# Package 'coreTDT'

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**Type** Package

**Title** TDT for compound heterozygous and recessive models

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**Description** Use to analysis case-
parent trio sequencing studies. Test the compound heterozygous and recessive disease models

**License** GPL-3

**NeedsCompilation** no

**Repository** CRAN

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## R topics documented:



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`compHet_TDT_v6` *compute p value for coreTDT*

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### Description

analysis after quanlity control

### Usage

```
compHet_TDT_v6(parent.geno, child.geno)
```

### Arguments

| | |
|---|---|
| parent.geno | array, parents genotype |
| child.geno | matrix, childs' genotype |

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coreTDT *Transimission Disequilibrium Test for compound heterozygous and recessive models*

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### Description

This program is used to compute the pvalues for Transimission Disequilibrium Test for compound heterozygous and recessive models

### Usage

```
coreTDT_geneset(samplePed, controlInf, useControlMAF = TRUE, maf.threshold = 1,
 qc.proportion = 0.8, geneList = c(),
 outputFile = "coreTDT_analysis.out",
 chrX = FALSE,writeFile=FALSE)
coreTDT_single(ped, maf.threshold = 1, qc.proportion = 0.8,
geneid = NA,control.maf = NULL)
```

### Arguments

| | |
|---|---|
| samplePed | plink file to input genetype informations, ref to PLINK recodeA |
| controlInf | Files form ATAV,contain information about variants,(evs dataset used) |
| useControlMAF | logical, if true, remove the variants with control MAF >= maf.threshold, else use parents MAF |
| maf.threshold | The allowed maximum of MAF that variants will be used in computation |
| qc.proportion | variants that have more than qc.proportion families with enough coverage will be used in computation |
| geneList | a vector containing gene names that used to analysis |
| outputFile | output file name |
| chrX | logical, if true, analyse chromosome X, not activated yet |
| writeFile | logical, if true, write the results to outputFile |
| ped | contain the genotype information for all samples,assume m families and n snps, 3m * n matrix, each column represents a variant, coded by 0/1/2 (number of alternative alleles);each row represents a sample, the first m rows are for child,the second m rows are for mother,the last m rows are for father |
| geneid | character, gene name |
| control.maf | vector contain the MAF of each variant in controls |

## Value

| | |
|---|---|
| pvalue_pr | pvalues computed from probabaility model |
| pvalue_lr | pvalues from likelihood ratio test with restricted alternative hypothesis |
| pvalue_lr2 | pvalues from likelihood ratio test |
| nmissing | number of variants is missing in data |
| nMedErr | number of loci contain mendel errors |
| nfamily | sample size |
| nsnp | number of variants used in analysis |
| N11 | number of family with parents compound genotype 1,1 |
| N12 | number of family with parents compound genotype 1,2 |
| N112 | number of family with compound genotype 1,1,2 |
| N122 | number of family with compound genotype 1,2,2 |

## Author(s)

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## References

Yu Jiang, Janice M McCarthy, Andrew S Allen,Testing the effect of rare compound-heterozygous and recessive mutations in case-parent sequencing studies (In Preparation)

## Examples

```
data(coreTDTexample)
attach(coreTDTexample)
coreTDT_geneset(samplePed, controlInf,maf.threshold=0.05,writeFile=FALSE)
```

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# coreTDT.results.2.df *convert coreTDT class to dataframe*

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## Description

convert coreTDT class, i.e., the result list generated inside coreTDT_geneset to dataframe

## Usage

```
coreTDT.results.2.df(coreTDTresults)
```

## Arguments

| | |
|---|---|
| coreTDTresults | result list generated inside coreTDT_geneset |

## Value

dataframe summarized the coreTDT result

## coreTDTexample *Example data for coreTDT*

### Description

Example data for coreTDT.

### Format

coreTDTexample contains the following objects:

**samplePed** dataframe, a numeric genotype matrix of 447 individuals and 244 SNPs. Each row represents a different individual, and each column represents a different SNP marker(from the 7th column), PLINK format

**evs** dataframe, rowname: varaint ID() chr_pos_ref_alt/rsID_); col1: gene ID; col2:varID;col3:indicator of variants included in analysis col4: number of samples have genotype 2;col5: number of samples have genotype 1;col6: number of samples have genotype 0;col7: mean coverage at this locus

## FamilyPhase *Phasing trios*

### Description

compute compound gentoype for trios from sequencing data

### Usage

```
FamilyPhase(parent.genotype, child.genotype)
FamilyPhaseII(parent.genotype, child.genotype)
FamilyPhaseIII(parent.genotype, child.genotype)
PairPhase(paternal.genotype, child.genotype)
```

### Arguments

`parent.genotype` matrix, parents genotype

`child.genotype` vector, child gentoype

`paternal.genotype` vector, genotype of one parent

### Details

PairPhase: sharing analysis between one parent and child FamilyPhase: compute cpmpound genotype for trios when parents do not share any variants FamilyPhaseII: compute cpmpound genotype for trios. when parents share heterozygous variants, remove shared variants and perform test FamilyPhaseIII: compute cpmpound genotype for trios. when parents share heterozygous variants, set family as missing data,used in current analysis

### Value

3 elements vector: paternal compound genotype, maternal compound genotype and child's compound genotype

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## pvalue_calculator *compute p value for exact coreTDT*

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### Description

compute all kinds of p values for exact coreTDT

### Usage

```
pvalue_calculator(y1, y2, n1, n2, theta1 = 0.25, theta2 = 0.5)
loglr_comp(x1, x2, n1, n2, theta1 = 0.25, theta2 = 0.5)
loglr_comp_2side(x1, x2, n1, n2, theta1 = 0.25, theta2 = 0.5)
```

### Arguments

| | |
|---|---|
| y1 | integer, N112 |
| y2 | integer, N122 |
| n1 | integer, N11 |
| n2 | integer, N12 |
| theta1 | float, probaliblity of N112 given N11 |
| theta2 | float, probaliblity of N122 given N12 |
| x1 | integer, N112 |
| x2 | integer, N122 |

### Value

| | |
|---|---|
| pvalue_pr | pvalues computed from probabaility model |
| pvalue_lr | pvalues from likelihood ratio test with restricted alternative hypothesis |
| pvalue_lr2 | pvalues from likelihood ratio test |

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