Package 'breakpointR'

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```
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Title Find breakpoints in Strand-seq data
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Description
     This package implements functions for finding breakpoints, plotting and export of Strand-seq data.
Depends R (>= 3.5), GenomicRanges, cowplot, breakpointRdata
Imports methods, utils, grDevices, stats, S4Vectors, GenomeInfoDb (>=
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     BiocGenerics, gtools, doParallel, foreach
Suggests knitr, BiocStyle, testthat
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biocViews Software, Sequencing, DNASeq, SingleCell, Coverage
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```

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breakpointR-package

Breakpoint detection in Strand-Seq data

Description

This package implements functions for finding breakpoints, plotting and export of Strand-seq data.

Details

The main function of this package is breakpointr and produces several plots and browser files. If you want to have more fine-grained control over the different steps check the vignette How to use breakpointR.

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Author(s)

David Porubsky, Ashley Sanders, Aaron Taudt

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Description

The BreakPoint object is output of the function runBreakpointr and is basically a list with various entries. The class() attribute of this list was set to "BreakPoint". Entries can be accessed with the list operators '[[]]' and '\$'.

Value

fragments A GRanges-class object with read fragments.

deltas A GRanges-class object with deltaWs.

breaks A GRanges-class object containing the breakpoint coordinates.

counts A GRanges-class object with the regions between breakpoints.

A vector with parameters that were used to obtain the results.

See Also

runBreakpointr

Description

This function is an easy-to-use wrapper to find breakpoints with runBreakpointr in parallel, write the results to file, plot results and find hotspots.

```
breakpointr(
  inputfolder,
  outputfolder,
  configfile = NULL,
  numCPU = 1,
  reuse.existing.files = FALSE,
  windowsize = 1e+06,
  binMethod = "size",
  multi.sizes = NULL,
  pairedEndReads = FALSE,
```

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```
pair2frgm = FALSE,
  chromosomes = NULL,
  min.mapq = 10,
  filtAlt = FALSE,
  genoT = "fisher",
  trim = 10,
  peakTh = 0.33,
  zlim = 3.291,
  background = 0.05,
  minReads = 10,
  maskRegions = NULL,
  callHotSpots = FALSE,
  conf = 0.99
)
```

Arguments

inputfolder Folder with BAM files.

outputfolder Folder to output the results. If it does not exist it will be created.

configfile A file specifying the parameters of this function (without inputfolder, outputfolder

and configfile). Having the parameters in a file can be handy if many samples with the same parameter settings are to be run. If a configfile is specified, it

will take priority over the command line parameters.

numCPU The numbers of CPUs that are used. Should not be more than available on your

machine.

reuse.existing.files

A logical indicating whether or not existing files in outputfolder should be

reused.

windowsize The window size used to calculate deltaWs, either number of reads or genomic

size depending on binMethod.

binMethod Method used to calculate optimal number of reads in the window ("size", "reads").

By default binMethod='size'.

multi.sizes User defined multiplications of the original window size.

pairedEndReads Set to TRUE if you have paired-end reads in your file.

pair2frgm Set to TRUE if every paired-end read should be merged into a single fragment.

chromosomes If only a subset of the chromosomes should be binned, specify them here.

min.mapq Minimum mapping quality when importing from BAM files.

filtAlt Set to TRUE if you want to filter out alternative alignments defined in 'XA' tag.

genoT A method ('fisher' or 'binom') to genotype regions defined by a set of break-

points.

trim The amount of outliers in deltaWs removed to calculate the stdev (10 will re-

move top 10% and bottom 10% of deltaWs).

peakTh The treshold that the peak deltaWs must pass to be considered a breakpoint (e.g.

0.33 is 1/3 of max(deltaW)).

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The number of stdev that the deltaW must pass the peakTh (ensures only significantly higher peaks are considered).

background The percent (e.g. 0.05 = 5%) of background reads allowed for WW or CC genotype calls.

minReads The minimal number of reads between two breaks required for genotyping.

maskRegions List of regions to be excluded from the analysis (tab-separated file: chromosomes start end).

callHotSpots Search for regions of high abundance of breakpoints in single cells.

conf Desired confidence interval of localized breakpoints.

Value

NULL

Author(s)

David Porubsky, Aaron Taudt, Ashley Sanders

Examples

```
## Not run:
## The following call produces plots and genome browser files for all BAM files in "my-data-folder"
breakpointr(inputfolder="my-data-folder", outputfolder="my-output-folder")
## End(Not run)
```

breakpointr2UCSC

Export UCSC browser formated files

Description

Write a bedfile or bedgraph from a breakpointR object for upload on to the UCSC Genome browser.

