

Package ‘SNPRelate’

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Type Package

Title Parallel Computing Toolset for Relatedness and Principal Component Analysis of SNP Data

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Imports methods

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Enhances SeqArray (>= 1.12.0)

Description Genome-wide association studies (GWAS) are widely used to investigate the genetic basis of diseases and traits, but they pose many computational challenges. We developed an R package SNPRelate to provide a binary format for single-nucleotide polymorphism (SNP) data in GWAS utilizing CoreArray Genomic Data Structure (GDS) data files. The GDS format offers the efficient operations specifically designed for integers with two bits, since a SNP could occupy only two bits. SNPRelate is also designed to accelerate two key computations on SNP data using parallel computing for multi-core symmetric multiprocessing computer architectures: Principal Component Analysis (PCA) and relatedness analysis using Identity-By-Descent measures. The SNP GDS format is also used by the GWASTools package with the support of S4 classes and generic functions. The extended GDS format is implemented in the SeqArray package to support the storage of single nucleotide variations (SNVs), insertion/deletion polymorphism (indel) and structural variation calls in whole-genome and whole-exome variant data.

License GPL-3

VignetteBuilder knitr

LazyData true

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Index**106****Description**

Genome-wide association studies are widely used to investigate the genetic basis of diseases and traits, but they pose many computational challenges. We developed SNPRelate (R package for multi-core symmetric multiprocessing computer architectures) to accelerate two key computations on SNP data: principal component analysis (PCA) and relatedness analysis using identity-by-descent measures. The kernels of our algorithms are written in C/C++ and highly optimized.

Details

Package: SNPRelate
 Type: Package
 License: GPL version 3
 Depends: gdsfmt (>= 1.0.4)

The genotypes stored in GDS format can be analyzed by the R functions in SNPRelate, which utilize the multi-core feature of machine for a single computer.

Webpage: <https://github.com/zhengxwen/SNPRelate>, <http://corearray.sourceforge.net/>

Tutorial: <http://corearray.sourceforge.net/tutorials/SNPRelate/>

Author(s)

Xiuwen Zheng <zhengxwen@gmail.com>

References

Zheng X, Levine D, Shen J, Gogarten SM, Laurie C, Weir BS. A High-performance Computing Toolset for Relatedness and Principal Component Analysis of SNP Data. *Bioinformatics* (2012); doi: 10.1093/bioinformatics/bts610

Examples

```
#####
# Convert the PLINK BED file to the GDS file
#

# PLINK BED files
bed.fn <- system.file("extdata", "plinkhapmap.bed.gz", package="SNPRelate")
fam.fn <- system.file("extdata", "plinkhapmap.fam.gz", package="SNPRelate")
bim.fn <- system.file("extdata", "plinkhapmap.bim.gz", package="SNPRelate")

# convert
snpgdsBED2GDS(bed.fn, fam.fn, bim.fn, "HapMap.gds")

#####
# Principal Component Analysis
#

# open
genofile <- snpgdsOpen("HapMap.gds")

RV <- snpgdsPCA(genofile)
plot(RV$eigenvect[,2], RV$eigenvect[,1], xlab="PC 2", ylab="PC 1",
      col=rgb(0,0,150, 50, maxColorValue=255), pch=19)

# close the file
```

```

snpgdsClose(genofile)

#####
# Identity-By-Descent (IBD) Analysis
#

# open
genofile <- snpgdsOpen(snpgdsExampleFileName())

RV <- snpgdsIBDMoM(genofile)
flag <- lower.tri(RV$k0)
plot(RV$k0[flag], RV$k1[flag], xlab="k0", ylab="k1",
      col=rgb(0,0,150, 50, maxColorValue=255), pch=19)
abline(1, -1, col="red", lty=4)

# close the file
snpgdsClose(genofile)

#####
# Identity-By-State (IBS) Analysis
#

# open
genofile <- snpgdsOpen(snpgdsExampleFileName())

RV <- snpgdsIBS(genofile)
m <- 1 - RV$ibs
colnames(m) <- rownames(m) <- RV$sample.id
GeneticDistance <- as.dist(m[1:45, 1:45])
HC <- hclust(GeneticDistance, "ave")
plot(HC)

# close the file
snpgdsClose(genofile)

#####
# Linkage Disequilibrium (LD) Analysis
#

# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

snpset <- read.gdsn(index.gdsn(genofile, "snp.id"))[1:200]
L1 <- snpgdsLDMat(genofile, snp.id=snpset, method="composite", slide=-1)

# plot
image(abs(L1$LD), col=terrain.colors(64))

# close the file
snpgdsClose(genofile)

```

| | |
|-------------|--|
| hapmap_geno | <i>SNP genotypes of HapMap samples</i> |
|-------------|--|

Description

A list object including the following components:

sample.id – a vector of sample ids;

snp.id – a vector of SNP ids;

snp.position – a vector of SNP positions;

snp.chromosome – a vector of chromosome indices;

snp.allele – a character vector of “reference / non-reference”;

genotype – a “# of SNPs” X “# of samples” genotype matrix.

Usage

```
hapmap_geno
```

Value

A list

| | |
|-----------------|----------------------------------|
| snpgdsAdmixPlot | <i>Plot Ancestry Proportions</i> |
|-----------------|----------------------------------|

Description

Plot the admixture proportions according to their ancestries.

Usage

```
snpgdsAdmixPlot(propmat, group=NULL, col=NULL, multiplot=TRUE, showgrp=TRUE,
  shownum=TRUE, ylim=TRUE, na.rm=TRUE)
snpgdsAdmixTable(propmat, group, sort=FALSE)
```

Arguments

| | |
|-----------|---|
| propmat | a sample-by-ancestry matrix of proportion estimates, returned from snpgdsAdmixProp() |
| group | a character vector of a factor according to the rows in propmat |
| col | specify colors; if group is not specified, it is a color for each sample; otherwise specify colors for the groups |
| multiplot | single plot or multiple plots |
| showgrp | show group names in the plot; applicable when group is used |

| | |
|---------|--|
| shownum | TRUE: show the number of each group on the X-axis in the figure; applicable when group is used |
| ylim | TRUE: y-axis is limited to [0, 1]; FALSE: ylim <- range(propmat); a 2-length numeric vector: ylim used in plot() |
| na.rm | TRUE: remove the sample(s) according to the missing value(s) in group |
| sort | TRUE: rearranges the rows of proportion matrices into descending order |

Details

The minor allele frequency and missing rate for each SNP passed in `snp.id` are calculated over all the samples in `sample.id`.

Value

`snpgdsAdmixPlot()`: none.

`snpgdsAdmixTable()`: a list of data.frame consisting of group, num, mean, sd, min, max

Author(s)

Xiuwen Zheng

References

Zheng X, Weir BS. Eigenanalysis on SNP Data with an Interpretation of Identity by Descent. Theoretical Population Biology. 2015 Oct 23. pii: S0040-5809(15)00089-1. doi: 10.1016/j.tpb.2015.09.004.

See Also

[snpgdsEIGMIX](#), [snpgdsAdmixProp](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

# get population information
# or pop_code <- scan("pop.txt", what=character())
# if it is stored in a text file "pop.txt"
pop_code <- read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))

# get sample id
samp.id <- read.gdsn(index.gdsn(genofile, "sample.id"))

# run eigen-analysis
RV <- snpgdsEIGMIX(genofile)

# define groups
groups <- list(CEU = samp.id[pop_code == "CEU"],
              YRI = samp.id[pop_code == "YRI"],
              CHB = samp.id[is.element(pop_code, c("HCB", "JPT"))])
```

```

prop <- snpgdsAdmixProp(RV, groups=groups, bound=TRUE)

# draw
snpgdsAdmixPlot(prop, group=pop_code)

# use user-defined colors for the groups
snpgdsAdmixPlot(prop, group=pop_code, multiplot=FALSE, col=c(3,2,4))

snpgdsAdmixTable(prop, group=pop_code)

# close the genotype file
snpgdsClose(genofile)

```

snpgdsAdmixProp *Estimate ancestral proportions from the eigen-analysis*

Description

Estimate ancestral (admixture) proportions based on the eigen-analysis.

Usage

```
snpgdsAdmixProp(eigobj, groups, bound=FALSE)
```

Arguments

| | |
|--------|---|
| eigobj | an object of snpgdsEigMixClass from snpgdsEIGMIX , or an object of snpgdsPCAClass from snpgdsPCA |
| groups | a list of sample IDs, such like groups = list(CEU = c("NA0101", "NA1022", ...), YRI = c("NAxxxx", ...), Asia = c("NA1234", ...)) |
| bound | if TRUE, the estimates are bounded in [0, 1], and the sum of proportions is one; bound=FALSE for unbiased estimates |

Details

The minor allele frequency and missing rate for each SNP passed in `snp.id` are calculated over all the samples in `sample.id`.

Value

Return a matrix of ancestral proportions with rows for study individuals (`rownames()` is sample ID).

Author(s)

Xiuwen Zheng

References

Zheng X, Weir BS. Eigenanalysis on SNP Data with an Interpretation of Identity by Descent. *Theoretical Population Biology*. 2015 Oct 23. pii: S0040-5809(15)00089-1. doi: 10.1016/j.tpb.2015.09.004. [Epub ahead of print]

See Also

[snpGDEIGMIX](#), [snpGDSPCA](#), [snpGDSAdmixPlot](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())

# get population information
# or pop_code <- scan("pop.txt", what=character())
# if it is stored in a text file "pop.txt"
pop_code <- read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))

# get sample id
samp.id <- read.gdsn(index.gdsn(genofile, "sample.id"))

# run eigen-analysis
RV <- snpGDEIGMIX(genofile)

# eigenvalues
RV$eigenval

# make a data.frame
tab <- data.frame(sample.id = samp.id, pop = factor(pop_code),
  EV1 = RV$eigenvect[,1], # the first eigenvector
  EV2 = RV$eigenvect[,2], # the second eigenvector
  stringsAsFactors = FALSE)
head(tab)

# draw
plot(tab$EV2, tab$EV1, col=as.integer(tab$pop),
  xlab="eigenvector 2", ylab="eigenvector 1")
legend("bottomleft", legend=levels(tab$pop), pch="o", col=1:4)

# define groups
groups <- list(CEU = samp.id[pop_code == "CEU"],
  YRI = samp.id[pop_code == "YRI"],
  CHB = samp.id[is.element(pop_code, c("HCB", "JPT"))])

prop <- snpGDSAdmixProp(RV, groups=groups)
head(prop)

# draw
plot(prop[, "YRI"], prop[, "CEU"], col=as.integer(tab$pop),
  xlab = "Admixture Proportion from YRI",
```

```
      ylab = "Admixture Proportion from CEU")
abline(v=0, col="gray25", lty=2)
abline(h=0, col="gray25", lty=2)
abline(a=1, b=-1, col="gray25", lty=2)
legend("topright", legend=levels(tab$pop), pch="o", col=1:4)

# draw
snpGDSAdmixPlot(prop, group=pop_code)

# close the genotype file
snpGDSClose(genofile)
```

snpGDSAlleleSwitch *Allele-switching*

Description

Switch alleles according to the reference if needed.

Usage

```
snpGDSAlleleSwitch(gdsobj, A.allele, verbose=TRUE)
```

Arguments

| | |
|-----------------------|---|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>A.allele</code> | characters, referring to A allele |
| <code>verbose</code> | if TRUE, show information |

Value

A logical vector with TRUE indicating allele-switching and NA when it is unable to determine. NA occurs when `A.allele = NA` or `A.allele` is not in the list of alleles.

Author(s)

Xiuwen Zheng

Examples

```
# the file name of SNP GDS
(fn <- snpGDSExampleFileName())

# copy the file
file.copy(fn, "test.gds", overwrite=TRUE)

# open the SNP GDS file
```

```

genofile <- snpGDSOpen("test.gds", readonly=FALSE)

# allelic information
allele <- read.gdsn(index.gdsn(genofile, "snp.allele"))
allele.list <- strsplit(allele, "/")

A.allele <- sapply(allele.list, function(x) { x[1] })
B.allele <- sapply(allele.list, function(x) { x[2] })

set.seed(1000)
flag <- rep(FALSE, length(A.allele))
flag[sample.int(length(A.allele), 50, replace=TRUE)] <- TRUE

A.allele[flag] <- B.allele[flag]
A.allele[sample.int(length(A.allele), 10, replace=TRUE)] <- NA
table(A.allele, exclude=NULL)

# allele switching
z <- snpGDSAlleleSwitch(genofile, A.allele)

table(z, exclude=NULL)

# close the file
snpGDSClose(genofile)

# delete the temporary file
unlink("test.gds", force=TRUE)

```

snpGDSApartSelection *Select SNPs with a basepair distance*

Description

Randomly selects SNPs for which each pair is at least as far apart as the specified basepair distance.

Usage

```
snpGDSApartSelection(chromosome, position, min.dist=100000,
                     max.n.snp.perchr=-1, verbose=TRUE)
```

Arguments

| | |
|------------|---|
| chromosome | chromosome codes |
| position | SNP positions in base pair |
| min.dist | A numeric value to specify minimum distance required (in basepairs) |

max.n.snp.perchr A numeric value specifying the maximum number of SNPs to return per chromosome, "-1" means no number limit

verbose if TRUE, show information

Value

A logical vector indicating which SNPs were selected.

Author(s)

Xiuwen Zheng

See Also

[snpGDSLdPruning](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())
genofile

chr <- read.gdsn(index.gdsn(genofile, "snp.chromosome"))
pos <- read.gdsn(index.gdsn(genofile, "snp.position"))

set.seed(1000)
flag <- snpGDSApartSelection(chr, pos, min.dist=250000, verbose=TRUE)
table(flag)

# close the genotype file
snpGSDSClose(genofile)
```

snpGDSBED2GDS

Conversion from PLINK BED to GDS

Description

Convert a PLINK binary ped file to a GDS file.

Usage

```
snpGDSBED2GDS.bed.fn, fam.fn, bim.fn, out.gdsfn, family=FALSE,
snpfirstdim=NA, compress.annotation="LZMA_RA", compress.geno="",
option=NULL, cvt.chr=c("int", "char"), cvt.snpid=c("auto", "int"),
verbose=TRUE)
```

Arguments

| | |
|---------------------|---|
| bed.fn | the file name of binary file, genotype information |
| fam.fn | the file name of first six columns of ".ped"; if it is missing, ".fam" is added to bed.fn |
| bim.fn | the file name of extended MAP file: two extra columns = allele names; if it is missing, ".bim" is added to bim.fn |
| out.gdsfn | the output file name of GDS file |
| family | if TRUE, to include family information in the sample annotation |
| snpfirstdim | if TRUE, genotypes are stored in the individual-major mode, (i.e, list all SNPs for the first individual, and then list all SNPs for the second individual, etc); NA, the dimension is determined by the BED file |
| compress.annotation | the compression method for the GDS variables, except "genotype"; optional values are defined in the function add.gdsn |
| compress.geno | the compression method for "genotype"; optional values are defined in the function add.gdsn |
| option | NULL or an object from snpGDSOption , see details |
| cvt.chr | "int" – chromosome code in the GDS file is integer; "char" – chromosome code in the GDS file is character |
| cvt.snpid | "int" – to create an integer snp.id starting from 1; "auto" – if SNP IDs in the PLINK file are not unique, to create an an integer snp.id, otherwise to use SNP IDs for snp.id |
| verbose | if TRUE, show information |

Details

GDS – Genomic Data Structures, the extended file name used for storing genetic data, and the file format is used in the [gdsfmt](#) package.

BED – the PLINK binary ped format.

The user could use `option` to specify the range of code for autosomes. For humans there are 22 autosomes (from 1 to 22), but dogs have 38 autosomes. Note that the default settings are used for humans. The user could call `option = snpGDSOption(autosome.end=38)` for importing the BED file of dog. It also allow define new chromosome coding, e.g., `option = snpGDSOption(Z=27)`.

Value

Return the file name of GDS format with an absolute path.

Author(s)

Xiuwen Zheng

References

Purcell S, Neale B, Todd-Brown K, Thomas L, Ferreira MAR, Bender D, Maller J, Sklar P, de Bakker PIW, Daly MJ & Sham PC. 2007. PLINK: a toolset for whole-genome association and population-based linkage analysis. *American Journal of Human Genetics*, 81.

See Also

[snpgdsOption](#), [snpgdsPED2GDS](#), [snpgdsGDS2PED](#)

Examples

```
# PLINK BED files
bed.fn <- system.file("extdata", "plinkhapmap.bed.gz", package="SNPRelate")
fam.fn <- system.file("extdata", "plinkhapmap.fam.gz", package="SNPRelate")
bim.fn <- system.file("extdata", "plinkhapmap.bim.gz", package="SNPRelate")

# convert
snpgdsBED2GDS(bed.fn, fam.fn, bim.fn, "HapMap.gds")

# open
genofile <- snpgdsOpen("HapMap.gds")
genofile

# close
snpgdsClose(genofile)

# delete the temporary file
unlink("HapMap.gds", force=TRUE)
```

snpgdsClose

Close the SNP GDS File

Description

Close the SNP GDS file

Usage

```
snpgdsClose(gdsobj)
```

Arguments

gdsobj an object of class [SNPGDSFileClass](#), a SNP GDS file

Details

It is suggested to call `snpgdsClose` instead of `closefn.gds`.

Value

None.

Author(s)

Xiuwen Zheng

See Also

[snpgdsOpen](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

genofile

# close the file
snpgdsClose(genofile)
```

snpgdsCombineGeno *Merge SNP datasets*

Description

To merge GDS files of SNP genotypes into a single GDS file

Usage

```
snpgdsCombineGeno(gds.fn, out.fn, method=c("position", "exact"),
  compress.annotation="ZIP_RA.MAX", compress.geno="ZIP_RA",
  same.strand=FALSE, snpfirstdim=FALSE, verbose=TRUE)
```

Arguments

| | |
|---------------------|--|
| gds.fn | a character vector of GDS file names to be merged |
| out.fn | the name of output GDS file |
| method | "exact": matching by all snp.id, chromosomes, positions and alleles; "position": matching by chromosomes and positions |
| compress.annotation | the compression method for the variables except genotype |
| compress.geno | the compression method for the variable genotype |
| same.strand | if TRUE, assuming the alleles on the same strand |
| snpfirstdim | if TRUE, genotypes are stored in the individual-major mode, (i.e, list all SNPs for the first individual, and then list all SNPs for the second individual, etc) |
| verbose | if TRUE, show information |

Details

This function calls [snpgdsSNPListIntersect](#) internally to determine the common SNPs. Allele definitions are taken from the first GDS file.

Value

None.

Author(s)

Xiuwen Zheng

See Also

[snpgdsCreateGeno](#), [snpgdsCreateGenoSet](#), [snpgdsSNPList](#), [snpgdsSNPListIntersect](#)

Examples

```
# get the file name of a gds file
fn <- snpgdsExampleFileName()

f <- snpgdsOpen(fn)
samp_id <- read.gdsn(index.gdsn(f, "sample.id"))
snp_id <- read.gdsn(index.gdsn(f, "snp.id"))
geno <- read.gdsn(index.gdsn(f, "genotype"), start=c(1,1), count=c(-1, 3000))
snpgdsClose(f)

# split the GDS file with different samples
snpgdsCreateGenoSet(fn, "t1.gds", sample.id=samp_id[1:10],
  snp.id=snp_id[1:3000])
snpgdsCreateGenoSet(fn, "t2.gds", sample.id=samp_id[11:30],
  snp.id=snp_id[1:3000])

# combine with different samples
snpgdsCombineGeno(c("t1.gds", "t2.gds"), "test.gds", same.strand=TRUE)
f <- snpgdsOpen("test.gds")
g <- read.gdsn(index.gdsn(f, "genotype"))
snpgdsClose(f)

identical(geno[1:30, ], g) # TRUE

# split the GDS file with different SNPs
snpgdsCreateGenoSet(fn, "t1.gds", snp.id=snp_id[1:100])
snpgdsCreateGenoSet(fn, "t2.gds", snp.id=snp_id[101:300])

# combine with different SNPs
snpgdsCombineGeno(c("t1.gds", "t2.gds"), "test.gds")
f <- snpgdsOpen("test.gds")
g <- read.gdsn(index.gdsn(f, "genotype"))
snpgdsClose(f)
```



```

identical(geno[, 1:300], g) # TRUE

# delete the temporary files
unlink(c("t1.gds", "t2.gds", "t3.gds", "t4.gds", "test.gds"), force=TRUE)

```

snpgdsCreateGeno *Create a SNP genotype dataset from a matrix*

Description

To create a GDS file of genotypes from a matrix.

