

Package ‘genetics’

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Description Classes and methods for handling genetic data. Includes classes to represent genotypes and haplotypes at single markers up to multiple markers on multiple chromosomes. Function include allele frequencies, flagging homo/heterozygotes, flagging carriers of certain alleles, estimating and testing for Hardy-Weinberg disequilibrium, estimating and testing for linkage disequilibrium, ...

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R topics documented:

ci.balance	2
Deprecated	3
diseq	4
expectedGenotypes	7
genotype	8
gregorius	13
groupGenotype	15
homozygote	17
HWE.chisq	19
HWE.exact	21
HWE.test	22

LD	24
locus	26
makeGenotypes	30
order.genotype	32
plot.genotype	35
print.LD	36
summary.genotype	38
undocumented	40
write.pop.file	40

Index	42
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ci.balance	<i>Experimental Function to Correct Confidence Intervals At or Near Boundaries of the Parameter Space by 'Sliding' the Interval on the Quantile Scale.</i>
------------	--

Description

Experimental function to correct confidence intervals at or near boundaries of the parameter space by 'sliding' the interval on the quantile scale.

Usage

```
ci.balance(x, est, confidence=0.95, alpha=1-confidence, minval, maxval,
          na.rm=TRUE)
```

Arguments

x	Bootstrap parameter estimates.
est	Observed value of the parameter.
confidence	Confidence level for the interval. Defaults to 0.95.
alpha	Type I error rate (size) for the interval. Defaults to 1-confidence.
minval	A numeric value specifying the lower bound of the parameter space. Leave unspecified (the default) if there is no lower bound.
maxval	A numeric value specifying the upper bound of the parameter space. Leave unspecified (the default) if there is no upper bound.
na.rm	logical. Should missing values be removed?

Details

EXPERIMENTAL FUNCTION:

This function attempts to compute a proper `conf*100%` confidence interval for parameters at or near the boundary of the parameter space using bootstrapped parameter estimates by 'sliding' the confidence interval on the quantile scale.

This is accomplished by attempting to place a `conf * 100%` interval symmetrically *on the quantile scale* about the observed value. If a symmetric interval would exceed the observed data at the upper (lower) end, a one-sided interval is computed with the upper (lower) boundary fixed at the the upper (lower) boundary of the parameter space.

Value

A list containing:

`ci` A 2-element vector containing the lower and upper confidence limits. The names of the elements of the vector give the actual quantile values used for the interval or one of the character strings "Upper Boundary" or "Lower Boundary".

`overflow.upper`, `overflow.lower`
The number of elements beyond those observed that would be needed to compute a symmetric (on the quantile scale) confidence interval.

`n.above`, `n.below`
The number of bootstrap values which are above (below) the observed value.

`lower.n`, `upper.n`
The index of the value used for the endpoint of the confidence interval or the character string "Upper Boundary" ("Lower Boundary").

Author(s)

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See Also

[boot](#), [bootstrap](#), Used by [diseq.ci](#).

Examples

```
# These are nonsensical examples which simply exercise the
# computation. See the code to diseq.ci for a real example.
#
# FIXME: Add real example using boot or bootstrap.

set.seed(7981357)
x <- abs(rnorm(100,1))
ci.balance(x,1, minval=0)
ci.balance(x,1)

x <- rnorm(100,1)
x <- ifelse(x>1, 1, x)
ci.balance(x,1, maxval=1)
ci.balance(x,1)
```

 Deprecated

Deprecated functions

Description

These functions are deprecated.

Usage

```
power.casectl(...)
```

Arguments

... All arguments are ignored

Details

The `power.casectl` function contained serious errors and has been replaced by [GPC](#), [GeneticPower.Quantitative](#) or [GeneticPower.Quantitative.Numeric](#) in the BioConductor GeneticsDesign package.

In specific, the `power.casectl` function used an expected contingency table to create the test statistic that was erroneously based on the underlying null, rather than on the marginal totals of the observed table. In addition, the modeling of dominant and recessive modes of inheritance had assumed a "perfect" genotype with no disease, whereas in reality a dominant or recessive mode of inheritance simply means that two of the genotypes will have an identical odds ratio compared to the 3rd genotype (the other homozygote).

 diseq

Estimate or Compute Confidence Interval for the Single-Marker Disequilibrium

Description

Estimate or compute confidence interval for single-marker disequilibrium.

Usage

```
diseq(x, ...)
## S3 method for class 'diseq':
print(x, show=c("D", "D'", "r", "R^2", "table"), ...)
diseq.ci(x, R=1000, conf=0.95, correct=TRUE, na.rm=TRUE, ...)
```

Arguments

<code>x</code>	genotype or haplotype object.
<code>show</code>	a character value or vector indicating which disequilibrium measures should be displayed. The default is to show all of the available measures. <code>show="table"</code> will display a table of observed, expected, and observed-expected frequencies.
<code>conf</code>	Confidence level to use when computing the confidence level for D-hat. Defaults to 0.95, should be in (0,1).
<code>R</code>	Number of bootstrap iterations to use when computing the confidence interval. Defaults to 1000.
<code>correct</code>	See details.
<code>na.rm</code>	logical. Should missing values be removed?
<code>...</code>	optional parameters passed to <code>boot.ci(diseq.ci)</code> or ignored.

Details

For a single-gene marker, `diseq` computes the Hardy-Weinberg (dis)equilibrium statistic D , D' , r (the correlation coefficient), and r^2 for each pair of allele values, as well as an overall summary value for each measure across all alleles. `print.diseq` displays the contents of a `diseq` object. `diseq.ci` computes a bootstrap confidence interval for this estimate.

For consistency, I have applied the standard definitions for D , D' , and r from the Linkage Disequilibrium case, replacing all marker probabilities with the appropriate allele probabilities.

Thus, for each allele pair,

D is defined as the half of the raw difference in frequency between the observed number of heterozygotes and the expected number:

$$D = \frac{1}{2}(p_{ij} + p_{ji}) - p_i p_j$$

D' rescales D to span the range $[-1,1]$

$$D' = \frac{D}{D_{max}}$$

where, if $D > 0$:

$$D_{max} = \min p_i p_j, p_j p_i = p_i p_j$$

or if $D < 0$:

$$D_{max} = \min p_i(1 - p_j), p_j(1 - p_i)$$

r is the correlation coefficient between two alleles, and can be computed by

$$r = \frac{-D}{\sqrt{(p_i * (1 - p_i) p(j)(1 - p_j))}}$$

where

- p_i defined as the observed probability of allele 'i',

- p_j defined as the observed probability of allele 'j', and
- p_{ij} defined as the observed probability of the allele pair 'ij'.

When there are more than two alleles, the summary values for these statistics are obtained by computing a weighted average of the absolute value of each allele pair, where the weight is determined by the expected frequency. For example:

$$D_{overall} = \sum_{i \neq j} |D_{ij}| * p_{ij}$$

Bootstrapping is used to generate confidence interval in order to avoid reliance on parametric assumptions, which will not hold for alleles with low frequencies (e.g. D' following a Chi-square distribution).

See the function `HWE.test` for testing Hardy-Weinberg Equilibrium, $D = 0$.

Value

`diseq` returns an object of class `diseq` with components

<code>call</code>	function call used to create this object
<code>data</code>	2-way table of allele pair counts
<code>D.hat</code>	matrix giving the observed count, expected count, observed - expected difference, and estimate of disequilibrium for each pair of alleles as well as an overall disequilibrium value.
<code>TODO</code>	more slots to be documented

`diseq.ci` returns an object of class `bootci`

Author(s)

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See Also

`genotype`, `HWE.test`, `boot`, `bootci`

Examples

```
example.data <- c("D/D", "D/I", "D/D", "I/I", "D/D",
                 "D/D", "D/D", "D/D", "I/I", "")
g1 <- genotype(example.data)
g1

diseq(g1)
diseq.ci(g1)
HWE.test(g1) # does the same, plus tests D-hat=0

three.data <- c(rep("A/A", 8),
```

```

rep("C/A",20),
rep("C/T",20),
rep("C/C",10),
rep("T/T",3))

g3 <- genotype(three.data)
g3

diseq(g3)
diseq.ci(g3, ci.B=10000, ci.type="bca")

# only show observed vs expected table
print(diseq(g3), show='table')
```

expectedGenotypes *Construct expected genotypes/haplotypes according to known allele variants*

Description

expectedGenotypes constructs expected genotypes according to known allele variants, which can be quite tedious with large number of allele variants. It can handle different level of ploidy.

Usage

```

expectedGenotypes(x, alleles=allele.names(x), ploidy=2, sort=TRUE,
                  haplotype=FALSE)
expectedHaplotypes(x, alleles=allele.names(x), ploidy=2, sort=TRUE,
                  haplotype=TRUE)
```

Arguments

x	genotype or haplotype
alleles	character, vector of allele names
ploidy	numeric, number of chromosome sets i.e. 2 for human autosomal genes
sort	logical, sort genotypes according to order of alleles in alleles argument
haplotype	logical, construct haplotypes i.e. ordered genotype

At least one of x or alleles must be given.