```
breakpointr2UCSC(
  index,
  outputDirectory,
  fragments = NULL,
  deltaWs = NULL,
  breakTrack = NULL,
  confidenceIntervals = NULL,
  breaksGraph = NULL
)
```

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Arguments

index A character used to name the bedfile(s).

outputDirectory

Location to write bedfile(s).

fragments A GRanges-class object with strand and mapq metadata, such as that generated

by readBamFileAsGRanges

deltaWs A GRanges-class object with metadata column "deltaW" generated by deltaWCalculator.

breakTrack A GRanges-class object with metadata "genoT" (e.g. newBreaks) will write a

bedtrack with refined breakpoints.

confidenceIntervals

A GRanges-class object with metadata "genoT" the same length as breakTrack (e.g. confint) will write a bedtrack with breakpoints confidence intervals.

breaksGraph A GRanges-class object.

Value

NULL

Author(s)

Ashley Sanders, David Porubsky, Aaron Taudt

Examples

```
## Get an example file
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
exampleFile <- list.files(exampleFolder, full.names=TRUE)[1]
## Load the file
brkpts <- get(load(exampleFile))
## Write results to BED files
breakpointr2UCSC(index='testfile', outputDirectory=tempdir(), breakTrack=brkpts$breaks)</pre>
```

breakSeekr

Find breakpoints from deltaWs

Description

Find breakpoints from deltaWs by localizing significant peaks based on z-score calculation.

```
breakSeekr(deltaWs, trim = 10, peakTh = 0.33, zlim = 3.291)
```

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Arguments

deltaWs	A GRanges-class object with metadata column "deltaW" generated by deltaWCalculator.
trim	The amount of outliers in deltaWs removed to calculate the stdev (10 will remove top 10% and bottom 10% of deltaWs).
peakTh	The treshold that the peak deltaWs must pass to be considered a breakpoint (e.g. 0.33 is 1/3 of max(deltaW)).
zlim	The number of stdev that the deltaW must pass the peakTh (ensures only significantly higher peaks are considered).

Value

A GRanges-class object containing breakpoint coordinates with various metadata columns.

Author(s)

David Porubsky, Aaron Taudt, Ashley Sanders

Examples

```
## Get an example file
exampleFolder <- system.file("extdata", "example_bams", package="breakpointRdata")
exampleFile <- list.files(exampleFolder, full.names=TRUE)[1]
## Load the file
fragments <- readBamFileAsGRanges(exampleFile, pairedEndReads=FALSE, chromosomes='chr22')
## Calculate deltaW values
dw <- deltaWCalculator(fragments)
## Get significant peaks in deltaW values
breaks <- breakSeekr(dw)</pre>
```

collapseBins

Collapse consecutive bins with the same ID value

Description

Collapse consecutive bins with the same value defined in 'id.field'.

Usage

```
collapseBins(gr, id.field = 3)
```

Arguments

gr A GRanges-class object.

id.field A number of metadata column to use for region merging.

Value

A GRanges-class object.

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confidenceInterval

Estimate confidence intervals for breakpoints

Description

Estimate confidence intervals for breakpoints by going outwards from the breakpoint read by read, and multiplying the probability that the read doesn't belong to the assigned segment.

Usage

```
confidenceInterval(breaks, fragments, background = 0.05, conf = 0.99)
```

Arguments

breaks Genotyped breakpoints as outputted from function GenotypeBreaks.

fragments Read fragments from function readBamFileAsGRanges.

background The percent (e.g. 0.05 = 5%) of background reads allowed for WW or CC

genotype calls.

conf Desired confidence interval of localized breakpoints.

Value

A GRanges-class object of breakpoint ranges for a given confidence interval in conf.

Author(s)

Aaron Taudt, David Porubsky

Examples

```
## Not run:
## Get an example file
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
exampleFile <- list.files(exampleFolder, full.names=TRUE)[1]
## Load the file
breakpoint.objects <- get(load(exampleFile))
## Calculate confidence intervals of genotyped breakpoints
confint <- confidenceInterval(breaks=breakpoint.objects$breaks, fragments=breakpoint.objects$fragments, background
## End(Not run)</pre>
```

confidenceInterval.binomial

confidenceInterval.binomial

Estimate confidence intervals for breakpoints

Description

Estimate confidence intervals for breakpoints by going outwards from the breakpoint read by read, and performing a binomial test of getting the observed or a more extreme outcome, given that the reads within the confidence interval belong to the other side of the breakpoint.

Usage

