Package 'ifCNVR'

October 13, 2022				
Title Isolation-Forest Based 'CNV' Detection from 'NGS' Data				
Version 0.1.0				
<pre>URL https://github.com/SimCab-CHU/ifCNVR</pre>				
Description Automatically detects Copy Number Variations (CNV) from Next Generation Sequencing data using a machine learning algorithm, Isolation forest. More details about the method can be found in the paper by Cabello-Aguilar (2022) <doi:10.1101 2022.01.03.474771="">.</doi:10.1101>				
License GPL-3				
Encoding UTF-8				
LazyData true				
RoxygenNote 7.1.2				
Imports data.table, rmarkdown, isotree				
Depends R (>= 2.10)				
NeedsCompilation no				
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Repository CRAN				
Date/Publication 2022-02-15 19:40:02 UTC				
R topics documented: abSamples abTargets calculateRatio calculateScore CreateReadsMatrix generateReport normalizeReads readsMatrixExample				

2 abTargets

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nples abSamples

Description

abSamples

Usage

```
abSamples(readsMatrix, conta = "auto", q = 0.99, verbose = TRUE)
```

Arguments

readsMatrix a matrix of the number of reads per target conta a parameter for the isotree function

q quantile verbose a boolean

Value

the aberrant and normal samples

Examples

```
reads \texttt{Matrix} = \texttt{data.frame}(\texttt{targets=paste0}(\texttt{"target\_"}, \texttt{seq(1,50)}), \ \texttt{matrix}(\texttt{runif(500)}, \texttt{nrow=50}, \texttt{ncol=10})) \\ \texttt{abSamples}(\texttt{readsMatrix})
```

Description

abTargets

Usage

```
abTargets(readsMatrix, abSamples, opt = "regular", pred = 0.6)
```

Arguments

readsMatrix the reads matrix

abSamples the abSamples list result of the abSamples function

opt "regular" or "extensive" a parameter

pred a threshold on the isolation forest outlier prediction (range=[0,1[)

calculateRatio 3

Value

a list of dataframes of the targets tagged as outliers

Examples

```
abTargets(readsMatrixExample,abSamples(readsMatrixExample))
```

calculateRatio

calculate Ratio

Description

calculate Ratio

Usage

```
calculateRatio(readsMatrix, abSamples, roi, soi)
```

Arguments

readsMatrix the reads matrix

abSamples the abSamples list result of the abSamples function

roi the region of interest soi the sample of interest

Value

the ratio associated with the abTargets

Examples

```
calculate Ratio (reads \texttt{MatrixExample}, \ ab \texttt{Samples} (reads \texttt{MatrixExample}), \ "\texttt{EGFR-Ex20"}, \ "sample\_2")
```

4 calculateScore

-	-		_	
cal	CUL	ate	Sco	re

calculate Score

Description

calculate Score

Usage

```
calculateScore(
  readsMatrix,
  abSamples,
  abTargets,
  roi = "Gene",
  sep = "-",
  thrScore = 7
)
```

Arguments

readsMatrix	a reads matrix with samples in columns and targets in lines (the first column are the targets)
abSamples	the abSamples list result of the abSamples function
abTargets	a list of dataframes of the targets tagged as outliers result of the abTargets() function
roi	the region of interest (Gene or Gene-Exon)
sep	a character the separator between roi in the bed file
thrScore	(default 0) a threshold on the localization score

Value

the score associated with the abSamples in the desired roi

Examples

```
abS <- abSamples(readsMatrixExample)
abT <- abTargets(readsMatrixExample,abSamples(readsMatrixExample))
calculateScore(readsMatrixExample, abS, abT, sep="-")</pre>
```

CreateReadsMatrix 5

CreateReadsMatrix C

CreateReadsMatrix

Description

CreateReadsMatrix

Usage

```
CreateReadsMatrix(
  bamPath,
  bedFile,
  bedtoolsPath,
  outputFile = "n",
  verbose = TRUE
)
```

Arguments

bamPath a path leading to the .bam and .bai files

bedFile a path leading to the .bed file (Warning replace chrX by X in the position)

bedtoolsPath the path leading to bedtools

outputFile (optional) a path leading to a text file

verbose a boolean

Value

a reads matrix

Examples

```
bamPath <- system.file("extdata/",package = "ifCNVR")
bed <- system.file("bedFile.bed",package = "ifCNVR")
bedtools <- 'n'
readsMatrix <- CreateReadsMatrix(bamPath, bed, bedtools)</pre>
```

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generateReport

generateReport

Description

generateReport

Usage

```
generateReport(outputFile = "n", readsMatrix, resTable, CNVpos)
```

Arguments

outputFile a path to the html output file

readsMatrix the reads matrix

resTable the table result of the CalculateScore() function
CNVpos the CNVpos list result of the abSamples function

Value

a html report

Examples

generateReport()

normalizeReads

normalizeReads

Description

normalizeReads

Usage

```
normalizeReads(readsMatrix)
```

Arguments

readsMatrix

a reads matrix with samples in columns and targets in lines (the first column are the targets)

Value

a normalized reads matrix

readsMatrixExample 7

Examples

```
normReads <- normalizeReads(readsMatrixExample)</pre>
```

readsMatrixExample

Example dataset

Description

A dataset for the examples

Usage

```
readsMatrixExample
```

Format

A data frame with 11 rows and 6 variables:

```
targets the targets
```

sample_1 the number of reads in sample 1

sample_2 the number of reads in sample 2

sample_3 the number of reads in sample 3

sample_4 the number of reads in sample 4

sample_5 the number of reads in sample 5 ...

scoring

scoring

Description

scoring

Usage

```
scoring(k, n, N)
```

Arguments

k	number of modifie	d targets on the region
K	number of mounte	u targets on the region

n number of targets on the region

N number of targets in the panel

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Value

the confidence score

Examples

scoring(10,20,150)

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