# Package 'comapr'

October 16, 2024

Title Crossover analysis and genetic map construction

Version 1.8.0

Description comapr detects crossover intervals for single gametes from their haplotype states sequences and stores the crossovers in GRanges object. The genetic distances can then be calculated via the mapping functions using estimated crossover rates for maker intervals. Visualisation functions for plotting interval-based genetic map or cumulative genetic distances are implemented, which help reveal the variation of crossovers landscapes across the genome and across individuals.

biocViews Software, SingleCell, Visualization, Genetics

**Depends** R (>= 4.1.0)

Imports methods, ggplot2, reshape2, dplyr, gridExtra, plotly, circlize, rlang, GenomicRanges, IRanges, foreach, BiocParallel, GenomeInfoDb, scales, RColorBrewer, tidyr, S4Vectors, utils, Matrix, grid, stats, SummarizedExperiment, plyr, Gviz

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# Description

change SNPs with genotype 'Fail' to NA

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### Usage

```
.change_missing(s_gt, missing = "Fail")
```

#### **Arguments**

s\_gt a column of labelled genotypes

missing the string used for encoding missing values default to Fail

#### **Details**

calculation.

#### Value

a vector of genotypes with Fail substituted by 'NA'

# Author(s)

Ruqian Lyu

.filterCOsExtra

Filter out doublet cells and uninformative SNPs

# Description

This function filter out cells that have been called too many crossovers due to diploid cell contamination or doublets. It also only keeps SNPs (rows) that ever contribute to a crossover interval. This function should be run for individual chromosomes and is called internally by 'readHapState'

```
.filterCOsExtra(
    se,
    minSNP = 30,
    minlogllRatio = 200,
    minCellSNP = 200,
    bpDist = 100,
    maxRawCO = 10,
    biasTol = 0.45,
    nmad = 1.5
)
```

4 .label\_gt

#### **Arguments**

the SummarizedExperiment object that contains the called haplotype state mase trix in the assay field and haplotype segment information in the metadata field. minSNP the crossover(s) will be filtered out if introduced by a segment that has fewer than 'minSNP' SNPs to support. minlogllRatio the crossover(s) will be filtered out if introduced by a segment that has lower than 'minlogllRatio' to its reversed state. minCellSNP the minimum number of SNPs detected for a cell to be kept, used with 'nmads' bpDist the crossover(s) will be filtered out if introduced by a segment that is shorter than 'bpDist' basepairs. if a cell has more than 'maxRawCO' number of raw crossovers called across a maxRawCO chromosome, the cell is filtered out biasTol the SNP's haplotype ratio across all cells is assumed to be 1:1. This argument can be used for removing SNPs that have a biased haplotype. i.e. almost always inferred to be haplotype state 1. It specifies a bias tolerance value, SNPs with haplotype ratios deviating from 0.5 smaller than this value are kept. Only effective when number of cells are larger than 10 how many mean absolute deviations lower than the median number of SNPs per nmad cellfor a cell to be considered as low coverage cell and filtered Only effective when number of cells are larger than 10. When effective, this or 'minCellSNP', whichever is larger, is applied

### **Details**

The 'logllRatio' value is returned by 'sgcocaller' for each haplotype segment formed by consecutive SNPs that are called to have a same state. It is calculated by taking log of ratio (likelihood of SNPs with inferred states) and (likelihood of SNPs with reversed states)

#### Value

A 'RangedSummarizedExperment' object that have different dims with input. the colnames are the cell barcodes, rowRanges specify the location of SNPs that contribute to crossovers.

#### Author(s)

Ruqian Lyu

.label_gt 'label_gt' for changing genotypes in alleles format to labels	

#### **Description**

It turns a vector of Genotypes to a vector of Labels consist of 'Homo\_ref', 'Homo\_alt', and 'Het' given the known genotypes for reference and alternative strains.

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#### Usage

```
.label_gt(s_gt, ref, alt, failed = "Fail")
```

### Arguments

s_gt	s_gt, a vector of genotypes for one sample across markers
ref	ref, a vector of genotypes for reference strain across markers
alt	alt, a vector of genotypes for alternative strain across markers
failed	what was used for encoding failed genotype calling such as "Fail" in example

#### **Details**

This function takes the a sample's genotype across each SNP marker in alleles and compare with genotypes of in-bred reference and alternative strains to. If the sample's genotype for a particular SNP marker is the same with the reference strain, it is labelled as Homo\_ref homogeneous reference for a particular SNP marker; if the sample's genotype is the same with the alternative strain it is labelled as Homo\_alt homogeneous alternative for a particular SNP marker; if the sample's genotype is heterozygous then it is labelled as Het heterozygous for this particular genotypes. If it does not fall in any of the three cases, it is labelled as the string specified by the argument 'missing'.

Note that the wrong/failed genotype is labelled as the string in 'missing' after this function. If there is a different label for failed genotype, provide the label using the 'missing' argument.

