TransMetaRare Package

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1 Overview

The TransMetaRare package contains functions to implement the score test for rare variant associations in trans-ethnic meta-analysis. The method uses summary level score statistics to carry out gene-based meta-analysis for rare variants.

2 Rare Varinats Trans-ethnic Meta-anlaysis in a Population-based Study Design

```
> ##############################################################################
> ####### To run this code, first download and unzip example files
> ##############################################################################
>
> library(TransMetaRare)
> y1 = as.matrix(read.table('y1.txt'))
> y2 = as.matrix(read.table('y2.txt'))
> y3 = as.matrix(read.table('y3.txt'))
> y4 = as.matrix(read.table('y4.txt'))
> y.list = list()
> y.list[[1]] = y1
> y.list[[2]] = y2
> y.list[[3]] = y3
> y.list[[4]] = y4;
> #########################
>
> x1 = as.matrix(read.table('x1.txt'))
> x2 = as.matrix(read.table('x2.txt'))
> x3 = as.matrix(read.table('x3.txt'))
> x4 = as.matrix(read.table('x4.txt'))
> x.list = list()
> x.list[[1]] = x1
> x.list[[2]] = as.matrix(x2[,1])
```

```
> x.list[[3]] = x3
> x.list[[4]] = x4
> ########################
> Kins_EUR = as.matrix(read.table('Kins_EUR.txt'))
> Kins_OW = as.matrix(read.table('Kins_OW.txt'))
> K.list = list()
> K.list[[1]] = Kins_EUR
> K.list[[2]] = Kins_OW
> K.list[[3]] = Kins_OW
> K.list[[4]] = Kins_OW
> ########################
> G.list = list()
> for(i in 1:50){
+         Z = as.matrix(read.table( paste("Z",i,".txt",sep="")) )
+         G.list[[i]] = Z
+ }
>

> # Compute a p-value of the first gene in a population-based study design
>
> Genetic_Kernel = Get_Kernel_Matrix(y.list, x.list, G.list, n.cohort = 4)
> obj = TransMeta_Rare_Null_Model(y.list, x.list, n.cohort=4, out_type="C")
> rs1 = TransMeta_Rare_wZ (G.list[[1]], obj, Kernel = Genetic_Kernel)
> rs1$p.value.normCopula.adj

[1] 2.684314e-34

>

> ### with groups
>
> Genetic_Kernel_G = Get_Kernel_Matrix(y.list, x.list, G.list, n.cohort = 4,
+                 Group_Idx=c(1,1,2,3))
> rs2 = TransMeta_Rare_wZ (G.list[[1]], obj, Kernel = Genetic_Kernel_G,
+                 Group_Idx=c(1,1,2,3))
> rs2$p.value.normCopula.adj

[1] 5.051275e-34

>
```

3 Rare Varinats Trans-ethnic Meta-anlaysis in a Family-based Study Design

```
> # Compute a p-value of the first gene in a family-based study design
>
```

```
> obj<-TransMeta_Rare_Null_Model_EmmaX(y.list, x.list, n.cohort=4, out_type="C", K.list)
> rs3 = TransMeta_Rare_wZ (G.list[[1]], obj, Kernel = Genetic_Kernel)
> rs3$p.value.normCopula.adj

[1] 6.334377e-34

>
>
>
>
```