# Package 'geneHapR'

July 22, 2025

| Type Package   |
|--|
| <b>Title</b> Gene Haplotype Statistics, Phenotype Association and Visualization  |
| <b>Description</b> Import genome variants data and perform gene haplotype Statistics, visualization and phenotype association with 'R'.  |
| biocViews NucleosomePositioning, DataImport  |
| Encoding UTF-8   |
| Maintainer Zhang Renliang <zhang_renliang@163.com></zhang_renliang@163.com>  |
| Version 1.2.4  |
| RoxygenNote 7.2.3  |
| LazyData True  |
| Imports ape, Biostrings, ggpubr, genetics, GenomicRanges, lolliplot, maps, methods, IRanges, pegas, reshape2, rlang, rtracklayer, shiny, shinyjs, stats, stringdist, stringr, tibble, sf, tidyr, utils, vcfR |
| <b>Depends</b> ggplot2, R (>= 4.0.0)   |
| Suggests mapdata, knitr, rmarkdown, testthat (>= 3.0.0)  |
| License GPL-3  |
| VignetteBuilder knitr  |
| Config/testthat/edition 3  |
| NeedsCompilation no  |
| Author Zhang Renliang [aut, cre], Jia Guanqing [aut]   |
| Repository CRAN  |
| <b>Date/Publication</b> 2024-03-01 14:32:40 UTC  |
| Contents   |
| addINFO  |

2 Contents

| plink.pedmap2hap         |    |
|--------------------------|----|
| network                  |    |
| LDheatmap                | 34 |
| import_vcf               | 33 |
| import_seqs              | 32 |
| import_plink.pedmap      | 31 |
| import_MultipleAlignment |    |
| import_hap               |    |
| import_gff               |    |
| • -                      |    |
| import_bed               |    |
| import_AccINFO           |    |
| hap_summary              | 26 |
| hapVsPhenos              | 24 |
| hapVsPhenoPerSite        | 23 |
| hapVsPheno               | 19 |
| hapDistribution          | 17 |
| hap2hmp                  | 16 |
| getGeneRanges            | 15 |
| getGenePOS               | 14 |
| filter_vcf               | 13 |
| filter_table             | 12 |
| filter_plink.pedmap      | 11 |
| filter_hmp               | 10 |
| filter_hap               | 9  |
| filterLargeVCF           | 8  |
| filterLargep.link        | 6  |
| displayVarOnGeneModel    | 5  |
| DataSet                  | 5  |
| ashaplotype              | 4  |
| l l - 4                  | 4  |

Index

addINFO 3

addINF0

Add Infomation to Haplotype Results

## **Description**

add annotations to INFO fields used for plotHapTable()

# Usage

```
addINFO(hap,
          tag = "", values = values,
          replace = FALSE, sep = ";")
sites(hap)
```

## **Arguments**

hap object of hapResult or hapSummary class
tag tag names, usually is a single word used before "="
values annotation for each site. Length of values must be equal with sites in hapResult
replace whether replace origin INFOs in hapResult or not. Default as FALSE
sep a character string to separate the terms. Not NA\_character\_.

## Value

object of hapSummary or hapResult class with added/replaced INFOs

#### See Also

```
plotHapTable()
plotHapTable()
```

```
data("geneHapR_test")

# length of values must be equal with number of sites in hap result
values <- paste0("newInfo",c(1:9))
hapResult <- addINFO(hapResult, tag = "new", values = values, replace = TRUE)

data("geneHapR_test")

# check how many sites were concluded in hapResult/hapSummary
sites(hapResult)</pre>
```

4 ashaplotype

addPromoter

add promoter to annotation

# Description

add promoter to annotation

# Usage

```
addPromoter(anno, PromoterLength = 1500, bedFile = NULL)
```

# Arguments

anno anotation, imported gff/bed

PromoterLength the length of promoter region, default as 1500

bedFile the output bed file name

# **Examples**

```
data("geneHapR_test")
bed <- addPromoter(gff)</pre>
```

ashaplotype

as.haplotype

# Description

convert hapSummary or hapResult class into haplotype class (pegas)

## Usage

```
as.haplotype(hap)
```

# Arguments

hap

object of hapSummary or hapResult class

## Value

haplotype class

## Note

It's not advised for hapSummary or hapResult with indels, due to indels will convert to SNPs with equal length of each indel.

DataSet 5

## **Examples**

```
data("geneHapR_test")
hap <- as.haplotype(hapResult)
hapSummary <- hap_summary(hapResult)
hap <- as.haplotype(hapSummary)</pre>
```

DataSet

Datasets gff contains a example of gff file used for test of visualization mutations on gene model.

## **Description**

pheno contains a simulated test pheno data used for test of comparison between different haps vcf, a vcfR object provide a data set for test of seq2hap(). vcf contains indels, snps, biallelic sites and multiallelic sites.

AccINFO a data.frame provide additional information of accessions, including accession type, source and location.

displayVarOnGeneModel Display Variants on Gene Model

## **Description**

show variants on gene model using hapSummary and gene annotations

## Usage

```
displayVarOnGeneModel(
  hapSummary,
  gff,
  Chr,
  startPOS,
  endPOS,
  type = "pin",
  cex = 0.7,
  CDS_h = 0.05,
  fiveUTR_h = 0.02,
  threeUTR_h = 0.01,
  geneElement = geneElement,
  hap
)
```

6 filterLargep.link

## Arguments

```
hapSummary, hap haplotype result

gff gff

Chr the chromosome name. If missing, the first element in the hapSummary will be used

startPOS If missing, will use the min position in hapSummary

endPOS If missing, will use the max position in hapSummary

type character. Could be "circle", "pie", "pin", "pie.stack" or "flag"

cex a numeric control the size of circle

CDS_h, fiveUTR_h, threeUTR_h

The height of CDS 5'UTR and 3'UTR in gene model
```

ploted elements, eg.: c("CDS", "five\_prime\_UTR")

#### Value

No return value

geneElement

## **Examples**

filterLargep.link

Pre-process of Large VCF File(s)

# Description

Filter/extract one or multiple gene(s)/range(s) from a large p.link file.

## Usage

```
filterLargeP.link(
  root,
  rootOut = rootOut,
  Chr = Chr,
  POS = NULL,
  start = start,
  end = end,
  override = TRUE,
  sep = "\t"
)
```

filterLargep.link 7

## Arguments

root The file name without suffix. This function only support p.link file format stored

in "map" and "ped" format, the file names after removed suffix should be same

with each other.

rootOut Path(s) of output p.link file stored in "ped&map" format.

Chr a single CHROM name or CHROM names vector.

POS, start, end provide the chromosome name should be extract from orignal p.link dataset.

POS: a vector consist with start and end position, eg.: c(1,200) indicates 3 ranges (1~200, 300~500 and 300~400). if POS is NULL, start and end are

needed.

override whether override existed file or not, default as TRUE.

sep a character indicate the separation of map and ped file, default is \t.

#### **Details**

This package import P.link files. However, import a large P.link file is time and memory consuming. It's suggested that extract variants in target range with filterLargeP.link() before identification of haplotype.

When filter/extract multi genes/ranges, the parameter of Chr and POS must have equal length. Results will save to a single file if the user provide a single file path or save to multiple P.link file(s) when a equal length vector consist with file paths is provided.

#### Value

No return value

```
# The filteration of P.link of regular size should be done with `filter_plink.pedmap()`.
# however, here, we use a mini vcf instead just for example and test
pedfile <- system.file("extdata",</pre>
                        "snp3kvars-CHR8-25947258-25951166-plink.ped",
                        package = "geneHapR")
mapfile <- system.file("extdata",</pre>
                        "snp3kvars-CHR8-25947258-25951166-plink.map",
                        package = "geneHapR")
oldDir <- getwd()
temp_dir <- tempdir()</pre>
if(! dir.exists(temp_dir))
 dir.create(temp_dir)
setwd(temp_dir)
file.copy(pedfile, "test.ped")
file.copy(mapfile, "test.map")
# extract a single gene/range from large vcf
filterLargeP.link(root = "test",
                  rootOut = "filtered_test",
```

8 filterLargeVCF

```
Chr = "scaffold_1", POS = c(4300,5000), override = TRUE)
setwd(oldDir)
# delete temp_dir
unlink(temp_dir, recursive = TRUE)
```

filterLargeVCF

Pre-process of Large VCF File(s)

## **Description**

Filter/extract one or multiple gene(s)/range(s) from a large \*.vcf/\*.vcf.gz file.

# Usage

## Arguments

VCFin Path of input \*.vcf/\*.vcf.gz file.

VCFout Path(s) of output \*.vcf/\*.vcf.gz file.

Chr a single CHROM name or CHROM names vector.