#### Value

a vector of labels Homo\_ref, Homo\_alt, Het indicating the progeny's genotypes across markers

#### Author(s)

Rugian Lyu

bootstrapDist bootstrapDist

### **Description**

Generating distribution of sample genetic distances

```
bootstrapDist(co_gr, B = 1000, mapping_fun = "k", group_by)
```

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### **Arguments**

co_gr	GRanges or RangedSummarizedExperiment object that contains the crossover counts for each marker interval across all samples. Returned by countCOs
В	integer the number of sampling times
mapping_fun	character default to "k" (kosambi mapping function). It can be one of the mapping functions: "k","h" $$
group_by	the prefix for each group that we need to generate distributions for(only when co_gr is a GRanges object). Or the column name for 'colData(co_gr)' that contains the group factor (only when co_gr is a RangedSummarizedExperiment object)

### **Details**

It takes the crossover counts for samples in multiple groups that is returned by 'countCO'. It then draws samples from a group with replacement and calculate the distribution of relevant statistics.

# Value

lists of numeric genetic distances for multiple samples

#### Author(s)

Ruqian Lyu

### **Examples**

```
data(coCount)
bootsDiff <- bootstrapDist(coCount, group_by = "sampleGroup",B=10)</pre>
```

calGeneticDist

cal Genetic Dist

# Description

Calculate genetic distances of marker intervals or binned-chromosome Given whether crossover happens in each marker interval, calculate the recombination fraction in samples and then derive the Haldane or Kosambi genetic distances via mapping functions

```
calGeneticDist(
  co_count,
  bin_size = NULL,
  mapping_fun = "k",
  ref_genome = "mm10",
  group_by = NULL,
```

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```
chrom_info = NULL
## S4 method for signature 'GRanges, missing, ANY, ANY, missing'
calGeneticDist(
  co_count,
 bin_size = NULL,
 mapping_fun = "k"
  ref_genome = "mm10",
  group_by = NULL,
  chrom_info = NULL
)
## S4 method for signature 'GRanges, numeric, ANY, ANY, missing'
calGeneticDist(
  co_count,
  bin_size = NULL,
  mapping_fun = "k",
  ref_genome = "mm10",
  group_by = NULL,
  chrom_info = NULL
)
## S4 method for signature 'GRanges, missing, ANY, ANY, character'
calGeneticDist(
  co_count,
 bin_size = NULL,
 mapping_fun = "k"
 ref_genome = "mm10",
 group_by = NULL,
  chrom_info = NULL
)
## S4 method for signature 'GRanges, numeric, ANY, ANY, character'
calGeneticDist(
  co_count,
  bin_size = NULL,
 mapping_fun = "k"
  ref_genome = "mm10",
  group_by = NULL,
  chrom_info = NULL
)
## S4 method for signature 'RangedSummarizedExperiment,missing,ANY,ANY,missing'
calGeneticDist(
  co_count,
  bin_size = NULL,
  mapping_fun = "k",
```

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```
ref_genome = "mm10",
  group_by = NULL,
  chrom_info = NULL
)
## S4 method for signature
## 'RangedSummarizedExperiment, missing, ANY, ANY, character'
calGeneticDist(
  co_count,
 bin_size = NULL,
 mapping_fun = "k",
 ref_genome = "mm10",
  group_by = NULL,
  chrom_info = NULL
)
## S4 method for signature
## 'RangedSummarizedExperiment, numeric, ANY, ANY, character'
calGeneticDist(
  co_count,
 bin_size = NULL,
 mapping_fun = "k",
 ref_genome = "mm10",
  group_by = NULL,
  chrom_info = NULL
)
## S4 method for signature 'RangedSummarizedExperiment,numeric,ANY,ANY,missing'
calGeneticDist(
  co_count,
  bin_size = NULL,
 mapping_fun = "k"
  ref_genome = "mm10",
  group_by = NULL,
  chrom_info = NULL
)
```

### **Arguments**

co_count	GRange or RangedSummarizedExperiment object, returned by countCO
bin_size	The binning size for grouping marker intervals into bins. If not supplied,the originial marker intervals are returned with converted genetic distancens based on recombination rate
mapping_fun	The mapping function to use, can be one of "k" or "h" (kosambi or haldane)
ref_genome	The reference genome name. It is used to fetch the chromosome size information from UCSC database.
group_by	character vector contains the unique prefix of sample names that are used for defining different sample groups. Or the column name in colData(co_count)

coCount 9

that specify the group factor. If missing all samples are assumed to be from one

group

chrom\_info A user supplied data.frame containing two columns with column names chrom

and size, describing the chromosome names and lengths if not using ref\_genome

from UCSC. If supplied, the 'ref\_genome' is ignored.

#### Value

GRanges object GRanges for marker intervals or binned intervals with Haldane or Kosambi centi-Morgans

### **Examples**

```
data(coCount)
dist_se <- calGeneticDist(coCount)
# dist_se <- calGeneticDist(coCount,group_by="sampleGroup")</pre>
```

coCount

RangedSummarizedExperiment object containing the crossover counts across samples for the list of SNP marker intervals

### **Description**

RangedSummarizedExperiment object containing the crossover counts across samples for the list of SNP marker intervals

### Usage

data(coCount)

#### **Format**

An object of class RangedSummarizedExperiment with 3 rows and 10 columns.

comapr	comapr package	

# Description

crossover inference package

#### **Details**

See the README on GitLab

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combineHapState

combineHapState

### Description

combine two 'RangedSummarizedExperiment' objects, each contains the haplotype state for a list of SNPs across a set of cells. The combined result will have cells from two individuals and merged list of SNPs from the two.

