

Package ‘POPPATHR’

August 8, 2019

Title Population-based pathway analysis of SNP-SNP coevolution

Version 1.0

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Depends R ($\geq 3.5.0$)

Imports plyr,
dplyr,
ggplot2,
data.table,
tools,
stringr,
reshape2,
gdata,
RColorBrewer,
gridExtra,
cowplot,
utils,
GenomicRanges,
snpStats

Description A bioinformatics package for determining pathway-level SNP coevolution driven by population-based positive selection.

License GPL (≥ 2)

Encoding UTF-8

URL <https://github.com/rosscm/POPPATHR>

RoxygenNote 6.1.1

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calcFST	<i>Calculates SNP-level FST values for use in setupGSEArun.R</i>
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Description

Calculates SNP-level FST values for use in setupGSEArun.R

Usage

```
calcFST(genoF, realFam, outDir, outF)
```

Arguments

genoF	(char) path to file with SNP genotype data (PLINK format).
realFam	(char) path to PLINK case/control coded fam file.
outDir	(char) directory to store output files.
outF	(char) path to write SNP-FST file.

Value

none

getPathStats	<i>Generates SNP lists per high-confidence pathway as determined by GSEA</i>
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Description

Generates SNP lists per high-confidence pathway as determined by GSEA

Usage

```
getPathStats(genoF, resF, gseaStatF, snp2geneF, enrichNES = 0.3,  
  unenrichNES = 0.1, enrichDir, unenrichDir)
```

Arguments

genoF	(char) path to file with SNP genotype data (PLINK format).
resF	(char) path to files with GSEA results. Structured to compare results of two population analyses i.e., CEU vs. YRI and CEU vs. LWK.
gseaStatF	(char) path to GSEA statistics file.
snp2geneF	(char) path to SNP-gene mapping file.
enrichNES	(integer) NES cutoff to select validated selection-enriched pathways (default=0.3)
unenrichNES	(integer) NES cutoff to select unenriched pathways (default=0.1)
enrichDir	(char) path to directory to store output files (PLINK files per validated selection-enriched pathway)
unenrichDir	(char) path to directory to store output files (PLINK files per unenriched pathway)

Value

none

popPCA	<i>Calculate population stratification between different ancestry groups (e.g., CEU vs YRI) via complete linkage clustering</i>
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Description

Calculate population stratification between different ancestry groups (e.g., CEU vs YRI) via complete linkage clustering

Usage

```
popPCA(genoF, famF, dimensions = 3L, outF)
```

Arguments

genoF	(char) path to file with SNP genotype data (PLINK format).
famF	(char) path to PLINK fam file (case/control population coded).
dimensions	(integer, default=3).
outF	(char) path to write PNG image.

Value

none

recodeFAM	<i>Recodes PLINK fam file to case/control format by population adapted from SP plink_baseSetup.R (part of GWAS2pathway package)</i>
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Description

Recodes PLINK fam file to case/control format by population adapted from SP plink_baseSetup.R (part of GWAS2pathway package)

Usage

```
recodeFAM(genoF, pop1, pop2, popsF, setSeed = 42L, caseCode = 2,
  ctrlCode = 1, outF = "case-ctrl")
```

Arguments

genoF	(char) path to file with SNP genotype data (PLINK format)
pop1	(char) character code for the first population (controls).
pop2	(char) character code for the second population (cases).
popsF	(char) path to file with population information. Gives the number of samples per population in the dataset.
setSeed	(integer) value for set.seed() before shuffling (default=42).
caseCode	(integer) value for case samples in fam phenotype column.
ctrlCode	(integer) value for control samples in fam phenotype column.
outF	(char) optional - name for fam file (default=case-ctrl). file extension added.

Value

none

setupGSEArun	<i>Sets up and runs GSEA using population-based FST table containing GSEA results (Geneset, Size, ES, NES, NominalP, FDR, FWER)</i>
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Description

Sets up and runs GSEA using population-based FST table containing GSEA results (Geneset, Size, ES, NES, NominalP, FDR, FWER)

Usage

```
setupGSEArun(realF, pathF, snp2geneF, setPerm = 1000L,
  snp2genedist = 500000L, minGene = 10L, maxGene = 300L,
  setSeed = 42L, outDir)
```

Arguments

realF	(char) path to file with real SNP association statistics
pathF	(char) path to pathway definitions GMT file
snp2geneF	(char) path to file with snp2gene mappings. Output of mapSNP2gene() (found in GWAS2Pathway)
setPerm	(integer) set cycle of permutations to run default=1000
snp2genedist	(integer) value for GSEA –distance. Max. distance between SNP and gene for their association in snp2geneF
minGene	(integer) value for GSEA –setmin. Min. number of genes in a gene set to be considered
maxGene	(integer) value for GSEA –setmax. Max. number of genes in a gene set to be considered
setSeed	(integer) value for GSEA –seed

Value

none

SNP2gene	<i>Maps SNPs to their nearest genes (for use in setupGSEArun.R) (adapted from map_SNP2gene from SP's GWAS2pathway package)</i>
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Description

NOTE: updated GenomicRanges to version 1.30.1 on 01/30/2018 Requires ignore.strand=TRUE param to properly run distanceToNearest() given unknown strand assignment for SNP array-based genotyping info

Usage

```
SNP2gene(inF, geneF, marg = 0L, outF)
```

Arguments

inF	(char) path to PLINK .bim file (only CHR, SNP, and BP columns considered).
geneF	(char) path to refseq table with header.
marg	(integer) region upstream and downstream of [txStart,txEnd].
outF	(char) path to write snp2gene mapping.

Value

none

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