Package 'wrapperQTL'

August 31, 2025

Title MatrixEQTL Wrapper for QTL Analysis

Version 1.0.0

Maintainer Lech Kaczmarczyk <drowsygoat1982@gmail.com></drowsygoat1982@gmail.com>	
Description MatrixEQTL wrapper for QTL analysis, including functions for running the analysis, processing results, and visualizing QTL data.	
License GPL-3	
Encoding UTF-8	
Roxygen list(markdown = TRUE)	
RoxygenNote 7.3.2	
Imports SummarizedExperiment, ggplot2, Matrix, data.table, ggbeeswarm	
Suggests testthat (>= 3.0.0)	
Config/testthat/edition 3	
Contents	
annotate_with_rowdata_peaks	2
checkpoint . CleanIDs convert_chromosome_ids ExportFeatureLocationsForMatrixEQTL extract_genotypes_from_vcf_by_pos extract_snp_gene_pairs get_model_constant grep_o infer_sex_from_mosdepth	33 34 4 5 6 7 8 9 9
init_logging	

```
      matrixEQTLwrapper
      12

      nearest_gene_from_gtf
      13

      nearest_gene_from_gtf_build_tss
      13

      peak_range
      14

      PlotClusterCovariateSummary
      16

      plotPeakMatrixBoxplot
      17

      prepareMatrixEQTLInputs
      19

      read_chr_pos_from_vcf
      20

      run_qtl_workflow
      21

      test_matrixEQTL_run
      22

      validate_sample_overlap
      23

Index
```

annotate_with_rowdata_peaks

Annotate a tibble with peak coordinates from a SummarizedExperiment

Description

Left-joins df to rowData(se) by matching df[[key_col]] to rownames(se). Appends four columns to df: peak_id (the matched key), peak_chr (from seqnames), peak_start (from start), and peak_end (from end). The number and order of rows in df are preserved.

Usage

```
annotate_with_rowdata_peaks(
   df,
   se,
   key_col = "peak_id",
   seq_field = "seqnames",
   start_field = "start",
   end_field = "end",
   warn_unmatched = TRUE
)
```

Arguments

checkpoint 3

Value

df with added columns: peak_id, peak_chr, peak_start, peak_end.

Examples

```
## Not run:
# df has "peak_ID" that matches rownames(se_atac_full)
df2 <- annotate_with_rowdata_peaks(
    df = df,
    se = se_atac_full,
    key_col = "peak_ID" # becomes df2$peak_id
)
## End(Not run)</pre>
```

checkpoint

Debug checkpoint logger

Description

Debug checkpoint logger

Usage

```
checkpoint(msg, verbose = TRUE)
```

CleanIDs

Clean and Standardize Sample IDs

Description

Cleans sample IDs by optionally selecting parts of IDs, removing duplicate halves, and converting to uppercase or lowercase. Can operate directly on a character vector or on the header line of a tab-delimited file (saving a repaired version).

Usage

```
CleanIDs(input, Capitalize = TRUE, pattern = NULL)
```

Arguments

input A character vector of IDs, or a file path to a tab-delimited text file where the first row contains IDs (first column is assumed to be a non-ID column name).

Capitalize Logical; if TRUE, converts IDs to uppercase, otherwise to lowercase. Default is

TRUE.

pattern Optional integer vector specifying which underscore-separated parts of the ID

to keep (by position). If NULL (default), keeps all parts, but if the ID consists of

two identical halves, only keeps the first half.

Details

If a file path is provided, the function:

- 1. Reads the file.
- 2. Cleans the sample IDs in the header.
- 3. Writes a new file with _names_repaired.txt appended to the name.

When cleaning IDs:

- IDs are split on underscores.
- If pattern is specified, only the specified parts are kept.
- If pattern is NULL and the ID has two identical halves, only the first half is kept.
- Otherwise, all parts are concatenated.

Value

- If input is a vector: a cleaned character vector.
- If input is a file path: (invisibly) returns the path to the repaired file.