Details

expectedHaplotypes() just calls expectedGenotypes() with argument haplotype=TRUE.

Value

A character vector with genotype names as "alele1/alele2" for diploid example. Length of output is $(n * (n + 1))/2$ for genotype (unordered genotype) and $n * n$ for haplotype (ordered genotype) for n allele variants.

Author(s)

Gregor Gorjanc

See Also[allele.names](#), [genotype](#)**Examples**

```
## On genotype
prp <- c("ARQ/ARQ", "ARQ/ARQ", "ARR/ARQ", "AHQ/ARQ", "ARQ/ARQ")
alleles <- c("ARR", "AHQ", "ARH", "ARQ", "VRR", "VRQ")
expectedGenotypes(as.genotype(prp))
expectedGenotypes(as.genotype(prp, alleles=alleles))
expectedGenotypes(as.genotype(prp, alleles=alleles, reorder="yes"))

## Only allele names
expectedGenotypes(alleles=alleles)
expectedGenotypes(alleles=alleles, ploidy=4)

## Haplotype
expectedHaplotypes(alleles=alleles)
expectedHaplotypes(alleles=alleles, ploidy=4)[1:20]
```

genotype

*Genotype or Haplotype Objects.***Description**`genotype` creates a genotype object.`haplotype` creates a haplotype object.`is.genotype` returns TRUE if x is of class genotype`is.haplotype` returns TRUE if x is of class haplotype`as.genotype` attempts to coerce its argument into an object of class genotype.`as.genotype.allele.count` converts allele counts (0,1,2) into genotype pairs ("A/A", "A/B", "B/B").`as.haplotype` attempts to coerce its argument into an object of class haplotype.`nallele` returns the number of alleles in an object of class genotype.**Usage**

```
genotype(a1, a2=NULL, alleles=NULL, sep="/", remove.spaces=TRUE,
         reorder = c("yes", "no", "default", "ascii", "freq"),
         allow.partial.missing=FALSE, locus=NULL,
         genotypeOrder=NULL)
```

```

haplotype(a1, a2=NULL, alleles=NULL, sep="/", remove.spaces=TRUE,
          reorder="no", allow.partial.missing=FALSE, locus=NULL,
          genotypeOrder=NULL)

is.genotype(x)

is.haplotype(x)

as.genotype(x, ...)

as.genotype.allele.count(x, alleles=c("A","B"), ... )

as.haplotype(x, ...)

## S3 method for class 'genotype':
print(x, ...)

nallele(x)

```

Arguments

<code>x</code>	either an object of class <code>genotype</code> or <code>haplotype</code> or an object to be converted to class <code>genotype</code> or <code>haplotype</code> .
<code>a1, a2</code>	vector(s) or matrix containing two alleles for each individual. See details, below.
<code>alleles</code>	names (and order if <code>reorder="yes"</code>) of possible alleles.
<code>sep</code>	character separator or column number used to divide alleles when <code>a1</code> is a vector of strings where each string holds both alleles. See below for details.
<code>remove.spaces</code>	logical indicating whether spaces and tabs will be removed from <code>a1</code> and <code>a2</code> before processing.
<code>reorder</code>	how should alleles within an individual be reordered. If <code>reorder="no"</code> , use the order specified by the <code>alleles</code> parameter. If <code>reorder="freq"</code> or <code>reorder="yes"</code> , sort alleles within each individual by observed frequency. If <code>reorder="ascii"</code> , reorder alleles in ASCII order (alphabetical, with all upper case before lower case). The default value for <code>genotype</code> is <code>"freq"</code> . The default value for <code>haplotype</code> is <code>"no"</code> .
<code>allow.partial.missing</code>	logical indicating whether one allele is permitted to be missing. When set to <code>FALSE</code> both alleles are set to <code>NA</code> when either is missing.
<code>locus</code>	object of class <code>locus</code> , <code>gene</code> , or <code>marker</code> , holding information about the source of this genotype.
<code>genotypeOrder</code>	character, vector of genotype/haplotype names so that further functions can sort genotypes/haplotypes in wanted order
<code>...</code>	optional arguments

Details

Genotype objects hold information on which gene or marker alleles were observed for different individuals. For each individual, two alleles are recorded.

The genotype class considers the stored alleles to be unordered, i.e., "C/T" is equivalent to "T/C". The haplotype class considers the order of the alleles to be significant so that "C/T" is distinct from "T/C".

When calling `genotype` or `haplotype`:

- If only `a1` is provided and is a character vector, it is assumed that each element encodes both alleles. In this case, if `sep` is a character string, `a1` is assumed to be coded as "Allele1<sep>Allele2". If `sep` is a numeric value, it is assumed that character locations `1 : sep` contain allele 1 and that remaining locations contain allele 2.
- If `a1` is a matrix, it is assumed that column 1 contains allele 1 and column 2 contains allele 2.
- If `a1` and `a2` are both provided, each is assumed to contain one allele value so that the genotype for an individual is obtained by `paste(a1, a2, sep="/")`.

If `remove.spaces` is `TRUE`, (the default) any whitespace contained in `a1` and `a2` is removed when the genotypes are created. If whitespace is used as the separator, (eg "C C", "C T", ...), be sure to set `remove.spaces` to `FALSE`.

When the alleles are explicitly specified using the `alleles` argument, all potential alleles not present in the list will be converted to `NA`.

NOTE: `genotype` assumes that the order of the alleles is not important (E.G., "A/C" == "C/A"). Use class `haplotype` if order is significant.

If `genotypeOrder=NULL` (the default setting), then `expectedGenotypes` is used to get standard sorting order. Only unique values in `genotypeOrder` are used, which in turns means that the first occurrence prevails. When `genotypeOrder` is given some genotype names, but not all that appear in the data, the rest (those in the data and possible combinations based on allele variants) is automatically added at the end of `genotypeOrder`. This puts "missing" genotype names at the end of sort order. This feature is especially useful when there are a lot of allele variants and especially in haplotypes. See examples.

Value

The `genotype` class extends "factor" and `haplotype` extends `genotype`. Both classes have the following attributes:

<code>levels</code>	character vector of possible genotype/haplotype values stored coded by <code>paste(allele1, "/", allele2, sep="")</code> .
<code>allele.names</code>	character vector of possible alleles. For a SNP, these might be <code>c("A","T")</code> . For a variable length dinucleotide repeat this might be <code>c("136","138","140","148")</code> .
<code>allele.map</code>	matrix encoding how the factor levels correspond to alleles. See the source code to <code>allele.genotype()</code> for how to extract allele values using this matrix. Better yet, just use <code>allele.genotype()</code> .
<code>genotypeOrder</code>	character, genotype/haplotype names in defined order that can used for sorting in various functions. Note that this slot stores both ordered and unordered genotypes i.e. "A/B" and "B/A".

Author(s)

Gregory R. Warnes (warnes@bst.rochester.edu) and Friedrich Leisch.

See Also

[HWE.test](#), [allele](#), [homozygote](#), [heterozygote](#), [carrier](#), [summary.genotype](#), [allele.count](#), [sort.genotype](#), [genotypeOrder](#), [locus](#), [gene](#), [marker](#), and [%in%](#) for default [%in%](#) method

Examples

```
# several examples of genotype data in different formats
example.data <- c("D/D","D/I","D/D","I/I","D/D",
                 "D/D","D/D","D/D","I/I","")
g1 <- genotype(example.data)
g1

example.data2 <- c("C-C","C-T","C-C","T-T","C-C",
                  "C-C","C-C","C-C","T-T","")
g2 <- genotype(example.data2,sep="-")
g2

example.nosep <- c("DD","DI","DD","II","DD",
                  "DD","DD","DD","II","")
g3 <- genotype(example.nosep,sep="")
g3

example.a1 <- c("D","D","D","I","D","D","D","D","I","")
example.a2 <- c("D","I","D","I","D","D","D","D","I","")
g4 <- genotype(example.a1,example.a2)
g4

example.mat <- cbind(a1=example.a1, a1=example.a2)
g5 <- genotype(example.mat)
g5

example.data5 <- c("D / D","D / I","D / D","I / I",
                  "D / D","D / D","D / D","D / D",
                  "I / I","")
g5 <- genotype(example.data5,rem=TRUE)
g5

# show how genotype and haplotype differ
data1 <- c("C/C","C/T","T/C")
data2 <- c("C/C","T/C","T/C")

test1 <- genotype( data1 )
test2 <- genotype( data2 )

test3 <- haplotype( data1 )
test4 <- haplotype( data2 )
```

```

test1==test2
test3==test4

test1=="C/T"
test1=="T/C"

test3=="C/T"
test3=="T/C"

## also
test1
test1
test3

test1
test1

test3
test3

## "Messy" example

m3 <- c("D D/\t D D","D\tD/ I", "D D/ D D","I/ I",
        "D D/ D D","D D/ D D","D D/ D D","D D/ D D",
        "I/ I","/ ","/I")

genotype(m3)
summary(genotype(m3))

m4 <- c("D D","D I","D D","I I",
        "D D","D D","D D","D D",
        "I I"," ", " I")

genotype(m4, sep=1)
genotype(m4, sep=" ", remove.spaces=FALSE)
summary(genotype(m4, sep=" ", remove.spaces=FALSE))

m5 <- c("DD","DI","DD","II",
        "DD","DD","DD","DD",
        "II"," ", " I")

genotype(m5, sep=1)
haplotype(m5, sep=1, remove.spaces=FALSE)

g5 <- genotype(m5, sep="")
h5 <- haplotype(m5, sep="")

heterozygote(g5)
homozygote(g5)
carrier(g5, "D")

g5[9:10] <- haplotype(m4, sep=" ", remove=FALSE)[1:2]
g5

