# Package 'sequoia'

May 22, 2021

Type Package  Title Pedigree Inference from SNPs  Version 2.3.5  Date 2021-05-22  Author Jisca Huisman [aut, cre]  Maintainer Jisca Huisman spisca.huisman@gmail.com>  Description Multi-generational pedigree inference from incomplete data on hundreds of SNPs, including parentage assignment and sibship clustering. See Huisman (2017) ( <doi:10.1111 1755-0998.12665="">) for more information.  License GPL-2  URL https://jiscah.github.io/  LazyData TRUE  Imports plyr (&gt;= 1.8.0), stats, utils, graphics  RoxygenNote 7.1.1  Suggests xlsx, knitr, rmarkdown, bookdown, kinship2, lattice, R.rsp  VignetteBuilder knitr, R.rsp  NeedsCompilation yes  Repository CRAN  Date/Publication 2021-05-22 15:10:02 UTC  R topics documented:  CalcBYprobs CalcMaxMismatch CalcOHLLR CalcOHLLR CalcPairLL CalcRped</doi:10.1111>
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2 CalcBYprobs

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CalcBYprobs Birth year probabilities

# Description

Estimate the probability that an individual with unknown birth year is born in year y, based on the (estimated) birth years of its parents and offspring and the age distribution of other parent-offspring pairs.

# Usage

CalcBYprobs(Pedigree = NULL, LifeHistData = NULL, AgePrior = NULL)

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#### **Arguments**

Pedigree dataframe with columns id-dam-sire.

LifeHistData dataframe with 3 columns (optionally 5):

**ID** max. 30 characters long

**Sex** 1 = female, 2 = male, 3 = unknown, 4 = hermaphrodite, other numbers or NA = unknown

**BirthYear** birth or hatching year, integer, with missing values as NA or any negative value.

**BY.min** minimum birth year, only used if BirthYear is missing **BY.max** maximum birth year, only used if BirthYear is missing

If the species has multiple generations per year, use an integer coding such that the candidate parents' 'Birth year' is at least one smaller than their putative offspring's. Column names are ignored, so ensure column order is ID - sex - birth year (- BY.min - BY.max). Individuals do not need to be in the same order as in 'Pedigree', nor do all individuals in the pedigree need to be included.

AgePrior a matrix with probability ratios for individuals with age difference A to have

relationship R, as generated by MakeAgePrior. If NULL, MakeAgePrior is called

using its default values.

#### **Details**

This function assists in estimating birth years of individuals for which these are unknown, provided they have at least one parent or one offspring in the pedigree. It is not a substitute for field-based estimates of age, only a method to summarise the pedigree + birth year based information.

No distinction is made between genotyped and non-genotyped or dummy individuals.

#### Value

A matrix with for each individual (rows) in the pedigree that has a missing birth year in LifeHistData, or that is not included in LifeHistData, the probability that it is born in y (columns). Probabilities are rounded to 3 decimal points and may therefore not sum exactly to 1.

## WARNING

Any errors in the pedigree or lifehistory data will cause errors in the birth year probabilities of their parents and offspring, and putatively also of more distant ancestors and descendants. If the ageprior is based on the same erroneous pedigree and lifehistory data, all birth year probabilities will be affected.

## See Also

MakeAgePrior to estimate effect of age on relationships.

4 CalcMaxMismatch

#### **Examples**

CalcMaxMismatch

Maximum Number of Mismatches

# **Description**

Calculate the maximum expected number of mismatches for duplicate samples, parent-offspring pairs, and parent-parent-offspring trios.

#### Usage

```
CalcMaxMismatch(Err, MAF, ErrFlavour = "version2.0", qntl = 1 - 1e-05)
```

## Arguments

Err estimated genotyping error rate, as a single number or 3x3 matrix (averaged

value(s) across SNPs), or a vector with the same length as MAF, or a nSnp x 3 x 3 array. If a matrix, this should be the probability of observed genotype (columns) conditional on actual genotype (rows). Each row must therefore sum

to 1. If an array, each 3x3 slice should abide this rule.

MAF vector with minor allele frequency at each SNP.

ErrFlavour function that takes Err as input, and returns a 3x3 matrix of observed (columns)

conditional on actual (rows) genotypes, or choose from inbuilt ones as used in sequoia 'version2.0', 'version1.3', or 'version1.1'. Ignored if Err is a matrix.

See ErrToM.

qnt1 quantile of binomial distribution to be used as the maximum, of individual-

level probability. For a desired dataset-level probability quantile Q, use qntl =

 $Q^{(1)}$ , where N is the number of individuals.

#### **Details**

The thresholds for maximum number of mismatches calculated here aim to minimise false negatives, i.e. to minimise the chance that any true duplicates or true parent-offspring pairs are already excluded during the filtering steps where these MaxMismatch values are used. Consequently, there is a high probability of false positives, i.e. it is likely that some sample pairs with fewer mismatches than the MaxMismatch threshold, are in fact not duplicate samples or parent-offspring pairs. Use of these MaxMismatch thresholds is therefore only the first step of pedigree reconstruction by sequoia.

# Value

A vector with three integers:

DUP Maximum number of differences between 2 samples from the same individual
OH Maximum number of Opposing Homozygous SNPs between a true parent-offspring

pair

ME Maximum number of Mendelian Errors among a true parent-parent- offspring

trio

.