Examples

```
# Clean a vector of IDs
CleanIDs(c("S1_A", "S2_A"))

# Keep only the first two parts of IDs
CleanIDs(c("S1_part1_part2_extra", "S2_part1_part2_extra"), pattern = 1:2)

# Clean IDs in a file and save repaired file
## Not run:
CleanIDs("matrix_eqtl_input/SNP.txt")

## End(Not run)
```

convert_chromosome_ids

Convert chromosome names by adding or removing "chr" prefix

Description

This function converts chromosome identifiers by either adding the "chr" prefix (e.g., $1 \rightarrow chr1$) or removing it (e.g., $chr1 \rightarrow 1$). It also handles the mitochondrial chromosome (MT <-> chrM).

```
convert_chromosome_ids(chroms, direction = c("add", "remove"))
```

Arguments

chroms A character vector of chromosome names.

direction A string specifying the conversion direction: "add" to add the "chr" prefix, or

"remove" to remove it.

Value

A character vector of converted chromosome names.

Examples

```
convert_chromosome_ids(c("1", "2", "MT"), direction = "add")
convert_chromosome_ids(c("chr1", "chr2", "chrM"), direction = "remove")
```

 ${\tt ExportFeatureLocationsForMatrixEQTL}$

Export gene feature locations for Matrix eQTL analysis

Description

This function extracts and formats genomic feature location data from a GTF file for a specified set of genes, and writes it in a format compatible with Matrix eQTL. It supports gene identifiers from Seurat objects, SummarizedExperiments, or character vectors.

Usage

```
ExportFeatureLocationsForMatrixEQTL(
  gtf_file,
  genes,
  output_dir = ".",
  use_gene_name = TRUE,
  add_chr_prefix = FALSE,
  filename = "feature_locations.txt"
)
```

Arguments

gtf_file	Path to a GTF file containing gene annotations.
genes	$\label{thm:continuous} A \ Seurat \ object, \ a \ Summarized \ Experiment \ object, \ or \ a \ character \ vector \ of \ gene \ identifiers.$
output_dir	Directory where the output file will be saved. Defaults to the current directory.
use_gene_name	$Logical. \ If \ TRUE, \ match \ genes \ by \ gene_name \ first, \ falling \ back \ to \ gene_id. \ If \ FALSE, \ match \ only \ by \ gene_id.$
add_chr_prefix	Logical. If TRUE, adds "chr" prefix to chromosome names (e.g., 1 -> chr1).
filename	Name of the output file. Defaults to "feature_locations.txt".

Details

The function filters GTF entries to keep only those of type "gene", and retrieves their chromosome, start, and end coordinates. If gene identifiers are duplicated between gene_name and gene_id, matches by gene_name are prioritized.

Value

(Invisibly) the path to the output file containing the feature locations.

See Also

convert_chromosome_ids for chromosome name formatting

Examples

```
## Not run:
ExportFeatureLocationsForMatrixEQTL(
  gtf_file = "annotation.gtf",
  genes = c("GeneA", "GeneB", "GeneC"),
  output_dir = "eqtl_files",
  use_gene_name = TRUE,
  add_chr_prefix = TRUE
)
## End(Not run)
```

```
extract_genotypes_from_vcf_by_pos
```

Extract Genotypes from VCF by Chromosome and Position

Description

Extracts genotype data for specific SNP positions from a VCF file and formats genotypes as unphased allele pairs (e.g., A/G, C/C).

```
extract_genotypes_from_vcf_by_pos(
  vcf_path,
  snp_df,
  chr_col = "CHR",
  pos_col = "POS",
  Capitalize = TRUE,
  pattern = NULL
)
```

extract_snp_gene_pairs 7

Arguments

vcf_path	Path to the VCF file (can be gzipped or plain text).
snp_df	A data frame containing SNP positions to extract. Must include chromosome and position columns.
chr_col	Name of the column in snp_df that contains chromosome identifiers. Default is "CHR".
pos_col	Name of the column in snp_df that contains position values. Default is "POS".
Capitalize	Logical; whether to capitalize cleaned sample names. Default is TRUE.
pattern	Optional integer vector specifying which parts of sample names (split by "_") to retain. Used for sample name cleaning.

