# Package 'ggmanh'

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Title Visualization Tool for GWAS Result
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**Description** Manhattan plot and QQ Plot are commonly used to visualize the end result of Genome Wide Association Study.

The ``ggmanh" package aims to keep the generation of these plots simple while maintaining customizability.

Main functions include manhattan\_plot, qqunif, and thinPoints.

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## Description

Manhattan plot and QQ Plot are commonly used to visualize the end result of Genome Wide Association Study. The "ggmanh" package aims to keep the generation of these plots simple while maintaining customizability. Main functions include manhattan\_plot, qqunif, and thinPoints.

## Author(s)

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#### **Description**

Contrary to the traditional manhattan plot, which plots all the points, the binned manhattan plot vertically and horizontally bins the variants into blocks. This speeds up plotting and reduces clutter in the case of high number of variants. The colors of the blocks can also be used to summarise the variants within each block and highlight certain features.

```
binned_manhattan_plot(x, ...)
## Default S3 method:
binned_manhattan_plot(x, ...)
## S3 method for class 'MPdataBinned'
binned_manhattan_plot(
  Х,
  outfn = NULL,
  signif.lwd = 1,
  bin.outline = FALSE,
  bin.outline.alpha = 0.2,
  highlight.colname = NULL,
  highlight.counts = TRUE,
  bin.palette = "viridis::plasma",
  bin.alpha = 0.9,
  palette.direction = 1,
  nonsignif.default = NULL,
  show.legend = TRUE,
  legend.title = NULL,
  background.col = c("grey90", "white"),
  background.alpha = 0.7,
  plot.title = ggplot2::waiver(),
  plot.subtitle = ggplot2::waiver(),
  plot.width = 10,
  plot.height = 5,
  plot.scale = 1,
)
## S3 method for class 'data.frame'
binned_manhattan_plot(
  х,
 bins.x = 10,
 bins.y = 100,
```

```
chr.gap.scaling = 0.4,
  signif = c(5e-08, 1e-05),
  pval.colname = "pval",
  chr.colname = "chr",
  pos.colname = "pos",
  chr.order = NULL,
  signif.col = NULL,
  preserve.position = TRUE,
  pval.log.transform = TRUE,
  summarise.expression.list = NULL,
  outfn = NULL,
  signif.lwd = 1,
  bin.outline = FALSE,
  bin.outline.alpha = 0.2,
  highlight.colname = NULL,
  highlight.counts = TRUE,
  bin.palette = "viridis::plasma",
  bin.alpha = 0.9,
  palette.direction = 1,
  nonsignif.default = NULL,
  show.legend = TRUE,
  legend.title = NULL,
  background.col = c("grey90", "white"),
  background.alpha = 0.7,
  plot.title = ggplot2::waiver(),
  plot.subtitle = ggplot2::waiver(),
  plot.width = 10,
  plot.height = 5,
 plot.scale = 1,
)
## S4 method for signature 'GRanges'
binned_manhattan_plot(
  Χ,
 bins.x = 10,
 bins.y = 100,
  chr.gap.scaling = 0.4,
  signif = c(5e-08, 1e-05),
  pval.colname = "pval",
  chr.order = NULL,
  signif.col = NULL,
  preserve.position = TRUE,
  pval.log.transform = TRUE,
  summarise.expression.list = NULL,
  outfn = NULL,
  signif.lwd = 1,
  bin.outline = FALSE,
```

```
bin.outline.alpha = 0.2,
  highlight.colname = NULL,
  highlight.counts = TRUE,
  bin.palette = "viridis::plasma",
  bin.alpha = 0.9,
  palette.direction = 1,
  nonsignif.default = NULL,
  show.legend = TRUE,
  legend.title = NULL,
  background.col = c("grey90", "white"),
  background.alpha = 0.7,
  plot.title = ggplot2::waiver(),
  plot.subtitle = ggplot2::waiver(),
  plot.width = 10,
  plot.height = 5,
  plot.scale = 1,
)
Х
```

## **Arguments**

a data.frame or any other extension of a data frame. It can also be a MPdata

object.