## See Also

```
SnpStats.
```

# Examples

```
CalcMaxMismatch(Err = 0.05, MAF = runif(n=100, min=0.3, max=0.5))
## Not run:
CalcMaxMismatch(Err = 0.02, MAF = SnpStats(MyGenoMatrix, Plot=FALSE)[,"AF"])
## End(Not run)
```

CalcOHLLR

Calculate OH and LLR

# **Description**

Count opposite homozygous (OH) loci between parent-offspring pairs and Mendelian errors (ME) between parent-parent-offspring trios, and calculate the parental log-likelihood ratios (LLR). Also returns estimate of actual genotypes.

# Usage

```
CalcOHLLR(
  Pedigree = NULL,
  GenoM = NULL,
  CalcLLR = TRUE,
  LifeHistData = NULL,
  AgePrior = FALSE,
  SeqList = NULL,
  Err = 1e-04,
  ErrFlavour = "version2.0",
  Tassign = 0.5,
  Tfilter = -2,
  Complex = "full",
  Herm = "no",
  quiet = FALSE
)
```

## **Arguments**

Pedigree dataframe with columns id-dam-sire. May include non-genotyped individu-

als, which will be treated as dummy individuals. If provided, any pedigree in

SeqList is ignored.

GenoM numeric matrix with genotype data: One row per individual, and one column

per SNP, coded as 0, 1, 2 or -9 (missing). See also GenoConvert.

CalcLLR calculate log-likelihood ratios for all assigned parents (genotyped + dummy/non-

> genotyped; parent vs. otherwise related). If FALSE, only number of mismatching SNPs are counted (OH & ME), and parameters LifeHistData, AgePrior, Err, Tassign, and Complex are **ignored**. Note also that calculating likelihood ratios

is much more time consuming than counting OH & ME.

LifeHistData dataframe with 3 columns (optionally 5):

**ID** max. 30 characters long

**Sex** 1 = female, 2 = male, 3 = unknown, 4 = hermaphrodite, other numbers or NA = unknown

**BirthYear** birth or hatching year, integer, with missing values as NA or any negative value.

**BY.min** minimum birth year, only used if BirthYear is missing

**BY.max** maximum birth year, only used if BirthYear is missing

If the species has multiple generations per year, use an integer coding such that the candidate parents' 'Birth year' is at least one smaller than their putative offspring's. Column names are ignored, so ensure column order is ID - sex birth year (- BY.min - BY.max). Individuals do not need to be in the same order

as in 'GenoM', nor do all genotyped individuals need to be included.

AgePrior logical (TRUE/FALSE) whether to estimate the ageprior from Pedigree and LifeHistData,

> or a matrix as generated by MakeAgePrior and included in the sequoia output. The AgePrior affects which relationships are considered possible: only those where P(A|R)/P(A) > 0. When TRUE, MakeAgePrior is called using its default values. When FALSE, all relationships are considered possible for all age differences, except that parent-offspring pairs cannot have age difference zero,

and grand-parental pairs have an age difference of at least two.

SeqList list with output from sequoia. If input parameter Pedigree=NULL, SeqList\$Pedigree

> will be used if present, and SeqList\$PedigreePar otherwise. If SeqList\$Specs is present, input parameters with the same name as its items are ignored, except 'CalcLLR' and 'AgePriors=FALSE'. The list elements 'LifeHist', 'AgePriors', and 'ErrM' are also used if present, and override the corresponding input pa-

rameters.

estimated genotyping error rate, as a single number or 3x3 matrix. Details below.

The error rate is presumed constant across SNPs, and missingness is presumed

random with respect to actual genotype.

ErrFlavour function that takes Err (single number) as input, and returns a 3x3 matrix of

> observed (columns) conditional on actual (rows) genotypes, or choose from inbuilt options 'version2.0', 'version1.3', or 'version1.1', referring to the sequoia

> version in which they were the default. Ignored if Err is a matrix. See ErrToM.

Err

Tassign minimum LLR required for acceptance of proposed relationship, relative to next

most likely relationship. Higher values result in more conservative assignments.

Must be zero or positive.

Tfilter threshold log10-likelihood ratio (LLR) between a proposed relationship versus

unrelated, to select candidate relatives. Typically a negative value, related to the fact that unconditional likelihoods are calculated during the filtering steps. More negative values may decrease non-assignment, but will increase computational

time.

Complex Breeding system complexity. Either "full" (default), "simp" (simplified, no ex-

plicit consideration of inbred relationships), "mono" (monogamous).

Herm Hermaphrodites, either "no", "A" (distinguish between dam and sire role, default

if at least 1 individual with sex=4), or "B" (no distinction between dam and sire

role). Both of the latter deal with selfing.

quiet logical, suppress messages

#### **Details**

Any individual in Pedigree that does not occur in GenoM is substituted by a dummy individual; these can be recognised by the value 0' in columns 'SNPd.id.dam' and 'SNPd.id.sire' in the output. For non-genotyped individuals the parental log-likelihood ratio can be calculated if they have at least one genotyped offspring (see also getAssignCat).

The birth years in LifeHistData and the AgePrior are not used in the calculation and do not affect the value of the likelihoods for the various relationships, but they \_are\_ used during some filtering steps, and may therefore affect the likelihood \_ratio\_. The default (AgePrior=FALSE) assumes all age-relationship combinations are possible, which may mean that some additional alternatives are considered compared to the sequoia default, resulting in somewhat lower LLR values.

A negative LLR for A's parent B indicates either that B is not truely the parent of A, or that B's parents are incorrect. The latter may cause B's presumed true, unobserved genotype to divert from its observed genotype, with downstream consequences for its offspring. In rare cases it may also be due to 'weird', non-implemented double or triple relationships between A and B.