Details

This function:

- Extracts VCF lines corresponding to provided chromosome-position pairs.
- Parses genotype fields (GT) and maps 0/1 alleles to REF/ALT bases.
- Cleans sample names using the provided pattern or automatic heuristics.

SNPs are identified by combining chromosome, position, and ALT allele (CHR:POS_ALT). Lines without valid genotypes are returned as NA.

Value

A data frame of genotypes with SNP IDs as rows and cleaned sample names as columns. Genotypes are in unphased format (e.g., A/T, G/G).

Examples

```
## Not run:
snp_df <- data.frame(CHR = c("1", "2"), POS = c("12345", "67890"))
vcf_file <- "variants.vcf.gz"
genotypes <- extract_genotypes_from_vcf_by_pos(vcf_file, snp_df)
## End(Not run)</pre>
```

```
{\tt extract\_snp\_gene\_pairs}
```

Extract SNP-Gene Pairs with Chromosome and Position

Description

Extracts unique SNP-gene pairs from a data frame and parses SNP identifiers into chromosome and position columns.

8 get_model_constant

Usage

```
extract_snp_gene_pairs(df, snp_col = "snp_id", gene_col = "peak_id")
```

Arguments

df A data frame containing SNP and gene identifiers.

snp_col Name of the column in df containing SNP identifiers in the format "CHR: POS"

or "CHR: POS_ALT". Default is "snp_id".

gene_col Name of the column in df containing gene or peak identifiers. Default is "peak_id".

Details

This function:

- Renames the input columns to standardized names (SNP_ID, GENE_ID).
- Ensures uniqueness of SNP-gene pairs.
- Extracts chromosome and position information from the SNP ID.

Value

A data frame with columns:

- SNP_ID: The SNP identifier.
- GENE_ID: The gene or peak identifier.
- CHR: Chromosome parsed from SNP_ID.
- POS: Position parsed from SNP_ID as an integer.

Examples

```
df <- data.frame(
   snp_id = c("1:12345_A", "2:67890_T"),
   peak_id = c("geneA", "geneB")
)
extract_snp_gene_pairs(df)</pre>
```

get_model_constant

Get model constant from name

Description

Get model constant from name

```
get_model_constant(model)
```

grep_o

grep_o

Extract First Regex Match from String

Description

A safer wrapper around regexpr() + regmatches() to extract the first pattern match from each string.

Usage

```
grep_o(strings, pattern)
```

Arguments

strings Character vector to search in.

pattern Regular expression pattern to match.

Value

Character vector of matched substrings. If no match is found, returns NA.

```
infer_sex_from_mosdepth
```

Infer Sex (ZW system) from mosdepth summaries and optionally write outputs

Description

Reads all summary.txt files under a mosdepth output directory, extracts mean coverage for chromosomes Z and W, computes the Z:W coverage ratio per sample, and calls sex under the avian ZW system (female = ZW, male = ZZ).

```
infer_sex_from_mosdepth(
  mosdepth_dir,
  out_dir = ".",
  threshold = 1.5,
  chrom_labels = c("Z", "W"),
  verbose = TRUE
)
```

10 load_sliced_file

Arguments

mosdepth_dir Character. Path to a directory containing mosdepth outputs.

out_dir Character. Directory where output files (covariates_sex.txt and ZW_coverage_ratio.pdf) will be written. Default: current working directory.

threshold Numeric. Z:W ratio cutoff (default 1.5).

chrom_labels Character vector of length 2 or named list with "Z" and "W".

verbose Logical. Print messages (default TRUE).

Details

Infer biological sex from mosdepth Z and W chromosome coverage

Value

A list with:

- results: data.frame with per-sample Z, W, ratio, and sex call.
- covariates: one-row data.frame suitable for Matrix eQTL.
- files: character vector of summary files used.
- plot_file: path to saved PDF (if written).
- covariate_file: path to covariate file (if written).

