

# Package ‘genomicper’

February 23, 2026

**Type** Package

**Title** Circular Genomic Permutation using Genome Wide Association  
p-Values

**Version** 1.8

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**Imports** stats,grDevices,utils,graphics

## Description

Circular genomic permutation approach uses genome wide association studies (GWAS) results to establish the significance of pathway/gene-set associations whilst accounting for genomic structure. All single nucleotide polymorphisms (SNPs) in the GWAS are placed in a 'circular genome' according to their location. Then the complete set of SNP association p-values are permuted by rotation with respect to the SNPs' genomic locations. Two testing frameworks are available: permutations at the gene level, and permutations at the SNP level. The permutation at the gene level uses Fisher's combination test to calculate a single gene p-value, followed by the hypergeometric test. The SNP count methodology maps each SNP to pathways/gene-sets and calculates the proportion of SNPs for the real and the permuted datasets above a pre-defined threshold. Genomicper requires a matrix of GWAS association p-values and SNPs annotation to genes. Pathways can be obtained from within the package or can be provided by the user. Cabrera et al (2012) <[doi:10.1534/g3.112.002618](https://doi.org/10.1534/g3.112.002618)> .

**License** GPL-2

**NeedsCompilation** no

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|                    |                                      |
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| genomicper-package | <i>Circular Genomic Permutations</i> |
|--------------------|--------------------------------------|

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## Description

Description: Circular genomic permutation approach uses genome wide association studies (GWAS) results to establish the significance of pathway/gene-set associations whilst accounting for genomic structure. All single nucleotide polymorphisms (SNPs) in the GWAS are placed in a 'circular genome' according to their location. Then the complete set of SNP association p-values are permuted by rotation with respect to the SNPs' genomic locations. Two testing frameworks are available: permutations at the gene level, and permutations at the SNP level. The permutation at the gene level uses Fisher's combination test to calculate a single gene p-value, followed by the hypergeometric test. The SNP count methodology maps each SNP to pathways/gene-sets and calculates the proportion of SNPs for the real and the permuted datasets above a pre-defined threshold. Genomicper requires a matrix of GWAS association p-values and SNPs annotation to genes. Pathways can be obtained from within the package or can be provided by the user.

## Details

|          |            |
|----------|------------|
| Package: | genomicper |
| Type:    | Package    |
| Version: | 1.8        |
| Date:    | 2026-02-21 |
| License: | GPL-2      |

## Author(s)

Claudia P. Cabrera, Pau Navarro, Chris S. Haley  
 Maintainer: Claudia Cabrera <c.cabrera@qmul.ac.uk>

## References

SNP-level Permutations:

Genomicper: genome-wide association SNP-set analysis

Claudia P. Cabrera\*, Pau Navarro\*, Jennifer E. Huffman, Alan F. Wright, Caroline Hayward, Harry Campbell, James F. Wilson, Igor Rudan, Nicholas D. Hastie, Veronique Vitart, Chris S. Haley\*

Gene-level Permutations:

Uncovering Networks from Genome-Wide Association Studies via

Circular Genomic Permutation. *G3: Genes|Genomes|Genetics* 2, 1067-1075.

Claudia P. Cabrera\*, Pau Navarro\*, Jennifer E. Huffman, Alan F. Wright, Caroline Hayward, Harry Campbell, James F. Wilson, Igor Rudan, Nicholas D. Hastie, Veronique Vitart, Chris S. Haley\*

## See Also

Genomicper functions: 1) `read_pvals`, 2) `genome_order`, 3) `read2_paths`, 4A) `snps_permutation`, 4B) `genes_permutation`, 5) `get_results`, 6) `plot_results`