Ignored

outfn a character. File name to save the Manhattan Plot. If outfn is supplied (i.e.

!is.null(outfn)), then the plot is not drawn in the graphics window.

a number. Line width of the significance threshold line. signif.lwd

bin.outline a logical. Outline each bin. The bins are colored black.

bin.outline.alpha

a number. Alpha value of the bin outline.

highlight.colname

a character. If you desire to color certain points (e.g. significant variants) rather than color by chromosome, you can specify the category in this column, and provide the color mapping in highlight.col. Ignored if NULL.

highlight.counts

a logical. If logical, the bins are colored based on the number of points in each

bin.palette a character. Palette to color the bins. Only palettes supported by the package

paletteer are supported. More in details.

bin.alpha a number. Alpha value of the bins.

palette.direction

a number. Direction of the palette. 1 for increasing and -1 for decreasing.

nonsignif.default

a character. Default color for bins that are not significant.

show.legend a logical. Show legend if bins are colored based on a variable. legend.title a character. Title of the legend.

background.col a character. Color of the background panels. Set to NULL for no color.

background.alpha

a number. Alpha value of the background panels.

plot.title a character. Plot title plot.subtitle a character. Plot subtitle

plot.width a numeric. Plot width in inches. Corresponds to width argument in ggsave

function.

plot.height a numeric. Plot height in inches. Corresponds to height argument in ggsave

function.

plot.scale a numeric. Plot scale. Corresponds to scale argument in ggsave function. bins.x an integer. number of blocks to horizontally span the longest chromosome

bins.y an integer. number of blocks to vertically span the plot

chr.gap.scaling

a numeric. scaling factor for gap between chromosome if you desire to change it if x is an MPdata object, then the gap will scale relative to the gap in the object.

signif a numeric vector. Significant p-value thresholds to be drawn for manhattan plot.

At least one value should be provided. Default value is c(5e-08, 1e-5). If signif is not NULL and x is an MPdata object, signif argument overrides the value

inside MPdata.

pval.colname a character. Column name of x containing p.value.

chr. colname a character. Column name of x containing chromosome.

pos. colname a character. Column name of x containing position.

chr. order a character vector. Order of chromosomes presented in manhattan plot.

signif.col a character vector of equal length as signif. It contains colors for the lines

drawn at signif. If NULL, the smallest value is colored black while others are

grey. If x is an MPdata object, behaves similarly to signif.

preserve.position

a logical. If TRUE, the width of each chromosome reflect the number of variants and the position of each variant is correctly scaled? If FALSE, the width of each chromosome is equal and the variants are equally spaced.

pval.log.transform

a logical. If TRUE, the p-value will be transformed to -log10(p-value).

summarise.expression.list

a list of formulas to summarise data for each bin. Check details for more information.

## Details

Similar to manhattan\_plot, this function accepts summary statistics from GWAS and plots manhattan plot. The difference is that the variants are binned. The number of blocks can be controlled by bins.x and bins.y. The blocks can be colored based on a column in the data frame.

Palette for coloring the bins can be chosen from the package paletteer. Only palettes available in paletteer are supported. Furthermore, what palette you can use depends on what kind of variable you are using to fill the bins. Use discrete palette for categorical variable and continuous palette for continuous variable.