#### Value

The Pedigree dataframe with additional columns:

LLRdam Log10-Likelihood Ratio (LLR) of this female being the mother, versus the next

most likely relationship between the focal individual and this female (see Details

for relationships considered)

LLRsire idem, for male parent

LLR for the parental pair, versus the next most likely configuration between the

three individuals (with one or neither parent assigned)

OHdam Number of loci at which the offspring and mother are opposite homozygotes

OHsire idem, for father

MEpair Number of Mendelian errors between the offspring and the parent pair, includes

OH as well as e.g. parents being opposing homozygotes, but the offspring not being a heterozygote. The offspring being OH with both parents is counted as 2

errors.

SNPd.id	Number of SNPs scored (non-missing) for the focal individual
SNPd.id.dam	Number of SNPs scored (non-missing) for both individual and dam
SNPd.id.sire	Number of SNPs scored for both individual and sire
Sexx	Sex in LifeHistData, or inferred Sex when assigned as part of parent-pair
BY.est	mode of birth year probability distribution
BY.lo	lower limit of 95% highest density region of birth year probability distribution
BY.hi	higher limit

The columns 'LLRdam', 'LLRsire' and 'LLRpair' are only included when CalcLLR=TRUE. When a parent or parent-pair is incompatible with the lifehistory data or presumed genotyping error rate, the error value '777' may be given.

The columns 'Sexx', 'BY.est', 'BY.lo' and 'BY.hi' are only included when LifeHistData is provided, and at least one genotyped individual has an unknown birth year or unknown sex.

## See Also

SummarySeq for visualisation of OH & LLR distributions; CalcPairLL for the likelihoods underlying the LLR, GenoConvert to read in various genotype data formats, CheckGeno; PedPolish to check and 'polish' the pedigree; getAssignCat to find which id-parent pairs are both genotyped or can be substituted by dummy individuals; sequoia for pedigree reconstruction.

```
data(Ped_HSg5, SimGeno_example, LH_HSg5, package="sequoia")
# count Mendelian errors in an existing pedigree
Ped.OH <- CalcOHLLR(Pedigree = Ped_HSg5, GenoM = SimGeno_example,</pre>
                    CalcLLR = FALSE)
Ped.OH[50:55,]
# view histograms
SummarySeq(Ped.OH, Panels="OH")
# Parent likelihood ratios in an existing pedigree, including for
# non-genotyped parents
Ped.LLR <- CalcOHLLR(Pedigree = Ped_HSg5, GenoM = SimGeno_example,</pre>
                     CalcLLR = TRUE, LifeHistData=LH_HSg5, AgePrior=TRUE)
SummarySeq(Ped.LLR, Panels="LLR")
# likelihood ratios change with presumed genotyping error rate:
Ped.LLR.B <- CalcOHLLR(Pedigree = Ped_HSg5, GenoM = SimGeno_example,</pre>
                     CalcLLR = TRUE, LifeHistData=LH_HSg5, AgePrior=TRUE,
                    Err = 0.005)
SummarySeq(Ped.LLR.B, Panels="LLR")
# run sequoia with CalcLLR=FALSE, and add OH + LLR later:
data(Ped_griffin, LH_griffin, package="sequoia")
Genotypes <- SimGeno(Ped_griffin, nSnp=400)</pre>
SeqOUT <- sequoia(Genotypes, LH_griffin, CalcLLR=FALSE,quiet=TRUE,Plot=FALSE)</pre>
PedA <- CalcOHLLR(Pedigree = SeqOUT[["Pedigree"]][, 1:3], GenoM = Genotypes,</pre>
```

```
LifeHistData = LH_griffin, AgePrior = TRUE, Complex = "full")
SummarySeq(PedA, Panels=c("LLR", "OH"))
```

CalcPairLL

Calculate Likelihoods for Alternative Relationships

# **Description**

For each specified pair of individuals, calculate the log10-likelihoods of being PO, FS, HS, GP, FA, HA, U (see Details). Individuals must be genotyped or have at least one genotyped offspring.

**NOTE** values > 0 are various NA types, see 'Likelihood special codes' in 'Value' section below.

## Usage

```
CalcPairLL(
 Pairs = NULL,
  GenoM = NULL,
  Pedigree = NULL,
  LifeHistData = NULL,
  AgePrior = TRUE,
  SeqList = NULL,
  Complex = "full",
 Herm = "no",
  Err = 1e-04,
  ErrFlavour = "version2.0",
 Tassign = 0.5,
 Tfilter = -2,
  quiet = FALSE,
 Plot = TRUE
)
```

## **Arguments**

Pairs

dataframe with columns ID1 and ID2, and optionally

**Sex1** Sex of ID1, 1=female, 2=male, 3=unknown, or NA to take from LifeHistData. The sex of individuals occurring as parent in Pedigree cannot be altered.

Sex2 Sex of ID2

**AgeDif** Age difference in whole time units, BirthYear1 - BirthYear2 (i.e. positive if ID2 is born before ID1). If NA, calculated from LifeHistData. Use '999' to explicitly specify 'unknown'.

**focal** relationship character abbreviation; PO, FS, HS, GP or U. See Details for its effect and explanation of abbreviations. Default: U

> patmat 1=maternal relatives, 2=paternal relatives. Only relevant for HS & GP, for which it defaults to Sex1, or 1 if Sex1=3, but is currently only predictably implemented for pairs of two genotyped individuals. Always equal to Sex2 for PO pairs when Sex2 is known.

