# Package 'CNVScope'

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Type Package

**Title** A Versatile Toolkit for Copy Number Variation Relationship Data Analysis and Visualization

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BugReports https://github.com/jamesdalg/CNVScope/issues/

**Depends** R (>= 4.1.0),ggplot2

**Imports** tidyr,reshape2,magrittr, jointseg,shiny,RCurl,foreach, GenomicInteractions,Matrix,OpenImageR,biomaRt,matrixStats, plyr,data.table,dplyr, doParallel,stringr,rtracklayer,Hmisc

**Suggests** knitr, remotes,pwr,ComplexHeatmap,rmarkdown,

HiCseg,igraph,visNetwork,circlize,plotly,

InteractionSet,GenomicRanges,GenomicFeatures,IRanges,rslurm, shinythemes,shinycssloaders,DT,logging,heatmaply, S4Vectors,BiocManager,shinyjs,htmltools,htmlwidgets,

GenomeInfoDb,BSgenome.Hsapiens.UCSC.hg19,tibble,smoothie

#### VignetteBuilder knitr

URL https://github.com/jamesdalg/CNVScope/

#### biocViews

#### **Description**

Provides the ability to create interaction maps, discover CNV map domains (edges), gene annotate interactions, and create interactive visualizations of these CNV interaction maps.

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averageMatrixEdges

Average edges of a matrix to facilitate downsampling.

## Description

Averages the columns and rows of a matrix by a certain amount.

## Usage

```
averageMatrixEdges(unchangedmatrix, nedges = 1, dimension = c("row", "column"))
```

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#### **Arguments**

unchangedmatrix

A matrix to have edges averaged with genomic coordinates in the form chr1\_50\_100

set as the column and row names.

nedges The number of edges to be averaged

dimension Selectively averages edges in one dimension. Performs symmetric edge averag-

ing by default.

#### Value

averaged\_matrix A matrix with edges averaged, which may be more amenable to downsampling

#### **Examples**

```
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
dim(nbl_result_matrix_sign_small)
nbl_result_matrix_sign_small_avg<-averageMatrixEdges(nbl_result_matrix_sign_small,
nedges=1,dimension="row")
dim(nbl_result_matrix_sign_small_avg)
nbl_result_matrix_sign_small_avg<-averageMatrixEdges(nbl_result_matrix_sign_small,
nedges=1,dimension="column")
dim(nbl_result_matrix_sign_small_avg)</pre>
```

calcCNVKernelProbDist Calculate the probability distribution of CNV concordance events with a fast kernel

## Description

This function produces several matrices, including a Z-score matrix from a matrix of the same size and a percentile matrix of these Z-scores

#### **Arguments**

submatrix A matrix of CNV data in an intrachromosomal region (e.g. chr1 vs chr1 or chr5

vs chr5)

win a window size for the matrix that calculates the windowed average using the

kernel function

debug extra output for debugging.

parallel use parallelization using mcmapply and doParallel?

mcmcores The number of cores used for parallelization.

```
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
mat_prob_dist<-calcCNVKernelProbDist(nbl_result_matrix_sign_small,parallel=FALSE)
mat_prob_dist</pre>
```

4 calc VecLMs

Create a linear regression matrix.

## Description

Creates a matrix of linear regression p-values, log transformed from every combination of columns in the parent matrix.

## Usage

```
calcVecLMs(
  bin_data,
  use_slurm = F,
  job_finished = F,
  slurmjob = NULL,
  n_nodes = NULL,
  cpus_on_each_node = 2,
  memory_per_node = "2g",
  walltime = "4:00:00",
  partitions = "ccr,quick"
)
```

#### **Arguments**

	bin_data	The parent matrix, with columns to have linear regression performed on them.	
	use_slurm	Paralleize over a number of slurm HPC jobs? If false, the program will simply run locally.	
	job_finished	Are all the slurm jobs finished and the results need retrieving?	
	slurmjob	the slurm job object produced by rslurm::slurm_apply(), after running the function initially.	
	n_nodes	the number of nodes used in your slurm job.	
cpus_on_each_node			
		The number of cpus used on each node	
	memory_per_node		
		the amount of ram per node (e.g. "32g" or "2g")	
	walltime	Time for job to be completed for SLURM scheduler in hh:mm:ss format. Defaults to $4h$ .	
	partitions	the partitions to which the jobs are to be scheduled, in order of priority.	

## Value

The output matrix, or if using slurm, the slurm job object (which should be saved as an rds file and reloaded when creating the output matrix).

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#### **Examples**

```
#small example
#bin_data<-matrix(runif(5*5),ncol=5)
foreach::registerDoSEQ()
#full_matrix<-suppressWarnings(calcVecLMs(bin_data))
#Please note that lm() will make a warning when there are two vectors that are too close
#numerically (this will always happen along the diagonal).
#This is normal behavior and is controlled & accounted for using this function as well as
#the postProcessLinRegMatrix function (which converts the infinite values to a maximum).</pre>
```

CNVScopeserver

Server component of the CNVScope plotly shiny application.