```

```

g5[9:10]
allele(g5[9:10],1)
allele(g5,1)[9:10]

# drop unused alleles
g5[9:10,drop=TRUE]
h5[9:10,drop=TRUE]

# Convert allele.counts into genotype

x <- c(0,1,2,1,1,2,NA,1,2,1,2,2,2)
g <- as.genotype.allele.count(x, alleles=c("C","T") )
g

# Use of genotypeOrder
example.data <- c("D/D","D/I","I/D","I/I","D/D",
                 "D/D","D/I","I/D","I/I","")
summary(genotype(example.data))
genotypeOrder(genotype(example.data))

summary(genotype(example.data, genotypeOrder=c("D/D", "I/I", "D/I")))
summary(genotype(example.data, genotypeOrder=c(
                                                "D/I")))
summary(haplotype(example.data, genotypeOrder=c(
                                                "I/D", "D/I")))
example.data <- genotype(example.data)
genotypeOrder(example.data) <- c("D/D", "I/I", "D/I")
genotypeOrder(example.data)

```

gregorius

Probability of Observing All Alleles with a Given Frequency in a Sample of a Specified Size.

Description

Probability of observing all alleles with a given frequency in a sample of a specified size.

Usage

```
gregorius(freq, N, missprob, tol = 1e-10, maxN = 10000, maxiter=100, showiter = FALSE)
```

Arguments

freq	(Minimum) Allele frequency (required)
N	Number of sampled genotypes
missprob	Desired maximum probability of failing to observe an allele.
tol	Omit computation for terms which contribute less than this value.
maxN	Largest value to consider when searching for N.
maxiter	Maximum number of iterations to use when searching for N.

`showiter` Boolean flag indicating whether to show the iterations performed when searching for `N`.

Details

If `freq` and `N` are provided, but `missprob` is omitted, this function computes the probability of failing to observe all alleles with true underlying frequency `freq` when `N` diploid genotypes are sampled. This is accomplished using the sum provided in Corollary 2 of Gregorius (1980), omitting terms which contribute less than `tol` to the result.

When `freq` and `missprob` are provide, but `N` is omitted. A binary search on the range of $[1, \text{maxN}]$ is performed to locate the smallest sample size, `N`, for which the probability of failing to observe all alleles with true underlying frequency `freq` is at most `missprob`. In this case, `maxiter` specifies the largest number of iterations to use in the binary search, and `showiter` controls whether the iterations of the search are displayed.

Value

A list containing the following values:

<code>call</code>	Function call used to generate this object.
<code>method</code>	One of the strings, "Compute <code>missprob</code> given <code>N</code> and <code>freq</code> ", or "Determine minimal <code>N</code> given <code>missprob</code> and <code>freq</code> ", indicating which type of computation was performed.
<code>retval\$freq</code>	Specified allele frequency.
<code>retval\$N</code>	Specified or computed sample size.
<code>retval\$missprob</code>	Computed probability of failing to observe all of the alleles with frequency <code>freq</code> .

Note

This code produces sample sizes that are slightly larger than those given in table 1 of Gregorius (1980). This appears to be due to rounding of the computed `missprobs` by the authors of that paper.

Author(s)

Code submitted by David Duffy <davidD@qumr.edu.au>, substantially enhanced by Gregory R. Warnes <warnes@bst.rochester.edu>.

References

Gregorius, H.R. 1980. The probability of losing an allele when diploid genotypes are sampled. *Biometrics* 36, 643-652.

Examples

```
# Compute the probability of missing an allele with frequency 0.15 when
# 20 genotypes are sampled:
gregorius(freq=0.15, N=20)

# Determine what sample size is required to observe all alleles with true
# frequency 0.15 with probability 0.95
gregorius(freq=0.15, missprob=1-0.95)
```

groupGenotype	<i>Group genotype values</i>
---------------	------------------------------

Description

groupGenotype groups genotype or haplotype values according to given "grouping/mapping" information

Usage

```
groupGenotype(x, map, haplotype=FALSE, factor=TRUE, levels=NULL, verbose=FALSE)
```

Arguments

x	genotype or haplotype
map	list, mapping information, see details and examples
haplotype	logical, should values in a map be treated as haplotypes or genotypes, see details
factor	logical, should output be a factor or a character
levels	character, optional vector of level names if factor is produced (factor=TRUE); the default is to use the sort order of the group names in map
verbose	logical, print genotype names that match entries in the map - mainly used for debugging

Details

Examples show how map can be constructed. This are the main points to be aware of:

- names of list components are used as new group names
- list components hold genotype names per each group
- genotype names can be specified directly i.e. "A/B" or abbreviated such as "A/*" or even "*/*", where "*" matches any possible allele, but read also further on
- all genotype names that are not specified can be captured with ".else" (note the dot!)

- genotype names that were not specified (and ".else" was not used) are changed to NA

map is inspected before grouping of genotypes is being done. The following steps are done during inspection:

- ".else" must be at the end (if not, it is moved) to match everything that has not yet been defined
- any specifications like "A/*", "*/A", or "*/*" are extended to all possible genotypes based on alleles in argument alleles - in case of haplotype=FALSE, "A/*" and "*/A" match the same genotypes
- since use of "*" and ".else" can cause duplicates along the whole map, duplicates are removed sequentially (first occurrence is kept)

Using ".else" or "*/*" at the end of the map produces the same result, due to removing duplicates sequentially.

Value

A factor or character vector with genotypes grouped

Author(s)

Gregor Gorjanc

See Also

[genotype](#), [haplotype](#), [factor](#), and [levels](#)

Examples

```
## --- Setup ---

x <- c("A/A", "A/B", "B/A", "A/C", "C/A", "A/D", "D/A",
      "B/B", "B/C", "C/B", "B/D", "D/B",
      "C/C", "C/D", "D/C",
      "D/D")
g <- genotype(x, reorder="yes")
## "A/A" "A/B" "A/B" "A/C" "A/C" "A/D" "A/D" "B/B" "B/C" "B/C" "B/D" "B/D"
## "C/C" "C/D" "C/D" "D/D"

h <- haplotype(x)
## "A/A" "A/B" "B/A" "A/C" "C/A" "A/D" "D/A" "B/B" "B/C" "C/B" "B/D" "D/B"
## "C/C" "C/D" "D/C" "D/D"

## --- Use of "A/A", "A/*" and ".else" ---

map <- list("homoG"=c("A/A", "B/B", "C/C", "D/D"),
           "heteroA*" =c("A/B", "A/C", "A/D"),
           "heteroB*" =c("B/*"),
           "heteroRest"=".else")

(tmpG <- groupGenotype(x=g, map=map, factor=FALSE))
```

```

(tmpH <- groupGenotype(x=h, map=map, factor=FALSE, haplotype=TRUE))

## Show difference between genotype and haplotype treatment
cbind(as.character(h), gen=tmpG, hap=tmpH, diff!=(tmpG == tmpH))
##           gen           hap           diff
## [1,] "A/A" "homoG"      "homoG"      "FALSE"
## [2,] "A/B" "heteroA*"   "heteroA*"   "FALSE"
## [3,] "B/A" "heteroA*"   "heteroB*"   "TRUE"
## [4,] "A/C" "heteroA*"   "heteroA*"   "FALSE"
## [5,] "C/A" "heteroA*"   "heteroRest" "TRUE"
## [6,] "A/D" "heteroA*"   "heteroA*"   "FALSE"
## [7,] "D/A" "heteroA*"   "heteroRest" "TRUE"
## [8,] "B/B" "homoG"      "homoG"      "FALSE"
## [9,] "B/C" "heteroB*"   "heteroB*"   "FALSE"
## [10,] "C/B" "heteroB*"   "heteroRest" "TRUE"
## [11,] "B/D" "heteroB*"   "heteroB*"   "FALSE"
## [12,] "D/B" "heteroB*"   "heteroRest" "TRUE"
## [13,] "C/C" "homoG"      "homoG"      "FALSE"
## [14,] "C/D" "heteroRest" "heteroRest" "FALSE"
## [15,] "D/C" "heteroRest" "heteroRest" "FALSE"
## [16,] "D/D" "homoG"      "homoG"      "FALSE"

map <- list("withA"="A/*", "rest"=".else")
groupGenotype(x=g, map=map, factor=FALSE)
## [1] "withA" "withA" "withA" "withA" "withA" "withA" "withA" "rest" "rest"
## [10] "rest" "rest" "rest" "rest" "rest" "rest" "rest" "rest"

groupGenotype(x=h, map=map, factor=FALSE, haplotype=TRUE)
## [1] "withA" "withA" "rest" "withA" "rest" "withA" "rest" "rest" "rest"
## [10] "rest" "rest" "rest" "rest" "rest" "rest" "rest" "rest"

## --- Use of "*/*" ---

map <- list("withA"="A/*", withB="*/*")
groupGenotype(x=g, map=map, factor=FALSE)
## [1] "withA" "withA" "withA" "withA" "withA" "withA" "withA" "withB" "withB"
## [10] "withB" "withB" "withB" "withB" "withB" "withB" "withB" "withB"

## --- Missing genotype specifications produces NA's ---

map <- list("withA"="A/*", withB="B/*")
groupGenotype(x=g, map=map, factor=FALSE)
## [1] "withA" "withA" "withA" "withA" "withA" "withA" "withA" "withB" "withB"
## [10] "withB" "withB" "withB" NA      NA      NA      NA

groupGenotype(x=h, map=map, factor=FALSE, haplotype=TRUE)
## [1] "withA" "withA" "withB" "withA" NA      "withA" NA      "withB" "withB"
## [10] NA      "withB" NA      NA      NA      NA      NA

```

Description

`homozygote` creates an vector of logicals that are true when the alleles of the corresponding observation are the identical.