```
confidenceInterval.binomial(breaks, fragments, background = 0.02, conf = 0.99)
```

Arguments

breaks Genotyped breakpoints as outputted from function GenotypeBreaks.

fragments Read fragments from function readBamFileAsGRanges.

background The percent (e.g. 0.05 = 5%) of background reads allowed for WW or CC

genotype calls.

conf Desired confidence interval of localized breakpoints.

Value

A GRanges-class object of breakpoint ranges for a given confidence interval in conf.

Author(s)

Aaron Taudt, David Porubsky

Examples

```
## Not run:
## Get an example file
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
exampleFile <- list.files(exampleFolder, full.names=TRUE)[1]
## Load the file
breakpoint.objects <- get(load(exampleFile))
## Calculate confidence intervals of genotyped breakpoints
confint <- confidenceInterval.binomial(breakpoint.objects$breaks, breakpoint.objects$fragments, background=0.02]
## End(Not run)</pre>
```

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Description

This function will move through BAM files in a folder, read in each individual file and go through each chromosome, determine if the chromosome is WW or CC based on WCcutoff, reverse complement all reads in the WW file, append to a new composite file for that chromosome, order the composite file of each chromosome based on position.

Usage

```
createCompositeFile(
  file.list,
  chromosomes = NULL,
  pairedEndReads = TRUE,
  pair2frgm = FALSE,
  min.mapq = 10,
  filtAlt = FALSE,
  WC.cutoff = 0.9,
  genoT = "fisher",
  background = 0.05
)
```

Arguments

file.list	A list of BAM fi	les to process.
-----------	------------------	-----------------

chromosomes If only a subset of the chromosomes should be binned, specify them here.

pairedEndReads Set to TRUE if you have paired-end reads in your file.

pair2frgm Set to TRUE if every paired-end read should be merged into a single fragment.

min.mapq Minimum mapping quality when importing from BAM files.

filtAlt Set to TRUE if you want to filter out alternative alignments defined in 'XA' tag.

WC.cutoff Percentage of WW or CC reads to consider chromosome being WW or CC

genoT A method ('fisher' or 'binom') to genotype regions defined by a set of break-

points.

background The percent (e.g. 0.05 = 5%) of background reads allowed for WW or CC

genotype calls.

Value

A GRanges-class object.

Author(s)

Ashley Sanders, David Porubsky

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deltaWCalculator

Calculate deltaWs

Description

This function will calculate deltaWs from a GRanges-class object with read fragments.

Usage

```
deltaWCalculator(frags, reads.per.window = 100)
```

Arguments

```
frags A GRanges-class with read fragments (see readBamFileAsGRanges).
reads.per.window
Number of reads in each dynamic window.
```

Value

The input frags with additional meta-data columns.

Author(s)

Aaron Taudt

See Also

readBamFileAsGRanges

Examples

```
## Get an example file
exampleFolder <- system.file("extdata", "example_bams", package="breakpointRdata")
exampleFile <- list.files(exampleFolder, full.names=TRUE)[1]
## Load the file
fragments <- readBamFileAsGRanges(exampleFile, pairedEndReads=FALSE, chromosomes='chr22')
## Calculate deltaW values
dw <- deltaWCalculator(fragments)</pre>
```

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deltaWCalculatorVariousWindows

Calculate deltaWs using various window sizes

Description

This function will calculate deltaWs from a GRanges-class object with read fragments.

Usage

```
deltaWCalculatorVariousWindows(
  frags,
  reads.per.window = 100,
  multi.sizes = c(2, 4, 6)
)
```

Arguments

```
\label{eq:class} Frags \qquad A \ GRanges-class \ with \ read \ fragments \ (see \ readBamFileAsGRanges). reads.per.window \qquad \qquad Number \ of \ reads \ in \ each \ dynamic \ window.
```

multi.sizes User defined multiplications of the original window size.

Value

The input frags with additional meta-data columns.

Author(s)

David Porubsky

See Also

deltaWCalculator

exportRegions

Function to print WC regions after breakpointR analysis

Description

Function to print WC regions after breakpointR analysis

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Usage