## Examples

```
#####
# Genomicper functions #####
# 1) read_pvals(data_name="", snps_ann="")
# 2) genome_order(all_data="")
# 3) read2_paths(ordered_alldata="", gs_locs="", sets_from="", sets_prefix="RHSA", level="")
# 4A) snps_permutation(ordered_alldata="", pers_ids="", ntraits="", nper="", saveto="",
# threshold="", gs_locs=gs_locs, envir = "")
# 4B) genes_permutation(ordered_alldata="", pers_ids="", pathways="",
# ntraits="", nper="", threshold="", saveto="", gs_locs=gs_locs, envir = "")
# 5) get_results(res_pattern="Perm", level="snp", from="workspace",
# threshold=0.05, envir = "")
# 6) plot_results(results = "", by = "", plot_all = TRUE, var = "", save_plot = TRUE,
# plot_name = "", bf = FALSE, save_qq = TRUE)
#####
##### DEMO: #####

#### SNP-level #####
# SNPs annotation and Pathways provided by user
# all data stored at the WORKSPACE

### Load files for analysis
data(demo, SNPsAnnotation)

# Read & format GWAS pvalues
all_data <- read_pvals(data_name=demo, snps_ann=SNPsAnnotation)

# Order data according to the genome
genome_results <- genome_order(all_data=all_data)
# Results from genome_order
ordered_alldata <- genome_results$ordered_alldata
gs_locs <- genome_results$gs_locs
```

```

# Create new environment to save variables (e.g. pathways, permutations):
gper.env <- new.env()

# Load example pathways into the new environment.
data(RHSA164843,RHSA446343,RHSA8876384,RHSA8964572,RHSA109582,RHSA1474244,envir=gper.env)

# Map SNPs to pathways
paths_res <- read2_paths(ordered_alldata=ordered_alldata,
gs_locs=gs_locs,sets_from="workspace",sets_prefix="RHSA",
level="snp",envir=gper.env)
# Results from read2_paths:
pers_ids <- paths_res$per_ors
pathways<- paths_res$pathways

# Perform permutations:
snps_permutation(ordered_alldata=ordered_alldata,
pers_ids=pers_ids,ntraits=c(7:13),nper=10,saveto="workspace",
threshold=0.05,gs_locs=gs_locs,envir = gper.env)

# Get results
results <- get_results(res_pattern="Permus",level="snp",
from="workspace",threshold=0.05,envir = gper.env)

# Plot results
## Not run:
#saves plots to working directory
qq <- plot_results(results=results,by="set",plot_all=TRUE)
qq <- plot_results(results=results,by="trait",
plot_all=FALSE,var="trait1")
# Displays interactive plot. Select a trait/set to plot and
# set arguments save_plot=FALSE, plot_all = FALSE
# IMPORTANT: to EXIT interactive plot, RIGHT CLICK on the
# plot and STOP.
qq <- plot_results(results=results,by="set",plot_all=FALSE,
var="RHSA109582",save_plot=FALSE)

## End(Not run)
# -- END OF DEMO
#####

```

---

demo

*GWAS p\_values demo data*

---

### Description

GWAS p-values (tab delimited file). First Column must contain the SNP ids and the column name = "name"

### Usage

data(demo)

**Format**

A data frame with SNPs identifiers and gwas p-values of association

name a character vector

Trait1 a numeric vector

Trait2 a numeric vector

Trait3 a numeric vector

Trait4 a numeric vector

Trait5 a numeric vector

Trait6 a numeric vector

Trait7 a numeric vector

Trait8 a numeric vector

Trait9 a numeric vector

| name       | Trait1    | Trait2     | Trait3    | Trait4    | Trait5    | Trait6     |
|------------|-----------|------------|-----------|-----------|-----------|------------|
| rs10000010 | 0.9122360 | 0.30088096 | 0.2332038 | 0.5193068 | 0.1255104 | 0.07253145 |
| rs10000023 | 0.8642906 | 0.52064064 | 0.9243443 | 0.7177759 | 0.9512171 | 0.81716250 |
| rs10000030 | 0.2832705 | 0.99021664 | 0.8359339 | 0.9662707 | 0.8491221 | 0.50208681 |

**Examples**

```
#Read input demo file for "read_pvals" function
data(demo)
```

---

genes\_permutation      *Gene-level Permutations*

---

**Description**

Performs gene-level circular genomic permutations. In each permutation, the complete set of SNP association p-values are permuted by rotation with respect to the SNPs' genomic locations. Once these 'simulated' p-values are assigned, the joint gene p-values are calculated using Fisher's combination test, and pathways' association tested using the hypergeometric test

**Usage**

```
genes_permutation(ordered_alldata = "", pers_ids = "", pathways = "",
ntraits = "", nper = 100, threshold = 0.05, seed=10, saveto = "workspace",
gs_locs="", envir = "")
```