Description

Initialize Logging

Usage

```
init_logging(logfile = NULL, verbose = TRUE)
```

Description

Load a SlicedData object from file

```
load_sliced_file(file, sliceSize = 2000, first_col_is_rownames = TRUE)
```

manhattan_plot_gg 11

manhattan_plot_gg

Manhattan Plot with Optional Region Highlighting

Description

Creates a Manhattan-style plot from a data frame using **ggplot2**, with optional highlighted genomic regions. Chromosome labels are naturally ordered (via gtools::mixedsort), basepair positions are auto-scaled to Mb when values look like basepairs, and a horizontal significance line can be added.

Usage

```
manhattan_plot_gg(
   df,
   chr_col = "chr",
   pos_col = "location",
   p_col = "FDR_recal",
   region_start_col = "RegionStart",
   region_end_col = "RegionEnd",
   sig.level = NA,
   show_regions = TRUE,
   region_alpha = 0.1,
   col = c("gray40", "gray60"),
   point_size = 1
)
```

Arguments

df	A data frame containing chromosome, position, and p-value columns.
chr_col	Column name (string) for chromosome identifiers. Default "Chromosome".
pos_col	Column name (string) for genomic position (bp or Mb). Default "PeakPosition".
p_col	Column name (string) for p-values. Default "PeakPvalue".
region_start_co	ol
	Column name (string) for region start (bp or Mb). Default "RegionStart".
region_end_col	Column name (string) for region end (bp or Mb). Default "RegionEnd".
sig.level	Numeric p-value threshold for a dashed horizontal line (e.g., 5e-8). Use NA (default) to omit the line.
show_regions	Logical; if TRUE (default) and both region columns are present in df, shaded rectangles are drawn over those regions.
region_alpha	Alpha for region shading (0-1). Default 0.1.
col	Two-color vector used to alternate chromosome point colors. Default $c("gray40", "gray60")$.
point_size	Point size for scatter layer. Default 1.

Details

If region columns are not present (or show_regions = FALSE), the plot is drawn without any high-lighted rectangles.

The function:

- 1. Strips a leading "chr" from chromosome labels and orders chromosomes using natural sort.
- 2. Converts positions to megabases if any positions exceed 1e6.
- 3. Computes cumulative genomic positions to lay chromosomes end-to-end.
- 4. Alternates point colors by chromosome.
- 5. Optionally shades regions if show_regions = TRUE and both region columns exist in df. If region columns are missing, the plot is drawn without regions.

Value

A ggplot object.

Examples

```
## Not run:
df <- data.frame(
   Chromosome = rep(paste0("chr", 1:3), each = 100),
   PeakPosition = rep(seq(1, 5e6, length.out = 100), 3),
   PeakPvalue = runif(300)
)
p <- manhattan_plot_gg(df, sig.level = 5e-8)
print(p)
## End(Not run)</pre>
```

 ${\tt matrixEQTLwrapper}$

Run MatrixEQTL

Description

Run MatrixEQTL

```
matrixEQTLwrapper(
  feature_locations_path,
  feature_data_path,
  snpFilePath,
  covFilePath,
  snpLocPath,
  group_name,
  resultsDir = getwd(),
```

```
cisDist = 1e+06,
pvOutputThreshold = 1e-05,
pvOutputThresholdCis = 1e-04,
useModel = "linear",
minPvByGeneSnp = TRUE,
noFDRsaveMemory = FALSE,
prefix = NULL,
pvalueHist = NULL,
verbose = TRUE,
dry_run = TRUE,
colNames_convention
)
```

Description

Nearest gene (by TSS) from a GTF for given chromosome/position(s) (patched to safely harmonize seqlevels)

