Package ‘traseR’

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Type Package

Title GWAS trait-associated SNP enrichment analyses in genomic intervals

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Author Li Chen, Zhaohui S. Qin

Maintainer li.chen@emory.edu

Description traseR performs GWAS trait-associated SNP enrichment analyses in genomic intervals using different hypothesis testing approaches, also provides various functionalities to explore and visualize the results.

License GPL

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biocViews Genetics, Sequencing, Coverage, Alignment, QualityControl, DataImport

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traseR-package  

GWAS trait-associated SNP enrichment analyses in genomic intervals

Description

Perform GWAS trait-associated SNP enrichment analyses in genomic intervals. Explore and visualize the results.

Details

Package: traseR  
Type: Package  
Version: 1.0  
Date: 2015-11-18  
License: GPL

Author(s)

Li Chen <li.chen@emory.edu>, Zhaohui S.Qin<zhaohui.qin@emory.edu>

CEU

Sampled SNPs from all SNPs of CEU population in 1000 genome project

Description

A GRange object CEU contains 5% of all SNPs from CEU by controlling genome-wide density is the same as all SNPs from CEU

Usage

data(CEU)

Value

The data frame CEU contains three columns,

<table>
<thead>
<tr>
<th>Column</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>SNP_ID</td>
<td>SNP rs number</td>
</tr>
<tr>
<td>seqnames</td>
<td>Chromosome number associated with rs number</td>
</tr>
<tr>
<td>ranges</td>
<td>Chromosomal position, in base pairs, associated with rs number</td>
</tr>
</tbody>
</table>

Author(s)

Li Chen <li.chen@emory.edu>, Zhaohui S.Qin<zhaohui.qin@emory.edu>
Visualize of trait-associated SNPs

Description
These are a group of functions to generate plot to visualize the trait-associated SNPs.

Usage

```r
plotContext(snpdb, region=NULL, keyword = NULL, pvalue = 1e-3)
plotPvalue(snpdb, region=NULL, keyword = NULL, plot.type = c("densityplot", "boxplot"), pvalue = 1e-3)
plotSNP(snpdb, snpid, ext = 10000)
plotGene(snpdb, gene, ext = 10000)
plotInterval(snpdb, interval, ext = 10000)
```

Arguments

- `snpdb`: A GRrange object or data frame, which is GWAS trait-associated SNPs downloaded from up-to-date dbGaP and NHGRI public database. It is maintained to be updated to the latest version. The data frame contains the following columns, Trait, SNP, p.value, Chr, Position, Context, GENE_NAME, GENE_START, GENE_END, GENE_STRAND. The data frame is in data subdirectory. Users are free to add more SNP records to the data frame for practical use.
- `region`: A data frame, which is genomic intervals with three columns, chromosome, genomic start position, genomic end position.
- `keyword`: The keyword is used when specific trait is of interest. If `keyword` is specified, only the SNPs associated to the trait are used for analyses. Otherwise, all traits will be analyzed.
- `snpid`: SNP rs number
- `gene`: Gene name
- `pvalue`: SNPs with p-value less than this threshold are used for analyses.
- `plot.type`: Either "densityplot" or "boxplot"
- `ext`: Bp extended upstream and downstream
- `xymax`: The maximum range on x-axis and y-axis
- `interval`: A data frame, genomic interval:chromosome, genomic start position, genomic end position

Value

- `plotContext`: A pie plot with the distribution of SNP function class
- `plotPvalue`: A density plot of -logPvalue of trait-associated SNPs
- `plotSNP`: A plot of trait-associated SNP on chromosome
print.traseR

plotGene A plot with the gene and possible nearby trait-associated SNPs
plotInterval A plot with chromosome interval with possible nearby genes and trait-associated SNPs

Author(s)
Li Chen <li.chen@emory.edu>, Zhaohui Qin<zhaohui.qin@emory.edu>

Examples
data(taSNP)
plotContext(snpdb= taSNP,keyword="Autoimmune")
plotGene(snpdb= taSNP,gene="ZFP92",ext=50000)
plotSNP(snpdb= taSNP,snpid="rs766420",ext=50000)
plotInterval(snpdb= taSNP,data.frame(chr="chrX",start=152633780,end=152737085))

print.traseR Print the outcome of taSNP enrichment analyses

Description
Print the outcome of taSNP enrichment analyses. Print the overall taSNP enrichment, trait-specific taSNP enrichment, trait-class-specific taSNP enrichment.

Usage
## S3 method for class 'traseR'
print(x,isTopK1=FALSE,topK1=10,isTopK2=FALSE,topK2=10,trait.threshold=10,traitclass.threshold=10,...)

