

Package ‘traseR’

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Type Package

Title GWAS trait-associated SNP enrichment analyses in genomic intervals

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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

LazyLoad yes

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

NeedsCompilation no

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|----------------|---|
| traseR-package | <i>GWAS trait-associated SNP enrichment analyses in genomic intervals</i> |
|----------------|---|

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)

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| | |
|-----|--|
| CEU | <i>Sampled SNPs from all SNPs of CEU population in 1000 genome project</i> |
|-----|--|

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,

| | |
|----------|--|
| SNP_ID | SNP rs number |
| seqnames | Chromosome number associated with rs number |
| ranges | Chromosomal position, in base pairs, associated with rs number |

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| | |
|-------|---|
| plots | <i>Visualize of trait-associated SNPs</i> |
|-------|---|

Description

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

Usage

```
plotContext(snpdb, region=NULL, keyword = NULL, pvalue = 1e-3)
```

```
plotPvalue(snpdb, region=NULL, keyword = NULL, plot.type = c("densityplot", "boxplot"), pvalue = 1e-
```

```
plotSNP(snpdb, snpid, ext = 10000)
```

```
plotGene(snpdb, gene, ext = 10000)
```

```
plotInterval(snpdb, interval, ext = 10000)
```

Arguments

| | |
|-----------|--|
| snpdb | A GRange 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 |

| | |
|--------------|--|
| 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 |

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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 | <i>Print the outcome of taSNP enrichment analyses</i> |
|--------------|---|

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

Author(s)

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Examples

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

| | |
|--------|---|
| querys | <i>Retrieve trait-associated SNPs based</i> |
|--------|---|

Description

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

Usage

```
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 GRange 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_S... |
| 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)

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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"))
```

| | |
|-------|---|
| taSNP | <i>trait-associated SNPs in dbGaP and NHGRI downloaded from Association Results Browser</i> |
|-------|---|

Description

A GRange 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

| | |
|-------------|--|
| Trait | Trait |
| Trait Class | Trait class which is formed based on the phenotype tree. Close traits are grouped together to form one class |
| SNP_ID | SNP rs number |
| p.value | GWAS SNP p-value |
| seqnames | Chromosome |
| ranges | Chromosome position |
| Context | SNP functional class |
| GENE_NAME | Nearest gene name |
| GENE_START | Gene start genomic position |
| GENE_END | Gene end genomic position |
| GENE_STRAND | Gene strand |

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| | |
|---------|---|
| taSNPLD | <i>linkage disequilibrium (>0.8) within 100kb SNPs of all trait-associated SNPs from dbGaP and NHGRI</i> |
|---------|---|

Description

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

Usage

```
data(taSNPLD)
```

Value

The data frame taSNPLD contains four columns,

| | |
|-------------|--|
| SNP_ID | SNP rs number |
| seqnames | Chromosome number associated with rs number |
| ranges | Chromosomal position, in base pairs, associated with rs number |
| Trait | Trait the SNP is associated with |
| Trait Class | Trait class which is formed based on the phenotype tree. Close traits are grouped together to form one class |

Author(s)

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| | |
|-------|---|
| Tcell | <i>Peak regions of H3K4me1 in Peripheral blood T cell</i> |
|-------|---|

Description

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

Usage

```
data(Tcell)
```

Value

The data frame Tcell contains three columns,

| | |
|----------|---------------------|
| seqnames | Chromosome id |
| ranges | Chromosome position |

Author(s)

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traseR

*TRait-Associated SNP EnRichment analyses***Description**

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

Usage

```
traseR(snpdb, region, snpdb.bg=NULL, keyword = NULL, rankby = c("pvalue", "odds.ratio"),
test.method = c("binomial", "fisher", "chisq", "nonparametric"), alternative = c("greater", "less",
ntimes=100, nbatch=1,
trait.threshold = 0, traitclass.threshold=0, pvalue = 1e-3)
```

Arguments

| | |
|----------------------|---|
| snpdb | A GRange 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 GRange object or data frame, which is genomic intervals with three columns, chromosome, genomic start position, genomic end position. |
| snpdb.bg | A GRange 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), chisq(Chi-squared test), chisq(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,

| | |
|-------------------------|--|
| <code>Trait</code> | Name of trait |
| <code>p.value</code> | P-value calculated from hypothesis testing |
| <code>q.value</code> | Adjusted p-value from multiple testing using FDR correction |
| <code>odds.ratio</code> | 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) |
| <code>taSNP.hits</code> | Number of trait-associated SNPs in genomic intervals |
| <code>taSNP.num</code> | Number of SNPs for specific trait |

Author(s)

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See Also

`print.traseR`

Examples

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

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