> dropPar1 Drop the parents of ID1 before calculating the pair likelihood, rather than conditioning on them; choose from 'none', 'dam', 'sire', or 'both'. See example. If e.g. the pair shares a common mother, 'none' and 'sire' will condition on this shared mother and not calculate the likelihood that they are maternal siblings, while dropPar1='dam' or 'both' will calculate that likelihood, and the other likelihoods as if the mother of ID1 were unknown.

dropPar2 as dropPar1, for ID2

numeric matrix with genotype data: One row per individual, and one column per SNP, coded as 0, 1, 2 or -9 (missing). See also GenoConvert.

dataframe with columns id-dam-sire; likelihoods will be calculated conditional on the pedigree. May include non-genotyped individuals, which will be treated as dummy individuals.

dataframe with 3 columns (optionally 5): LifeHistData

**ID** max. 30 characters long

**Sex** 1 = female, 2 = male, 3 = unknown, 4 = hermaphrodite, other numbers or NA = unknown

**BirthYear** birth or hatching year, integer, with missing values as NA or any negative value.

**BY.min** minimum birth year, only used if BirthYear is missing **BY.max** maximum birth year, only used if BirthYear is missing

If the species has multiple generations per year, use an integer coding such that the candidate parents' 'Birth year' is at least one smaller than their putative offspring's. Column names are ignored, so ensure column order is ID - sex birth year (- BY.min - BY.max). Individuals do not need to be in the same order as in 'GenoM', nor do all genotyped individuals need to be included.

logical (TRUE/FALSE) whether to estimate the ageprior from Pedigree and LifeHistData,

or a matrix as generated by MakeAgePrior and included in the sequoia output. The AgePrior affects which relationships are considered possible: only those where P(A|R)/P(A) > 0. When TRUE, MakeAgePrior is called using its default values. When FALSE, all relationships are considered possible for all age differences, except that parent-offspring pairs cannot have age difference zero, and grand-parental pairs have an age difference of at least two.

list with output from sequoia. If input parameter Pedigree=NULL, SeqList\$Pedigree will be used if present, and SeqList\$PedigreePar otherwise. If SeqList\$Specs is present, input parameters with the same name as its items are ignored. The list elements 'LifeHist', 'AgePriors', and 'ErrM' are also used if present, and

override the corresponding input parameters.

Breeding system complexity. Either "full" (default), "simp" (simplified, no explicit consideration of inbred relationships), "mono" (monogamous).

Hermaphrodites, either "no", "A" (distinguish between dam and sire role, default if at least 1 individual with sex=4), or "B" (no distinction between dam and sire role). Both of the latter deal with selfing.

GenoM

Pedigree

AgePrior

SeqList

Complex

Herm

Err estimated genotyping error rate, as a single number or 3x3 matrix. Details below.

The error rate is presumed constant across SNPs, and missingness is presumed

random with respect to actual genotype.

ErrFlavour function that takes Err (single number) as input, and returns a 3x3 matrix of

observed (columns) conditional on actual (rows) genotypes, or choose from inbuilt options 'version2.0', 'version1.3', or 'version1.1', referring to the sequoia version in which they were the default. Ignored if Err is a matrix. See ErrToM.

Tassign minimum LLR required for acceptance of proposed relationship, relative to next

most likely relationship. Higher values result in more conservative assignments.

Must be zero or positive.

Tfilter threshold log10-likelihood ratio (LLR) between a proposed relationship versus

unrelated, to select candidate relatives. Typically a negative value, related to the fact that unconditional likelihoods are calculated during the filtering steps. More negative values may decrease non-assignment, but will increase computational

time.

quiet logical, suppress messages

Plot logical, display scatter plots by PlotPairLL.

## **Details**

The same pair may be included multiple times, e.g. with different sex, age difference, or focal relationship, to explore their effect on the likelihoods. Likelihoods are only calculated for relationships that are possible given the age difference, e.g. PO (parent-offspring) is not calculated for pairs with an age difference of 0.

Non-genotyped individuals can be included if they have at least one genotyped offspring and can be turned into a dummy (see getAssignCat); to establish this a pedigree must be provided.

**Warning 1**: There is no check whether the input pedigree is genetically sensible, it is simply conditioned upon. Checking whether a pedigree is compatible with the SNP data can be done with CalcOHLLR.

Warning 2: Conditioning on a Pedigree can make computation orders of magnitude slower.

## Value

The Pairs dataframe including all optional columns listed above, plus the additional columns:

LL\_xx Log10-Likelihood of this pair having relationship xx, with xx being one of PO,

FS, etc. as detailed below.

TopRel Abbreviation of most likely relationship

LLR Likelihood ratio between most-likely and second most likely relationships

#### **Relationship abbreviations:**

PO Parent - offspring
FS Full siblings
HS Half siblings
GP Grandparent

FA	Full avuncular
НА	Half avuncular and other 3rd degree relationships
U	Unrelated
2nd	Unclear which type of 2nd degree relatives (HS, GP, or FA)
??	Unclear which type of 1st, 2nd or 3rd degree relatives

#### Likelihood special codes:

222	Maybe (via) other parent (e.g. focal="GP", but as likely to be maternal as paternal grandparent, and therefore not assignable)
333	Excluded from comparison (shouldn't occur)
444	Not implemented (e.g. would create an odd double/triple relationship in combination with the provided pedigree)
777	Impossible (e.g. cannot be both full sibling and grandparent)
888	Already assigned in the provided pedigree (see dropPar arguments)
999	NA

#### **Double relationships & focal relationship**

Especially when Complex='full', not only the seven relationship alternatives listed above are considered, but a whole range of possible double and even triple relationships. For example, mother A and offspring B (PO) may also be paternal half-siblings (HS, A and A's mother mated with same male), grandmother and grand-offspring (GP, B's father is A's son), or paternal aunt (B's father is a full or half sib of A).