Usage

```

snpgdsCreateGeno(gds.fn, genmat, sample.id=NULL, snp.id=NULL, snp.rs.id=NULL,
  snp.chromosome=NULL, snp.position=NULL, snp.allele=NULL, snpfirstdim=TRUE,
  compress.annotation="ZIP_RA.max", compress.geno="", other.vars=NULL)

```

Arguments

| | |
|---------------------|--|
| gds.fn | the file name of gds |
| genmat | a matrix of genotypes |
| sample.id | the sample ids, which should be unique |
| snp.id | the SNP ids, which should be unique |
| snp.rs.id | the rs ids for SNPs, which can be not unique |
| snp.chromosome | the chromosome indices |
| snp.position | the SNP positions in basepair |
| snp.allele | the reference/non-reference alleles |
| snpfirstdim | if TRUE, genotypes are stored in the individual-major mode, (i.e, list all SNPs for the first individual, and then list all SNPs for the second individual, etc) |
| compress.annotation | the compression method for the variables except genotype |
| compress.geno | the compression method for the variable genotype |
| other.vars | a list object storing other variables |

Details

There are possible values stored in the variable genmat: 0, 1, 2 and other values. "0" indicates two B alleles, "1" indicates one A allele and one B allele, "2" indicates two A alleles, and other values indicate a missing genotype.

If snpfirstdim is TRUE, then genmat should be "# of SNPs X # of samples"; if snpfirstdim is FALSE, then genmat should be "# of samples X # of SNPs".

The typical variables specified in other.vars are "sample.annot" and "snp.annot", which are data.frame objects.

Value

None.

Author(s)

Xiuwen Zheng

See Also

[snpgdsCreateGenoSet](#), [snpgdsCombineGeno](#)

Examples

```
# load data
data(hapmap_geno)

# create a gds file
with(hapmap_geno, snpgdsCreateGeno("test.gds", genmat=genotype,
  sample.id=sample.id, snp.id=snp.id, snp.chromosome=snp.chromosome,
  snp.position=snp.position, snp.allele=snp.allele, snpfirstdim=TRUE))

# open the gds file
genofile <- snpgdsOpen("test.gds")

RV <- snpgdsPCA(genofile)
plot(RV$eigenvect[,2], RV$eigenvect[,1], xlab="PC 2", ylab="PC 1")

# close the file
snpgdsClose(genofile)
```

snpgdsCreateGenoSet *Create a SNP genotype dataset from a GDS file*

Description

To create a GDS file of genotypes from a specified GDS file.

Usage

```
snpgdsCreateGenoSet(src.fn, dest.fn, sample.id=NULL, snp.id=NULL,
  snpfirstdim=NULL, compress.annotation="ZIP_RA.max", compress.geno="",
  verbose=TRUE)
```

Arguments

src.fn the file name of a specified GDS file
 dest.fn the file name of output GDS file
 sample.id a vector of sample id specifying selected samples; if NULL, all samples are used
 snp.id a vector of snp id specifying selected SNPs; if NULL, all SNPs are used
 snpfirstdim if TRUE, genotypes are stored in the individual-major mode, (i.e, list all SNPs for the first individual, and then list all SNPs for the second individual, etc)
 compress.annotation the compression method for the variables except genotype
 compress.geno the compression method for the variable genotype
 verbose if TRUE, show information

Value

None.

Author(s)

Xiuwen Zheng

See Also

[snpgdsCreateGeno](#), [snpgdsCombineGeno](#)

Examples

```

# open an example dataset (HapMap)
(genofile <- snpgdsOpen(snpgdsExampleFileName()))
# + [ ] *
# |--+ sample.id { VStr8 279 ZIP(29.9%), 679B }
# |--+ snp.id { Int32 9088 ZIP(34.8%), 12.3K }
# |--+ snp.rs.id { VStr8 9088 ZIP(40.1%), 36.2K }
# |--+ snp.position { Int32 9088 ZIP(94.7%), 33.6K }
# |--+ snp.chromosome { UInt8 9088 ZIP(0.94%), 85B } *
# |--+ snp.allele { VStr8 9088 ZIP(11.3%), 4.0K }
# |--+ genotype { Bit2 279x9088, 619.0K } *
# \--+ sample.annot [ data.frame ] *
# |---+ family.id { VStr8 279 ZIP(34.4%), 514B }
# |---+ father.id { VStr8 279 ZIP(31.5%), 220B }
# |---+ mother.id { VStr8 279 ZIP(30.9%), 214B }
# |---+ sex { VStr8 279 ZIP(17.0%), 95B }
# \--+ pop.group { VStr8 279 ZIP(6.18%), 69B }

set.seed(1000)
snpset <- unlist(snpgdsLDpruning(genofile))
length(snpset)
# 6547

# close the file

```

```

snpgdsClose(genofile)

snpgdsCreateGenoSet(snpgdsExampleFileName(), "test.gds", snp.id=snpset)

#####
# check

(gfile <- snpgdsOpen("test.gds"))
# + [ ] *
# |--+ sample.id { Str8 279 ZIP_ra(31.2%), 715B }
# |--+ snp.id { Int32 6547 ZIP_ra(34.9%), 8.9K }
# |--+ snp.rs.id { Str8 6547 ZIP_ra(41.5%), 27.1K }
# |--+ snp.position { Int32 6547 ZIP_ra(94.9%), 24.3K }
# |--+ snp.chromosome { Int32 6547 ZIP_ra(0.45%), 124B }
# |--+ snp.allele { Str8 6547 ZIP_ra(11.5%), 3.0K }
# \--+ genotype { Bit2 279x6547, 446.0K } *

# close the file
snpgdsClose(gfile)

unlink("test.gds", force=TRUE)

```

snpgdsCutTree

Determine clusters of individuals

Description

To determine sub groups of individuals using a specified dendrogram from hierarchical cluster analysis

Usage

```

snpgdsCutTree(hc, z.threshold=15, outlier.n=5, n.perm = 5000, samp.group=NULL,
  col.outlier="red", col.list=NULL, pch.outlier=4, pch.list=NULL,
  label.H=FALSE, label.Z=TRUE, verbose=TRUE)

```

Arguments

| | |
|-------------|--|
| hc | an object of snpgdsHCluster |
| z.threshold | the threshold of Z score to determine whether split the node or not |
| outlier.n | the cluster with size less than or equal to outlier.n is considered as outliers |
| n.perm | the times for permutation |
| samp.group | if NULL, determine groups by Z score; if a vector of factor, assign each individual in dendrogram with respect to samp.group |
| col.outlier | the color of outlier |
| col.list | the list of colors for different clusters |

| | |
|-------------|---|
| pch.outlier | plotting 'character' for outliers |
| pch.list | plotting 'character' for different clusters |
| label.H | if TRUE, plotting heights in a dendrogram |
| label.Z | if TRUE, plotting Z scores in a dendrogram |
| verbose | if TRUE, show information |

Details

The details will be described in future.

Value

Return a list:

| | |
|-------------|--|
| sample.id | the sample ids used in the analysis |
| z.threshold | the threshold of Z score to determine whether split the node or not |
| outlier.n | the cluster with size less than or equal to outlier.n is considered as outliers |
| samp.order | the order of samples in the dendrogram |
| samp.group | a vector of factor, indicating the group of each individual |
| dmat | a matrix of pairwise group dissimilarity |
| dendrogram | the dendrogram of individuals |
| merge | a data.frame of (z, n1, n2) describing each combination: z, the Z score; n1, the size of the first cluster; n2, the size of the second cluster |
| clust.count | the counts for clusters |

Author(s)

Xiuwen Zheng

See Also

[snpgdsHCluster](#), [snpgdsDrawTree](#), [snpgdsIBS](#), [snpgdsDiss](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

pop.group <- as.factor(read.gdsn(index.gdsn(
  genofile, "sample.annot/pop.group")))
pop.level <- levels(pop.group)

diss <- snpgdsDiss(genofile)
hc <- snpgdsHCluster(diss)

# close the genotype file
snpgdsClose(genofile)
```

```
#####
# cluster individuals
#

set.seed(100)
rv <- snpgdsCutTree(hc, label.H=TRUE, label.Z=TRUE)

# the distribution of Z scores
snpgdsDrawTree(rv, type="z-score", main="HapMap Phase II")

# draw dendrogram
snpgdsDrawTree(rv, main="HapMap Phase II",
  edgePar=list(col=rgb(0.5,0.5,0.5, 0.75), t.col="black"))

#####
# or cluster individuals by ethnic information
#

rv2 <- snpgdsCutTree(hc, samp.group=pop.group)

# cluster individuals by Z score, specifying 'clust.count'
snpgdsDrawTree(rv2, rv$clust.count, main="HapMap Phase II",
  edgePar = list(col=rgb(0.5,0.5,0.5, 0.75), t.col="black"),
  labels = c("YRI", "CHB/JPT", "CEU"), y.label=0.1)
legend("bottomleft", legend=levels(pop.group), col=1:nlevels(pop.group),
  pch=19, ncol=4, bg="white")

#####
# zoom in ...
#

snpgdsDrawTree(rv2, rv$clust.count, dend.idx = c(1),
  main="HapMap Phase II -- YRI",
  edgePar=list(col=rgb(0.5,0.5,0.5, 0.75), t.col="black"),
  y.label.kinship=TRUE)

snpgdsDrawTree(rv2, rv$clust.count, dend.idx = c(2,2),
  main="HapMap Phase II -- CEU",
  edgePar=list(col=rgb(0.5,0.5,0.5, 0.75), t.col="black"),
  y.label.kinship=TRUE)

snpgdsDrawTree(rv2, rv$clust.count, dend.idx = c(2,1),
  main="HapMap Phase II -- CHB/JPT",
  edgePar=list(col=rgb(0.5,0.5,0.5, 0.75), t.col="black"),
  y.label.kinship=TRUE)
```

snpGDSdiss *Individual dissimilarity analysis*

Description

Calculate the individual dissimilarities for each pair of individuals.

Usage

```
snpGDSdiss(gdsobj, sample.id=NULL, snp.id=NULL, autosome.only=TRUE,
           remove.monosnp=TRUE, maf=NaN, missing.rate=NaN, num.thread=1, verbose=TRUE)
```

Arguments

| | |
|----------------|--|
| gdsobj | an object of class SNPGDSFileClass , a SNP GDS file |
| sample.id | a vector of sample id specifying selected samples; if NULL, all samples are used |
| snp.id | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| autosome.only | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| remove.monosnp | if TRUE, remove monomorphic SNPs |
| maf | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| missing.rate | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| num.thread | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| verbose | if TRUE, show information |

Details

The minor allele frequency and missing rate for each SNP passed in `snp.id` are calculated over all the samples in `sample.id`.

`snpGDSdiss()` returns $1 - \beta_{ij}$ which is formally described in Weir&Goudet (2017).

Value

Return a class "snpGDSdissClass":

| | |
|-----------|--------------------------------------|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| diss | a matrix of individual dissimilarity |

Author(s)

Xiuwen Zheng

References

Zheng, Xiuwen. 2013. Statistical Prediction of HLA Alleles and Relatedness Analysis in Genome-Wide Association Studies. PhD dissertation, the department of Biostatistics, University of Washington.

Weir BS, Zheng X. SNPs and SNVs in Forensic Science. 2015. Forensic Science International: Genetics Supplement Series.

Weir BS, Goudet J. A Unified Characterization of Population Structure and Relatedness. *Genetics*. 2017 Aug;206(4):2085-2103. doi: 10.1534/genetics.116.198424.

See Also

[snpGDSHCluster](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())

pop.group <- as.factor(read.gdsn(index.gdsn(
  genofile, "sample.annot/pop.group")))
pop.level <- levels(pop.group)

diss <- snpGDSDisss(genofile)
diss
hc <- snpGDSHCluster(diss)
names(hc)
plot(hc$dendrogram)

# close the genotype file
snpGSDSClose(genofile)

# split
set.seed(100)
rv <- snpGDSCutTree(hc, label.H=TRUE, label.Z=TRUE)

# draw dendrogram
snpGDSDrawTree(rv, main="HapMap Phase II",
  edgePar=list(col=rgb(0.5,0.5,0.5, 0.75), t.col="black"))
```

snpGDSDrawTree

Draw a dendrogram

Description

To draw a dendrogram or the distribution of Z scores

Usage

```
snpGdsDrawTree(obj, clust.count=NULL, dend.idx=NULL,
  type=c("dendrogram", "z-score"), yaxis.height=TRUE, yaxis.kinship=TRUE,
  y.kinship.baseline=NaN, y.label.kinship=FALSE, outlier.n=NULL,
  shadow.col=c(rgb(0.5, 0.5, 0.5, 0.25), rgb(0.5, 0.5, 0.5, 0.05)),
  outlier.col=rgb(1, 0.50, 0.50, 0.5), leaflab="none",
  labels=NULL, y.label=0.2, ...)
```

Arguments

| | |
|---------------------------------|--|
| <code>obj</code> | an object returned by snpGdsCutTree |
| <code>clust.count</code> | the counts for clusters, drawing shadows |
| <code>dend.idx</code> | the index of sub tree, plot <code>obj\$dendrogram[[dend.idx]]</code> , or <code>NULL</code> for the whole tree |
| <code>type</code> | "dendrogram", draw a dendrogram; or "z-score", draw the distribution of Z score |
| <code>yaxis.height</code> | if <code>TRUE</code> , draw the left Y axis: height of tree |
| <code>yaxis.kinship</code> | if <code>TRUE</code> , draw the right Y axis: kinship coefficient |
| <code>y.kinship.baseline</code> | the baseline value of kinship; if <code>NaN</code> , it is the height of the first split from top in a dendrogram; only works when <code>yaxis.kinship = TRUE</code> |
| <code>y.label.kinship</code> | if <code>TRUE</code> , show 'PO/FS' etc on the right axis |
| <code>outlier.n</code> | the cluster with size less than or equal to <code>outlier.n</code> is considered as outliers; if <code>NULL</code> , let <code>outlier.n = obj\$outlier.n</code> |
| <code>shadow.col</code> | two colors for shadow |
| <code>outlier.col</code> | the colors for outliers |
| <code>leaflab</code> | a string specifying how leaves are labeled. The default "perpendicular" write text vertically (by default). "textlike" writes text horizontally (in a rectangle), and "none" suppresses leaf labels. |
| <code>labels</code> | the legend for different regions |
| <code>y.label</code> | y positions of labels |
| <code>...</code> | Arguments to be passed to the method " <code>plot(, ...)</code> ", such as graphical parameters. |

Details

The details will be described in future.

Value

None.

Author(s)

Xiuwen Zheng

See Also

[snpgdsCutTree](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

pop.group <- as.factor(read.gdsn(index.gdsn(
  genofile, "sample.annot/pop.group")))
pop.level <- levels(pop.group)

diss <- snpgdsDiss(genofile)
hc <- snpgdsHCluster(diss)

# close the genotype file
snpgdsClose(genofile)

# split
set.seed(100)
rv <- snpgdsCutTree(hc, label.H=TRUE, label.Z=TRUE)

# draw dendrogram
snpgdsDrawTree(rv, main="HapMap Phase II",
  edgePar=list(col=rgb(0.5,0.5,0.5, 0.75), t.col="black"))
```

snpgdsEIGMIX

Eigen-analysis on SNP genotype data

Description

Eigen-analysis on IBD matrix based SNP genotypes.

Usage

```
snpgdsEIGMIX(gdsobj, sample.id=NULL, snp.id=NULL, autosome.only=TRUE,
  remove.monosnp=TRUE, maf=NaN, missing.rate=NaN, num.thread=1L,
  eigen.cnt=32L, diagadj=TRUE, ibdmat=FALSE, verbose=TRUE)
## S3 method for class 'snpgdsEigMixClass'
plot(x, eig=c(1L,2L), ...)
```

Arguments

| | |
|------------------------|--|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |

| | |
|-----------------------------|---|
| <code>autosome.only</code> | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| <code>remove.monosnp</code> | if TRUE, remove monomorphic SNPs |
| <code>maf</code> | to use the SNPs with " <code>>= maf</code> " only; if NaN, no MAF threshold |
| <code>missing.rate</code> | to use the SNPs with " <code><= missing.rate</code> " only; if NaN, no missing threshold |
| <code>num.thread</code> | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| <code>eigen.cnt</code> | output the number of eigenvectors; if <code>eigen.cnt < 0</code> , returns all eigenvectors; if <code>eigen.cnt==0</code> , no eigen calculation |
| <code>diagadj</code> | TRUE for diagonal adjustment by default |
| <code>ibdmat</code> | if TRUE, returns the IBD matrix |
| <code>verbose</code> | if TRUE, show information |
| <code>x</code> | a <code>snpGDS<i>EigMixClass</i></code> object |
| <code>eig</code> | indices of eigenvectors, like <code>1:2</code> or <code>1:4</code> |
| <code>...</code> | the arguments passed to or from other methods, like <code>pch</code> , <code>col</code> |

Value

Return a `snpGDSEigMixClass` object, and it is a list:

| | |
|------------------------|--|
| <code>sample.id</code> | the sample ids used in the analysis |
| <code>snp.id</code> | the SNP ids used in the analysis |
| <code>eigenval</code> | eigenvalues |
| <code>eigenvect</code> | eigenvectors, "# of samples" x "eigen.cnt" |
| <code>afreq</code> | allele frequencies |
| <code>ibd</code> | the IBD matrix when <code>ibdmat=TRUE</code> |
| <code>diagadj</code> | the argument <code>diagadj</code> |

Author(s)

Xiuwen Zheng

References

Zheng X, Weir BS. Eigenanalysis on SNP Data with an Interpretation of Identity by Descent. *Theoretical Population Biology*. 2016 Feb;107:65-76. doi: 10.1016/j.tpb.2015.09.004

See Also

[snpGDS*AdmixProp*](#), [snpGDS*AdmixPlot*](#), [snpGDS*PCA*](#), [snpGDS*PCASNPLoading*](#), [snpGDS*PCASampLoading*](#)

Examples

```

# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())

# get population information
# or pop_code <- scan("pop.txt", what=character())
# if it is stored in a text file "pop.txt"
pop_code <- read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))

# get sample id
samp.id <- read.gdsn(index.gdsn(genofile, "sample.id"))

# run eigen-analysis
RV <- snpGDS_EIGMIX(genofile)
RV

# eigenvalues
RV$eigenval

# make a data.frame
tab <- data.frame(sample.id = samp.id, pop = factor(pop_code),
  EV1 = RV$eigenvect[,1], # the first eigenvector
  EV2 = RV$eigenvect[,2], # the second eigenvector
  stringsAsFactors = FALSE)
head(tab)

# draw
plot(tab$EV2, tab$EV1, col=as.integer(tab$pop),
  xlab="eigenvector 2", ylab="eigenvector 1")
legend("topleft", legend=levels(tab$pop), pch="o", col=1:4)

# define groups
groups <- list(CEU = samp.id[pop_code == "CEU"],
  YRI = samp.id[pop_code == "YRI"],
  CHB = samp.id[is.element(pop_code, c("HCB", "JPT"))])

prop <- snpGDSAdmixProp(RV, groups=groups)

# draw
plot(prop[, "YRI"], prop[, "CEU"], col=as.integer(tab$pop),
  xlab = "Admixture Proportion from YRI",
  ylab = "Admixture Proportion from CEU")
abline(v=0, col="gray25", lty=2)
abline(h=0, col="gray25", lty=2)
abline(a=1, b=-1, col="gray25", lty=2)
legend("topright", legend=levels(tab$pop), pch="o", col=1:4)

# close the genotype file
snpGDS_Close(genofile)

```

| | |
|--------------|---------------------------------------|
| snpGdsErrMsg | <i>Get the last error information</i> |
|--------------|---------------------------------------|

Description

Return the last error message.

Usage

```
snpGdsErrMsg()
```

Value

Characters

Author(s)

Xiuwen Zheng

Examples

```
snpGdsErrMsg()
```

| | |
|-----------------------|-------------------------|
| snpGdsExampleFileName | <i>Example GDS file</i> |
|-----------------------|-------------------------|

Description

Return the file name of example data

Usage

```
snpGdsExampleFileName()
```

Details

A GDS genotype file was created from a subset of HapMap Phase II dataset consisting of 270 individuals and duplicates.

Value

Characters

Author(s)

Xiuwen Zheng

Examples

```
snpGdsExampleFileName()
```

| | |
|-----------------|------------------------|
| snpGDSFileClass | <i>SNPGDSFileClass</i> |
|-----------------|------------------------|

Description

A `SNPGDSFileClass` object provides access to a GDS file containing genome-wide SNP data. It extends the class `gds.class` in the `gdsfmt` package.

Author(s)

Xiuwen Zheng

See Also

[snpGdsOpen](#), [snpGdsClose](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())
genofile

class(genofile)
# "SNPGDSFileClass" "gds.class"

# close the file
snpGdsClose(genofile)
```

| | |
|-----------|--|
| snpGdsFst | <i>F-statistics (fixation indices)</i> |
|-----------|--|

Description

Calculate relatedness measures F-statistics (also known as fixation indices) for given populations

Usage

```
snpGdsFst(gdsobj, population, method=c("W&C84", "W&H02"), sample.id=NULL,
  snp.id=NULL, autosome.only=TRUE, remove.monosnp=TRUE, maf=NaN,
  missing.rate=NaN, with.id=FALSE, verbose=TRUE)
```

Arguments

| | |
|----------------|---|
| gdsobj | an object of class <code>SNPGDSFileClass</code> , a SNP GDS file |
| population | a factor, indicating population information for each individual |
| method | "W&C84" – Fst estimator in Weir & Cockerham 1984 (by default), "W&H02" – relative beta estimator in Weir & Hill 2002, see details |
| sample.id | a vector of sample id specifying selected samples; if NULL, all samples are used |
| snp.id | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| autosome.only | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| remove.monosnp | if TRUE, remove monomorphic SNPs |
| maf | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| missing.rate | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| with.id | if TRUE, the returned value with sample.id and sample.id |
| verbose | if TRUE, show information |

Details

The minor allele frequency and missing rate for each SNP passed in `snp.id` are calculated over all the samples in `sample.id`.

