# Package 'SPAr'

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Title Perform rare variants association analysis based on summation of

Type Package

partition approaches

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<b>Description</b> This package performs robust nonparametric tests for rare variants association analysis using summation of partition approaches that incorporate gene-gene and gene-environmental interactions
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print.SPA

SPAr-package	Rare variants association tests based on summation of partition approaches
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# Description

Perform nonparametric association tests between a set of rare variants and continous or dichotomous outcomes incorporation gene-gene and gene-environmental interactions

#### **Details**

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# Author(s)

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#### References

Fan, R., Lo, S-H (2013). A robust model-free approach for rare variantss association studies incorporating gene-gene and gene-environmental interactions. *PLOS ONE* 8(12):e83057.

print.SPA

Print out the values and significance of SPA test scores.

# Description

This function prints out the numerical values of SPA test statistics and their p-values.

# Usage

```
## S3 method for class 'SPA'
print(x,...)
```

SPA.I

#### **Arguments**

```
x an SPA object, result form SPA.I or SPA.I.GE.
... See print.
```

# Author(s)

Ruixue Fan rf2283@columbia.edu

#### References

Fan, R., Lo, S-H (2013). A robust model-free approach for rare variantss association studies incorporating gene-gene and gene-environmental interactions. *PLOS ONE* 8(12):e83057.

#### See Also

```
SPA.I,SPA.I.GE
```

# **Examples**

```
data(SPA_example)
attach(SPA_example)
I1_dich <- SPA.I(x,y.d,interaction=1,nperm=100) # only compute I1 and its pvalue
print(I1_dich)</pre>
```

SPA.I

Compute rare variants association test scores with and(or) without gene-gene interactions

# Description

Compute the nonparametric association test scores between a set of rare variants and continous or dichotomous outcomes with and(or) without gene-gene interactions

# Usage

```
SPA.I(x, y, nperm = 100, type = "dichotomous", interaction = 0)
```

# Arguments

X	a matrix of numeric genotypes with each row as an individual and each column as a separate rare SNP. Each genotype should be coded as 0,1 and 2.
У	a vector of phenotypes. The length of $y$ should be the same as the number of rows of $x$ .
nperm	the number of permutations used to obtain the significance level.
type	a string to indicate if the phenoype is continuous or dichotomous. It could take values "dichotomous" or "continuous".
interaction	an integer indicating if the interaction is considered when computing the information score. See Details.

SPA.I

#### **Details**

There are three types of interactions: 1 - compute the score without interaction (score I1); 2 - compute the score with interaction (score I2); 0 - compute both scores with and without interaction as well as the adaptive p\* score.

# Value

I The numeric value of I score. Could be a vector of 3 values if interaction=0.

pvalue The p-value of corresponding I score. Could be a vector of 3 values if interac-

tion=0.

nperm Number of permutations.

call The matched call.

type Type of methods used. Could be "I1", "I2" or "p\*".

#### Author(s)

Ruixue Fan rf2283@columbia.edu

#### References

Fan, R., Lo, S-H (2013). A robust model-free approach for rare variantss association studies incorporating gene-gene and gene-environmental interactions. *PLOS ONE* 8(12):e83057.

# **Examples**

```
data(SPA_example)
attach(SPA_example)

## compute p-values of dichotomous traits

pstar_dich <- SPA.I(x,y.d,nperm=10) #compute I1,I2 and p* and their p-values
print(pstar_dich)

I1_dich <- SPA.I(x,y.d,interaction=1,nperm=10) # only compute I1 and its pvalue
print(I1_dich)

## compute p-values of continuous traits

pstar_cont <- SPA.I(x,y.c,nperm=10,type="cont") #compute I1,I2 and p* and their p-values
print(pstar_cont)

I1_cont <- SPA.I(x,y.c,type="cont",interaction=1,nperm=10) # only compute I1 and its pvalue
print(I1_cont)</pre>
```

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SPA.I.GE	Rare variants association test score to evaulate the effect of gene-
	environmental interactions

# Description

Compute the nonparametric association information score between a set of rare variants and dichotomous outcomes, incorporating interactions with environmental factors.

# Usage

```
SPA.I.GE(x, y, E, nperm = 100)
```

# Arguments

X	a matrix of numeric genotypes with each row as an individual and each column as a separate rare SNP. Each genotype should be coded as 0,1 and 2.
У	a vector of phenotypes. The length of $\boldsymbol{y}$ should be the same as the number of rows of $\boldsymbol{x}$ .
Е	a vector of environmental factors. It must be a categorical variable.
nperm	the number of permutations used to obtain the significance level. See details.

#### **Details**

This function evaluates the gene-environmental intearction effects associated with the dichotomous trait. The phenotype y must be dichotomous. The significance is computed using both global permutation and local permutation.

#### Value

I2star	The numerical value of the information score.
pvalue.global	The p-value from global permutation.
pvalue.local	The p-value from local permutation.
call	The matched call.
type	Type of the method used. It is "env".

#### Author(s)

Ruixue Fan rf2283@columbia.edu

#### References

Fan, R., Lo, S-H (2013). A robust model-free approach for rare variantss association studies incorporating gene-gene and gene-environmental interactions. *PLOS ONE* 8(12):e83057.

SPA\_example

# **Examples**

```
data(SPA_example)
attach(SPA_example)
SPA.I.GE <- SPA.I.GE(x,y.d,E,nperm=10)
print(SPA.I.GE)</pre>
```

SPA\_example

Example data for SPA with continuous and dichotomous traits and an environmental factor.

# Description

Example data for SPA with continuous and dichotomous traits and an environmental factor.

#### **Format**

SPA\_example contains the following objects:

**x** a numeric genotype matrix of 1000 individuals and 20 SNPs. Each row represents a different individual and each column represents a different SNP marker.

y.c a numeric vector of continuous phenotypes.

y.d a numeric vector of dichotomous phenotypes.

E a numeric vector of an environmental factor.

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