The likelihood reported as 'LL\_PO' is the most-likely one of the possible alternatives, among those that are not impossible due to age differences or due to the pedigree (as reconstructed up to that point). Whether e.g. the likelihood to be both PO & HS is counted as PO or as HS, depends on the situation and is determined by the variable 'focal': During parentage assignment, it is counted as PO but not HS, while during sibship clustering, it is counted as HS but not PO – not omitting from the alternative relationship would result in a deadlock.

## See Also

PlotPairLL to plot alternative relationship pairs from the output; CalcOHLLR to calculate LLR for parents & parent-pairs in a pedigree; GetRelM to find all pairwise relatives according to the pedigree; GetMaybeRel to get likely relative pairs not in the pedigree.

CalcRped 13

```
# LLRdam & LLRsire:
CalcPairLL(Pairs, SimGeno_example)
# LLRpair is min. of dam & sire LLR, conditional on co-parent:
CalcPairLL(cbind(Pairs, dropPar1=rep(c("dam", "sire"), each=3)),
           SimGeno_example, SeqList = Seq.HSg5)
# likelihoods underlying LLR in getMaybeRel output:
data(Ped_griffin, SeqOUT_griffin, package="sequoia")
Geno.griffin <- SimGeno(Ped_griffin, nSnp=200, SnpError = 0.01, ParMis=0.4)
MR <- GetMaybeRel(GenoM = Geno.griffin,</pre>
                  LifeHistData = SeqOUT_griffin$LifeHist,
                  Module = "par", Err = 0.001)
FivePairs <- MR$MaybePar[1:5, c("ID1", "ID2", "Sex1", "Sex2")]</pre>
FivePairs$AgeDif <- NA  # pretend unknown age differences
PairLL <- CalcPairLL(Pairs = rbind( cbind(FivePairs, focal = "PO"),</pre>
                                     cbind(FivePairs, focal = "HS"),
                                     cbind(FivePairs, focal = "GP")),
                     GenoM = Geno.griffin,
                     Err = 0.005, Plot=FALSE)
PairLL[c(1, 6, 11), ]
# LL(FS)==222 : HSHA, HSGP, FAHA more likely than FS
# LL(GP) higher when focal=HS: GP via 'other' parent also considered
# LL(FA) higher when focal=PO: FAHA, or FS of 'other' parent
```

CalcRped

Calculate Pedigree Relatedness

## **Description**

Morph pedigree into a **kinship2** compatible format and use **kinship** to calculate kinship coefficients; relatedness = 2\*kinship.

## Usage

```
CalcRped(Pedigree, OUT = "DF")
```

## **Arguments**

Pedigree dataframe with columns id-dam-sire.

OUT desired output format, 'M' for matrix or 'DF' for dataframe with columns IID1 - IID2 - R.ped.

#### Value

A matrix or dataframe.

14 CheckGeno

CheckGeno

Check Genotype Matrix

## **Description**

Check that the provided genotype matrix is in the correct format, and check for low call rate samples and SNPs.

## Usage

```
CheckGeno(
   GenoM,
   quiet = FALSE,
   Plot = FALSE,
   Return = "GenoM",
   DumPrefix = c("F0", "M0")
)
```

# **Arguments**

GenoM the genotype matrix. quiet suppress messages.

Plot display the plots of SnpStats.

Return either 'GenoM' to return the cleaned-up genotype matrix, or 'excl' to return a

list with excluded SNPs and individuals (see Value).

DumPrefix length 2 vector, to check if these don't occur among genotyped individuals.

#### Value

If Return='excl' a list with, if any are found:

ExcludedSNPs SNPs scored for <10 excluded when running sequoia

ExcludedSnps-mono

monomorphic (fixed) SNPs; automatically excluded when running sequoia. This includes nearly-fixed SNPs with MAF = 1/2N. Column numbers are

\*after\* removal of ExcludedSNPs, if any.

ExcludedIndiv Individuals scored for <5 reliably included during pedigree reconstruction. In-

dividual call rate is calculated after removal of 'Excluded SNPs'

Snps-LowCallRate

SNPs scored for 10 recommended to be filtered out

Indiv-LowCallRate

individuals scored for <50 recommended to be filtered out

When Return='excl' the return is invisible, i.e. a check is run and warnings or errors are always displayed, but nothing may be returned.

#### **Thresholds**

Appropriate call rate thresholds for SNPs and individuals depend on the total number of SNPs, distribution of call rates, genotyping errors, and the proportion of candidate parents that are SNPd (sibship clustering is more prone to false positives). Note that filtering first on SNP call rate tends to keep more individuals in.

#### See Also

SnpStats to calculate SNP call rates; CalcOHLLR to count the number of SNPs scored in both focal individual and parent.