binned\_manhattan\_preprocess

Preprocess GWAS Result for Binned Manhattan Plot

## **Description**

Preprocess a result from Genome Wide Association Study before creating a binned manhattan plot. Works similar to manhattan\_data\_preprocess. Returns a MPdataBinned object. It can be created using a data.frame or a MPdata object. Go to details to read how to use summarise.expression.list.

```
binned_manhattan_preprocess(x, ...)
## Default S3 method:
binned_manhattan_preprocess(x, ...)
## S3 method for class 'MPdata'
binned_manhattan_preprocess(
  Х,
 bins.x = 10,
 bins.y = 100,
  chr.gap.scaling = 0.4,
  summarise.expression.list = NULL,
  show.message = TRUE,
)
## S3 method for class 'data.frame'
binned_manhattan_preprocess(
  х,
 bins.x = 10,
 bins.y = 100,
  chr.gap.scaling = 0.4,
  signif = c(5e-08, 1e-05),
  pval.colname = "pval",
  chr.colname = "chr",
  pos.colname = "pos",
  chr.order = NULL,
  signif.col = NULL,
  preserve.position = TRUE,
  pval.log.transform = TRUE,
```

```
summarise.expression.list = NULL,
)
## S4 method for signature 'GRanges'
binned_manhattan_preprocess(
  х,
 bins.x = 10,
 bins.y = 100,
  chr.gap.scaling = 0.4,
  signif = c(5e-08, 1e-05),
  pval.colname = "pval",
  chr.order = NULL,
  signif.col = NULL,
  preserve.position = TRUE,
  pval.log.transform = TRUE,
  summarise.expression.list = NULL,
)
```

#### **Arguments**

a data.frame or any other extension of a data frame. It can also be a MPdata Х object. Ignored . . . bins.x an integer. number of blocks to horizontally span the longest chromosome bins.v an integer. number of blocks to vertically span the plot chr.gap.scaling a number. scaling factor for the gap between chromosomes summarise.expression.list a list of formulas to summarise data for each bin. Check details for more information. a logical. Show warning if MPdata directly used. Set to FALSE to suppress show.message warning. a numeric vector. Significant p-value thresholds to be drawn for manhattan plot. signif At least one value should be provided. Default value is c(5e-08, 1e-5) pval.colname a character. Column name of x containing p.value. chr.colname a character. Column name of x containing chromosome. pos.colname a character. Column name of x containing position. chr.order a character vector. Order of chromosomes presented in manhattan plot. a character vector of equal length as signif. It contains colors for the lines signif.col drawn at signif. If NULL, the smallest value is colored black while others are grey. preserve.position

a logical. If TRUE, the width of each chromosome reflect the number of variants and the position of each variant is correctly scaled? If FALSE, the width of each chromosome is equal and the variants are equally spaced.

```
pval.log.transform a logical. If TRUE, the p-value will be transformed to -log10(p-value).
```

#### **Details**

If x is a data frame or something alike, then it creates a MPdata object first and then builds MPdataBinned S3 object.

x can also be a MPdata object. Be sure to check if thin has been applied because this can affect what's being aggregated such as number of variables in each bin.

Positions of each point relative to the plot are first calculated via manhattan\_data\_preprocess. Then the data is binned into blocks. bins.x indicates number of blocks allocated to the chromsome with the widest width. The number of blocks for other chromosomes is proportional to the widest chromosome. bins.y indicates the number of blocks allocated to the y-axis. The number may be slightly adjusted to have the block height end exactly at the significance threshold.

Since points are aggregated into bins, users have the choice to freely specify expressions to summarise the data for each bin through summarise.expression.list argument. This argument takes a list of two-sided formulas, where the left side is the name of the new column and the right side is the expression to calculate the column. This expression is then passed to summarise. For example, to calculate the mean, min, max of a column named beta in each bin, summarise.expression.list arument would be

```
# inside binned_manhattan_preprocess function
summarise.expression.list = list(
  mean_beta ~ mean(beta),
  min_beta ~ min(beta),
  max_beta ~ max(beta)
)
```

#### Value

a MPdataBinned object. This object contains necessary components for creating a binned manhattan plot.