**Arguments**

|                 |  |
|-----------------|--|
| ordered_alldata | Return variable from "genome_order". Ordered genome and trait p-values   |
| gs_locs         | Return variable from "genome_order". SNP indexes   |
| pers_ids        | Return variable "per_ors" from "read2_paths". Gene indexes   |
| pathways        | Return variable "pathways" from "read2_paths"  |
| ntraits         | Traits INDEX to be analysed. Index according to "ordered_alldata". Trait Columns index must start at 7. Example: ntraits=c(7:9), ntraits=7 |
| nper            | Number of permutations.Example: nper=1000  |
| threshold       | Threshold to be set by the hypergeometric test. threshold=0.05   |
| seed            | Set a number for random sampling   |
| saveto          | Save permutation results to "workspace" OR "directory"   |
| envir           | R environment to save the data to when saveto is set to "workspace"  |

**Value**

Returns "Permus\_trait" variables or files (permutation datasets).

**References**

Imports phyper (from stats)

**See Also**

[snps\\_permutation](#)

**Examples**

```
#load data
data(demo,SNPsAnnotation)
all_data <- read_pvals(data_name=demo,snps_ann=SNPsAnnotation)

# Prepare Genome
genome_results <-genome_order(all_data=all_data)
# Results from genome_order
ordered_alldata <- genome_results$ordered_alldata
gs_locs <- genome_results$gs_locs

# Create new environment to save data:
gper.env <- new.env()

# Get pathways
data(RHSA164843,RHSA446343,RHSA8876384,RHSA8964572,RHSA109582,RHSA1474244,envir=gper.env)

# Map Genes to pathways
paths_res <- read2_paths(ordered_alldata=ordered_alldata,gs_locs=gs_locs,
sets_from="workspace",sets_prefix="RHSA",level="gene",envir=gper.env)
pers_ids <- paths_res$per_ors
```

```

pathways<- paths_res$pathways

# Perform Permutations:
genes_permutation(ordered_alldata=ordered_alldata,
pers_ids=pers_ids,pathways=pathways,ntraits=c(7:9),
nper=10,threshold=0.05, saveto="workspace",
gs_locs=gs_locs,envir = gper.env)

# Results
results <- get_results(res_pattern="Permus",level="gene",
from="workspace",threshold=0.05,envir= gper.env)

```

genome\_order

*Genome Order***Description**

Orders the SNPs according to their genomic location

**Usage**

```
genome_order(all_data = "")
```

**Arguments**

all\_data           SNPs to Genes Annotation and Trait Pvalues of Association  
all\_data = (read\_pvals output) OR matrix/dataframe.

**Details**

Input Columns with "\*" must be included for analysis

NOTE: Trait p-values must start at Column #7

```

# *Column 1: "name" (SNP_IDs - any SNP ID as character)
# *Column 2: Chromosome Location
# *Column 3: SNP Location
# *Column 4: Gene ID
# Column 5: Symbol (OR Annotation Field 1)
# Column 6: Annotaiton Field 2
# *Column 7: First trait pvalues of association
# Column N: Next trait pvalues of association
# Example Input Data:
name           Chromosome   Location   GENE_ID   Symbol   Orientation   abpi
rs10000010         4   21618674   80333   KCNIP4         -   0.91
rs10000023         4   95733906   658   BMPR1B         +   0.86
rs10000092         4   21895517   80333   KCNIP4         -   0.20
rs1000022           13  100461219  171425   CLYBL           +   0.26
rs10000300         4   40466547   54502   RBM47           -   0.58

```

**Value**

ordered\_alldata           SNPs annotated to Genes and Trait p-values

gs\_locs                   Gene annotations, location indexes and number of observations

**Format**

SNPs annotated to Genes and Trait p-values

```
#ordered_alldata[1:5,1:8]
name  Chromosome Location GENE_ID  Symbol Orientation Trait1 Trait2
rs3934834 1 1005806 NA <NA> <NA> 0.97 0.92
rs3737728 1 1021415 54991 C1orf159 - 0.91 0.69
rs6687776 1 1030565 54991 C1orf159 - 0.71 0.45
rs9651273 1 1031540 54991 C1orf159 - 0.22 0.60
rs4970405 1 1048955 54991 C1orf159 - 0.77 0.56
```

Gene annotations, location indexes and number of observations

```
#gs_locs[1:5,]
# Symbol Chromosome Location Gene_ID Start_Indx Observations
# [1,] "A1BG" "19" "58864479" "1" "293976" "1"
# [2,] "A2M" "12" "9232268" "2" "215264" "5"
# [3,] "NAT1" "8" "18077310" "9" "151804" "1"
# [4,] "NAT2" "8" "18257280" "10" "151831" "2"
# [5,] "SERPINA3" "14" "95080803" "12" "249519" "2"
```