## **Description**

Server function of the CNVScope shiny application. run with runCNVScopeShiny

## **Arguments**

session The shiny session object for the application.

input shiny server input

output shiny server output

debug enable debugging mode

#### Value

None

```
## Not run:
runCNVScopeShiny()
## End(Not run)
```

createChromosomalMatrixSet

Create chromosomal interaction matrices for CNVScope shiny application.

## Description

Takes a linear regression matrix and sets infinites to a finite value, and changes the sign to match the sign of the correlation for each value.

#### Usage

```
createChromosomalMatrixSet(
  whole_genome_mat,
  output_dir = NULL,
  prefix = "nbl_"
)
```

#### **Arguments**

whole\_genome\_mat

The matrix containing all of the data, from which the individual matrices will be

spii

output\_dir the folder where the matrices in RData format, will be written.

prefix filename prefix for individual matrices. Default: "nbl\_"

## Value

The list of files already written to disk, with full filenames and paths.

```
#examples for this function would be too large to
#include and should be run on an HPC machine node.
#illustration of this process is shown clearly in
#the vignette and can be done if a user properly
#follows the instructions.
# The function is intended to be run on a whole interactome matrix (chr1-X).
```

divisors 7

divisors List of Divisors

## **Description**

Generates a list of divisors of an integer number. Identical to the same function within the numbers package. The code has been modified from the numbers package, following GPL 3.0 guidelines on 3/30/2022, section 5. Reference for GPL v3.0 LICENSE: https://www.gnu.org/licenses/gpl-3.0.en.html.

## Usage

```
divisors(n)
```

## **Arguments**

n

an integer whose divisors will be generated.

#### Value

Returns a vector integers.

#### See Also

[numbers::divisors()]

## **Examples**

```
divisors(1) # 1
divisors(2) # 1 2
divisors(3) # 1 2 3
divisors(2^5) # 1 2 4 8 16 32
divisors(1000) # 1 2 4 5 8 10 ... 100 125 200 250 500 1000
divisors(1001) # 1 7 11 13 77 91 143 1001
```

 ${\tt downsample\_genomic\_matrix}$ 

Rescale positive and negative data, preserving sign information.

## **Description**

Downsamples a matrix by a specified factor.

8 extractNegLogPval

#### Arguments

whole\_matrix A matrix to be downsampled, on a single chromosome downsamplefactor

A factor by which to reduce the matrix. Must be something that both the row and columns can be divisible by.

singlechromosome

Single chromosome mode; Multi-chromosome not yet implemented (leave T)

#### Value

whole\_matrix\_dsamp A downsampled matrix.

#### **Examples**

```
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
downsample_genomic_matrix(whole_matrix=nbl_result_matrix_sign_small,
downsamplefactor=5,singlechromosome=TRUE)
```

extractNegLogPval

Find the negative log p-value of a pair of vectors.

#### **Description**

Finds the negative log p-value of a matrix, if it exists. Checks first to see if there is a p-value to return.

#### Usage

```
extractNegLogPval(x, y, repval = 300, lowrepval = 0, signed = F)
```

#### **Arguments**

x a vector that is regressed in the fashion y~x. y a vector that is regressed in the fashion y~x.

repval the replacement value if the regression cannot be performed, default 300 (the

vectors are identical if this is used).

lowrepval The low replacement value in the case that a regression p-value is undefined.

signed change the sign of the negative log p-value based on the sign of beta? e.g. if the

line has a negative slope, so will the returned value. If there is a positive slope, there will be a positive negative log p-value. if this option is disabled, then no

sign changes will happen based on the sign of the slope.

#### Value

The negative log p-value or replacement value.

#### **Examples**

```
#small example
xval < -c(1,1,1,1,1)
yval < -c(1,2,3,4,5)
a < -c(3,4,5,6,7)
extractNegLogPval(x=xval,y=yval) #no possible p-value if one vector is constant.
#Some edge cases this may not be correct (if the data lies near a constant),
# but the indiviual sample data should reveal true trends.
suppressWarnings(cor(xval,yval)) #you can't get a correlation value either.
cor(a,a) #gives correlation of 1.
extractNegLogPval(a,a)
#gives replacement value.
suppressWarnings(extractNegLogPval(x=a,y=yval))
#gives 107.3909 and warns about a nearly perfect fit.
```

 $form {\tt Sample Matrix From Raw GDCD} at a$ 

Form sample matrix from GDC copy number data files.

## **Description**

Reads a GDC segmetnation files, adds sample information, and forms a data matrix of samples and bins of a specified size.

