

Package ‘topr’

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Title Create Custom Plots for Viewing Genetic Association Results

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BugReports <https://github.com/totajuliusd/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.

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Author Thorhildur Juliusdottir [cph, aut, cre],
Andri Stefansson [aut],
Kyle Scott [ctb]

Maintainer Thorhildur Juliusdottir <totajuliusd@gmail.com>

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Contents

annotate_with_nearest_gene	2
CD_FINNGEN	3
CD_UKBB	4
create_snpset	5
create_snpset_code	6
effectplot	6
effect_plot	9
flip_to_positive_allele_for_dat1	10
foresttopr	11
get_best_snp_per_MB	14
get_gene	15
get_genes_by_Gene_Symbol	16
get_genes_in_region	16
get_gene_coords	17
get_lead_snps	18
get_overlapping_snps_by_pos	19
get_sign_and_sugg_loci	20
get_snpset	21
get_snpset_code	22
get_snps_within_region	22
get_topr_colors	23
get_topr_old_colors	24
get_top_snp	24
locuszoom	25
manhattan	30
manhattanExtra	35
match_alleles	37
match_by_alleles	38
match_by_pos	38
qqtopr	39
R2_CD_UKBB	41
regionplot	41
UC_UKBB	47
Index	48

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,
  build = 38,
  .chr_map = NULL
)

```

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)
build	A number representing the genome build. Set to 37 to change to build (GRCh37). The default is build 38 (GRCh38).
.chr_map	An internally used list which maps chromosome names to numbers.

Value

the input dataframe with Gene_Symbol as an additional column

Examples

```

## Not run:
variants <- get_lead_snps(CD_UKBB)
annotate_with_nearest_gene(variants)

## End(Not run)

```

CD_FINNGEN

Finngen r7 Crohn's disease (K11_CROHNS)

Description

Dataset retrieved from the Finngen database (version 7) including 3147 crohn's cases (K50) and 296,100 controls. The dataset has been filtered on variants with $P < 1e-03$. FinnGen data are publicly available and were downloaded from <https://finngen.fi>.

Usage

```
CD_FINNGEN
```

Format

A data frame with 32,303 rows and 8 variables:

CHROM Chromosome, written as for example chr1 or 1

POS genetic position of the variant

REF the reference allele

ALT the alternative allele

P P-value from Plink run, additive model, regression model GLM_FIRTH

BETA Variant effect

ID Variant identifier, e.g. rsid

AF Allele frequency

Source

Crohn's K50 (K11_CROHNS), 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 21,717 rows and 8 variables:

CHROM Chromosome, written as for example chr1 or 1

POS genetic position of the variant

REF the reference allele

ALT the alternative allele

ID Variant identifier, e.g. rsid

P P-value from Plink run, additive model, regression model GLM_FIRTH

OR Odds Ratio

AF Allele frequency

Source

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

create_snpset	<i>Create a dataframe that can be used as input for making effect plots</i>
---------------	---

Description

```
create_snpset()
```

This method is deprecated and will be removed in future versions. use [get_snpset](#) instead.

Usage

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

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

```
## Not run:  
create_snpset(CD_UKBB,CD_FINNGEN, thresh=1e-09)  
  
## End(Not run)
```

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

Description

This method is deprecated and will be removed in future versions. use [get_snpset_code](#) instead.

```
create_snpset_code()
```

Usage

```
create_snpset_code()
```

Value

Dataframe containing the top hit

Examples

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

effectplot *Create a plot comparing variant effects in two datasets*

Description

```
effectplot()
```

Usage

```
effectplot(  
  df,  
  pheno_x = "x_pheno",  
  pheno_y = "y_pheno",  
  annotate_with = "Gene_Symbol",  
  thresh = 5e-08,  
  ci_thresh = 1,  
  gene_label_thresh = 5e-08,  
  color = get_topr_colors()[1],  
  scale = 1,  
  build = 38,  
  label_fontface = "italic",  
  label_family = "",
```

```

nudge_y = 0.001,
nudge_x = 0.001,
size = 2,
segment.size = 0.2,
segment.linetype = "solid",
segment.color = "transparent",
angle = 0,
title = NULL,
axis_text_size = 10,
axis_title_size = 12,
title_text_size = 13,
subtitle_text_size = 11,
gene_label_size = 3.2,
snpsset_thresh = 5e-08,
snpsset_region_size = 1e+06,
max.overlaps = 10,
annotate = 0,
label_color = NULL
)

```

Arguments

df	The input dataframe (snpsset) containing one row per variant and P values (P1 and P2) and effects (E1 and E2) from two datasets/phenotypes OR a list containing two datasets.
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	Deprecated: A number, 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)
build	A number representing the genome build or a data frame. Set to 37 to change to build (GRCh37). The default is build 38 (GRCh38).
label_fontface	A string or a vector of strings. Label font "plain", "bold", "italic", "bold.italic" (ggrepel argument)
label_family	A string or a vector of strings. Label font name (default ggrepel argument is "")

nudge_y	A number to horizontally adjust the starting position of each gene label (this is a ggrepel parameter)
nudge_x	A number to vertically adjust the starting position of each gene label (this is a ggrepel parameter)
size	A number or a vector of numbers, setting the size of the plot points (default: size=1.2)
segment.size	line segment color (ggrepel argument)
segment.linetype	line segment solid, dashed, etc.(ggrepel argument)
segment.color	line segment thickness (ggrepel argument)
angle	A number, the angle of the text label
title	A string to set the plot title
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)
subtitle_text_size	A number setting the text size of the subtitle (default: 11)
gene_label_size	A number setting the size of the gene labels shown at the bottom of the plot
snpset_thresh	A number representing the threshold used to create the snpset used for plotting (Only applicable if the input dataframe is a list containing two datasets)
snpset_region_size	A number representing the region size to use when creating the snpset used for plotting (Only applicable if the input dataframe is a list containing two datasets)
max.overlaps	Exclude text labels that overlap too many things. Defaults to 10 (ggrepel argument)
annotate	A number, label datapoints with p-value below below this number (in the second df) by their nearest gene
label_color	A string or a vector of strings. To change the color of the gene or variant labels

Value

ggplot object

Examples

