Package ‘topr’

October 14, 2022

Title  Create Custom Plots for Viewing Genetic Association Results
Version  1.0.0

URL  https://github.com/GenuityScience/topr

BugReports  https://github.com/GenuityScience/topr/issues

Description  A collection of functions for visualizing, exploring and annotating genetic association results. Association results from multiple traits can be viewed simultaneously along with gene annotation, over the entire genome (Manhattan plot) or in the more detailed regional view.

License  LGPL (>= 3)

Encoding  UTF-8

LazyData  true

LazyDataCompression  xz

RoxygenNote  7.1.2

Suggests  testthat (>= 3.0.0), knitr, rmarkdown, markdown

Config/testthat/edition  3

Imports  ggplot2 (>= 3.3.2), dplyr (>= 1.0.2), stringr (>= 1.4.0), readr (>= 1.4.0), scales (>= 1.1.1), ggrepel, egg, grid, gridExtra, magrittr (>= 1.5), utils, toprdata

Depends  R (>= 3.5.0)

NeedsCompilation  no

Author  Genuity Science [cph],
  Thorhildur Juliusdottir [aut, cre],
  Andri Stefansson [aut]

Maintainer  Thorhildur Juliusdottir <thorhildur@genuitysci.com>

Repository  CRAN

Date/Publication  2022-01-12 11:00:02 UTC
**annotate_with_nearest_gene**

*Get the nearest gene for one or more snps*

**Description**

annotate_with_nearest_gene() Annotate the variant/snp with their nearest gene. Required parameters is a dataframe of SNPs (with the columns CHROM and POS)

**Usage**

annotate_with_nearest_gene(variants, protein_coding_only = FALSE)

**Arguments**

variants a dataframe of variant positions (CHROM and POS)

protein_coding_only Logical, if set to TRUE only annotate with protein coding genes (the default value is FALSE)
Value

the input dataframe with Gene_Symbol as an additional column

Examples

variants <-get_best_snp_per_MB(CD_UKBB)
annotate_with_nearest_gene(variants)

<table>
<thead>
<tr>
<th>CD_FINNGEN</th>
<th>Finngen v5 Crohn's disease (CHRONSMALL)</th>
</tr>
</thead>
</table>

Description

Dataset retrieved from the Finngen database (version 5) including 968 crohn’s cases and 210,100 controls. The dataset has been filtered on variants with P <1e-03.

Usage

CD_FINNGEN

Format

A data frame with 29,926 rows and 9 variables:

- **CHROM** Chromosome, written as for example chr1 or 1
- **POS** genetic position of the variant
- **ID** Variant identifier, e.g. rsid
- **P** P-value from Plink run, additive model, regression model GLM_FIRTH
- **beta** Variant effect

Source

Crohn’s small intestines (CHRONSMALL), only including variants with P<1e-03
CD_UKBB

**UKBB Crohns disease (ICD 10 code K50)**

### Description

Dataset retrieved from the UK biobank consisting of 2,799 crohn’s cases (K50) and 484,515 controls. The dataset has been filtered on variants with P <1e-03.

### Usage

CD_UKBB

### Format

A data frame with 26,824 rows and 10 variables:

- **CHROM** Chromosome, written as for example chr1 or 1
- **POS** genetic position of the variant
- **ID** Variant identifier, e.g. rsid
- **P** P-value from Plink run, additive model, regression model GLM_FIRTH
- **OR** Odds Ratio

### Source

Crohn’s UKBB ICD10 code K50, only including variants with P<1e-03

---

**create_snpset**

Create a dataframe that can be used as input for making effect plots

### Description

create_snpset()

### Usage

```
create_snpset(  
    df1,  
    df2,  
    thresh = 1e-08,  
    protein_coding_only = TRUE,  
    region_size = 1e+06,  
    verbose = FALSE
  )
```
create_snpset_code

**Arguments**

- `df1` The dataframe to extract the top snps from (with p-value below thresh)
- `df2` The dataframe in which to search for overlapping SNPs from dataframe1
- `thresh` Numeric, the p-value threshold used for extracting the top snps from dataset 1
- `protein_coding_only` Logical, set this variable to TRUE to only use protein_coding genes for the annotation
- `region_size` Integer, the size of the interval which to extract the top snps from
- `verbose` Logical, (default: FALSE). Assign to TRUE to get information on which alleles are matched and which are not.