POS, start, end provide the range should be extract from orignal vcf. POS: a vector consist with

start and end position or a list with length equal to Chr, eg.: list(c(1,200), c(300,500), c(300,400)) indicates 3 ranges (1~200, 300~500 and 300~400). if POS is NULL, start and end are needed, eg.: start = c(1, 30) and end =  $\frac{1}{2}$ 

c(200, 150) indicates 2 ranges (1~200 and 30~150)

override whether override existed file or not, default as TRUE.

## **Details**

This package import VCF files with 'vcfR' which is more efficient to import/manipulate VCF files in 'R'. However, import a large VCF file is time and memory consuming. It's suggested that filter/extract variants in target range with filterLargeVCF().

When filter/extract multi genes/ranges, the parameter of Chr and POS must have equal length. Results will save to a single file if the user provide a single file path or save to multiple VCF file(s) when a equal length vector consist with file paths is provided.

However, if you have hundreds gene/ranges need to extract from very large VCF file(s), it's prefer to process with other linux tools in a script on server, such as: 'vcftools' and 'bcftools'.

filter\_hap 9

## Value

No return value

## **Examples**

```
# The filteration of small vcf should be done with `filter_vcf()`.
 # however, here, we use a mini vcf instead just for example and test.
 vcfPath <- system.file("extdata", "var.vcf.gz", package = "geneHapR")</pre>
 oldDir <- getwd()
 temp_dir <- tempdir()</pre>
 if(! dir.exists(temp_dir))
  dir.create(temp_dir)
 setwd(temp_dir)
 # extract a single gene/range from large vcf
 filterLargeVCF(VCFin = vcfPath, VCFout = "filtered.vcf.gz",
                Chr = "scaffold_1", POS = c(4300,5000), override = TRUE)
 # extract multi genes/ranges from large vcf
 filterLargeVCF(VCFin = vcfPath,
                VCFout = c("filtered1.vcf.gz",
                            "filtered2.vcf.gz",
                           "filtered3.vcf.gz"),
                Chr = rep("scaffold_1", 3),
                POS = list(c(4300, 5000),
                           c(5000, 6000),
                           c(5000, 7000)),
                override = TRUE)
setwd(oldDir)
```

filter\_hap

Filter hap

## **Description**

filter hapResult or hapSummary by remove positions or accessions or haplotypes

# Usage

10 filter\_hmp

## **Arguments**

| hap          | object of hapSummary or hapResult class   |
|--------------|---|
| rm.mode      | filter mode, one of "position", "accession", "haplotype"  |
| position.rm  | numeric vector contains positions need to be removed  |
| accession.rm | character vector contains accessions need to be removed, only hapResult can be filtered by accessions |
| haplotype.rm | character vector contains haplotypes need to be removed   |
| freq.min     | numeric, hapltypes with accessions number less than freq.min will be removed                          |

## Value

hapSummary or hapResult depend input

## **Examples**

filter\_hmp

filter variants in hapmap format

## **Description**

filter variants in hapmap format

# Usage

```
filter_hmp(
    x,
    mode = c("POS", "type", "both"),
    Chr = Chr,
    start = start,
    end = end,
    gff = gff,
    type = type,
    cusTyp = cusTyp,
    geneID = geneID
)
```

filter\_plink.pedmap 11

# Arguments

| X      | genotype dataset in hapmap format, object of data.frame class   |
|--------|---|
| mode   | filter mode, one of "POS", "type", "both"   |
| Chr    | chromosome name, needed if mode set to "POS" or "both"  |
| start  | start position, needed if mode set to "POS" or "both"   |
| end    | end position, needed if mode set to "POS" or "both"   |
| gff    | object of GRanges class, genome annotations imported by import_gff()  |
| type   | filter type, needed if mode set to "type" or "both", one of "CDS", "exon", "gene", "genome", "custom", if type was set to "custom", then custom_type is needed. |
| cusTyp | character vector, custom filter type, needed if type set to "custom"  |
| geneID | gene ID   |
|        |   |

# **Examples**

```
filter_plink.pedmap
```

# Description

used for filtration of p.link

# Usage

# Arguments

| Χ          | a list stored the p.link information                                    |
|------------|---|
| mode       | filtration mode, one of c("POS", "type", "both")                        |
| Chr        | the chromosome name, need if mode set as POS or both                    |
| start, end | numeric, the range of filtration, and the start should smaller than end |
| gff        | the imported gff object   |

filter\_table

```
type should be in unique(gff$type), usually as "CDS", "genome".

cusTyp if type set as custom, then cusTyp is needed
geneID geneID
```

## Value

list, similar with x, but filtered

## **Examples**

filter\_table

filter variants stored in table

# Description

filter variants stored in table

## Usage

```
filter_table(
    x,
    mode = c("POS", "type", "both"),
    Chr = Chr,
    start = start,
    end = end,
    gff = gff,
    type = type,
    cusTyp = cusTyp,
    geneID = geneID
)
```

filter\_vcf

## **Arguments**

| X      | genotype dataset in hapmap format, object of data.frame class   |
|--------|---|
| mode   | filter mode, one of "POS", "type", "both"   |
| Chr    | chromosome name, needed if mode set to "POS" or "both"  |
| start  | start position, needed if mode set to "POS" or "both"   |
| end    | end position, needed if mode set to "POS" or "both"   |
| gff    | object of GRanges class, genome annotations imported by import_gff()  |
| type   | filter type, needed if mode set to "type" or "both", one of "CDS", "exon", "gene", "genome", "custom", if type was set to "custom", then custom_type is needed. |
| cusTyp | character vector, custom filter type, needed if type set to "custom"  |
| geneID | gene ID   |
|        |   |

# **Examples**

filter\_vcf

Filter VCF

# Description

filter VCF by GFF annotation or by position or both

# Usage

## Arguments

| vcf   | object of vcfR class, VCF file imported by import_vcf()              |
|-------|--|
| gff   | object of GRanges class, genome annotations imported by import_gff() |
| mode  | filter mode, one of "POS", "type", "both"                            |
| Chr   | chromosome name, needed if mode set to "POS" or "both"               |
| start | start position, needed if mode set to "POS" or "both"                |
| end   | end position, needed if mode set to "POS" or "both"                  |

14 getGenePOS

#### Value

vcfR

## **Examples**

getGenePOS

Get Gene Position

# Description

Get Gene Position

#### Usage

## **Arguments**

```
gff imported gff
geneID target geneID

type vector consist with one or more types in gff
gffTermContaingeneID
 which term contains the geneID in your gff, defalt is Parent
```

getGeneRanges 15

## Value

named vectors contains start, end and strand

# **Examples**

getGeneRanges

Get Gene Ranges

## **Description**

Get Gene Ranges

# Usage

# **Arguments**

```
gff imported gff
geneID target geneID

type vector consist with one or more types in gff
gffTermContaingeneID
 which term contains the geneID in your gff, defalt is Parent
```

# Value

GRanges

16 hap2hmp

| hap2hmp | Convert hapResult object to hapmap (hmp) format, for interact with other packages |
|---------|---|
|         |   |

## **Description**

Convert hapResult object to hapmap (hmp) format, for interact with other packages

## Usage

```
hap2hmp(hap)
hmp2hap(hmp, hapPrefix = "H", hetero_remove = TRUE, na_drop = TRUE, ...)
```

## **Arguments**

hap object of "hapResult" class

hmp object of "data.frame" class in hapmap format

hapPrefix prefix of haplotype names

hetero\_remove whether remove accessions contains hyb-sites, Character not A T C G

na\_drop whether drop accessions contains missing data ("N", "NA", ".")

... Arguments passed on to table2hap

x a data.frame contains variants information. The first file column are fix as Chrome name, position, reference nuclieotide, alter nuclieotide and INFO.
 Accession genotype should be in followed columns. "-" will be treated as Indel. "." and "N" will be treated as missing data. Heterozygotes should be

"A/T", "AAA/A"

pad The number length in haplotype names should be extend to.