Usage

```
nearest_gene_from_gtf(
  gtf_file,
  chr,
  pos,
  output = c("gene", "distance", "both"),
  tss_gr = NULL
)
```

```
nearest\_gene\_from\_gtf\_build\_tss\\ Build\ (and\ reuse)\ a\ TSS\ GRanges\ from\ a\ GTF\ (helper\ for\ speed)
```

Description

Build (and reuse) a TSS GRanges from a GTF (helper for speed)

```
nearest_gene_from_gtf_build_tss(gtf_file)
```

14 peak_range

Arguments

```
gtf_file Path to GTF.
```

Value

A GRanges of width-1 TSS with gene_name (and possibly gene_id) in mcols. Pass this as tss_gr= to nearest_gene_from_gtf() for repeated queries.

peak_range

Derive Significant QTL Ranges Around Local Peaks

Description

Iteratively identifies local peaks (smallest p-values) per chromosome and grows significant ranges around each peak using one of two strategies: *DistanceFromPeak* (bounded by absolute genomic distance from the peak) or *AdjacentPoints* (bounded by the largest allowed gap between adjacent significant positions). After collecting candidate ranges, overlapping/nearby ranges are merged (controlled by MINGAPSIZE).

Usage

```
peak_range(
  qtl_data,
  METHOD = "DistanceFromPeak",
  SIGTHRESHOLD = 0.01,
  SEARCHDISTANCE = 3e+06,
  LOCALPEAKTHRESHOLD = 0.01,
  MINGAPSIZE = 0
)
```

Arguments

qtl_data

A data frame/data.table/tibble with at least the columns:

chr_snp Chromosome identifier (character/factor).

location Genomic position (integer/numeric, increasing within chromosome).

FDR Adjusted p-value (numeric); smaller is more significant.

METHOD

Character scalar. Range-building strategy:

Additional columns are ignored.

- "DistanceFromPeak": Define the region as the farthest significant points within SEARCHDISTANCE bp upstream/downstream of the peak. Significance within the region is evaluated vs. a local dynamic threshold (PEAKVALUE / LOCALPEAKTHRESHOLD).
- "AdjacentPoints": Walk upstream and downstream from the peak, extending the region while adjacent significant points are no farther apart than SEARCHDISTANCE bp. Uses the same local dynamic threshold for significance.

peak_range 15

Default "DistanceFromPeak".

SIGTHRESHOLD Numeric in (0,1]. Global significance cutoff for peaks. Iterations stop when the

current best (minimum) p-value exceeds this threshold. Default 0.01.

 ${\tt SEARCHDISTANCE} \quad Numeric \ (bp). \ For \ "{\tt DistanceFromPeak}", the \ maximum \ absolute \ distance \ from \ an extraction of the property of the prope$

the peak position. For "AdjacentPoints", the maximum gap allowed between consecutive significant positions while extending upstream/downstream. De-

fault 3e6.

LOCALPEAKTHRESHOLD

Numeric > 0. The dynamic threshold factor relative to the local peak p-value; a value of 0.01 means points are considered locally significant if p <= PEAKVALUE / 0.01 (i.e., up to two orders of magnitude less significant than the peak). De-

fault 0.01.

MINGAPSIZE Non-negative numeric (bp). Adjacent/overlapping ranges with a gap \leq MINGAPSIZE

are merged. Default 0.

Details

The algorithm loops per chromosome: pick the minimum p-value (the peak); construct a candidate range around it using METHOD and local dynamic threshold; remove covered rows; repeat until no positions with p <= SIGTHRESHOLD remain. Finally, overlapping/nearby ranges are merged with tolerance MINGAPSIZE.

Value

A data.frame with columns:

Chromosome id.

PeakPosition Position of local peak.

PeakPvalue Local peak p-value.

RegionStart Start coordinate (inclusive).

RegionEnd End coordinate (inclusive).

NPeaks Number of peaks merged into the range.

NSignificant Count of positions with p <= SIGTHRESHOLD inside the range.

RegionLength Computed as RegionEnd - RegionStart + 1.