**Value**

Dataframe containing the top hit

**Examples**

```r
CD_UKBB_index_snps <- get_best_snp_per_MB(CD_UKBB)
create_snpset(CD_UKBB_index_snps, CD_FINNGEN)
```

---

create_snpset_code  
*Show the code/functions used to create a snpset*

---

**Description**

create_snpset_code()

**Usage**

create_snpset_code()

**Value**

Dataframe containing the top hit

**Examples**

create_snpset_code()
dat_column_check_and_set

dat_column_check_and_set

Description

This function is used to standardize the column names in the input dataframe

Usage

dat_column_check_and_set(dat)

Arguments

dat A data frame or a list of data frames

---

effect_plot

Create a plot comparing effects within two datasets

description

effect_plot()

Usage

effect_plot(
    dat,
    pheno_x = "x_pheno",
    pheno_y = "y_pheno",
    annotate_with = "Gene_Symbol",
    thresh = 1e-08,
    ci_thresh = 1,
    gene_label_thresh = 1e-08,
    color = get_topr_colors()[[1]],
    scale = 1
)

Arguments

dat The input dataframe (snpset) containing one row per variant and P values (P1 and P2) and effects (E1 and E2) from two datasets/phenotypes

pheno_x A string representing the name of the phenotype whose effect is plotted on the x axis

pheno_y A string representing the name of the phenotype whose effect is plotted on the y axis
annotate_with A string. The name of the column that contains the label for the datapoints (default value is Gene_Symbol)

thresh A number. Threshold cutoff, datapoints with P2 below this threshold are shown as filled circles whereas datapoints with P2 above this threshold are shown as open circles

ci_thresh A number, Show the confidence intervals if the P-value is below this threshold

gene_label_thresh A string, label datapoints with P2 below this threshold

color A string, default value is the first of the top colors

scale A number, to change the size of the title and axes labels and ticks at the same time (default = 1)

Value

ggplot object

Examples

CD_UKBB_index_snps <- get_best_snp_per_MB(CD_UKBB)
snpset <- create_snpset(CD_UKBB_index_snps, CD_FINNGEN)
effect_plot(snpset)

flip_to_positive_allele_for_dat1

Flip to the positive allele for dataset 1

Description

flip_to_positive_allele_for_dat1()

Usage

flip_to_positive_allele_for_dat1(df)

Arguments

df A dataframe that is in the snpset format (like returned by the get_snpset() function)

Value

The input dataframe after flipping to the positive effect allele in dataframe 1
get_best_snp_per_MB

Examples

CD_UKBB_index_snps <- get_best_snp_per_MB(CD_UKBB)
snpset <- create_snpset(CD_UKBB_index_snps, CD_FINNGEN)
flip_to_positive_allele_for_dat1(snpset)

get_best_snp_per_MB Get the index/lead variants

Description

get_best_snp_per_MB() Get the top variants within 1 MB windows of the genome with association p-values below the given threshold

Usage

get_best_snp_per_MB(
  df,
  thresh = 1e-09,
  region_size = 1e+06,
  protein_coding_only = FALSE,
  chr = NULL,
  .checked = FALSE,
  verbose = FALSE
)

Arguments

df Dataframe
thresh A number. P-value threshold, only extract variants with p-values below this threshold (1e-09 by default)
region_size An integer (default = 1000000) indicating the window size for variant labelling. Increase this number for sparser annotation and decrease for denser annotation.
protein_coding_only Logical, set this variable to TRUE to only use protein_coding genes for annotation
chr String, get the top variants from one chromosome only, e.g. chr="chr1"
.checked Logical, if the input data has already been checked, this can be set to TRUE so it wont be checked again (FALSE by default)
verbose Logical, set to TRUE to get printed information on number of SNPs extracted

Value

Dataframe of lead variants. Returns the best variant per MB (by default, change the region size with the region argument) with p-values below the input threshold (thresh=1e-09 by default)
get_gene

Examples
    get_best_snp_per_MB(CD_UKBB)

---

get_gene

Get the genetic position of a gene by gene name

Description
    get_gene() Get the gene coordinates for a gene Required parameter is gene name

Usage
    get_gene(gene_name, chr = NULL)