## Value

a data.frame in hapmap format.

```
data("geneHapR_test")
hmp <- hap2hmp(hapResult)
hap <- hmp2hap(hmp)</pre>
```

hapDistribution 17

hapDistribution

Display of Geography Distribution

## **Description**

show distribution of intereted haplotypes on maps

# Usage

```
hapDistribution(
  hap,
 AccINFO,
 LON.col,
 LAT.col,
  hapNames,
  database = "world",
  regions = ".",
  hap.color = hap.color,
  zColours = zColours,
  legend = TRUE,
  symbolSize = 1,
  symbol.lim = c(1, 10),
  ratio = 1,
  cex.legend = 0.8,
  lwd.pie = 1,
  borderCol.pie = NA,
  lty.pie = 1,
  showlabel = TRUE,
  label.col = "black",
  label.cex = 0.8,
  label.font = 1,
  label.adj = c(0.5, 0.5),
 map.fill.color = 1,
)
```

# **Arguments**

database

hap an object of hapResult class

AccINFO a data.frame contains accession information

LON.col, LAT.col

column names of longitude(LON.col) and latitude(LAT.col)

hapNames haplotype names used for display

character string naming a geographical database, a list of x, y, and names obtained from a previous call to map or a spatial object of class SpatialPolygons or SpatialLines. The string choices include a world map, three USA databases

18 hapDistribution

> (usa, state, county), and more (type help(package='maps') to see the package index). If the requied database is in a different package that has not been attached, the string may be started with "packagename::". The location of the map databases may be overridden by setting the R\_MAP\_DATA\_DIR environment variable.

regions

character vector that names the polygons to draw. Each database is composed of a collection of polygons, and each polygon has a unique name. When a region is composed of more than one polygon, the individual polygons have the name of the region, followed by a colon and a qualifier, as in michigan: north and michigan: south. Each element of regions is matched against the polygon names in the database and, according to exact, a subset is selected for drawing. The regions may also be defined using (perl) regular expressions. This makes it possible to use 'negative' expressions like "Norway(?!:Svalbard)", which means Norway and all islands except Svalbard. All entries are case insensitive. The default selects all polygons in the database.

hap.color, zColours

colors to apply to the pie section for each attribute column, "zColours" will be

detached in future.

legend a keyword specified the position of legend, one of "bottomright", "bottom", "bot-

tomleft", "left", "topleft", "top", "topright", "right" and "center"; or a numeric

vector of length two contains x,y coordinate of the legend

symbolSize a numeric specified the symbol size. It will be detached in future. Please use

"symbol.lim" instead.

symbol.lim a numeric vector give the maximum and minimum size of each symbol

the ratio of Y to N in the output map, set to 1 as default ratio

cex.legend character expansion factor for legend relative to current par("cex")

lwd.pie line width of the pies

The color of pie's border, default is NA, which means no border will be plotted

lty.pie the line type of pie border

showlabel a bool vector indicates whether show the labels which represens number of in-

dividuals. Default as TRUE.

label.col color of the labels, default as "black"

label.cex a number indicates the text size in label, default as 0.8

Font of label, 1 for normal, 2 for bold, 3 for italica, 4 for bold-italica

label.adi the position of label, default as c(0.5, 0.5)

map.fill.color vector of colors. If fill is FALSE, the first color is used for plotting all lines, and

any other colors are ignored. Otherwise, the colors are matched one-one with the polygons that get selected by the region argument (and are reused cyclically, if necessary). If fill = TRUE, the default boundary line colour is given by par("fg"). To change this, you can use the border argument (see '...'). A color of NA causes the corresponding region to be deleted from the list of polygons to be drawn. Polygon colors are assigned after polygons are deleted due to values of the xlim

and ylim arguments

Extra arguments passed to polygon or lines. Of particular interest may be

the options border and ty that control the color and line type of the polygon

borders when fill = TRUE.

borderCol.pie

label.font

## Value

No return value

## **Examples**

hapVsPheno

hapVsPheno

# Description

hapVsPheno

## Usage

```
hapVsPheno(
  hap,
  pheno,
  phenoName,
  hapPrefix = "H",
  title = "",
  comparisons = comparisons,
 method = "t.test",
 method.args = list(),
  symnum.args = list(),
 mergeFigs = FALSE,
  angle = angle,
  hjust = hjust,
  vjust = vjust,
 minAcc = minAcc,
  freq.min = freq.min,
  outlier.rm = TRUE,
)
```

# Arguments

```
hap object of hapResult class, generate withvcf2hap() or seqs2hap()
pheno object of data.frame class, imported by import_pheno()
phenoName pheno name for plot, should be one column name of pheno
```

hapPrefix prefix of hapotypes, default as "H"

title a charater which will used for figure title

comparisons a list contains comparison pairs eg. list(c("H001", "H002"), c("H001",

"H004")), or a character vector contains haplotype names for comparison, or

"none" indicates do not add comparisons.

method a character string indicating which method to be used for comparing means.

method.args a list of additional arguments used for the test method. For example one might

use method.args = list(alternative = "greater") for wilcoxon test.

symnum.args a list of arguments to pass to the function symnum for symbolic number coding

of p-values. For example, symnum.args <- list(cutpoints = c(0, 0.0001, 0.001, 0.001, 0.005, Inf), symbols = c("\*\*\*\*", "\*\*", "\*\*", "\*\*", "ns")).

In other words, we use the following convention for symbols indicating statistical significance:

• ns: p > 0.05

• \*: p <= 0.05

• \*\*: p <= 0.01

• \*\*\*: p <= 0.001

• \*\*\*\*: p <= 0.0001

mergeFigs bool type, indicate whether merge the heat map and box plot or not. Default as

**FALSE** 

angle the angle of x labels

hjust, vjust hjust and vjust of x labels

minAcc, freq.min

If observations number of a Hap less than this number will not be compared with others or be ploted. Should not less than 3 due to the t-test will meaninglessly.

Default as 5

outlier.rm whether remove ouliers, default as TRUE

... Arguments passed on to ggpubr::ggviolin

data a data frame

x character string containing the name of x variable.

y character vector containing one or more variables to plot

combine logical value. Default is FALSE. Used only when y is a vector containing multiple variables to plot. If TRUE, create a multi-panel plot by combining the plot of y variables.

merge logical or character value. Default is FALSE. Used only when y is a vector containing multiple variables to plot. If TRUE, merge multiple y variables in the same plotting area. Allowed values include also "asis" (TRUE) and "flip". If merge = "flip", then y variables are used as x tick labels and the x variable is used as grouping variable.

color outline color.

fill fill color.

palette the color palette to be used for coloring or filling by groups. Allowed values include "grey" for grey color palettes; brewer palettes e.g. "RdBu", "Blues", ...; or custom color palette e.g. c("blue", "red"); and scientific journal palettes from ggsci R package, e.g.: "npg", "aaas", "lancet", "jco", "ucscgb", "uchicago", "simpsons" and "rickandmorty".

alpha color transparency. Values should be between 0 and 1.

xlab character vector specifying x axis labels. Use xlab = FALSE to hide xlab. ylab character vector specifying y axis labels. Use ylab = FALSE to hide ylab. facet.by character vector, of length 1 or 2, specifying grouping variables for faceting the plot into multiple panels. Should be in the data.

panel.labs a list of one or two character vectors to modify facet panel labels. For example, panel.labs = list(sex = c("Male", "Female")) specifies the labels for the "sex" variable. For two grouping variables, you can use for example panel.labs = list(sex = c("Male", "Female"), rx = c("Obs", "Lev", "Lev2")).

short.panel.labs logical value. Default is TRUE. If TRUE, create short labels for panels by omitting variable names; in other words panels will be labelled only by variable grouping levels.

linetype line types.

trim If TRUE (default), trim the tails of the violins to the range of the data. If FALSE, don't trim the tails.

size Numeric value (e.g.: size = 1). change the size of points and outlines. width violin width.

draw\_quantiles If not(NULL) (default), draw horizontal lines at the given quantiles of the density estimate.

select character vector specifying which items to display.

remove character vector specifying which items to remove from the plot.

order character vector specifying the order of items.

add character vector for adding another plot element (e.g.: dot plot or error bars). Allowed values are one or the combination of: "none", "dotplot", "jitter", "boxplot", "point", "mean, "mean\_se", "mean\_sd", "mean\_ci", "mean\_range", "median, "median\_iqr", "median\_hilow", "median\_q1q3", "median\_mad", "median\_range"; see ?desc\_statby for more details.

add.params parameters (color, shape, size, fill, linetype) for the argument 'add'; e.g.: add.params = list(color = "red").

error.plot plot type used to visualize error. Allowed values are one of c("pointrange", "linerange", "crossbar", "errorbar", "upper\_errorbar", "lower\_errorbar", "upper\_pointrange", "lower\_pointrange", "upper\_linerange", "lower\_linerange"). Default value is "pointrange" or "errorbar". Used only when add != "none" and add contains one "mean\_\*" or "med\_\*" where "\*" = sd, se, ....

label the name of the column containing point labels. Can be also a character vector with length = nrow(data).

font.label a list which can contain the combination of the following elements: the size (e.g.: 14), the style (e.g.: "plain", "bold", "italic", "bold.italic") and the color (e.g.: "red") of labels. For example font.label = list(size = 14, face = "bold", color = "red"). To specify only the size and the style, use font.label = list(size = 14, face = "plain").

label.select can be of two formats:

- a character vector specifying some labels to show.
- a list containing one or the combination of the following components:
  - top.up and top.down: to display the labels of the top up/down points. For example, label.select = list(top.up = 10, top.down = 4).
  - criteria: to filter, for example, by x and y variabes values, use this:
     label.select = list(criteria = "`y` > 2 & `y` < 5 & `x` %in% c('A',
     'B')").</pre>

repel a logical value, whether to use ggrepel to avoid overplotting text labels or not.

label.rectangle logical value. If TRUE, add rectangle underneath the text, making it easier to read.

position Position adjustment, either as a string naming the adjustment (e.g. "jitter" to use position\_jitter), or the result of a call to a position adjustment function. Use the latter if you need to change the settings of the adjustment.

ggtheme function, ggplot2 theme name. Default value is theme\_pubr(). Allowed values include ggplot2 official themes: theme\_gray(), theme\_bw(), theme minimal(), theme classic(), theme void(), ....

