Package: rolypoly (via r-universe)

November 16, 2026

Type Package

Title Identifying Trait-Relevant Functional Annotations

Version 0.1.0.9001

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Description Using enrichment of genome-wide association summary statistics to identify trait-relevant cellular functional annotations. Compatibility updates by Renova.

Depends R (>= 3.1.3),

Imports data.table (>= 1.9.6), dplyr (>= 0.4.3), foreach (>= 1.4.3), ggplot2 (>= 1.0.1), glmnet (>= 2.0-5), MASS (>= 7.3-45), Matrix (>= 1.2-6),

Suggests CompQuadForm (>= 1.4.1), knitr, testthat, rmarkdown

License GPL-3

VignetteBuilder knitr

LazyData TRUE

URL https://github.com/dcalderon/rolypoly

BugReports https://github.com/dcalderon/rolypoly/issues

RoxygenNote 6.0.1

NeedsCompilation no

Repository https://renova-r.r-universe.dev

Date/Publication 2026-11-16 17:34:43 UTC

RemoteUrl https://github.com/renova-r/rolypoly

RemoteRef renova-0.1.0.9001

RemoteSha 908c2536a29cb2917382dd7ae3d3fc2dff86973a

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`bootstrap_estimator` *Bootstrap parameter estimates for confidence intervals.*

Description

Bootstrap parameter estimates for confidence intervals.

Usage

```
bootstrap_estimator(rolypoly, bootstrap_iters, run_light, run_parallel)
```

Arguments

rolypoly	rolypoly object
bootstrap_iters	number of bootstrap iterations to run
run_light	if we throw away bootstrap data
run_parallel	if we want to collect bootstrap data in parallel

Examples

```
## Not run: bootstrap_estimator(rolypoly)
```

---

calculate_annotation_block_heritability

Caclulate the contribution of block annotations to the heritability of a trait.

Description

A vector of indenpendent heritability contributions of block annotations is returned. Sum the vector to get total explained heritability and divide by sum to get proportion.

Usage

```
calculate_annotation_block_heritability(block_data, params)
```

Arguments

block_data	functional information of blocks
params	parameter fit

Examples

```
## Not run: calculate_annotation_block_heritability(block_data, params)
```

`calculate_block_values` *Caclulate predicted block values based on block information and model fit.*

Description

From a model fit we can predict expected variance of a block based on information we have about the block. In the example of gene expression this would equate to predicting the importance of a gene based on its signature of expression.

Usage

```
calculate_block_values(block_data, params)
```

Arguments

`block_data`	block_data
`params`	parameter fit

Value

returns block values

Examples

```
## Not run: calculate_gene_values(block_data, params)
```

`calculate_expected_block_values_given_ld` *Caclulate predicted block values based on block information and model fit.*

Description

From a model fit we can predict expected variance of a block based on information we have about the block. In the example of gene expression this would equate to predicting the importance of a gene based on its signature of expression.

Usage

```
calculate_expected_block_values_given_ld(rolypoly, block_values)
```

Arguments

rolypoly	rolypoly object
block_values	estimated block values.

Examples

```
## Not run: calculate_expected_block_values_given_ld(rolypoly, block_values)
```

---

cv_regularized_parameter_estimator *Perform regularization inference.*

Description

Use CV to find appropriate values of lambda for either feature selection or for prediction.

Usage

```
cv_regularized_parameter_estimator(vectorized_rolypoly_data, n_folds = 10,
  ...)
```

Arguments

vectorized_rolypoly_data	rolypoly data used for inference
n_folds	number of folds for cross validation
...	other arguments to pass to cv.glmnet

Value

results from cross validation

Examples

```
## Not run: cv_regularized_parameter_estimator(vectorized_rolypoly_data)
```

---

data_io *Functions for opening and organizing data.*

Description

We must open block annotation data, gwas data, snp annotations, gene annotations. Here, you'll find functions that to this and organize these data into a rolypoly object.

`inference` *Inference functions.*

Description

Functions for inferring relevant annotations using the polyTest model.

`main_wrapper` *Main wrapper functions.*

Description

The main endpoint user functions.

`make_ld_matrix` *Helper function to pull LD data from NCBI.*

Description

Given the path of a gwas file open it into a data.table object

Usage

```
make_ld_matrix(all_snps, ld_data)
```

Arguments

`all_snps`	The snps that were queried
`ld_data`	A returned LD matrix with SNP, Proxy, and RSquared columns

Value

an LD matrix where query snps will be the first columns in the correct order

Examples

```
## Not run: make_ld_matrix(all_snps, ld_data)
```

make_results_df *Helper function to make a summary table of results from bootstrap data.*

Description

Helper function to make a summary table of results from bootstrap data.

Usage

```
make_results_df(value_collection, annotations, model_estimates)
```

Arguments

value_collection	collection of bootstrapped value estimates
annotations	vector of annotation names
model_estimates	estimates for bias parameter estimates

Value

data frame with results summary

Examples

```
## Not run: make_results_df(value_collection)
```

parameter_estimator *Find parameter estimates for the data.*

Description

Find parameter estimates for the data.