**See Also**

[read2\\_paths](#)

**Examples**

```
## DEMO WORKSPACE

data(demo, SNPsAnnotation)
all_data<-read_pvals(data_name=demo,snps_ann=SNPsAnnotation)
# GENOME ORDER
genome_results <- genome_order(all_data=all_data)
ordered_alldata <- genome_results$ordered_alldata
gs_locs <- genome_results$gs_locs
```

---

get\_results

*Circular Permutation Results*

---

**Description**

Creates a summary dataframe of the genomic permutations datasets

**Usage**

```
get_results(res_pattern="Permus", level="snp", from="workspace",
threshold=0.05, envir = "")
```

**Arguments**

|             |   |
|-------------|---|
| res_pattern | Pattern of the Permutation files/variable. eg. res=pattern="Permus"         |
| level       | Permutation level performed.level values "snp" or "gene"                    |
| from        | Location of the permutation datasets.from values "workspace" or "directory" |
| threshold   | Threshold of significance set   |
| envir       | R environment where save the data to  |

**Value**

|         |   |
|---------|---|
| results | Data frame with Pathway ID, Trait, Threshold set by permutations, Gene results include the theoretical hypergeometric p-value and the, observed (Empirical Hypergeometric p-values) SNP results include the count of significant SNPs and the overall score Score is the proportion of tests observed with more significant results |
|---------|---|

**Format**

```
## SNP level results
  PathID  Trait Threshold RealCount Score
1 hsa00010  abpi         0         0 0.037
2 hsa00010 abpildfa         0         0 0.040
3 hsa04720  abpi         2         0 0.311
## Gene level results
  PathID Trait  Threshold  P-Value  Observed
1 hsa00010  abpi 0.040441176 0.058823529 1.0000000
2 hsa00020  abpi 0.000000000 0.000000000 0.1666667
3 hsa00030  abpi 0.040441176 0.058823529 1.0000000
```

**Examples**

```
data(demo, SNPsAnnotation)
all_data <- read_pvals(data_name=demo, snps_ann=SNPsAnnotation)
genome_results <- genome_order(all_data=all_data)
# Results from genome_order
ordered_alldata <- genome_results$ordered_alldata
gs_locs <- genome_results$gs_locs

# Create new environment to save data
gper.env <- new.env()

# Get pathways
data(RHSA164843, RHSA446343, RHSA8876384, RHSA8964572, RHSA109582,
RHSA1474244, envir=gper.env)
```

```
paths_res <- read2_paths(ordered_alldata=ordered_alldata,gs_locs=gs_locs,
sets_from="workspace",sets_prefix="RHSA",level="snp",envir=gper.env)
pers_ids <- paths_res$per_ors
pathways<- paths_res$pathways

snps_permutation(ordered_alldata=ordered_alldata,pers_ids=pers_ids,
ntraits=c(7,9),nper=10,saveto="workspace",threshold=0.05,
gs_locs=gs_locs,envir= gper.env)

results <- get_results(res_pattern="Permus",level="snp",
from="workspace",threshold=0.05,envir = gper.env)
```

---

hyprbg

*Hypergeometric Test (phyper)*

---

### **Description**

Performs Hypergeometric test (phyper() from R)

### **Usage**

```
hyprbg(Sig_in_Paths, uniSig, gns_in_Paths, universe)
```

### **Arguments**

|              |  |
|--------------|--|
| Sig_in_Paths | Number of significant genes in the pathway |
| uniSig       | Number of significant genes in the dataset |
| gns_in_Paths | Number of genes in the pathway             |
| universe     | Number of genes in the dataset             |

### **Value**

Returns hypergeometric test

### **References**

hyprbg Imports phyper() (from stats)

---

|              |  |
|--------------|--|
| plot_results | <i>Plot Results Circular Permutation</i> |
|--------------|--|

---

**Description**

QQ plots

**Usage**

```
plot_results(results="",by="",plot_all=TRUE, var = "", save_plot=TRUE, plot_name="",
bf= FALSE , save_qq = TRUE)
```