#### Value

list. A list contains a character vector with Haps were applied student test, a mattrix contains p-value of each compare of Haps and a ggplot2 object named as figs if mergeFigs set as TRUE, or two ggplot2 objects names as fig\_pvalue and fig\_Violin

hapVsPhenoPerSite 23

hapVsPhenoPerSite hapVsPhenoPerSite

hap Vs Pheno Per Site

# **Description**

Comparie phenotype site by site.

## Usage

```
hapVsPhenoPerSite(
  hap,
  pheno,
  phenoName,
  sitePOS,
  fileName,
  fileType = NULL,
  freq.min = 5,
  ...
)
```

# Arguments

```
data("geneHapR_test")
hapVsPhenoPerSite(hapResult, pheno, sitePOS = "4300")
```

hapVsPhenos

hapVsPhenos

## **Description**

hapVsPhenos

# Usage

```
hapVsPhenos(
  hap,
  pheno,
  outPutSingleFile = TRUE,
  hapPrefix = "H",
  title = "Seita.0G000000",
 width = 12,
  height = 8,
  res = 300,
  compression = "lzw",
  filename.prefix = filename.prefix,
  filename.surfix = "pdf",
  filename.sep = "_",
  outlier.rm = TRUE,
 mergeFigs = TRUE,
)
```

# **Arguments** hap

pheno object of data.frame class, imported by import\_pheno() outPutSingleFile TRUE or FALSE indicate whether put all figs into to each pages of single file or generate multi-files. Only worked while file type is pdf hapPrefix prefix of hapotypes, default as "H" a charater which will used for figure title title width manual option for determining the output file width in inches. (default: 12) height manual option for determining the output file height in inches. (default: 8) The nominal resolution in ppi which will be recorded in the bitmap file, if a res positive integer. Also used for units other than the default, and to convert points to pixels compression the type of compression to be used.

object of hapResult class, generate withvcf2hap() or seqs2hap()

filename.prefix, filename.surfix, filename.sep

if multi files generated, file names will be formed by prefix filename.prefix, a seperate charcter filename.sep, pheno name, a dot and surfix filename.surfix, and file type was decide by filename.surfix; if single file was generated, file name will be formed by prefix filename.prefix, a dot and surfix filename.surfix

outlier.rm

whether remove ouliers, default as TRUE

mergeFigs

bool type, indicate whether merge the heat map and box plot or not. Default as  $\ensuremath{\mathsf{FALSE}}$ 

Arguments passed on to hapVsPheno

phenoName pheno name for plot, should be one column name of pheno minAcc, freq.min If observations number of a Hap less than this number will not be compared with others or be ploted. Should not less than 3 due to the t-test will meaninglessly. Default as 5

angle the angle of x labels

hjust, vjust hjust and vjust of x labels

comparisons a list contains comparison pairs eg. list(c("H001", "H002"), c("H001", "H004")), or a character vector contains haplotype names for comparison, or "none" indicates do not add comparisons.

method a character string indicating which method to be used for comparing means.

method.args a list of additional arguments used for the test method. For example one might use method.args = list(alternative = "greater") for wilcoxon test.

symnum.args a list of arguments to pass to the function symnum for symbolic number coding of p-values. For example, symnum.args <- list(cutpoints = c(0, 0.0001, 0.001, 0.01, 0.05, Inf), symbols = c("\*\*\*\*", "\*\*", "\*\*", "\*\*", "s")).

In other words, we use the following convention for symbols indicating statistical significance:

- ns: p > 0.05
- \*:  $p \le 0.05$
- \*\*: p <= 0.01
- \*\*\*: p <= 0.001
- \*\*\*: p <= 0.0001

## Value

No return value

```
data("geneHapR_test")

oriDir <- getwd()
temp_dir <- tempdir()
if(! dir.exists(temp_dir))
  dir.create(temp_dir)</pre>
```

26 hap\_summary

hap\_summary

Summary Hap Results

#### **Description**

A function used for summarize hapResult to visualization and calculation.

## Usage

## **Arguments**

hap object of hapResult class, generated by vcf2hap() or seqs2hap or import\_hap()

hapPrefix prefix of hap names, default as "H"

file file path where to save the hap summary result. If missing, nothing will be saved

to disk.

#### Details

It is suggested to use the result of vcf2hap() or seqs2hap() as input directly. However the user can import previously hap result from local file with import\_hap()

#### Value

hapSummary, first four rows are fixed to meta information: CHR, POS, INFO, ALLELE Hap names were placed in first column, Accessions and freqs were placed at the last two columns.

## Note

If the user have changed the default hapPrefix in vcf2hap() or seqs2hap(), then the parameter hapPrefix is needed. Furthermore, a multi-letter prefix of hap names is possible.

import\_AccINFO 27

## **Examples**

```
data("geneHapR_test")
hapSummary <- hap_summary(hapResult, hapPrefix = "H")</pre>
```

import\_AccINFO

Import Accession Information from File

## **Description**

import accession information including phenotype data, accession group, location from a tab delimed table file

# Usage

# **Arguments**

file file path, this file should be a tab delimed table

comment.char character: a character vector of length one containing a single character or an

empty string. Use "" to turn off the interpretation of comments altogether.

check.names logical. If TRUE then the names of the variables in the data frame are checked

to ensure that they are syntactically valid variable names. If necessary they are adjusted (by make.names) so that they are, and also to ensure that there are no

duplicates.

row.names a vector of row names. This can be a vector giving the actual row names, or a

single number giving the column of the table which contains the row names, or character string giving the name of the table column containing the row names.

If there is a header and the first row contains one fewer field than the number of columns, the first column in the input is used for the row names. Otherwise if

row. names is missing, the rows are numbered.

Using row.names = NULL forces row numbering. Missing or NULL row.names generate row names that are considered to be 'automatic' (and not preserved by

as.matrix).

Further arguments to be passed to read.table.

## **Details**

First column should be Accessions; phenos/accession information should begin from second column, phenoName/group/locations should located at the first row, If a dot '.' is located in pheno name, then the part before the dot will be set as y axis name and the latter will be set as foot when plot figures.

28 import\_bed

#### Value

data.frame, Accession names were set as rownames and columns were named by pheno/info names

## **Examples**

```
oldDir <- getwd()
temp_dir <- tempdir()
if(! dir.exists(temp_dir))
    dir.create(temp_dir)
setwd(temp_dir)
data("geneHapR_test")
write.table(pheno, file = "test.pheno.txt", sep = "\t")
pheno <- import_AccINFO("test.pheno.txt")
pheno
setwd(oldDir)</pre>
```

import\_bed

import annotation files in BED format

## Description

import bed files contains annotations into R as GRanges object

## Usage

```
import_bed(con, quite = FALSE)
```

## **Arguments**

con

A path, URL, connection or BEDFile object. For the functions ending in .bed, .bedGraph and .bed15, the file format is indicated by the function name. For the base export and import functions, the format must be indicated another way. If con is a path, URL or connection, either the file extension or the format argument needs to be one of "bed", "bed15", "bedGraph", "bedpe", "narrow-Peak", or "broadPeak". Compressed files ("gz", "bz2" and "xz") are handled transparently.

quite

whether show message

## **Details**

If there is no genome annotation file in GFF format for your interest species, a BED file is convenient to custom a simple annotation file for a gene. Here we suggest two type of BED format: BED6 and BED4.

As the definition of UCSC. The BED6 contains 6 columns, which are 1) chrom, 2) chromStart, 3) chromEnd, 4) name, 5) score and 6) strand. The BED4 format contains the first 4 column of BED6 format.

import\_gff 29

However, in gene haplotype statistics, we only care about the type of each site. Thus we use the fourth column to definition the transcripts name and "CDS" or "URTs", separated by a space, eg.:

```
Chr8 678 890 HD1.1 CDS . -
Chr8 891 989 HD1.1 five_prime_UTR . -
Chr8 668 759 HD1.2 CDS . -
Chr8 908 989 HD1.2 CDS . -
```

This example indicate a small gene named as HD1 have two transcripts, named as HD1.1 and HD1.2, separately. HD1 has a CDS and a UTR region; while HD1.2 has two CDS region.

#### Value

GRange object

## **Examples**

```
bed.Path <- system.file("extdata", "annotation.bed6", package = "geneHapR")
bed <- import_bed(bed.Path)
bed</pre>
```

import\_gff

Import Annotations from GFF Format File

## **Description**

import genome annotations in GFF/GFF3 format

## Usage

```
import_gff(gffFile, format = "gff")
```

## **Arguments**

```
gffFile the gff file path
```

format should be one of "gff", "gff1", "gff2", "gff3", "gvf", or "gtf". Default as gff

## Value

GRange object

```
gff.Path <- system.file("extdata", "annotation.gff", package = "geneHapR")
gff <- import_gff(gff.Path, format = "gff")
gff</pre>
```

30 import\_hap

Import hapResult/hapSummary

# Description

This function could be used for import hap result or hap summary result. The type of returned object is decided by input file, see details.