The "W&H02" option implements the calculation in Buckleton et. al. 2016.

Value

Return a list:

| | |
|-----------|--|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| Fst | weighted Fst estimate |
| MeanFst | the average of Fst estimates across SNPs |
| FstSNP | a vector of Fst for each SNP |
| Beta | Beta matrix |

Author(s)

Xiuwen Zheng

References

- Weir, BS. & Cockerham, CC. Estimating F-statistics for the analysis of population structure. (1984).
 Weir, BS. & Hill, WG. Estimating F-statistics. Annual review of genetics 36, 721-50 (2002).
 Population-specific FST values for forensic STR markers: A worldwide survey. Buckleton J, Curran J, Goudet J, Taylor D, Thiery A, Weir BS. Forensic Sci Int Genet. 2016 Jul;23:91-100. doi: 10.1016/j.fsigen.2016.03.004.

Examples

```

# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

group <- as.factor(read.gdsn(index.gdsn(
  genofile, "sample.annot/pop.group")))

# Fst estimation
v <- snpgdsFst(genofile, population=group, method="W&C84")
v$Fst
v$MeanFst
summary(v$FstSNP)

# or
v <- snpgdsFst(genofile, population=group, method="W&H02")
v$Fst
v$MeanFst
v$Beta
summary(v$FstSNP)

# close the genotype file
snpgdsClose(genofile)

```

snpgdsGDS2BED

Conversion from GDS to PLINK BED

Description

Convert a GDS file to a PLINK binary ped (BED) file.

Usage

```
snpgdsGDS2BED(gdsobj, bed.fn, sample.id=NULL, snp.id=NULL, snpfirstdim=NULL,
  verbose=TRUE)
```

Arguments

| | |
|-------------|--|
| gdsobj | an object of class SNPGDSFileClass , a SNP GDS file; or characters, the file name of GDS |
| bed.fn | the file name of output, without the filename extension ".bed" |
| sample.id | a vector of sample id specifying selected samples; if NULL, all samples are used |
| snp.id | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| snpfirstdim | if TRUE, genotypes are stored in the individual-major mode, (i.e, list all SNPs for the first individual, and then list all SNPs for the second individual, etc); if NULL, determine automatically |
| verbose | if TRUE, show information |

Details

GDS – Genomic Data Structures, the extended file name used for storing genetic data, and the file format used in the [gdsfmt](#) package.

BED – the PLINK binary ped format.

Value

None.

Author(s)

Xiuwen Zheng

References

Purcell S, Neale B, Todd-Brown K, Thomas L, Ferreira MAR, Bender D, Maller J, Sklar P, de Bakker PIW, Daly MJ & Sham PC. 2007. PLINK: a toolset for whole-genome association and population-based linkage analysis. *American Journal of Human Genetics*, 81.

<http://corearray.sourceforge.net/>

See Also

[snpGDS2BED](#), [snpGDS2PED](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())

snpset <- snpGDSSelectSNP(genofile, missing.rate=0.95)
snpGDS2BED(genofile, bed.fn="test", snp.id=snpset)

# close the genotype file
snpGDSClose(genofile)

# delete the temporary files
unlink(c("test.bed", "test.bim", "test.fam"), force=TRUE)
```

snpGDS2Eigen

Conversion from GDS to Eigen (EIGENSTRAT)

Description

Convert a GDS file to an EIGENSTRAT file.

Usage

```
snpGDS2Eigen(gdsobj, eigen.fn, sample.id=NULL, snp.id=NULL, verbose=TRUE)
```

Arguments

| | |
|------------------------|--|
| <code>gdsobj</code> | an object of class <code>SNPGDSFileClass</code> , a SNP GDS file |
| <code>eigen.fn</code> | the file name of EIGENSTRAT |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| <code>verbose</code> | if TRUE, show information |

Details

GDS – Genomic Data Structures, the extended file name used for storing genetic data, and the file format used in the `gdsfmt` package.

Eigen – the text format used in EIGENSTRAT.

Value

None.

Author(s)

Xiuwen Zheng

References

Patterson N, Price AL, Reich D (2006) Population structure and eigenanalysis. *PLoS Genetics* 2:e190.

Price AL, Patterson NJ, Plenge RM, Weinblatt ME, Shadick NA, Reich D (2006) Principal components analysis corrects for stratification in genome-wide association studies. *Nat Genet.* 38, 904-909.

<http://corearray.sourceforge.net/>

See Also

[snpGDS2PED](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())

snpset <- snpGDSSelectSNP(genofile, missing.rate=0.95)
snpGDS2Eigen(genofile, eigen.fn="tmpeigen", snp.id=snpset)

# close the genotype file
snpGSDSClose(genofile)
```

```
# delete the temporary files
unlink(c("tmpeigen.eigenstratgeno", "tmpeigen.ind", "tmpeigen.snp"), force=TRUE)
```

snpGDS2PED *Conversion from GDS to PED*

Description

Convert a GDS file to a PLINK text ped file.

Usage

```
snpGDS2PED(gdsobj, ped.fn, sample.id=NULL, snp.id=NULL, use.snp.rs.id=TRUE,
           format=c("A/G/C/T", "A/B", "1/2"), verbose=TRUE)
```

Arguments

| | |
|---------------|--|
| gdsobj | a GDS file object (gds.class) |
| ped.fn | the file name of output |
| sample.id | a vector of sample id specifying selected samples; if NULL, all samples are used |
| snp.id | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| use.snp.rs.id | if TRUE, use "snp.rs.id" instead of "snp.id" if available |
| format | specify the coding: "A/G/C/T" – allelic codes stored in "snp.allele" of the GDS file; "A/B" – A and B codes; "1/2" – 1 and 2 codes |
| verbose | if TRUE, show information |

Details

GDS – Genomic Data Structures, the extended file name used for storing genetic data, and the file format used in the [gdsfmt](#) package.

PED – the PLINK text ped format.

Value

None.

Author(s)

Xiuwen Zheng

References

Purcell S, Neale B, Todd-Brown K, Thomas L, Ferreira MAR, Bender D, Maller J, Sklar P, de Bakker PIW, Daly MJ & Sham PC. 2007. PLINK: a toolset for whole-genome association and population-based linkage analysis. *American Journal of Human Genetics*, 81.

<http://corearray.sourceforge.net/>

See Also[snpgdsGDS2BED](#)**Examples**

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

# GDS ==> PED
snpgdsGDS2PED(genofile, ped.fn="tmp")

# close the GDS file
snpgdsClose(genofile)
```

snpgdsGEN2GDS

*Conversion from Oxford GEN format to GDS***Description**

Convert an Oxford GEN file (text format) to a GDS file.

Usage

```
snpgdsGEN2GDS(gen.fn, sample.fn, out.fn, chr.code=NULL,
  call.threshold=0.9, version=c(">=2.0", "<=1.1.5"),
  snpfirstdim=FALSE, compress.annotation="ZIP_RA.max", compress.geno="",
  verbose=TRUE)
```

Arguments

| | |
|----------------------------------|--|
| <code>gen.fn</code> | the file name of Oxford GEN text file(s), it could be a vector indicate merging all files |
| <code>sample.fn</code> | the file name of sample annotation |
| <code>out.fn</code> | the output GDS file |
| <code>chr.code</code> | a vector of chromosome code according to <code>gen.fn</code> , indicating chromosomes. It could be either numeric or character-type |
| <code>call.threshold</code> | the threshold to determine missing genotypes |
| <code>version</code> | either ">=2.0" or "<=1.1.5", see details |
| <code>snpfirstdim</code> | if TRUE, genotypes are stored in the individual-major mode, (i.e, list all SNPs for the first individual, and then list all SNPs for the second individual, etc) |
| <code>compress.annotation</code> | the compression method for the GDS variables, except "genotype"; optional values are defined in the function <code>add.gdsn</code> |
| <code>compress.geno</code> | the compression method for "genotype"; optional values are defined in the function <code>add.gdsn</code> |
| <code>verbose</code> | if TRUE, show information |

Details

GDS – Genomic Data Structures, the extended file name used for storing genetic data, and the file format is used in the [gdsfmt](#) package.

NOTE : the sample file format (`sample.fn`) has changed with the release of SNPTEST v2. Specifically, the way in which covariates and phenotypes are coded on the second line of the header file has changed. `version` has to be specified, and the function uses "`>=2.0`" by default.

Value

Return the file name of GDS format with an absolute path.

Author(s)

Xiuwen Zheng

References

<https://code.enkre.net/bgen>

See Also

[snpGDSBED2GDS](#), [snpGDSVCF2GDS](#)

Examples

```
cat("running snpGDSGEN2GDS ...\\n")
## Not run:
snpGDSGEN2GDS("test.gen", "test.sample", "output.gds", chr.code=1)

## End(Not run)
```

| | |
|---------------|---------------------------------|
| snpGDSGetGeno | <i>To get a genotype matrix</i> |
|---------------|---------------------------------|

Description

To get a genotype matrix from a specified GDS file

Usage

```
snpGDSGetGeno(gdsobj, sample.id=NULL, snp.id=NULL, snpfirstdim=NA,
              .snpread=NA, with.id=FALSE, verbose=TRUE)
```

Arguments

| | |
|--------------------------|--|
| <code>gdsobj</code> | an object of class <code>SNPGDSFileClass</code> , a SNP GDS file; or characters to specify the file name of SNP GDS |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| <code>snpfirstdim</code> | if TRUE, genotypes are stored in the individual-major mode, (i.e, list all SNPs for the first individual, and then list all SNPs for the second individual, etc); FALSE for snp-major mode; if NA, determine automatically |
| <code>.snpread</code> | internal use |
| <code>with.id</code> | if TRUE, return <code>sample.id</code> and <code>snp.id</code> |
| <code>verbose</code> | if TRUE, show information |

Value

The function returns an integer matrix with values 0, 1, 2 or NA representing the number of reference allele when `with.id=FALSE`; or `list(genotype, sample.id, snp.id)` when `with.id=TRUE`. The orders of sample and SNP IDs in the genotype matrix are actually consistent with `sample.id` and `snp.id` in the GDS file, which may not be as the same as the arguments `sample.id` and `snp.id` specified by users.

Author(s)

Xiuwen Zheng

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

set.seed(1000)
snpset <- sample(read.gdsn(index.gdsn(genofile, "snp.id")), 1000)

mat1 <- snpgdsGetGeno(genofile, snp.id=snpset, snpfirstdim=TRUE)
dim(mat1)
# 1000 279
table(c(mat1), exclude=NULL)

mat2 <- snpgdsGetGeno(genofile, snp.id=snpset, snpfirstdim=FALSE)
dim(mat2)
# 279 1000
table(c(mat2), exclude=NULL)

identical(t(mat1), mat2)
# TRUE

# close the file
snpgdsClose(genofile)
```

snpGDSGRM

*Genetic Relationship Matrix (GRM) for SNP genotype data***Description**

Calculate Genetic Relationship Matrix (GRM) using SNP genotype data.

Usage

```
snpGDSGRM(gdsobj, sample.id=NULL, snp.id=NULL,
  autosome.only=TRUE, remove.monosnp=TRUE, maf=NaN, missing.rate=NaN,
  method=c("GCTA", "Eigenstrat", "EIGMIX", "Weighted", "Corr", "IndivBeta"),
  num.thread=1L, useMatrix=FALSE, out.fn=NULL, out.prec=c("double", "single"),
  out.compress="LZMA_RA", with.id=TRUE, verbose=TRUE)
```

Arguments

| | |
|-----------------------------|---|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| <code>autosome.only</code> | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| <code>remove.monosnp</code> | if TRUE, remove monomorphic SNPs |
| <code>maf</code> | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| <code>missing.rate</code> | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| <code>method</code> | "GCTA" – genetic relationship matrix defined in CGTA; "Eigenstrat" – genetic covariance matrix in EIGENSTRAT; "EIGMIX" – two times coancestry matrix defined in Zheng&Weir (2016), "Weighted" – weighted GCTA, as the same as "EIGMIX", "Corr" – Scaled GCTA GRM (dividing each i,j element by the product of the square root of the i,i and j,j elements), "IndivBeta" – two times individual beta estimate relative to the minimum of beta; see details |
| <code>num.thread</code> | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| <code>useMatrix</code> | if TRUE, use <code>Matrix::dspMatrix</code> to store the output square matrix to save memory |
| <code>out.fn</code> | NULL for no GDS output, or a file name |
| <code>out.prec</code> | double or single precision for storage |
| <code>out.compress</code> | the compression method for storing the GRM matrix in the GDS file |
| <code>with.id</code> | if TRUE, the returned value with <code>sample.id</code> and <code>sample.id</code> |
| <code>verbose</code> | if TRUE, show information |

Details

"GCTA": the genetic relationship matrix in GCTA is defined as $G_{ij} = \text{avg}_l [(g_{il} - 2*p_l)*(g_{jl} - 2*p_l) / 2*p_l*(1 - p_l)]$ for individuals i,j and locus l ;

"Eigenstrat": the genetic covariance matrix in EIGENSTRAT $G_{ij} = \text{avg}_l [(g_{il} - 2*p_l)*(g_{jl} - 2*p_l) / 2*p_l*(1 - p_l)]$ for individuals i,j and locus l ; the missing genotype is imputed by the dosage mean of that locus.

"EIGMIX" / "Weighted": it is the same as '2 * snpGDS EIGMIX(, ibdmat=TRUE, diagadj=FALSE)\$ibd': $G_{ij} = [\sum_l (g_{il} - 2*p_l)*(g_{jl} - 2*p_l)] / [\sum_l 2*p_l*(1 - p_l)]$ for individuals i,j and locus l ;

"IndivBeta": 'beta = snpGDS IndivBeta(, inbreeding=TRUE)' (Weir&Goudet, 2017), and beta-based GRM is $g_{ij} = 2 * (\beta_{ij} - \beta_{\min}) / (1 - \beta_{\min})$ for $i \neq j$, $g_{ij} = 1 + (\beta_i - \beta_{\min}) / (1 - \beta_{\min})$ for $i=j$. It is relative to the minimum value of beta estimates.

Value

Return a list if with.id = TRUE:

| | |
|-----------|---|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| method | characters, the method used |
| grm | the genetic relationship matrix; different methods might have different meanings and interpretation for estimates |

If with.id = FALSE, this function returns the genetic relationship matrix (GRM) without sample and SNP IDs.

Author(s)

Xiuwen Zheng

References

- Patterson, N., Price, A. L. & Reich, D. Population structure and eigenanalysis. *PLoS Genet.* 2, e190 (2006).
- Yang, J., Lee, S. H., Goddard, M. E. & Visscher, P. M. GCTA: a tool for genome-wide complex trait analysis. *American journal of human genetics* 88, 76-82 (2011).
- Zheng X, Weir BS. Eigenanalysis on SNP Data with an Interpretation of Identity by Descent. *Theoretical Population Biology.* 2016 Feb;107:65-76. doi: 10.1016/j.tpb.2015.09.004
- Weir BS, Zheng X. SNPs and SNVs in Forensic Science. *Forensic Science International: Genetics Supplement Series.* 2015. doi:10.1016/j.fsigss.2015.09.106
- Weir BS, Goudet J. A Unified Characterization of Population Structure and Relatedness. *Genetics.* 2017 Aug;206(4):2085-2103. doi: 10.1534/genetics.116.198424.

See Also

[snpGDS PCA](#), [snpGDS EIGMIX](#), [snpGDS IndivBeta](#), [snpGDS IndInb](#), [snpGDS Fst](#), [snpGDS MergeGRM](#)

Examples

```

# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpGdsExampleFileName())

rv <- snpgdsGRM(genofile, method="GCTA")
eig <- eigen(rv$grm) # Eigen-decomposition

# output to a GDS file
snpGdsGRM(genofile, method="GCTA", out.fn="test.gds")

pop <- factor(read.gdsn(index.gdsn(genofile, "sample.annot/pop.group")))
plot(eig$vector[,1], eig$vector[,2], col=pop)
legend("topleft", legend=levels(pop), pch=19, col=1:4)

# close the file
snpGdsClose(genofile)

# delete the temporary file
unlink("test.gds", force=TRUE)

```

snpGdsHCluster

Hierarchical cluster analysis

Description

Perform hierarchical cluster analysis on the dissimilarity matrix.

Usage

```
snpGdsHCluster(dist, sample.id=NULL, need.mat=TRUE, hang=0.25)
```

Arguments

| | |
|------------------------|--|
| <code>dist</code> | an object of "snpGdsDissClass" from snpGdsDiss , an object of "snpGdsIBSClass" from snpGdsIBS , or a square matrix for dissimilarity |
| <code>sample.id</code> | to specify sample id, only work if <code>dist</code> is a matrix |
| <code>need.mat</code> | if TRUE, store the dissimilarity matrix in the result |
| <code>hang</code> | The fraction of the plot height by which labels should hang below the rest of the plot. A negative value will cause the labels to hang down from 0. |

Details

Call the function [hclust](#) to perform hierarchical cluster analysis, using `method="average"`.

Value

Return a list (class "snpGdsHCClass"):

| | |
|------------|--|
| sample.id | the sample ids used in the analysis |
| hclust | an object returned from hclust |
| dendrogram | |
| dist | the dissimilarity matrix, if need.mat = TRUE |

Author(s)

Xiuwen Zheng

See Also

[snpGdsIBS](#), [snpGdsDiss](#), [snpGdsCutTree](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

pop.group <- read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))
pop.group <- as.factor(pop.group)
pop.level <- levels(pop.group)

diss <- snpGdsDiss(genofile)
hc <- snpGdsHCluster(diss)
rv <- snpGdsCutTree(hc)
rv

# call 'plot' to draw a dendrogram
plot(rv$dendrogram, leaflab="none", main="HapMap Phase II")

# the distribution of Z scores
snpGdsDrawTree(rv, type="z-score", main="HapMap Phase II")

# draw dendrogram
snpGdsDrawTree(rv, main="HapMap Phase II",
  edgePar=list(col=rgb(0.5,0.5,0.5, 0.75), t.col="black"))

# close the file
snpGdsClose(genofile)
```

`snpgdsHWE`*Statistical test of Hardy-Weinberg Equilibrium*

Description

Calculate the p-values for the exact SNP test of Hardy-Weinberg Equilibrium.

Usage

```
snpgdsHWE(gdsobj, sample.id=NULL, snp.id=NULL, with.id=FALSE)
```

Arguments

| | |
|------------------------|--|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples will be used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs will be used |
| <code>with.id</code> | if TRUE, the returned value with sample and SNP IDs |

Value

If `with.id=FALSE`, return a vector of numeric values (p-value); otherwise, return a list with three components "pvalue", "sample.id" and "snp.id".

Author(s)

Xiuwen Zheng, Janis E. Wigginton

References

Wigginton, J. E., Cutler, D. J. & Abecasis, G. R. A note on exact tests of Hardy-Weinberg equilibrium. *Am. J. Hum. Genet.* 76, 887-93 (2005).

See Also

[snpgdsSNPRateFreq](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

# Japanese samples
sample.id <- read.gdsn(index.gdsn(genofile, "sample.id"))
pop <- read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))
(samp.sel <- sample.id[pop=="JPT"])
samp.sel <- samp.sel[nchar(samp.sel) == 7]
```

```

# chromosome 1
snp.id <- snpGDSselectSNP(genofile, sample.id=samp.sel, autosome.only=1L)

# HWE test
p <- snpGDSHWE(genofile, sample.id=samp.sel, snp.id=snp.id)
summary(p)

# QQ plot
plot(-log10((1:length(p))/length(p)), -log10(p[order(p)]),
     xlab="-log10(expected P)", ylab="-log10(observed P)", main="QQ plot")
abline(a=0, b=1, col="blue")

# close the genotype file
snpGDSclose(genofile)

```

snpGDSIBDKING

KING method of moment for the identity-by-descent (IBD) analysis

Description

Calculate IBD coefficients by KING method of moment.

