

Package ‘breakpointR’

January 2, 2025

Type Package

Title Find breakpoints in Strand-seq data

Version 1.24.0

Date 2021-11-20

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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 (>= 1.12.3), IRanges, Rsamtools, GenomicAlignments, ggplot2, BiocGenerics, gtools, doParallel, foreach

Suggests knitr, BiocStyle, testthat

License file LICENSE

LazyLoad yes

VignetteBuilder knitr

biocViews Software, Sequencing, DNaseq, SingleCell, Coverage

URL <https://github.com/daewoooo/BreakPointR>

RoxygenNote 7.2.3

git_url <https://git.bioconductor.org/packages/breakpointR>

git_branch RELEASE_3_20

git_last_commit ba27b90

git_last_commit_date 2024-10-29

Repository Bioconductor 3.20

Date/Publication 2025-01-02

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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](#).

Author(s)

David Porubsky, Ashley Sanders, Aaron Taudt

| | |
|------------|--------------------------|
| BreakPoint | <i>BreakPoint object</i> |
|------------|--------------------------|

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

| | |
|------------------------|--|
| <code>fragments</code> | A <code>GRanges-class</code> object with read fragments. |
| <code>deltas</code> | A <code>GRanges-class</code> object with deltaWs. |
| <code>breaks</code> | A <code>GRanges-class</code> object containing the breakpoint coordinates. |
| <code>counts</code> | A <code>GRanges-class</code> object with the regions between breakpoints. |
| <code>params</code> | A vector with parameters that were used to obtain the results. |

See Also

`runBreakpointR`

| | |
|--------------------------|---|
| <code>breakpointR</code> | <i>Main function for the <code>breakpointR</code> package</i> |
|--------------------------|---|

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.

Usage

```
breakpointR(
  inputfolder,
  outputfolder,
  configfile = NULL,
  numCPU = 1,
  reuse.existing.files = FALSE,
  windowsize = 1e+06,
  binMethod = "size",
  multi.sizes = NULL,
  pairedEndReads = FALSE,
  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 breakpoints. |
| trim | The amount of outliers in deltaWs removed to calculate the stdev (10 will remove top 10% and bottom 10% of deltaWs). |
| peakTh | The threshold 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). |
| 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 formatted files
```

Description

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

Usage

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

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)
```

breakSeekr

*Find breakpoints from deltaWs***Description**

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

Usage

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

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 threshold 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)
```

| | |
|--------------|---|
| collapseBins | <i>Collapse consecutive bins with the same ID value</i> |
|--------------|---|

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.

| | |
|--------------------|--|
| confidenceInterval | <i>Estimate confidence intervals for breakpoints</i> |
|--------------------|--|

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, back
## End(Not run)
```

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)
## End(Not run)
```

createCompositeFile *Create composite Strand-seq file*

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 files 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 breakpoints. |
| 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

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)
```

`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

`frags` A [GRanges-class](#) with read fragments (see [readBamFileAsGRanges](#)).
`reads.per.window` 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

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='')
```

genotype.binom *Assign states to any given region using binomial test.*

Description

Assign states to any given region using binomial test.

Usage

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

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)
```

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. |

Value

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

Author(s)

David Porubsky, Aaron Taudt

Examples

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

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 breakpoints. |

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)
```

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

| | |
|----------------------|---|
| <code>gr.list</code> | A list or GRangesList-class with GRanges-class object containing the coordinates of the genomic events. |
| <code>bw</code> | Bandwidth used for kernel density estimation (see density). |
| <code>pval</code> | P-value cutoff for hotspots. |

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)
```

| | |
|-----------|---|
| insertchr | <i>Insert chromosome for in case it's missing</i> |
|-----------|---|

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'.

| | |
|---------------|--|
| loadFromFiles | <i>Load breakpointR objects from file</i> |
|---------------|--|

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 objects 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)
```

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.

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)
```

plotBreakpointsPerChr *Plotting chromosome specific ideograms* **breakpointR**

Description

This function will create chromosome specific genome-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')
```

plotHeatmap

Genome wide heatmap of template inheritance states

Description

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

Usage

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

Arguments

| | |
|-------------------------|---|
| <code>files2plot</code> | A list of files that contains <code>BreakPoint</code> objects or a single <code>BreakPoint</code> object. |
| <code>file</code> | Name of the file to plot to. |
| <code>hotspots</code> | A <code>GRanges-class</code> 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)
```

| | |
|-------------|---|
| ranges2UCSC | <i>Generates a bedfile from an input GRanges file</i> |
|-------------|---|

Description

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

Usage

```
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 formatted file
ranges2UCSC(gr=counts[counts$states == 'wc'], index='testfile', outputDirectory=tempdir())
```

| | |
|----------------------|-------------------------------------|
| readBamFileAsGRanges | <i>Import BAM file into GRanges</i> |
|----------------------|-------------------------------------|

Description

Import aligned reads from a BAM file into a [GRanges-class](#) object.

Usage

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

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')
```

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.

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.

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)
```

runBreakpointR

Find breakpoints in Strand-seq data

Description

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

Usage

```
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
)
```

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 stdev (10 will remove top 10% and bottom 10% of deltaWs). |
| peakTh | The threshold 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). |
| 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 breakpoints. |
| minReads | The minimal number of reads between two breaks required for genotyping. |
| maskRegions | List of regions to be excluded from the analysis in GRanges-class object. |
| conf | Desired confidence interval of localized breakpoints. |

Details

Breakpoints are located in the following way:

1. calculate deltaWs chromosome-by-chromosome
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
5. write a file for each index with all chromosomes included -> can upload on to UCSC Genome browser

Value

A [BreakPoint](#) object.

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)
```

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)
```

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)
```

| | |
|------------|--------------------------------------|
| transCoord | <i>Transform genomic coordinates</i> |
|------------|--------------------------------------|

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.

Value

The input [GRanges-class](#) with two additional metadata columns 'start.genome' and 'end.genome'.

| | |
|-------------|---|
| writeConfig | <i>Write breakpointR configuration file</i> |
|-------------|---|

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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