`heterozygote` creates an vector of logicals that are true when the alleles of the corresponding observation differ.

`carrier` create a logical vector or matrix of logicals indicating whether the specified alleles are present.

`allele.count` returns the number of copies of the specified alleles carried by each observation.

`allele` extract the specified allele(s) as a character vector or a 2 column matrix.

`allele.names` extract the set of allele names.

Usage

```
homozygote(x, allele.name, ...)
heterozygote(x, allele.name, ...)
carrier(x, allele.name, ...)
## S3 method for class 'genotype':
carrier(x, allele.name=allele.names(x),
        any=!missing(allele.name), na.rm=FALSE, ...)
allele.count(x, allele.name=allele.names(x), any=!missing(allele.name),
            na.rm=FALSE)
allele(x, which=c(1,2) )
allele.names(x)
```

Arguments

<code>x</code>	genotype object
<code>...</code>	optional parameters (ignored)
<code>allele.name</code>	character value or vector of allele names
<code>any</code>	logical value. When <code>TRUE</code> , a single count or indicator is returned by combining the results for all of the elements of <code>allele</code> . If <code>FALSE</code> separate counts or indicators should be returned for each element of <code>allele</code> . Defaults to <code>FALSE</code> if <code>allele</code> is missing. Otherwise defaults to <code>TRUE</code> .
<code>na.rm</code>	logical value indicating whether to remove missing values. When true, any NA values will be replaced by 0 or <code>FALSE</code> as appropriate. Defaults to <code>FALSE</code> .
<code>which</code>	selects which allele to return. For first allele use 1. For second allele use 2. For both (the default) use <code>c(1,2)</code> .

Details

When the `allele.name` argument is given, `heterozygote` and `homozygote` return `TRUE` if *exactly* one or both alleles, respectively, match the specified `allele.name`.

Value

`homozygote` and `heterozygote` return a vector of logicals.

`carrier` returns a logical vector if only one allele is specified, or if any is TRUE. Otherwise, it returns matrix of logicals with one row for each element of `allele`.

`allele.count` returns a vector of counts if only one allele is specified, or if any is TRUE. Otherwise, it returns matrix of counts with one row for each element of `allele`.

`allele` returns a character vector when one allele is specified. When 2 alleles are specified, it returns a 2 column character matrix.

`allele.names` returns a character vector containing the set of allele names.

Author(s)

Gregory R. Warnes <warnes@bst.rochester.edu>

See Also

[genotype](#), [HWE.test](#), [summary.genotype](#), [locus gene marker](#)

Examples

```
example.data <- c("D/D", "D/I", "D/D", "I/I", "D/D", "D/D", "D/D", "D/D", "I/I", "")
g1 <- genotype(example.data)
g1

heterozygote(g1)
homozygote(g1)

carrier(g1, "D")
carrier(g1, "D", na.rm=TRUE)

# get count of one allele
allele.count(g1, "D")

# get count of each allele
allele.count(g1) # equivalent to
allele.count(g1, c("D", "I"), any=FALSE)

# get combined count for both alleles
allele.count(g1, c("I", "D"))

# get second allele
allele(g1, 2)

# get both alleles
allele(g1)
```

`HWE.chisq`*Perform Chi-Square Test for Hardy-Weinberg Equilibrium*

Description

Test the null hypothesis that Hardy-Weinberg equilibrium holds using the Chi-Square method.

Usage

```
HWE.chisq(x, ...)  
## S3 method for class 'genotype':  
HWE.chisq(x, simulate.p.value=TRUE, B=10000, ...)
```

Arguments

<code>x</code>	genotype or haplotype object.
<code>simulate.p.value</code>	a logical value indicating whether the p-value should be computed using simulation instead of using the χ^2 approximation. Defaults to TRUE.
<code>B</code>	Number of simulation iterations to use when <code>simulate.p.value=TRUE</code> . Defaults to 10000.
<code>...</code>	optional parameters passed to <code>chisq.test</code>

Details

This function generates a 2-way table of allele counts, then calls `chisq.test` to compute a p-value for Hardy-Weinberg Equilibrium. By default, it uses an unadjusted Chi-Square test statistic and computes the p-value using a simulation/permutation method. When `simulate.p.value=FALSE`, it computes the test statistic using the Yates continuity correction and tests it against the asymptotic Chi-Square distribution with the appropriate degrees of freedom.

Note: The Yates continuity correction is applied *only* when `simulate.p.value=FALSE`, so that the reported test statistics when `simulate.p.value=FALSE` and `simulate.p.value=TRUE` will differ.

Value

An object of class `htest`.

See Also

[HWE.exact](#), [HWE.test](#), [diseq](#), [diseq.ci](#), [allele](#), [chisq.test](#), [boot](#), [bootci](#)

Examples

```
example.data <- c("D/D", "D/I", "D/D", "I/I", "D/D",
                 "D/D", "D/D", "D/D", "I/I", "")
g1 <- genotype(example.data)
g1

HWE.chisq(g1)
# compare with
HWE.exact(g1)
# and
HWE.test(g1)

three.data <- c(rep("A/A", 8),
                rep("C/A", 20),
                rep("C/T", 20),
                rep("C/C", 10),
                rep("T/T", 3))

g3 <- genotype(three.data)
g3

HWE.chisq(g3, B=10000)
```

`HWE.exact`*Exact Test of Hardy-Weinberg Equilibrium for 2-Allele Markers*

Description

Exact test of Hardy-Weinberg Equilibrium for 2 Allele Markers.

Usage

```
HWE.exact(x)
```

Arguments

x Genotype object

Value

Object of class 'htest'.

Note

This function only works for genotypes with exactly 2 alleles.

Author(s)

David Duffy <davidD@qimr.edu.au> with modifications by Gregory R. Warnes <warnes@bst.rochester.edu>

References

Emigh TH. (1980) "Comparison of tests for Hardy-Weinberg Equilibrium", *Biometrics*, 36, 627-642.

See Also

[HWE.chisq](#), [HWE.test](#), [diseq](#), [diseq.ci](#)

Examples

```
example.data <- c("D/D", "D/I", "D/D", "I/I", "D/D",
                 "D/D", "D/D", "D/D", "I/I", "")
g1 <- genotype(example.data)
g1

HWE.exact(g1)
# compare with
HWE.chisq(g1)

g2 <- genotype(sample( c("A", "C"), 100, p=c(100,10), rep=TRUE),
               sample( c("A", "C"), 100, p=c(100,10), rep=TRUE) )
HWE.exact(g2)
```

HWE.test

Estimate Disequilibrium and Test for Hardy-Weinberg Equilibrium

Description

Estimate disequilibrium parameter and test the null hypothesis that Hardy-Weinberg equilibrium holds.

Usage

```
HWE.test(x, ...)
## S3 method for class 'genotype':
HWE.test(x, exact = nallele(x)==2, simulate.p.value=!exact,
         B=10000, conf=0.95, ci.B=1000, ... )
## S3 method for class 'data.frame':
HWE.test(x, ..., do.Allele.Freq=TRUE, do.HWE.test=TRUE)
## S3 method for class 'HWE.test':
print(x, show=c("D", "D'", "r", "table"), ...)
```

Arguments

<code>x</code>	genotype or haplotype object.
<code>exact</code>	a logical value indicated whether the p-value should be computed using the exact method, which is only available for 2 allele genotypes.
<code>simulate.p.value</code>	a logical value indicating whether the p-value should be computed using simulation instead of using the χ^2 approximation. Defaults to TRUE.
<code>B</code>	Number of simulation iterations to use when <code>simulate.p.value=TRUE</code> . Defaults to 10000.
<code>conf</code>	Confidence level to use when computing the confidence level for D-hat. Defaults to 0.95, should be in (0,1).
<code>ci.B</code>	Number of bootstrap iterations to use when computing the confidence interval. Defaults to 1000.
<code>show</code>	a character vector containing the names of HWE test statistics to display from the set of "D", "D'", "r", and "table".
<code>...</code>	optional parameters passed to <code>HWE.test</code> (data.frame method) or <code>chisq.test</code> (base method).
<code>do.Allele.Freq</code>	logical indication whether to summarize allele frequencies.
<code>do.HWE.test</code>	logical indication whether to perform HWE tests

Details

HWE.test calls `diseq` to compute the Hardy-Weinberg (dis)equilibrium statistics D, D', and r (correlation coefficient). Next it calls `diseq.ci` to compute a bootstrap confidence interval for these estimates. Finally, it calls `chisq.test` to compute a p-value for Hardy-Weinberg Equilibrium using a simulation/permutation method.