Arguments
    gene_name  A string representing a gene name (e.g. "FTO")
    chr       A string, search for the genes on this chromosome only, (e.g chr="chr1")

Value
    Dataframe of genes

Examples
    get_gene("FTO")

---

get_genes_by_Gene_Symbol

Get the genetic position of a gene by its gene name

Description
    get_genes_by_Gene_Symbol() Get genes by their gene symbol/name Required parameters is on
gene name or a vector of gene names

Usage
    get_genes_by_Gene_Symbol(genes, chr = NULL)

Arguments
    genes  A string or vector of strings representing gene names, (e.g. "FTO") or (c("FTO","NOD2"))
    chr    A string, search for the genes on this chromosome only, (e.g chr="chr1")
Value

Dataframe of genes

Examples

genes_by_Gene_Symbol(c("FTO","THADA"))

df1 <- get_best.snp.per.MB(CD_UKBB)
df2 <- CD_FINNGEN
get_overlapping_snps_by_pos(df1, df2, verbose = TRUE)
get_snps_within_region

Get SNPs/variants within region

Description

get_snps_within_region()

Usage

get_snps_within_region(df, region, chr = NULL, xmin = NULL, xmax = NULL)

Arguments

df  data frame of association results with the columns CHR and POS
region A string representing the genetic region (e.g chr16:50693587-50734041)
chr A string, chromosome (e.g. chr16)
xmin An integer, include variants with POS larger than xmin
xmax An integer, include variants with POS smaller than xmax

Value

the variants within the requested region

Examples

get_snps_within_region(CD_UKBB, "chr16:50593587-50834041")

get_topr_colors

Get the top hit from the dataframe

Description

get_topr_colors() Get the top hit from the dataframe All other input parameters are optional

Usage

get_topr_colors()

Value

Vector of colors used for plotting

Examples

get_topr_colors()
get_top_snp

*Get the top hit from the dataframe*

**Description**

get_top_snp() Get the top hit from the dataframe All other input parameters are optional

**Usage**

get_top_snp(df, chr = NULL)

**Arguments**

- **df**: Dataframe containing association results
- **chr**: String, get the top hit in the data frame for this chromosome. If chromosome is not provided, the top hit from the entire dataset is returned.

**Value**

Dataframe containing the top hit

**Examples**

get_top_snp(CD_UKBB, chr="chr1")

locuszoom

*Create a locuszoom-like plot*

**Description**

locuszoom() displays the association results for a smaller region within one chromosome. Required parameter is at least one dataset (dataframe) containing the association data (with columns CHROM, POS, P in upper or lowercase)

**Usage**

locuszoom(
    df,
    annotate = NULL,
    ntop = 3,
    xmin = 0,
    size = 2,
    shape = 19,
    alpha = 1,
    label_size = 4,
annotate_with = "ID",
color = NULL,
axis_text_size = 11,
axis_title_size = 12,
title_text_size = 13,
show_genes = FALSE,
show_overview = FALSE,
show_exons = FALSE,
max_genes = 200,
sign_thresh = 5e-09,
sign_thresh_color = "red",
sign_thresh_label_size = 3.5,
xmax = NULL,
ymin = NULL,
ymax = NULL,
protein_coding_only = FALSE,
region_size = 1e+06,
gene_padding = 1e+05,
angle = 0,
legend_title_size = 12,
legend_text_size = 12,
nudge_x = 0.01,
nudge_y = 0.01,
rsids = NULL,
variant = NULL,
rsids_color = "gray40",
legend_name = "Data:",
legend_position = "right",
chr = NULL,
vline = NULL,
show_gene_names = NULL,
legend_labels = NULL,
gene = NULL,
title = NULL,
label_color = "gray40",
region = NULL,
scale = 1,
rsids_with_vline = NULL,
annotate_with_vline = NULL,
sign_thresh_size = 0.5,
unit_main = 7,
unit_gene = 2
)