### Usage

```
combineHapState(rse1, rse2, groupName = c("Sample1", "Sample2"))
```

#### **Arguments**

rse1 the first 'RangedSummarizedExperiment'
rse2 the second 'RangedSummarizedExperiment'

groupName a character vector of length 2 that contains the first and the second group's names

#### Value

A 'RangedSummarizedExperiment' that contains the cells and SNPs in both 'rse'

#### Author(s)

Ruqian Lyu

correctGT 11

|--|

### **Description**

function for formatting and correction genotypes of markers

### Usage

```
correctGT(gt_matrix, ref, alt, failed = "Fail", wrong_label = "Homo_ref")
```

### **Arguments**

gt_matrix	the input genotype matrix of markers by samples with rownames as marker IDs and column names as sample IDs
ref	a vector of genotypes of the inbred reference strain
alt	a vector of genotypes of the inbred alternative strain
failed	what was used for encoding failed genotype calling such as "Fail" in example data snp_geno
wrong_label	what would be considered a wrong genotype label for example Homo_ref which should not be in the possible genotypes of BC1F1 samples

#### **Details**

This function changes genotype in alleles to genotype labels, change Homo\_ref to Hets/Fail, infer Failed genotype, and change "Failed" to NA for counting crossover later

This function changes genotype in alleles to labels by calling internal functions lable\_gt, and changes the wrong genotype Homo\_ref to Fail by calling .change\_missing.

### Value

a genotype data.frame of sample genotypes with dimension as the input 'gt\_matrix' with genotypes converted to labels and failed calls are changed to NA.

### Author(s)

Ruqian Lyu

12 countBinState

### **Description**

Bins the chromosome into supplied number of bins and find the state of the chromosome bins across all gamete cells

### Usage

```
countBinState(chr, snpAnno, viState, genomeRange, ntile = 5)
```

### **Arguments**

chr	character, the chromosome to check
snpAnno	data.frame, the SNP annotation for the supplied chromosome
viState	dgTMatrix/Matrix, the viterbi state matrix, output from 'sgcocaller'
genomeRange	GRanges object with seqlengths information for the genome
ntile	integer, how many tiles the chromosome is binned into

#### **Details**

This function is used for checking whether chromosome segregation pattern obeys the expected ratio.

#### Value

a data.frame that contains chromosome bin segregation ratio

### Author(s)

Ruqian Lyu

```
library(IRanges)
library(S4Vectors)

chrom_info <- GenomeInfoDb::getChromInfoFromUCSC("mm10")
seq_length <- chrom_info$size
names(seq_length) <- chrom_info$chrom

dna_mm10_gr <- GenomicRanges::GRanges(
    seqnames = Rle(names(seq_length)),
    ranges = IRanges(1, end = seq_length, names = names(seq_length)),
    seqlengths = seq_length)

GenomeInfoDb::genome(dna_mm10_gr) <- "mm10"</pre>
```

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countC0s

countCOs

### **Description**

Count number of COs within each marker interval COs identified in the interval overlapping missing markers are distributed according to marker interval base-pair sizes. Genotypes encoded with "0" are treated as missing value.

### Usage

```
countCOs(geno)
## S4 method for signature 'GRanges'
countCOs(geno)
## S4 method for signature 'RangedSummarizedExperiment'
countCOs(geno)
```

# **Arguments**

geno

GRanges object or RangedSummarizedExperiment object with genotype matrix that has SNP positions in the rows and cells/samples in the columns

#### Value

GRanges object or RangedSummarizedExperiment with markers-intervals as rows and samples in columns, values as the number of COs estimated for each marker interval

### Author(s)

Ruqian Lyu

14 countGT

### **Examples**

```
data(twoSamples)
cocount <- countCOs(twoSamples)</pre>
```

countGT

countGT

# Description

count how many samples have genotypes calls across markers and count how many markers that each individual has called genotypes for. This function helps identify poor samples or poor markers for filtering. It can also generate plots that help identify outlier samples/markers

### Usage

```
countGT(geno, plot = TRUE, interactive = FALSE)
```

# Arguments

geno the genotype data.frame of markers by samples from output of function correctGT

plot it determines whether a plot will be generated, defaults to TRUE

interactive it determines whether an interactive plot will be generated

### Value

A list of two elements including n\_markers and n\_samples

# Author(s)

Ruqian Lyu

```
data(snp_geno_gr)
genotype_counts <- countGT(GenomicRanges::mcols(snp_geno_gr))</pre>
```

fill\_fail

C:11 C :1	
fill_fail	Infer the genotype of failed SNPs If we have a Fail in the genotype
	data and the Fail in a block of either Home_alt, or Het, we fill in the
	Fails using values of the ones adjacent to it, otherwise they remain as
	"Fail" to indicate missing values.

### **Description**

Infer the genotype of failed SNPs If we have a Fail in the genotype data and the Fail in a block of either Home\_alt, or Het, we fill in the Fails using values of the ones adjacent to it, otherwise they remain as "Fail" to indicate missing values.