Using bootstrapping for the confidence interval and simulation for the p-value avoids reliance on the assumptions the underlying Chi-square approximation. This is particularly important when some allele pairs have small counts.

For details on the definition of D, D', and r, see the help page for `diseq`.

Value

An object of class `HWE.test` with components

<code>diseq</code>	A <code>diseq</code> object providing details on the disequilibrium estimates.
<code>ci</code>	A <code>diseq.ci</code> object providing details on the bootstrap confidence intervals for the disequilibrium estimates.
<code>test</code>	A <code>htest</code> object providing details on the permutation based Chi-square test.
<code>call</code>	function call used to create this object.
<code>conf, B, ci.B, simulate.p.value</code>	values used for these arguments.

Author(s)

Gregory R. Warnes (warnes@bst.rochester.edu)

See Also

[genotype](#), [diseq](#), [diseq.ci](#), [HWE.chisq](#), [HWE.exact](#),

Examples

```
example.data <- c("D/D", "D/I", "D/D", "I/I", "D/D",
                 "D/D", "D/D", "D/D", "I/I", "")
g1 <- genotype(example.data)
g1

HWE.test(g1)

#compare with
diseq(g1)
diseq.ci(g1)
HWE.chisq(g1)
HWE.exact(g1)

three.data <- c(rep("A/A", 8),
                rep("C/A", 20),
                rep("C/T", 20),
                rep("C/C", 10),
                rep("T/T", 3))

g3 <- genotype(three.data)
g3

HWE.test(g3, ci.B=10000)
```

LD

Pairwise linkage disequilibrium between genetic markers.

Description

Compute pairwise linkage disequilibrium between genetic markers

Usage

```
LD(g1, ...)
## S3 method for class 'genotype':
LD(g1, g2, ...)
## S3 method for class 'data.frame':
LD(g1, ...)
```

Arguments

g1	genotype object or dataframe containing genotype objects
g2	genotype object (ignored if g1 is a dataframe)
...	optional arguments (ignored)

Details

Linkage disequilibrium (LD) is the non-random association of marker alleles and can arise from marker proximity or from selection bias.

`LD.genotype` estimates the extent of LD for a single pair of genotypes. `LD.data.frame` computes LD for all pairs of genotypes contained in a data frame. Before starting, `LD.data.frame` checks the class and number of alleles of each variable in the dataframe. If the data frame contains non-genotype objects or genotypes with more or less than 2 alleles, these will be omitted from the computation and a warning will be generated.

Three estimators of LD are computed:

D raw difference in frequency between the observed number of AB pairs and the expected number:

$$D = p_{AB} - p_A p_B$$

D' scaled D spanning the range [-1,1]

$$D' = \frac{D}{D_{max}}$$

where, if $D > 0$:

$$D_{max} = \min(p_A p_b, p_a p_B)$$

or if $D < 0$:

$$D_{max} = \max(-p_A p_b, -p_a p_B)$$

r correlation coefficient between the markers

$$r = \frac{-D}{\sqrt{(p_A * p_a * p_B * p_b)}}$$

where

- p_A is defined as the observed probability of allele 'A' for marker 1,
- $p_a = 1 - p_A$ is defined as the observed probability of allele 'a' for marker 1,
- p_B is defined as the observed probability of allele 'B' for marker 2, and
- $p_b = 1 - p_B$ is defined as the observed probability of allele 'b' for marker 2, and
- p_{AB} is defined as the probability of the marker allele pair 'AB'.

For genotype data, AB/ab cannot be distinguished from aB/Ab. Consequently, we estimate p_{AB} using maximum likelihood and use this value in the computations.

Value

`LD.genotype` returns a 5 element list:

<code>call</code>	the matched call
<code>D</code>	Linkage disequilibrium estimate
<code>Dprime</code>	Scaled linkage disequilibrium estimate
<code>corr</code>	Correlation coefficient
<code>nobs</code>	Number of observations
<code>chisq</code>	Chi-square statistic for linkage equilibrium (i.e., $D=D'=corr=0$)
<code>p.value</code>	Chi-square p-value for marker independence

`LD.data.frame` returns a list with the same elements, but each element is a matrix where the upper off-diagonal elements contain the estimate for the corresponding pair of markers. The other matrix elements are NA.

Author(s)

Gregory R. Warnes (warnes@bst.rochester.edu)

See Also

[genotype](#), [HWE.test](#)

Examples

```
g1 <- genotype( c('T/A',    NA, 'T/T',    NA, 'T/A',    NA, 'T/T', 'T/A',
                  'T/T', 'T/T', 'T/A', 'A/A', 'T/T', 'T/A', 'T/A', 'T/T',
                  NA, 'T/A', 'T/A',    NA) )

g2 <- genotype( c('C/A', 'C/A', 'C/C', 'C/A', 'C/C', 'C/A', 'C/A', 'C/A',
                  'C/A', 'C/C', 'C/A', 'A/A', 'C/A', 'A/A', 'C/A', 'C/C',
                  'C/A', 'C/A', 'C/A', 'A/A') )

g3 <- genotype( c('T/A', 'T/A', 'T/T', 'T/A', 'T/T', 'T/A', 'T/A', 'T/A',
                  'T/A', 'T/T', 'T/A', 'T/T', 'T/A', 'T/A', 'T/A', 'T/T',
                  'T/A', 'T/A', 'T/A', 'T/T') )

# Compute LD on a single pair

LD(g1,g2)

# Compute LD table for all 3 genotypes

data <- makeGenotypes(data.frame(g1,g2,g3))
LD(data)
```

Description

`locus`, `gene`, and `marker` create objects to store information, respectively, about genetic loci, genes, and markers.

`is.locus`, `is.gene`, and `ismarker` test whether an object is a member of the respective class.

`as.character.locus`, `as.character.gene`, `as.character.marker` return a character string containing a compact encoding the object.

`getlocus`, `getgene`, `getmarker` extract locus data (if present) from another object.

`locus<-`, `marker<-`, and `gene<-` adds locus data to an object.

Usage

```
locus(name, chromosome, arm=c("p", "q", "long", "short", NA),
      index.start, index.end=NULL)
```

```
gene(name, chromosome, arm=c("p", "q", "long", "short"),
     index.start, index.end=NULL)
```

```
marker(name, type, locus.name, bp.start, bp.end = NULL,
       relative.to = NULL, ...)
```

```
is.locus(x)
```

```
is.gene(x)
```

```
is.marker(x)
```

```
## S3 method for class 'locus':
as.character(x, ...)
```

```
## S3 method for class 'gene':
as.character(x, ...)
```

```
## S3 method for class 'marker':
as.character(x, ...)
```

```
getlocus(x, ...)
```

```
locus(x) <- value
```

```
marker(x) <- value
```

```
gene(x) <- value
```

Arguments

<code>name</code>	character string giving locus, gene, or marker name
<code>chromosome</code>	integer specifying chromosome number (1:23 for humans).
<code>arm</code>	character indicating long or short arm of the chromosome. Long is specified by "long" or "p". Short is specified by "short" or "q".
<code>index.start</code>	integer specifying location of start of locus or gene on the chromosome.
<code>index.end</code>	optional integer specifying location of end of locus or gene on the chromosome.
<code>type</code>	character string indicating marker type, e.g. "SNP"
<code>locus.name</code>	either a character string giving the name of the locus or gene (other details may be specified using <code>...</code>) or a <code>locus</code> or <code>gene</code> object.
<code>bp.start</code>	start location of marker, in base pairs
<code>bp.end</code>	end location of marker, in base pairs (optional)
<code>relative.to</code>	location (optional) from which <code>bp.start</code> and <code>bp.end</code> are calculated.
<code>...</code>	parameters for <code>locus</code> used to fill in additional details on the locus or gene within which the marker is located.
<code>x</code>	an object of class <code>locus</code> , <code>gene</code> , or <code>marker</code> , or (for <code>getlocus</code> , <code>locus<-</code> , <code>marker<-</code> , and <code>gene<-</code>) an object that may contain a locus attribute or field, notably a <code>genotype</code> object.
<code>value</code>	<code>locus</code> , <code>marker</code> , or <code>gene</code> object

Value

Object of class `locus` and `gene` are lists with the elements:

<code>name</code>	character string giving locus, gene, or marker name
<code>chromosome</code>	integer specifying chromosome number (1:23 for humans).
<code>arm</code>	character indicating long or short arm of the chromosome. Long is specified by "long" or "p". Short is specified by "short" or "q".
<code>index.start</code>	integer specifying location of start of locus or gene on the chromosome.
<code>index.end</code>	optional integer specifying location of end of locus or gene on the chromosome.
<code>marker.name</code>	character string giving the name of the marker
<code>bp.start</code>	start location of marker, in base pairs
<code>bp.end</code>	end location of marker, in base pairs (optional)
<code>relative.to</code>	location (optional) from which <code>bp.start</code> and <code>bp.end</code> are calculated.