Arguments

x Object returned from traseR
isTopK1 If isTopK1 is TRUE, topK1 traits are printed; otherwise, traits with p-value below Bonferroni correction threshold are printed. Default is FALSE.
topK1 Top K1 traits are printed. Default is 10.
isTopK2 If isTopK2 is TRUE, topK2 trait class are printed; otherwise, trait class with p-value below Bonferroni correction threshold are printed. Default is FALSE.
topK2 Top K2 trait class are printed. Default is 10.
trait.threshold Traits above this threshold are reported. Default is 10.
traitclass.threshold Trait class above this threshold are reported. Default is 10.
... Other parameters to print

Value
Print a data frame of traits ranked by p-value
**Description**

These are a group of functions to retrieve the trait-associated SNPs based on input.

**Usage**

```r
queryKeyword(snpdb, region=NULL, keyword = NULL, returnby = c("SNP_ID", "trait"), pvalue = 1e-3)
queryGene(snpdb, genes = NULL)
querySNP(snpdb, snpid, region = NULL)
```

**Arguments**

- **snpdb**: A GRanges object or data frame, which is GWAS trait-associated SNPs downloaded from up-to-date dbGaP and NHGRI public database. It is maintained to be updated to the latest version. The data frame contains the following columns: Trait, SNP_ID, p.value, Chr, Position, Context, GENE_NAME, GENE_START, GENE_END, GENE_STRAND. The data frame is in data subdirectory. Users are free to add more SNP records to the data frame for practical use.

- **region**: A data frame, which is genomic intervals with three columns, chromosome, genomic start position, genomic end position.

- **keyword**: The keyword is used when specific trait is of interest. If keyword is specified, only the SNPs associated to the trait are used for analyses. Otherwise, all traits will be analyzed.

- **snpid**: SNP rs number

- **genes**: Gene name

- **pvalue**: SNPs with p-value less than this threshold are used for analyses.

- **returnby**: Either SNP or trait. If returnby is specified as ’SNP_ID’, a data frame based on ’SNP_ID’ is returned. If returnby is specified as ’trait’, a data frame based on ’trait’ is returned.
Value

queryKeyword: Return a data frame of traits containing the keyword
queryGene: Return a data frame of traits associated with the gene
querySNP: Return a data frame of traits associated with the SNP

Author(s)

Li Chen <li.chen@emory.edu>, Zhaohui Qin<zhaohui.qin@emory.edu>

Examples

data(taSNP)
data(Tcell)
x=queryKeyword(snpdb=taSNP,region=Tcell,keyword="Autoimmune",returnby="SNP_ID")
x=queryGene(snpdb=taSNP,genes=c("AGRN","UBE2J2","SSU72"))
x=querySNP(snpdb=taSNP,snpid=c("rs3766178","rs880051"))

Description

A GR range object taSNP contains trait-associated SNPs from dbGaP and NHGRI downloaded from Association Results Browser.

Usage

data(taSNP)

Value

The data frame taSNP contains the following columns:

<table>
<thead>
<tr>
<th>Column</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Trait</td>
<td>Trait</td>
</tr>
<tr>
<td>Trait Class</td>
<td>Trait class which is formed based on the phenotype tree. Close traits are grouped together to form one class</td>
</tr>
<tr>
<td>SNP_ID</td>
<td>SNP rs number</td>
</tr>
<tr>
<td>p.value</td>
<td>GWAS SNP p-value</td>
</tr>
<tr>
<td>seqnames</td>
<td>Chromosome</td>
</tr>
<tr>
<td>ranges</td>
<td>Chromosome position</td>
</tr>
<tr>
<td>Context</td>
<td>SNP functional class</td>
</tr>
<tr>
<td>GENE_NAME</td>
<td>Nearest gene name</td>
</tr>
<tr>
<td>GENE_START</td>
<td>Gene start genomic position</td>
</tr>
<tr>
<td>GENE_END</td>
<td>Gene end genomic position</td>
</tr>
<tr>
<td>GENE_STRAND</td>
<td>Gene strand</td>
</tr>
</tbody>
</table>

Author(s)

Li Chen <li.chen@emory.edu>, Zhaohui Qin<zhaohui.qin@emory.edu>
### taSNPLD

**linkage disequilibrium (>0.8) within 100kb SNPs of all trait-associated SNPs from dbGaP and NHGRI**

**Description**

A GRange object `taSNPLD` contains linkage disequilibrium (>0.8) SNPs of all trait-associated SNPs from dbGaP and NHGRI.

**Usage**

```r
data(taSNPLD)
```

**Value**

The data frame `taSNPLD` contains four columns,

<table>
<thead>
<tr>
<th>SNP.ID</th>
<th>SNP rs number</th>
</tr>
</thead>
<tbody>
<tr>
<td>seqnames</td>
<td>Chromosome number associated with rs number</td>
</tr>
<tr>
<td>ranges</td>
<td>Chromosomal position, in base pairs, associated with rs number</td>
</tr>
<tr>
<td>Trait</td>
<td>Trait the SNP is associated with</td>
</tr>
<tr>
<td>Trait Class</td>
<td>Trait class which is formed based on the phenotype tree. Close traits are grouped together to form one class</td>
</tr>
</tbody>
</table>

**Author(s)**

Li Chen <li.chen@emory.edu>, Zhaohui S.Qin<zhaohui.qin@emory.edu>

### Tcell

**Peak regions of H3K4me1 in Peripheral blood T cell**

**Description**

A GRange object `Tcell` contains three columns: chromosome, genomic start position and genomic end position.