Usage

```
parameter_estimator(vectorized_rolypoly_data)
```

Arguments

vectorized_rolypoly_data	rolypoly data that has been vectorized

Value

results of inference

Examples

```
## Not run: parameter_estimator(rolypoly)
```

`plot_rolypoly_annotation_estimates`
Visualize parameter estimates after running inference

Description

Visualize parameter estimates after running inference

Usage

```
plot_rolypoly_annotation_estimates(rolypoly)
```

Arguments

`rolypoly`	a rolypoly object

Value

ggplot2 object

Examples

```
## Not run: plot_rolypoly_annotation_estimates(rolypoly)
```

`plot_rolypoly_annotation_ranking`
Rank annotations by p-value after running inference

Description

Rank annotations by p-value after running inference

Usage

```
plot_rolypoly_annotation_ranking(rolypoly)
```

Arguments

`rolypoly`	a rolypoly object

Value

ggplot2 object

Examples

```
## Not run: plot_rolypoly_annotation_ranking(rolypoly)
```

`regularized_inference` *Inference functions that include regularization*

Description

Functions for inferring relevant annotations using the polyTest model.

`robust_parameter_estimator`
Find robust parameter estimates for the data.

Description

Find robust parameter estimates for the data.

Usage

```
robust_parameter_estimator(vectorized_rolypoly_data, outlier_threshold = 10)
```

Arguments

`vectorized_rolypoly_data`
: vectorized rolypoly data

`outlier_threshold`
: outlier threshold for robust inference

Examples

```
## Not run: parameter_estimator(rolypoly)
```

`rolypoly` *The rolypoly package for identifying annotations associated with complex traits.*

Description

The rolypoly package for identifying annotations associated with complex traits.

`rolypoly_add_ld_corrected_gwas_block_scores`
Add LD corrected block scores to rolypoly.

Description

Add LD corrected block scores to rolypoly.

Usage

```
rolypoly_add_ld_corrected_gwas_block_scores(rolypoly, fast_calculation = T)
```

Arguments

`rolypoly`	rolypoly data
`fast_calculation`	if F then LD deconvolution else quadratic form.

Value

rolypoly object with LD corrected gwas scores attached

Examples

```
## Not run: rolypoly_add_ld_corrected_gwas_block_scores(rolypoly)
```

`rolypoly_link_blocks_and_gwas`
Link blocks and gwas

Description

Takes block information, potentially independent LD blocks or gene blocks, and gwas data and organizes the data for interenal processing

Usage

```
rolypoly_link_blocks_and_gwas(rolypoly, ld_folder, r2_threshold = 0.2,
  run_parallel = F)
```

Arguments

`rolypoly`	a rolypoly object
`ld_folder`	path to a folder with ld data
`r2_threshold`	LD threshold to look at data
`run_parallel`	check if user wants to run in parallel

Value

rolypoly object with data attached

Examples

```
## Not run: rolypoly_link_blocks_and_gwas(rolypoly, ld_folder, r2_threshold)
```

`rolypoly_load_block_annotation`

Load annotations for blocks of LD, in some cases this is a gene annotation with a window around a gene.

Description

Load annotations for blocks of LD, in some cases this is a gene annotation with a window around a gene.

Usage

```
rolypoly_load_block_annotation(rolypoly, block_annotation, genes = T)
```

Arguments

`rolypoly`	rolypoly data object
`block_annotation`	annotation information for block
`genes`	if these are genes

Value

rolypoly data with block annotations attached

Examples

```
## Not run: rolypoly_load_block_annotation(rolypoly, block_annotation)
```

rolypoly_load_block_data

Block annotations, usually gene model.

Description

Block annotations, usually gene model.

Usage

```
rolypoly_load_block_data(rolypoly, block_data)
```

Arguments

rolypoly	a rolypoly object
block_data	a data frame of block information, usually gene expression. Requires rownames that are identitcal to block labels loaded previously.

Value

a rolypoly object with block information loaded

Examples

```
## Not run: rolypoly_load_block_data(rolypoly, block_data)
```

rolypoly_load_gwas *Load gwas data*

Description

Load gwas data

Usage

```
rolypoly_load_gwas(rolypoly, gwas_data, snp_annotations = NULL,
  gwas_z_filter = -1, add_spline = F, n_knots = 1, add_poly = F,
  n_degree = 2)
```

Arguments

rolypoly	rolypoly data
gwas_data	gwas data
snp_annotations	if there are additional snp annotations included
gwas_z_filter	if we want to remove large effect SNPs
add_spline	for fitting a spline to maf
n_knots	number of knots for spline
add_poly	for fitting a polynomial to maf
n_degree	degree of polynomial to fit

Value

rolypoly object with gwas data loaded

Examples

```
## Not run: rolypoly_load_gwas(rolypoly, gwas_data)
```

---

rolypoly_perform_inference *Run inference.*

Description

Coordinates running inference.