**Arguments**

|           |  |
|-----------|--|
| results   | Results datarame from "get_results()"  |
| by        | Visualize results by "trait" OR by "set"   |
| plot_all  | plot_all = TRUE plots all the variables in the results dataframe and saves a pdf file in the working directory. Setting plot_all to FALSE plots a single variable(trait or set). The argument "var" must be declared.  |
| var       | Variable name to plot  |
| save_plot | save_plot = TRUE saves the plots in the working directory.<br>save_plot = FALSE the plot is visualized at the console.<br>save_plot = FALSE can be used only when plot_all is set to FALSE.<br>The plot displayed at the console is interactive, clicking on a point displays the points name. |
| plot_name | Argument used to save the file name for the plots. Default value = Results_genomicper_[set/trait]  |
| bf        | Displays the bonferroni correction   |
| save_qq   | TRUE returns the qq plot values  |

**Value**

|    |                                |
|----|--------------------------------|
| qq | Data frame with qq plot values |
|----|--------------------------------|

**See Also**

[get\\_results](#)

**Examples**

```
data(demo,SNPsAnnotation)
all_data <- read_pvals(data_name=demo,snps_ann=SNPsAnnotation)
genome_results <-genome_order(all_data=all_data)
# Results from genome_order
ordered_alldata <- genome_results$ordered_alldata
gs_locs <- genome_results$gs_locs
```

```

# Create new environment to save the data:
gper.env <- new.env()

# Load Pathways
data(RHSA164843,RHSA446343,RHSA8876384,RHSA8964572,RHSA109582,RHSA1474244,envir=gper.env)

paths_res <- read2_paths(ordered_alldata=ordered_alldata,gs_locs=gs_locs,
sets_from="workspace",sets_prefix="RHSA",level="snp",envir=gper.env)
pers_ids <- paths_res$per_ors
pathways<- paths_res$pathways

snps_permutation(ordered_alldata=ordered_alldata,pers_ids=pers_ids,
ntraits=c(7,9),nper=10,saveto="workspace",threshold=0.05,
gs_locs=gs_locs,envir = gper.env)

results <- get_results(res_pattern="Permus",level="snp",
from="workspace",threshold=0.05,envir = gper.env)

#saves plots to working directory
## Not run:
qq <- plot_results(results=results,by="set",plot_all=TRUE)
qq <- plot_results(results=results,by="trait",plot_all=FALSE,var="trait1")
qq <- plot_results(results=results,by="set",
plot_all=FALSE,var="R-HSA-8964572",
save_plot=FALSE) ## IMPORTANT: to EXIT interactive plot
## right click on the plot to stop

## End(Not run)

```

---

read2\_paths

*Read to SNPs to sets; Map SNPs to gene-sets/pathways*


---

## Description

Reads the sets/pathways, map the SNPs and genes to the gene-sets/pathways read2\_paths uses the "genome\_order" output(ordered\_alldata, gs\_locs) to assign genomic location indexes to each element in the gene-set. The permutation method must be defined (i.e. level = "snp" OR level = "gene").

## Usage

```
read2_paths(ordered_alldata="",gs_locs="",sets_from="workspace",
sets_prefix="RHSA",level="snp",envir="")
```

## Arguments

ordered\_alldata

Ordered data according to the SNPs genomic location. Traits start at column 7

Return variable from:

```

genome_results <-genome_order(all_data=all_data)
ordered_alldata <- genome_results$ordered_alldata

gs_locs      Gene annotation,indexes and number of observations
              Return variable from genome_order():
              genome_results <-genome_order(all_data=all_data)
              gs_locs <- genome_results$gs_locs

sets_from    Location of the gene-sets. Default set to "workspace"
              sets_from="workspace" OR sets_from="directory"
              "directory", only will search for information in the working directory.

sets_prefix  Prefix of the gene-set variables or files.
              Default set to sets_prefix= "RHSAs" e.g. Variables "RHSAs164843","RHSAs446343","RHSAs8876384"
              each variable/file contains the list of gene identifiers part of that pathway

level       The level at which the permutations will be performed. Assigns the indexes
              according to snps or genes
              Default value "snp" level values = "snp" OR "gene"

envir       R environment where pathway data is stored. e.g(envir=.GlobalEnv, envir=gper.env)

```

**Value**

```

pathways    Pathway Id, Description,Number of Genes in the pathway, Number of genes
              found in the dataset, Number of SNPs found in the dataset

per_ors     A list of identifiers mapped to each pathway