## **Examples**

```
gwasdat <- data.frame(
  "chromosome" = rep(1:5, each = 1500),
  "position" = c(replicate(5, sample(1:15000, 30))),
  "pvalue" = rbeta(7500, 1, 1)^5,
  "beta" = rnorm(7500)
)

tmp <- binned_manhattan_preprocess(
  gwasdat, pval.colname = "pvalue", chr.colname = "chromosome",
  pos.colname = "position", chr.order = as.character(1:5),
  bins.x = 10, bins.y = 50,
  summarise.expression.list = list(
    mean_beta ~ mean(beta, na.rm = TRUE),
    max_abs_beta ~ max(abs(beta), na.rm = TRUE)
)</pre>
```

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```
)
print(tmp)
```

calc\_new\_pos

Calculate new x-position of each point

## **Description**

Calculate the actual x-positions of each point used for the manhattan plot. MPdata object contains the unscaled positions that has not been positioned according to the relative position and width of each chromosome.

## Usage

```
calc_new_pos(mpdata)
```

#### **Arguments**

mpdata

an MPdata object.

### **Details**

This is used calculate the actual positions used for the inside manhattan\_plot function. It was designed this way should the scaling and relative positioning of each chromosome be changed (e.g. gap between the )

#### Value

a numeric vector containing the scaled x-positions.

## **Examples**

```
gwasdat <- data.frame(
  "chromosome" = rep(1:5, each = 30),
  "position" = c(replicate(5, sample(1:300, 30))),
  "pvalue" = rbeta(150, 1, 1)^5
)

mpdata <- manhattan_data_preprocess(
  gwasdat, pval.colname = "pvalue", chr.colname = "chromosome", pos.colname = "position",
  chr.order = as.character(1:5)
)

calc_new_pos(mpdata)</pre>
```

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default\_gds\_path

Path to Default GDS File

## **Description**

Find path to the default gds file.

## Usage

```
default_gds_path()
```

## Value

A character vector.

## **Examples**

```
default_gds_path()
```

gds\_annotate

Annotation with GDS File

## **Description**

Retrieve variant annotation stored in a GDS file with chromosome location or rs.id.

```
gds_annotate(
    x,
    gdsfile = NULL,
    annot.method = "position",
    chr = NULL,
    pos = NULL,
    ref = NULL,
    alt = NULL,
    rs.id = NULL,
    concat_char = "/",
    verbose = TRUE,
    annotation_names = c("annotation/info/symbol", "annotation/info/consequence",
        "annotation/info/LoF")
)
```

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#### Arguments

a data. frame object to be annotated. a character for GDS filename. If NULL, the default GDS file included with the gdsfile package is used. a method for searching variants. "position" requires chr, pos, ref, and alt. annot.method "rs.id" requires rs.id. chr, pos, ref, alt, rs.id column names of x that contain chromosome, position, reference allele, alternate allele, and rs.id, respectively. concat\_char a character used to separate multiple annotations returned from the gds file. verbose output messages. annotation\_names a character vector of nodes of the gdsfile that are to be extracted.

#### Value

A character vector the length of nrow(x) if  $concat\_char$  is a character. A data frame with nrow(x) rows and  $length(annotation\_names)$  if  $concat\_char$  is null.

#### **Examples**

```
vardata <- data.frame(
  chr = c(11,20,14),
  pos = c(12261002, 10033792, 23875025),
  ref = c("G", "G", "CG"),
  alt = c("A", "A", "C")
)
annotations <- gds_annotate(
  x = vardata, annot.method = "position",
  chr = "chr", pos = "pos", ref = "ref", alt = "alt"
)
print(annotations)</pre>
```

ggmanh

ggmanh: A package for visualization of GWAS results.

#### **Description**

ggmanh provides flexible tools for visualizing GWAS result for downstream analysis.

#### **Details**

Manhattan plot is commonly used to display significant Single Nucleotide Polymorphisms (SNPs) in Genome Wide Association Study (GWAS) This package comes with features useful for manhattan plot creation, including annotation with <code>ggrepel</code>, truncating data for faster plot generation, and manual rescaling of the y-axis. The manhattan plot is generated in two steps: data preprocessing and plotting. This allows the user to iteratively customize the plot without having the process the GWAS summary data over and over again. Currently, data.frame and GRanges from GenomicRanges are supported.

A vignette detailing the usage of the package is accessible by vignette("ggmanh")

#### Author(s)

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Authors:

• John Lee < john.lee@abbvie.com> (AbbVie)

Other contributors:

• Xiuwen Zheng <xiuwen.zheng@abbvie.com> [contributor, data contributor]

ggmanh\_annotation\_gds gnomAD Variant Annotation in SeqArray Format

#### **Description**

ggmanh provides a GDS file whose path is accessible by default\_gds\_path. The original annotation file is from the gnomAD browser v2.1.1 release, available in this link: https://gnomad.broadinstitute.org/downloads. This gds file contains variants in the exome with the global minor allele frequency  $\geq 0.0002$ , and has been manually curated to fit the file size requirement for R Bioconductor packages.

## Format

A GDS file with 1015430 variants with chromosome, position, allele, gene symbol, Ensembl VEP Consequence, and predicted LoF.

manhattan\_data\_preprocess

Preprocess GWAS Result

#### **Description**

Preprocesses a result from Genome Wide Association Study before making a manhattan plot. It accepts a data.frame, which at bare minimum should contain a chromosome, position, and p-value. Additional options, such as chromosome color, label column names, and colors for specific variants, are provided here.