Arguments

| df          | Dataframe or a list of dataframes (required columns are CHROM, POS, P), in upper- or lowercase) of association results. |
annotate: A number (p-value). Display annotation for variants with p-values below this threshold.
nadapt: An integer, number of datasets (GWAS results) to show on the top plot.
xmin: Integer, setting the chromosomal range to display on the x-axis.
size: An integer setting the size of the plot points (default: size=1.2).
shape: A number of vector of numbers setting the shape of the plotted points.
label: A number or vector of numbers setting the transparency of the plotted points.
label_size: An integer to set the size of the plot labels (default: label_size=3).
annotate_with: A string. Annotate the variants with either Gene_Symbol or ID (default: "Gene_Symbol").
color: A string or a vector of strings, for setting the color of the datapoints on the plot.
axis_text_size: A number, size of the x and y axes tick labels (default: 12).
axis_title_size: A number, size of the x and y title labels (default: 12).
title_text_size: A number, size of the plot title (default: 13).
show_genes: A logical scalar, show genes instead of exons (default show_genes=FALSE).
show_overview: A logical scalar, shows/hides the overview plot (default=TRUE).
show_exons: A logical scalar, show exons instead of genes (default show_exons=FALSE).
max_genes: An integer, only label the genes if they are fewer than max_genes (default values is 200).
sign_thresh: A number or vector of numbers, setting the horizontal significance threshold (default: sign_thresh=5.1e-9). Set to NULL to hide the significance threshold.
sign_thresh_color: A string or vector of strings to set the color/s of the significance threshold/s.
sign_thresh_label_size: A number setting the text size of the label for the significance thresholds (default text size is 3.5).
xmax: Integer, setting the chromosomal range to display on the x-axis.
ymin: Integer, min and max of the y-axis, (default values: ymin=0, ymax=max(-log10(df$P))).
ymax: Integer, min and max of the y-axis, (default values: ymin=0, ymax=max(-log10(df$P))).
protein_coding_only: A logical scalar, if TRUE, only protein coding genes are used for annotation.
region_size: An integer (default = 1000000) indicating the window size for variant labelling. Increase this number for sparser annotation and decrease for denser annotation.
gene_padding: An integer representing size of the region around the gene, if the gene argument was used (default = 100000).
angle: A number, the angle of the text label.
legend_title_size: A number, size of the legend title.
legend_text_size  
A number, size of the legend text

nudge_x  
A number to vertically adjust the starting position of each gene label (this is a ggrepel parameter)

nudge_y  
A number to horizontally adjust the starting position of each gene label (this is a ggrepel parameter)

rsids  
A string (rsid) or vector of strings to highlight on the plot, e.g. rsids=c("rs1234, rs45898")

variant  
A string representing the variant to zoom in on. Can be either an rsid, or a dataframe (with the columns CHROM,POS,P)

rsids_color  
A string, the color of the variants in variants_id (default color is red)

legend_name  
A string, use to change the name of the legend (default: None)

legend_position  
A string, top, bottom, left or right

chr  
A string or integer, the chromosome to plot (i.e. chr15), only required if the input dataframe contains results from more than one chromosome

vline  
A number or vector of numbers to add a vertical line to the plot at a specific chromosomal position, e.g. vline=204000066. Multiple values can be provided in a vector, e.g. vline=c(204000066, 100500188)

show_gene_names  
A logical scalar, if set to TRUE, gene names are shown even though they exceed the max_genes count

legend_labels  
A string or vector of strings representing legend labels for the input dataset/s

gene  
A string representing the gene to zoom in on (e.g. gene=FTO)

title  
A string

label_color  
A string. To change the color of the gene or variant labels

region  
A string representing a genetic region, e.g. chr1:67038906-67359979

scale  
A number, to change the size of the title and axes labels and ticks at the same time (default = 1)

rsids_with_vline  
A string (rsid) or vector of strings to highlight on the plot with their rsids and vertical lines further highlighting their positions

annotate_with_vline  
A number (p-value). Display annotation and vertical lines for variants with p-values below this threshold

sign_thresh_size  
A number, sets the size of the horizontal significance threshold line (default = 1)

unit_main  
the height unit of the main plot (default = 7)

unit_gene  
the height unit of the gene plot (default= 2 )

Value

plots using egg (https://cran.r-project.org/web/packages/egg/vignettes/Ecosystem.html)
Examples

locuszoom(R2_CD_UKBB)

manhattan

Create a Manhattan plot

Description

manhattan() displays association results for the entire genome on a Manhattan plot. Required parameter is at least one dataset (dataframe) containing the association data (with columns CHROM, POS, P in upper or lowercase)