```
## Not run:
effectplot(list(CD_UKBB, CD_FINNGEN))

## End(Not run)
```

 effect_plot

 Create a plot comparing effects within two datasets

Description

```
effect_plot()
```

This method is deprecated and will be removed in future versions. use [effectplot](#) instead.

Usage

```
effect_plot(
  dat,
  pheno_x = "pheno_x",
  pheno_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 (snpsset) 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 topr colors
scale	A number, to change the size of the title and axes labels and ticks at the same time (default = 1)

Examples

```
## Not run:  
effect_plot(dat)  
  
## End(Not run)
```

```
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

Examples

```
## Not run:  
CD_UKBB_index_snps <- get_lead_snps(CD_UKBB)  
snpset <- get_snpset(CD_UKBB_index_snps, CD_FINNGEN)  
flip_to_positive_allele_for_dat1(snpset$matched)  
  
## End(Not run)
```

`foresttopr`*Create a forest plot from one or more association result tables*

Description

`foresttopr()` creates a forest plot visualizing effect estimates and confidence intervals across one or more datasets. The function supports odds ratios (OR) and regression coefficients (beta), allows matching rows across datasets by a key column, and optionally displays human-readable labels from a separate annotation column.

Effect estimates are automatically standardized across input datasets, and confidence intervals are derived preferentially from explicit bounds, standard errors, or p-values when necessary.

Usage

```
foresttopr(  
  dat = NULL,  
  legend_labels = NULL,  
  colors = NULL,  
  key_col = "ID",  
  label_col = NULL,  
  effect_type = c("OR", "beta"),  
  xlim = NULL,  
  xbreaks = NULL,  
  xlabel = NULL,  
  size = 2.5,  
  shape = 16,  
  alpha = 1,  
  points_dist = 0.6,  
  band_color = "grey96",  
  band_border_color = "grey96",  
  band_border_linewidth = 0.01,  
  sign_thresh = NULL,  
  ylabel_order = NULL,  
  scale = 1,  
  title = NULL,  
  title_text_size = 15,  
  axis_text_size = 12,  
  axis_title_size = 14,  
  show_shape_legend = TRUE,  
  show_color_legend = TRUE,  
  legend_position = "right",  
  legend_nrow = NULL,  
  legend_name = NULL,  
  legend_title_size = axis_text_size * 0.95,  
  legend_text_size = axis_text_size * 0.85,  
  match_on_gene = FALSE  
)
```

Arguments

<code>dat</code>	A data frame or a list of data frames containing association results. Each data frame must contain an effect estimate column (e.g. OR or BETA) and a p-value column. If a single data frame is provided, it is internally wrapped into a list.
<code>legend_labels</code>	A character vector of labels corresponding to each dataset in <code>dat</code> . These labels are used in the plot legend. Defaults to "Set1", "Set2", etc.
<code>colors</code>	A character vector of colors to use for each dataset. If NULL, a default color palette is used.
<code>key_col</code>	Character scalar giving the column name used to match rows across datasets (e.g. gene identifier or variant ID). Defaults to "gene".
<code>label_col</code>	Optional character scalar giving the column name in the reference dataset (the first element of <code>dat</code>) to use for labeling rows on the y-axis. If NULL, <code>key_col</code> is used for labeling.
<code>effect_type</code>	Character scalar specifying the effect scale to plot. Either "OR" (odds ratio; default) or "beta" (regression coefficient). Matching is case-insensitive. When required, effect estimates are automatically converted between scales.
<code>xlim</code>	Numeric length-2 vector giving x-axis limits. If NULL, limits are computed automatically from the data.
<code>xbreaks</code>	Numeric vector or function specifying x-axis breaks. If NULL, reasonable defaults are chosen based on <code>effect_type</code> .
<code>xlabel</code>	Character scalar giving the x-axis label. If NULL, a default label is chosen based on <code>effect_type</code> .
<code>size</code>	Numeric scalar or vector controlling point sizes for each dataset.
<code>shape</code>	Integer scalar or vector specifying point shapes for each dataset.
<code>alpha</code>	Numeric scalar or vector specifying point transparency.
<code>points_dist</code>	Numeric scalar controlling horizontal separation of points from different datasets within the same row.
<code>band_color</code>	Background color for alternating row bands.
<code>band_border_color</code>	Color for row band borders.
<code>band_border_linewidth</code>	Numeric scalar giving the line width for row band borders.
<code>sign_thresh</code>	Optional numeric scalar specifying a p-value threshold for highlighting statistically significant points via shape encoding.
<code>ylabel_order</code>	Optional character vector specifying the order of rows on the y-axis. If NULL, rows are ordered as they appear in the reference dataset.
<code>scale</code>	Numeric scalar used to globally scale text and point sizes.
<code>title</code>	Optional character scalar giving the plot title.
<code>title_text_size</code>	Numeric scalar controlling title text size.
<code>axis_text_size</code>	Numeric scalar controlling axis text size.

<code>axis_title_size</code>	Numeric scalar controlling axis title text size.
<code>show_shape_legend</code>	Logical; whether to display the shape legend.
<code>show_color_legend</code>	Logical; whether to display the color legend.
<code>legend_position</code>	Character string specifying legend position. One of "right", "top", or "bottom".
<code>legend_nrow</code>	Optional integer specifying the number of rows in the legend.
<code>legend_name</code>	Optional character scalar giving the legend title.
<code>legend_title_size</code>	Numeric scalar controlling legend title text size.
<code>legend_text_size</code>	Numeric scalar controlling legend text size.
<code>match_on_gene</code>	Logical; if FALSE and variant-level columns (e.g. REF/ALT) are detected, matching is performed at the variant level. Otherwise, matching is performed using <code>key_col</code> .

Value

A `ggplot2` object representing the forest plot.

See Also

[ggplot](#)

Examples

