# Package 'traseR'

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Author Li Chen, Zhaohui S.Qin
Maintainer li chen <li>li .chen@emory.edu&gt;</li>
<b>Description</b> 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.
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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 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 controling genome-wide density is the same as all SNPs from CEU

# Usage

data(CEU)

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# 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

# Author(s)

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

plots

Visualize of trait-associated SNPs

# **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-3,
plotSNP(snpdb, snpid, ext = 10000)
plotGene(snpdb, gene, ext = 10000)
plotInterval(snpdb,interval,ext = 10000)
```

#### **Arguments**

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snpdb	A GRange object or data frame, which is GWAS trait-associated SNPs down-loaded 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

snpid SNP rs number

will be analyzed.

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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

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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))
```

#### **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,...
```

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# **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.threshol	d								
	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)

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

# **Examples**

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

querys

Retrieve trait-associated SNPs based

# 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)
```

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#### **Arguments**

A GRange object or data frame, which is GWAS trait-associated SNPs down-loaded 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\_STRA

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"))
```

taSNP trait-associated SNPs in dbGaP and NHGRI downloaded from Association Results Browser

#### Description

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

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#### 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

segnames 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

#### Author(s)

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

taSNPLD linkage disequilibrium (>0.8) within 100kb SNPs of all traitassociated 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

data(taSNPLD)

### Value

The data frame taSNPLD contains four columns,

SNP\_ID SNP rs number

segnames Chromosome number associated with rs number

ranges Chromosomal position, in base pairs, associated with rs number

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

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#### 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

```
data(Tcell)
```

#### Value

The data frame Tcell contains three columns,

seqnames

Chromosome id

ranges

Chromosome position

#### Author(s)

Li Chen <a href="mailto:clicken@emory.edu">Li Chen <a href="mailto:c

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", "tw
ntimes=100, nbatch=1,
trait.threshold = 0, traitclass.threshold=0, pvalue = 1e-3)
```

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#### 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, Posit

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 back-

ground 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 adds.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.

Trait Name of trait

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

# 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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