All other input parameters are optional

Usage

manhattan(
  df,
  ntop = 3,
  title = "",
  annotate = NULL,
  color = get_topr_colors(),
  sign_thresh = 5e-09,
  sign_thresh_color = "red",
  sign_thresh_label_size = 3.5,
  label_size = 3.5,
  size = 0.8,
  shape = 19,
  alpha = 1,
  highlight_genes_color = "green",
  highlight_genes_ypos = 1,
  axis_text_size = 12,
  axis_title_size = 14,
  title_text_size = 15,
  legend_title_size = 13,
  legend_text_size = 12,
  protein_coding_only = TRUE,
  angle = 0,
  legend_labels = NULL,
  chr = NULL,
  annotate_with = "Gene_Symbol",
  region_size = 1e+06,
  legend_name = NULL,
  legend_position = "bottom",
  nudge_x = 0.1,
  nudge_y = 0.2,
  xmin = NULL,
Arguments

df
- Dataframe or a list of dataframes (required columns are CHROM, POS, P), in upper- or lowercase) of association results.

ntop
- An integer, number of datasets (GWAS results) to show on the top plot

title
- A string

annotate
- A number (p-value). Display annotation for variants with p-values below this threshold

color
- A string or a vector of strings, for setting the color of the datapoints on the plot

sign_thresh
- A number or vector of numbers, setting the horizontal significance threshold (default: sign_thresh=5.1e-9). Set to NULL to hide the significance threshold.

sign_thresh_color
- A string or vector of strings to set the color/s of the significance threshold/s

sign_thresh_label_size
- A number setting the text size of the label for the significance thresholds (default text size is 3.5)

label_size
- An number to set the size of the plot labels (default: label_size=3)

size
- An integer setting the size of the plot points (default: size=1.2)

shape
- A number of vector of numbers setting the shape of the plotted points

alpha
- A number or vector of numbers setting the transparency of the plotted points

highlight_genes_color
- A string, color for the highlighted genes (default: green)

highlight_genes_ypos
- An integer, controlling where on the y-axis the highlighted genes are placed (default value is 1)

axis_text_size
- A number, size of the x and y axes tick labels (default: 12)
axis_title_size
A number, size of the x and y title labels (default: 12)
title_text_size
A number, size of the plot title (default: 13)
legend_title_size
A number, size of the legend title
legend_text_size
A number, size of the legend text
protein_coding_only
A logical scalar, if TRUE, only protein coding genes are used for annotation
angle
A number, the angle of the text label
legend_labels
A string or vector of strings representing legend labels for the input dataset/s
chr
A string or integer, the chromosome to plot (i.e. chr15), only required if the input dataframe contains results from more than one chromosome
annotate_with
A string. Annotate the variants with either Gene_Symbol or ID (default: "Gene_Symbol")
region_size
An integer (default = 1000000) indicating the window size for variant labelling. Increase this number for sparser annotation and decrease for denser annotation.
legend_name
A string, use to change the name of the legend (default: None)
legend_position
A string, top, bottom, left or right
nudge_x
A number to vertically adjust the starting position of each gene label (this is a ggrepel parameter)
nudge_y
A number to horizontally adjust the starting position of each gene label (this is a ggrepel parameter)
xmin, xmax
Integer, setting the chromosomal range to display on the x-axis
ymin, ymax
Integer, min and max of the y-axis, (default values: ymin=0, ymax=max(-log10(df$P)))
highlight_genes
A string or vector of strings, gene or genes to highlight at the bottom of the plot
label_color
A string. To change the color of the gene or variant labels
legend_nrow
An integer, sets the number of rows allowed for the legend labels
gene_label_size
A number setting the size of the gene labels shown at the bottom of the plot
gene_label_angle
A number setting the angle of the gene label shown at the bottom of the plot (default: 0)
scale
A number, to change the size of the title and axes labels and ticks at the same time (default = 1)
show_legend
A logical scalar, set to FALSE to hide the legend (default value is TRUE)
sign_thresh_linetype
A string, the linetype of the horizontal significance threshold (default = dashed)
sign_thresh_size
A number, sets the size of the horizontal significance threshold line (default = 1)
match_alleles