```
manhattan_data_preprocess(x, ...)
## Default S3 method:
manhattan_data_preprocess(x, ...)
## S3 method for class 'data.frame'
manhattan_data_preprocess(
  Х,
  chromosome = NULL,
  signif = c(5e-08, 1e-05),
  pval.colname = "pval",
  chr.colname = "chr",
  pos.colname = "pos",
  highlight.colname = NULL,
  chr.order = NULL,
  signif.col = NULL,
  chr.col = NULL,
  highlight.col = NULL,
  preserve.position = FALSE,
  thin = NULL,
  thin.n = 1000,
  thin.bins = 200,
  pval.log.transform = TRUE,
  chr.gap.scaling = 1,
)
## S4 method for signature 'GRanges'
manhattan_data_preprocess(
  chromosome = NULL,
  signif = c(5e-08, 1e-05),
  pval.colname = "pval",
  highlight.colname = NULL,
```

```
chr.order = NULL,
  signif.col = NULL,
  chr.col = NULL,
  highlight.col = NULL,
  preserve.position = FALSE,
  thin = NULL,
  thin.n = 100,
  thin.bins = 200,
  pval.log.transform = TRUE,
  chr.gap.scaling = 1,
  ...
)
```

#### **Arguments**

x a data frame or any other extension of data frame (e.g. a tibble). At bare mini-

mum, it should contain chromosome, position, and p-value.

. . . Additional arguments for manhattan\_data\_preprocess.

chromosome a character. This is supplied if a manhattan plot of a single chromosome is

desired. If NULL, then all the chromosomes in the data will be plotted.

signif a numeric vector. Significant p-value thresholds to be drawn for manhattan plot.

At least one value should be provided. Default value is c(5e-08, 1e-5)

pval.colname a character. Column name of x containing p.value.

chr.colname a character. Column name of x containing chromosome.

pos.colname a character. Column name of x containing position.

highlight.colname

a character. If you desire to color certain points (e.g. significant variants) rather than color by chromosome, you can specify the category in this column, and

provide the color mapping in highlight.col. Ignored if NULL.

chr.order a character vector. Order of chromosomes presented in manhattan plot.

signif.col a character vector of equal length as signif. It contains colors for the lines

drawn at signif. If NULL, the smallest value is colored black while others are

grey.

chr.col a character vector of equal length as chr.order. It contains colors for the chro-

mosomes. Name of the vector should match chr. order. If NULL, default colors

are applied using RColorBrewer.

highlight.col a character vector. It contains color mapping for the values from highlight.colname.

preserve.position

a logical. If TRUE, the width of each chromosome reflect the number of variants and the position of each variant is correctly scaled? If FALSE, the width of each

chromosome is equal and the variants are equally spaced.

thin a logical. If TRUE, thinPoints will be applied. Defaults to TRUE if chromosome

is NULL. Defaults to FALSE if chromosome is supplied.

thin.n an integer. Number of max points per horizontal partitions of the plot. Defaults

to 1000.

#### **Details**

manhattan\_data\_preprocess gathers information needed to plot a manhattan plot and organizes the information as MPdata S3 object.

New positions for each points are calculated, and stored in the data.frame as "new\_pos". By default, all chromosomes will have the same width, with each point being equally spaced. This behavior is changed when preserve.position = TRUE. The width of each chromosome will scale to the number of points and the points will reflect the original positions.

chr.col and highlight.col, maps the data values to colors. If they are an unnamed vector, then the function will try its best to match the values of chr.colname or highlight.colname to the colors. If they are a named vector, then they are expected to map all values to a color. If highlight.colname is supplied, then chr.col is ignored.

While feeding a data.frame directly into manhattan\_plot does preprocessing & plotting in one step. If you plan on making multiple plots with different graphic options, you have the choice to preprocess separately and then generate plots.

#### Value

a MPdata object. This object contains all the necessary components for constructing a manhattan plot.

#### **Examples**