```
exportRegions(
  datapath,
  file = NULL,
  collapseInversions = FALSE,
  collapseRegionSize = 5e+06,
  minRegionSize = 5e+06,
  state = "wc"
)
```

Arguments

datapath A path to that

file A filename to print exported regions to.

collapseInversions

Set to TRUE if you want to collapse putative inverted regions.

collapseRegionSize

Upper range of what sized regions should be collapsed.

minRegionSize Minimal size of the region to be reported.

state A genotype of the regions to be exported ('ww', 'cc' or 'wc').

Value

A data. frame object containing all regions with user defined 'state'.

Author(s)

David Porubsky

Examples

```
## Get an example file
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
## To export regions genotyped as 'wc'
wc.regions <- exportRegions(datapath=exampleFolder, collapseInversions=FALSE, minRegionSize=5000000, state='wc')</pre>
```

genotype.binom

Assign states to any given region using binomial test.

Description

Assign states to any given region using binomial test.

```
genotype.binom(wReads, cReads, background = 0.05, minReads = 10, log = FALSE)
```

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Arguments

wReads Number of Watson reads. cReads Number of Crick reads.

background The percent (e.g. 0.05 = 5%) of background reads allowed for WW or CC

genotype calls.

minReads The minimal number of reads between two breaks required for genotyping.

log Set to TRUE if you want to calculate probability in log space.

Value

A list with the \$bestFit and \$pval.

Author(s)

David Porubsky

Examples

```
## Get Crick and Watson read counts
## Crick read count
cReads <- 30
## Watson read count
wReads <- 5
genotype.binom(cReads = cReads, wReads = wReads, background = 0.05, minReads = 10, log = TRUE)</pre>
```

genotype.fisher

Assign states to any given region using Fisher Exact Test.

Description

Assign states to any given region using Fisher Exact Test.

Usage

```
genotype.fisher(cReads, wReads, roiReads, background = 0.05, minReads = 10)
```

Arguments

cReads Number of Crick reads. wReads Number of Watson reads.

roiReads Total number of Crick and Watson reads.

background The percent (e.g. 0.05 = 5%) of background reads allowed for WW or CC

genotype calls.

minReads The minimal number of reads between two breaks required for genotyping.

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Value

A list with the \$bestFit and \$pval.

Author(s)

David Porubsky, Aaron Taudt

Examples

```
## Get Crick and Watson read count
## Crick read count
cReads <- 30
## Watson read count
wReads <- 5
genotype.fisher(cReads = cReads, wReads = wReads, roiReads = cReads + wReads, background = 0.05, minReads = 10)</pre>
```

genotyping

Set of functions to genotype regions in between localized breakpoints

Description

Each defined region is given one of the three states ('ww', 'cc' or 'wc') Consecutive regions with the same state are collapsed

Usage

```
GenotypeBreaks(
  breaks,
  fragments,
  background = 0.05,
  minReads = 10,
  genoT = "fisher"
)
```

Arguments

breaks A GRanges-class object with breakpoint coordinates.

fragments A GRanges-class object with read fragments.

background The percent (e.g. 0.05 = 5%) of background reads allowed for WW or CC

genotype calls.

minReads The minimal number of reads between two breaks required for genotyping.

genoT A method ('fisher' or 'binom') to genotype regions defined by a set of break-

points.

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Details

Function GenotypeBreaks exports states of each region defined by breakpoints. Function genotype.fisher assigns states to each region based on expected counts of Watson and Crick reads. Function genotype.binom assigns states to each region based on expected counts of Watson and Crick reads.

Value

A GRanges-class object with genotyped breakpoint coordinates.

Functions

• GenotypeBreaks: Genotypes breakpoint defined regions.

Author(s)

David Porubsky, Ashley Sanders, Aaron Taudt

Examples

```
## Get an example file
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
exampleFile <- list.files(exampleFolder, full.names=TRUE)[1]
## Load the file
breakpoint.objects <- get(load(exampleFile))
## Genotype regions between breakpoints
gbreaks <- GenotypeBreaks(breaks=breakpoint.objects$breaks, fragments=breakpoint.objects$fragments)</pre>
```

hotspotter

Find hotspots of genomic events

Description

Find hotspots of genomic events by using kernel density estimation.

Usage