## Usage

```
import_hap(file, type = "auto", ...)
```

## Arguments

```
file hapSummary or hapResult file path.

type the content type of imported file, should be one of c("hapResult", "hapSummary")

... extras will pass to read.delim()
```

## **Details**

The hap result and hap summary result have common features. The common features of these two types are: First four rows contains extra information: CHR, POS, INFO and ALLELE Hap names were in the first column. The differences are: Hap summary result have a freq column while hap result not. Rows represent haplotypes in hap summary result, while rows represent accessions in hap result. In addition, the accessions of each haplotype in hap summary result were separated by ":".

#### Value

hapSummary or hapResult

```
oldDir <- getwd()
temp_dir <- tempdir()
if(! dir.exists(temp_dir))
    dir.create(temp_dir)
setwd(temp_dir)
data("geneHapR_test")
write.hap(hapResult, file = "test.pheno.txt", sep = "\t")
hap <- import_hap("test.pheno.txt")
hap
setwd(oldDir)</pre>
```

```
import_MultipleAlignment
```

Import MultipleAlignment Result

## **Description**

import sequences algned results

## Usage

```
import_MultipleAlignment(filepath, format = "fasta", type = "DNA")
```

## **Arguments**

filepath A character vector (of arbitrary length when reading, of length 1 when writing)

containing the paths to the files to read or write. Note that special values like "" or "|cmd" (typically supported by other I/O functions in R) are not supported

here. Also filepath cannot be a connection.

format Either "fasta" (the default), stockholm, or "clustal".

type one of "DNA" and "Protein"

#### Value

object of DNAMultipleAlignment

## **Examples**

import\_plink.pedmap

import\_plink.pedmap

## **Description**

used for import regular p.link file stored in map and ped format

32 import\_seqs

## Usage

## Arguments

root The file name without suffix. This function only support p.link file format stored

in "map" and "ped" format, the file names after removed suffix should be same

with each other.

sep\_ped a character indicate the separation of ped file sep\_map a character indicate the separation of map file

pedfile, mapfile

if root is missing then pedfile and mapfile are needed

## Value

list, contains map information stored in data.frame and ped information stored in data.frame

## **Examples**

import\_seqs

Import Sequences

#### **Description**

import DNA sequences in FASTA format

## Usage

```
import_seqs(filepath, format = "fasta")
```

import\_vcf 33

## **Arguments**

filepath A character vector containing the path to the DNA sequences file. Reading files

in gzip format (which usually have the '.gz' extension) is supported. Note that

only DNA supported here.

format Either "fasta" (the default) or "fastq"

#### Value

object of DNAStringSet class

## **Examples**

```
seqPath <- system.file("extdata", "seqs.fa", package = "geneHapR")
geneSeqs <- import_seqs(filepath = seqPath, format = "fasta")</pre>
```

import\_vcf

Import VCF from File

# Description

import \*.vcf structured text format, as well as the compressed \*.vcf.gz format.

## Usage

```
import_vcf(file = file, ...)
import_vcf(file = file, ...)
```

## **Arguments**

```
file file path of VCF file
... pass to vcfR::read.vcfR()
```

## Value

vcfR object

## Author(s)

Zhangrenl

## See Also

```
vcfR::read.vcfR()
```

34 LDheatmap

## **Examples**

```
vcfPath <- system.file("extdata", "var.vcf.gz", package = "geneHapR")
vcf <- import_vcf(file = vcfPath)
vcf</pre>
```

LDheatmap

This function produces a pairwise LD plot.

## **Description**

LDheatmap() is used to produce a graphical display, as a heat map, of pairwise linkage disequilibrium (LD) measurements for SNPs. The heat map is a false color image in the upper-left diagonal of a square plot. Optionally, a line parallel to the diagonal of the image indicating the physical or genetic map positions of the SNPs may be added, along with text reporting the total length of the genomic region considered.

## Usage

```
plot_LDheatmap(
  hap,
  gff,
 Chr,
  start,
  end,
  geneID = NULL,
 distances = "physical",
 LDmeasure = "r",
  title = "Pairwise LD",
  add.map = TRUE,
 map.height = 1,
  colorLegend = TRUE,
  geneMapLocation = 0.15,
  geneMapLabelX = NULL,
  geneMapLabelY = NULL,
  SNP.name = TRUE,
  color = NULL,
  color_gmodel = "grey",
  color_snp = "grey",
  color_snpname = "grey40",
  cex_snpname = 0.8,
  snpmarks_height = NULL,
  newpage = TRUE,
  name = "ldheatmap",
  vp.name = NULL,
 pop = FALSE,
  text = FALSE
)
```

LDheatmap 35

#### **Arguments**

hap R object of hapSummary or hapResult class.

gff annotations Chr, start, end, geneID

chromosome, start and end position, gene ID for extract annotation in target

range.

distances A character string to specify whether the provided map locations are in physical

or genetic distances. If distances = "physical" (default), the text describing the total length of the region will be "Physical Length:XXkb" where XX is the length of the region in kilobases. If distances = "genetic", the text will be "Genetic Map Length:YYcM" where YY is the length of the region in centi-Morgans. If gdat is an object of class LDheatmap, distances is taken from

gdat.

LDmeasure A character string specifying the measure of LD

• either allelic correlation  $r^2$  or Lewontin's |D'|; default = "r" for  $r^2$ ; type "D'" for |D'|. This argument is ignored when the user has already supplied calculated LD measurements through gdat (i.e., when gdat is a matrix of

pairwise LD measurements or an object of class "LDheatmap").

title A character string for the main title of the plot. Default is "Pairwise LD".

add.map If TRUE (default) a diagonal line indicating the physical or genetic map positions

of the SNPs will be added to the plot, along with text indicating the total length

of the genetic region.

map.height the height of gene map, default is 0.02

colorLegend If TRUE (default) the color legend is drawn.

geneMapLocation

A numeric value specifying the position of the line parallel to the diagonal of the matrix; the larger the value, the farther it lies from the matrix diagonal. Ignored

when add.map = FALSE.

geneMapLabelX A numeric value specifying the x-coordinate of the text indicating the total

length of the genomic region being considered. Ignored when add.map = FALSE.

geneMapLabelY A numeric value specifying the y-coordinate of the text indicating the total

length of the genomic region being considered. Ignored when add.map = FALSE.

SNP. name a logical vector indicated wherther display SNP names using positions.

color A range of colors to be used for drawing the heat map. Default is grDevices::colorRampPalette(c("re

"grey"))(30).

color\_gmodel, color\_snp, color\_snpname

the color of gene model and snp and snp names respectively, default as grey80.

cex\_snpname the size of snp names/labels

snpmarks\_height

the height of snp marks, if set as NULL, nothing will display on gene model

where the heat map is going to be drawn.

newpage If TRUE (default), the heat map will be drawn on a new page.

name A character string specifying the name of the LDheatmap graphical object (grob)

to be produced.

vp. name A character string specifying the name of the viewport

pop If TRUE, the viewport where the heat map is drawn is popped (i.e. removed) from

the viewport tree after drawing. Default = FALSE.

text If TRUE, the LD measurements are printed on each cell.

#### **Details**

The input object gdat can be a data frame of genotype objects (a data structure from the **genetics** package), a SnpMatrix object (a data structure from the **snpStats** package), or any square matrix with values between 0 and 1 inclusive. LD computation is much faster for SnpMatrix objects than for genotype objects. In the case of a matrix of LD values between 0 and 1, the values above the diagonal will be plotted. In the display of LD, SNPs appear in the order supplied by the user as the horizontal and vertical coordinates are increased and one moves along the off-diagonal line, from the bottom-left to the top-right corner. To achieve this, the conventions of the image() function have been adopted, in which horizontal coordinates correspond to the rows of the matrix and vertical coordinates correspond to columns, and vertical coordinates are indexed in increasing order from bottom to top. See the package vignette LDheatmap for more examples and details of the implementation. Examples of adding "tracks" of genomic annotation above a flipped heatmap are in the package vignette addTracks.

#### Value

An object of class "LDheatmap" which contains the following components:

LDmatrix The matrix of pairwise LD measurements plotted in the heat map.

LDheatmapGrob A grid graphical object (grob) representing the produced heat map.

heatmapVP The viewport in which the heat map is drawn. See viewport.

genetic.distances

The vector of the supplied physical or genetic map locations, or the vector of

equispaced marker distances when no distance vector is supplied.

distances A character string specifying whether the provided map distances are physical

or genetic.

color The range of colors used for drawing the heat map.