## **Arguments**

tcga_files	GDC files to be read
format	file format, TCGA or TARGET.
binsize	the binsize, in base pairs (default 1Mb or 1e6). This value provides a good balance of resolution and speed with memory sensitive applications.
freadskip	the number of lines to skip in the GDC files, typically 14 (the first 13 lines are metadata and the first is a blank line in NBL data). Adjust as needed.
debug	debug mode enable (allows specific breakpoints to be checked).
chromosomes	A vector of chromosomes to be used. Defaults to chr1-chrX, but others can be added e.g. chrY or chrM for Y chromosome or mitochondrial DNA. Format expected is a character vector, e.g. c("chr1", "chr2", "chr3").
sample_pat	Pattern used to extract sample name from filename. Use "" to use the filename.
sample_col	The name of the sample column (for custom format input).
chrlabel	The name of the chromosome column (for custom format input).
startlabel	The name of the start column (for custom format input).
endlabel	The name of the end column (for custom format input).

#### Value

A dataframe containing the aggregated copy number values, based on the parameters provided.

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#### **Examples**

```
#Pipeline examples would be too large to include in package checks.
#please see browseVignettes("CNVScope") for a demonstration.
```

freadGDCfile

Read GDC segmentation datafile for low-pass sequencing data.

#### **Description**

Reads a GDC segmetnation file and extract the segmetnation data.

## Usage

```
freadGDCfile(
   file,
   fread_skip = NULL,
   format = "TARGET",
   CN_colname = "log2",
   sample_pattern = "[^_]+",
   sample_colname = NULL
)
```

#### **Arguments**

file GDC file to be read

fread\_skip The number of metadata lines to be skipped(typically 14) format The format of the files (TCGA,TARGET, or custom).

CN\_colname The name of the column containing the copy number values. sample\_pattern Regex pattern to obtain the sample ID from the filename.

sample\_colname Alternatively, a column can be specified with the sample ID on each line.

#### Value

input\_tsv\_with\_sample\_info A data frame containing the sample information extracted from the filename, including sample name & comparison type.

#### References

https://docs.gdc.cancer.gov/Encyclopedia/pages/TCGA\_Barcode/

```
freadGDCfile(file =
system.file("extdata","somaticCnvSegmentsDiploidBeta_TARGET-30-PANRVJ_NormalVsPrimary.tsv",
package = "CNVScope"))
```

getAnnotationMatrix 11

 $\begin{tabular}{ll} {\it Get the genes in the genomic ranges indicated by the row and column} \\ {\it labels.} \end{tabular}$ 

## **Description**

Gets the genes in the ranges within each cell of the matrix.

## Usage

```
getAnnotationMatrix(
  genomic_matrix,
  prot_only = T,
  sequential = F,
  flip_row_col = F
)
```

#### **Arguments**

```
genomic_matrix A matrix with row and column names of the format chr1_100_200 (chr,start,end)
prot_only Inlcude only the protein coding genes from ensembl?
sequential Turn off parallelism with doParallel?
flip_row_col Give column genes along the rows and row genes down columns?
```

#### Value

concatenated\_gene\_matrix A matrix with row and column genes

## **Examples**

```
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
load(system.file("extdata","ensembl_gene_tx_table_prot.rda",package = "CNVScope"))
load(system.file("extdata","grch37.rda",package = "CNVScope"))
getAnnotationMatrix(genomic_matrix=nbl_result_matrix_sign_small[1:5,1:5],sequential=TRUE,prot_only=TRUE)
```

```
getAsymmetricBlockIndices
```

Get Block Indices from an asymmetric (or symmetric) matrix.

#### **Description**

This function segments a matrix, including asymmetric matrices using multiple imputation (MI) techniques and a segmentation algorithm to generate breakpoints for column and row.

#### Usage

```
getAsymmetricBlockIndices(
  genomicmatrix = NULL,
  algorithm = "HiCseg",
  nb_change_max = 100,
  distrib = "G",
  model = "D",
  MI_strategy = "average",
  transpose = T
)
```

#### **Arguments**

genomicmatrix the large, whole matrix from which blocks are taken

algorithm Algorithm to be used: HiCseg or jointSeg.

nb\_change\_max the maximal number of changepoints, passed to HiCseg (if this algorithm is

used). Note: HiCseg doesn't actually obey this limit. Rather, use it as a param-

eter to increase/decrease segmentation extent.

distrib Passed to Hicseg\_linkC\_R, from their documentation: Distribution of the data:

"B" is for Negative Binomial distribution, "P" is for the Poisson distribution and

"G" is for the Gaussian distribution."

model Passed on to HiCseg\_linkC\_R: "Type of model: "D" for block-diagonal and

"Dplus" for the extended block-diagonal model."

MI\_strategy strategy to make the matrix temporarily symmetric. "average" adds a number of

values equal to the average of the matrix, while copy copies part of the matrix

to the shorter side, making a square matrix.

transpose transpose the matrix and output the breakpoints? Some segmentation algorithms

(e.g. HiCseg) produces different results when used against the transposed version of the matrix, as it expects symmetry. This allows the output of additional breakpoints Users can choose to take intersect() or union() on the results to get

conserved changepoints or additional changepoints, depending on need.

