
data.table,
magrittr,
tidyr,
GenomicRanges,
SummarizedExperiment,
ggplot2

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add_gnomAD_AF	<i>Add allele frequencies from gnomAD</i>
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Description

Add allele frequency information from gnomAD.

Usage

```
add_gnomAD_AF(data, gene_assembly = c("hg19", "hg38"),
  max_af_cutoff = 0.001, populations = c("AF", "AF_afr", "AF_amr",
    "AF_eas", "AF_nfe"))
```

Arguments

`data` A data.frame containing allelic counts.

`gene_assembly` either 'hg19' or 'hg38' indicating the genome assembly of the variants.

`max_af_cutoff` cutoff for a variant to be considered rare. Default is .001.

`populations`

Value

A data.table with the original contents plus columns containing allele frequencies from different gnomAD populations.

Author(s)

Vicente Yepez

Examples

```
file <- system.file("extdata", "demo_MAE_counts.tsv", package = "tMAE", mustWork = TRUE)
maeCounts <- read.table(file)
maeRes <- run_deseq_all_mae(maeCounts)
maeRes <- add_gnomAD_AF(maeCounts, gene_assembly = 'hg19')
```

DESeq4MAE

Run deseq test for MAE

Description

Uses a negative binomial test to determine if a variant is mono-allelically expressed.

Usage

```
DESeq4MAE(data, minCoverage = 10, disp = 0.05,
  independentFiltering = FALSE)
```

Arguments

`data` A data.frame containing allelic counts.

`minCoverage` minimum total allelic count. Default is 10.

`disp` Gene dispersion for the NB test. Default is 0.05.

`independentFiltering` Parameter that affects the multiple testing. Default is FALSE.

Value

Mono-allelic results table containing original counts plus p-value, p-adjusted and freqALT columns.

Author(s)

Vicente Yepez, Christian Mertes

Examples

```
file <- system.file("extdata", "demo_MAE_counts.tsv", package = "tMAE", mustWork = TRUE)
maeCounts <- read.table(file)
run_deseq_all_mae(maeCounts)
```

plotMA	<i>plotMA</i>
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Description

Creates an MA plot, ie, Fold Change (ALT/REF) vs Coverage colored by significance (from p adjusted and allelic ratio) or significance and minor allele frequency (if rare_column is provided).

Usage

```
plotMA(data, title = NULL, padjCutoff = 0.05,
        allelicRatioCutoff = 0.8, rare_column = NULL)
```

Arguments

data	A data.frame containing the results table from DESeq4MAE function
title	The plot's title
padjCutoff	The significance level
allelicRatioCutoff	The minimum allelic ratio ALT/(ALT+REF) to be considered significant
rare_column	The name of the column that indicates if a variant is rare or not. Default is null which means it won't be plotted.

Value

A ggplot object containing the MA plot.

Author(s)

Vicente Yepez

Examples

```
file <- system.file("extdata", "demo_MAE_counts.tsv",
  package = "tMAE", mustWork = TRUE)
maeCounts <- read.table(file)
res <- run_deseq_all_mae(maeCounts)
plotMA(res)
```

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