```

**Format**

```

Input: Ordered_alldata
name      Chromosome  Location  GENE_ID  Symbol  Orientation  Trait1  Trait2
rs1001567      1  9194614   <NA>    <NA>    <NA>  0.96  0.89
rs1000313      1 15405489  23254  KIAA1026  +  0.93  0.57
rs1002365      1 19797248  <NA>    <NA>    <NA>  0.68  0.58
rs1002706      1 25051153  <NA>    <NA>    <NA>  0.71  0.02
rs1002487      1 26865971  6195   RPS6KA1  +  0.98  0.78

```

```

Input:gs_locs
      Symbol  Chromosome  Location  Gene_ID  Start_Indx  Observations
[1,] "ACYP2"  "2"         "54399633" "98"      "35"        "1"
[2,] "AMPD3"  "11"        "10514707" "272"     "898"       "1"
[3,] "ANK2"   "4"         "113830885" "287"     "479"       "4"

```

```

Input:pathway example
RHSAs8964572
[1] 1149 128486 161247 29923 345275 63924

```

```

Output:pathways
      ID          GenesInPath  GenesFound  SNPsInPath

```

```
"RHS109582" "681" "8" "11"
"RHS1474244" "418" "7" "10"
"RHS164843" "11" "0" "0"
"RHS446343" "4" "1" "1"
"RHS8876384" "32" "1" "1"
"RHS8964572" "6" "1" "1"
```

## See Also

[genes\\_permutation](#) [snps\\_permutation](#) [genome\\_order](#)

## Examples

```
## DEMO - SNP Level data stored in workspace #####
# library(genomicper)
data(demo, SNPsAnnotation)
all_data <- read_pvals(data_name=demo, snps_ann=SNPsAnnotation)
genome_results <- genome_order(all_data=all_data)
ordered_alldata <- genome_results$ordered_alldata
gs_locs <- genome_results$gs_locs

# Create new environment to save variables (e.g. pathways, permutations):
gper.env <- new.env()

data(RHS164843, RHS446343, RHS8876384, RHS8964572, RHS109582, RHS1474244, envir=gper.env)

paths_res <- read2_paths(ordered_alldata=ordered_alldata,
gs_locs=gs_locs, sets_from="workspace", sets_prefix="RHS",
level="snp", envir=gper.env)
pers_ids <- paths_res$per_ors
pathways <- paths_res$pathways
#####
```

---

read\_pvals

*Read GWAS p-values of association and Merge with SNP annotations*

---

## Description

Read GWAS p-values of association and Merge with SNP annotations for analysis

## Usage

```
read_pvals(data_name="", snps_ann="", from="workspace")
```

**Arguments**

|           |   |
|-----------|---|
| data_name | GWAS p_values (tab delimited file)(SNP_IDs Trait1 Trait2 ...TraitN)   |
| snps_ann  | SNPs Annotation (SNPsAnnotation). Genomicper uses entrez gene ids to annotate<br>associate SNPs-to genes-pathways.<br>The annotation MUST match your data input (coordinates and chromosome format)<br>Any SNP ID is valid, as long the ID is set as character<br>The examples below show an option on how to annotate the SNPs prior the use of genomicper |
| from      | Datasets location. Values "workspace" OR "directory"  |

**Value**

Dataframe: name; chromosome; Location; GeneID; Symbol; Orientation; Trait1; TraitN

**Formats**

```
GWAS p_values (tab delimited file)(SNP_IDs Trait1 Trait2 ...TraitN)
name      Trait1      Trait2      TraitN
rs10000010 0.9122360 0.30088096 0.2332038
rs10000023 0.8642906 0.52064064 0.9243443
rs10000030 0.2832705 0.99021664 0.8359339
```

```
SNPs Annotation (SNPsAnnotation)
name      Chromosome  Location  GENE_ID  Symbol  Orientation
rs1000313 1             15405489 23254    KIAA1026 +
rs1000533 1             168282491 9095     TBX19    +
rs1000731 1             231963491 27185    DISC1    +
```

Output:

```
name      Chromosome  Location  GENE_ID  Symbol  Orientation  Trait1
rs10000010 4          21618674 80333    KCNIP4  -           0.9122360
rs10000023 4          95733906 658      BMPR1B  +           0.8642906
rs10000030 4          103374154 NA       <NA>     <NA>      0.2832705
```

**See Also**

[genome\\_order](#)

**Examples**

```
## DEMO // WORKSPACE
data(demo, SNPsAnnotation)
all_data <- read_pvals(data_name=demo, snps_ann=SNPsAnnotation)
```

---

 RHSAXXXX

*Reactome Pathway examples*


---

**Description**

Each file "RHSAXXXX" contains the gene identifiers.