The grob LDheatmapGrob has three grobs as its children (components). They are listed below along with their own children and respectively represent the color image with main title, genetic map and color key of the heat map: "heatMap" - "heatmap", "title"; "geneMap" - "diagonal", "segments", "title", "symbols", "SNPnames"; and "Key" - "colorKey", "title", "labels", "ticks", "box".

## Note

The produced heat map can be modified in two ways. First, it is possible to edit *interactively* the grob components of the heat map, by using the function <code>grid.edit</code>; the function will not work if there is no open graphical device showing the heat map. Alternatively, the user can use the

network 37

function editGrob and work with the grob LDheatmapGrob returned by LDheatmap. See Examples for usage. LDheatmap() uses Grid, which does not respond to par() settings. Hence modifying par() settings of mfrow and mfcol will not work with LDheatmap(). The Examples section shows how to display multiple heat maps on one plot without the use of par().

#### References

Shin J-H, Blay S, McNeney B and Graham J (2006). LDheatmap: An R Function for Graphical Display of Pairwise Linkage Disequilibria Between Single Nucleotide Polymorphisms. Journal of Statistical Software, **16** Code Snippet 3

#### **Examples**

network

Generate Haplotype Net Relationshop with Haplotype Result

#### **Description**

computes a haplotype network with haplotype summary result

#### Usage

#### Arguments

hapSummary object of hapSummary class, generated by hap\_summary()

AccINFO

data.frame, specified groups of each accession. Used for pie plot. If missing, pie will not draw in plotHapNet. Or you can supplied a hap\_group mattrix with plot(hapNet, pie = hap\_group).

38 plink.pedmap2hap

groupName the group name used for pie plot, should be in AccINFO column names, default

as the first column name

na.label the label of NAs

#### Value

hapNet class

#### References

```
Mark P.J. van der Loo (2014) doi:10.32614/RJ2014011;
E. Paradis (2010) doi:10.1093/bioinformatics/btp696
```

## See Also

```
plotHapNet() and hap_summary().
```

#### **Examples**

```
data("geneHapR_test")
hapSummary <- hap_summary(hapResult)</pre>
# calculate haploNet
hapNet <- get_hapNet(hapSummary,</pre>
                     AccINFO = AccINFO, # accession types
                     groupName = colnames(AccINFO)[2])
# plot haploNet
plot(hapNet)
# plot haploNet
plotHapNet(hapNet,
           size = "freq", # circle size
           scale = "log10", # scale circle with 'log10(size + 1)'
           cex = 1, # size of hap symbol
           col.link = 2, # link colors
           link.width = 2, # link widths
           show.mutation = 2, # mutation types one of c(0,1,2,3)
           legend = FALSE) # legend position
```

plink.pedmap2hap

plink.pedmap2hap

# Description

convert p.link format data into hapResult

plotEFF 39

#### Usage

```
plink.pedmap2hap(
  p.link,
  hapPrefix = "H",
  pad = 3,
  hetero_remove = TRUE,
  na_drop = TRUE
)
```

## **Arguments**

p.link list contains p.link information
 hapPrefix prefix of haplotype names
 pad The number length in haplotype names should be extend to.
 hetero\_remove whether remove accessions contains hyb-sites

na\_drop whether drop accessions contains missing data ("N", NA)

#### Value

object of hapSummary class

#### **Examples**

plotEFF

plotEFF

## Description

plotEFF

40 plotEFF

#### Usage

```
plotEFF(
  siteEFF,
  gff = gff,
  Chr = Chr,
  start = start,
  end = end,
  showType = c("five_prime_UTR", "CDS", "three_prime_UTR"),
  CDS.height = CDS.height,
  cex = 0.1,
  col = c("red", "yellow"),
  pch = 20,
 main = main,
  legend.cex = 0.8,
  gene.legend = TRUE,
 markMutants = TRUE,
 mutants.col = 1,
  mutants.type = 1,
  y = c("pvalue", "effect"),
  ylab = ylab,
  legendtitle = legendtitle,
  par.restore = TRUE
)
```

#### **Arguments**

mutants.type

```
siteEFF
                  matrix, column name are pheno names and row name are site position
gff
                  gff annotation
Chr
                  the chromosome name
start
                  start position
                  end position
end
                  character vector, eg.: "CDS", "five_prime_UTR", "three_prime_UTR"
showType
CDS.height
                  numeric indicate the height of CDS in gene model, range: [0,1]
cex
                  a numeric control the size of point
col
                  vector specified the color bar
pch
                  vector controls points type, see par()
                  main title
main
                  a numeric control the legend size
legend.cex
gene.legend
                  whether add legend for gene model
markMutants
                  whether mark mutants on gene model, default as TRUE
mutants.col
                  color of lines which mark mutants
```

a vector of line types

plotHapNet 41

```
y, ylab, legendtitle
y: indicate either pvalue or effect should be used as y axix, ylab,legendtitle:,character, if missing, the value will be decide by y.

par.restore default as TRUE, wether restore the origin par after ploted EFF.
```

#### Value

No return value, called for side effects

## **Examples**

```
data("geneHapR_test")

# calculate site functional effect
# siteEFF <- siteEFF(hapResult, pheno, names(pheno))
# plotEFF(siteEFF, gff = gff, Chr = "scaffold_1")</pre>
```

plotHapNet

plotHapNet

## **Description**

plotHapNet

#### Usage

```
plotHapNet(
 hapNet,
  size = "freq",
  scale = 1,
  cex = 0.8,
  cex.legend = 0.6,
  col.link = 1,
  link.width = 1,
  show.mutation = 2,
  backGround = backGround,
  hapGroup = hapGroup,
  legend = FALSE,
  show_size_legend = TRUE,
  show_color_legend = TRUE,
  pie.lim = c(0.5, 2),
 main = main,
  labels = TRUE,
  legend_version = 0,
  labels.cex = 0.8,
  labels.col = "blue",
  labels.adj = NULL,
```

42 plotHapNet

```
labels.font = 2,
...
)
```

#### **Arguments**

hapNet an object of class "haploNet"

size a numeric vector giving the diameter of the circles representing the haplotypes:

this is in the same unit than the links and eventually recycled.

scale a numeric indicate the ratio of the scale of the links representing the number

of steps on the scale of the circles representing the haplotypes or a character one of c('log10', 'log2') indicate the scale method by log10(size) or

log2(size), respectively. Default as 1

cex character expansion factor relative to current par("cex")

cex.legend same as cex, but for text in legend

col.link a character vector specifying the colours of the links; eventually recycled.

link.width a numeric vector giving the width of the links; eventually recycled.

show.mutation an integer value:

if 0, nothing is drawn on the links;

if 1, the mutations are shown with small segments on the links;

if 2, they are shown with small dots;

if 3, the number of mutations are printed on the links.

backGround a color vector with length equal to number of Accession types

hapGroup a matrix used to draw pie charts for each haplotype; its number of rows must be

equal to the number of haplotypes

legend a logical specifying whether to draw the legend, or a vector of length two giving

the coordinates where to draw the legend; FALSE by default. If TRUE, the user is

asked to click where to draw the legend.

show\_size\_legend, show\_color\_legend

wether show size or color legend

pie.lim A numeric vector define the maximum and minmum pie size, which will be

avoid the pie to samll or too large

main The main title (on top) using font, size (character expansion) and color par(c("font.main",

"cex.main", "col.main")).

labels a logical specifying whether to identify the haplotypes with their labels (default

as TRUE)

legend\_version the size legened style, default as 0

labels.cox the size of labels labels.col the labels color

labels.adj a named list contains two length vectors defining the adjustment of labels. The

names should be exactly matched with the haplotype names. default as NULL.

labels. font the font of labels, default as 2

... other parameters will pass to plot function

plotHapNet 43

#### **Details**

Additional parameters control the network features: labels.cex = 1, labels.font = 2, link.color = "black", link.type = 1, link.type.alt = 2, link.width = 1, link.width.alt = 1, altlinks = TRUE, threshold = c(1,2), haplotype.inner.color = "white", haplotype.outer.color = "black", mutations.cex = 1, mutations.font = 1, mutations.frame.background = "#0000FF4D", mutations.frame.border = "black", mutations.text.color = 1, mutations.arrow.color = "black", mutations.arrow.type = "triangle", mutations.sequence.color = "#BFBFBF4D", mutations.sequence.end = "round", mutations.sequence.length = 0.3, mutations.sequence.width = 5, pie.inner.segments.color = "black", pie.colors.function = rainbow, scale.ratio = 1, show.mutation = 2

The alter links could be eliminated by set the 'threshold' to 0 or set 'altlinks' as FALSE.