<table>
<thead>
<tr>
<th>Argument</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>rsids</td>
<td>A string (rsid) or vector of strings to highlight on the plot, e.g. <code>rsids=c(&quot;rs1234, rs45898&quot;)</code></td>
</tr>
<tr>
<td>rsids_color</td>
<td>A string, the color of the variants in <code>variants_id</code> (default color is red)</td>
</tr>
<tr>
<td>rsids_with_vline</td>
<td>A string (rsid) or vector of strings to highlight on the plot with their rsids and vertical lines further highlighting their positions</td>
</tr>
<tr>
<td>annotate_with_vline</td>
<td>A number (p-value). Display annotation and vertical lines for variants with p-values below this threshold</td>
</tr>
</tbody>
</table>

Value

ggplot object

Examples

manhattan(CD_UKBB)

match_alleles

Match the variants in the snpset by their alleles

Description

match_alleles()

Usage

match_alleles(df, verbose = FALSE)

Arguments

<table>
<thead>
<tr>
<th>Argument</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>df</td>
<td>A dataframe that is in the snpset format (like returned by the get_snpset() function)</td>
</tr>
<tr>
<td>verbose</td>
<td>A logical scalar (default: FALSE). Assign to TRUE to get information on which alleles are matched and which are not.</td>
</tr>
</tbody>
</table>

Value

The input dataframe containing only those variants with matched alleles in the snpset

Examples

CD_UKBB_index_snps <- get_best.snp_per_MB(CD_UKBB)
snpset <- create_snpset(CD_UKBB_index_snps, CD_FINNGEN)
match_alleles(snpset)
qqtopr

Create a QQ plot

Description

qqtopr() displays QQ plots for association data. Required parameter is at least one dataset (dataframe) containing the association data (with columns CHROM, POS, P).

Usage

```r
qqtopr(
  dat,
  scale = 1,
  n_variants = 0,
  breaks = 15,
  title = NULL,
  color = get_topr_colors(),
  size = 1,
  legend_name = "",
  legend_position = "right",
  legend_labels = NULL,
  axis_text_size = 11,
  axis_title_size = 12,
  title_text_size = 13,
  legend_title_size = 12,
  legend_text_size = 12
)
```

Arguments

dat         Dataframe or a list of dataframes (required columns is P) of association results.
scale       An integer, plot elements scale, default: 1
n_variants  An integer, total number of variants used in the study
breaks      A number setting the breaks for the axes
title       A string
color       A string or vector of strings setting the color/s for the input dataset/s
size        An integer setting the size of the plot points (default: size=1.2)
legend_name A string, use to change the name of the legend (default: None)
legend_position A string, top, bottom, left or right
legend_labels A string or vector of strings representing legend labels for the input dataset/s
axis_text_size A number, size of the x and y axes tick labels (default: 12)
axis_title_size A number, size of the x and y title labels (default: 12)
R2_CD_UKBB

**title_text_size**
A number, size of the plot title (default: 13)

**legend_title_size**
A number, size of the legend title

**legend_text_size**
A number, size of the legend text

**Value**

`ggplot`

---

**Description**

The dataset is a subset of CD_UKBB and only includes variants above and near the IL23R gene on chromosome 1.

**Usage**

`R2_CD_UKBB`

**Format**

A data frame with 26,824 rows and 10 variables:

- **CHROM** Chromosome, written as for example chr1 or 1
- **POS** genetic position of the variant
- **ID** Variant identifier, e.g. rsid
- **P** P-value from Plink run, additive model, reggresion model GLM_FIRTH
- **R2** variant correlation ($r^2$)

**Source**

A subset of the CD_UKBB dataset
regionplot

Create a regionplot

Description

regionplot() displays the association results for a smaller genetic regions within one chromosome. Required parameter is at least one dataset (dataframe) containing the association data (with columns CHROM, POS, P in upper or lowercase) and either a variant ID, gene name or the genetic region represented as a chromosome together with start and stop positions (either as a single string or as three separate arguments).