```
hotspotter(gr.list, bw, pval = 1e-08)
```

Arguments

gr.list A list or GRangesList-class with GRanges-class object containing the coor-

dinates of the genomic events.

bw Bandwidth used for kernel density estimation (see density).

pval P-value cutoff for hotspots.

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Details

The hotspotter uses density to perform a KDE. A p-value is calculated by comparing the density profile of the genomic events with the density profile of a randomly subsampled set of genomic events. Due to this random sampling, the result can vary for each function call, most likely for hotspots whose p-value is close to the specified pval.

Value

A GRanges-class object containing coordinates of hotspots with p-values.

Author(s)

Aaron Taudt

Examples

```
## Get example BreakPoint objects
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
exampleFiles <- list.files(exampleFolder, full.names=TRUE)
breakpoint.objects <- loadFromFiles(exampleFiles)
## Extract breakpoint coordinates
breaks <- lapply(breakpoint.objects, '[[', 'breaks')
## Get hotspot coordinates
hotspots <- hotspotter(gr.list=breaks, bw=1e6)</pre>
```

insertchr

Insert chromosome for in case it's missing

Description

Add two columns with transformed genomic coordinates to the GRanges-class object. This is useful for making genomewide plots.

Usage

```
insertchr(gr)
```

Arguments

gr

A GRanges-class object.

Value

The input GRanges-class object with an additional metadata column containing chromosome name with 'chr'.

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Load breakpointR objects from file

Description

Wrapper to load **breakpointR** objects from file and check the class of the loaded objects.

Usage

```
loadFromFiles(files, check.class = c("GRanges", "BreakPoint"))
```

Arguments

files A list of GRanges-class or BreakPoint objects or a vector of files that contain

such objects.

check.class Any combination of c('GRanges', 'BreakPoint'). If any of the loaded ob-

jects does not belong to the specified class, an error is thrown.

Value

A list of GRanges-class or BreakPoint objects.

Examples

```
## Get some files that you want to load
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
exampleFiles <- list.files(exampleFolder, full.names=TRUE)
## Load the processed data
breakpoint.objects <- loadFromFiles(exampleFiles)</pre>
```

plotBreakpoints

Plotting genome-wide ideograms breakpointR

Description

This function will create genome-wide ideograms from a BreakPoint object.

Usage

```
plotBreakpoints(files2plot, file = NULL)
```

Arguments

files2plot A list of files that contains BreakPoint objects or a single BreakPoint object.

file Name of the file to plot to.

plotBreakpointsPerChr

Value

A list with ggplot objects.

Author(s)

David Porubsky, Aaron Taudt, Ashley Sanders

Examples

```
## Get an example file
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
exampleFile <- list.files(exampleFolder, full.names=TRUE)[1]
## Plot the file
plotBreakpoints(files2plot=exampleFile)</pre>
```

plotBreakpointsPerChr Plotting chromosome specific ideograms breakpointR

Description

This function will create chromsome specific enome-wide ideograms from a BreakPoint object.

Usage

```
plotBreakpointsPerChr(files2plot, plotspath = NULL, chromosomes = NULL)
```

Arguments

files2plot A list of files that contains BreakPoint objects or a single BreakPoint object.

plotspath Directory to store plots.

chromosomes Set specific chromosome(s) to be plotted.

Value

A list with ggplot objects.

Author(s)

David Porubsky

Examples

```
## Get an example file
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
exampleFiles <- list.files(exampleFolder, full.names=TRUE)
## Plot results
plotBreakpointsPerChr(exampleFiles, chromosomes='chr7')</pre>
```

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Description

Plot a genome-wide heatmap of template inheritance states from a BreakPoint object.

Usage

```
plotHeatmap(files2plot, file = NULL, hotspots = NULL)
```

Arguments

files2plot A list of files that contains BreakPoint objects or a single BreakPoint object.

file Name of the file to plot to.

hotspots A GRanges-class object with locations of breakpoint hotspots.

Value

A ggplot object.

Author(s)

David Porubsky, Aaron Taudt, Ashley Sanders

Examples

```
## Get example BreakPoint objects to plot
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
exampleFiles <- list.files(exampleFolder, full.names=TRUE)
breakpoint.objects <- loadFromFiles(exampleFiles)
## Plot the heatmap
plotHeatmap(breakpoint.objects)</pre>
```

ranges2UCSC

Generates a bedfile from an input GRanges file

Description

Write a bedfile from Breakpoint.R files for upload on to UCSC Genome browser