Assumptions

- Positions are on the same coordinate system and comparable within a chromosome.
- For reproducibility and performance, positions within each chromosome should be sorted ascending.
- qtl_data\$FDR contains the p-values to be used (adjusted). If you wish to use raw p-values, pass them in the same column or rename accordingly.

Examples

PlotClusterCovariateSummary

Plot Cluster-Sample Summary (Mean/Median of Non-Zero Values)

Description

Reads a serialized SummarizedExperiment (.rds), computes per **cluster x sample** summaries of a chosen assay (mean and median of non-zero entries), and saves a combined boxplot with jittered points.

Usage

```
PlotClusterCovariateSummary(
    se_path,
    save_dir,
    assay_name = NULL,
    filename = "cluster_summary_plot.pdf"
)
```

Arguments

se_path Character path to an .rds file containing a SummarizedExperiment.

save_dir Directory where the plot file will be written (created if it does not exist).

Optional character name of the assay to use. If NULL (default), the first assay in assayNames(se) is used.

Output filename for the saved plot (PDF/PNG depending on extension). Default: "cluster_summary_plot.pdf".

Details

The input SummarizedExperiment must contain at least two columns in colData(se):

- cluster: cluster identifier (coerced to character)
- sample_id: sample identifier (coerced to character)

For each cluster, samples present in that cluster are considered. For each cluster x sample subset, all *non-zero* entries of the assay are taken, and their mean and median are computed. These statistics are reshaped to long format and visualized as boxplots with quasi-random points on top.

If assay_name is NULL, the first assay is selected and a note is messaged. The plot is saved with ggplot2::ggsave() to file.path(save_dir, filename).

Value

Invisibly returns NULL. The function is used for its side effects (writing a plot).

Package Requirements

This function uses **SummarizedExperiment**, **Matrix**, **ggplot2**, **ggbeeswarm**, **dplyr**, and **tidyr**. The function checks for these packages at runtime.

Examples

```
## Not run:
PlotClusterCovariateSummary(
    se_path = "path/to/object.rds",
    save_dir = "results/plots",
    assay_name = NULL,
    filename = "cluster_summary_plot.pdf"
)
## End(Not run)
```

plotPeakMatrixBoxplot Plot PeakMatrix Boxplots per Sample

Description

Generates a per-sample boxplot of peak signals from a SummarizedExperiment object. Optionally subsets to the top N peaks per sample before plotting. Can save the plot to a PDF and optionally return the subsetted object.

```
plotPeakMatrixBoxplot(
    se,
    assay_name = "PeakMatrix",
    top_n_peaks = NULL,
    title = NULL,
    outfile = NULL,
    lim_y = 1
)
```

Arguments

se	A SummarizedExperiment containing the peak signal assay.
assay_name	Name of the assay to use. Default is "PeakMatrix".
top_n_peaks	Optional integer. If set, selects the top N peaks (by signal) per sample before plotting. The function will return the subsetted SummarizedExperiment invisibly in this case.
title	Optional plot title. If NULL, a default title is generated.
outfile	Optional file path. If provided, saves the plot as a high-resolution PDF.
lim_y	Numeric; upper y-axis limit for plotting. Default is 1.

Details

The function:

- 1. Validates that the input is a SummarizedExperiment with the specified assay.
- 2. Optionally subsets to the top N peaks per sample.
- 3. Converts the peak matrix to a long format (supports dense and sparse matrices).
- 4. Creates a combined boxplot + quasi-random scatter plot of peak signals.

Sparse matrices (dgCMatrix or dgTMatrix) are converted internally for plotting. Missing values are ignored by ggplot2.

Value

- If top_n_peaks is provided: the subsetted SummarizedExperiment (invisibly).
- If top_n_peaks is NULL: NULL (invisibly).