All other input parameters are optional

Usage

regionplot(
  df,
  ntop = 3,
  annotate = NULL,
  xmin = 0,
  size = 2,
  shape = 19,
  alpha = 1,
  label_size = 4,
  annotate_with = "ID",
  color = get_topr_colors(),
  axis_text_size = 11,
  axis_title_size = 12,
  title_text_size = 13,
  show_genes = FALSE,
  show_overview = TRUE,
  show_exons = FALSE,
  max_genes = 200,
  sign_thresh = 5e-09,
  sign_thresh_color = "red",
  sign_thresh_label_size = 3.5,
  xmax = NULL,
  ymin = NULL,
  ymax = NULL,
  protein_coding_only = FALSE,
  region_size = 1e+06,
  gene_padding = 1e+05,
  angle = 0,
  legend_title_size = 12,
  legend_text_size = 11,
  nudge_x = 0.01,
  nudge_y = 0.01,
regionplot

rsids = NULL,
variant = NULL,
rsids_color = NULL,
legend_name = "",
legend_position = "right",
chr = NULL,
vline = NULL,
show_gene_names = NULL,
legend_labels = NULL,
gene = NULL,
title = NULL,
label_color = NULL,
locuszoomplot = FALSE,
region = NULL,
legend_nrow = NULL,
gene_label_size = NULL,
scale = 1,
show_legend = TRUE,
sign_thresh_linetype = "dashed",
sign_thresh_size = 0.5,
rsids_with_vline = NULL,
annotate_with_vline = NULL,
show_gene_legend = TRUE,
unit_main = 7,
unit_gene = 2,
unit_overview = 1.25,
verbose = TRUE
)

Arguments

df          Dataframe or a list of dataframes (required columns are CHROM, POS, P), in upper- or lowercase) of association results.
ntop        An integer, number of datasets (GWAS results) to show on the top plot
annotate    A number (p-value). Display annotation for variants with p-values below this threshold
xmin        Integer, setting the chromosomal range to display on the x-axis
size        An integer setting the size of the plot points (default: size=1.2)
shape       A number of vector of numbers setting the shape of the plotted points
alpha       A number or vector of numbers setting the transparency of the plotted points
label_size  An number to set the size of the plot labels (default: label_size=3)
annotate_with A string. Annotate the variants with either Gene_Symbol or ID (default: "Gene_Symbol")
color       A string or a vector of strings, for setting the color of the datapoints on the plot
axis_text_size A number, size of the x and y axes tick labels (default: 12)
axis_title_size A number, size of the x and y title labels (default: 12)
title_text_size
   A number, size of the plot title (default: 13)

show_genes
   A logical scalar, show genes instead of exons (default show_genes=FALSE)

show_overview
   A logical scalar, shows/hides the overview plot (default= TRUE)

show_exons
   A logical scalar, show exons instead of genes (default show_exons=FALSE)

max_genes
   An integer, only label the genes if they are fewer than max_genes (default values is 200).

sign_thresh
   A number or vector of numbers, setting the horizontal significance threshold (default: sign_thresh=5.1e-9). Set to NULL to hide the significance threshold.

sign_thresh_color
   A string or vector of strings to set the color/s of the significance threshold/s

sign_thresh_label_size
   A number, setting the text size of the label for the significance thresholds (default text size is 3.5)

xmax
   Integer, setting the chromosomal range to display on the x-axis

ymn
   Integer, min and max of the y-axis, (default values: ymn=0, ymax=max(-log10(df$P)))

ymax
   Integer, min and max of the y-axis, (default values: ymn=0, ymax=max(-log10(df$P)))

protein_coding_only
   A logical scalar, if TRUE, only protein coding genes are used for annotation

region_size
   An integer (default = 1000000) indicating the window size for variant labelling. Increase this number for sparser annotation and decrease for denser annotation.

gene_padding
   An integer representing size of the region around the gene, if the gene argument was used (default = 100000)

angle
   A number, the angle of the text label

legend_title_size
   A number, size of the legend title

legend_text_size
   A number, size of the legend text

nudge_x
   A number to vertically adjust the starting position of each gene label (this is a ggrepel parameter)

nudge_y
   A number to horizontally adjust the starting position of each gene label (this is a ggrepel parameter)

rsids
   A string (rsid) or vector of strings to highlight on the plot, e.g. rsids=c("rs1234, rs45898")

variant
   A string representing the variant to zoom in on. Can be either an rsid, or a dataframe (with the columns CHROM,POS,P)

rsids_color
   A string, the color of the variants in variants_id (default color is red)

legend_name
   A string, use to change the name of the legend (default: None)