```
gwasdat <- data.frame(
  "chromosome" = rep(1:5, each = 30),
  "position" = c(replicate(5, sample(1:300, 30))),
  "pvalue" = rbeta(150, 1, 1)^5
)

manhattan_data_preprocess(
  gwasdat, pval.colname = "pvalue", chr.colname = "chromosome", pos.colname = "position",
  chr.order = as.character(1:5)
)</pre>
```

manhattan\_plot

Manhattan Plotting

#### **Description**

A generic function for manhattan plot.

```
manhattan_plot(x, ...)
manhattan_plot.default(x, ...)
## S3 method for class 'data.frame'
manhattan_plot(
  Х,
  chromosome = NULL,
  outfn = NULL,
  signif = c(5e-08, 1e-05),
  pval.colname = "pval",
  chr.colname = "chr",
  pos.colname = "pos",
  label.colname = NULL,
  highlight.colname = NULL,
  chr.order = NULL,
  signif.col = NULL,
  chr.col = NULL,
  highlight.col = NULL,
  rescale = TRUE,
  rescale.ratio.threshold = 5,
  signif.rel.pos = 0.2,
  chr.gap.scaling = 1,
  color.by.highlight = FALSE,
  preserve.position = FALSE,
  thin = NULL,
  thin.n = 1000,
  thin.bins = 200,
  pval.log.transform = TRUE,
  plot.title = ggplot2::waiver(),
  plot.subtitle = ggplot2::waiver(),
  plot.width = 10,
  plot.height = 5,
  plot.scale = 1,
  point.size = 0.75,
  label.font.size = 2,
  max.overlaps = 20,
  x.label = "Chromosome",
```

```
y.label = expression(-log[10](p)),
)
## S3 method for class 'MPdata'
manhattan_plot(
 х,
  chromosome = NULL,
 outfn = NULL,
  signif = NULL,
  signif.col = NULL,
  rescale = TRUE,
  rescale.ratio.threshold = 5,
  signif.rel.pos = 0.2,
  chr.gap.scaling = NULL,
  color.by.highlight = FALSE,
  label.colname = NULL,
  x.label = "Chromosome",
 y.label = expression(-log[10](p)),
  point.size = 0.75,
  label.font.size = 2,
 max.overlaps = 20,
 plot.title = ggplot2::waiver(),
  plot.subtitle = ggplot2::waiver(),
 plot.width = 10,
 plot.height = 5,
 plot.scale = 1,
)
## S4 method for signature 'GRanges'
manhattan_plot(
  Х,
  chromosome = NULL,
  outfn = NULL,
  signif = c(5e-08, 1e-05),
  pval.colname = "pval",
  label.colname = NULL,
  highlight.colname = NULL,
  chr.order = NULL,
  signif.col = NULL,
  chr.col = NULL,
  highlight.col = NULL,
  rescale = TRUE,
  rescale.ratio.threshold = 5,
  signif.rel.pos = 0.2,
  chr.gap.scaling = 1,
  color.by.highlight = FALSE,
```

