

Package ‘BOBaFIT’

April 3, 2023

Type Package

Title Refitting diploid region profiles using a clustering procedure

Version 1.2.0

Description This package provides a method to refit and correct the diploid region in copy number profiles. It uses a clustering algorithm to identify pathology-specific normal (diploid) chromosomes and then use their copy number signal to refit the whole profile. The package is composed by three functions: DRrefit (the main function), ComputeNormalChromosome and PlotCluster.

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Encoding UTF-8

LazyData true

RoxygenNote 7.1.2

URL <https://github.com/andrea-poletti-unibo/BOBaFIT>

BugReports <https://github.com/andrea-poletti-unibo/BOBaFIT/issues>

Imports dplyr, NbClust, ggplot2, ggbio, grDevices, stats, tidyr, GenomicRanges, ggforce, stringr, plyranges, methods, utils, magrittr

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Config/testthat/edition 3

biocViews CopyNumberVariation, Clustering, Visualization, Normalization, Software

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R topics documented:

computeNormalChromosomes	2
DRrefit	3
DRrefit_plot	4
PlotChrCluster	5
Popeye	6
TCGA_BRCA_CN_segments	6
Index	8

computeNormalChromosomes
<i>computeNormalChromosomes</i>

Description

This function compute the DRrefits' input "chromosome list". It is a vector that contains the chromosomal arms considered "normal" in the cohort of samples tested (BED file), under a specific tolerance value

Usage

```
computeNormalChromosomes(  
  segments,  
  tolerance_val = 0.15,  
  maxCN = 6,  
  min_threshold = 1.6,  
  max_threshold = 2.4,  
  verbose = FALSE  
)
```

Arguments

segments	data.frame formatted with correct column names
tolerance_val	decimal value of alteration frequency. By default is 0.15
maxCN	threshold of max copy number to consider. By default is 6
min_threshold	minimum threshold to define a normal CN. By default is 1.60
max_threshold	maximum threshold to define a normal CN. By default is 2.40
verbose	print information about the processes of the function. By default is FALSE

Value

vector with chromosome names and plot with the alteration rate of each chromosomal arms

Examples

```
data("TCGA_BRCA_CN_segments")
chr_list <- computeNormalChromosomes(segments = TCGA_BRCA_CN_segments)
```

DRrefit	<i>DRrefit</i>
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Description

This function refits the diploid region of input copy number profiles (segments - BED file)

Usage

```
DRrefit(
  segments_chort,
  chrlist,
  maxCN = 6,
  clust_method = "ward.D2",
  verbose = FALSE
)
```

Arguments

segments_chort	data.frame formatted with correct column names
chrlist	list of normal chromosome arms (pathology-specific)
maxCN	threshold of max copy number to consider. By default is 6
clust_method	clustering method. By default is "ward.D2"
verbose	print information about the processes of the function. By default is FALSE

Value

Return two data frames, one is the DRrefit-corrected segments and the other is the samples report. See the vignette for data frame descriptions.

Examples

```
data("TCGA_BRCA_CN_segments")

chr_list <- c("10q", "11p", "12p", "19q", "1p", "21q", "2q", "3p", "4p", "4q", "6p", "6q", "7p" )

results <- DRrefit(segments_chort = TCGA_BRCA_CN_segments,
  chrlist = chr_list)
```

DRrefit_plot	<i>DRrefit_plot</i>
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Description

The function plot the copy number profile before and after DRrefit recalibration

Usage

```
DRrefit_plot(
  corrected_segments,
  DRrefit_report,
  plot_viewer = F,
  plot_save = F,
  plot_format = "png",
  plot_path
)
```

Arguments

corrected_segments	DRrefit output dataframe.
DRrefit_report	DRrefit output dataframe.
plot_viewer	Logical parameter. When it is TRUE, the function print the output plot in the R viewer. By default is FALSE.
plot_save	Logical parameter. When it is TRUE, the function save the plot in the chosen path and format. By default is FALSE.
plot_format	File format for the output plots (accepts "png", "jpg", "pdf", "tiff"). By default is "png"
plot_path	Path to save output plots.