legend_position
   A string, top, bottom, left or right

chr
   A string or integer, the chromosome to plot (i.e. chr15), only required if the input dataframe contains results from more than one chromosome
regionplot

vline  A number or vector of numbers to add a vertical line to the plot at a specific chromosomal position, e.g. vline=204000066. Multiple values can be provided in a vector, e.g. vline=c(204000066,100500188)

show_gene_names  A logical scalar, if set to TRUE, gene names are shown even though they exceed the max_genes count

legend_labels  A string or vector of strings representing legend labels for the input dataset/s

gene  A string representing the gene to zoom in on (e.g. gene=FTO)

title  A string

label_color  A string. To change the color of the gene or variant labels

locuszoomplot  A logical scalar set to FALSE. Only set to TRUE by calling the locuszoom function

region  A string representing a genetic region, e.g. chr1:67038906-67359979

legend_nrow  An integer, sets the number of rows allowed for the legend labels

gene_label_size  A number setting the size of the gene labels shown at the bottom of the plot

scale  A number, to change the size of the title and axes labels and ticks at the same time (default = 1)

show_legend  A logical scalar, set to FALSE to hide the legend (default value is TRUE)

sign_thresh_linetype  A string, the linetype of the horizontal significance threshold (default = dashed)

sign_thresh_size  A number, sets the size of the horizontal significance threshold line (default = 1)

rsids_with_vline  A string (rsid) or vector of strings to highlight on the plot with their rsids and vertical lines further highlighting their positions

annotate_with_vline  A number (p-value). Display annotation and vertical lines for variants with p-values below this threshold

show_gene_legend  A logical scalar, set to FALSE to hide the gene legend (default value is TRUE)

unit_main  the height unit of the main plot (default = 7)

unit_gene  the height unit of the gene plot (default = 2)

unit_overview  the height unit of the overview plot (default = 1.25)

verbose  Logical, set to FALSE to get suppress printed information

Value

plots within ggplotGrobs, arranged with egg::gtable_frame

Examples

regionplot(CD_UKBB, gene="IL23R")
Description

A package for viewing and annotating genetic association data

topr functions

The main plotting functions are:

- `manhattan` to create Manhattan plot of association results
- `regionplot` to create regional plots of association results for smaller genetic regions

Examples

```r
library(topr)
# Create a manhattan plot using
manhattan(CD_UKBB)

# Create a regional plot
regionplot(CD_UKBB, gene="IL23R")

# Get the lead/index snps (the top snp per MB window)
get_best_snp_per_MB(CD_UKBB)

# Annotate the index snps with their nearest gene
index_snps <- get_best_snp_per_MB(CD_UKBB)
annotate_with_nearest_gene(index_snps)
```

UC_UKBB

UKBB Ulcerative colitis (ICD 10 code K51)

Description

Dataset retrieved from the UK biobank including of 5,452 UC cases (K51) and 481,862 controls. The dataset has been filtered on variants with P<1e-03.

Usage

UC_UKBB
Format

A data frame with 57,383 rows and 10 variables

- **CHROM**: Chromosome, written as for example chr1 or 1
- **POS**: Genetic position of the variant
- **ID**: Variant identifier, e.g. rsid
- **P**: P-value from Plink run, additive model, regression model GLM_FIRTH
- **OR**: Odds Ratio

Source

Ulcerative Colitis UKBB ICD10 code K51, only including variants with P<1e-03
Index

* datasets
  - CD_FINNGEN, 3
  - CD_UKBB, 4
  - R2_CD_UKBB, 21
  - UC_UKBB, 26

annotate_with_nearest_gene, 2

CD_FINNGEN, 3
CD_UKBB, 4
create_snpset, 4
create_snpset_code, 5

dat_column_check_and_set, 6

effect_plot, 6

flip_to_positive_allele_for_dat1, 7

get_best_snp_per_MB, 8
get_gene, 9
get_genes_by_Gene_Symbol, 9
get_overlapping_snps_by_pos, 10
get_snps_within_region, 11
get_top_snp, 12
get_topr_colors, 11

locuszoom, 12

manhattan, 16, 26
match_alleles, 19

qqtopr, 20

R2_CD_UKBB, 21
regionplot, 22, 26

topr, 26

UC_UKBB, 26