```
ranges2UCSC(gr, outputDirectory = ".", index = "bedFile", colorRGB = "0,0,0")
```

Arguments

gr A GRanges-class object with genomic ranges to be exported into UCSC format.

outputDirectory

Location to write bedfile(s).

index A character used to name the bedfile(s).

colorRGB An RGB color to be used for submitted ranges.

Value

NULL

Author(s)

David Porubsky

Examples

```
## Get an example file
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
exampleFile <- list.files(exampleFolder, full.names=TRUE)[1]
## Load the file
counts <- get(load(exampleFile))[['counts']]
## Export 'wc' states into a UCSC formated file
ranges2UCSC(gr=counts[counts$states == 'wc'], index='testfile', outputDirectory=tempdir())</pre>
```

Description

Import aligned reads from a BAM file into a GRanges-class object.

```
readBamFileAsGRanges(
   file,
   bamindex = file,
   chromosomes = NULL,
   pairedEndReads = FALSE,
   min.mapq = 10,
   remove.duplicate.reads = TRUE,
   pair2frgm = FALSE,
   filtAlt = FALSE
)
```

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Arguments

file Bamfile with aligned reads.

bamindex Bam-index file with or without the .bai ending. If this file does not exist it will

be created and a warning is issued.

chromosomes If only a subset of the chromosomes should be binned, specify them here.

pairedEndReads Set to TRUE if you have paired-end reads in your file.

min.mapq Minimum mapping quality when importing from BAM files.

remove.duplicate.reads

A logical indicating whether or not duplicate reads should be kept.

pair2frgm Set to TRUE if every paired-end read should be merged into a single fragment.

filtAlt Set to TRUE if you want to filter out alternative alignments defined in 'XA' tag.

Value

A GRanges-class object.

Author(s)

David Porubsky, Aaron Taudt, Ashley Sanders

Examples

```
## Get an example file
exampleFolder <- system.file("extdata", "example_bams", package="breakpointRdata")
exampleFile <- list.files(exampleFolder, full.names=TRUE)[1]
## Load the file
fragments <- readBamFileAsGRanges(exampleFile, pairedEndReads=FALSE, chromosomes='chr22')</pre>
```

readConfig

Read breakpointR configuration file

Description

Read an breakpointR configuration file into a list structure. The configuration file has to be specified in INI format. R expressions can be used and will be evaluated.

Usage

```
readConfig(configfile)
```

Arguments

configfile Path to the configuration file

Value

A list with one entry for each element in configfile.

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Author(s)

Aaron Taudt

removeDoubleSCEs

Process double SCE chromosomes: with internal WC region.

Description

This function will take from a double SCE chromosome only WW or CC region (Longer region is taken).

Usage

```
removeDoubleSCEs(gr, collapseWidth = 5e+06)
```

Arguments

gr A GRanges-class object.

collapseWidth A segment size to be collapsed with neighbouring segments.

Value

The input GRanges-class object with only WW or CC region retained.

removeReadPileupSpikes

Remove large spikes in short reads coverage

Description

This function takes a GRanges-class object of aligned short reads and removes pockets of reads that are stacked on top of each other based on the maximum number of reads allowed to pileup in 'max.pileup' parameter.

Usage

```
removeReadPileupSpikes(gr = NULL, max.pileup = 30)
```

Arguments

gr A GRanges-class object.

max.pileup A maximum number of reads overlapping each other to be kept.

Value

A GRanges-class object.

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Author(s)

David Porubsky

Examples

```
## Get some files that you want to load
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
infile <- list.files(exampleFolder, full.names=TRUE)[1]
## Read in the reads
breakP.obj <- get(load(infile))
frags <- breakP.obj$fragments
## Remove read spikes
frags <- removeReadPileupSpikes(gr=frags)</pre>
```

runBreakpointr

Find breakpoints in Strand-seq data

Description

Find breakpoints in Strand-seq data. See section Details on how breakpoints are located.