```
foresttopr(
  dat = list(
    CD_UKBB |>
      dplyr::arrange(P) |>
      head(n = 10) |>
      annotate_with_nearest_gene(),
    CD_FINNGEN
  ),
  key_col = "ID",
  label_col = "Gene_Symbol",
  legend_labels = c("CD_UKBB", "CD_FINNGEN"),
  effect_type = "beta"
)
```

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

This method is deprecated and will be removed in future versions. use [get_lead_snps](#) instead.

Usage

```
get_best_snp_per_MB(
  df,
  thresh = 5e-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 (5e-09 by default)
region_size	An integer (default = 2000000) (or a string represented as 200kb or 2MB) indicating the window size for variant labeling. 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=5e-09 by default)

Examples

```
## Not run:  
  get_best_snp_per_MB(CD_UKBB)  
  
## End(Not run)
```

get_gene	<i>Get the genetic position of a gene by gene name</i>
----------	--

Description

get_gene() Get the gene coordinates for a gene Required parameter is gene name
This method is deprecated and will be removed in future versions. use [get_gene_coords](#) instead.

Usage

```
get_gene(gene_name, chr = NULL, build = 38)
```

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")
build	A string, genome build, choose between builds 37 (GRCh37) and 38 (GRCh38) (default is 38)

Value

Dataframe with the gene name and its genetic coordinates

Examples

```
## Not run:  
  get_gene("FTO")  
  
## End(Not run)
```

```
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, build = 38)
```

Arguments

<code>genes</code>	A string or vector of strings representing gene names, (e.g. "FTO") or (c("FTO","NOD2"))
<code>chr</code>	A string, search for the genes on this chromosome only, (e.g chr="chr1")
<code>build</code>	A string, genome build, choose between builds 37 (GRCh37) and 38 (GRCh38) (default is 38)

Value

Dataframe of genes

Examples

```
## Not run:
  get_genes_by_Gene_Symbol(c("FTO", "THADA"))

## End(Not run)
```

```
get_genes_in_region
```

Get SNPs/variants within region

Description

```
get_genes_in_region()
```

Usage

```

get_genes_in_region(
  chr = chr,
  xmin = xmin,
  xmax = xmax,
  protein_coding_only = F,
  show_exons = F,
  show_genes = T,
  build = 38,
  region = NULL
)

```

Arguments

chr	A string, chromosome (e.g. chr16)
xmin	An integer representing genetic position
xmax	An integer representing genetic position
protein_coding_only	A logical scalar, if TRUE, only protein coding genes are used for annotation
show_exons	Deprecated : A logical scalar, show exons instead of genes (default show_exons=FALSE)
show_genes	A logical scalar, show genes instead of exons (default show_genes=FALSE)
build	A number representing the genome build or a data frame. Set to 37 to change to build (GRCh37). The default is build 38 (GRCh38).
region	A string representing the genetic region (e.g chr16:50693587-50734041)

Value

the genes the requested region

Examples

```

## Not run:
get_genes_in_region(region="chr16:50593587-50834041")

## End(Not run)

```

get_gene_coords	<i>Get the genetic position of a gene by gene name</i>
-----------------	--

Description

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

Usage

```
get_gene_coords(gene_name, chr = NULL, build = 38)
```

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")
build	A string, genome build, choose between builds 37 (GRCh37) and 38 (GRCh38) (default is 38)

Value

Dataframe with the gene name and its genetic coordinates

Examples

```
## Not run:
get_gene_coords("FTO")

## End(Not run)
```

get_lead_snps	<i>Get the index/lead variants</i>
---------------	------------------------------------

Description

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

Usage

```
get_lead_snps(
  df,
  thresh = 5e-08,
  region_size = 1e+06,
  protein_coding_only = FALSE,
  chr = NULL,
  .checked = FALSE,
  verbose = NULL,
  keep_chr = TRUE
)
```

Arguments

df	Dataframe
thresh	A number. P-value threshold, only extract variants with p-values below this threshold (5e-08 by default)
region_size	An integer (default = 2000000) (or a string represented as 200kb or 2MB) indicating the window size for variant labeling. 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
keep_chr	Logical, set to FALSE to remove the "chr" prefix before each chromosome if present (TRUE by default)

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=5e-08 by default)

Examples

```
## Not run:
get_lead_snps(CD_UKBB)

## End(Not run)
```

get_overlapping_snps_by_pos

Get variants that overlap between two datasets

Description

get_overlapping_snps_by_pos()

This method is deprecated and will be removed in future versions. use [match_by_pos](#) instead.