#### Value

An output list of the following:

breakpoints col A vector of breakpoints for the columns.

breakpoints\_row A vector of breakpoints for the rows.

breakpoints\_col A vector of breakpoints for columns on the transposed genomic matrix.

breakpoints\_row A vector of breakpoints for the rows on the transposed genomic matrix.

```
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
submatrix_tiny<-nbl_result_matrix_sign_small
tiny_test<-getAsymmetricBlockIndices(submatrix_tiny,nb_change_max=10,algorithm="jointSeg")</pre>
```

```
## Not run:
submatrix_wide<-submatrix_tiny[1:5,]</pre>
submatrix_narrow<-submatrix_tiny[,1:5]</pre>
wide_test<-getAsymmetricBlockIndices(submatrix_wide,distrib = "G",model = "Dplus",</pre>
nb\_change\_max = 1e4)
 #the below work, but the time to run all of these would be greater than 10 seconds..
random_wide<-matrix(runif(n = 400*200),ncol=400,nrow=200)</pre>
random_narrow<-matrix(runif(n = 400*200),ncol=200,nrow=400)</pre>
random_wide_test_avg<-getAsymmetricBlockIndices(random_wide,</pre>
distrib = "G",model = "Dplus",nb_change_max = 1e4)
random_narrow_test_avg<-getAsymmetricBlockIndices(random_narrow,</pre>
distrib = "G",model = "Dplus",nb_change_max = 1e4)
random_wide_test_copy<-getAsymmetricBlockIndices(random_wide,</pre>
distrib = "G",model = "Dplus",nb_change_max = 1e4,MI_strategy = "copy")
random_narrow_test_copy<-getAsymmetricBlockIndices(random_narrow,</pre>
distrib = "G",model = "Dplus",nb_change_max = 1e4,MI_strategy = "copy")
genomicmatrix=random_narrow
nb_change_max=100
model = "D"
distrib = "G"
MI_strategy="copy"
#question-- does it pick different breakpoints if transposed first?
#Answer: yes, at least in Dplus model.
rm(genomicmatrix)
rm(model)
rm(distrib)
rm(MI_strategy)
random_wide_test_copy<-getAsymmetricBlockIndices(genomicmatrix = random_wide,</pre>
                                                   distrib = "G",
                                model = "Dplus",nb_change_max = 1e2,MI_strategy = "copy")
random_narrow_test_copy<-getAsymmetricBlockIndices(random_narrow,distrib = "G",
                                                     model = "Dplus",
                                                 nb_change_max = 1e2,MI_strategy = "copy")
random_wide_test_copy_t<-getAsymmetricBlockIndices(genomicmatrix = t(random_wide),</pre>
                                                    distrib = "G", model = "Dplus",
                                                 nb_change_max = 1e2,MI_strategy = "copy")
random_narrow_test_copy_t<-getAsymmetricBlockIndices(genomicmatrix = t(random_narrow),</pre>
                                                      distrib = "G",model = "Dplus",
                                                 nb_change_max = 1e2,MI_strategy = "copy")
length(intersect(random_wide_test_copy$breakpoints_col,
random_wide_test_copy_t$breakpoints_row))/length(unique(c(random_wide_test_copy$breakpoints_col,
random_wide_test_copy_t$breakpoints_row)))
random_wide_test_copy_with_transpose<-getAsymmetricBlockIndices(genomicmatrix = random_wide,</pre>
distrib = "G",model = "Dplus",nb_change_max = 1e2,MI_strategy = "copy",transpose = T)
random_narrow_test_copy_with_transpose<-getAsymmetricBlockIndices(genomicmatrix = random_narrow,
 distrib = "G",model = "Dplus",nb_change_max = 1e2,MI_strategy = "copy",transpose = T)
random_narrow_test_copy_with_transpose<-getAsymmetricBlockIndices(genomicmatrix = random_narrow,</pre>
distrib = "G", model = "Dplus", nb_change_max = 1e2, MI_strategy = "copy", transpose = T)
conserved_breakpoints_col<-intersect(random_narrow_test_copy_with_transpose$breakpoints_col,
 random_narrow_test_copy_with_transpose$t_breakpoints_row)
conserved_breakpoints_row<-intersect(random_narrow_test_copy_with_transpose$breakpoints_row,
 random_narrow_test_copy_with_transpose$t_breakpoints_col)
random_wide_test_copy_with_transpose<-getAsymmetricBlockIndices(genomicmatrix = random_wide,</pre>
```

```
distrib = "G",model = "Dplus",nb_change_max = 1e2,MI_strategy = "copy",transpose = T)
conserved_breakpoints_col<-intersect(random_wide_test_copy_with_transpose$breakpoints_col,
    random_wide_test_copy_with_transpose$t_breakpoints_row)
conserved_breakpoints_row<-intersect(random_wide_test_copy_with_transpose$breakpoints_row,
    random_wide_test_copy_with_transpose$t_breakpoints_col)
## End(Not run)</pre>
```

getBlockAverageMatrixFromBreakpoints

Calculate block averages and areas in a matrix given breakpoints.