Usage

```
rolypoly_perform_inference(rolypoly, bootstrap_iters = 50,
  outlier_threshold = -1, run_light = F, run_parallel = F)
```

Arguments

rolypoly	rolypoly object
bootstrap_iters	number of bootstrap iterations to perform
outlier_threshold	threshold for performing robust regression, still experimental.
run_light	if we throw out bootstrap data
run_parallel	if we collect bootstraps in parallel

Value

rolypoly object with inference information attached

Examples

```
## Not run: run_inference(rolypoly)
```

`rolypoly_perform_regularized_inference` *Run inference with added regularization.*

Description

If p-values are desired use the other inference function. This for prediction purposes.

Usage

```
rolypoly_perform_regularized_inference(rolypoly, ...)
```

Arguments

`rolypoly`	a rolypoly object
`...`	other arguments to pass to cv.glmnet

Value

rolypoly object with regularization results

Examples

```
## Not run: rolypoly_perform_regularized_inference(rolypoly)
```

`rolypoly_plots` *Plot functions.*

Description

Functions for rolypoly results.

`rolypoly_roll` *Main rolypoly wrapper function.*

Description

The entry point for rolypoly analysis. If no expression data, we assume that we are running just the vegas score processing.

Usage

```
rolypoly_roll(rolypoly = NULL, gwas_data = NULL, block_annotation = NULL,
  block_data = NULL, ld_folder = NULL, bootstrap_iters = 50,
  outlier_threshold = -1, perform_cv = F, n_folds = 10,
  gwas_z_filter = -1, add_spline = F, n_knots = 1, add_poly = F,
  n_degree = 2, run_light = T, gwas_link_parallel = F,
  bootstrap_parallel = F, keep_model = F, keep_gwas = F, ...)
```

Arguments

`rolypoly`	Previous rolypoly run to parts of pipeline.
`gwas_data`	Gwas data for a trait, including snp annotations.
`block_annotation`	Start and end points for blocks
`block_data`	Information about blocks.
`ld_folder`	Folder with LD information.
`bootstrap_iters`	Number bootstrap iterations to perform for inference.
`outlier_threshold`	Set to positive if we want to run robusted regression.
`perform_cv`	If we want to interpret annotation effects do not set this to T. However, if our goal is prediction accuracy then set this to T.
`n_folds`	number of folds for cross validation
`gwas_z_filter`	Z-score filter for SNPs, helps prevent large effects biasing inference.
`add_spline`	If we want to fit a spline to maf.
`n_knots`	number of knots to add to the spline.
`add_poly`	If we want to fit a polynomial to maf.
`n_degree`	the degree of the polynomial.
`run_light`	if we want to throw away bootstrap data, and save memory
`gwas_link_parallel`	if user wants to run in gwas linking in parallel, registerDoParallel must have been run in advance.

bootstrap_parallel	if user wants to run in bootstraps in parallel, registerDoParallel must have been run in advance.
keep_model	if we should keep the regression model, can be large.
keep_gwas	set to T if we want to include gwas in returned rolypoly object.
...	other arguments to pass to cv.glmnet

Value

rolypoly object

Examples

```
## Not run: rolypoly_roll(rolypoly)
```

---

sim_block_annotation *Simulated block data annotation.*

Description

A dataset containing simulated block data annotation for use in the rolypoly vignette.

Usage

```
sim_block_annotation
```

Format

A data frame with 1000 rows and 4 variables:

chrom chromosome, we only use autosomes

start base pair position of variant

end rsid identifier of variant

label effect size, univariate regression coefficient

Source

I generated these fields to link with SNP positions

sim_expression_data_normalized *Simulated expression data.*

Description

A dataset containing simulated expression data for use in the rolypoly vignette.

Usage

```
sim_expression_data_normalized
```

Format

A data frame with 1000 rows and 5 variables:

Liver simulated expression data for tissue. This tissue has a significant effect on the simulated gwas.

Adrenal.Gland simulated expression data for tissue

Blood simulated expression data for tissue. This tissue has a significant effect on the simulated gwas.

Heart simulated expression data for tissue

Lung simulated expression data for tissue

Source

I generated this dataset

sim_gwas_data *Simulated GWAS summary statistics*

Description

A dataset containing simulated genome-wide association summary statistics for use in the rolypoly vignette.

Usage

```
sim_gwas_data
```

Format

A data frame with 14934 rows and 6 variables:

chrom chromosome, we only use autosomes

pos base pair position of variant

rsid rsid identifier of variant

beta effect size, univariate regression coefficient

se standard error of effect size

maf minor allele frequency

Source

rsids were from 1000g and I generated the other fields

`vectorize_rolypoly` *Take a list of rolypoly data and vectorize it for inference.*

Description

Take a list of rolypoly data and vectorize it for inference.

Usage

```
vectorize_rolypoly(data)
```

Arguments

`data`	the list of block information from rolypoly object

Value

list of necessary information for inference

Examples

```
## Not run: vectorize_rolypoly(data)
```

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