Usage

```
get_overlapping_snps_by_pos(df1, df2, verbose = F)
```

Arguments

df1	A dataframe of variants, has to contain CHROM and POS
df2	A dataframe of variants, has to contain CHROM and POS
verbose	A logical scalar (default: FALSE). Assign to TRUE to get information on which alleles are matched and which are not.

Value

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

Examples

```
## Not run:  
get_overlapping_snps_by_pos(dat1, dat2)  
  
## End(Not run)
```

```
get_sign_and_sugg_loci
```

Get the index/lead variants

Description

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

Usage

```
get_sign_and_sugg_loci(  
  df,  
  genome_wide_thresh = 5e-08,  
  suggestive_thresh = 1e-06,  
  flank_size = 1e+06,  
  region_size = 1e+06  
)
```

Arguments

df	Dataframe, GWAS summary statistics
genome_wide_thresh	A number. P-value threshold for genome wide significant loci (5e-08 by default)
suggestive_thresh	A number. P-value threshold for suggestive loci (1e-06 by default)
flank_size	A number (default = 1e6). The size of the flanking region for the significant and suggestitive snps.
region_size	A number (default = 1e6). The size of the region for top snp search. Only one snp per region is returned.

Value

List of genome wide and suggestive loci.

Examples

```
## Not run:  
get_sign_and_sugg_loci(CD_UKBB)  
  
## End(Not run)
```

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

Description

get_snpset()

Usage

```
get_snpset(
  df1,
  df2,
  thresh = 5e-08,
  protein_coding_only = TRUE,
  region_size = 1e+06,
  verbose = NULL,
  show_full_output = FALSE,
  build = 38,
  format = "wide"
)
```

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	A number. P-value threshold, only extract variants with p-values below this threshold (5e-08 by default)
protein_coding_only	Logical, set this variable to TRUE to only use protein_coding genes for annotation
region_size	An integer (default = 20000000) (or a string represented as 200kb or 2MB) indicating the window size for variant labeling. Increase this number for sparser annotation and decrease for denser annotation.
verbose	Logical, (default: FALSE). Assign to TRUE to get information on which alleles are matched and which are not.
show_full_output	A logical scalar (default:FALSE). Assign to TRUE to show the full output from this function
build	A string, genome build, choose between builds 37 (GRCh37) and 38 (GRCh38) (default is 38)
format	A string, representing either wide or long format (default : "wide"). By default a snpset created from two dataframes is returned in a wide format.

Value

Dataframe of overlapping snps (snpset)

Examples

```
## Not run:  
CD_UKBB_index_snps <-get_lead_snps(CD_UKBB)  
get_snpset(CD_UKBB_index_snps, CD_FINNGEN)  
  
## End(Not run)
```

get_snpset_code	<i>Show the code/functions used to get a snpset</i>
-----------------	---

Description

```
get_snpset_code()
```

Usage

```
get_snpset_code()
```

Value

Dataframe containing the top hit

Examples

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

get_snps_within_region	<i>Get SNPs/variants within region</i>
------------------------	--

Description

```
get_snps_within_region()
```

Usage

```
get_snps_within_region(  
  df,  
  region,  
  chr = NULL,  
  xmin = NULL,  
  xmax = NULL,  
  keep_chr = 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
keep_chr	Deprecated: Logical, set to FALSE to remove the "chr" prefix before each chromosome if present (TRUE by default)

Value

the variants within the requested region

Examples

```
## Not run:
get_snps_within_region(CD_UKBB, "chr16:50593587-50834041")

## End(Not run)
```

get_topr_colors	<i>Get the topr custom colors</i>
-----------------	-----------------------------------

Description

get_topr_colors() Get topr custom colors

Usage

```
get_topr_colors()
```

Value

Vector of colors used for plotting

Examples

```
## Not run:
get_topr_colors()

## End(Not run)
```

get_topr_old_colors *Get the topr custom colors from v. 2.0.2*

Description

get_topr_old_colors() Get topr custom colors from v. 2.0.2

Usage

```
get_topr_old_colors()
```

Value

Vector of colors used for plotting

Examples

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

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

```
## Not run:  
get_top_snp(CD_UKBB, chr="chr1")  
  
## End(Not run)
```

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 = NULL,  
  show_overview = FALSE,  
  show_exons = FALSE,  
  max_genes = 200,  
  sign_thresh = 5e-08,  
  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,
gene_color = NULL,
segment.size = 0.2,
segment.color = "black",
segment.linetype = "solid",
show_gene_legend = TRUE,
max.overlaps = 10,
extract_plots = FALSE,
label_fontface = "plain",
label_family = "",
gene_label_fontface = "plain",
gene_label_family = "",
build = 38,
verbose = NULL,
show_legend = TRUE,
label_alpha = 1,
gene_label_size = NULL,
vline_color = "grey",
vline_linetype = "dashed",
vline_alpha = 1,
vline_size = 0.5,
log_trans_p = TRUE
)