Usage

```

snpGDSIBDKING(gdsobj, sample.id=NULL, snp.id=NULL, autosome.only=TRUE,
              remove.monosnp=TRUE, maf=NaN, missing.rate=NaN,
              type=c("KING-robust", "KING-homo"), family.id=NULL, num.thread=1L,
              useMatrix=FALSE, verbose=TRUE)

```

Arguments

| | |
|-----------------------------|---|
| <code>gdsobj</code> | an object of class SNPGDSfileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| <code>autosome.only</code> | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| <code>remove.monosnp</code> | if TRUE, remove monomorphic SNPs |
| <code>maf</code> | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| <code>missing.rate</code> | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| <code>type</code> | "KING-robust" – relationship inference in the presence of population stratification (by default); "KING-homo" – relationship inference in a homogeneous population |
| <code>family.id</code> | if NULL, all individuals are treated as singletons; if family id is given, within- and between-family relationship are estimated differently. If <code>sample.id=NULL</code> , <code>family.id</code> should have the same length as "sample.id" in the GDS file, otherwise <code>family.id</code> should have the same length and order as the argument <code>sample.id</code> |

| | |
|------------|--|
| num.thread | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| useMatrix | if TRUE, use <code>Matrix::dspMatrix</code> to store the output square matrix to save memory |
| verbose | if TRUE, show information |

Details

KING IBD estimator is a moment estimator, and it is computationally efficient relative to MLE method. The approaches include "KING-robust" – robust relationship inference within or across families in the presence of population substructure, and "KING-homo" – relationship inference in a homogeneous population.

With "KING-robust", the function would return the proportion of SNPs with zero IBS (IBS0) and kinship coefficient (kinship). With "KING-homo" it would return the probability of sharing one IBD (k1) and the probability of sharing zero IBD (k0).

The minor allele frequency and missing rate for each SNP passed in `snp.id` are calculated over all the samples in `sample.id`.

Value

Return a list:

| | |
|-----------|---|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| k0 | a matrix for IBD coefficients, the probability of sharing zero IBD, if type="KING-homo" |
| k1 | a matrix for IBD coefficients, the probability of sharing one IBD, if type="KING-homo" |
| IBS0 | a matrix for the proportions of SNPs with zero IBS, if type="KING-robust" |
| kinship | a matrix for the estimated kinship coefficients, if type="KING-robust" |

Author(s)

Xiuwen Zheng

References

Manichaikul A, Mychaleckyj JC, Rich SS, Daly K, Sale M, Chen WM. Robust relationship inference in genome-wide association studies. *Bioinformatics*. 2010 Nov 15;26(22):2867-73.

See Also

[snpGdsIBDMLE](#), [snpGdsIBDMoM](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

# CEU population
samp.id <- read.gdsn(index.gdsn(genofile, "sample.id"))
```

```

CEU.id <- samp.id[
  read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))=="CEU"]

#### KING-robust:
#### relationship inference in the presence of population stratification
#### robust relationship inference across family

ibd.robust <- snpGdsIBDKING(genofile, sample.id=CEU.id)
names(ibd.robust)
# [1] "sample.id" "snp.id" "afreq" "IBS0" "kinship"

# select a set of pairs of individuals
dat <- snpGdsIBDSelection(ibd.robust, 1/32)
head(dat)

plot(dat$IBS0, dat$kinship, xlab="Proportion of Zero IBS",
      ylab="Estimated Kinship Coefficient (KING-robust)")

# using Matrix
ibd.robust <- snpGdsIBDKING(genofile, sample.id=CEU.id, useMatrix=TRUE)
is(ibd.robust$IBS0) # dspMatrix
is(ibd.robust$kinship) # dspMatrix

#### KING-robust:
#### relationship inference in the presence of population stratification
#### within- and between-family relationship inference

# incorporate with pedigree information
family.id <- read.gdsn(index.gdsn(genofile, "sample.annot/family.id"))
family.id <- family.id[match(CEU.id, samp.id)]

ibd.robust2 <- snpGdsIBDKING(genofile, sample.id=CEU.id, family.id=family.id)
names(ibd.robust2)

# select a set of pairs of individuals
dat <- snpGdsIBDSelection(ibd.robust2, 1/32)
head(dat)

plot(dat$IBS0, dat$kinship, xlab="Proportion of Zero IBS",
      ylab="Estimated Kinship Coefficient (KING-robust)")

#### KING-homo: relationship inference in a homogeneous population

ibd.homo <- snpGdsIBDKING(genofile, sample.id=CEU.id, type="KING-homo")
names(ibd.homo)
# "sample.id" "snp.id" "afreq" "k0" "k1"

```

```

# select a subset of pairs of individuals
dat <- snpGDSIBDSelection(ibd.homo, 1/32)
head(dat)

plot(dat$k0, dat$kinship, xlab="Pr(IBD=0)",
      ylab="Estimated Kinship Coefficient (KING-homo)")

# using Matrix
ibd.homo <- snpGDSIBDKING(genofile, sample.id=CEU.id, type="KING-homo",
                          useMatrix=TRUE)
is(ibd.homo$k0) # dspMatrix
is(ibd.homo$k1) # dspMatrix

# close the genotype file
snpGDSClose(genofile)

```

| | |
|--------------|---|
| snpGDSIBDMLE | <i>Maximum likelihood estimation (MLE) for the Identity-By-Descent (IBD) Analysis</i> |
|--------------|---|

Description

Calculate the three IBD coefficients (k0, k1, k2) for non-inbred individual pairs by Maximum Likelihood Estimation.

Usage

```

snpGDSIBDMLE(gdsobj, sample.id=NULL, snp.id=NULL, autosome.only=TRUE,
             remove.monosnp=TRUE, maf=NaN, missing.rate=NaN, kinship=FALSE,
             kinship.constraint=FALSE, allele.freq=NULL,
             method=c("EM", "downhill.simplex", "Jacquard"), max.niter=1000L,
             reltol=sqrt(.Machine$double.eps), coeff.correct=TRUE,
             out.num.iter=TRUE, num.thread=1, verbose=TRUE)

```

Arguments

| | |
|-----------------------------|--|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| <code>autosome.only</code> | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| <code>remove.monosnp</code> | if TRUE, remove monomorphic SNPs |
| <code>maf</code> | to use the SNPs with " \geq maf" only; if NaN, no any MAF threshold |
| <code>missing.rate</code> | to use the SNPs with " \leq missing.rate" only; if NaN, no any missing threshold |

| | |
|--------------------|--|
| kinship | if TRUE, output the estimated kinship coefficients |
| kinship.constraint | if TRUE, constrict IBD coefficients (k_0, k_1, k_2) in the genealogical region ($k_0 k_1 \geq k_2^2$) |
| allele.freq | to specify the allele frequencies; if NULL, determine the allele frequencies from gdsobj using the specified samples; if snp.id is specified, allele.freq should have the same order as snp.id |
| method | "EM", "downhill.simplex", "Jacquard", see details |
| max.niter | the maximum number of iterations |
| reltol | relative convergence tolerance; the algorithm stops if it is unable to reduce the value of log likelihood by a factor of $\text{reltol} * (\text{abs}(\log \text{likelihood with the initial parameters}) + \text{reltol})$ at a step. |
| coeff.correct | TRUE by default, see details |
| out.num.iter | if TRUE, output the numbers of iterations |
| num.thread | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| verbose | if TRUE, show information |

Details

The minor allele frequency and missing rate for each SNP passed in `snp.id` are calculated over all the samples in `sample.id`.

The PLINK moment estimates are used as the initial values in the algorithm of searching maximum value of log likelihood function. Two numeric approaches can be used: one is Expectation-Maximization (EM) algorithm, and the other is Nelder-Mead method or downhill simplex method. Generally, EM algorithm is more robust than downhill simplex method. "Jacquard" refers to the estimation of nine Jacquard's coefficients.

If `coeff.correct` is TRUE, the final point that is found by searching algorithm (EM or downhill simplex) is used to compare the six points (fullsib, offspring, halfsib, cousin, unrelated), since any numeric approach might not reach the maximum position after a finite number of steps. If any of these six points has a higher value of log likelihood, the final point will be replaced by the best one.

Although MLE estimates are more reliable than MoM, MLE is much more computationally intensive than MoM, and might not be feasible to estimate pairwise relatedness for a large dataset.

Value

Return a `snpGdsIBDClass` object, which is a list:

| | |
|--------------------------|--|
| <code>sample.id</code> | the sample ids used in the analysis |
| <code>snp.id</code> | the SNP ids used in the analysis |
| <code>afreq</code> | the allele frequencies used in the analysis |
| <code>k0</code> | IBD coefficient, the probability of sharing ZERO IBD, if <code>method="EM"</code> or <code>"downhill.simplex"</code> |
| <code>k1</code> | IBD coefficient, the probability of sharing ONE IBD, if <code>method="EM"</code> or <code>"downhill.simplex"</code> |
| <code>D1, ..., D8</code> | Jacquard's coefficients, if <code>method="Jacquard"</code> , $D9 = 1 - D1 - \dots - D8$ |
| <code>kinship</code> | the estimated kinship coefficients, if the parameter <code>kinship=TRUE</code> |

Author(s)

Xiuwen Zheng

References

Milligan BG. 2003. Maximum-likelihood estimation of relatedness. *Genetics* 163:1153-1167.

Weir BS, Anderson AD, Hepler AB. 2006. Genetic relatedness analysis: modern data and new challenges. *Nat Rev Genet.* 7(10):771-80.

Choi Y, Wijsman EM, Weir BS. 2009. Case-control association testing in the presence of unknown relationships. *Genet Epidemiol* 33(8):668-78.

Jacquard, A. *Structures Genetiques des Populations* (Masson & Cie, Paris, 1970); English translation available in Charlesworth, D. & Charlesworth, B. *Genetics of Human Populations* (Springer, New York, 1974).

See Also

[snpGdsIBDMLELogLik](#), [snpGdsIBDMoM](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

YRI.id <- read.gdsn(index.gdsn(genofile, "sample.id"))[
  read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))=="YRI"]
YRI.id <- YRI.id[1:30]

# SNP pruning
set.seed(10)
snpset <- snpGdsLDPruning(genofile, sample.id=YRI.id, maf=0.05,
  missing.rate=0.05)
snpset <- sample(unlist(snpset), 250)
mibd <- snpGdsIBDMLE(genofile, sample.id=YRI.id, snp.id=snpset)
mibd

# select a set of pairs of individuals
d <- snpGdsIBDSelection(mibd, kinship.cutoff=1/8)
head(d)

# log likelihood

loglik <- snpGdsIBDMLELogLik(genofile, mibd)
loglik0 <- snpGdsIBDMLELogLik(genofile, mibd, relatedness="unrelated")

# likelihood ratio test
p.value <- pchisq(loglik - loglik0, 1, lower.tail=FALSE)

flag <- lower.tri(mibd$k0)
```

```

plot(NaN, xlim=c(0,1), ylim=c(0,1), xlab="k0", ylab="k1")
lines(c(0,1), c(1,0), col="red", lty=3)
points(mibd$k0[f1ag], mibd$k1[f1ag])

# specify the allele frequencies
afreq <- snpgdsSNPRateFreq(genofile, sample.id=YRI.id,
  snp.id=snpset)$AlleleFreq
subibd <- snpgdsIBDMLE(genofile, sample.id=YRI.id[1:25], snp.id=snpset,
  allele.freq=afreq)
summary(c(subibd$k0 - mibd$k0[1:25, 1:25]))
# ZERO
summary(c(subibd$k1 - mibd$k1[1:25, 1:25]))
# ZERO

# close the genotype file
snpgdsClose(genofile)

```

| | |
|--------------------|--|
| snpgdsIBDMLELogLik | <i>Log likelihood for MLE method in the Identity-By-Descent (IBD) Analysis</i> |
|--------------------|--|

Description

Calculate the log likelihood values from maximum likelihood estimation.

Usage

```

snpgdsIBDMLELogLik(gdsobj, ibdobj, k0 = NaN, k1 = NaN,
  relatedness=c("", "self", "fullsib", "offspring",
    "halfsib", "cousin", "unrelated"))

```

Arguments

| | |
|--------------------------|--|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>ibdobj</code> | the <code>snpgdsIBDClass</code> object returned from snpgdsIBDMLE |
| <code>k0</code> | specified IBD coefficient |
| <code>k1</code> | specified IBD coefficient |
| <code>relatedness</code> | specify a relatedness, otherwise use the values of <code>k0</code> and <code>k1</code> |

Details

If (`relatedness == ""`) and (`k0 == NaN` or `k1 == NaN`), then return the log likelihood values for each (`k0`, `k1`) stored in `ibdobj`. \ If (`relatedness == ""`) and (`k0 != NaN`) and (`k1 != NaN`), then return the log likelihood values for a specific IBD coefficient (`k0`, `k1`). \ If `relatedness` is: "self", then `k0 = 0`, `k1 = 0`; "fullsib", then `k0 = 0.25`, `k1 = 0.5`; "offspring", then `k0 = 0`, `k1 = 1`; "halfsib", then `k0 = 0.5`, `k1 = 0.5`; "cousin", then `k0 = 0.75`, `k1 = 0.25`; "unrelated", then `k0 = 1`, `k1 = 0`.

Value

Return a n-by-n matrix of log likelihood values, where n is the number of samples.

Author(s)

Xiuwen Zheng

References

Milligan BG. 2003. Maximum-likelihood estimation of relatedness. *Genetics* 163:1153-1167.

Weir BS, Anderson AD, Hepler AB. 2006. Genetic relatedness analysis: modern data and new challenges. *Nat Rev Genet.* 7(10):771-80.

Choi Y, Wijsman EM, Weir BS. 2009. Case-control association testing in the presence of unknown relationships. *Genet Epidemiol* 33(8):668-78.

See Also

[snpgdsIBDMLE](#), [snpgdsIBDMoM](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

YRI.id <- read.gdsn(index.gdsn(genofile, "sample.id"))[
  read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))=="YRI"]
YRI.id <- YRI.id[1:30]

# SNP pruning
set.seed(10)
snpset <- snpgdsLDpruning(genofile, sample.id=YRI.id, maf=0.05,
  missing.rate=0.05)
snpset <- sample(unlist(snpset), 250)
mibd <- snpgdsIBDMLE(genofile, sample.id=YRI.id, snp.id=snpset)
names(mibd)

# select a set of pairs of individuals
d <- snpgdsIBDSelection(mibd, kinship.cutoff=1/8)
head(d)

# log likelihood

loglik <- snpgdsIBDMLELogLik(genofile, mibd)
loglik0 <- snpgdsIBDMLELogLik(genofile, mibd, relatedness="unrelated")

# likelihood ratio test
p.value <- pchisq(loglik - loglik0, 1, lower.tail=FALSE)

flag <- lower.tri(mibd$k0)
```

```

plot(NaN, xlim=c(0,1), ylim=c(0,1), xlab="k0", ylab="k1")
lines(c(0,1), c(1,0), col="red", lty=3)
points(mibd$k0[f1ag], mibd$k1[f1ag])

# specify the allele frequencies
afreq <- snpGDSRateFreq(genofile, sample.id=YRI.id,
  snp.id=snpset)$AlleleFreq
subibd <- snpGDSIBDMLE(genofile, sample.id=YRI.id[1:25], snp.id=snpset,
  allele.freq=afreq)
summary(c(subibd$k0 - mibd$k0[1:25, 1:25]))
# ZERO
summary(c(subibd$k1 - mibd$k1[1:25, 1:25]))
# ZERO

# close the genotype file
snpGDSClose(genofile)

```

| | |
|--------------|--|
| snpGDSIBDMoM | <i>PLINK method of moment (MoM) for the Identity-By-Descent (IBD) Analysis</i> |
|--------------|--|

Description

Calculate three IBD coefficients for non-inbred individual pairs by PLINK method of moment (MoM).

Usage

```

snpGDSIBDMoM(gdsobj, sample.id=NULL, snp.id=NULL, autosome.only=TRUE,
  remove.monosnp=TRUE, maf=NaN, missing.rate=NaN, allele.freq=NULL,
  kinship=FALSE, kinship.constraint=FALSE, num.thread=1L, useMatrix=FALSE,
  verbose=TRUE)

```

Arguments

| | |
|-----------------------------|--|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| <code>autosome.only</code> | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| <code>remove.monosnp</code> | if TRUE, remove monomorphic SNPs |
| <code>maf</code> | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| <code>missing.rate</code> | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| <code>allele.freq</code> | to specify the allele frequencies; if NULL, determine the allele frequencies from <code>gdsobj</code> using the specified samples; if <code>snp.id</code> is specified, <code>allele.freq</code> should have the same order as <code>snp.id</code> |

| | |
|--------------------|---|
| kinship | if TRUE, output the estimated kinship coefficients |
| kinship.constraint | if TRUE, constrict IBD coefficients (k_0, k_1, k_2) in the genealogical region ($2k_0k_1 \geq k_2^2$) |
| num.thread | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| useMatrix | if TRUE, use <code>Matrix::dspMatrix</code> to store the output square matrix to save memory |
| verbose | if TRUE, show information |

Details

PLINK IBD estimator is a moment estimator, and it is computationally efficient relative to MLE method. In the PLINK method of moment, a correction factor based on allele counts is used to adjust for sampling. However, if allele frequencies are specified, no correction factor is conducted since the specified allele frequencies are assumed to be known without sampling.

The minor allele frequency and missing rate for each SNP passed in `snp.id` are calculated over all the samples in `sample.id`.

Value

Return a list:

| | |
|------------------------|--|
| <code>sample.id</code> | the sample ids used in the analysis |
| <code>snp.id</code> | the SNP ids used in the analysis |
| <code>k0</code> | IBD coefficient, the probability of sharing ZERO IBD |
| <code>k1</code> | IBD coefficient, the probability of sharing ONE IBD |
| <code>kinship</code> | the estimated kinship coefficients, if the parameter <code>kinship=TRUE</code> |

Author(s)

Xiuwen Zheng

References

Purcell S, Neale B, Todd-Brown K, Thomas L, Ferreira MAR, Bender D, Maller J, Sklar P, de Bakker PIW, Daly MJ & Sham PC. 2007. PLINK: a toolset for whole-genome association and population-based linkage analysis. *American Journal of Human Genetics*, 81.

See Also

[snpGdsIBDMLE](#), [snpGdsIBDMLELogLik](#)

Examples

```

# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpGDSExampleFileName())

#####
# CEU population

CEU.id <- read.gdsn(index.gdsn(genofile, "sample.id"))[
  read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))=="CEU"]
pibd <- snpgdsIBDMoM(genofile, sample.id=CEU.id)
names(pibd)

flag <- lower.tri(pibd$k0)
plot(NaN, xlim=c(0,1), ylim=c(0,1), xlab="k0", ylab="k1")
lines(c(0,1), c(1,0), col="red", lty=3)
points(pibd$k0[flag], pibd$k1[flag])

# select a set of pairs of individuals
d <- snpgdsIBDSelection(pibd, kinship.cutoff=1/8)
head(d)

#####
# YRI population

YRI.id <- read.gdsn(index.gdsn(genofile, "sample.id"))[
  read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))=="YRI"]
pibd <- snpgdsIBDMoM(genofile, sample.id=YRI.id)
flag <- lower.tri(pibd$k0)
plot(NaN, xlim=c(0,1), ylim=c(0,1), xlab="k0", ylab="k1")
lines(c(0,1), c(1,0), col="red", lty=3)
points(pibd$k0[flag], pibd$k1[flag])

# specify the allele frequencies
afreq <- snpgdsSNPRateFreq(genofile, sample.id=YRI.id)$AlleleFreq
aibd <- snpgdsIBDMoM(genofile, sample.id=YRI.id, allele.freq=afreq)
flag <- lower.tri(aibd$k0)
plot(NaN, xlim=c(0,1), ylim=c(0,1), xlab="k0", ylab="k1")
lines(c(0,1), c(1,0), col="red", lty=3)
points(aibd$k0[flag], aibd$k1[flag])