Value

Return the sample copy number profile before and after DRrefit recalibration. The function can output the figure in the R viewer or save it in a specific path.

Examples

```
data("TCGA_BRCA_CN_segments")

chr_list <- c("10q", "11p", "12p", "19q", "1p", "21q", "2q", "3p", "4p", "4q", "6p", "6q", "7p" )

results <- DRrefit(segments_chort = TCGA_BRCA_CN_segments, chrlist = chr_list)

my_segments <- results$corrected_segments
my_report <- results$report
```

```
DRrefit_plot(corrected_segments = my_segments,
             DRrefit_report = my_report,
             plot_viewer= FALSE,
             plot_save = FALSE)
```

PlotChrCluster

PlotChrCluster

Description

The function clusters chromosomes based on the copy number (CN) and returns a graph where it is possible to observe the different groups and two data frames (report and plot_table). See the vignette for the data frame descriptions.

Usage

```
PlotChrCluster(
  segs,
  clust_method = "ward.D2",
  plot_output = TRUE,
  plot_viewer = TRUE,
  plot_save = FALSE,
  plot_format = "png",
  plot_path,
  verbose = FALSE
)
```

Arguments

segs	data.frame with segments of samples. It must be formatted with correct column names (start, end, ID)
clust_method	clustering method. Default is "ward.D2"
plot_output	Whether to plot refitted profiles (logical)
plot_viewer	Logical parameter. When it is TRUE, the function print the output plot in the R viewer. By default is TRUE.
plot_save	Logical parameter. When it is TRUE, the function save the plot in the chosen path and format. By default is TRUE.
plot_format	File format for the output plots (accepts "png", "jpg", "pdf", "tiff"). By default is "png"
plot_path	Path to save output plots.
verbose	print information about the processes of the function. By default is FALSE

Value

Plot with chromosomes clustered

Examples

```
data(TCGA_BRCA_CN_segments)
Cluster <- PlotChrCluster(segs=TCGA_BRCA_CN_segments,
                          clust_method= "ward.D2",
                          plot_output=FALSE)
```

Popeye

Popeye

Description

The function assign the chromosomal arm to each segment.

Usage

```
Popeye(segments)
```

Arguments

segments data.frame formatted with correct column names (see package vignette)

Value

Return a data frame containg segments with the arm annotation.

Examples

```
data("TCGA_BRCA_CN_segments")
data <- TCGA_BRCA_CN_segments[1:9] #as it already presents the arm column
data_annotated <- Popeye(segments = data)
```

TCGA_BRCA_CN_segments *Segments of 100 Breast Cancer samples, downloaded from TCGA-BRCA.*

Description

Segments of 100 Breast Cancer samples, downloaded from TCGA-BRCA.

Usage

```
TCGA_BRCA_CN_segments
```

Format

A data frame with 79,607 rows and 12 variables:

chr Chromosome which the segment belong

start Starting point of the segment, in Mb

end Ending point of the segment, in Mb

width Width of the segment, in Mb

strand Strand of the segment

ID Sample name

Num_Probes Probes involved

Segment_Mean LogR of the segments

Sample Barcode of TCGA-BRCA database

arm Arm information, p or q

chrarm Chromosomal arm which the segment belong

CN Segments Copy Number value obtained by the logR

Source

<https://portal.gdc.cancer.gov/projects/TCGA-BRCA>

Index

* **datasets**

TCGA_BRCA_CN_segments, [6](#)

computeNormalChromosomes, [2](#)

DRrefit, [3](#)

DRrefit_plot, [4](#)

PlotChrCluster, [5](#)

Popeye, [6](#)

TCGA_BRCA_CN_segments, [6](#)