```

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
ntop	An integer, number of datasets (GWAS results) to show on the top plot
xmin, xmax	Integer, setting the chromosomal range to display on the x-axis
size	A number or a vector of numbers, setting the size of the plot points (default: size=1.2)
shape	A number or a vector of numbers setting the shape of the plotted points
alpha	A number or a 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	Deprecated : 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=5e-8). 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)
ymin, 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 = 20000000) (or a string represented as 200kb or 2MB) indicating the window size for variant labeling. 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 datasets
gene	A string representing the gene to zoom in on (e.g. gene=FTO)
title	A string to set the plot title
label_color	A string or a vector of strings. 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)
gene_color	A string representing a color, can be used to change the color of the genes/exons on the geneplot
segment.size	line segment color (ggrepel argument)
segment.color	line segment thickness (ggrepel argument)

<code>segment.linetype</code>	line segment solid, dashed, etc.(ggrepel argument)
<code>show_gene_legend</code>	A logical scalar, set to FALSE to hide the gene legend (default value is TRUE)
<code>max.overlaps</code>	Exclude text labels that overlap too many things. Defaults to 10 (ggrepel argument)
<code>extract_plots</code>	Logical, FALSE by default. Set to TRUE to extract the three plots separately in a list
<code>label_fontface</code>	A string or a vector of strings. Label font “plain”, “bold”, “italic”, “bold.italic” (ggrepel argument)
<code>label_family</code>	A string or a vector of strings. Label font name (default ggrepel argument is “”)
<code>gene_label_fontface</code>	Gene label font “plain”, “bold”, “italic”, “bold.italic” (ggrepel argument)
<code>gene_label_family</code>	Gene label font name (default ggrepel argument is “”)
<code>build</code>	A number representing the genome build or a data frame. Set to 37 to change to build (GRCh37). The default is build 38 (GRCh38).
<code>verbose</code>	Logical, set to FALSE to get suppress printed information
<code>show_legend</code>	A logical scalar, set to FALSE to hide the legend (default : TRUE)
<code>label_alpha</code>	An number or vector of numbers to set the transparency of the plot labels (default: label_alpha=1)
<code>gene_label_size</code>	A number setting the size of the gene labels shown at the bottom of the plot
<code>vline_color</code>	A string. The color of added vertical line/s (default: grey)
<code>vline_linetype</code>	A string. The linetype of added vertical line/s (default : dashed)
<code>vline_alpha</code>	A number. The alpha of added vertical line/s (default : 1)
<code>vline_size</code>	A number.The size of added vertical line/s (default : 0.5)
<code>log_trans_p</code>	A logical scalar (default: TRUE). By default the p-values in the input datasets are log transformed using -log10. Set this argument to FALSE if the p-values in the datasets have already been log transformed.

Value

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

Examples

```
## Not run:
locuszoom(R2_CD_UKBB)

## End(Not run)
```

`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 = 4,  
  title = "",  
  annotate = NULL,  
  color = NULL,  
  sign_thresh = 5e-08,  
  sign_thresh_color = "darkred",  
  sign_thresh_label_size = 3.5,  
  label_size = 3.5,  
  size = 0.8,  
  shape = 19,  
  alpha = 1,  
  highlight_genes_color = "darkred",  
  highlight_genes_ypos = 1.5,  
  axis_text_size = 12,  
  axis_title_size = 14,  
  title_text_size = 15,  
  legend_title_size = axis_text_size * 0.95,  
  legend_text_size = axis_text_size * 0.85,  
  protein_coding_only = TRUE,  
  angle = 0,  
  legend_labels = NULL,  
  chr = NULL,  
  annotate_with = "Gene_Symbol",  
  region_size = 2e+07,  
  legend_name = NULL,  
  legend_position = "bottom",  
  nudge_x = 0.1,  
  nudge_y = 0.7,  
  xmin = NULL,  
  xmax = NULL,  
  ymin = NULL,  
  ymax = NULL,
```

