

Package ‘PSCAN’

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Type Package

Title Protein-structure-guided scan tests for detecting gene-level associations and signal variants

Version 1.0

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Description Protein-structure-guided scan (PSCAN) tests for evaluating the gene-level association with complex traits and detecting signal clusters in protein space.

License GPL (>=2)

Depends R (>= 3.1.1), MASS, SKAT, igraph, rgl

NeedsCompilation no

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PSCAN-package	<i>Protein-structure-guided scan tests for detecting gene-level associations and signal variants</i>
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Description

PSCAN package has the main PSCAN function that implements protein-structure-guided scan (PSCAN) methods for detecting gene-level associations and signal variants.

Details

Package: PSCAN
Type: Package
Version: 1.0
Date: 2020-04-06
License: GPL (>=2)

PSCAN

Author(s)

Zheng-Zheng Tang

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References

Tang, Z.-Z., Sliwoski, G., Chen, G., Jin, B., Bush, W., Li, B., and Capra, T. (2020). Spatial scan tests guided by protein structures improve complex disease gene discovery and signal variant detection. Submitted.

Examples

```
data(one.gene)
U = one.gene$U
V = one.gene$V
MAC = one.gene$MAC
weight = one.gene$weight
# perform PSCAN mean-based test and signal region detection
PSCAN(type="mean", U=U, V=V, MAC=MAC, weight=weight, N.MC=1000, FWER=0.05, plot3D=TRUE)
```

one.gene

Score statistics, covariance matrix, and SNP weights for a gene

Description

The data are derived from the genetic association study for a gene.

Usage

```
data("one.gene")
```

Format

one.gene contains the following objects:

- U: a vector of score statistics
- V: a covariance matrix
- weight: a vector of SNP weights

Source

Tang, Z.-Z., Sliwoski, G., Chen, G., Jin, B., Bush, W., Li, B., and Capra, T. (2020). Spatial scan tests guided by protein structures improve complex disease gene discovery and signal variant detection. Submitted.

Examples

```
data(one.gene)
```

PSCAN	<i>Protein-structure-guided scan tests for detecting gene-level associations and signal variants</i>
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Description

This function allows users to (a) perform PSCAN gene-level association tests; (b) run PSCAN procedures to identify trait-associated variants.

Usage

```
PSCAN(type="mean", p.comb="Cauchy", U, V, MAC, weight=NULL, N.MC = 1000, FWER = 0.05,
f=0, plot3D = FALSE)
```

Arguments

type	type="mean" is the scan test for testing the mean of the effects and type="variance" is the scan test for testing the variance
p.comb	p.comb="Cauchy" or "minP" is to use Cauchy method or minimum p-value method to obtain the gene-level p-value
U	a vector of score statistics with names as SNP ID in chr:position format
V	a covariance matrix for the score statistics
MAC	a vector of minor allele count
weight	a vector contains the weights for SNPs (default = NULL, flat weight)
N.MC	the number of Monte Carlo simulations for minP gene-level test and signal region detection.
FWER	family wise error rate for the PSCAN signal region detection
f	overlap fraction cutoff in the signal detection algorithm ($0 \leq f \leq 1$). When $f = 0$, algorithm identifies nonoverlapping signal regions. When $f = 1$, algorithm essentially keeps every region passing the significance threshold as the detected signal regions.
plot3D	whether generate interactive 3D plot for signal regions in protein space. In the plot, the purple spheres represent signal variants and grey spheres represent non-signal variants. The plot will not be generated if no signal region is detected

Value

pscan.pval	a p-value of PSCAN mean-based or variance-based test
signal	a list of vector with each vector contains SNP IDs in a signal region
signal.pval	a vector of p-values with each p-value corresponds to the mean-based or variance-based test p-value for a signal region

Author(s)

Zheng-Zheng Tang

References

Tang, Z.-Z., Sliwoski, G., Chen, G., Jin, B., Bush, W., Li, B., and Capra, T. (2020). Spatial scan tests guided by protein structures improve complex disease gene discovery and signal variant detection. Submitted.

Examples

```
data(one.gene)
U = one.gene$U
V = one.gene$V
MAC = one.gene$MAC
weight = one.gene$weight
# perform PSCAN mean-based test and signal region detection
PSCAN(type="mean", U=U, V=V, MAC=MAC, weight=weight, N.MC=1000, FWER=0.05, plot3D=TRUE)
```

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