Author(s)

Gregory R. Warnes (warnes@bst.rochester.edu)

See Also

[genotype](#),

Examples

```
ar2 <- gene("AR2", chromosome=7, arm="q", index.start=35)
ar2

par <- locus(name="AR2 Pseudogene",
             chromosome=1,
             arm="q",
             index.start=32,
             index.end=42)
par

c109t <- marker(name="C-109T",
                type="SNP",
                locus.name="AR2",
                chromosome=7,
                arm="q",
                index.start=35,
                bp.start=-109,
                relative.to="start of coding region")
c109t

c109t <- marker(name="C-109T",
                type="SNP",
                locus=ar2,
                bp.start=-109,
                relative.to="start of coding region")
c109t

example.data <- c("D/D", "D/I", "D/D", "I/I", "D/D",
                  "D/D", "D/D", "D/D", "I/I", "")
g1 <- genotype(example.data, locus=ar2)
g1

getlocus(g1)

summary(g1)
HWE.test(g1)

g2 <- genotype(example.data, locus=c109t)
summary(g2)

getlocus(g2)

heterozygote(g2)
homozygote(g1)
```

```

allele(g1, 1)

carrier(g1, "I")

heterozygote(g2)

```

makeGenotypes	<i>Convert columns in a dataframe to genotypes or haplotypes</i>
---------------	--

Description

Convert columns in a dataframe to genotypes or haplotypes.

Usage

```

makeGenotypes(data, convert, sep = "/", tol = 0.5, ..., method=as.genotype)
makeHaplotypes(data, convert, sep = "/", tol = 0.9, ...)

```

Arguments

data	Dataframe containing columns to be converted
convert	Vector or list of pairs specifying which columns contain genotype/haplotype data. See below for details.
sep	Genotype separator
tol	See below.
...	Optional arguments to as.genotype function
method	Function used to perform the conversion.

Details

The functions `makeGenotypes` and `makeHaplotypes` allow the conversion of all of the genetic variables in a dataset to genotypes or haplotypes in a single step.

The parameter `convert` may be missing, a vector of column names, indexes or true/false indicators, or a list of column name or index pairs.

When the argument `convert` is not provided, the function will look for columns where at least `tol*100%` of the records contain the separator character `sep` (‘/’ by default). These columns will then be assumed to contain both of the genotype/haplotype alleles and will be converted in-place to genotype variables.

When the argument `convert` is a vector of column names, indexes or true/false indicators, the corresponding columns will be assumed to contain both of the genotype/haplotype alleles and will be converted in-place to genotype variables.

When the argument `convert` is a list containing column name or index pairs, the two elements of each pair will be assumed to contain the individual alleles of a genotype/haplotype. The first column specified in each pair will be replaced with the new genotype/haplotype variable named `name1 + sep + name2`. The second column will be removed.

Note that the `method` argument may be used to supply a non-standard conversion function, such as `as.genotype.allele.count`, which converts from `[0,1,2]` to `['A/A','A/B','A/C']` (or the specified allele names). See the example below.

Value

Dataframe containing converted genotype/haplotype variables. All other variables will be unchanged.

Author(s)

Gregory R. Warnes `<warnes@bst.rochester.edu >`

See Also

[genotype](#)

Examples

```
## Not run:
# common case
data <- read.csv(file="genotype_data.csv")
data <- makeGenotypes(data)
## End(Not run)

# Create a test data set where there are several genotypes in columns
# of the form "A/T".
test1 <- data.frame(Tmt=sample(c("Control","Trt1","Trt2"),20, replace=TRUE),
  G1=sample(c("A/T","T/T","T/A",NA),20, replace=TRUE),
  N1=rnorm(20),
  I1=sample(1:100,20,replace=TRUE),
  G2=paste(sample(c("134","138","140","142","146"),20,
    replace=TRUE),
    sample(c("134","138","140","142","146"),20,
    replace=TRUE),
    sep=" / "),
  G3=sample(c("A /T","T /T","T /A"),20, replace=TRUE),
  comment=sample(c("Possible Bad Data/Lab Error",""),20,
    rep=TRUE)
)

test1

# now automatically convert genotype columns
geno1 <- makeGenotypes(test1)
geno1

# Create a test data set where there are several haplotypes with alleles
# in adjacent columns.
test2 <- data.frame(Tmt=sample(c("Control","Trt1","Trt2"),20, replace=TRUE),
  G1.1=sample(c("A","T",NA),20, replace=TRUE),
  G1.2=sample(c("A","T",NA),20, replace=TRUE),
  N1=rnorm(20),
```

```

I1=sample(1:100,20,replace=TRUE),
G2.1=sample(c("134","138","140","142","146"),20,
            replace=TRUE),
G2.2=sample(c("134","138","140","142","146"),20,
            replace=TRUE),
G3.1=sample(c("A ","T ","T "),20, replace=TRUE),
G3.2=sample(c("A ","T ","T "),20, replace=TRUE),
comment=sample(c("Possible Bad Data/Lab Error",""),20,
              rep=TRUE)
)
test2

# specify the locations of the columns to be paired for haplotypes
makeHaplotypes(test2, convert=list(c("G1.1","G1.2"),6:7,8:9))

# Create a test data set where the data is coded as numeric allele
# counts (0-2).
test3 <- data.frame(Tmt=sample(c("Control","Trt1","Trt2"),20, replace=TRUE),
                   G1=sample(c(0:2,NA),20, replace=TRUE),
                   N1=rnorm(20),
                   I1=sample(1:100,20,replace=TRUE),
                   G2=sample(0:2,20, replace=TRUE),
                   comment=sample(c("Possible Bad Data/Lab Error",""),20,
                                 rep=TRUE)
)
test3

# specify the locations of the columns, and a non-standard conversion
makeGenotypes(test3, convert=c('G1','G2'), method=as.genotype.allele.count)

```

order.genotype	<i>Order/sort genotype/haplotype object</i>
----------------	---

Description

Order/sort genotype or haplotype object according to order of allele names or genotypes

Usage

```

## S3 method for class 'genotype':
order(..., na.last=TRUE, decreasing=FALSE,
      alleleOrder=allele.names(x), genotypeOrder=NULL)

## S3 method for class 'genotype':
sort(x, decreasing=FALSE, na.last=NA, ...,
     alleleOrder=allele.names(x), genotypeOrder=NULL)

```

```
genotypeOrder(x)
genotypeOrder(x) <- value
```

Arguments

...	genotype or haplotype in <code>order</code> method; not used for <code>sort</code> method
x	genotype or haplotype in <code>sort</code> method
na.last	as in default <code>order</code> or <code>sort</code>
decreasing	as in default <code>order</code> or <code>sort</code>
alleleOrder	character, vector of allele names in wanted order
genotypeOrder	character, vector of genotype/haplotype names in wanted order
value	the same as in argument <code>order.genotype</code>

Details

Argument `genotypeOrder` can be useful, when you want that some genotypes appear "together", whereas they are not "together" by allele order.

Both methods (`order` and `sort`) work with genotype and haplotype classes.

If `alleleOrder` is given, `genotypeOrder` has no effect.

Genotypes/haplotypes, with missing alleles in `alleleOrder` are treated as NA and ordered according to `order` arguments related to NA values. In such cases a warning is issued ("Found data values not matching specified alleles. Converting to NA.") and can be safely ignored. Genotypes present in `x`, but not specified in `genotypeOrder`, are also treated as NA.

Value of `genotypeOrder` such as "B/A" matches also "A/B" in case of genotypes.

Only unique values in argument `alleleOrder` or `genotypeOrder` are used i.e. first occurrence prevails.