```
highlight_genes = NULL,
label_color = NULL,
legend_nrow = NULL,
gene_label_size = NULL,
gene_label_angle = 0,
scale = 1,
show_legend = TRUE,
sign_thresh_linetype = "dashed",
sign_thresh_size = 0.5,
rsids = NULL,
rsids_color = NULL,
rsids_with_vline = NULL,
annotate_with_vline = NULL,
shades_color = NULL,
shades_alpha = 0.5,
segment.size = 0.2,
segment.color = "black",
segment.linetype = "dashed",
max.overlaps = 10,
label_fontface = "plain",
label_family = "",
gene_label_fontface = "plain",
gene_label_family = "",
build = 38,
verbose = NULL,
label_alpha = 1,
shades_line_alpha = 1,
vline = NULL,
vline_color = "grey",
vline_linetype = "dashed",
vline_alpha = 1,
vline_size = 0.5,
region = NULL,
theme_grey = FALSE,
xaxis_label = "Chromosome",
use_shades = FALSE,
even_no_chr_lightness = 0.8,
get_chr_lengths_from_data = TRUE,
log_trans_p = TRUE,
chr_ticknames = NULL,
show_all_chrticks = FALSE,
hide_chrticks_from_pos = 17,
hide_chrticks_to_pos = NULL,
hide_every_nth_chrtick = 2,
downsample_cutoff = 0.05,
downsample_prop = 0.1
)
```

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 to set the plot title
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=5e-8). 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	A number or a vector of numbers, setting the size of the plot points (default: size=1.2)
shape	A number or a vector of numbers setting the shape of the plotted points
alpha	A number or a vector of numbers setting the transparency of the plotted points
highlight_genes_color	A string, color for the highlighted genes (default: darkred)
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 datasets
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 = 20000000) (or a string represented as 200kb or 2MB) indicating the window size for variant labeling. 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 or a vector of strings. 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 : TRUE)
sign_thresh_linetype	A string, the line-type of the horizontal significance threshold (default : dashed)
sign_thresh_size	A number, sets the size of the horizontal significance threshold line (default : 1)
rsids	A string (rsid) or vector of strings to highlight on the plot, e.g. rsids=c("rs1234, rs45898")
rsids_color	A string, the color of the variants in variants_id (default color is red)
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
shades_color	The color of the rectangles (shades) representing the different chromosomes on the Manhattan plot
shades_alpha	The transparency (alpha) of the rectangles (shades)
segment.size	line segment color (ggrepel argument)
segment.color	line segment thickness (ggrepel argument)

<code>segment.linetype</code>	line segment solid, dashed, etc.(ggrepel argument)
<code>max.overlaps</code>	Exclude text labels that overlap too many things. Defaults to 10 (ggrepel argument)
<code>label_fontface</code>	A string or a vector of strings. Label font “plain”, “bold”, “italic”, “bold.italic” (ggrepel argument)
<code>label_family</code>	A string or a vector of strings. Label font name (default ggrepel argument is “”)
<code>gene_label_fontface</code>	Gene label font “plain”, “bold”, “italic”, “bold.italic” (ggrepel argument)
<code>gene_label_family</code>	Gene label font name (default ggrepel argument is “”)
<code>build</code>	A number representing the genome build or a data frame. Set to 37 to change to build (GRCh37). The default is build 38 (GRCh38).
<code>verbose</code>	A logical scalar (default: NULL). Set to FALSE to suppress printed messages
<code>label_alpha</code>	An number or vector of numbers to set the transparency of the plot labels (default: label_alpha=1)
<code>shades_line_alpha</code>	The transparency (alpha) of the lines around the rectangles (shades)
<code>vline</code>	A number or vector of numbers to add a vertical line to the plot at a specific chromosomal position, e.g <code>vline="chr1:204000066"</code> . Multiple values can be provided in a vector, e.g <code>vline=c("chr1:204000066", "chr5:100500188")</code>
<code>vline_color</code>	A string. The color of added vertical line/s (default: grey)
<code>vline_linetype</code>	A string. The linetype of added vertical line/s (default : dashed)
<code>vline_alpha</code>	A number. The alpha of added vertical line/s (default : 1)
<code>vline_size</code>	A number.The size of added vertical line/s (default : 0.5)
<code>region</code>	A string representing a genetic region, e.g. <code>chr1:67038906-67359979</code>
<code>theme_grey</code>	A logical scalar (default: FALSE). Use gray rectangles (instead of white to distinguish between chromosomes)
<code>xaxis_label</code>	A string. The label for the x-axis (default: Chromosome)
<code>use_shades</code>	A logical scalar (default: FALSE). Use shades/rectangles to distinguish between chromosomes
<code>even_no_chr_lightness</code>	Lightness value for even numbered chromosomes. A number or vector of numbers between 0 and 1 (default: 0.8). If set to 0.5, the same color as shown for odd numbered chromosomes is displayed. A value below 0.5 will result in a darker color displayed for even numbered chromosomes, whereas a value above 0.5 results in a lighter color.
<code>get_chr_lengths_from_data</code>	A logical scalar (default: TRUE). If set to FALSE, use the inbuilt chromosome lengths (from hg38), instead of chromosome lengths based on the max position for each chromosome in the input dataset/s.
<code>log_trans_p</code>	A logical scalar (default: TRUE). By default the p-values in the input datasets are log transformed using $-\log_{10}$. Set this argument to FALSE if the p-values in the datasets have already been log transformed.

`chr_ticknames` A vector containing the chromosome names displayed on the x-axis. If NULL, the following format is used: `chr_ticknames <- c(1:16, "", 18, "", 20, "", 22, 'X')`

`show_all_chrticks`
A logical scalar (default : FALSE). Set to TRUE to show all the chromosome names on the ticks on the x-axis

`hide_chrticks_from_pos`
A number (default: 17). Hide every nth chromosome name on the x-axis FROM this position (chromosome number)

`hide_chrticks_to_pos`
A number (default: NULL). Hide every nth chromosome name on the x-axis TO this position (chromosome number). When NULL this variable will be set to the number of numeric chromosomes in the input dataset.

`hide_every_nth_chrtick`
A number (default: 2). Hide every nth chromosome tick on the x-axis (from the `hide_chr_ticks_from_pos` to the `hide_chr_ticks_to_pos`).

`downsample_cutoff`
A number (default: 0.05) used to downsample the input dataset prior to plotting. Sets the fraction of high p-value (default: $P > 0.05$) markers to display on the plot.

`downsample_prop`
A number (default: 0.1) used to downsample the input dataset prior to plotting. Only a proportion of the variants (10% by default) with P-values higher than the `downsample_cutoff` will be displayed on the plot.

Value

ggplot object

Examples

```
## Not run:
manhattan(CD_UKBB)

## End(Not run)
```

manhattanExtra	<i>Create a Manhattan plot highlighting genome-wide significant and suggestive loci</i>
----------------	---

Description

`manhattanExtra()` displays association results for the entire genome on a Manhattan plot, highlighting genome-wide significant and suggestive loci. 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