# **Examples**

```
data(Ped_HSg5)
GenoM <- SimGeno(Ped_HSg5, nSnp=400, CallRate = runif(400, 0.2, 0.8))</pre>
# quick alternative:
GenoM.checked <- CheckGeno(GenoM)</pre>
# user supervised alternative:
Excl <- CheckGeno(GenoM, Return = "excl")</pre>
GenoM.orig <- GenoM # make a 'backup' copy</pre>
if ("ExcludedSnps" %in% names(Excl))
  GenoM <- GenoM[, -Excl[["ExcludedSnps"]]]</pre>
if ("ExcludedInd" %in% names(Excl))
  GenoM <- GenoM[!rownames(GenoM) %in% Excl[["ExcludedInd"]], ]</pre>
if ("ExcludedIndiv" %in% names(Excl))
  GenoM <- GenoM[!rownames(GenoM) %in% Excl[["ExcludedIndiv"]], ]</pre>
# warning about SNPs scored for <50% of individuals ?
SnpCallRate <- apply(GenoM, MARGIN=2,</pre>
                      FUN = function(x) sum(x!=-9)) / nrow(GenoM)
hist(SnpCallRate, breaks=50, col="grey")
GenoM <- GenoM[, SnpCallRate > 0.6]
# to be on the safe side, filter out low call rate individuals
IndivCallRate <- apply(GenoM, MARGIN=1,</pre>
                        FUN = function(x) sum(x!=-9)) / ncol(GenoM)
hist(IndivCallRate, breaks=50, col="grey")
GoodSamples <- rownames(GenoM)[ IndivCallRate > 0.8]
```

ComparePairs

Compare Pairwise Relationships

#### **Description**

Compare, count and identify different types of relative pairs between two pedigrees, or within one pedigree.

## Usage

```
ComparePairs(
  Ped1 = NULL,
  Ped2 = NULL,
  Pairs2 = NULL,
  GenBack = 1,
  patmat = FALSE,
  ExcludeDummies = TRUE,
  DumPrefix = c("F0", "M0"),
  Return = "Counts"
)
```

#### **Arguments**

Ped1 first (e.g. original/reference) pedigree, dataframe with 3 columns: id-dam-sire.

Ped2 optional second (e.g. inferred) pedigree.

Pairs2 optional dataframe with relationships categories between pairs of individuals,

instead of or in addition to Ped2, e.g. as returned by GetMaybeRe1. First three columns: ID1-ID2-relationship, column names and any additional columns are

ignored.

GenBack number of generations back to consider; 1 returns parent-offspring and sibling

relationships, 2 also returns grandparental, avuncular and first cousins. GenBack

>2 is not implemented.

patmat logical, distinguish between paternal versus maternal relative pairs?

ExcludeDummies logical, exclude dummy IDs from output? Individuals with e.g. the same

dummy father will still be counted as paternal halfsibs. No attempt is made to match dummies in one pedigree to individuals in the other pedigree; for that

use PedCompare.

DumPrefix character vector with the prefixes identifying dummy individuals. Use 'F0'

('M0') to avoid matching to regular individuals with IDs starting with 'F' ('M'),

provided Ped2 has fewer than 999 dummy females (males).

Return return a matrix with Counts or a Summary of the number of identical relation-

ships and mismatches per relationship, or detailed results as a 2xNxN Array or

as a Dataframe. All returns a list with all four.

## Details

If Pairs2 is as returned by GetMaybeRel (identified by the additional column names 'LLR' and 'OH'), these relationship categories are appended with an '?' in the output, to distinguish them from those derived from Ped2.

When Pairs2\$TopRel contains values other than the ones listed among the return values for the combination of patmat and GenBack, they are prioritised in decreasing order of factor levels, or in decreasing alphabetical order, and before the default (ped2 derived) levels.

The matrix returned by DyadCompare [Deprecated] is a subset of the matrix returned here using default settings.

#### Value

Array

Depending on Return, one of the following, or a list with all:

Counts (the default), a matrix with counts, with the classification in Ped1 on rows and

that in Ped2 in columns. Counts for 'symmetrical' pairs ("FS", "HS", "MHS",

"PHS", "FC1", "DFC1", "U", "X") are divided by two.

Summary a matrix with one row per relationship type and four columns, named as if Ped1

is the true pedigree:

n total number of pairs with that relationship in Ped1, and occurring in Ped2

OK Number of pairs with same relationship in Ped2 as in Ped1

**hi** Number of pairs with 'higher' relationship in Ped2 as in Ped1 (e.g. FS instead of HS; ranking is the order given below)

**lo** Number of pairs with 'lower' relationship in Ped2 as in Ped1, but not unrelated in Ped2

a 2xNxN array (if Ped2 or Pairs2 is specified) or a NxN matrix, where N is the

total number of individuals occurring in Ped1 and/or Ped2.

Dataframe a dataframe with  $N^2$  rows and four columns:

id.A First individual of the pairid.B Second individual of the pair

 $\pmb{RC1} \ \ \text{the relationship category in Ped1, as a factor with all considered categories}$ 

as levels, including those with 0 count

RC2 the relationship category in Ped2

Each pair is listed twice, e.g. once as P and once as O, or twice as FS.

## Relationship abbreviations and ranking

By default (GenBack=1, patmat=FALSE) the following 7 relationships are distinguished:

- S: Self (not included in Counts)
- MP: Parent
- **O**: Offspring (not included in Counts)
- **FS**: Full sibling
- HS: Half sibling
- U: Unrelated, or otherwise related
- X: Either or both individuals not occurring in both pedigrees

In the array and dataframe, 'MP' indicates that the second (column) individual is the parent of the first (row) individual, and 'O' indicates the reverse.

When GenBack=1, patmat=TRUE the categories are (S)-M-P-(O)-FS-MHS-PHS- U-X.