#### Value

No return value

#### See Also

```
hap_summary() and get_hapNet().
```

```
data("geneHapR_test")
hapSummary <- hap_summary(hapResult)</pre>
# calculate haploNet
hapNet <- get_hapNet(hapSummary,</pre>
                     AccINFO = AccINFO, # accession types
                     groupName = colnames(AccINFO)[2])
# plot haploNet
plot(hapNet)
# plot haploNet
plotHapNet(hapNet,
           size = "freq", # circle size
           scale = "log10", # scale circle with 'log10(size + 1)'
           cex = 1, # size of hap symbol
           col.link = 2, # link colors
           link.width = 2, # link widths
           show.mutation = 2, # mutation types one of c(0,1,2,3)
           legend = FALSE) # legend position
```

44 plotHapTable

# Description

display hap result as a table-like figure

# Usage

# Arguments

| hapSummary         | object of hapSummary class   |  |  |  |
|--------------------|--|--|--|--|
| hapPrefix          | prefix of haplotype names. Default as "H"  |  |  |  |
| title              | the main title of the final figure   |  |  |  |
| geneName           | character, will be used for filter INFO filed of ANN   |  |  |  |
| INFO_tag           | The annotations in the INFO field are represented as tag-value pairs, where the tag and value are separated by an equal sign, ie "=", and pairs are separated by colons, ie ";". For more information please see details.                        |  |  |  |
| tag_split          | usually, the value of tag-value contains one information. However, if a tag contains more than one fields, eg "ANN", then tag_split is needed. When INFO_tag was set as "ANN" or "SNPEFF", tag_split will be set as "I" by default, see details. |  |  |  |
| tag_field          | integer, if a tag-value contains more than one fields, user need to specified which field should be display. If tag_field set as 0, the whole contents will be displayed. Default as 0.  |  |  |  |
| tag_name           | tag name is displayed in Hap figure. If tag_name is missing, will take the value of INFO_tag.  |  |  |  |
| displayIndelSize   |  |  |  |  |
|                    | display indels with max size of displayIndelSize, If set as 0, all indels will convert into "i*" of which "i" represents "indel".  |  |  |  |
| angle              | the angle of coordinates, should be one of 0, 45 and 90  |  |  |  |
| replaceMultiAllele |  |  |  |  |
|                    | whether to replace MultiAllele with "T*", default as TRUE.   |  |  |  |
| ALLELE.color       | the color of ALLELE row, default as "grey90"   |  |  |  |

plotHapTable 45

#### **Details**

In **VCF** files, the INFO field are represented as tag-value pairs, where the tag and value are separated by an equal sign, ie "=", and pairs are separated by colons, ie ";".

If hapSummarys were generated from sequences, INFO row is null. If hapSummarys were generated from VCF, INFO was take from the INFO column in the source VCF file. Some tag-values may contains more than one value separated by "I", eg.: "ANN" or "snpEFF" added by 'snpeff' or other software. For those fields we need specified value of tag\_field = "ANN" and tag\_split = "[\]", it's suggest specified the value of tag\_name for display in figure.

'snpeff', a toolbox for genetic variant annotation and functional effect prediction, will add annotations to INFO filed in VCF file under a tag named as "ANN". The annotations contains several fields separated by "I". eg.:

- 1. Allele
- 2. Annotation
- 3. Annotation\_Impact
- 4. Gene Name
- 5. Gene\_ID
- 6. Feature\_Type
- 7. Feature\_ID
- 8. Transcript\_BioType
- 9. Rank
- 10. HGVS.c
- 11. HGVS.p
- 12. cDNA.pos/cDNA.length ... ...

However, the INFO in hapResults may missing annotations that we need. In this case, we can custom INFOs in hapSummarys with addINFO(). Once the needed annotations were included in hap results, we can display them with plotHapTable() by specify the value of INFO\_tag.

#### Value

```
ggplot2 object
```

#### See Also

```
addINFO()
```

```
data("geneHapR_test")
plotHapTable(hapResult)
```

46 plotHapTable2

plotHapTable2

plotHapTable2

#### **Description**

plot the hapResult in table like style using grid system. This function is under development and may not stable. Some parameters may deleted or renamed in future.

## Usage

```
plotHapTable2(
  hapSummary,
  show_indel_size = 1,
  replaceMultiAllele = TRUE,
  angle = 0,
  show_INFO = FALSE,
  INFO_split = c(";", ",", "\setminus |"),
  INFO_tag = "ANN",
  geneID = NA,
  tag_field = -1,
  title = "",
  gff = NULL,
  show_chr_name = TRUE,
  Chr = NULL,
  start = NULL,
  end = NULL,
 model_rect_col = "black",
 model_rect_fill = "grey80",
 model_line_col = "black",
 model_anno_txt = NULL,
 model_anno_col = "black",
 model\_anno\_cex = 1,
  table_txt_col = "black",
  table_txt_cex = 1,
 model\_anno\_pos = c(-1, -1),
 model\_anno\_adj = c(0, 1),
  gene_model_height = 0.2,
  space_height = 0.1,
  table_height = NULL,
  CDS_height = 0.3,
  link_line_type = 3,
  headrows = 1,
  equal_col_width = FALSE,
  head\_anno = 1,
  col_annots = 0,
  row_labels = 1,
  row_annots = 1,
```

plotHapTable2 47

table\_line\_col = "white",

```
annot_for_each_transcrips = TRUE,
      labels_fill = "white",
      annot_fill = "grey90",
      head_fill = NULL,
      cell_fill = NULL,
      style = gpar(fontfamily = "sans", fontface = 1, cex = 0.7),
      footbar = "",
    )
Arguments
    hapSummary
                      the hapSummary or hapResult object
    show_indel_size
                      the Indel length longer will be replaced by "i1,i2,i3,..."
    replaceMultiAllele
                      replace multi-allele title by 'T1,T2,...' or not
    angle
                      the angle of number positions
    show_INFO
                      show annotation field or not, default as FALSE
    INFO_split, INFO_tag, geneID, tag_field
                      used to set annotation in haplotype table. And the geneID was used to fileter
                      annotation in INFO field.
    title
                      title of plot
    gff
                      gff or bed annotation
    show_chr_name
                      show chromosome name at left-top cell or not
                      which range should be plotted in gene model
    Chr, start, end
    model_rect_col, model_rect_fill, model_line_col
                      a string specified the color for line/rectangle in gene model
    table_txt_col, table_txt_cex
                      controls the color and size of texts in genotype table
    model_anno_pos,
                       model_anno_adj,
                                            model_anno_cex,
                                                                model_anno_col,
    model_anno_txt
                      the position (x,y), just (hjust, vjust), color, size and content of annotation text in
                      gene model
    gene_model_height, table_height, space_height
                      the plotting range height of gene model, table and spacer
                      a numeric vector specified the height of CDS, and the height of utr is half of
    CDS_height
                      that, only useful when gff is provided,
    link_line_type the type of link lines for mutations in gene model and genotype table
    equal_col_width
                      a bool or numeric vector specified whether column with should be equal
    col_annots, head_anno, headrows, row_annots, row_labels
```

the column or row number of annotation or labels or heads

48 seqs2hap

## **Examples**

```
#
data(geneHapR_test)
plotHapTable2(hapResult)
plotHapTable2(hapResult, gff = gff)
```

seqs2hap

Generate Hap Results from Seqs

## **Description**

generate hapResults from aligned and trimed sequences

# Usage

SetATGas0 49

#### **Arguments**

seqs object of DNAStringSet or DNAMultipleAlignment class

Ref the name of reference sequences. Default as the name of the first sequence hetero\_remove whether remove accessions contains hybrid site or not. Default as TRUE

na\_drop whether drop sequeces contain "N" Default as TRUE.

maxGapsPerSeq value in [0, 1] that indicates the maximum fraction of gaps allowed in each

seq after alignment (default as 0.25). Seqs with gap percent exceed that will be

dropped

hapPrefix prefix of hap names. Default as "H"

The number length in haplotype names should be extend to.

chrName the Name should be used for haplotype

... Parameters not used.

minFlankFraction

A value in [0, 1] that indicates the minimum fraction needed to call a gap in

the consensus string (default as 0.1).

#### Value

object of hapResult class

#### **Examples**

SetATGas0

Set Position of ATG as Zero

#### Description

Set position of ATG as zero in hap result and gff annotation. The upstream was negative while the gene range and downstream was positive.

50 SetATGas0

#### Usage

#### **Arguments**

gff gene annotations

hap object of hapResult or hapSummary class

geneID geneID

Chr Chromsome name

POS vector consist with start and end position

#### **Details**

Filter hap result and gff annotation according to provided information. And then set position of ATG as zero in hap result and gff annotation. The upstream was negative while the gene range and downstream was positive.

**Notice:** the position of "ATG" after modified was 0, 1 and 2 separately. The site in hap result exceed the selected range will be **dropped**.