# analysis on a subset
subibd <- snpgdsIBDMoM(genofile, sample.id=YRI.id[1:25], allele.freq=afreq)
summary(c(subibd$k0 - aibd$k0[1:25, 1:25]))
# ZERO
summary(c(subibd$k1 - aibd$k1[1:25, 1:25]))
# ZERO

# close the genotype file
snpGDSClose(genofile)

```

snpGDSIBDSelection *Get a table of IBD coefficients*

Description

Return a data frame with IBD coefficients.

Usage

```
snpGDSIBDSelection(ibdobj, kinship.cutoff=NaN, samp.sel=NULL)
```

Arguments

`ibdobj` an object of `snpGDSIBDClass` returned by `snpGDSIBDMLE` or `snpGDSIBDMoM`
`kinship.cutoff` select the individual pairs with kinship coefficients \geq `kinship.cutoff`; no filter if `kinship.cutoff = NaN`
`samp.sel` a logical vector or integer vector to specify selection of samples

Value

Return a data.frame:

| | |
|----------------------|---|
| <code>ID1</code> | the id of the first individual |
| <code>ID2</code> | the id of the second individual |
| <code>k0</code> | the probability of sharing ZERO alleles |
| <code>k1</code> | the probability of sharing ONE alleles |
| <code>kinship</code> | kinship coefficient |

Author(s)

Xiuwen Zheng

See Also

[snpGDSIBDMLE](#), [snpGDSIBDMoM](#), [snpGDSIBDKING](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())

# YRI population
YRI.id <- read.gdsn(index.gdsn(genofile, "sample.id"))[
  read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))=="YRI"]
pibd <- snpGDSIBDMoM(genofile, sample.id=YRI.id)
flag <- lower.tri(pibd$k0)
plot(NaN, xlim=c(0,1), ylim=c(0,1), xlab="k0", ylab="k1")
```

```

lines(c(0,1), c(1,0), col="red", lty=3)
points(pibd$k0[flag], pibd$k1[flag])

# close the genotype file
snpGDSClose(genofile)

# IBD coefficients
dat <- snpGDSIBSSelection(pibd, 1/32)
head(dat)
#      ID1      ID2      k0      k1      kinship
# 1 NA19152 NA19154 0.010749154 0.9892508 0.24731271
# 2 NA19152 NA19093 0.848207777 0.1517922 0.03794806
# 3 NA19139 NA19138 0.010788047 0.9770181 0.25035144
# 4 NA19139 NA19137 0.012900661 0.9870993 0.24677483
# 5 NA18912 NA18914 0.008633077 0.9913669 0.24784173
# 6 NA19160 NA19161 0.008635754 0.9847777 0.24948770

```

snpGDSIBS

Identity-By-State (IBS) proportion

Description

Calculate the fraction of identity by state for each pair of samples

Usage

```

snpGDSIBS(gdsobj, sample.id=NULL, snp.id=NULL, autosome.only=TRUE,
  remove.monosnp=TRUE, maf=NaN, missing.rate=NaN, num.thread=1L,
  useMatrix=FALSE, verbose=TRUE)

```

Arguments

| | |
|-----------------------------|--|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| <code>autosome.only</code> | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| <code>remove.monosnp</code> | if TRUE, remove monomorphic SNPs |
| <code>maf</code> | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| <code>missing.rate</code> | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| <code>num.thread</code> | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| <code>useMatrix</code> | if TRUE, use <code>Matrix::dspMatrix</code> to store the output square matrix to save memory |
| <code>verbose</code> | if TRUE, show information |

Details

The minor allele frequency and missing rate for each SNP passed in `snp.id` are calculated over all the samples in `sample.id`.

The values of the IBS matrix range from ZERO to ONE, and it is defined as the average of $1 - |g_{\{1, i\}} - g_{\{2, i\}}| / 2$ across the genome for the first and second individuals and SNP `i`.

Value

Return a list (class "snpGdsIBSClass"):

| | |
|------------------------|---|
| <code>sample.id</code> | the sample ids used in the analysis |
| <code>snp.id</code> | the SNP ids used in the analysis |
| <code>ibs</code> | a matrix of IBS proportion, "# of samples" x "# of samples" |

Author(s)

Xiuwen Zheng

See Also

[snpGdsIBSNum](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

# perform identity-by-state calculations
ibs <- snpGdsIBS(genofile)

# perform multidimensional scaling analysis on
# the genome-wide IBS pairwise distances:
loc <- cmdscale(1 - ibs$ibs, k = 2)
x <- loc[, 1]; y <- loc[, 2]
race <- as.factor(read.gdsn(index.gdsn(genofile, "sample.annot/pop.group")))
plot(x, y, col=race, xlab = "", ylab = "", main = "cmdscale(IBS Distance)")
legend("topleft", legend=levels(race), text.col=1:nlevels(race))

# close the file
snpGdsClose(genofile)
```

snpgdsIBSNum *Identity-By-State (IBS)*

Description

Calculate the number of SNPs for identity by state for each pair of samples.

Usage

```
snpgdsIBSNum(gdsobj, sample.id = NULL, snp.id = NULL, autosome.only = TRUE,
  remove.monosnp = TRUE, maf = NaN, missing.rate = NaN, num.thread = 1L,
  verbose = TRUE)
```

Arguments

| | |
|----------------|--|
| gdsobj | an object of class SNPGDSFileClass , a SNP GDS file |
| sample.id | a vector of sample id specifying selected samples; if NULL, all samples are used |
| snp.id | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| autosome.only | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| remove.monosnp | if TRUE, remove monomorphic SNPs |
| maf | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| missing.rate | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| num.thread | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| verbose | if TRUE, show information |

Details

The minor allele frequency and missing rate for each SNP passed in `snp.id` are calculated over all the samples in `sample.id`.

Value

Return a list (n is the number of samples):

| | |
|-----------|---|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| ibs0 | a n-by-n matrix, the number of SNPs sharing 0 IBS |
| ibs1 | a n-by-n matrix, the number of SNPs sharing 1 IBS |
| ibs2 | a n-by-n matrix, the number of SNPs sharing 2 IBS |

Author(s)

Xiuwen Zheng

See Also[snpgdsIBS](#)**Examples**

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

RV <- snpgdsIBSNum(genofile)
pop <- read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))
L <- order(pop)
image(RV$ibs0[L, L]/length(RV$snp.id))

# close the genotype file
snpgdsClose(genofile)
```

snpgdsIndInb

*Individual Inbreeding Coefficients***Description**

To calculate individual inbreeding coefficients using SNP genotype data

Usage

```
snpgdsIndInb(gdsobj, sample.id=NULL, snp.id=NULL,
  autosome.only=TRUE, remove.monosnp=TRUE, maf=NaN, missing.rate=NaN,
  method=c("mom.weir", "mom.visscher", "mle", "gcta1", "gcta2", "gcta3"),
  allele.freq=NULL, out.num.iter=TRUE, reltol=.Machine$double.eps^0.75,
  verbose=TRUE)
```

Arguments

| | |
|-----------------------------|--|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| <code>autosome.only</code> | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| <code>remove.monosnp</code> | if TRUE, remove monomorphic SNPs |
| <code>maf</code> | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| <code>missing.rate</code> | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| <code>method</code> | see details |
| <code>allele.freq</code> | to specify the allele frequencies; if NULL, the allele frequencies are estimated from the given samples |
| <code>out.num.iter</code> | output the numbers of iterations |

`reltol` relative convergence tolerance used in MLE; the algorithm stops if it is unable to reduce the value of log likelihood by a factor of $\$reltol * (abs(log likelihood with the initial parameters) + reltol)$ at a step.

`verbose` if TRUE, show information

Details

The method can be: "mom.weir": a modified Visscher's estimator, proposed by Bruce Weir; "mom.visscher": Visscher's estimator described in Yang et al. (2010); "mle": the maximum likelihood estimation; "gcta1": F^I in GCTA, $avg [(g_i - 2p_i)^2 / (2 * p_i * (1 - p_i)) - 1]$; "gcta2": F^{II} in GCTA, $avg [1 - g_i * (2 - g_i) / (2 * p_i * (1 - p_i))]$; "gcta3": F^{III} in GCTA, the same as "mom.visscher", $avg [g_i^2 - (1 + 2p_i) * g_i + 2 * p_i^2] / (2 * p_i * (1 - p_i))$.

Value

Return estimated inbreeding coefficient.

Author(s)

Xiuwen Zheng

References

Yang J, Benyamin B, McEvoy BP, Gordon S, Henders AK, Nyholt DR, Madden PA, Heath AC, Martin NG, Montgomery GW, Goddard ME, Visscher PM. 2010. Common SNPs explain a large proportion of the heritability for human height. *Nat Genet.* 42(7):565-9. Epub 2010 Jun 20.

Yang, J., Lee, S. H., Goddard, M. E. & Visscher, P. M. GCTA: a tool for genome-wide complex trait analysis. *American journal of human genetics* 88, 76-82 (2011).

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

rv <- snpGdsIndInb(genofile, method="mom.visscher")
head(rv$inbreeding)
summary(rv$inbreeding)

# close the genotype file
snpGdsClose(genofile)
```

snpGdsIndInbCoef

Individual Inbreeding Coefficient

Description

To calculate an individual inbreeding coefficient using SNP genotype data

Usage

```
snpGdsIndInbCoef(x, p, method = c("mom.weir", "mom.visscher", "mle"),
  reltol=.Machine$double.eps^0.75)
```

Arguments

| | |
|--------|---|
| x | SNP genotypes |
| p | allele frequencies |
| method | see details |
| reltol | relative convergence tolerance used in MLE; the algorithm stops if it is unable to reduce the value of log likelihood by a factor of $\$reltol * (\text{abs}(\text{log likelihood with the initial parameters}) + reltol)$ at a step. |

Details

The method can be: "mom.weir": a modified Visscher's estimator, proposed by Bruce Weir; "mom.visscher": Visscher's estimator described in Yang et al. (2010); "mle": the maximum likelihood estimation.

Value

Return estimated inbreeding coefficient.

Author(s)

Xiuwen Zheng

References

Yang J, Benyamin B, McEvoy BP, Gordon S, Henders AK, Nyholt DR, Madden PA, Heath AC, Martin NG, Montgomery GW, Goddard ME, Visscher PM. 2010. Common SNPs explain a large proportion of the heritability for human height. *Nat Genet.* 42(7):565-9. Epub 2010 Jun 20.

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

chr1 <- read.gdsn(index.gdsn(genofile, "snp.id"))[
  read.gdsn(index.gdsn(genofile, "snp.chromosome"))==1]
chr1idx <- match(chr1, read.gdsn(index.gdsn(genofile, "snp.id")))

AF <- snpGdsSNPRateFreq(genofile)
g <- read.gdsn(index.gdsn(genofile, "genotype"), start=c(1,1), count=c(-1,1))

snpGdsIndInbCoef(g[chr1idx], AF$AlleleFreq[chr1idx], method="mom.weir")
snpGdsIndInbCoef(g[chr1idx], AF$AlleleFreq[chr1idx], method="mom.visscher")
snpGdsIndInbCoef(g[chr1idx], AF$AlleleFreq[chr1idx], method="mle")

# close the genotype file
```

```
snpgdsClose(genofile)
```

snpgdsIndivBeta *Individual inbreeding and relatedness estimation (beta estimator)*

Description

Calculate individual inbreeding and relatedness estimation (beta estimator) using SNP genotype data.

Usage

```
snpgdsIndivBeta(gdsobj, sample.id=NULL, snp.id=NULL, autosome.only=TRUE,
  remove.monosnp=TRUE, maf=NaN, missing.rate=NaN, method=c("weighted"),
  inbreeding=TRUE, num.thread=1L, with.id=TRUE, useMatrix=FALSE, verbose=TRUE)
snpgdsIndivBetaRel(beta, beta_rel, verbose=TRUE)
```

Arguments

| | |
|-----------------------------|--|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| <code>autosome.only</code> | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| <code>remove.monosnp</code> | if TRUE, remove monomorphic SNPs |
| <code>maf</code> | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| <code>missing.rate</code> | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| <code>method</code> | "weighted" estimator |
| <code>inbreeding</code> | TRUE, the diagonal is a vector of inbreeding coefficients; otherwise, individual variance estimates |
| <code>num.thread</code> | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| <code>with.id</code> | if TRUE, the returned value with <code>sample.id</code> and <code>sample.id</code> |
| <code>useMatrix</code> | if TRUE, use <code>Matrix::dspMatrix</code> to store the output square matrix to save memory |
| <code>beta</code> | the object returned from <code>snpgdsIndivBeta()</code> |
| <code>beta_rel</code> | the beta-based matrix is generated relative to <code>beta_rel</code> |
| <code>verbose</code> | if TRUE, show information |

Value

Return a list if with.id = TRUE:

| | |
|------------|--|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| inbreeding | a logical value; TRUE, the diagonal is a vector of inbreeding coefficients; otherwise, individual variance estimates |
| beta | beta estimates |
| avg_val | the average of M_B among all loci, it could be used to calculate each M_ij |

If with.id = FALSE, this function returns the genetic relationship matrix without sample and SNP IDs.

Author(s)

Xiuwen Zheng

References

Weir BS, Zheng X. SNPs and SNVs in Forensic Science. *Forensic Science International: Genetics Supplement Series*. 2015. doi:10.1016/j.fsigss.2015.09.106

Weir BS, Goudet J. A Unified Characterization of Population Structure and Relatedness. *Genetics*. 2017 Aug;206(4):2085-2103. doi: 10.1534/genetics.116.198424.

See Also

[snpGdsGRM](#), [snpGdsIndInb](#), [snpGdsFst](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

b <- snpGdsIndivBeta(genofile, inbreeding=FALSE)
b$beta[1:10, 1:10]

z <- snpGdsIndivBetaRel(b, min(b$beta))

# close the file
snpGdsClose(genofile)
```

snpGDSLDMat

*Linkage Disequilibrium (LD) analysis***Description**

Return a LD matrix for SNP pairs.

Usage

```
snpGDSLDMat(gdsobj, sample.id=NULL, snp.id=NULL, slide=250L,
            method=c("composite", "r", "dprime", "corr", "cov"), mat.trim=FALSE,
            num.thread=1L, with.id=TRUE, verbose=TRUE)
```

Arguments

| | |
|------------|---|
| gdsobj | an object of class SNPGDSFileClass , a SNP GDS file |
| sample.id | a vector of sample id specifying selected samples; if NULL, all samples are used |
| snp.id | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| slide | # of SNPs, the size of sliding window; if $slide < 0$, return a full LD matrix; see details |
| method | "composite", "r", "dprime", "corr", "cov", see details |
| mat.trim | if TRUE, trim the matrix when $slide > 0$: the function returns a "num_slide x (n_snp - slide)" matrix |
| num.thread | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| with.id | if TRUE, the returned value with sample.id and snp.id |
| verbose | if TRUE, show information |

Details

Four methods can be used to calculate linkage disequilibrium values: "composite" for LD composite measure, "r" for R coefficient (by EM algorithm assuming HWE, it could be negative), "dprime" for D' , and "corr" for correlation coefficient. The method "corr" is equivalent to "composite", when SNP genotypes are coded as: 0 – BB, 1 – AB, 2 – AA.

If $slide \leq 0$, the function returns a n-by-n LD matrix where the value of i row and j column is LD of i and j SNPs. If $slide > 0$, it returns a m-by-n LD matrix where n is the number of SNPs, m is the size of sliding window, and the value of i row and j column is LD of j and j+i SNPs.

Value

Return a list:

| | |
|-----------|-------------------------------------|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| LD | a matrix of LD values |
| slide | the size of sliding window |

Author(s)

Xiuwen Zheng

References

Weir B: Inferences about linkage disequilibrium. *Biometrics* 1979; 35: 235-254.

Weir B: *Genetic Data Analysis II*. Sunderland, MA: Sinauer Associates, 1996.

Weir BS, Cockerham CC: Complete characterization of disequilibrium at two loci; in Feldman MW (ed): *Mathematical Evolutionary Theory*. Princeton, NJ: Princeton University Press, 1989.

See Also

[snpGdsLDpair](#), [snpGdsLDpruning](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

# missing proportion and MAF
ff <- snpGdsSNPRateFreq(genofile)

# chromosome 15
snpset <- read.gdsn(index.gdsn(genofile, "snp.id"))[
  ff$MissingRate==0 & ff$MinorFreq>0 &
  read.gdsn(index.gdsn(genofile, "snp.chromosome"))==15]
length(snpset)

# LD matrix without sliding window
ld.noslide <- snpGdsLDMat(genofile, snp.id=snpset, slide=-1, method="composite")
# plot
image(t(ld.noslide$LD^2), col=terrain.colors(16))

# LD matrix with a sliding window
ld.slide <- snpGdsLDMat(genofile, snp.id=snpset, method="composite")
# plot
image(t(ld.slide$LD^2), col=terrain.colors(16))

# close the genotype file
snpGdsClose(genofile)
```

snpGdsLDpair

Linkage Disequilibrium (LD)

Description

Return a LD value between snp1 and snp2.

Usage

```
snpgdsLDpair(snp1, snp2, method = c("composite", "r", "dprime", "corr"))
```

Arguments

| | |
|--------|--|
| snp1 | a vector of SNP genotypes (0 – BB, 1 – AB, 2 – AA) |
| snp2 | a vector of SNP genotypes (0 – BB, 1 – AB, 2 – AA) |
| method | "composite", "r", "dprime", "corr", see details |

Details

Four methods can be used to calculate linkage disequilibrium values: "composite" for LD composite measure, "r" for R coefficient (by EM algorithm assuming HWE, it could be negative), "dprime" for D' , and "corr" for correlation coefficient. The method "corr" is equivalent to "composite", when SNP genotypes are coded as: 0 – BB, 1 – AB, 2 – AA.

Value

Return a numeric vector:

| | |
|----|-------------------------------------|
| ld | a measure of linkage disequilibrium |
|----|-------------------------------------|

if method = "r" or "dprime",

| | |
|------|---|
| pA_A | haplotype frequency of AA, the first locus is A and the second locus is A |
| pA_B | haplotype frequency of AB, the first locus is A and the second locus is B |
| pB_A | haplotype frequency of BA, the first locus is B and the second locus is A |
| pB_B | haplotype frequency of BB, the first locus is B and the second locus is B |

Author(s)

Xiuwen Zheng

References

Weir B: Inferences about linkage disequilibrium. *Biometrics* 1979; 35: 235-254.

Weir B: *Genetic Data Analysis II*. Sunderland, MA: Sinauer Associates, 1996.

Weir BS, Cockerham CC: Complete characterization of disequilibrium at two loci; in Feldman MW (ed): *Mathematical Evolutionary Theory*. Princeton, NJ: Princeton University Press, 1989.