Examples

```
## Not run:
library(SummarizedExperiment)

# Example SE object with assay "PeakMatrix"
plotPeakMatrixBoxplot(se, top_n_peaks = 1000, outfile = "peaks.pdf")

## End(Not run)
```

```
prepareMatrixEQTLInputs
```

Prepare Matrix eQTL Input Files

Description

Prepares and filters expression, SNP, and covariate matrices for Matrix eQTL analysis. Supports SummarizedExperiment or raw matrices as input, optional GTF-based annotation, and automatic chunking for large datasets. Saves intermediate results as RDS files.

Usage

```
prepareMatrixEQTLInputs(
  verbose = TRUE,
  features,
  sampleMetadata = NULL,
  groupCol = NULL,
  sampleCol = "sample_id",
  snpPaths,
  covPaths,
 resultsDir,
 minFeatureFrac = 0.8,
 minFeatureMean = 0,
 matrixName = NULL,
 nChunks = 1,
  topNrows = NULL,
  topNSNPs = NULL,
  groupSubset = NULL,
  sliceSize = 2000,
 gtfFile = NULL,
  useGeneName = FALSE
)
```

Arguments

verbose	Logical; whether to print progress messages.
features	SummarizedExperiment object or expression matrix (genes x samples).
${\tt sampleMetadata}$	Optional data.frame with sample annotations (required if features is not a SE).
groupCol	Column in metadata indicating group assignment.
sampleCol	Column in metadata indicating sample identifiers (default: "sample_id").
snpPaths	Character vector of paths to SNP matrices or directories.
covPaths	Character vector of paths to covariate matrices or directories.
resultsDir	Directory to save all output files.
minFeatureFrac	Minimum fraction of samples a gene must be detected in (default: 0.8).

minFeatureMean Minimum mean expression for features to be retained (default: 0).

matrixName Name of assay to extract from SummarizedExperiment (optional).

nChunks Number of chunks to split SNP and expression matrices into (default: 5).

topNrows Optional limit on top expressed features to retain.
topNSNPs Optional limit on top SNPs by variance to retain.
groupSubset Optional vector of group names to include.

sliceSize Slice size for SlicedData export (default: 2000).

gtfFile Optional GTF file to retrieve genomic coordinates for features.

useGeneName Logical; whether to prioritize gene name over gene ID (default: FALSE).

Value

Invisible TRUE; writes RDS and text files to resultsDir.

read_chr_pos_from_vcf Read Chromosome and Position from a VCF File

Description

Extracts chromosome and position columns from a VCF file, excluding header lines.

Usage

```
read_chr_pos_from_vcf(vcf_path)
```

Arguments

vcf_path Path to the VCF file (can be gzipped or plain text).

Details

This function:

- Uses grep -v '^##' to skip metadata header lines.
- Reads only the first two columns from the VCF: chromosome and position.
- Renames them to chr_snp and location_snp.
- $\bullet \ \ Applies \ convert_chromosome_ids() \ function \ to \ standardize \ chromosome \ names.$

Value

A tibble with two columns:

- chr_snp: Chromosome identifiers, optionally converted using convert_chromosome_ids().
- location_snp: SNP positions as character strings.

run_qtl_workflow 21

Note

This function assumes that a helper function convert_chromosome_ids() is available in the environment.

Examples

```
## Not run:
vcf_file <- "variants.vcf.gz"
chr_pos_tbl <- read_chr_pos_from_vcf(vcf_file)
## End(Not run)</pre>
```

run_qtl_workflow

Run the QTL post-processing workflow (+ optional nearest-gene annotation)

Description

Run the QTL post-processing workflow (+ optional nearest-gene annotation)

Usage

```
run_qtl_workflow(
  results,
 vcf_file,
 apply_peak_range = TRUE,
 peak_params = list(METHOD = "DistanceFromPeak", SIGTHRESHOLD = 0.01, SEARCHDISTANCE =
    3e+06, LOCALPEAKTHRESHOLD = 0.01, MINGAPSIZE = 0),
  recalc_fdr = TRUE,
  annotate_peaks = FALSE,
  se = NULL,
  key_col = "peak_id",
 nearest_genes = FALSE,
 gtf_file = NULL,
 nearest_on = c("peak", "snp"),
 nearest_output = c("both", "gene", "distance"),
  tss_gr = NULL,
 output_dir = ".",
 prefix = "qtl",
 write_outputs = FALSE
)
```

Arguments

results

QTL results data.frame/tibble with columns: p_value, chr_snp, location_snp, peak_id, QTL_type (and optionally FDR).