```
runBreakpointr(
 bamfile,
  ID = basename(bamfile),
 pairedEndReads = TRUE,
  chromosomes = NULL,
 windowsize = 1e+06,
 binMethod = "size",
 multi.sizes = NULL,
  trim = 10,
 peakTh = 0.33,
  zlim = 3.291,
  background = 0.05,
 min.mapq = 10,
 pair2frgm = FALSE,
  filtAlt = FALSE,
  genoT = "fisher",
 minReads = 20,
 maskRegions = NULL,
  conf = 0.99
)
```

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Arguments

bamfile A file with aligned reads in BAM format.

ID A character string that will serve as identifier in downstream functions.

pairedEndReads Set to TRUE if you have paired-end reads in your file.

chromosomes If only a subset of the chromosomes should be binned, specify them here.

windowsize The window size used to calculate deltaWs, either number of reads or genomic

size depending on binMethod.

binMethod Method used to calculate optimal number of reads in the window ("size", "reads").

By default binMethod='size'.

multi.sizes User defined multiplications of the original window size.

trim The amount of outliers in deltaWs removed to calculate the stdey (10 will re-

move top 10% and bottom 10% of deltaWs).

peakTh The treshold that the peak deltaWs must pass to be considered a breakpoint (e.g.

0.33 is 1/3 of max(deltaW)).

zlim The number of stdev that the deltaW must pass the peakTh (ensures only signif-

icantly higher peaks are considered).

background The percent (e.g. 0.05 = 5%) of background reads allowed for WW or CC

genotype calls.

min.mapq Minimum mapping quality when importing from BAM files.

pair2frgm Set to TRUE if every paired-end read should be merged into a single fragment.

filtAlt Set to TRUE if you want to filter out alternative alignments defined in 'XA' tag.

genoT A method ('fisher' or 'binom') to genotype regions defined by a set of break-

points.

minReads The minimal number of reads between two breaks required for genotyping.

maskRegions List of regions to be excluded from the analysis (tab-separated file: chromo-

somes start end).

conf Desired confidence interval of localized breakpoints.

Details

Breakpoints are located in the following way:

- 1. calculate deltaWs chromosome-by-chromsome
- 2. localize breaks that pass zlim above the threshold
- 3. genotype both sides of breaks to confirm whether strand state changes
- 4. write a file of _reads, _deltaWs and _breaks in a chr fold -> can upload on to UCSC Genome browser
- write a file for each index with all chromosomes included -> can upload on to UCSC Genome browser

Value

A BreakPoint object.

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Author(s)

David Porubsky, Ashley Sanders, Aaron Taudt

Examples

```
## Get an example file
exampleFolder <- system.file("extdata", "example_bams", package="breakpointRdata")
exampleFile <- list.files(exampleFolder, full.names=TRUE)[1]
## Run breakpointR
brkpts <- runBreakpointr(exampleFile, chromosomes='chr22', pairedEndReads=FALSE)</pre>
```

summarizeBreaks

Compile breakpoint summary table

Description

This function will calculate deltaWs from a GRanges-class object with read fragments.

Usage

```
summarizeBreaks(breakpoints)
```

Arguments

breakpoints A list containing breakpoints stored in GRanges-class object.

Value

A data. frame of compiled breakpoints together with confidence intervals.

Author(s)

David Porubsky

Examples

```
## Get some files that you want to load
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
file <- list.files(exampleFolder, full.names=TRUE)[1]
breakpoints <- get(load(file))[c('breaks', 'confint')]
summarizeBreaks(breakpoints)</pre>
```

synchronizeReadDir 27

synchronizeReadDir Synchronize Strand-seq read directionality

Description

This function aims to synchronize strand directionality of reads that fall into WW and CC regions.

Usage

```
synchronizeReadDir(files2sync, collapseWidth = 5e+06)
```

Arguments

files2sync A list of files that contains BreakPoint objects.

collapseWidth A segment size to be collapsed with neighbouring segments.

Value

A GRanges-class object that reads synchronized by directionality.

Author(s)

David Porubsky

Examples

```
## Get some files that you want to load
exampleFolder <- system.file("extdata", "example_results", package="breakpointRdata")
files2sync <- list.files(exampleFolder, full.names=TRUE)[1]
synchronizeReadDir(files2sync=files2sync)</pre>
```

transCoord

Transform genomic coordinates

Description

Add two columns with transformed genomic coordinates to the GRanges-class object. This is useful for making genomewide plots.

Usage

```
transCoord(gr)
```

Arguments

gr

A GRanges-class object.

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Value

The input GRanges-class with two additional metadata columns 'start.genome' and 'end.genome'.

writeConfig

Write breakpointR configuration file

Description

Write an breakpointR configuration file from a list structure.

Usage

```
writeConfig(config, configfile)
```

Arguments

config

A list structure with parameter values. Each entry will be written in one line.

configfile

Filename of the outputfile.

Value

NULL

Author(s)

Aaron Taudt

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