

Package ‘CASMAP’

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Type Package

Title Detection of Statistically Significant Combinations of SNPs in Association Mapping

Version 0.6.1

Description A significant pattern mining-based toolbox for region-based genome-wide association studies and higher-order epistasis analyses, implementing the methods described in Llinares-López et al. (2017) <doi:10.1093/bioinformatics/btx071>.

Depends R (>= 3.0.2)

Imports methods, Rcpp

LinkingTo Rcpp

Encoding UTF-8

LazyData true

License GPL (>= 2)

NeedsCompilation yes

RoxygenNote 6.0.1

SystemRequirements C++11

Suggests testthat, knitr, rmarkdown

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R topics documented:

CASMAP-class	2
getExampleCovariatesFilename	4
getExampleDataFilename	5
getExampleLabelsFilename	5
Index	7

CASMAP-class	<i>Constructor for CASMAP class object.</i>
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Description

Constructor for CASMAP class object.

Details

Constructor for CASMAP class object, which needs the mode parameter to be set by the user. Please see the examples.

Fields

mode Either 'regionGWAS' or 'higherOrderEpistasis'.

alpha A numeric value setting the Family-wise Error Rate (FWER). Must be strictly between 0 and 1. Default value is 0.05.

max_comb_size A numeric specifying the maximum length of combinations. For example, if set to 4, then only combinations of size between 1 and 4 (inclusive) will be considered. To consider combinations of arbitrary (maximal) length, use value 0, which is the default value.

Base method, for both modes

readFiles Read the data, label and possibly covariates files. Parameters are genotype_file, for the data, phenotype_file for the labels and (optional) covariates_file for the covariates. The option plink_file_root is not supported in the current version, but will be supported in future versions.

setMode Can set/change the mode, but note that any data files will need to read in again using the readFiles command.

setTargetFWER Can set/change the Family-wise Error Rate (FWER). Takes a numeric parameter alpha, strictly between 0 and 1.

execute Once the data files have been read, can execute the algorithm. Please note that, depending on the size of the data files, this could take a long time.

getSummary Returns a data frame with a summary of the results from the execution, but not any significant regions/itemsets. See getSignificantRegions, getSignificantInteractions, and getSignificantClusterRepresentatives.

writeSummary Directly write the information from getSummary to file.

regionGWAS Methods

`getSignificantRegions` Returns a data frame with the the significant regions. Only valid when `mode='regionGWAS'`.

`getSignificantClusterRepresentatives` Returns a data frame with the the representatives of the significant clusters. This will be a subset of the regions returned from `getSignificantRegions`. Only valid when `mode='regionGWAS'`.

`writeSignificantRegions` Writes the data from `getSignificantRegions` to file, which must be specified in the parameter path. Only valid when `mode='regionGWAS'`.

`writeSignificantClusterRepresentatives` Writes the data from `getSignificantClusterRepresentatives` to file, which must be specified in the parameter path. Only valid when `mode='regionGWAS'`.

higherOrderEpistasis Methods

`getSignificantInteractions` Returns the frame from `getSignificantInteractions` to file, which must be specified in the parameter path. Only valid when `mode='higherOrderEpistasis'`.

`writeSignificantInteractions` Writes a data frame with the significant interactions. Only valid when `mode='higherOrderEpistasis'`.

References

A. Terada, M. Okada-Hatakeyama, K. Tsuda and J. Sese *Statistical significance of combinatorial regulations*, Proceedings of the National Academy of Sciences (2013) 110 (32): 12996-13001

F. Llinares-Lopez, D. G. Grimm, D. Bodenham, U. Gieraths, M. Sugiyama, B. Rowan and K. Borgwardt, *Genome-wide detection of intervals of genetic heterogeneity associated with complex traits*, ISMB 2015, Bioinformatics (2015) 31 (12): i240-i249

L. Papaxanthos, F. Llinares-Lopez, D. Bodenham, K. Borgwardt, *Finding significant combinations of features in the presence of categorical covariates*, Advances in Neural Information Processing Systems 29 (NIPS 2016), 2271-2279.