See Also

[snpgdsLDMat](#), [snpgdsLDpruning](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())

snp1 <- read.gdsn(index.gdsn(genofile, "genotype"), start=c(1,1), count=c(1,-1))
snp2 <- read.gdsn(index.gdsn(genofile, "genotype"), start=c(2,1), count=c(1,-1))

snpGDSLDPair(snp1, snp2, method = "composite")
snpGDSLDPair(snp1, snp2, method = "r")
snpGDSLDPair(snp1, snp2, method = "dprime")
snpGDSLDPair(snp1, snp2, method = "corr")

# close the genotype file
snpGSDSClose(genofile)
```

snpGDSLDP pruning

*Linkage Disequilibrium (LD) based SNP pruning***Description**

Recursively removes SNPs within a sliding window

Usage

```
snpGDSLDP pruning(gdsobj, sample.id=NULL, snp.id=NULL, autosome.only=TRUE,
  remove.monosnp=TRUE, maf=0.005, missing.rate=0.05,
  method=c("composite", "r", "dprime", "corr"), slide.max.bp=500000L,
  slide.max.n=NA, ld.threshold=0.2,
  start.pos=c("random.f500", "random", "first", "last"),
  num.thread=1L, autosave=NULL, verbose=TRUE)
```

Arguments

| | |
|-----------------------------|--|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| <code>autosome.only</code> | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| <code>remove.monosnp</code> | if TRUE, remove monomorphic SNPs |
| <code>maf</code> | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| <code>missing.rate</code> | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| <code>method</code> | "composite", "r", "dprime", "corr", see details |
| <code>slide.max.bp</code> | the maximum basepairs in the sliding window |
| <code>slide.max.n</code> | the maximum number of SNPs in the sliding window |
| <code>ld.threshold</code> | the LD threshold |

| | |
|-------------------------|--|
| <code>start.pos</code> | "random.f500", a starting position randomly selected from the first 500 markers (by default); "random": a random starting position; "first": start from the first position; "last": start from the last position. "random.f500" is applicable for \geq v1.37.2 |
| <code>num.thread</code> | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| <code>autosave</code> | NULL or a file name for autosaving a single R object (saving via saveRDS) |
| <code>verbose</code> | if TRUE, show information |

Details

The minor allele frequency and missing rate for each SNP passed in `snp.id` are calculated over all the samples in `sample.id`.

Four methods can be used to calculate linkage disequilibrium values: "composite" for LD composite measure, "r" for R coefficient (by EM algorithm assuming HWE, it could be negative), "dprime" for D' , and "corr" for correlation coefficient. The method "corr" is equivalent to "composite", when SNP genotypes are coded as: 0 – BB, 1 – AB, 2 – AA. The argument `ld.threshold` is the absolute value of measurement.

It is useful to generate a pruned subset of SNPs that are in approximate linkage equilibrium with each other. The function `snpGdsLDpruning` recursively removes SNPs within a sliding window based on the pairwise genotypic correlation. SNP pruning is conducted chromosome by chromosome, since SNPs in a chromosome can be considered to be independent with the other chromosomes.

The pruning algorithm on a chromosome is described as follows (n is the total number of SNPs on that chromosome):

- 1) Randomly select a starting position i (`start.pos="random"`), $i=1$ if `start.pos="first"`, or $i=\text{last}$ if `start.pos="last"`; and let the current SNP set $S=\{i\}$;
- 2) For each right position j from $i+1$ to n : if any LD between j and k is greater than `ld.threshold`, where k belongs to S , and both of j and k are in the sliding window, then skip j ; otherwise, let S be $S + \{j\}$;
- 3) For each left position j from $i-1$ to 1: if any LD between j and k is greater than `ld.threshold`, where k belongs to S , and both of j and k are in the sliding window, then skip j ; otherwise, let S be $S + \{j\}$;
- 4) Output S , the final selection of SNPs.

Value

Return a list of SNP IDs stratified by chromosomes.

Author(s)

Xiuwen Zheng

References

- Weir B: Inferences about linkage disequilibrium. *Biometrics* 1979; 35: 235-254.
 Weir B: *Genetic Data Analysis II*. Sunderland, MA: Sinauer Associates, 1996.

Weir BS, Cockerham CC: Complete characterization of disequilibrium at two loci; in Feldman MW (ed): Mathematical Evolutionary Theory. Princeton, NJ: Princeton University Press, 1989.

See Also

[snpgdsLDMat](#), [snpgdsLDpair](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

set.seed(1000)
snpset <- snpgdsLDpruning(genofile)
str(snpset)
names(snpset)
# [1] "chr1" "chr2" "chr3" "chr4" "chr5" "chr6" "chr7" "chr8" "chr9"
# [10] "chr10" "chr11" "chr12" "chr13" "chr14" "chr15" "chr16" "chr17" "chr18"
# .....

# get SNP ids
snp.id <- unlist(unname(snpset))

# close the genotype file
snpgdsClose(genofile)
```

snpgdsMergeGRM

Merge Multiple Genetic Relationship Matrices (GRM)

Description

Combine multiple genetic relationship matrices with weighted averaging.

Usage

```
snpgdsMergeGRM(filelist, out.fn=NULL, out.prec=c("double", "single"),
  out.compress="LZMA_RA", weight=NULL, verbose=TRUE)
```

Arguments

| | |
|--------------|--|
| filelist | a character vector, list of GDS file names |
| out.fn | NULL, return a GRM object; or characters, the output GDS file name |
| out.prec | double or single precision for storage |
| out.compress | the compression method for storing the GRM matrix in the GDS file |
| weight | NULL, weights proportional to the numbers of SNPs; a numeric vector, or a logical vector (FALSE for excluding some GRMs with a negative weight, weights proportional to the numbers of SNPs) |
| verbose | if TRUE, show information |

Details

The final GRM is the weighted averaged matrix combining multiple GRMs. The merged GRM may not be identical to the GRM calculated using full SNPs, due to missing genotypes or the internal weighting strategy of the specified GRM calculation.

Value

None or a GRM object if `out.fn=NULL`.

Author(s)

Xiuwen Zheng

See Also

[snpgdsGRM](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

snpid <- read.gdsn(index.gdsn(genofile, "snp.id"))
snpid <- snpid[snpgdsSNPRateFreq(genofile)$MissingRate == 0]

# there is no missing genotype
grm <- snpgdsGRM(genofile, snp.id=snpid, method="GCTA")

# save two GRMs
set1 <- grm$snp.id[1:(length(grm$snp.id)/2)]
set2 <- setdiff(grm$snp.id, set1)
snpgdsGRM(genofile, method="GCTA", snp.id=set1, out.fn="tmp1.gds")
snpgdsGRM(genofile, method="GCTA", snp.id=set2, out.fn="tmp2.gds")

# merge GRMs and export to a new GDS file
snpgdsMergeGRM(c("tmp1.gds", "tmp2.gds"), "tmp.gds")

# return the GRM
grm2 <- snpgdsMergeGRM(c("tmp1.gds", "tmp2.gds"))

# check
f <- openfn.gds("tmp.gds")
m <- read.gdsn(index.gdsn(f, "grm"))
closefn.gds(f)

summary(c(m - grm$grm)) # ~zero
summary(c(m - grm2$grm)) # zero

# close the file
```

```
snpgdsClose(genofile)

# delete the temporary file
unlink(c("tmp1.gds", "tmp2.gds", "tmp.gds"), force=TRUE)
```

snpgdsOpen*Open a SNP GDS File*

Description

Open a SNP GDS file

Usage

```
snpgdsOpen(filename, readonly=TRUE, allow.duplicate=FALSE, allow.fork=TRUE)
```

Arguments

| | |
|-----------------|---|
| filename | the file name |
| readonly | whether read-only or not |
| allow.duplicate | if TRUE, it is allowed to open a GDS file with read-only mode when it has been opened in the same R session, see openfn.gds |
| allow.fork | TRUE for parallel environment using forking, see openfn.gds |

Details

It is strongly suggested to call `snpgdsOpen` instead of [openfn.gds](#), since `snpgdsOpen` will perform internal checking for data integrity.

Value

Return an object of class `SNPGDSFileClass`.

Author(s)

Xiuwen Zheng

See Also

[snpgdsClose](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

genofile

# close the file
snpgdsClose(genofile)
```

snpgdsOption

Option settings: chromosome coding, etc

Description

Return an option list used by the SNPRelate package or a GDS file

Usage

```
snpgdsOption(gdsobj=NULL, autosome.start=1L, autosome.end=22L, ...)
```

Arguments

gdsobj an object of class [SNPGDSFileClass](#), a SNP GDS file
autosome.start the starting index of autosome
autosome.end the ending index of autosome
... optional arguments for new chromosome coding

Value

A list

Author(s)

Xiuwen Zheng

Examples

```
# define the new chromosomes 'Z' and 'W'
snpgdsOption(Z=27L, W=28L)

# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

snpgdsOption(genofile)

# close the genotype file
snpgdsClose(genofile)
```

snpGDSPairIBD *Calculate Identity-By-Descent (IBD) Coefficients*

Description

Calculate the three IBD coefficients (k0, k1, k2) for non-inbred individual pairs by Maximum Likelihood Estimation (MLE) or PLINK Method of Moment (MoM).

Usage

```
snpGDSPairIBD(geno1, geno2, allele.freq,
              method=c("EM", "downhill.simplex", "MoM", "Jacquard"),
              kinship.constraint=FALSE, max.niter=1000L, reltol=sqrt(.Machine$double.eps),
              coeff.correct=TRUE, out.num.iter=TRUE, verbose=TRUE)
```

Arguments

| | |
|--------------------|--|
| geno1 | the SNP genotypes for the first individual, 0 – BB, 1 – AB, 2 – AA, other values – missing |
| geno2 | the SNP genotypes for the second individual, 0 – BB, 1 – AB, 2 – AA, other values – missing |
| allele.freq | the allele frequencies |
| method | "EM", "downhill.simplex", "MoM" or "Jacquard", see details |
| kinship.constraint | if TRUE, constrict IBD coefficients (k_0, k_1, k_2) in the genealogical region ($k_0 k_1 \geq k_2^2$) |
| max.niter | the maximum number of iterations |
| reltol | relative convergence tolerance; the algorithm stops if it is unable to reduce the value of log likelihood by a factor of $\text{reltol} * (\text{abs}(\log \text{likelihood with the initial parameters}) + \text{reltol})$ at a step. |
| coeff.correct | TRUE by default, see details |
| out.num.iter | if TRUE, output the numbers of iterations |
| verbose | if TRUE, show information |

Details

If method = "MoM", then PLINK Method of Moment without a allele-count-based correction factor is conducted. Otherwise, two numeric approaches for maximum likelihood estimation can be used: one is Expectation-Maximization (EM) algorithm, and the other is Nelder-Mead method or downhill simplex method. Generally, EM algorithm is more robust than downhill simplex method. "Jacquard" refers to the estimation of nine Jacquard's coefficients.

If coeff.correct is TRUE, the final point that is found by searching algorithm (EM or downhill simplex) is used to compare the six points (fullsib, offspring, halfsib, cousin, unrelated), since any numeric approach might not reach the maximum position after a finite number of steps. If any of these six points has a higher value of log likelihood, the final point will be replaced by the best one.

Value

Return a data.frame:

| | |
|--------|--|
| k0 | IBD coefficient, the probability of sharing ZERO IBD |
| k1 | IBD coefficient, the probability of sharing ONE IBD |
| loglik | the value of log likelihood |
| niter | the number of iterations |

Author(s)

Xiuwen Zheng

References

Milligan BG. 2003. Maximum-likelihood estimation of relatedness. *Genetics* 163:1153-1167.

Weir BS, Anderson AD, Hepler AB. 2006. Genetic relatedness analysis: modern data and new challenges. *Nat Rev Genet.* 7(10):771-80.

Choi Y, Wijsman EM, Weir BS. 2009. Case-control association testing in the presence of unknown relationships. *Genet Epidemiol* 33(8):668-78.

Purcell S, Neale B, Todd-Brown K, Thomas L, Ferreira MAR, Bender D, Maller J, Sklar P, de Bakker PIW, Daly MJ & Sham PC. 2007. PLINK: a toolset for whole-genome association and population-based linkage analysis. *American Journal of Human Genetics*, 81.

See Also

[snpGdsPairIBDMLELogLik](#), [snpGdsIBDMLE](#), [snpGdsIBDMLELogLik](#), [snpGdsIBDMoM](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

YRI.id <- read.gdsn(index.gdsn(genofile, "sample.id"))[
  read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))=="YRI"]

# SNP pruning
set.seed(10)
snpset <- snpGdsLDPruning(genofile, sample.id=YRI.id, maf=0.05,
  missing.rate=0.05)
snpset <- unname(sample(unlist(snpset), 250))

# the number of samples
n <- 25

# specify allele frequencies
RF <- snpGdsSNPRateFreq(genofile, sample.id=YRI.id, snp.id=snpset,
  with.id=TRUE)
summary(RF$AlleleFreq)
```

```

subMLE <- snpgdsIBDMLE(genofile, sample.id=YRI.id[1:n], snp.id=RF$snp.id,
  allele.freq=RF$AlleleFreq)
subMoM <- snpgdsIBDMoM(genofile, sample.id=YRI.id[1:n], snp.id=RF$snp.id,
  allele.freq=RF$AlleleFreq)
subJac <- snpgdsIBDMLE(genofile, sample.id=YRI.id[1:n], snp.id=RF$snp.id,
  allele.freq=RF$AlleleFreq, method="Jacquard")

#####

# genotype matrix
mat <- snpgdsGetGeno(genofile, sample.id=YRI.id[1:n], snp.id=snpset,
  snpfirstdim=TRUE)

rv <- NULL
for (i in 2:n)
{
  rv <- rbind(rv, snpgdsPairIBD(mat[,1], mat[,i], RF$AlleleFreq, "EM"))
  print(snpGdsPairIBDMLELogLik(mat[,1], mat[,i], RF$AlleleFreq,
    relatedness="unrelated", verbose=TRUE))
}
rv
summary(rv$k0 - subMLE$k0[1, 2:n])
summary(rv$k1 - subMLE$k1[1, 2:n])
# ZERO

rv <- NULL
for (i in 2:n)
  rv <- rbind(rv, snpgdsPairIBD(mat[,1], mat[,i], RF$AlleleFreq, "MoM"))
rv
summary(rv$k0 - subMoM$k0[1, 2:n])
summary(rv$k1 - subMoM$k1[1, 2:n])
# ZERO

rv <- NULL
for (i in 2:n)
  rv <- rbind(rv, snpgdsPairIBD(mat[,1], mat[,i], RF$AlleleFreq, "Jacquard"))
rv
summary(rv$D1 - subJac$D1[1, 2:n])
summary(rv$D2 - subJac$D2[1, 2:n])
# ZERO

# close the genotype file
snpGdsClose(genofile)

```

snpGdsPairIBDMLELogLik

Log likelihood for MLE method in the Identity-By-Descent (IBD) Analysis

Description

Calculate the log likelihood values from maximum likelihood estimation.

Usage

```
snpGdsPairIBDMLELogLik(geno1, geno2, allele.freq, k0=NaN, k1=NaN,
  relatedness=c("", "self", "fullsib", "offspring", "halfsib",
  "cousin", "unrelated"), verbose=TRUE)
```

Arguments

| | |
|-------------|---|
| geno1 | the SNP genotypes for the first individual, 0 – BB, 1 – AB, 2 – AA, other values – missing |
| geno2 | the SNP genotypes for the second individual, 0 – BB, 1 – AB, 2 – AA, other values – missing |
| allele.freq | the allele frequencies |
| k0 | specified IBD coefficient |
| k1 | specified IBD coefficient |
| relatedness | specify a relatedness, otherwise use the values of k0 and k1 |
| verbose | if TRUE, show information |

Details

If (relatedness == "") and (k0 == NaN or k1 == NaN), then return the log likelihood values for each (k0, k1) stored in ibdobj.

If (relatedness == "") and (k0 != NaN) and (k1 != NaN), then return the log likelihood values for a specific IBD coefficient (k0, k1).

If relatedness is: "self", then k0 = 0, k1 = 0; "fullsib", then k0 = 0.25, k1 = 0.5; "offspring", then k0 = 0, k1 = 1; "halfsib", then k0 = 0.5, k1 = 0.5; "cousin", then k0 = 0.75, k1 = 0.25; "unrelated", then k0 = 1, k1 = 0.

Value

The value of log likelihood.

Author(s)

Xiuwen Zheng

References

- Milligan BG. 2003. Maximum-likelihood estimation of relatedness. *Genetics* 163:1153-1167.
- Weir BS, Anderson AD, Hepler AB. 2006. Genetic relatedness analysis: modern data and new challenges. *Nat Rev Genet.* 7(10):771-80.
- Choi Y, Wijsman EM, Weir BS. 2009. Case-control association testing in the presence of unknown relationships. *Genet Epidemiol* 33(8):668-78.

See Also

[snpGdsPairIBD](#), [snpGdsIBDMLE](#), [snpGdsIBDMLELogLik](#), [snpGdsIBDMoM](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

YRI.id <- read.gdsn(index.gdsn(genofile, "sample.id"))[
  read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))=="YRI"]

# SNP pruning
set.seed(10)
snpset <- snpGdsLDpruning(genofile, sample.id=YRI.id, maf=0.05,
  missing.rate=0.05)
snpset <- unname(sample(unlist(snpset), 250))

# the number of samples
n <- 25

# specify allele frequencies
RF <- snpGdsSNPRateFreq(genofile, sample.id=YRI.id, snp.id=snpset,
  with.id=TRUE)
summary(RF$AlleleFreq)

subMLE <- snpGdsIBDMLE(genofile, sample.id=YRI.id[1:n], snp.id=RF$snp.id,
  allele.freq=RF$AlleleFreq)
subMoM <- snpGdsIBDMoM(genofile, sample.id=YRI.id[1:n], snp.id=RF$snp.id,
  allele.freq=RF$AlleleFreq)

# genotype matrix
mat <- snpGdsGetGeno(genofile, sample.id=YRI.id[1:n], snp.id=snpset,
  snpfirstdim=TRUE)

#####

rv <- NULL
for (i in 2:n)
{
  rv <- rbind(rv, snpGdsPairIBD(mat[,1], mat[,i], RF$AlleleFreq, "EM"))
  print(snpGdsPairIBDMLELogLik(mat[,1], mat[,i], RF$AlleleFreq,
    relatedness="unrelated", verbose=TRUE))
}
rv
summary(rv$k0 - subMLE$k0[1, 2:n])
summary(rv$k1 - subMLE$k1[1, 2:n])
# ZERO

rv <- NULL
for (i in 2:n)
```

```

    rv <- rbind(rv, snpgdsPairIBD(mat[,1], mat[,i], RF$AlleleFreq, "MoM"))
rv
summary(rv$k0 - subMoM$k0[1, 2:n])
summary(rv$k1 - subMoM$k1[1, 2:n])
# ZERO

# close the genotype file
snpgdsClose(genofile)

```

snpgdsPairScore

Genotype Score for Pairs of Individuals

Description

Calculate the genotype score for pairs of individuals based on identity-by-state (IBS) measure

Usage

```

snpgdsPairScore(gdsobj, sample1.id, sample2.id, snp.id=NULL,
  method=c("IBS", "GVH", "HVG", "GVH.major", "GVH.minor", "GVH.major.only",
    "GVH.minor.only"), type=c("per.pair", "per.snp", "matrix", "gds.file"),
  dosage=TRUE, with.id=TRUE, output=NULL, verbose=TRUE)

```

Arguments

| | |
|------------|--|
| gdsobj | an object of class SNPGDSFileClass , a SNP GDS file |
| sample1.id | a vector of sample id specifying selected samples; if NULL, all samples are used |
| sample2.id | a vector of sample id specifying selected samples; if NULL, all samples are used |
| snp.id | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| method | "IBS" – identity-by-state score, "GVH" or "HVG", see Details |
| type | "per.pair", "per.snp" or "matrix", see Value |
| dosage | TRUE, uses dosages 0, 1, 2; FALSE, uses 0, 1 (changing a return value of 1 or 2 to be 1) |
| with.id | if TRUE, returns "sample.id" and "snp.id"; see Value |
| output | if type="gds.file", the file name |
| verbose | if TRUE, show information |

Details

| sample1.id | sample2.id | IBS | GVH | HVG | GVH.major | GVH.minor | GVH.major.only | GVH.minor.only |
|------------|------------|-----|-----|-----|-----------|-----------|----------------|----------------|
| Patient | Donor | | | | | | | |
| AA / 2 | AA / 2 | 2 | 0 | 0 | 0 | 0 | 0 | 0 |
| AA / 2 | AB / 1 | 1 | 0 | 1 | 0 | 0 | 0 | 0 |
| AA / 2 | BB / 0 | 0 | 2 | 2 | 1 | 0 | 1 | NA |

| | | | | | | | | |
|--------|--------|---|---|---|---|---|----|----|
| AB / 1 | AA / 2 | 1 | 1 | 0 | 0 | 1 | NA | 1 |
| AB / 1 | AB / 1 | 2 | 0 | 0 | 0 | 0 | 0 | 0 |
| AB / 1 | BB / 0 | 1 | 1 | 0 | 1 | 0 | 1 | NA |
| BB / 0 | AA / 2 | 0 | 2 | 2 | 0 | 1 | NA | 1 |
| BB / 0 | AB / 1 | 1 | 0 | 1 | 0 | 0 | 0 | 0 |
| BB / 0 | BB / 0 | 2 | 0 | 0 | 0 | 0 | 0 | 0 |

Value

Return a list:

| | |
|-----------|---|
| sample.id | the sample ids used in the analysis, if with.id=TRUE |
| snp.id | the SNP ids used in the analysis, if with.id=TRUE |
| score | a matrix of genotype score: if type="per.pair", a data.frame with the first column for average scores, the second column for standard deviation and the third column for the valid number of SNPs; the additional columns for pairs of samples. if type="per.snp", a 3-by-# of SNPs matrix with the first row for average scores, the second row for standard deviation and the third row for the valid number of individual pairs; if type="matrix", a # of pairs-by-# of SNPs matrix with rows for pairs of individuals |

Author(s)

Xiuwen Zheng

References

Warren, E. H., Zhang, X. C., Li, S., Fan, W., Storer, B. E., Chien, J. W., Boeckh, M. J., et al. (2012). Effect of MHC and non-MHC donor/recipient genetic disparity on the outcome of allogeneic HCT. *Blood*, 120(14), 2796-806. doi:10.1182/blood-2012-04-347286