## Description

This function produces several matrix outputs of averages and areas of matrix blocks, given a pair of vectors for breakpoints.

#### **Arguments**

whole\_matrix the large, whole matrix from which blocks are taken

breakpoints\_col

An integer list of column breakpoints, including 1 and the number of columns in the whole matrix.

breakpoints\_row

outputs

An integer list of row breakpoints, including 1 and the number of rows in the

A list of the following possible outputs (default all): "blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index","blockaverages\_reformatted\_by\_index\_in

whole matrix.

or "blockaverages\_matrix\_label\_area"

#### Value

An output list of the following:

blockaverages\_reformatted\_by\_index a matrix of the block averages and areas, in long format, with indexes used to generate the averages.

blockaverages\_reformatted\_by\_label a matrix of the block averages and areas, in long format, with labels of the indexes used to generate the averages.

blockaverages\_matrix\_idx\_area a matrix of the block areas, with indexes based on the original row/col index used to generate the data.

blockaverages\_matrix\_idx\_avg a matrix of the block averages, with indexes based on the original row/col index used to generate the data.

blockaverages\_matrix\_label\_area a matrix of the block areas, with indexes based on the original row/col label used to generate the data.

blockaverages\_matrix\_label\_avg a matrix of the block averages, with indexes based on the original row/col label used to generate the data.

getGlobalRescalingStats

#### **Examples**

```
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
set.seed(303)
mat<-matrix(data=runif(n = 25),nrow=5,ncol=5,dimnames = list(c("chr1_0_5000",</pre>
"chr1 5000 10000"."chr1 10000 15000"."chr1 15000 20000"."chr1 20000 25000").
c("chr1_0_5000","chr1_5000_10000","chr1_10000_15000","chr1_15000_20000","chr1_20000_25000")))
breakpoints_col < -c(1,2,4,5)
breakpoints_row < -c(1,2,4,5)
foreach::registerDoSEQ()
getBlockAverageMatrixFromBreakpoints(whole_matrix=mat,breakpoints_col=breakpoints_col,
breakpoints_row=breakpoints_row)
## Not run: #extra examples
mat<-matrix(data=round(runif(min = 0,max=100,n = 25)),nrow=5,ncol=5,</pre>
dimnames = list(c("chr1_0_5000","chr1_5000_10000","chr1_10000_15000","chr1_15000_20000",
"chr1_20000_25000"),c("chr2_0_50000","chr2_50000_100000",
"chr2_100000_150000", "chr2_150000_200000", "chr2_200000_250000")))
breakpoints_col < -c(1,2,4,5)
breakpoints_row<-c(1,2,4,5)</pre>
avg_results<-getBlockAverageMatrixFromBreakpoints(whole_matrix=mat,</pre>
breakpoints_col=breakpoints_col,breakpoints_row=breakpoints_row)
avg_results$blockaverages_reformatted_by_label
avg_results$blockaverages_reformatted_by_index
whole_matrix=mat
mat<-matrix(data=round(runif(min = 0, max=100, n = 25)), nrow=5, ncol=5,</pre>
dimnames = list(c("chr1_0_5000","chr1_5000_10000","chr1_10000_15000",
"chr1_15000_20000", "chr1_20000_25000"), c("chr2_0_50000",
"chr2_50000_100000", "chr2_100000_150000",
"chr2_150000_200000","chr2_200000_250000")))
breakpoints_col <-c(1,2,4,5)
breakpoints_row < -c(1,2,4,5)
avg_results<-getBlockAverageMatrixFromBreakpoints(whole_matrix=mat,</pre>
breakpoints_col=breakpoints_col,breakpoints_row=breakpoints_row)
avg_results$blockaverages_reformatted_by_label
avg_results$blockaverages_reformatted_by_index
whole_matrix=mat
submatrix<-nbl_result_matrix_sign_small</pre>
breakpoints_row_jointseg<-jointseg::jointSeg(submatrix,K=5)$bestBkp</pre>
breakpoints_col_jointseg<-jointseg::jointSeg(t(submatrix),K=5)$bestBkp</pre>
submatrix_avg_results<-getBlockAverageMatrixFromBreakpoints(whole_matrix=submatrix,
breakpoints_col=breakpoints_col_jointseg,breakpoints_row=breakpoints_row_jointseg)
## End(Not run)
```

getGlobalRescalingStats

Calculate several base statistics for color rescaling.

#### **Description**

calculates several statistics from a large matrix that can then be applied to smaller submatrices without needing to load the entire matrix into memmory

#### Usage

```
getGlobalRescalingStats(whole_matrix, saveToDisk = F, output_fn = NULL)
```

#### **Arguments**

whole\_matrix the whole matrix to get stats for.

saveToDisk Save the statistics to disk as an RDS file in the local directory?

output\_fn the name of the output file.