When GenBack=2, patmat=TRUE, the following relationships are distinguished:

- S: Self (not included in Counts)
- M: Mother
- P: Father

- **O**: Offspring (not included in Counts)
- **FS**: Full sibling
- MHS: Maternal half-sibling
- PHS: Paternal half-sibling
- MGM: Maternal grandmother
- MGF: Maternal grandfather
- PGM: Paternal grandmother
- PGF: Paternal grandfather
- **GO**: Grand-offspring (not included in Counts)
- FA: Full avuncular; maternal or paternal aunt or uncle
- HA: Half avuncular
- **FN**: Full nephew/niece (not included in Counts)
- HN: Half nephew/niece (not included in Counts)
- FC1: Full first cousin
- DFC1: Double full first cousin
- U: Unrelated, or otherwise related
- X: Either or both individuals not occurring in both pedigrees

Note that for avuncular and cousin relationships no distinction is made between paternal versus maternal, as this may differ between the two individuals and would generate a large number of sub-classes. When a pair is related via multiple paths, the first-listed relationship is returned.

When GenBack=2, patmat=FALSE, MGM, MGF, PGM and PGF are combined into GP, with the rest of the categories analogous to the above.

#### See Also

PedCompare for individual-based comparison; GetRelM for a pairwise relationships matrix of a single pedigree; PlotRelPairs for visualisation of relationships within each pedigree.

To estimate P(actual relationship (Ped1) | inferred relationship (Ped2)), see examples at EstConf.

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```
RelDF[RelDF$id.A=="i190_2010_M" & RelDF$id.B=="i168_2009_F", ]
# Colony-style lists of full sib dyads & half sib dyads:
FullSibDyads <- with(RelDF, RelDF[Ped1 == "FS" & id.A < id.B, ])
HalfSibDyads <- with(RelDF, RelDF[Ped1 == "HS" & id.A < id.B, ])
# Use 'id.A < id.B' because each pair is listed 2x</pre>
```

DyadCompare

Compare Dyads

# Description

Count the number of half and full sibling pairs correctly and incorrectly assigned. DEPRECATED - PLEASE USE ComparePairs

## Usage

```
DyadCompare(Ped1 = NULL, Ped2 = NULL, na1 = c(NA, "0"))
```

## **Arguments**

Ped1 original pedigree, dataframe with 3 columns: id-dam-sire.

Ped2 second (inferred) pedigree.

na1 the value for missing parents in Ped1.

#### Value

A 3x3 table with the number of pairs assigned as full siblings (FS), half siblings (HS) or unrelated (U, including otherwise related) in the two pedigrees, with the classification in Ped1 on rows and that in Ped2 in columns.

#### See Also

ComparePairs which supersedes this function; PedCompare

20 ErrToM

**ErrToM** 

Generate Genotyping Error Matrix

## Description

Generate a matrix with the probabilities of observed genotypes (columns) conditional on actual genotypes (rows), or return a function to generate such matrices (using a single value Err as input to that function).

# Usage

```
ErrToM(Err = NA, flavour = "version2.0", Return = "matrix")
```

## **Arguments**

Err estimated genotyping error rate, as a single number or 3x3 or 4x4 matrix. If a

single number, an error model is used that aims to deal with scoring errors typical for SNP arrays. If a matrix, this should be the probability of observed genotype (columns) conditional on actual genotype (rows). Each row must therefore

sum to 1. If Return='function', this may be NA.

flavour matrix-generating function, or one of 'version2.0', 'version1.3' (='SNPchip'),

'version1.1' (='version111'), referring to the sequoia version in which it was used as default. Ignored if Err is a matrix and Return='matrix' (in which case

the matrix will only be checked for validity).

Return output, 'matrix' (always 3x3) or 'function'.

#### **Details**

By default (flavour = "SNPchip"), Err is interpreted as a locus-level error rate (rather than allele-level), and equals the probability that an actual heterozygote is observed as either homozygote (i.e., the probability that it is observed as AA = probability that observed as aa = Err/2). The probability that one homozygote is observed as the other is  $(\text{Err}/2)^2$ .

The inbuilt 'flavours' correspond to the presumed and simulated error structures, which have changed with sequoia versions. The most appropriate error structure will depend on the genotyping platform; 'version0.9' and 'version1.1' were inspired by SNP array genotyping while 'version1.3' and 'version2.0' are intended to be more general.

Pr(observed genotype (columns) | actual genotype (rows)):

version2.0:

$$\begin{array}{ccccc} & \mathbf{0} & \mathbf{1} & \mathbf{2} \\ \mathbf{0} & (1-E/2)^2 & E(1-E/2) & (E/2)^2 \\ \mathbf{1} & E/2 & 1-E & E/2 \\ \mathbf{2} & (E/2)^2 & E(1-E/2) & (1-E/2)^2 \end{array}$$

version1.3

$$\begin{array}{ccccc} & \mathbf{0} & \mathbf{1} & \mathbf{2} \\ \mathbf{0} & 1 - E - (E/2)^2 & E & (E/2)^2 \\ \mathbf{1} & E/2 & 1 - E & E/2 \\ \mathbf{2} & (E/2)^2 & E & 1 - E - (E/2)^2 \end{array}$$

version1.1

version0.9 (not recommended)

$$\begin{array}{cccccc} & \mathbf{0} & \mathbf{1} & \mathbf{2} \\ \mathbf{0} & 1-E & E & 0 \\ \mathbf{1} & E/2 & 1-E & E/2 \\ \mathbf{2} & 0 & E & 1-E \end{array}$$

# Value

Either a 3x3 matrix, or a function generating a 3x3 matrix.

EstConf

Confidence Probabilities

# **Description**

Estimate confidence probabilities ('backward') and assignment error rates ('forward') per category (genotyped/dummy) by repeatedly simulating genotype data from a reference pedigree using SimGeno, reconstruction a pedigree from this using sequoia, and counting the number of mismatches using PedCompare.

## Usage