See Also

[snpGdsIBS](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

# autosomal SNPs
selsnp <- snpGdsSelectSNP(genofile, autosome.only=TRUE, remove.monosnp=FALSE)

# sample ID
sample.id <- read.gdsn(index.gdsn(genofile, "sample.id"))
father.id <- read.gdsn(index.gdsn(genofile, "sample.annot/father.id"))
```

```

offspring.id <- sample.id[father.id != ""]
father.id <- father.id[father.id != ""]

# calculate average genotype scores
z1 <- snpgdsPairScore(genofile, offspring.id, father.id, snp.id=selsnp,
  method="IBS", type="per.pair")
str(z1)
head(z1$score)

# calculate average genotype scores
z1 <- snpgdsPairScore(genofile, offspring.id, father.id, snp.id=selsnp,
  method="IBS", type="per.pair", dosage=FALSE)
str(z1)
head(z1$score)

# calculate average genotype scores
z2 <- snpgdsPairScore(genofile, offspring.id, father.id, snp.id=selsnp,
  method="IBS", type="per.snp")
str(z2)
z2$score[, 1:4]
mean(z2$score["Avg",])
mean(z2$score["SD",])

plot(z2$score["Avg",], pch=20, cex=0.75, xlab="SNP Index", ylab="IBS score")

# calculate a matrix of genotype scores over samples and SNPs
z3 <- snpgdsPairScore(genofile, offspring.id, father.id, snp.id=selsnp,
  method="IBS", type="matrix")
str(z3)

# output the score matrix to a GDS file
snpgdsPairScore(genofile, offspring.id, father.id, snp.id=selsnp,
  method="IBS", type="gds.file", output="tmp.gds")
(f <- snpgdsOpen("tmp.gds"))
snpgdsClose(f)

# close the file
snpgdsClose(genofile)

unlink("tmp.gds", force=TRUE)

```

snpGDS_PCA

Principal Component Analysis (PCA) on SNP genotype data

Description

To calculate the eigenvectors and eigenvalues for principal component analysis in GWAS.

Usage

```
snpGDSPCA(gdsobj, sample.id=NULL, snp.id=NULL,
  autosome.only=TRUE, remove.monosnp=TRUE, maf=NaN, missing.rate=NaN,
  algorithm=c("exact", "randomized"),
  eigen.cnt=ifelse(identical(algorithm, "randomized"), 16L, 32L),
  num.thread=1L, bayesian=FALSE, need.genmat=FALSE,
  genmat.only=FALSE, eigen.method=c("DSPEVX", "DSPEV"),
  aux.dim=eigen.cnt*2L, iter.num=10L, verbose=TRUE)
## S3 method for class 'snpGDSPCAclass'
plot(x, eig=c(1L,2L), ...)
```

Arguments

| | |
|-----------------------------|---|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples are used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| <code>autosome.only</code> | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| <code>remove.monosnp</code> | if TRUE, remove monomorphic SNPs |
| <code>maf</code> | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| <code>missing.rate</code> | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| <code>eigen.cnt</code> | output the number of eigenvectors; if eigen.cnt \leq 0, then return all eigenvectors |
| <code>algorithm</code> | "exact", traditional exact calculation; "randomized", fast PCA with randomized algorithm introduced in Galinsky et al. 2016 |
| <code>num.thread</code> | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| <code>bayesian</code> | if TRUE, use bayesian normalization |
| <code>need.genmat</code> | if TRUE, return the genetic covariance matrix |
| <code>genmat.only</code> | return the genetic covariance matrix only, do not compute the eigenvalues and eigenvectors |
| <code>eigen.method</code> | "DSPEVX" – compute the top eigen.cnt eigenvalues and eigenvectors using LAPACK::DSPEVX; "DSPEV" – to be compatible with SNPRelate_1.1.6 or earlier, using LAPACK::DSPEV; "DSPEVX" is significantly faster than "DSPEV" if only top principal components are of interest |
| <code>aux.dim</code> | auxiliary dimension used in fast randomized algorithm |
| <code>iter.num</code> | iteration number used in fast randomized algorithm |
| <code>verbose</code> | if TRUE, show information |
| <code>x</code> | a <code>snpGDSPCAclass</code> object |
| <code>eig</code> | indices of eigenvectors, like 1:2 or 1:4 |
| <code>...</code> | the arguments passed to or from other methods, like <code>pch</code> , <code>col</code> |

Details

The minor allele frequency and missing rate for each SNP passed in `snp.id` are calculated over all the samples in `sample.id`.

Value

Return a snpGDS-PCAClass object, and it is a list:

| | |
|-----------|--|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| eigenval | eigenvalues |
| eigenvect | eigenvectors, "# of samples" x "eigen.cnt" |
| varprop | variance proportion for each principal component |
| TraceXTX | the trace of the genetic covariance matrix |
| Bayesian | whether use bayesian normalization |
| genmat | the genetic covariance matrix |

Author(s)

Xiuwen Zheng

References

Patterson N, Price AL, Reich D. Population structure and eigenanalysis. PLoS Genet. 2006 Dec;2(12):e190.

Galinsky KJ, Bhatia G, Loh PR, Georgiev S, Mukherjee S, Patterson NJ, Price AL. Fast Principal-Component Analysis Reveals Convergent Evolution of ADH1B in Europe and East Asia. Am J Hum Genet. 2016 Mar 3;98(3):456-72. doi: 10.1016/j.ajhg.2015.12.022. Epub 2016 Feb 25.

See Also

[snpGDS-PCACorr](#), [snpGDS-PCASNPLoading](#), [snpGDS-PCASampLoading](#), [snpGDS-AdmixProp](#), [snpGDS-EIGMIX](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())

# run PCA
RV <- snpGDSPCA(genofile)
RV

# eigenvalues
head(RV$eigenval)

# variance proportion (%)
head(round(RV$varprop*100, 2))
# [1] 12.23 5.84 1.01 0.95 0.84 0.74

# draw
plot(RV)
plot(RV, 1:4)
```

```

#### there is no population information ####

# make a data.frame
tab <- data.frame(sample.id = RV$sample.id,
  EV1 = RV$eigenvect[,1], # the first eigenvector
  EV2 = RV$eigenvect[,2], # the second eigenvector
  stringsAsFactors = FALSE)
head(tab)
#   sample.id      EV1      EV2
# 1 NA19152 -0.08411287 -0.01226860
# 2 NA19139 -0.08360644 -0.01085849
# 3 NA18912 -0.08110808 -0.01184524
# 4 NA19160 -0.08680864 -0.01447106
# 5 NA07034  0.03109761  0.07709255
# 6 NA07055  0.03228450  0.08155730

# draw
plot(tab$EV2, tab$EV1, xlab="eigenvector 2", ylab="eigenvector 1")

#### there are population information ####

# get population information
# or pop_code <- scan("pop.txt", what=character())
# if it is stored in a text file "pop.txt"
pop_code <- read.gdsn(index.gdsn(genofile, "sample.annot/pop.group"))

# get sample id
samp.id <- read.gdsn(index.gdsn(genofile, "sample.id"))

# assume the order of sample IDs is as the same as population codes
cbind(samp.id, pop_code)
#      samp.id      pop_code
# [1,] "NA19152"      "YRI"
# [2,] "NA19139"      "YRI"
# [3,] "NA18912"      "YRI"
# [4,] "NA19160"      "YRI"
# [5,] "NA07034"      "CEU"
# ...

# make a data.frame
tab <- data.frame(sample.id = RV$sample.id,
  pop = factor(pop_code)[match(RV$sample.id, samp.id)],
  EV1 = RV$eigenvect[,1], # the first eigenvector
  EV2 = RV$eigenvect[,2], # the second eigenvector
  stringsAsFactors = FALSE)
head(tab)
#   sample.id pop      EV1      EV2
# 1 NA19152 YRI -0.08411287 -0.01226860
# 2 NA19139 YRI -0.08360644 -0.01085849
# 3 NA18912 YRI -0.08110808 -0.01184524

```

```
# 4 NA19160 YRI -0.08680864 -0.01447106
# 5 NA07034 CEU 0.03109761 0.07709255
# 6 NA07055 CEU 0.03228450 0.08155730

# draw
plot(tab$EV2, tab$EV1, col=as.integer(tab$pop),
      xlab="eigenvector 2", ylab="eigenvector 1")
legend("bottomright", legend=levels(tab$pop), pch="o", col=1:4)

# close the file
snpgdsClose(genofile)
```

snpgdsPCACorr

PC-correlated SNPs in principal component analysis

Description

To calculate the SNP correlations between eigenvectors and SNP genotypes

Usage

```
snpgdsPCACorr(pcaobj, gdsobj, snp.id=NULL, eig.which=NULL, num.thread=1L,
              with.id=TRUE, outgds=NULL, verbose=TRUE)
```

Arguments

| | |
|------------|--|
| pcaobj | a snpgdsPCAclass object returned from the function snpgdsPCA , a snpgdsEigMixClass from snpgdsEIGMIX , or an eigenvector matrix with row names (sample id) |
| gdsobj | an object of class SNPGDSFileClass , a SNP GDS file |
| snp.id | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| eig.which | a vector of integers, to specify which eigenvectors to be used |
| num.thread | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| with.id | if TRUE, the returned value with <code>sample.id</code> and <code>sample.id</code> |
| outgds | NULL or a character of file name for exporting correlations to a GDS file, see details |
| verbose | if TRUE, show information |

Details

If an output file name is specified via `outgds`, "sample.id", "snp.id" and "correlation" will be stored in the GDS file. The GDS node "correlation" is a matrix of correlation coefficients, and it is stored with the format of packed real number ("packedreal16" preserving 4 digits, 0.0001 is the smallest number greater zero, see [add.gdsn](#)).

Value

Return a list if outgds=NULL,

| | |
|-----------|---|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| snpcorr | a matrix of correlation coefficients, "# of eigenvectors" x "# of SNPs" |

Author(s)

Xiuwen Zheng

References

Patterson N, Price AL, Reich D (2006) Population structure and eigenanalysis. PLoS Genetics 2:e190.

See Also

[snpgdsPCA](#), [snpgdsPCASampLoading](#), [snpgdsPCASNPLoading](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())
# get chromosome index
chr <- read.gdsn(index.gdsn(genofile, "snp.chromosome"))

pca <- snpgdsPCA(genofile)
cr <- snpgdsPCACorr(pca, genofile, eig.which=1:4)
plot(abs(cr$snpcorr[3,]), xlab="SNP Index", ylab="PC 3", col=chr)

# output to a gds file if limited memory
snpgdsPCACorr(pca, genofile, eig.which=1:4, outgds="test.gds")

(f <- openfn.gds("test.gds"))
m <- read.gdsn(index.gdsn(f, "correlation"))
closefn.gds(f)

# check
summary(c(m - cr$snpcorr)) # should < 1e-4

# close the file
snpgdsClose(genofile)

# delete the temporary file
unlink("test.gds", force=TRUE)
```

snpgdsPCASampLoading *Project individuals onto existing principal component axes*

Description

To calculate the sample eigenvectors using the specified SNP loadings

Usage

```
snpgdsPCASampLoading(loadobj, gdsobj, sample.id=NULL, num.thread=1L,
  verbose=TRUE)
```

Arguments

| | |
|------------|--|
| loadobj | a snpgdsPCASNPLoadingClass or snpgdsEigMixSNPLoadingClass object returned from snpgdsPCASNPLoading |
| gdsobj | an object of class SNPGDSFileClass , a SNP GDS file |
| sample.id | a vector of sample id specifying selected samples; if NULL, all samples are used |
| num.thread | the number of CPU cores used |
| verbose | if TRUE, show information |

Details

The sample.id are usually different from the samples used in the calculation of SNP loadings.

Value

Returns a snpgdsPCAClass object, and it is a list:

| | |
|-----------|--|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| eigenval | eigenvalues |
| eigenvect | eigenvectors, “# of samples” x “eigen.cnt” |
| TraceXTX | the trace of the genetic covariance matrix |
| Bayesian | whether use bayerisan normalization |

Or returns a snpgdsEigMixClass object, and it is a list:

| | |
|-----------|--|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| eigenval | eigenvalues |
| eigenvect | eigenvectors, “# of samples” x “eigen.cnt” |
| afreq | allele frequencies |

Author(s)

Xiuwen Zheng

References

Patterson N, Price AL, Reich D (2006) Population structure and eigenanalysis. *PLoS Genetics* 2:e190.

Zhu, X., Li, S., Cooper, R. S., and Elston, R. C. (2008). A unified association analysis approach for family and unrelated samples correcting for stratification. *Am J Hum Genet*, 82(2), 352-365.

See Also

[snpGdsPCA](#), [snpGdsPCACorr](#), [snpGdsPCASNPLoading](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGdsOpen(snpGdsExampleFileName())

sample.id <- read.gdsn(index.gdsn(genofile, "sample.id"))

# first PCA
pca <- snpGdsPCA(genofile, eigen.cnt=8)
snp_load <- snpGdsPCASNPLoading(pca, genofile)

# calculate sample eigenvectors from SNP loadings
snp_load <- snpGdsPCASampLoading(snp_load, genofile, sample.id=sample.id[1:100])

diff <- pca$eigenvect[1:100,] - snp_load$eigenvect
summary(c(diff))
# ~ ZERO

# combine eigenvectors
allpca <- list(
  sample.id = c(pca$sample.id, snp_load$sample.id),
  snp.id = pca$snp.id,
  eigenval = c(pca$eigenval, snp_load$eigenval),
  eigenvect = rbind(pca$eigenvect, snp_load$eigenvect),
  varprop = c(pca$varprop, snp_load$varprop),
  TraceXTX = pca$TraceXTX
)
class(allpca) <- "snpGdsPCAClass"
allpca

# close the genotype file
snpGdsClose(genofile)
```

snpgdsPCASNPLoading *SNP loadings in principal component analysis*

Description

To calculate the SNP loadings in Principal Component Analysis

Usage

```
snpgdsPCASNPLoading(pcaobj, gdsobj, num.thread=1L, verbose=TRUE)
```

Arguments

| | |
|------------|---|
| pcaobj | a snpgdsPCAClass object returned from the function snpgdsPCA or a snpgdsEigMixClass from snpgdsEIGMIX |
| gdsobj | an object of class SNPGDSFileClass , a SNP GDS file |
| num.thread | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| verbose | if TRUE, show information |

Details

Calculate the SNP loadings (or SNP eigenvectors) from the principal component analysis conducted in snpgdsPCA.

Value

Returns a snpgdsPCASNPLoading object if pcaobj is snpgdsPCAClass, which is a list:

| | |
|-----------|--|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| eigenval | eigenvalues |
| snploding | SNP loadings, or SNP eigenvectors |
| TraceXTX | the trace of the genetic covariance matrix |
| Bayesian | whether use bayerisan normalization |
| avgfreq | two times allele frequency used in snpgdsPCA |
| scale | internal parameter |

Or returns a snpgdsEigMixSNPLoadingClass object if pcaobj is snpgdsEigMixClass, which is a list:

| | |
|-----------|-------------------------------------|
| sample.id | the sample ids used in the analysis |
| snp.id | the SNP ids used in the analysis |
| eigenval | eigenvalues |
| snploding | SNP loadings, or SNP eigenvectors |
| afreq | allele frequency |

Author(s)

Xiuwen Zheng

References

Patterson N, Price AL, Reich D (2006) Population structure and eigenanalysis. *PLoS Genetics* 2:e190.

Price AL, Patterson NJ, Plenge RM, Weinblatt ME, Shadick NA, Reich D (2006) Principal components analysis corrects for stratification in genome-wide association studies. *Nat Genet.* 38, 904-909.

Zhu, X., Li, S., Cooper, R. S., and Elston, R. C. (2008). A unified association analysis approach for family and unrelated samples correcting for stratification. *Am J Hum Genet*, 82(2), 352-365.

See Also

[snpGDSPCA](#), [snpGDS EIGMIX](#), [snpGDSPCASampLoading](#), [snpGDSPCACorr](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())

PCARV <- snpGDSPCA(genofile, eigen.cnt=8)
SnpLoad <- snpGDSPCASNPLoading(PCARV, genofile)

names(SnpLoad)
# [1] "sample.id" "snp.id" "eigenval" "snploading" "TraceXTX"
# [6] "Bayesian" "avgfreq" "scale"
dim(SnpLoad$snploading)
# [1] 8 8722

plot(SnpLoad$snploading[1,], type="h", ylab="PC 1")

# close the genotype file
snpGSDSClose(genofile)
```

snpGDSPED2GDS

Conversion from PLINK PED to GDS

Description

Convert a PLINK PED text file to a GDS file.

Usage

```
snpGDSPED2GDS(ped.fn, map.fn, out.gdsfn, family=TRUE, snpfirstdim=FALSE,
  compress.annotation="ZIP_RA.max", compress.geno="", verbose=TRUE)
```

Arguments

| | |
|---------------------|--|
| ped.fn | the file name of PED file, genotype information |
| map.fn | the file name of MAP file |
| out.gdsfn | the output GDS file |
| family | if TRUE, to include family information in the sample annotation |
| snpfirstdim | if TRUE, genotypes are stored in the individual-major mode, (i.e, list all SNPs for the first individual, and then list all SNPs for the second individual, etc) |
| compress.annotation | the compression method for the GDS variables, except "genotype"; optional values are defined in the function add.gdsn |
| compress.geno | the compression method for "genotype"; optional values are defined in the function add.gdsn |
| verbose | if TRUE, show information |

Details

GDS – Genomic Data Structures, the extended file name used for storing genetic data, and the file format is used in the [gdsfmt](#) package.

PED – PLINK PED format.

Value

None.

Author(s)

Xiuwen Zheng

References

Purcell S, Neale B, Todd-Brown K, Thomas L, Ferreira MAR, Bender D, Maller J, Sklar P, de Bakker PIW, Daly MJ & Sham PC. 2007. PLINK: a toolset for whole-genome association and population-based linkage analysis. *American Journal of Human Genetics*, 81.

See Also

[snpgdsGDS2PED](#), [snpgdsBED2GDS](#), [snpgdsGDS2BED](#)

Examples

```
# open
genofile <- snpgdsOpen(snpgdsExampleFileName())

snpgdsGDS2PED(genofile, "tmp")

# close
snpgdsClose(genofile)
```

```
# PED ==> GDS
snpgdsPED2GDS("tmp.ped", "tmp.map", "test.gds")

# delete the temporary file
unlink(c("tmp.ped", "tmp.map", "test.gds"), force=TRUE)
```

snpgdsSampMissRate *Missing Rate of Samples*

Description

Return the missing fraction for each sample

Usage

```
snpgdsSampMissRate(gdsobj, sample.id=NULL, snp.id=NULL, with.id=FALSE)
```

Arguments

| | |
|-----------|--|
| gdsobj | an object of class SNPGDSFileClass , a SNP GDS file |
| sample.id | a vector of sample id specifying selected samples; if NULL, all samples will be used |
| snp.id | a vector of snp id specifying selected SNPs; if NULL, all SNPs will be used |
| with.id | if TRUE, the returned value with sample id |

Value

A vector of numeric values.

Author(s)

Xiuwen Zheng

See Also

[snpgdsSNPRateFreq](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

RV <- snpgdsSampMissRate(genofile)
summary(RV)

# close the genotype file
snpgdsClose(genofile)
```

snpgdsSelectSNP *SNP selection*

Description

Create a list of candidate SNPs based on specified criteria

Usage

```
snpgdsSelectSNP(gdsobj, sample.id=NULL, snp.id=NULL, autosome.only=TRUE,
  remove.monosnp=TRUE, maf=NaN, missing.rate=NaN, verbose=TRUE)
```

Arguments

| | |
|-----------------------------|--|
| <code>gdsobj</code> | an object of class SNPGDSFileClass , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples will be used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs will be used |
| <code>autosome.only</code> | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| <code>remove.monosnp</code> | if TRUE, remove monomorphic SNPs |
| <code>maf</code> | to use the SNPs with " \geq maf" only; if NaN, no any MAF threshold |
| <code>missing.rate</code> | to use the SNPs with " \leq missing.rate" only; if NaN, no any missing threshold |
| <code>verbose</code> | if TRUE, show information |

Value

Return a list of snp ids.