#### Value

A list of the output statistics, including: the global min, max, length, sigma (matrix variance), pos\_sigma (variance of the positive values), neg\_sigma(variance of the negative values), global mean (global\_mu), est\_max\_cap (global\_mu+global\_sigma\_pos\*2), as well as the number of rows and columns of the matrix.

#### **Examples**

```
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
getGlobalRescalingStats(nbl_result_matrix_sign_small)
```

getInterchromosomalInteractivePlot

Create an HTML widget for use in shiny or webshot for a given pair of chromosomes.

#### **Description**

This function requires a matrix with genomic coordinates in the row and column names, and produces a heatmap with a tooltip

#### Arguments

whole\_matrix the large, whole genomic matrix from which the submatrix is taken (rows)

chrom1 The first chromsome used for the map (columns).

chrom2 The second chromsome used for a map axis.

#### Value

An HTML widget.

#### **Examples**

```
## Not run:
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
getInterchromosomalInteractivePlot(whole_matrix=nbl_result_matrix_sign_small,chrom1=1,
chrom2=1)
## End(Not run)
```

GRanges\_to\_underscored\_pos

Convert GRanges object to underscord positions.

## **Description**

This function converts row or column names (or any character vector of the format) into a GenomicRanges object.

#### Usage

```
GRanges_to_underscored_pos(input_gr, minusOneToEnd = T)
```

#### **Arguments**

input\_gr A GenomicRanges object

minusOneToEnd Minus one position to end of each Genomic Range?

## **Examples**

```
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
col_gr<-underscored_pos_to_GRanges(colnames(nbl_result_matrix_sign_small))
GRanges_to_underscored_pos(col_gr)</pre>
```

 ${\tt importBreakpointBed}$ 

Import a breakpoint BED file.

#### **Description**

Imports a BED file with breakpoints or other interactions, in a dual position format.

#### **Arguments**

breakpoint\_fn the filename of the breakpoint bed file

## Value

a Genomic Interactions Object

#### **Examples**

```
importBreakpointBed(breakpoint_fn = system.file("extdata",
"sample_breakpoints.bed",package = "CNVScope"))
```

mathead

Gets a small piece of a matrix (top left corner) for viewing, rather than pulling the first n rows.

## **Description**

Gives a small square of a matrix to get an idea of content rather than grabbing the entire row. When this row is thousands of numbers long, this can be a problem.

#### Usage

```
mathead(mat, n = 6L)
```

#### **Arguments**

mat A matrix.

The length and width of the piece to view.

## Value

averaged\_matrix a small matrix of size n.

## **Examples**

```
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
mathead(nbl_result_matrix_sign_small)
```

```
nbl_result_matrix_sign_small
```

Neuroblastoma sample CNV relationship matrix

## Description

The first 25 Mb of chromosome 1, neuroblastoma copy number signed relation matrix.

#### **Format**

A matrix with 25 rows and 25 variables

#### Source

```
https://gdc.cancer.gov/
```

```
postProcessLinRegMatrix
```

Postprocess linear regression matrix.

#### **Description**

Takes a linear regression matrix and sets infinites to a finite value, and changes the sign to match the sign of the correlation for each value.

## Usage

```
postProcessLinRegMatrix(
   input_matrix,
   LM_mat,
   cor_type = "pearson",
   inf_replacement_val = 300
)
```

## Arguments

input\_matrix The input matrix, which consists of bins and samples (no LM or correlation has been done on the segmentation values)
 LM\_mat The linear regression matrix, with rows and columns consisting of bins and the values being the negative log p-value between them.
 cor\_type The correlation type ("pearson" (linear), "spearman" (rank), "kendall" (also rank-based)). Rank correlations capture nonlinear relationships as well as linear. Passed to stats::cor's method parameter.

inf\_replacement\_val

the value for which infinites are replaced, by default 300.

#### Value

The output matrix, or if using slurm, the slurm job object (which should be saved as an rds file and reloaded when creating the output matrix).

```
inputmat<-matrix(runif(15),nrow=3)
colnames(inputmat)<-c("chr2_1_1000","chr2_1001_2000","chr2_2001_3000","chr2_3001_4000",
"chr2_4001_5000")
rownames(inputmat)<-c("PAFPJK","PAKKAT","PUFFUM")
outputmat<-matrix(runif(15),nrow=3)
outputmat<-cor(inputmat)*matrix(runif(25,-30,500),nrow=5)
diag(outputmat)<-Inf
postProcessLinRegMatrix(input_matrix=t(inputmat),LM_mat=outputmat,cor_type="pearson",
inf_replacement_val=300)</pre>
```

20 rebinGenomicInteractions

rebinGenomicInteractions

Assign GenomicInteractions to a predefined series of bins for row and column, corresponding to a genomic matrix.