**Usage**

```r
data(Tcell)
```

**Value**

The data frame `Tcell` contains three columns,

<table>
<thead>
<tr>
<th>seqnames</th>
<th>Chromosome id</th>
</tr>
</thead>
<tbody>
<tr>
<td>ranges</td>
<td>Chromosome position</td>
</tr>
</tbody>
</table>

**Author(s)**

Li Chen <li.chen@emory.edu>, Zhaohui S.Qin<zhaohui.qin@emory.edu>
traseR

TRait-Associated SNP EnRichment analyses

Description

Perform GWAS trait-associated SNP enrichment analyses in genomic intervals using different approaches.

Usage

\[
\text{traseR(snpdb, region, snpdb.bg=NULL, keyword = NULL, rankby = c("pvalue", "odds.ratio"),}
\]

\[
\text{test.method = c("binomial", "fisher", "chisq", "nonparametric"), alternative = c("greater", "less",}
\]

\[
\text{ntimes=100,nbatch=1,}
\]

\[
\text{trait.threshold = 0, traitclass.threshold=0, pvalue = 1e-3)}
\]

Arguments

**snpdb**
A GRRange object. It could be GWAS trait-associated SNPs downloaded from up-to-date dbGaP and NHGRI public database. It is maintained to be updated to the latest version. The data frame contains the following columns, Source, Trait, SNP, p.value, Chr, Position. The data frame is in data subdirectory. Users are free to add more SNP records to the data frame for practical use. It could also be a data frame with columns as, SNP, Chr, Position.

**region**
A GRRange object or data frame, which is genomic intervals with three columns, chromosome, genomic start position, genomic end position.

**snpdb.bg**
A GRRange object contains non-trait-associated SNPs. They are treated as background for statistical testing instead of whole genome as background if specified.

**keyword**
The keyword is used when specific trait is of interest. If keyword is specified, only the SNPs associated to the trait are used for analyses. Otherwise, all traits will be analyzed.

**rankby**
Traits could be ranked by either p-value or odds.ratio based on the enrichment level of trait-associated SNPs in genomic intervals.

**test.method**
Several hypothesis testing options are provided: binomial(binomial test), fisher(Fisher’s exact test), chi sq(Chi-squared test), chi sq(nonparametric test). Default is binomial(binomial test).

**alternative**
Indicate the alternative hypothesis. If greater, test if the genomic intervals are enriched in trait-associated SNPs than background. If less, test if the genomic intervals are depleted in trait-associated SNPs than background. If two.sided, test if there is difference between the enrichment of trait-associated SNPs in genomic intervals and in background.

**ntimes**
The number of shuffling time for one batch. See nbatch.

**nbatch**
The number of batches. The product of ntimes and nbatch is the total number of shuffling time.

**trait.threshold**
Test traits with number of SNPs more than the threshold.

**traitclass.threshold**
Test trait class with number of SNPs more than the threshold.

**pvalue**
SNPs with p-value less than this threshold are used for analyses.
Details

Return a list that contains three data frames. One data frame `tb.all` contains the results of enrichment analyses for all trait-associated SNPs in genomic intervals. Another data frame `tb1` contains the results of enrichment analyses for each trait-associated SNPs in genomic intervals separately. Another data frame `tb2` contains the results of enrichment analyses for each trait-class-associated SNPs in genomic intervals separately.

Value

The data frame `tb1` has columns,

<table>
<thead>
<tr>
<th>Trait</th>
<th>Name of trait</th>
</tr>
</thead>
<tbody>
<tr>
<td>p.value</td>
<td>P-value calculated from hypothesis testing</td>
</tr>
<tr>
<td>q.value</td>
<td>Adjusted p-value from multiple testing using FDR correction</td>
</tr>
<tr>
<td>odds.ratio</td>
<td>Odds ratio calculated based on number of trait-associated SNPs in genomic intervals, number of trait-associated SNPs across whole genome, genomic intervals size (bps) and genome size (bps)</td>
</tr>
<tr>
<td>taSNP.hits</td>
<td>Number of trait-associated SNPs in genomic intervals</td>
</tr>
<tr>
<td>taSNP.num</td>
<td>Number of SNPs for specific trait</td>
</tr>
</tbody>
</table>

Author(s)

Li Chen <li.chen@emory.edu>, Zhaohui S.Qin<zhaohui.qin@emory.edu>

See Also

`print.traseR`

Examples

data(taSNP)
data(Tcell)
x=traseR(snpdb=taSNP,region=Tcell)
print(x)
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