#### Usage

```
fill_fail(s_gt, fail = "Fail", chr = NULL)
```

### **Arguments**

s_gt	a column of labelled genotypes
fail	the string that is used for encoding failed genotype results, default to Fail
chr	the factor vector indicating which chromosomes the markers are on, default to
	NULL which means the input markers are all on the same chromosome.

### Value

a vector of genotypes with Failed genotype imputed or changed to 'NA' if not imputable

### Author(s)

Ruqian Lyu

filter GT
-----------

#### **Description**

Filter markers or samples that have too many missing values

```
filterGT(geno, min_markers = 5, min_samples = 3)
## S4 method for signature 'matrix,numeric,numeric'
filterGT(geno, min_markers = 5, min_samples = 3)
## S4 method for signature 'GRanges,numeric,numeric'
filterGT(geno, min_markers = 5, min_samples = 3)
```

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### **Arguments**

geno the genotype data.frame of markers by samples from output of function correctGT

min\_markers the minimum number of markers for a sample to be kept min\_samples the minimum number of samples for a marker to be kept

#### **Details**

This function takes the geno data.frame and filter the data.frame by the provided cut-offs.

#### Value

The filtered genotype matrix

### Author(s)

Ruqian Lyu

### **Examples**

```
data(snp_geno_gr)
corrected_geno <- filterGT(snp_geno_gr, min_markers = 30,min_samples = 2)</pre>
```

findDupSamples

findDupSamples

#### **Description**

Find the duplicated samples by look at the number of matching genotypes in all pair-wise samples

#### Usage

```
findDupSamples(geno, threshold = 0.99, in_text = FALSE)
```

### **Arguments**

the genotype data.frame of markers by samples from output of function correctGT geno

threshold the frequency cut-off of number of matching genotypes out of all geneotypes

for determining whether the pair of samples are duplicated, defaults to 0.99. NAs are regarded as same genotypes for two samples if they both have NA for

a marker.

in\_text whether text of frequencies should be displayed in the heatmap cells

### Value

The paris of duplicated samples.

getAFTracks 17

#### Author(s)

Ruqian Lyu

#### **Examples**

getAFTracks

getAFTracks

### **Description**

Generate the raw alternative allele frequencies tracks for all cells in the columns of provided 'co\_count'

### Usage

```
getAFTracks(
  chrom = "chr1",
  path_loc = "./output/firstBatch/WC_522/",
  sampleName = "WC_522",
  nwindow = 80,
  barcodeFile,
  co_count,
  snp_track = NULL
)
```

### **Arguments**

chrom the chromosome path\_loc the path prefix to the output files from sscocaller including "\*\_totalCount.mtx" and "\_altCount.mtx" the sample name, which is the prefix of sscocaller's output files sampleName nwindow the number of windows for binning the chromosome barcodeFile the barcode file containing the list of cell barcodes used as the input file for sscocaller 'GRange' or 'RangedSummarizedExperiment' object, returned by countCO that co\_count contains the crossover intervals and the number of crossovers in each cell. snp\_track the SNP position track which is used for obtaining the SNP chromosome locations. It could be omitted and the SNP positions will be acquired from the "\*\_snpAnnot.txt" file.

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#### Value

a list object, in which each element is a list of two items with the cell's alternative allele frequency DataTrack and the called crossover ranges.

#### Author(s)

Ruqian Lyu

### **Examples**

getCellAFTrack

getCellAFTrack Generates the DataTracks for plotting AF and crossover regions

### Description

It plots the raw alternative allele frequencies and highlight the crossover regions for the selected cell.

It plots the raw alternative allele frequencies and highlight the crossover regions for the selected cell.

```
getCellAFTrack(
  chrom = "chr1",
  path_loc = "./output/firstBatch/WC_522/",
  sampleName = "WC_522",
  nwindow = 80,
  barcodeFile,
  cellBarcode,
  co_count,
  snp_track = NULL,
```

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```
chunk = 1000L
)

getCellAFTrack(
  chrom = "chr1",
  path_loc = "./output/firstBatch/WC_522/",
  sampleName = "WC_522",
  nwindow = 80,
  barcodeFile,
  cellBarcode,
  co_count,
  snp_track = NULL,
  chunk = 1000L
)
```

### **Arguments**

chrom the chromosome

path\_loc the path prefix to the output files from sscocaller including "\*\_totalCount.mtx"

and "\_altCount.mtx"

sampleName the sample name, which is the prefix of sscocaller's output files

nwindow the number of windows for binning the chromosome

barcodeFile the barcode file containing the list of cell barcodes used as the input file for

sscocaller

cellBarcode the selected cell barcode

co\_count 'GRange' or 'RangedSummarizedExperiment' object, returned by countC0 that

contains the crossover intervals and the number of crossovers in each cell.

snp\_track the SNP position track which is used for obtaining the SNP chromosome lo-

cations. It could be omitted and the SNP positions will be acquired from the

"\*\_snpAnnot.txt" file.

chunk An integer scalar indicating the chunk size to use, i.e., number of rows to read

at any one time.

#### Value

The DataTrack object defined in DataTrack
The DataTrack object defined in DataTrack

### Author(s)

Ruqian Lyu

20 getCellCORange