F. Llinares-Lopez, L. Papaxanthos, D. Bodenham, D. Roqueiro and K. Borgwardt, *Genome-wide genetic heterogeneity discovery with categorical covariates*. Bioinformatics 2017, 33 (12): 1820-1828.

Examples

```
## An example using the "regionGWAS" mode
fastcmh <- CASMAP(mode="regionGWAS")      # initialise object

datafile <- getExampleDataFilename()      # file name of example data
labelsfile <- getExampleLabelsFilename()  # file name of example labels
covfile <- getExampleCovariatesFilename() # file name of example covariates

# read the data, labels and covariate files
fastcmh$readFiles(genotype_file=getExampleDataFilename(),
                  phenotype_file=getExampleLabelsFilename(),
                  covariate_file=getExampleCovariatesFilename() )

# execute the algorithm (this may take some time)
```

```

fastcmh$execute()

#get the summary results
summary_results <- fastcmh$getSummary()

#get the significant regions
sig_regions <- fastcmh$getSignificantRegions()

#get the clustered representatives for the significant regions
sig_cluster_rep <- fastcmh$getSignificantClusterRepresentatives()

## Another example of regionGWAS
fais <- CASMAP(mode="regionGWAS")      # initialise object

# read the data and labels, but no covariates
fastcmh$readFiles(genotype_file=getExampleDataFilename(),
                  phenotype_file=getExampleLabelsFilename())

## Another example, doing higher order epistasis search
facs <- CASMAP(mode="higherOrderEpistasis")      # initialise object

```

```
getExampleCovariatesFilename
```

Get the path to the example covariates file for regionGWAS mode

Description

Path to CASMAP_example_covariates_1.txt in inst/extdata. The covariates categories for the data set CASMAP_example_data_1.txt, the path to which is given by getExampleDataFilename.

Usage

```
getExampleCovariatesFilename()
```

Format

A single column vector of 100 labels, each of which is 0 or 1 (same format as labels file).

Details

Path to the file containing the labels, for reading in to CASMAP object using the readFiles function.

See Also

getExampleDataFilename, getExampleLabelsFilename

Examples

```
covfile <- getExampleCovariatesFilename()
```

getExampleDataFilename

Get the path to the example data file for regionGWAS mode

Description

Path to CASMAP_example_data_1.txt in inst/extdata. A dataset containing binary samples for the regionGWAS method. There are accompanying labels and covariates dataset.

Usage

```
getExampleDataFilename()
```

Format

A matrix of 0s and 1s, with 1000 rows (features) and 100 columns (samples). In other words, each column is a sample, and each sample has 1000 binary features.

Details

Path to the file containing the data, for reading in to CASMAP object using the readFiles function. Note that the significant region is [99, 102].

See Also

getExampleLabelsFilename, getExampleCovariatesFilename

Examples

```
datafile <- getExampleDataFilename()
```

getExampleLabelsFilename

Get the path to the example labels file for regionGWAS mode

Description

Path to CASMAP_example_labels_1.txt in inst/extdata. A dataset containing the binary labels for the data in the file CASMAP_example_data_1.txt, the path to which is given by getExampleDataFilename.

Usage

```
getExampleLabelsFilename()
```

Format

A single column of 100 labels, each of which is either 0 or 1.

Details

Path to the file containing the labels, for reading in to CASMAP object using the `readFiles` function.

See Also

`getExampleDataFilename`, `getExampleCovariatesFilename`

Examples

```
labelsfile <- getExampleLabelsFilename()
```

Index

CASMAP (CASMAP-class), [2](#)
CASMAP-class, [2](#)

getExampleCovariatesFilename, [4](#)
getExampleDataFilename, [5](#)
getExampleLabelsFilename, [5](#)