```
preserve.position = FALSE,
  thin = NULL,
  thin.n = 1000,
  thin.bins = 200,
  pval.log.transform = TRUE,
  plot.title = ggplot2::waiver(),
  plot.subtitle = ggplot2::waiver(),
  plot.width = 10,
  plot.height = 5,
  plot.scale = 1,
  point.size = 0.75.
  label.font.size = 2,
 max.overlaps = 20,
  x.label = "Chromosome",
 y.label = expression(-log[10](p)),
)
```

#### Arguments

x a data.frame, an extension of data.frame object (e.g. tibble), or an MPdata

object.

... additional arguments to be passed onto geom\_label\_repel

chromosome a character. This is supplied if a manhattan plot of a single chromosome is

desired. If NULL, then all the chromosomes in the data will be plotted.

outfn a character. File name to save the Manhattan Plot. If outfn is supplied (i.e.

!is.null(outfn)), then the plot is not drawn in the graphics window.

signif a numeric vector. Significant p-value thresholds to be drawn for manhattan plot.

At least one value should be provided. Default value is c(5e-08, 1e-5). If signif is not NULL and x is an MPdata object, signif argument overrides the value

inside MPdata.

pval.colname a character. Column name of x containing p.value.

chr. colname a character. Column name of x containing chromosome.

pos.colname a character. Column name of x containing position.

label.colname a character. Name of the column in MPdata\$data to be used for labeling.

highlight.colname

a character. If you desire to color certain points (e.g. significant variants) rather than color by chromosome, you can specify the category in this column, and

provide the color mapping in highlight.col. Ignored if NULL.

chr. order a character vector. Order of chromosomes presented in manhattan plot.

signif.col a character vector of equal length as signif. It contains colors for the lines

drawn at signif. If NULL, the smallest value is colored black while others are

grey. If x is an MPdata object, behaves similarly to signif.

chr.col a character vector of equal length as chr.order. It contains colors for the chro-

mosomes. Name of the vector should match chr.order. If NULL, default colors

are applied using RColorBrewer.

highlight.col a character vector. It contains color mapping for the values from highlight.colname.

rescale a logical. If TRUE, the plot will rescale itself depending on the data. More on

this in details.

rescale.ratio.threshold

a numeric. Threshold of that triggers the rescale.

signif.rel.pos a numeric between 0.1 and 0.9. If the plot is rescaled, where should the signifi-

cance threshold be positioned?

chr.gap.scaling

a numeric. scaling factor for gap between chromosome if you desire to change it if x is an MPdata object, then the gap will scale relative to the gap in the object.

color.by.highlight

a logical. Should the points be colored based on a highlight column?

preserve.position

a logical. If TRUE, the width of each chromosome reflect the number of variants and the position of each variant is correctly scaled? If FALSE, the width of each chromosome is equal and the variants are equally spaced.

thin a logical. If TRUE, thinPoints will be applied. Defaults to TRUE if chromosome

is NULL. Defaults to FALSE if chromosome is supplied.

thin.n an integer. Number of max points per horizontal partitions of the plot. Defaults

to 1000.

thin.bins an integer. Number of bins to partition the data. Defaults to 200.

pval.log.transform

a logical. If TRUE, the p-value will be transformed to -log10(p-value).

plot.title a character. Plot title plot.subtitle a character. Plot subtitle

plot.width a numeric. Plot width in inches. Corresponds to width argument in ggsave

function.

plot.height a numeric. Plot height in inches. Corresponds to height argument in ggsave

function.

plot.scale a numeric. Plot scale. Corresponds to scale argument in ggsave function.

point.size a numeric. Size of the points.

label.font.size

a numeric. Size of the labels.

max.overlaps an integer. Exclude text labels that overlaps too many things.

x.label a character. x-axis labely.label a character. y-axis label

#### **Details**

This generic function accepts a result of a GWAS in the form of data.frame or a MPdata object produced by manhattan\_data\_preprocess. The function will throw an error if another type of object is passed.

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Having rescale = TRUE is useful when there are points with very high -log10(p.value). In this case, the function attempts to split the plot into two different scales, with the split happening near the strictest significance threshold. More precisely, the plot is rescaled when