```
minlogllRatio = 50,
                              bpDist = 100,maxRawCO=10,
                              minCellSNP = 0)
s1_counts <- countCOs(s1_rse_state)</pre>
af_co_tracks <- getCellAFTrack(chrom ="chr1",</pre>
                                 path_loc = demo_path,
                                 sampleName = "s1",
                                 barcodeFile = paste0(demo_path,
                                                       "s1_barcodes.txt"),
                                 cellBarcode = "BC1",
                                 co_count = s1_counts)
demo_path <-paste0(system.file("extdata",package = "comapr"),"/")</pre>
s1_rse_state <- readHapState("s1",chroms=c("chr1"),</pre>
                              path=demo_path,barcodeFile=NULL,minSNP = 0,
                              minlogllRatio = 50,
                              bpDist = 100,maxRawCO=10,
                              minCellSNP = 0)
s1_counts <- countCOs(s1_rse_state)</pre>
af_co_tracks <- getCellAFTrack(chrom ="chr1",</pre>
                                 path_loc = demo_path,
                                 sampleName = "s1",
                                 barcodeFile = paste0(demo_path,
                                                       "s1_barcodes.txt"),
                                 cellBarcode = "BC1",
                                 co_count = s1_counts)
```

getCellCORange

getCellCORange

### **Description**

It finds the crossover intervals for a selected cell

#### **Usage**

```
getCellCORange(co_count, cellBarcode)
```

### **Arguments**

co\_count 'GRanges' or 'RangedSummarizedExperiment' object,
cellBarcode the selected cell's barcode

#### Value

GRange object containing the crossover intervals for the selected cell

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#### Author(s)

Ruqian Lyu

### **Examples**

getCellDPTrack

getCellDPTrack Generates the DataTrack for plotting DP of a selected cell

# Description

It plots the total allele counts for the selected cell.

### Usage

```
getCellDPTrack(
  chrom = "chr1",
  path_loc = "./output/firstBatch/WC_522/",
  sampleName = "WC_522",
  nwindow = 80,
  barcodeFile,
  cellBarcode,
  snp_track = NULL,
  chunk = 1000L,
  log = TRUE,
  plot_type = "hist"
)
```

### **Arguments**

chrom the chromosome

path\_loc the path prefix to the output files from sscocaller including "\*\_totalCount.mtx"

sampleName the sample name, which is the prefix of sscocaller's output files

nwindow the number of windows for binning the chromosome

barcodeFile the barcode file containing the list of cell barcodes used as the input file for

sscocaller

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the selected cell barcode

snp\_track the SNP position track which is used for obtaining the SNP chromosome locations. It could be omitted and the SNP positions will be acquired from the "\*\_snpAnnot.txt" file.

chunk A integer scalar indicating the chunk size to use, i.e., number of rows to read at any one time.

whether the histogram of SNP density should be plotted on log scale (log10)

plot\_type the DataTrack plot type, default to be 'hist'

#### Value

log

The DataTrack object defined in DataTrack

### Author(s)

Ruqian Lyu

# **Examples**

getDistortedMarkers getDistortedMarkers

#### **Description**

Marker segregation distortion detection using chisq-test

```
getDistortedMarkers(geno, p = c(0.5, 0.5), adj.method = "BH")
```

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### **Arguments**

geno	the genotype data.frame of markers by samples from output of function correctGT
р	the expected geneotype ratio in a numeric vector, defaults to $c(0.5,0.5)$
adj.method	Methods to adjust for multiple comparisons, defaults to "BH"

#### **Details**

We expect the genotypes to appear with the frequencies of 1:1 homo\_alt:hets. We usechisq.test for finding markers that have genotypes among samples that are significantly different from the 1:1 ratio and report them

#### Value

data.frame with each row representing one SNP marker and columns containing the chisq.test results

#### Author(s)

Ruqian Lyu

#### **Examples**

```
data(parents_geno)
data(snp_geno_gr)
corrected_geno <- correctGT(gt_matrix = GenomicRanges::mcols(snp_geno_gr),
ref = parents_geno$ref,alt = parents_geno$alt,fail = "Fail",
wrong_label = "Homo_ref")
GenomicRanges::mcols(snp_geno_gr) <- corrected_geno
corrected_geno <- filterGT(snp_geno_gr, min_markers = 30,min_samples = 2)
pvalues <- getDistortedMarkers(GenomicRanges::mcols(corrected_geno))</pre>
```

getMeanDPTrack

getMeanDPTrack

### **Description**

Generate the mean DP (Depth) DataTrack (from Gviz) for cells

```
getMeanDPTrack(
  chrom = "chr1",
  path_loc,
  nwindow = 80,
  sampleName,
  barcodeFile,
  plot_type = "hist",
```

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```
selectedBarcodes = NULL,
snp_track = NULL,
log = TRUE
)
```

### Arguments

chrom the chromosome

path\_loc the path prefix to the output files from sscocaller including "\*\_totalCount.mtx"

and "\_altCount.mtx"

nwindow the number of windows for binning the chromosome

sampleName the sample name, which is the prefix of sscocaller's output files

barcodeFile the barcode file containing the list of cell barcodes used as the input file for

sscocaller

plot\_type the DataTrack plot type, default to be 'hist'

selectedBarcodes

the selected cell barcodes which should be the barcodes that have been called

crossovers for. If not supplied then all cells are counted.