#### Value

```
gffSetATGas0: filtered gff with position of ATG was as zero hapSetATGas0: hap results with position of ATG was set as zero
```

#### See Also

```
displayVarOnGeneModel()
```

siteEFF 51

```
# set position of ATG as zero in hap results newhapResult <- hapSetATGas0(gff = gff, hap = hapResult, geneID = "test1G0387", Chr = "scaffold_1", POS = c(4300, 7910))
```

siteEFF

Calculation of Sites Effective

## **Description**

Calculation of Sites Effective

#### Usage

#### **Arguments**

| hap        | object of "hapResult" class  |
|------------|--|
| pheno      | phenotype data, with column names as pheno name and row name as accessions.  |
| phenoNames | pheno names used for analysis, if missing, will use all pheno names in pheno   |
| quality    | bool type, indicate whther the type of phenos are quality or quantitative. Length of quality could be 1 or equal with length of phenoNames. Default as FALSE   |
| method     | character or character vector with length equal with phenoNames indicate which method should be performed towards each phenotype. Should be one of "t.test", "chi.test", "wilcox.test" and "auto". Default as "auto", see details. |
| p.adj      | character, indicate correction method. Could be "BH", "BY", "none"   |

#### **Details**

The site **EFF** was determinate by the phenotype difference between each site geno-type.

The *p* was calculated with statistical analysis method as designated by the parameter method. If method set as "auto", then chi.test will be selected for quantity phenotype, eg.: color; for quantity phynotype, eg.: height, with at least 30 observations per geno-type and fit Gaussian distribution t.test will be performed, otherwise wilcox.test will be performed.

#### Value

a list containing two matrix names as "p" and "EFF", with column name are pheno names and row name are site position. The matrix names as "p" contains all *p*-value. The matrix named as "EFF" contains scaled difference between each geno-types per site.

52 sites\_compar

## **Examples**

```
data("geneHapR_test")

# calculate site functional effect
# siteEFF <- siteEFF(hapResult, pheno, names(pheno))
# plotEFF(siteEFF, gff = gff, Chr = "scaffold_1")</pre>
```

sites\_compar

sites comparison

# Description

Used for all allele effect compare once

# Usage

```
compareAllSites(
  hap,
  pheno,
  phenoName = names(pheno)[1],
  hetero_remove = TRUE,
  title = "",
  file = file
)
```

# Arguments

hap object of hapResult class

pheno a data.frame contains phenotypes

phenoName the name of used phenotype

 $\mbox{hetero\_remove} \quad \mbox{removing the heter-sites or not, default as TRUE}$ 

title the title of the figure

file if provieds a file path the comparing results will saved to file.

table2hap 53

## **Description**

convert variants stored in table format into hapResult

# Usage

```
table2hap(x, hapPrefix = "H", pad = 3, hetero_remove = TRUE, na_drop = TRUE)
```

#### Arguments

x a data.frame contains variants information. The first file column are fix as Chrome name, position, reference nuclicotide, alter nuclicotide and INFO. Accession genotype should be in followed columns. "-" will be treated as Indel. "." and "N" will be treated as missing data. Heterozygotes should be "A/T",

"AAA/A"

hapPrefix prefix of haplotype names

pad The number length in haplotype names should be extend to.

hetero\_remove whether remove accessions contains hyb-sites, Character not A T C G

na\_drop whether drop accessions contains missing data ("N", "NA", ".")

## Value

object of hapSummary class

# **Examples**

vcf2hap Generat Haps from VCF

# Description

Generate hapResult from vcfR object A simple filter by position was provided in this function, however it's prefer to filter VCF (vcfR object) through filter\_vcf().

54 vcf2hap

#### Usage

```
vcf2hap(
  vcf,
  hapPrefix = "H",
  filter_Chr = FALSE,
  filter_POS = FALSE,
  pad = 3,
  hetero_remove = TRUE,
  na_drop = TRUE,
  ...
)
```

## **Arguments**

vcf vcfR object imported by import\_vcf()
hapPrefix prefix of hap names, default as "H"
filter\_Chr not used
filter\_POS not used
pad The number length in haplotype names should be extend to.

hetero\_remove whether remove accessions contains hybrid site or not. Default as TRUE

na\_drop whether remove accessions contains unknown allele site or not Default as TRUE.

... Parameters not used

# Value

object of hapResult class

#### Author(s)

Zhangrenl

#### See Also

```
extract genotype from vcf: vcfR::extract_gt_tidy(), import vcf files: import_vcf() (preferred) and vcfR::read.vcfR(), filter vcf according position and annotations: filter_vcf()
```

```
data("geneHapR_test")
hapResult <- vcf2hap(vcf)</pre>
```

write.hap 55

| write. | han   |  |
|--------|-------|--|
| write. | . nab |  |

Save Haplotype Results to Disk

#### Description

This function will write hap result into a txt file.

## Usage

```
write.hap(x, file = file, sep = "\t^{"}, pheno = pheno, phenoName = phenoName)
```

## **Arguments**

x objec of hapResult or hapSummary class

file file path, where to save the hap result/summary

sep the field separator string. Values within each row of x are separated by this

string. Default as "\t"

pheno, phenoName

the phenotype data.frame, only used for export hapResult object.

#### **Details**

The hap result and hap summary result have common features. The common features of these two types are: First four rows contains extra information: CHR, POS, INFO and ALLELE Hap names were in the first column. The differences are: Hap summary result have a freq column while hap result not. Rows represent haplotypes in hap summary result, while rows represent accessions in hap result. In addition, the accessions of each haplotype in hap summary result were separated by ";".

#### Value

No return value

```
oriDir <- getwd()
  temp_dir <- tempdir()
  if(! dir.exists(temp_dir))
    dir.create(temp_dir)
  setwd(temp_dir)
data("geneHapR_test")
write.hap(hapResult, file = "hapResult.txt")
setwd(oriDir)</pre>
```

# **Index**

| * datasets   | gt.geno(DataSet), 5                     |
|--|---|
| DataSet, 5   |   |
|  | hap2hmp, 16                             |
| AccINFO (DataSet), 5                               | hap_summary, 26                         |
| addINFO, 3   | $hap\_summary(), 38, 43$                |
| addINFO(), 45                                      | hapDistribution, 17                     |
| addPromoter, 4                                     | hapResult (DataSet), 5                  |
| as.haplotype(ashaplotype),4                        | hapSetATGas0 (SetATGas0), 49            |
| as.matrix, 27                                      | hapVsPheno, 19, 25                      |
| ashaplotype, 4                                     | hapVsPhenoPerSite, 23                   |
| 1: (D + C +) 5                                     | hapVsPhenos, 24                         |
| china (DataSet), 5                                 | hmp2hap (hap2hmp), 16                   |
| compareAllSites(sites_compar), 52                  |   |
| county, 18   | import_AccINFO, 27                      |
| DataSet, 5   | import_bed, 28                          |
| displayVarOnGeneModel, 5                           | <pre>import_gff, 29</pre>               |
| displayVarOnGeneModel(), 50                        | <pre>import_hap, 30</pre>               |
| display var ondenehodel(), 30                      | <pre>import_MultipleAlignment, 31</pre> |
| editGrob, 37                                       | <pre>import_plink.pedmap, 31</pre>      |
|  | <pre>import_seqs, 32</pre>              |
| filter_hap, 9                                      | <pre>import_vcf, 33</pre>               |
| filter_hmp, 10                                     | <pre>import_vcf(), 54</pre>             |
| filter_plink.pedmap, 11                            |   |
| filter_table, 12                                   | LDheatmap, 34                           |
| filter_vcf, 13                                     |   |
| filter_vcf(), 53, 54                               | make.names, 27                          |
| <pre>filterLargeP.link(filterLargep.link), 6</pre> |   |
| filterLargep.link,6                                | NA_character_, 3                        |
| filterLargeVCF,8                                   | network, 37                             |
| <pre>get_hapNet (network), 37</pre>                | par(), <i>40</i>                        |
| <pre>get_hapNet(), 43</pre>                        | pheno (DataSet), 5                      |
| getGenePOS, 14                                     | plink.pedmap2hap, 38                    |
| getGeneRanges, 15                                  | plot_LDheatmap (LDheatmap), 34          |
| getHapGroup (network), 37                          | plotEFF, 39                             |
| gff (DataSet), 5                                   | plotHapNet, 41                          |
| gffSetATGas0 (SetATGas0), 49                       | plotHapNet(), $38$                      |
| ggpubr::ggviolin,20                                | plotHapTable, 44                        |
| Grid, <i>37</i>                                    | plotHapTable(), $3$                     |
| grid.edit,36                                       | plotHapTable2, 46                       |

INDEX 57

```
seqs (DataSet), 5
seqs2hap, 48
SetATGas0, 49
siteEFF, 51
sites(addINFO), 3
sites_compar, 52
state, 18
symnum, 20, 25
table2hap, 16, 53
trimSeqs(seqs2hap), 48
usa, 18
vcf (DataSet), 5
vcf2hap, 53
vcfR::extract_gt_tidy(), 54
vcfR::read.vcfR(), 33, 54
viewport, 36
world, 17
write.hap, 55
```