Value

The same as in `order` or `sort`

Author(s)

Gregor Gorjanc

See Also

[genotype](#), [allele.names](#), [order](#), and [sort](#)

Examples

```

x <- c("C/C", "A/C", "A/A", NA, "C/B", "B/A", "B/B", "B/C", "A/C")
alleles <- c("A", "B", "C")

g <- genotype(x, alleles=alleles, reorder="yes")
## "C/C" "A/C" "A/A" NA "B/C" "A/B" "B/B" "B/C" "A/C"

h <- haplotype(x, alleles=alleles)
## "C/C" "A/C" "A/A" NA "C/B" "B/A" "B/B" "B/C" "A/C"

## --- Standard usage ---

sort(g)
## "A/A" "A/B" "A/C" "A/C" "B/B" "B/C" "B/C" "C/C" NA

sort(h)
## "A/A" "A/C" "A/C" "B/A" "B/B" "B/C" "C/B" "C/C" NA

## --- Reversed order of alleles ---

sort(g, alleleOrder=c("B", "C", "A"))
## "B/B" "B/C" "B/C" "A/B" "C/C" "A/C" "A/C" "A/A" NA
## note that A/B comes after B/C since it is treated as B/A;
## order of alleles (not in alleleOrder!) does not matter for a genotype

sort(h, alleleOrder=c("B", "C", "A"))
## "B/B" "B/C" "B/A" "C/B" "C/C" "A/C" "A/C" "A/A" NA

## --- Missing allele(s) in alleleOrder ---

sort(g, alleleOrder=c("B", "C"))
## "B/B" "B/C" "B/C" "C/C" "A/C" "A/A" NA "A/B" "A/C"

sort(g, alleleOrder=c("B"))
## "B/B" "C/C" "A/C" "A/A" NA "B/C" "A/B" "B/C" "A/C"
## genotypes with missing allele are treated as NA

sort(h, alleleOrder=c("B", "C"))
## "B/B" "B/C" "C/B" "C/C" "A/C" "A/A" NA "B/A" "A/C"

sort(h, alleleOrder=c("B"))
## "B/B" "C/C" "A/C" "A/A" NA "C/B" "B/A" "B/C" "A/C"

## --- Use of genotypeOrder ---

sort(g, genotypeOrder=c("A/A", "C/C", "B/B", "A/B", "A/C", "B/C"))
## "A/A" "C/C" "B/B" "A/B" "A/C" "A/C" "B/C" "B/C" NA

sort(h, genotypeOrder=c("A/A", "C/C", "B/B",
                        "A/C", "C/B", "B/A", "B/C"))
## "A/A" "C/C" "B/B" "A/C" "A/C" "C/B" "B/A" "B/C" NA

```

```
## --- Missing genotype(s) in genotypeOrder ---

sort(g, genotypeOrder=c("C/C", "A/B", "A/C", "B/C"))
## "C/C" "A/B" "A/C" "A/C" "B/C" "B/C" "A/A" NA "B/B"

sort(h, genotypeOrder=c("C/C", "A/B", "A/C", "B/C"))
## "C/C" "A/C" "A/C" "B/C" "A/A" NA "C/B" "B/A" "B/B"
```

plot.genotype *Plot genotype object*

Description

plot.genotype can plot genotype or allele frequency of a genotype object.

Usage

```
## S3 method for class 'genotype':
plot(x, type=c("genotype", "allele"),
     what=c("percentage", "number"), ...)
```

Arguments

x	genotype object, as genotype.
type	plot "genotype" or "allele" frequency, as character.
what	show "percentage" or "number", as character
...	Optional arguments for barplot.

Value

The same as in barplot.

Author(s)

Gregor Gorjanc

See Also

[genotype](#), [barplot](#)

Examples

```
set <- c("A/A", "A/B", "A/B", "B/B", "B/B", "B/B",
        "B/B", "B/C", "C/C", "C/C")
set <- genotype(set, alleles=c("A", "B", "C"), reorder="yes")
plot(set)
plot(set, type="allele", what="number")
```

print.LD

Textual and graphical display of linkage disequilibrium (LD) objects

Description

Textual and graphical display of linkage disequilibrium (LD) objects

Usage

```
## S3 method for class 'LD':
print(x, digits = getOption("digits"), ...)
## S3 method for class 'LD.data.frame':
print(x, ...)

summary.LD.data.frame(object, digits = getOption("digits"),
                      which = c("D", "D'", "r", "X^2", "P-value", "n", " "),
                      rowsep, show.all = FALSE, ...)
## S3 method for class 'summary.LD.data.frame':
print(x, digits = getOption("digits"), ...)

## S3 method for class 'LD.data.frame':
plot(x,digits=3, colorcut=c(0,0.01, 0.025, 0.5, 0.1, 1),
     colors=heat.colors(length(colorcut)), textcol="black",
     marker, which="D'", distance, ...)

LDtable(x, colorcut=c(0,0.01, 0.025, 0.5, 0.1, 1),
        colors=heat.colors(length(colorcut)), textcol="black",
        digits=3, show.all=FALSE, which=c("D", "D'", "r", "X^2",
        "P-value", "n"), colorize="P-value", cex, ...)

LDplot(x, digits=3, marker, distance, which=c("D", "D'", "r", "X^2",
        "P-value", "n", " "), ... )
```

Arguments

x, object	LD or LD.data.frame object
digits	Number of significant digits to display
which	Name(s) of LD information items to be displayed
rowsep	Separator between rows of data, use NULL for no separator.
colorcut	P-value cutoffs points for colorizing LDtable
colors	Colors for each P-value cutoff given in colorcut for LDtable
textcol	Color for text labels for LDtable
marker	Marker used as 'comparator' on LDplot. If omitted separate lines for each marker will be displayed

distance	Marker location, used for locating of markers on LDplot.
show.all	If TRUE, show all rows/columns of matrix. Otherwise omit completely blank rows/columns.
colorize	LD parameter used for determining table cell colors
cex	Scaling factor for table text. If absent, text will be scaled to fit within the table cells.
...	Optional arguments (plot.LD.data.frame passes these to LDtable and LDplot)

Value

None.

Author(s)

Gregory R. Warnes (warnes@bst.rochester.edu)

See Also

LD, genotype, HWE.test

Examples

```

g1 <- genotype( c('T/A',    NA, 'T/T',    NA, 'T/A',    NA, 'T/T', 'T/A',
                  'T/T', 'T/T', 'T/A', 'A/A', 'T/T', 'T/A', 'T/A', 'T/T',
                  NA, 'T/A', 'T/A',    NA) )

g2 <- genotype( c('C/A', 'C/A', 'C/C', 'C/A', 'C/C', 'C/A', 'C/A', 'C/A',
                  'C/A', 'C/C', 'C/A', 'A/A', 'C/A', 'A/A', 'C/A', 'C/C',
                  'C/A', 'C/A', 'C/A', 'A/A') )

g3 <- genotype( c('T/A', 'T/A', 'T/T', 'T/A', 'T/T', 'T/A', 'T/A', 'T/A',
                  'T/A', 'T/T', 'T/A', 'T/T', 'T/A', 'T/A', 'T/A', 'T/T',
                  'T/A', 'T/A', 'T/A', 'T/T') )

data <- makeGenotypes(data.frame(g1,g2,g3))

# Compute & display LD for one marker pair
ld <- LD(g1,g2)
print(ld)

# Compute LD table for all 3 genotypes
ldt <- LD(data)

# display the results
print(ldt)                                # textual display
LDtable(ldt)                              # graphical color-coded table
LDplot(ldt, distance=c(124, 834, 927))    # LD plot vs distance

# more markers makes prettier plots!

```

```

data <- list()
nobs <- 1000
ngene <- 20
s <- seq(0,1,length=ngene)
a1 <- a2 <- matrix("", nrow=nobs, ncol=ngene)
for(i in 1:length(s) )
{

  rallele <- function(p) sample( c("A","T"), 1, p=c(p, 1-p))

  if(i==1)
  {
    a1[,i] <- sample( c("A","T"), 1000, p=c(0.5,0.5), replace=TRUE)
    a2[,i] <- sample( c("A","T"), 1000, p=c(0.5,0.5), replace=TRUE)
  }
  else
  {
    p1 <- pmax( pmin( 0.25 + s[i] * as.numeric(a1[,i-1]=="A"),1 ), 0 )
    p2 <- pmax( pmin( 0.25 + s[i] * as.numeric(a2[,i-1]=="A"),1 ), 0 )
    a1[,i] <- sapply(p1, rallele )
    a2[,i] <- sapply(p2, rallele )
  }

  data[[paste("G",i,sep="")] ] <- genotype(a1[,i],a2[,i])
}
data <- data.frame(data)
data <- makeGenotypes(data)

ldt <- LD(data)
plot(ldt, digits=2, marker=19) # do LDtable & LDplot on in a single
                               # graphics window

```

summary.genotype *Allele and Genotype Frequency from a Genotype or Haplotype Object*

Description

summary.genotype creates an object containing allele and genotype frequency from a genotype or haplotype object. print.summary.genotype displays a summary.genotype object.

Usage

```

## S3 method for class 'genotype':
summary(object, ..., maxsum)
## S3 method for class 'summary.genotype':
print(x, ..., round=2)

```

Arguments

object, x	an object of class <code>genotype</code> or <code>haplotype</code> (for <code>summary.genotype</code>) or an object of class <code>summary.genotype</code> (for <code>print.summary.genotype</code>)
...	optional parameters. Ignored by <code>summary.genotype</code> , passed to <code>print.matrix</code> by <code>print.summary.genotype</code> .
maxsum	specifying any value for the parameter <code>maxsum</code> will cause <code>summary.genotype</code> to fall back to <code>summary.factor</code> .
round	number of digits to use when displaying proportions.