snp\_track the SNP position track which is used for obtaining the SNP chromosome lo-

cations. It could be omitted and the SNP positions will be acquired from the

"\*\_snpAnnot.txt" file.

log whether the histogram of SNP density should be plotted on log scale (log10)

#### Value

DataTrack object plotting the mean DP histogram for windowed chromosomes

### Author(s)

Ruqian Lyu

getSNPDensityTrack 25

getSNPDensityTrack	getSNPDensityTrack

# Description

Generate the SNP density DataTrack (from 'Gviz') for selected chromosome

# Usage

```
getSNPDensityTrack(
  chrom = "chr1",
  sampleName = "s1",
  path_loc = ".",
  nwindow = 80,
  plot_type = "hist",
  log = TRUE
)
```

# Arguments

chrom	the chromosome
sampleName	the sample name, which is the prefix of sscocaller's output files
path_loc	the path prefix to the output files from sscocaller including "*_totalCount.mtx" and "_altCount.mtx"
nwindow	the number of windows for binning the chromosome
plot_type	the DataTrack plot type, default to be 'hist'
log	whether the histogram of SNP density should be plotted on log scale (log10)

### Value

DataTrack object plotting the SNP density histogram

### Author(s)

Ruqian Lyu

26 perCellChrQC

parents\_geno

Parents' genotype for F1 samples in 'snp\_geno'

# Description

Parents' genotype for F1 samples in 'snp\_geno'

### Usage

```
data(parents_geno)
```

#### **Format**

A data.frame:

**C57BL.6J** genotype of reference mouse train across markers **FVB.NJ..i.** genotype of alternative mouse train across markers

perCellChrQC

perCellChrQC

### **Description**

A function that parses output ('\_viSegInfo.txt') from 'sgcocaller' https://gitlab.svi.edu.au/biocellgen-public/sgcocaller and generate cell (per chr) summary statistics

#### Usage

```
perCellChrQC(
  sampleName,
  chroms = c("chr1", "chr7", "chr15"),
  path,
  barcodeFile = NULL,
  doPlot = TRUE
)
```

# Arguments

sampleName	the name of the sam	nle to parse	which is used as	prefix for finding relevant files

for the underlying sample

chroms the character vectors of chromosomes to parse. Multiple chromosomes' results

will be concated together.

path the path to the files, with name patterns \*chrom\_vi.mtx, \*chrom\_viSegInfo.txt,

end with slash

barcodeFile defaults to NULL, it is assumed to be in the same directory as the other files

and with name sampleName\_barcodes.txt

doPlot whether a plot should returned, default to TRUE

permuteDist 27

### Value

a list object that contains the data.frame with summarised statistics per chr per cell and a plot (if doPlot)

### Author(s)

Ruqian Lyu

### **Examples**

```
demo_path <-system.file("extdata",package = "comapr")
pcQC <- perCellChrQC(sampleName="s1",chroms=c("chr1"),
path=demo_path,
barcodeFile=NULL)</pre>
```

permuteDist

permuteDist

### **Description**

Permutation test of two sample groups

### Usage

```
permuteDist(co_gr, B = 100, mapping_fun = "k", group_by)
```

### **Arguments**

co_gr	GRanges or RangedSummarizedExperiment object that contains the crossover counts for each marker interval across all samples. Returned by countCOs
В	integer the number of sampling times
mapping_fun	character default to "k" (kosambi mapping function). It can be one of the mapping functions: "k","h" $$
group_by	the prefix for each group that we need to generate distributions for(only when co_gr is a GRanges object). Or the column name for 'colData(co_gr)' that contains the group factor (only when co_gr is a RangedSummarizedExperiment object)

# **Details**

It shuffles the group labels for the samples and calculate a difference between two groups after shuffling.

### Value

A list of three elements. 'permutes' of length B with numeric differences of permuted group differences, 'observed\_diff' the observed genetic distances of two groups, 'nSample', the number of samples in the first and second group.

28 perSegChrQC

#### Author(s)

Ruqian Lyu

#### **Examples**

```
data(coCount)
perms <- permuteDist(coCount, group_by = "sampleGroup",B=10)</pre>
```

perSegChrQC

perSegChrQC

### **Description**

Plots the summary statistics of segments that are generated by 'sgcocaller' https://gitlab.svi.edu.au/biocellgen-public/sgcocaller which have been detected by finding consequtive viter states along the list of SNP markers.

### Usage

```
perSegChrQC(
  sampleName,
  chroms = c("chr1", "chr7", "chr15"),
  path,
  barcodeFile = NULL,
  maxRawCO = 10
)
```

### **Arguments**

sampleName the name of the sample to parse which is used as prefix for finding relevant files

for the underlying sample

chroms the vector of chromosomes

path the path to the files, with name patterns \*chrom\_vi.mtx, \*chrom\_viSegInfo.txt,

end with slash

barcodeFile defaults to NULL, it is assumed to be in the same directory as the other files and

with name sampleName\_barcodes.txt

maxRawCO if a cell has more than 'maxRawCO' number of raw crossovers called across a

chromosome, the cell is filtered out#'

### **Details**

It provides guidance in filtering out close double crossovers that are not likely biological but due to technical reasons as well as crossovers that are supported by fewer number of SNPs at the ends of the chromosomes.