```
-log10(pvalue)/(strictest significance threshold) \ge rescale.ratio.threshold
```

If you wish to add annotation to the points, provide the name of the column to label.colname. The labels are added with ggrepel.

Be careful though: if the annotation column contains a large number of variants, then the plotting could take a long time, and the labels will clutter up the plot. For those points with no annotation, you have the choice to set them as NA or "".

#### Value

```
gg object if is.null(outfn), NULL if !is.null(outf)
```

#### **Examples**

```
gwasdat <- data.frame(
    "chromosome" = rep(1:5, each = 30),
    "position" = c(replicate(5, sample(1:300, 30))),
    "pvalue" = rbeta(150, 1, 1)^5
)

manhattan_plot(
    gwasdat, pval.colname = "pvalue", chr.colname = "chromosome", pos.colname = "position",
    chr.order = as.character(1:5)
)

mpdata <- manhattan_data_preprocess(
    gwasdat, pval.colname = "pvalue", chr.colname = "chromosome", pos.colname = "position",
    chr.order = as.character(1:5)
)

manhattan_plot(mpdata)</pre>
```

qqunif

Plot Quantile-Quantile Plot of p-values against uniform distribution.

## Description

Plot Quantile-Quantile Plot of p-values against uniform distribution.

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#### Usage

```
qqunif(
   x,
   outfn = NULL,
   conf.int = 0.95,
   plot.width = 5,
   plot.height = 5,
   thin = TRUE,
   thin.n = 500,
   zero.pval = "replace"
)
```

#### **Arguments**

x a numeric vector of p-values. All values should be between 0 and 1.

outfn a character. File name to save the QQ Plot. If outfn is supplied (i.e. !is.null(outfn)),

then the plot is not drawn in the graphics window.

conf.int a numeric between 0 and 1. Confidence band to draw around reference line. Set

to NA to leave it out.

plot.width a numeric. Plot width in inches. plot.height a numeric. Plot height in inches.

thin a logical. Reduce number of data points when they are cluttered?

thin.n an integer. Number of max points per horizontal partitions of the plot. Defaults

to 500.

zero.pval a character. Determine how to treat 0 pvals. "replace" will replace the p-value

of zero with the non-zero minimum. "remove" will remove the p-value of zero.

#### Value

a ggplot object

## **Examples**

```
x <- rbeta(1000, 1, 1)
qqunif(x)</pre>
```

thinPoints

Thin Data Points

## **Description**

Reduce the number of cluttered data points.

```
thinPoints(dat, value, n = 1000, nbins = 200, groupBy = NULL)
```

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## Arguments

dat	a data frame
value	column name of dat to be used for partitioning (see details)
n	number of points to sample for each partition
nbins	number of partitions
groupBy	column name of dat to group by before partitioning (e.g. chromosome)

#### **Details**

The result of Genome Wide Association Study can be very large, with the majority of points being being clustered below significance threshold. This unnecessarily increases the time to plot while making almost no difference. This function reduces the number of points by partitioning the points by a numeric column value into nbins and sampling n points.

#### Value

```
a data.frame
```

## **Examples**

```
dat <- data.frame(
   A1 = c(1:20, 20, 20),
   A2 = c(rep(1, 12), rep(1,5), rep(20, 3), 20, 20),
   B = rep(c("a", "b", "c", "d"), times = c(5, 7, 8, 2))
)
# partition "A1" into 2 bins and then sample 6 data points
thinPoints(dat, value = "A1", n = 6, nbins = 2)
# partition "A2" into 2 bins and then sample 6 data points
thinPoints(dat, value = "A2", n = 6, nbins = 2)
# group by "B", partition "A2" into 2 bins and then sample 3 data points
thinPoints(dat, value = "A2", n = 3, nbins = 2, groupBy = "B")</pre>
```

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