```
manhattanExtra(
  df,
  genome_wide_thresh = 5e-08,
  suggestive_thresh = 1e-06,
  flank_size = 1e+06,
  region_size = 1e+06,
  sign_thresh_color = NULL,
  sign_thresh_label_size = NULL,
  show_legend = TRUE,
  label_fontface = NULL,
  nudge_y = NULL,
  ymax = NULL,
  sign_thresh = NULL,
  label_color = NULL,
  color = NULL,
  legend_labels = NULL,
  annotate = NULL,
  ...
)
```

Arguments

<code>df</code>	Dataframe, GWAS summary statistics
<code>genome_wide_thresh</code>	A number. P-value threshold for genome wide significant loci (5e-08 by default)
<code>suggestive_thresh</code>	A number. P-value threshold for suggestive loci (1e-06 by default)
<code>flank_size</code>	A number (default = 1e6). The size of the flanking region for the significant and suggestitive snps.
<code>region_size</code>	A number (default = 1e6). The size of the region for gene annotation. Increase this number for sparser annotation and decrease for denser annotation.
<code>sign_thresh_color</code>	A string or vector of strings to set the color/s of the significance threshold/s
<code>sign_thresh_label_size</code>	A number setting the text size of the label for the significance thresholds (default text size is 3.5)
<code>show_legend</code>	A logical scalar, set to FALSE to hide the legend (default : TRUE)
<code>label_fontface</code>	A string or a vector of strings. Label font “plain”, “bold”, “italic”, “bold.italic” (ggrepel argument)
<code>nudge_y</code>	A number to horizontally adjust the starting position of each gene label (this is a ggrepel parameter)
<code>ymax</code>	Integer, max of the y-axis, (default value: $ymax = (\max(-\log_{10}(df\$P)) + \max(-\log_{10}(df\$P)) * .2)$)
<code>sign_thresh</code>	A number or vector of numbers, setting the horizontal significance threshold (default: <code>sign_thresh=5e-8</code>). Set to NULL to hide the significance threshold.

label_color	A string or a vector of strings. To change the color of the gene or variant labels
color	A string or a vector of strings, for setting the color of the datapoints on the plot
legend_labels	A string or vector of strings representing legend labels for the input datasets
annotate	A number (p-value). Display annotation for variants with p-values below this threshold
...	Additional arguments passed to other plotting functions.

Value

ggplot object

Examples

```
## Not run:
manhattanExtra(df)

## End(Not run)
```

match_alleles	<i>Match the variants in the snpset by their alleles</i>
---------------	--

Description

match_alleles()

This method is deprecated and will be removed in future versions. use [match_by_alleles](#) instead.

Usage

```
match_alleles(df, verbose = F)
```

Arguments

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

Value

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

Examples

```
## Not run:
match_alleles(df)

## End(Not run)
```

match_by_alleles	<i>Match the variants in the snpset by their alleles</i>
------------------	--

Description

```
match_by_alleles()
```

Usage

```
match_by_alleles(df, verbose = NULL, show_full_output = FALSE)
```

Arguments

df	A dataframe that is in the snpset format (like returned by the get_snpset function)
verbose	A logical scalar (default: FALSE). Assign to TRUE to get information on which alleles are matched and which are not.
show_full_output	A logical scalar (default:FALSE). Assign to TRUE to show the full output from this function

Value

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

Examples

```
## Not run:
CD_UKBB_lead_snps <- get_lead_snps(CD_UKBB)
snpset <- get_snpset(CD_UKBB_lead_snps, CD_FINNGEN)
match_by_alleles(snpset$found)

## End(Not run)
```

match_by_pos	<i>Get variants that overlap between two datasets</i>
--------------	---

Description

```
match_by_pos()
```

Usage

```
match_by_pos(df1, df2, verbose = NULL, show_full_output = FALSE)
```

Arguments

df1	A dataframe of variants, has to contain CHROM and POS
df2	A dataframe of variants, has to contain CHROM and POS
verbose	A logical scalar (default: FALSE). Assign to TRUE to get information on which alleles are matched and which are not.
show_full_output	A logical scalar (default:FALSE). Assign to TRUE to show the full output from this function

Value

A list containing two dataframes, one of overlapping snps and the other snps not found in the second input dataset

Examples

```
## Not run:
CD_UKBB_index_snps <- get_lead_snps(CD_UKBB)
match_by_pos(CD_UKBB_index_snps, CD_FINNGEN)

## End(Not run)
```

qqtopr

Create a quantile quantile (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

```
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,
    verbose = NULL,
    diagonal_line_color = "#808080"
)

```

Arguments

<code>dat</code>	Dataframe or a list of dataframes (required columns is P)) of association results.
<code>scale</code>	An integer, plot elements scale, default: 1
<code>n_variants</code>	An integer, total number of variants used in the study
<code>breaks</code>	A number setting the breaks for the axes
<code>title</code>	A string to set the plot title
<code>color</code>	A string or vector of strings setting the color's for the input datasets
<code>size</code>	A number or a vector of numbers, setting the size of the plot points (default: size=1.2)
<code>legend_name</code>	A string, use to change the name of the legend (default: None)
<code>legend_position</code>	A string, top,bottom,left or right
<code>legend_labels</code>	A string or vector of strings representing legend labels for the input datasets
<code>axis_text_size</code>	A number, size of the x and y axes tick labels (default: 12)
<code>axis_title_size</code>	A number, size of the x and y title labels (default: 12)
<code>title_text_size</code>	A number, size of the plot title (default: 13)
<code>legend_title_size</code>	A number, size of the legend title
<code>legend_text_size</code>	A number, size of the legend text
<code>verbose</code>	A logical scalar (default: NULL). Set to FALSE to suppress printed messages
<code>diagonal_line_color</code>	A string setting the color of the diagonal line on the plot

Value

ggplot

Examples