#### **Description**

This function allows the user to assign a set of genomicinteractions to a pre-existing matrix with known dimensions and column/row names. It finds the row/column index of each point and produces a merged dataframe with the original annotation columns that correspond to each bin in the matrix, with appropriate labels & indexes.

## **Arguments**

gint A GenomicInteractions object needing to be binned.				
whole_genome_matrix				
	A matrix with underscored positions for column and rownames e.g. chr1_1_5000,chr1_5001_10000. If this is provided, it will override rown/column names and GRanges objects.			
rownames_gr	A Genomic Ranges object created from the whole genome matrix row names in chr_start_end format, e.g. chr1_1_5000. No effect if whole_genome_mattrix is specified.			
colnames_gr	A Genomic Ranges object created from the whole genome matrix column names in chr_start_end format. No effect if whole_genome_mattrix is specified.			
rownames_mat	The row names of the whole_genome_matrix in chr_start_end format.			
colnames_mat	The column names of the whole_genome_matrix in chr_start_end format.			
method	Method to rebin with— can use overlap and nearest methods.Default: nearest.			

```
foreach::registerDoSEQ()
gint_small_chr1<-importBreakpointBed(breakpoint_fn = system.file("extdata",
    "sample_breakpoints_chr1.bed",package = "CNVScope"))
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
rebinGenomicInteractions(gint=gint_small_chr1,whole_genome_matrix=NULL,
    rownames_gr=underscored_pos_to_GRanges(rownames(nbl_result_matrix_sign_small)),
    colnames_gr=underscored_pos_to_GRanges(colnames(nbl_result_matrix_sign_small)),
    rownames_mat = rownames(nbl_result_matrix_sign_small),
    colnames_mat = colnames(nbl_result_matrix_sign_small),
    method="nearest")</pre>
```

runCNVScopeLocal 21

runCNVScopeLocal

Runs the CNVScope plotly shiny application.

## Description

Runs the interactive suite of tools locally.

## Usage

```
runCNVScopeLocal()
```

#### Value

none. Runs the application if the correct files are present.

## **Examples**

```
## Not run:
CNVScope::runCNVScopeLocal()
## End(Not run)
```

runCNVScopeShiny

Runs the CNVScope plotly shiny application.

## **Description**

Runs the interactive suite of tools locally or on a server if called in a script file (e.g. App.R). Data sources are required. For a simple installation, please use the runCNVScopeLocal function.

## Usage

```
runCNVScopeShiny(
  baseurl = NULL,
  basefn = NULL,
  osteofn = NULL,
  debug = F,
  useCNVScopePublicData = F
)
```

22 signedRescale

## **Arguments**

baseurl the url of the source files for the application (e.g. the contents of plotly\_dashboard\_ext).

This will be pulled from remotely.

basefn the linux file path of the same source files.

osteofn the linux file path of the OS files.

debug Enable debugging output.

useCNVScopePublicData

Use files from the CNVScopePublicData package.

#### Value

none. Runs the application if the correct files are present.

## **Examples**

```
#see runCNVScopeLocal(useCNVScopePublicData=T).
## Not run:
runCNVScopeShiny(useCNVScopePublicData=T)
## End(Not run)
```

signedRescale

Rescale positive and negative data, preserving sign information.

## Description

Performs a signed rescale on the data, shrinking the negative and positive ranges into the [0,1] space, such that negative is always less than 0.5 and positive is always greater.

#### **Usage**

```
signedRescale(
  matrix,
  global_max = NULL,
  global_min = NULL,
  global_sigma = NULL,
  global_mu = NULL,
  max_cap = NULL,
  method = "minmax",
  tan_transform = F,
  global_sigma_pos = NULL,
  global_sigma_neg = NULL,
  asymptotic_max = T
)
```

#### **Arguments**

	matrix	A matrix to be transformed
	global_max	the global maximum (used if scaling using statistics from a large matrix upon a submatrix).
	global_min	the global minimum
	global_sigma	the global signma
	global_mu	the global mu
	max_cap	the maximum saturation—decreases the ceiling considered for the scaling function. Useful to see greater differences if an image is too white, increase it if there is too much color to tell apart domains.
	method	method to perform the rescaling. Options are "minmax" (default), "tan" for tangent, and "sd" for standard devation $$
	tan_transform	apply a tangent transformation?
global_sigma_pos		
		The positive global sigma. See getGlobalRescalingStats.
global_sigma_neg		
		The negative global sigma. See getGlobalRescalingStats.
	asymptotic_max	make the maximum value in the matrix not 1, but rather something slightly below.

## Value

transformedmatrix A transformed matrix.

## **Examples**

```
mat<-matrix(c(5,10,15,20,0,40,-45,300,-50),byrow=TRUE,nrow=3)
rescaled_mat<-signedRescale(mat)
mat
rescaled_mat<-signedRescale(abs(mat))</pre>
```

```
underscored\_pos\_to\_GRanges
```

Convert coordinates in underscored format to a GRanges object.