Author(s)

Xiuwen Zheng

See Also

[snpgdsSampMissRate](#), [snpgdsSNPRateFreq](#), [snpgdsLDpruning](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

snpset <- snpgdsSelectSNP(genofile, maf=0.05, missing.rate=0.95)
length(snpset)
# 7502

# close the genotype file
snpgdsClose(genofile)
```

snpGDSslidingWindow *Sliding window*

Description

Apply a user-defined function with a sliding window.

Usage

```
snpGDSslidingWindow(gdsobj, sample.id=NULL, snp.id=NULL,
  FUN=NULL, winsize=100000L, shift=10000L, unit=c("basepair", "locus"),
  winstart=NULL, autosome.only=FALSE, remove.monosnp=TRUE, maf=NaN,
  missing.rate=NaN, as.is=c("list", "numeric", "array"),
  with.id=c("snp.id", "snp.id.in.window", "none"), num.thread=1,
  verbose=TRUE, ...)
```

Arguments

| | |
|----------------|---|
| gdsobj | an object of class SNPGDSFileClass , a SNP GDS file |
| sample.id | a vector of sample id specifying selected samples; if NULL, all samples are used |
| snp.id | a vector of snp id specifying selected SNPs; if NULL, all SNPs are used |
| FUN | a character or a user-defined function, see details |
| winsize | the size of sliding window |
| shift | the amount of shifting the sliding window |
| unit | "basepair" – winsize and shift are applied with SNP coordinate of basepair; "locus" – winsize and shift are applied according to the SNP order in the GDS file |
| winstart | NULL – no specific starting position; an integer – a starting position for all chromosomes; or a vector of integer – the starting positions for each chromosome |
| autosome.only | if TRUE, use autosomal SNPs only; if it is a numeric or character value, keep SNPs according to the specified chromosome |
| remove.monosnp | if TRUE, remove monomorphic SNPs |
| maf | to use the SNPs with " \geq maf" only; if NaN, no MAF threshold |
| missing.rate | to use the SNPs with " \leq missing.rate" only; if NaN, no missing threshold |
| as.is | save the value returned from FUN as "list" or "numeric"; "array" is equivalent to "numeric" except some cases, see details |
| with.id | "snp.id", "snp.id.in.window" or "none" |
| num.thread | the number of (CPU) cores used; if NA, detect the number of cores automatically |
| verbose | if TRUE, show information |
| ... | optional arguments to FUN |

Details

If FUN="snpGDSFst", two additional arguments "population" and "method" should be specified. "population" and "method" are defined in [snpGDSFst](#). "as.is" could be "list" (returns a list of the values from [snpGDSFst](#)), "numeric" (population-average Fst, returns a vector) or "array" (population-average and -specific Fst, returns a '# of pop + 1'-by-'# of windows' matrix, and the first row is population-average Fst).

Value

Return a list

Author(s)

Xiuwen Zheng

Examples

```
# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())

# sliding windows
rv <- snpGDSslidingWindow(genofile, winsize=500000, shift=100000,
  FUN=function(...) NULL)

# plot
plot(rv$chr1.num, ylab="# of SNPs in the sliding window")

# close the genotype file
snpGDSClose(genofile)
```

snpGDSNPList

Create a SNP list object

Description

A list object of SNP information including rs, chr, pos, allele and allele frequency.

Usage

```
snpGDSNPList(gdsobj, sample.id=NULL)
```

Arguments

gdsobj an object of class [SNPGDSFileClass](#), a SNP GDS file
 sample.id a vector of sample id specifying selected samples; if NULL, all samples are used

Value

Return an object of `snpGDS_SNPListClass` including the following components:

| | |
|-------------------------|-----------------------------------|
| <code>snp.id</code> | SNP id |
| <code>chromosome</code> | SNP chromosome index |
| <code>position</code> | SNP physical position in basepair |
| <code>allele</code> | reference / non-ref alleles |
| <code>afreq</code> | allele frequency |

Author(s)

Xiuwen Zheng

See Also

[snpGDS_SNPListIntersect](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpGDSOpen(snpGDSExampleFileName())

# to get a snp list object
snplist <- snpGDS_SNPList(genofile)
head(snplist)

# close the file
snpGDS_Close(genofile)
```

`snpGDS_SNPListClass` *the class of a SNP list*

Description

the class of a SNP list, and its instance is returned from [snpGDS_SNPList](#).

Value

Return an object of “`snpGDS_SNPListClass`” including the following components:

| | |
|-------------------------|-----------------------------------|
| <code>snp.id</code> | SNP id |
| <code>chromosome</code> | SNP chromosome index |
| <code>position</code> | SNP physical position in basepair |
| <code>allele</code> | reference / non-ref alleles |
| <code>afreq</code> | allele frequency |

Author(s)

Xiuwen Zheng

See Also[snpGDSNPList](#), [snpGDSNPListIntersect](#)

`snpGDSNPListIntersect`*Get a common SNP list between/among SNP list objects*

Description

Get a common SNP list by comparing their snp id, chromosome, positions and allele frequency if needed.

Usage

```
snpGDSNPListIntersect(snpList1, snpList2, ..., method=c("position", "exact"),
  na.rm=TRUE, same.strand=FALSE, verbose=TRUE)
```

Arguments

| | |
|--------------------------|--|
| <code>snpList1</code> | the SNP list object snpGDSNPListClass |
| <code>snpList2</code> | the SNP list object snpGDSNPListClass |
| <code>...</code> | the other SNP list objects |
| <code>method</code> | "exact": matching by all snp.id, chromosomes, positions and alleles; "position": matching by chromosomes and positions |
| <code>na.rm</code> | if TRUE, remove mismatched alleles |
| <code>same.strand</code> | if TRUE, assuming the alleles on the same strand |
| <code>verbose</code> | if TRUE, show information |

Value

Return a list of `snpGDSNPListClass` including the following components:

| | |
|---------------------|--|
| <code>idx1</code> | the indices of common SNPs in the first GDS file |
| <code>idx2</code> | the indices of common SNPs in the second GDS file |
| <code>idx...</code> | |
| <code>idxn</code> | the indices of common SNPs in the n-th GDS file |
| <code>flag2</code> | an integer vector, flip flag for each common SNP for the second GDS file (assuming a value <code>v</code>): <code>bitwAnd(v, 1)</code> : 0 – no flip of allele names, 1 – flip of allele names; <code>bitwAnd(v, 2)</code> : 0 – on the same strand, 2 – on the different strands, comparing with the first GDS file; <code>bitwAnd(v, 4)</code> : 0 – no strand ambiguity, 4 – ambiguous allele names, determined by allele frequencies; NA – mismatched allele names (there is no NA if <code>na.rm=TRUE</code>) |


```
flag...
flagn          flip flag for each common SNP for the n-th GDS file
```

Author(s)

Xiuwen Zheng

See Also

[snpgdsSNPList](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

# to get a snp list object
snplist1 <- snpgdsSNPList(genofile)
snplist2 <- snpgdsSNPList(genofile)

# a common snp list, exactly matching
v <- snpgdsSNPListIntersect(snplist1, snplist2)
names(v)
# "idx1" "idx2"

# a common snp list, matching by position
v <- snpgdsSNPListIntersect(snplist1, snplist2, method="pos")
names(v)
# "idx1" "idx2" "flag2"

table(v$flag2, exclude=NULL)

# close the file
snpgdsClose(genofile)
```

snpgdsSNPRateFreq *Allele Frequency, Minor Allele Frequency, Missing Rate of SNPs*

Description

Calculate the allele frequency, minor allele frequency and missing rate per SNP.

Usage

```
snpgdsSNPRateFreq(gdsobj, sample.id=NULL, snp.id=NULL, with.id=FALSE,
  with.sample.id=FALSE, with.snp.id=FALSE)
```

Arguments

| | |
|-----------------------------|--|
| <code>gdsobj</code> | an object of class <code>SNPGDSFileClass</code> , a SNP GDS file |
| <code>sample.id</code> | a vector of sample id specifying selected samples; if NULL, all samples will be used |
| <code>snp.id</code> | a vector of snp id specifying selected SNPs; if NULL, all SNPs will be used |
| <code>with.id</code> | if TRUE, return both sample and SNP IDs |
| <code>with.sample.id</code> | if TRUE, return sample IDs |
| <code>with.snp.id</code> | if TRUE, return SNP IDs |

Value

Return a list:

| | |
|--------------------------|---|
| <code>AlleleFreq</code> | allele frequencies |
| <code>MinorFreq</code> | minor allele frequencies |
| <code>MissingRate</code> | missing rates |
| <code>sample.id</code> | sample id, if <code>with.id=TRUE</code> or <code>with.sample.id=TRUE</code> |
| <code>snp.id</code> | SNP id, if <code>with.id=TRUE</code> or <code>with.snp.id=TRUE</code> |

Author(s)

Xiuwen Zheng

See Also

[snpgdsSampMissRate](#)

Examples

```
# open an example dataset (HapMap)
genofile <- snpgdsOpen(snpgdsExampleFileName())

RV <- snpgdsSNPRateFreq(genofile, with.snp.id=TRUE)
head(data.frame(RV))

hist(RV$AlleleFreq, breaks=128)
summary(RV$MissingRate)

# close the file
snpgdsClose(genofile)
```

| | |
|---------------|-------------------------------------|
| snpGDSsummary | <i>Summary of GDS genotype file</i> |
|---------------|-------------------------------------|

Description

Print the information stored in the gds object

Usage

```
snpGDSsummary(gds, show=TRUE)
```

Arguments

| | |
|------|--|
| gds | a GDS file name, or an object of class SNPGDSfileClass |
| show | if TRUE, show information |

Value

Return a list:

| | |
|-----------|--------------------------|
| sample.id | the IDs of valid samples |
| snp.id | the IDs of valid SNPs |

Author(s)

Xiuwen Zheng

Examples

```
snpGDSsummary(snpGDSexampleFileName())
```

| | |
|-----------------|-----------------------------------|
| snpGDSTranspose | <i>Transpose genotypic matrix</i> |
|-----------------|-----------------------------------|

Description

Transpose the genotypic matrix if needed.

Usage

```
snpGDSTranspose(gds.fn, snpfirstdim=FALSE, compress=NULL, optimize=TRUE,  
verbose=TRUE)
```

Arguments

| | |
|-------------|---|
| gds.fn | the file name of SNP GDS format |
| snpfirstdim | if TRUE, genotypes are stored in snp-by-sample; if FALSE, sample-by-snp mode; if NA, force to transpose the SNP matrix |
| compress | the compression mode for SNP genotypes, optional values are defined in the function of add.gdsn; if NULL, to use the compression mode |
| optimize | if TRUE, call <code>cleanup.gds</code> after transposing |
| verbose | if TRUE, show information |

Value

None.

Author(s)

Xiuwen Zheng

Examples

```
# the file name of SNP GDS
(fn <- snpgdsExampleFileName())

# copy the file
file.copy(fn, "test.gds", overwrite=TRUE)

# summary
snpgdsSummary("test.gds")

# transpose the SNP matrix
snpgdsTranspose("test.gds", snpfirstdim=TRUE)

# summary
snpgdsSummary("test.gds")

# delete the temporary file
unlink("test.gds", force=TRUE)
```

snpgdsVCF2GDS

Reformat VCF file(s)

Description

Reformat Variant Call Format (VCF) file(s)

Usage

```
snpGDSVCF2GDS(vcf.fn, out.fn, method=c("biallelic.only", "copy.num.of.ref"),
  snpfirstdim=FALSE, compress.annotation="LZMA_RA", compress.geno="",
  ref.allele=NULL, ignore.chr.prefix="chr", verbose=TRUE)
```

Arguments

| | |
|----------------------------------|--|
| <code>vcf.fn</code> | the file name of VCF format, <code>vcf.fn</code> can be a vector, see details |
| <code>out.fn</code> | the file name of output GDS |
| <code>method</code> | either "biallelic.only" by default or "copy.num.of.ref", see details |
| <code>snpfirstdim</code> | if TRUE, genotypes are stored in the individual-major mode, (i.e, list all SNPs for the first individual, and then list all SNPs for the second individual, etc) |
| <code>compress.annotation</code> | the compression method for the GDS variables, except "genotype"; optional values are defined in the function <code>add.gdsn</code> |
| <code>compress.geno</code> | the compression method for "genotype"; optional values are defined in the function <code>add.gdsn</code> |
| <code>ref.allele</code> | NULL or a character vector indicating reference allele (like "A", "G", "T", NA, ...) for each site where NA to use the original reference allele in the VCF file(s). The length of character vector should be the total number of variants in the VCF file(s). |
| <code>ignore.chr.prefix</code> | a vector of character, indicating the prefix of chromosome which should be ignored, like "chr"; it is not case-sensitive |
| <code>verbose</code> | if TRUE, show information |

Details

GDS – Genomic Data Structures used for storing genetic array-oriented data, and the file format used in the [gdsfmt](#) package.

VCF – The Variant Call Format (VCF), which is a generic format for storing DNA polymorphism data such as SNPs, insertions, deletions and structural variants, together with rich annotations.

If there are more than one file names in `vcf.fn`, `snpGDSVCF2GDS` will merge all dataset together if they all contain the same samples. It is useful to combine genetic/genomic data together if VCF data are divided by chromosomes.

`method = "biallelic.only"`: to exact bi-allelic and polymorphic SNP data (excluding monomorphic variants); `method = "copy.num.of.ref"`: to extract and store dosage (0, 1, 2) of the reference allele for all variant sites, including bi-allelic SNPs, multi-allelic SNPs, indels and structural variants.

Haploid and triploid calls are allowed in the transfer, the variable `snp.id` stores the original the row index of variants, and the variable `snp.rs.id` stores the rs id.

When `snp.chromosome` in the GDS file is character, `SNPRelate` treats a chromosome as auto-some only if it can be converted to a numeric value (like "1", "22"). It uses "X" and "Y" for non-autosomes instead of numeric codes. However, some software format chromosomes in VCF

files with a prefix "chr". Users should remove that prefix when importing VCF files by setting `ignore.chr.prefix = "chr"`.

The extended GDS format is implemented in the SeqArray package to support the storage of single nucleotide variation (SNV), insertion/deletion polymorphism (indel) and structural variation calls. It is strongly suggested to use SeqArray for large-scale whole-exome and whole-genome sequencing variant data instead of SNPRelate.

Value

Return the file name of GDS format with an absolute path.

Author(s)

Xiuwen Zheng

References

The variant call format and VCFtools. Danecek P, Auton A, Abecasis G, Albers CA, Banks E, DePristo MA, Handsaker RE, Lunter G, Marth GT, Sherry ST, McVean G, Durbin R; 1000 Genomes Project Analysis Group. *Bioinformatics*. 2011 Aug 1;27(15):2156-8. Epub 2011 Jun 7.

<http://corearray.sourceforge.net/>

See Also

[snpgdsBED2GDS](#)

Examples

```
# the VCF file
vcf.fn <- system.file("extdata", "sequence.vcf", package="SNPRelate")
cat(readLines(vcf.fn), sep="\n")

snpgdsVCF2GDS(vcf.fn, "test1.gds", method="biallelic.only")
snpgdsSummary("test1.gds")

snpgdsVCF2GDS(vcf.fn, "test2.gds", method="biallelic.only", snpfirstdim=TRUE)
snpgdsSummary("test2.gds")

snpgdsVCF2GDS(vcf.fn, "test3.gds", method="copy.num.of.ref", snpfirstdim=TRUE)
snpgdsSummary("test3.gds")

snpgdsVCF2GDS(vcf.fn, "test4.gds", method="copy.num.of.ref")
snpgdsSummary("test4.gds")

snpgdsVCF2GDS(vcf.fn, "test5.gds", method="copy.num.of.ref",
  ref.allele=c("A", "T", "T", "T", "A"))
snpgdsSummary("test5.gds")

# open "test1.gds"
```

```
(genofile <- snpGDSOpen("test1.gds"))

read.gdsn(index.gdsn(genofile, "sample.id"))
read.gdsn(index.gdsn(genofile, "snp.rs.id"))
read.gdsn(index.gdsn(genofile, "genotype"))

# close the file
snpGDSClose(genofile)

# open "test2.gds"
(genofile <- snpGDSOpen("test2.gds"))

read.gdsn(index.gdsn(genofile, "sample.id"))
read.gdsn(index.gdsn(genofile, "snp.rs.id"))
read.gdsn(index.gdsn(genofile, "genotype"))

# close the file
snpGDSClose(genofile)

# open "test3.gds"
(genofile <- snpGDSOpen("test3.gds"))

read.gdsn(index.gdsn(genofile, "sample.id"))
read.gdsn(index.gdsn(genofile, "snp.rs.id"))
read.gdsn(index.gdsn(genofile, "genotype"))

# close the file
snpGDSClose(genofile)

# open "test4.gds"
(genofile <- snpGDSOpen("test4.gds"))

read.gdsn(index.gdsn(genofile, "sample.id"))
read.gdsn(index.gdsn(genofile, "snp.rs.id"))
read.gdsn(index.gdsn(genofile, "snp.allele"))
read.gdsn(index.gdsn(genofile, "genotype"))

# close the file
snpGDSClose(genofile)

# open "test5.gds"
(genofile <- snpGDSOpen("test5.gds"))

read.gdsn(index.gdsn(genofile, "sample.id"))
read.gdsn(index.gdsn(genofile, "snp.rs.id"))
read.gdsn(index.gdsn(genofile, "snp.allele"))
read.gdsn(index.gdsn(genofile, "genotype"))

# close the file
```

```
snpGDSclose(genofile)

# delete the temporary files
unlink(paste("test", 1:5, ".gds", sep=""), force=TRUE)
```

snpGDSVCF2GDS_R *Reformat a VCF file (R implementation)*

Description

Reformat a Variant Call Format (VCF) file

Usage

```
snpGDSVCF2GDS_R(vcf.fn, out.fn, nblock=1024,
  method = c("biallelic.only", "copy.num.of.ref"),
  compress.annotation="LZMA_RA", snpfirstdim=FALSE, option = NULL,
  verbose=TRUE)
```

Arguments

| | |
|---------------------|--|
| vcf.fn | the file name of VCF format, vcf.fn can be a vector, see details |
| out.fn | the output gds file |
| nblock | the buffer lines |
| method | either "biallelic.only" by default or "copy.num.of.ref", see details |
| compress.annotation | the compression method for the GDS variables, except "genotype"; optional values are defined in the function add.gdsn |
| snpfirstdim | if TRUE, genotypes are stored in the individual-major mode, (i.e, list all SNPs for the first individual, and then list all SNPs for the second individual, etc) |
| option | NULL or an object from snpGDSOption , see details |
| verbose | if TRUE, show information |

Details

GDS – Genomic Data Structures used for storing genetic array-oriented data, and the file format used in the [gdsfmt](#) package.

VCF – The Variant Call Format (VCF), which is a generic format for storing DNA polymorphism data such as SNPs, insertions, deletions and structural variants, together with rich annotations.

If there are more than one file name in vcf.fn, snpGDSVCF2GDS will merge all dataset together once they all contain the same samples. It is useful to combine genetic data if VCF data are divided by chromosomes.

method = "biallelic.only": to exact bi-allelic and polymorphic SNP data (excluding monomorphic variants); method = "biallelic.only": to exact bi-allelic and polymorphic SNP data; method

= "copy.num.of.ref": to extract and store dosage (0, 1, 2) of the reference allele for all variant sites, including bi-allelic SNPs, multi-allelic SNPs, indels and structural variants.

Haploid and triploid calls are allowed in the transfer, the variable `snp.id` stores the original the row index of variants, and the variable `snp.rs.id` stores the rs id.

The user could use `option` to specify the range of code for autosomes. For humans there are 22 autosomes (from 1 to 22), but dogs have 38 autosomes. Note that the default settings are used for humans. The user could call `option = snpGDSOption(autosome.end=38)` for importing the VCF file of dog. It also allows defining new chromosome coding, e.g., `option = snpGDSOption(Z=27)`, then "Z" will be replaced by the number 27.

Value

None.

Author(s)

Xiuwen Zheng

References

The variant call format and VCFtools. Danecek P, Auton A, Abecasis G, Albers CA, Banks E, DePristo MA, Handsaker RE, Lunter G, Marth GT, Sherry ST, McVean G, Durbin R; 1000 Genomes Project Analysis Group. *Bioinformatics*. 2011 Aug 1;27(15):2156-8. Epub 2011 Jun 7.

See Also

[snpGDSVCF2GDS_R](#), [snpGDSOption](#), [snpGDSBED2GDS](#)

Examples

```
# The VCF file
vcf.fn <- system.file("extdata", "sequence.vcf", package="SNPRelate")
cat(readLines(vcf.fn), sep="\n")

snpGDSVCF2GDS_R(vcf.fn, "test1.gds", method="biallelic.only")
snpGDSSummary("test1.gds")

snpGDSVCF2GDS_R(vcf.fn, "test2.gds", method="biallelic.only")
snpGDSSummary("test2.gds")

snpGDSVCF2GDS_R(vcf.fn, "test3.gds", method="copy.num.of.ref")
snpGDSSummary("test3.gds")

snpGDSVCF2GDS_R(vcf.fn, "test4.gds", method="copy.num.of.ref")
snpGDSSummary("test4.gds")
```

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