```

## Not run:
qqtopr(CD_UKBB)

## End(Not run)

```

R2_CD_UKBB	<i>Example dataset including the R2 column for the locuszoom plot function</i>
------------	--

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 329 rows and 5 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

R2 variant correlation (r^2)

Source

A subset of the CD_UKBB dataset

regionplot	<i>Create a regionplot</i>
------------	----------------------------

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 = 10,  
  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 = NULL,  
  show_overview = TRUE,  
  show_exons = NULL,  
  max_genes = 200,  
  sign_thresh = 5e-08,  
  sign_thresh_color = "darkred",  
  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,  
  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 = NULL,
gene_color = NULL,
segment.size = 0.2,
segment.color = "black",
segment.linetype = "solid",
max.overlaps = 10,
unit_ratios = NULL,
extract_plots = FALSE,
label_fontface = "plain",
label_family = "",
gene_label_fontface = "plain",
gene_label_family = "",
build = 38,
label_alpha = 1,
vline_color = "grey",
vline_linetype = "dashed",
vline_alpha = 1,
vline_size = 0.5,
log_trans_p = TRUE,
lz_color = 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
annotate	A number (p-value). Display annotation for variants with p-values below this threshold
xmin, xmax	Integer, setting the chromosomal range to display on the x-axis
size	A number or a vector of numbers, setting the size of the plot points (default: size=1.2)
shape	A number or a vector of numbers setting the shape of the plotted points
alpha	A number or a 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	Deprecated : 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=5e-8). 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)
ymin, 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 = 2000000) (or a string represented as 200kb or 2MB) indicating the window size for variant labeling. 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 datasets
gene	A string representing the gene to zoom in on (e.g. gene=FTO)
title	A string to set the plot title
label_color	A string or a vector of strings. 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 : TRUE)
sign_thresh_linetype	A string, the line-type 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
gene_color	A string representing a color, can be used to change the color of the genes/exons on the geneplot
segment.size	line segment color (ggrepel argument)

<code>segment.color</code>	line segment thickness (ggrepel argument)
<code>segment.linetype</code>	line segment solid, dashed, etc.(ggrepel argument)
<code>max.overlaps</code>	Exclude text labels that overlap too many things. Defaults to 10 (ggrepel argument)
<code>unit_ratios</code>	A string of three numbers separated by ":", for the overview, main and gene plots height ratios e.g 1.25:7:2
<code>extract_plots</code>	Logical, FALSE by default. Set to TRUE to extract the three plots separately in a list
<code>label_fontface</code>	A string or a vector of strings. Label font "plain", "bold", "italic", "bold.italic" (ggrepel argument)
<code>label_family</code>	A string or a vector of strings. Label font name (default ggrepel argument is "")
<code>gene_label_fontface</code>	Gene label font "plain", "bold", "italic", "bold.italic" (ggrepel argument)
<code>gene_label_family</code>	Gene label font name (default ggrepel argument is "")
<code>build</code>	A number representing the genome build or a data frame. Set to 37 to change to build (GRCh37). The default is build 38 (GRCh38).
<code>label_alpha</code>	An number or vector of numbers to set the transparency of the plot labels (default: label_alpha=1)
<code>vline_color</code>	A string. The color of added vertical line/s (default: grey)
<code>vline_linetype</code>	A string. The linetype of added vertical line/s (default : dashed)
<code>vline_alpha</code>	A number. The alpha of added vertical line/s (default : 1)
<code>vline_size</code>	A number.The size of added vertical line/s (default : 0.5)
<code>log_trans_p</code>	A logical scalar (default: TRUE). By default the p-values in the input datasets are log transformed using -log10. Set this argument to FALSE if the p-values in the datasets have already been log transformed.
<code>lz_color</code>	A string, to set the base color in the locuszoom plot

Value

plots within ggplotGrobs, arranged with `egg::gtable_frame`

Examples

```
## Not run:
regionplot(CD_UKBB, gene="IL23R")

## End(Not run)
```

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 45,012 rows and 8 variables

CHROM Chromosome, written as for example chr1 or 1

POS genetic position of the variant

REF the reference allele

ALT the alternative allele

ID Variant identifier, e.g. rsid

P P-value from Plink run, additive model, regression model GLM_FIRTH

OR Odds Ratio

AF Allele frequency

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, 41
UC_UKBB, 47

annotate_with_nearest_gene, 2

CD_FINNGEN, 3
CD_UKBB, 4
create_snpset, 5
create_snpset_code, 6

effect_plot, 9
effectplot, 6, 9

flip_to_positive_allele_for_dat1, 10
foresttopr, 11

get_best_snp_per_MB, 14
get_gene, 15
get_gene_coords, 15, 17
get_genes_by_Gene_Symbol, 16
get_genes_in_region, 16
get_lead_snps, 14, 18
get_overlapping_snps_by_pos, 19
get_sign_and_sugg_loci, 20
get_snps_within_region, 22
get_snpset, 5, 21, 38
get_snpset_code, 6, 22
get_top_snp, 24
get_topr_colors, 23
get_topr_old_colors, 24
ggplot, 13

locuszoom, 25

manhattan, 30
manhattanExtra, 35
match_alleles, 37
match_by_alleles, 37, 38

match_by_pos, 19, 38

qqtopr, 39

R2_CD_UKBB, 41
regionplot, 41

UC_UKBB, 47