## Description

This function creates a new GRanges object from a character vector of coordinates in the form "chr1\_0\_5000" and creates a GRanges object from them.

#### Usage

```
underscored_pos_to_GRanges(
  underscored_positions = NULL,
  extended_data = NULL,
  zeroToOneBasedStart = T,
  zeroToOneBasedEnd = F
)
```

#### Arguments

## Value

A GRanges object

#### **Examples**

```
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
underscored_pos_to_GRanges(colnames(nbl_result_matrix_sign_small))
```

writeAsymmetricMeltedChromosomalMatrixToDisk

Write a matrix, with genes, of a submatrix of a whole genome interaction matrix to disk.

#### **Description**

Writes an RData file with a ggplot2 object within.

## Usage

```
writeAsymmetricMeltedChromosomalMatrixToDisk(
  whole_genome_matrix,
  chrom1,
  chrom2,
  extra_data_matrix = NULL,
  transpose = F,
  sequential = T,
```

```
debug = T,
  desired_range_start = 50,
  desired_range_end = 300,
  saveToDisk = T,
  max_cap = NULL,
  rescale = T
)
```

#### **Arguments**

whole\_genome\_matrix

A matrix to have edges averaged with genomic coordinates in the form chr1\_50\_100

set as the column and row names.

chrom1 first chromosome of the two which will subset the matrix. (this is done in row-

column fasion).

chrom2 second chromosome of the two which will subset the matrix. (this is done in

row-column fasion).

extra\_data\_matrix

A matrix with additional variables about each point, one position per row with

as many variables as remaining columns.

transpose the matrix?

sequential disable parallelization with registerDoSEQ()?

debug extra output

desired\_range\_start

start of range for width and height of matrix for downsampling

desired\_range\_end

end of range for width and height of matrix for downsampling

saveToDisk saves the matrix to disk

max\_cap maximum saturation cap, passed to signedRescale

rescale perform signedRescale() on matrix?

#### Value

ggplotmatrix a matrix with values sufficient to create a ggplot2 heatmap with geom\_tile() or with ggiraph's geom\_tile\_interactive()

```
load(system.file("extdata","grch37.rda",package = "CNVScope"))
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
load(system.file("extdata","ensembl_gene_tx_table_prot.rda",package = "CNVScope"))
writeAsymmetricMeltedChromosomalMatrixToDisk(whole_genome_matrix = nbl_result_matrix_sign_small,
chrom1 = 1,chrom2 = 1,desired_range_start = 25, desired_range_end = 25)
file.remove("chr1_chr1_melted.RData")
```

writeMeltedChromosomalMatrixToDisk

Write a matrix, with genes, of a submatrix of a whole genome interaction matrix to disk.

## Description

Writes an RData file with a ggplot2 object within the current directory.

#### Usage

```
writeMeltedChromosomalMatrixToDisk(
  whole_genome_matrix,
  chrom1,
  chrom2,
  filename,
  extra_data_matrix = NULL,
  transpose = F,
  sequential = T,
  debug = T,
  desired_range_start = 50,
  desired_range_end = 300
)
```

## **Arguments**

whole\_genome\_matrix

A matrix to have edges averaged with genomic coordinates in the form chr1\_50\_100

set as the column and row names.

chrom1 first chromosome of the two which will subset the matrix. (this is done in row-

column fasion).

chrom2 second chromosome of the two which will subset the matrix. (this is done in

row-column fasion).

filename the filename to be written

extra\_data\_matrix

A matrix with additional variables about each point, one position per row with

as many variables as remaining columns.

transpose the matrix?

sequential Disable parallelization with doParallel? registerDoSEQ() is used for this.

debug verbose output for debugging

desired\_range\_start

the downsampled matrix must be of this size (rows & cols) at minimum

desired\_range\_end

the downsampled matrix must be of this size (rows & cols) at maximum

## Value

ggplotmatrix a matrix with values sufficient to create a ggplot2 heatmap with geom\_tile() or with ggiraph's geom\_tile\_interactive()

```
load(system.file("extdata","grch37.rda",package = "CNVScope"))
load(system.file("extdata","nbl_result_matrix_sign_small.rda",package = "CNVScope"))
load(system.file("extdata","ensembl_gene_tx_table_prot.rda",package = "CNVScope"))
writeMeltedChromosomalMatrixToDisk(whole_genome_matrix = nbl_result_matrix_sign_small,
chrom1 = 1,chrom2 = 1,desired_range_start = 25, desired_range_end = 25)
file.remove("chr1_chr1_melted.RData")
```

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 $\label{lem:constraint} write A symmetric Melted Chromosomal Matrix To Disk, \\ \frac{24}{\text{write} Melted Chromosomal Matrix To Disk}, \\ 26$