**Usage**

```
data(RHSA164843)
```

**Format**

The format is: num [1:6] 11168 155030 155348 155459 155908 2547...

**Source**

reactome.db

**Examples**

```
data(RHSA164843)
```

---

SNPsAnnotation

*SNPs-Genes annotation to Distance 0 (SNPs within a gene)*


---

**Description**

SNPs annotated to genes. Annotation only when the SNPs fall within start and end of transcription of the genes.

**Usage**

```
data(SNPsAnnotation)
```

**Format**

Sample data frame with 339096 SNP observations on the following 6 variables.

name a character vector

Chromosome a character vector

Location a numeric vector of the SNP location

GENE\_ID a numeric vector with entrez geneID

Symbol a character vector ; other annotation slot 1

Orientation a character vector; other annotation slot 2

| name      | Chromosome | Location  | GENE_ID | Symbol   | Orientation |
|-----------|------------|-----------|---------|----------|-------------|
| rs1000313 | 1          | 15405489  | 23254   | KIAA1026 | +           |
| rs1000533 | 1          | 168282491 | 9095    | TBX19    | +           |
| rs1000731 | 1          | 231963491 | 27185   | DISC1    | +           |

### Source

NCBI Gene database,(<http://www.ncbi.nlm.nih.gov/gene> ; Build.37.1).

### Examples

```
data(SNPsAnnotation)
```

---

|                  |                               |
|------------------|-------------------------------|
| snps_permutation | <i>SNP-level permutations</i> |
|------------------|-------------------------------|

---

### Description

Performs SNP-level circular genomic permutations. In each permutation, the complete set of SNP association p-values are permuted by rotation with respect to the SNPs' genomic locations.

Once these 'simulated' p-values are assigned, the proportion of SNPs per set above a pre-defined threshold is calculated

### Usage

```
snps_permutation(ordered_alldata = "", pers_ids = "", ntraits = "",
nper = 100, threshold = 0.05, seed=10,saveto = "workspace",
gs_locs = "",envir = "")
```

### Arguments

|                 |  |
|-----------------|--|
| ordered_alldata | Return variable from "genome_order". Ordered genome and trait p-values   |
| gs_locs         | Return variable from "genome_order". SNP indexes   |
| pers_ids        | Return variable "per_ors" from "read2_paths". SNP indexes  |
| ntraits         | Traits INDEX to be analysed. Index according to "ordered_alldata". Trait Columns index must start at 7. Example: ntraits=c(7:9), ntraits=7 |
| nper            | Number of permutations.Example: nper=1000  |
| threshold       | Threshold to be set by the hypergeometric test. threshold=0.05   |
| seed            | Set number for random sampling   |
| saveto          | Save permutation results to "workspace" OR "directory"   |
| envir           | R environment to save the Permutations to when saveto is set to "workspace"  |

**Value**

Returns "Permus\_genesetsname" variables or files (permutation datasets).

**See Also**

[genes\\_permutation](#)

**Examples**

```
data(demo, SNPsAnnotation)
all_data <- read_pvals(data_name=demo, snps_ann=SNPsAnnotation)
genome_results <- genome_order(all_data=all_data)
# Results from genome_order
ordered_alldata <- genome_results$ordered_alldata
gs_locs <- genome_results$gs_locs

# Create new environment to save the permutations to:
gper.env <- new.env()

data(RHSA164843, RHSA446343, RHSA8876384, RHSA8964572,
RHSA109582, RHSA1474244, envir=gper.env)

paths_res <- read2_paths(ordered_alldata=ordered_alldata, gs_locs=gs_locs,
sets_from="workspace", sets_prefix="RHSA", level="snp", envir=gper.env)
pers_ids <- paths_res$per_ors
pathways <- paths_res$pathways

# SNP permutations
snps_permutation(ordered_alldata=ordered_alldata, pers_ids=pers_ids,
ntraits=c(7,9), nper=10, saveto="workspace", threshold=0.05,
gs_locs=gs_locs, envir = gper.env)

# Get results
results <- get_results(res_pattern="Permus", level="snp",
from="workspace", threshold=0.05, envir = gper.env)
```

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