22 test_matrixEQTL_run

vcf_file Path to VCF used to filter SNPs (via read_chr_pos_from_vcf).

apply_peak_range

Logical; run peak_range() per cis/trans and join back. Default TRUE.

peak_params List of params passed to peak_range(). Default: list(METHOD="DistanceFromPeak",

SIGTHRESHOLD=0.01, SEARCHDISTANCE=3e6, LOCALPEAKTHRESHOLD=0.01, MINGAPSIZE=0).

recalc_fdr Logical; if TRUE, add FDR_recal := p.adjust(p_value, "BH"). Default TRUE.

annotate_peaks Logical; if TRUE, call annotate_with_rowdata_peaks() on cis/trans. Default

FALSE.

se SummarizedExperiment for annotate_with_rowdata_peaks() (required if annotate_peaks=TRUE).

key_col Peak ID column in results for annotation. Default "peak_id".

nearest_genes Logical; if TRUE, annotate nearest gene by TSS from a GTF. Default FALSE. gtf_file Path to GTF (plain or .gz). Required if nearest_genes=TRUE and tss_gr is

NULL.

nearest_on Compute nearest gene relative to "peak" (columns chr, location) or "snp"

(chr_snp, location_snp). Default "peak".

nearest_output What to add: "gene", "distance", or "both". Default "both".

tss_gr Optional precomputed TSS GRanges (from nearest_gene_from_gtf_build_tss())

to speed up repeated calls.

output_dir Directory for outputs when write_outputs=TRUE. Default ".".

prefix Filename prefix for outputs. Default "qtl".
write_outputs Logical; write CSVs and PDFs. Default FALSE.

Value

List:

- filtered VCF-filtered results.
- peak_counts counts per QTL type.
- cis_ranges, trans_ranges outputs from peak_range() (if used).
- cis, trans final tables (with optional FDR recalculation, rowData & nearest gene annotations).
- plot_files written plot paths (if any).
- tss the TSS GRanges used (if nearest_genes=TRUE).

test_matrixEQTL_run Run test harness for MatrixEQTL

Description

Run test harness for MatrixEQTL

```
test_matrixEQTL_run()
```

```
validate_sample_overlap
```

Validate and reorder sample columns if needed, based on trimmed sample names Trims suffixes from sample names (e.g., removes '_C1_RNA') before checking

Description

Validate and reorder sample columns if needed, based on trimmed sample names Trims suffixes from sample names (e.g., removes '_C1_RNA') before checking

```
validate_sample_overlap(
  feature_data,
   snps,
   covs,
   group_name,
   colNames_convention,
   verbose = TRUE
)
```

Index

```
annotate_with_rowdata_peaks, 2
checkpoint, 3
CleanIDs, 3
convert_chromosome_ids, 4, 6
{\tt ExportFeatureLocationsForMatrixEQTL}, {\tt 5}
\verb|extract_genotypes_from_vcf_by_pos, 6|
extract_snp_gene_pairs, 7
get_model_constant, 8
grep_o, 9
infer_sex_from_mosdepth, 9
init_logging, 10
load_sliced_file, 10
manhattan_plot_gg, 11
matrixEQTLwrapper, 12
nearest_gene_from_gtf, 13
{\tt nearest\_gene\_from\_gtf\_build\_tss}, 13
peak_range, 14
PlotClusterCovariateSummary, 16
plotPeakMatrixBoxplot, 17
prepareMatrixEQTLInputs, 19
read_chr_pos_from_vcf, 20
run_qtl_workflow, 21
test_matrixEQTL_run, 22
validate_sample_overlap, 23
```