

Package ‘GVB’

June 13, 2024

Title GVB - Gene-wise Variant Burden

Version 1.0.0

Description Gene-wise Variant Burden(GVB) is an integrated gene-level measure of the cumulative impact of the multitude of deleterious variants on a given gene in a population data-free manner.
The 'GVB' package facilitates preprocessing and calculation of GVB scores. In addition, it supports visualization of GVB scores.

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Encoding UTF-8

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Imports psych, dplyr, magrittr

Depends R (>= 2.10)

LazyData true

R topics documented:

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GVB	<i>GVB() Function</i>
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Description

This function calculates the GVB score.

Usage

```
GVB(
  input,
  score = "SIFT",
  threshold = 0.7,
  direction = "or_less",
  homozygote = FALSE,
  method = "gm",
  indelScore = NULL
)
```

Arguments

input	a list, the output of GVB_preProcessing() function.
score	a character vector for in-silico variant score (e.g.: "SIFT", "PP2", "CADD") which is a column name of the data frame as elements of 'input' (Default = "SIFT").
threshold	a numeric vector specifying cut-off of the in-silico variant score. The default value for "SIFT" is 0.7, for "PP2" it's 0.85, and for "CADD" it's 15.
direction	The direction of scores to be included based on the threshold. Only one of "or_less" or "or_more" is allowed. "or_less" means \leq the threshold and "or_more" means \geq the threshold (Default = "or_less").
homozygote	logical value. If TRUE, consider homozygotes for GVB calculation. (Default = FALSE).
method	allowed values are one of "gm" or "hm". "gm" means geometric mean and "hm" means harmonic mean (Default = "gm").
indelScore	a numeric value to assign the in-silico variant score of indel variants when the 'score' parameter is "SIFT" or "PP2" (Default = NULL).

Examples

```
VDF_list <- GVB_preProcessing(VariantDataFrame)
GVB(input = VDF_list, score = "SIFT", threshold = 0.7, direction = "or_less", homozygote = TRUE,
method = "hm", indelScore)
```

GVB_converting

GVB_converting() Function

Description

This function converts a VEP tab-delimited output to a GVB input.

Usage

```
GVB_converting(vep_tab)
```

Arguments

vep_tab	a character vector for a file path of the VEP tab output.
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Examples

```
GVB_converting(vep_tab = "VEP-tab_Annotation.txt")
```

GVB_histogram	<i>GVB_histogram() Function</i>
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Description

This function plots the GVB score for one specific gene as a histogram from merged GVB data, which is the output of the GVB() function.

Usage

```
GVB_histogram(data, geneSymbol, outputDirectory = NULL)
```

Arguments

data	a matrix, the output of GVB_merging() function.
geneSymbol	a character vector as gene symbol which is a column name of the 'data'.
outputDirectory	a character vector as directory for saving the plot. The file name of plot is geneSymbol_histogram.jpg.

Examples

```
GVB_histogram(data = merged_gvb, geneSymbol = "Gene1")
```

GVB_merging	<i>GVB_merging() Function</i>
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Description

This function merges GVB data frames of multiple samples derived from GVB() function.

Usage

```
GVB_merging(x)
```

Arguments

x	a vector that includes object names of GVB data frames as global variables.
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Examples

```
GVB_merging(x = GVB_DataFrames_Vector)
```

GVB_preCalculation	<i>GVB_preCalculation()</i> Function
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Description

This function is for calculating the GVB score.

Usage

```
GVB_preCalculation(
  input,
  score,
  threshold,
  direction,
  homozygote,
  method,
  indelScore
)
```

Arguments

input	a list, the output of GVB_preProcessing() function.
score	a character vector for in-silico variant score (e.g.: "SIFT", "PP2", "CADD") which is a column name of the data frame as elements of 'input' (Default = "SIFT").
threshold	a numeric vector specifying cut-off of the in-silico variant score. The default value for "SIFT" is 0.7, for "PP2" it's 0.85, and for "CADD" it's 15.
direction	The direction of scores to be included based on the threshold. Only one of "or_less" or "or_more" is allowed. "or_less" means \leq the threshold and "or_more" means \geq the threshold (Default = "or_less").
homozygote	logical value. If TRUE, consider homozygotes for GVB calculation. (Default = FALSE).
method	allowed values are one of "gm" or "hm". "gm" means geometric mean and "hm" means harmonic mean (Default = "gm").
indelScore	a numeric value to assign the in-silico variant score of indel variants when the 'score' parameter is "SIFT" or "PP2" (Default = NULL).

Examples

```
VDF_list <- GVB_preProcessing(VariantDataFrame)
GVB_preCalculation(input = VDF_list, score = "SIFT", threshold = 0.7, direction = "or_less",
homozygote = TRUE, method = "hm", indelScore)
```

GVB_preProcessing	<i>GVB_preProcessing() Function</i>
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Description

This function preprocesses the input and converts it to a list where each element is a data frame for each gene before calculating the GVB score.

Usage

```
GVB_preProcessing(x, indel = FALSE)
```

Arguments

x	a data frame derived from individual variant annotated file.
indel	a logical value. If TRUE, include indel variants (Default = FALSE).

Examples

```
GVB_preProcessing(x, indel=FALSE)
```

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