plotCount 29

### Value

Histogram plots for statistics summarized across all Viterbi state segments

#### Author(s)

Ruqian Lyu

### **Examples**

plotCount

plotCount

### **Description**

Plot the number of COs per sample group or per chromosome

```
plotCount(
  co_count,
  by_chr = FALSE,
 group_by = "sampleGroup",
 plot_type = "error_bar"
)
## S4 method for signature 'RangedSummarizedExperiment, missing, missing'
plotCount(
  co_count,
 by_chr = FALSE,
 group_by = "sampleGroup",
 plot_type = "error_bar"
)
## S4 method for signature 'RangedSummarizedExperiment,missing,character'
plotCount(
  co_count,
 by_chr = FALSE,
 group_by = "sampleGroup",
 plot_type = "error_bar"
```

30 plotCount

```
## S4 method for signature 'RangedSummarizedExperiment,logical,character'
plotCount(
  co_count,
 by_chr = FALSE,
 group_by = "sampleGroup",
 plot_type = "error_bar"
)
## S4 method for signature 'RangedSummarizedExperiment,logical,missing'
plotCount(
  co_count,
 by_chr = FALSE,
 group_by = "sampleGroup",
 plot_type = "error_bar"
)
## S4 method for signature 'GRanges,logical,missing'
plotCount(
 co_count,
 by_chr = FALSE,
 group_by = "sampleGroup",
 plot_type = "error_bar"
)
## S4 method for signature 'GRanges, missing, missing'
plotCount(
  co_count,
 by_chr = FALSE,
 group_by = "sampleGroup",
 plot_type = "error_bar"
)
## S4 method for signature 'GRanges, missing, character'
plotCount(
 co_count,
 by_chr = FALSE,
 group_by = "sampleGroup",
 plot_type = "error_bar"
## S4 method for signature 'GRanges, logical, character'
plotCount(
 co_count,
 by_chr = FALSE,
 group_by = "sampleGroup",
 plot_type = "error_bar"
)
```

plotGeneticDist 31

### **Arguments**

co_count	GRange or RangedSummarizedExperiment object, returned by countCO
by_chr	whether it should plot each chromosome separately
group_by	the column name in 'colData(co_count)' that specify the grouping factor. Or the character vector contains the unique prefix of sample names that are used for defining different sample groups. If missing all samples are assumed to be from one group
plot_type	determins what type the plot will be, choose from "error_bar" or "hist". Only

### Value

ggplot object

# **Examples**

plotGeneticDist

plotGeneticDist

relevant when by\_chr=TRUE

# Description

Plotting the calculated genetic distanced for each bin or marker interval supplied by the GRanges object

# Usage

```
plotGeneticDist(gr, bin = TRUE, chr = NULL, cumulative = FALSE)
```

the cumulative distances

### **Arguments**

gr	GRanges object with genetic distances calculated for marker intervals
bin	TRUE or FALSE, indicating whether the supplied GRange objecct is for binned interval
chr	the specific chrs selected to plot
cumulative	TRUE or FALSE, indicating whether it plots the bin-wise genetic distances or

32 plotGTFreq

### Value

```
ggplot2 plot
```

#### Author(s)

Ruqian Lyu

# **Examples**

```
data(coCount)
dist_se <- calGeneticDist(coCount)
plotGeneticDist(SummarizedExperiment::rowRanges(dist_se))</pre>
```

plotGTFreq

plotGTFreq

### **Description**

Function to plot the genotypes for all samples faceted by genotype

#### Usage

```
plotGTFreq(geno)
```

#### **Arguments**

geno

the genotype data.frame of markers by samples from output of function correctGT

### Value

A ggplot object

#### Author(s)

Ruqian Lyu

plotWholeGenome 33

plotWholeGenome	Plot cumulative genetic distances across the genome

### **Description**

This function takes the calculated genetic distances for all marker intervals across all chromosomes provided and plot the cumulative genetic distances

# Usage

```
plotWholeGenome(gr)
```

# **Arguments**

gr

GRanges object with genetic distances calculated for marker intervals

#### Value

A ggplot object

# Examples

```
data(coCount)
dist_se <- calGeneticDist(coCount)
plotWholeGenome(SummarizedExperiment::rowRanges(dist_se))</pre>
```

readColMM

readColMM

### **Description**

Modified the 'Matrix::readMM' function for reading matrices stored in the Harwell-Boeing or MatrixMarket formats but only reads selected column.

# Usage

```
readColMM(file, which.col, chunk = 1000L)
```

### **Arguments**

fi]	Le t	he name of	the file to	be read	l from as a c	haracter scalar	. Those storing matrices
-----	------	------------	-------------	---------	---------------	-----------------	--------------------------

in the MatrixMarket format usually end in ".mtx".

which.col An integer scalar, the column index

chunk An integer scalar indicating the chunk size to use, i.e., number of rows to read

at any one time.

readHapState

### **Details**

See readMM

### Value

A sparse matrix object that inherits from the "Matrix" class which the original dimensions. To get the vector of the specified column, one need to subset the matrix to select the column with the same index.