Details

Specifying any value for the parameter `maxsum` will cause fallback to `summary.factor`. This is so that the function `summary.dataframe` will give reasonable output when it contains a genotype column. (Hopefully we can figure out something better to do in this case.)

Value

The returned value of `summary.genotype` is an object of class `summary.genotype` which is a list with the following components:

<code>locus</code>	locus information field (if present) from <code>x</code>
<code>allele.names</code>	vector of allele names
<code>allele.freq</code>	A two column matrix with one row for each allele, plus one row for NA values (if present). The first column, <code>Count</code> , contains the frequency of the corresponding allele value. The second column, <code>Proportion</code> , contains the fraction of alleles with the corresponding allele value. Note each observation contains two alleles, thus the <code>Count</code> field sums to twice the number of observations.
<code>genotype.freq</code>	A two column matrix with one row for each genotype, plus one row for NA values (if present). The first column, <code>Count</code> , contains the frequency of the corresponding genotype. The second column, <code>Proportion</code> , contains the fraction of genotypes with the corresponding value.

`print.summary.genotype` silently returns the object `x`.

Author(s)

Gregory R. Warnes (warnes@bst.rochester.edu)

See Also

[genotype](#), [HWE.test](#), [allele](#), [homozygote](#), [heterozygote](#), [carrier](#), [allele.count](#)
[locus gene marker](#)

Examples

```

example.data <- c("D/D", "D/I", "D/D", "I/I", "D/D",
                 "D/D", "D/D", "D/D", "I/I", "")
g1 <- genotype(example.data)
g1

summary(g1)

```

undocumented *Undocumented functions*

Description

These functions are undocumented. Some are internal and not intended for direct use. Some are not yet ready for end users. Others simply haven't been documented yet.

Author(s)

Gregory R. Warnes

write.pop.file *Create genetics data files*

Description

write.pop.file creates a 'pop' data file, as used by the GenePop (<http://wbiomed.curtin.edu.au/genepop/>) and LinkDos (<http://wbiomed.curtin.edu.au/genepop/linkdos.html>) software packages.

write.pedigree.file creates a 'pedigree' data file, as used by the QTDT software package (<http://www.sph.umich.edu/statgen/abecasis/QTDT/>).

write.marker.file creates a 'marker' data file, as used by the QTDT software package (<http://www.sph.umich.edu/statgen/abecasis/QTDT/>).

Usage

```

write.pop.file(data, file = "", digits = 2, description = "Data from R")
write.pedigree.file(data, family, pid, father, mother, sex,
                   file="pedigree.txt")
write.marker.file(data, location, file="marker.txt")

```

Arguments

<code>data</code>	Data frame containing genotype objects to be exported
<code>file</code>	Output filename
<code>digits</code>	Number of digits to use in numbering genotypes, either 2 or 3.
<code>description</code>	Description to use as the first line of the 'pop' file.
<code>family, pid, father, mother</code>	Vector of family, individual, father, and mother id's, respectively.
<code>sex</code>	Vector giving the sex of the individual (1=Male, 2=Female)
<code>location</code>	Location of the marker relative to the gene of interest, in base pairs.

Details

The format of 'Pop' files is documented at http://wbiomed.curtin.edu.au/genepop/help_input.html, the format of 'pedigree' files is documented at <http://www.sph.umich.edu/csg/abecasis/GOLD/docs/pedigree.html> and the format of 'marker' files is documented at <http://www.sph.umich.edu/csg/abecasis/GOLD/docs/map.html>.

Value

No return value.

Author(s)

Gregory R. Warnes <warnes@bst.rochester.edu>

See Also

[write.table](#)

Examples

```
# TBA
```

Index

- *Topic **IO**
 - write.pop.file, 40
- *Topic **hplot**
 - plot.genotype, 35
- *Topic **manip**
 - expectedGenotypes, 7
 - groupGenotype, 15
 - order.genotype, 32
- *Topic **misc**
 - ci.balance, 2
 - Deprecated, 3
 - diseq, 4
 - genotype, 8
 - gregorius, 13
 - groupGenotype, 15
 - homozygote, 17
 - HWE.chisq, 19
 - HWE.exact, 21
 - HWE.test, 22
 - LD, 24
 - locus, 26
 - makeGenotypes, 30
 - order.genotype, 32
 - print.LD, 36
 - summary.genotype, 38
 - undocumented, 40
- ==.genotype (*genotype*), 8
- ==.haplotype (*genotype*), 8
- [.genotype (*genotype*), 8
- [.haplotype (*genotype*), 8
- [<-.genotype (*genotype*), 8
- [<-.haplotype (*genotype*), 8
- %in%, 10
- %in% (*genotype*), 8

- allele, 10, 20, 39
- allele (*homozygote*), 17
- allele.count, 10, 39
- allele.count.2.genotype (*undocumented*), 40

- allele.count.genotype (*genotype*), 8
- allele.names, 7, 33
- as.character.gene (*locus*), 26
- as.character.locus (*locus*), 26
- as.character.marker (*locus*), 26
- as.factor (*undocumented*), 40
- as.factor.allele.genotype (*undocumented*), 40
- as.factor.default (*undocumented*), 40
- as.factor.genotype (*undocumented*), 40
- as.genotype (*genotype*), 8
- as.genotype.allele.count (*genotype*), 8
- as.genotype.character (*genotype*), 8
- as.genotype.default (*genotype*), 8
- as.genotype.factor (*genotype*), 8
- as.genotype.genotype (*genotype*), 8
- as.genotype.haplotype (*genotype*), 8
- as.genotype.table (*genotype*), 8
- as.haplotype (*genotype*), 8

- barplot, 35
- boot, 3, 6, 20
- bootci, 6, 20
- bootstrap, 3

- carrier, 10, 39
- carrier (*homozygote*), 17
- chisq.test, 20, 23
- ci.balance, 2

- Deprecated, 3
- diseq, 4, 20, 21, 23
- diseq.ci, 3, 20, 21, 23
- expectedGenotypes, 7, 10

- expectedHaplotypes
(*expectedGenotypes*), 7
- factor, 16
- gene, 10, 19, 39
- gene (*locus*), 26
- gene<- (*locus*), 26
- GeneticPower.Quantitative.Factor,
4
- GeneticPower.Quantitative.Numeric,
4
- geno.as.array (*undocumented*), 40
- genotype, 6, 7, 8, 16, 19, 23, 26, 28, 31, 33,
35, 39
- genotypeOrder, 10
- genotypeOrder (*order.genotype*), 32
- genotypeOrder<- (*order.genotype*),
32
- getgene (*locus*), 26
- getlocus (*locus*), 26
- getmarker (*locus*), 26
- GPC, 4
- gregorius, 13
- groupGenotype, 15
- hap (*undocumented*), 40
- hapambig (*undocumented*), 40
- hapenum (*undocumented*), 40
- hapfreq (*undocumented*), 40
- haplotype, 16
- haplotype (*genotype*), 8
- hapmcmc (*undocumented*), 40
- hapshuffle (*undocumented*), 40
- heterozygote, 10, 39
- heterozygote (*homozygote*), 17
- heterozygote.genotype (*genotype*),
8
- homozygote, 10, 17, 39
- homozygote.genotype (*genotype*), 8
- hstest, 23
- HWE.chisq, 19, 21, 23
- HWE.exact, 20, 21, 23
- HWE.test, 5, 6, 10, 19–21, 22, 26, 39
- is.gene (*locus*), 26
- is.genotype (*genotype*), 8
- is.haplotype (*genotype*), 8
- is.locus (*locus*), 26
- is.marker (*locus*), 26
- LD, 24
- LDplot (*print.LD*), 36
- LDtable (*print.LD*), 36
- levels, 16
- locus, 10, 19, 26, 39
- locus<- (*locus*), 26
- makeGenotypes, 30
- makeHaplotypes (*makeGenotypes*), 30
- marker, 10, 19, 39
- marker (*locus*), 26
- marker<- (*locus*), 26
- mknum (*undocumented*), 40
- mourant (*undocumented*), 40
- nallele (*genotype*), 8
- order, 33
- order (*order.genotype*), 32
- order.genotype, 32
- plot.genotype, 35
- plot.LD.data.frame (*print.LD*), 36
- power.casectl (*Deprecated*), 3
- print.allele.count (*genotype*), 8
- print.allele.genotype (*genotype*),
8
- print.diseq (*diseq*), 4
- print.gene (*locus*), 26
- print.genotype (*genotype*), 8
- print.HWE.test (*HWE.test*), 22
- print.LD, 36
- print.locus (*locus*), 26
- print.marker (*locus*), 26
- print.summary.genotype
(*summary.genotype*), 38
- print.summary.LD.data.frame
(*print.LD*), 36
- shortsummary.genotype
(*undocumented*), 40
- sort, 33
- sort.genotype, 10
- sort.genotype (*order.genotype*), 32
- summary.genotype, 10, 19, 38
- summary.LD.data.frame (*print.LD*),
36

undocumented, [40](#)

write.marker.file
 (*write.pop.file*), [40](#)

write.pedigree.file
 (*write.pop.file*), [40](#)

write.pop.file, [40](#)

write.table, [41](#)