### Author(s)

Ruqian Lyu

# **Examples**

```
demo_path <-paste0(system.file("extdata",package = "comapr"),"/")
readColMM(file = paste0(demo_path,"s1_chr1_vi.mtx"), which.col=2,chunk=2)</pre>
```

readHapState

readHapState

### **Description**

A function that parses the viterbi state matrix (in .mtx format), barcode.txt and snpAnno.txt files for each individual.

```
readHapState(
  sampleName,
  chroms = c("chr1"),
  path,
  barcodeFile = NULL,
  minSNP = 30,
  minlogllRatio = 200,
  bpDist = 100,
  maxRawCO = 10,
  nmad = 1.5,
  minCellSNP = 200,
  biasTol = 0.45
)
```

readHapState 35

### **Arguments**

sampleName	the name of the sample to parse which is used as prefix for finding relevant files for the underlying sample
chroms	the character vectors of chromosomes to parse. Multiple chromosomes' results will be concated together.
path	the path to the files, with name patterns *chrom_vi.mtx, *chrom_viSegInfo.txt, end with slash
barcodeFile	if NULL, it is assumed to be in the same directory as the other files and with name sampleName_barcodes.txt
minSNP	the crossover(s) will be filtered out if introduced by a segment that has fewer than 'minSNP' SNPs to support.
minlogllRatio	the crossover(s) will be filtered out if introduced by a segment that has lower than 'minlogllRatio' to its reversed state.
bpDist	the crossover(s) will be filtered out if introduced by a segment that is shorter than 'bpDist' basepairs. It can be a single value or a vector that is the same length and order with 'chroms'.
maxRawCO	if a cell has more than 'maxRawCO' number of raw crossovers called across a chromosome, the cell is filtered out
nmad	how many mean absolute deviations lower than the median number of SNPs per cellfor a cell to be considered as low coverage cell and filtered Only effective when number of cells are larger than 10. When effective, this or 'minCellSNP', whichever is larger, is applied
minCellSNP	the minimum number of SNPs detected for a cell to be kept, used with 'nmads'
biasTol	the SNP's haplotype ratio across all cells is assumed to be 1:1. This argument can be used for removing SNPs that have a biased haplotype. i.e. almost always inferred to be haplotype state 1. It specifies a bias tolerance value, SNPs with haplotype ratios deviating from 0.5 smaller than this value are kept. Only

### Value

a RangedSummarizedExperiment with rowRanges as SNP positions that contribute to crossovers in any cells. colData contains cells annotation including barcodes and sampleName.

effective when number of cells are larger than 10

### Author(s)

Ruqian Lyu

```
demo_path <- system.file("extdata",package = "comapr")
s1_rse_state <- readHapState(sampleName="s1",chroms=c("chr1"),
path=paste0(demo_path,"/"),
barcodeFile=NULL,minSNP = 0, minlogllRatio = 50,
bpDist = 100,maxRawCO=10,minCellSNP=3)
s1_rse_state</pre>
```

36 snp\_geno

snp\_geno

Markers by genotype results for a group of samples

### Description

Markers by genotype results for a group of samples

### Usage

```
data(snp_geno)
```

#### **Format**

A data frame with columns:

C57BL.6J genotype of reference mouse train across markers

FVB.NJ..i. genotype of alternative mouse train across markers

**POS** SNP marker base-pair location

**CHR** SNP marker chromosome location

X100 a mouse sample

X101 a mouse sample

X102 a mouse sample

X103 a mouse sample

X104 a mouse sample

X105 a mouse sample

X106 a mouse sample

X107 a mouse sample

X108 a mouse sample

X109 a mouse sample

X110 a mouse sample

X111 a mouse sample

X112 a mouse sample

X113 a mouse sample

X92 a mouse sample

X93 a mouse sample

X94 a mouse sample

X95 a mouse sample

X96 a mouse sample

X97 a mouse sample

X98 a mouse sample

**X99** a mouse sample

A) a mouse samp

rsID the SNP ID

snp\_geno\_gr 37

#### **Source**

Statistics Canada. Table 001-0008 - Production and farm value of maple products, annual. http://www5.statcan.gc.ca/cansim/

snp\_geno\_gr

Markers by genotype results for a group of samples

### **Description**

Markers by genotype results for a group of samples

### Usage

```
data(snp_geno_gr)
```

#### **Format**

A GRanges object:

X100 a mouse sample

X101 a mouse sample

X102 a mouse sample

X103 a mouse sample

X104 a mouse sample

X105 a mouse sample

X106 a mouse sample

X107 a mouse sample

X108 a mouse sample

X109 a mouse sample

X110 a mouse sample

X111 a mouse sample

X112 a mouse sample

X113 a mouse sample

X92 a mouse sample

X93 a mouse sample

X94 a mouse sample

X95 a mouse sample

X96 a mouse sample

X97 a mouse sample

X98 a mouse sample

**X99** a mouse sample

rsID the SNP ID

38 twoSamples

### Source

TBD

twoSamples	RangedSummarizedExperiment object containing the Viterbi states SNP markers for samples from two groups. 'colData(twoSamples)' contains the sample group factor.

# Description

RangedSummarizedExperiment object containing the Viterbi states SNP markers for samples from two groups. 'colData(twoSamples)' contains the sample group factor.

# Usage

data(twoSamples)

### **Format**

An object of class RangedSummarizedExperiment with 6 rows and 10 columns.

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