```
EstConf(
  Pedigree = NULL,
  LifeHistData = NULL,
  args.sim = list(nSnp = 400, SnpError = 0.001, ParMis = c(0.4, 0.4)),
  args.seq = list(Module = "ped", Err = 0.001, Tassign = 0.5, CalcLLR = FALSE),
  nSim = 10,
```

```
nCores = 1,
quiet = TRUE
)
```

## **Arguments**

Pedigree reference pedigree from which to simulate, dataframe with columns id-dam-sire.

Additional columns are ignored.

LifeHistData dataframe with id, sex (1=female, 2=male, 3=unknown), and birth year.

args.sim list of arguments to pass to SimGeno, such as nSnp (number of SNPs), SnpError

(genotyping error rate) and ParMis (proportion of non-genotyped parents). Set

to NULL to use all default values.

args.seq list of arguments to pass to sequoia, such as Module ('par' or 'ped'), Err (as-

sumed genotyping error rate), and Complex. May include (part of) SeqList, a list of sequoia output (i.e. as a list-within-a-list). Set to NULL to use all default

values.

nSim number of iterations of simulate - reconstruct - compare to perform, i.e. number

of simulated datasets.

nCores number of computer cores to use. If >1, package **parallel** is used. Set to NULL

to use all but one of the available cores, as detected by parallel::detectCores()

(using all cores tends to freeze up your computer).

quiet suppress messages. TRUE runs SimGeno and sequoia quietly, 'very' also sup-

presses other messages and the iteration counter when nCores=1 (there is no

iteration counter when nCores>1).

#### Details

The confidence probability is taken as the number of correct (matching) assignments, divided by all assignments made in the *observed* (inferred-from-simulated) pedigree. In contrast, the false negative & false positive assignment rates are proportions of the number of parents in the *true* (reference) pedigree. Each rate is calculated separatedly for dams & sires, and separately for each category (Genotyped/Dummy(fiable)/X (none)) of individual, parent and co-parent.

This function does not know which individuals in the actual Pedigree are genotyped, so the confidence probabilities need to be added to the Pedigree as shown in the example at the bottom.

A confidence of 1 means all assignments on simulated data were correct for that category-combination. It should be interpreted as (and perhaps modified to) > 1 - 1/N, where sample size N is given in the last column of the ConfProb and PedErrors dataframes in the output. The same applies for a false negative/positive rate of 0 (i.e. to be interpreted as < 1/N).

#### Value

A list, with elements:

ConfProb See below
PedErrors See below
Pedigree.reference

the pedigree from which data was simulated

LifeHistData

Pedigree.inferred

a list with for each iteration the inferred pedigree based on the simulated data

SimSNPd a list with for each iteration the IDs of the individuals simulated to have been

genotyped

PedComp. fwd Counts from the 'forward' PedCompare, from which PedErrors is calculated

RunParams a list with the call to EstConf, as well as the default parameter values for

SimGeno, and sequoia.

RunTime sequoia runtime per simulation in seconds, as measured by system.time()['elapsed'].

Dataframe ConfProb has 7 columns:

id.cat, dam.cat, sire.cat

Category of the focal individual, dam, and sire, in the pedigree inferred based

on the simulated data. Coded as G=genotyped, D=dummy, X=none

dam.conf Probability that the dam is correct, given the categories of the assigned dam and

sire (ignoring whether or not the sire is correct)

sire.conf as dam.conf, for the sire

pair.conf Probability that both dam and sire are correct, given their categories

N Number of individuals per category-combination, across all nSim iterations

Array PedErrors has three dimensions:

class

• FalseNeg(atives): could have been assigned but was not (individual + parent both genotyped or dummyfiable; Plonly in PedCompare).

• FalsePos(itives): no parent in reference pedigree, but one was assigned based on the simulated data (P2only)

• Mismatch: different parents between the pedigrees

cat

Category of individual + parent, as a two-letter code where the first letter indi-

cates the focal individual and the second the parent; G=Genotyped, D=Dummy,

T=Total

parent dam or sire

## **Assumptions**

Because the actual true pedigree is (typically) unknown, the provided reference pedigree is used as a stand-in and assumed to be the true pedigree, with unrelated founders. It is also assumed that the probability to be genotyped is equal for all parents; in each iteration, a new random set of parents (proportion set by ParMis) is mimicked to be non-genotyped. In addition, SNPs are assumed to segregate independently.

## Object size

The size in Kb of the returned list can become pretty big, as each of the inferred pedigrees is included. When running EstConf many times for a range of parameter values, it may be prudent to save the required summary statistics for each run rather than the full output.

#### See Also

SimGeno, sequoia, PedCompare.

```
data(Ped_HSg5, LH_HSg5, package="sequoia")
## Example A: parentage assignment only
conf.A <- EstConf(Pedigree = Ped_HSg5, LifeHistData = LH_HSg5,</pre>
   args.sim = list(nSnp = 100, SnpError = 5e-3, ParMis=c(0.2, 0.5)),
   args.seq = list(Module="par", Err=1e-3, Tassign=0.5), nSim = 3)
# parent-pair confidence, per category:
conf.A$ConfProb
# calculate (correct) assignment rates (ignores co-parent)
1 - apply(conf.A$PedErrors, c(1,3), sum, na.rm=TRUE)
## Example B: with sibship clustering, based on sequoia inferred pedigree
RealGenotypes <- SimGeno(Ped = Ped_HSg5, nSnp = 100,</pre>
                         ParMis=c(0.19, 0.53), SnpError = 6e-3)
SeqOUT <- sequoia(GenoM = RealGenotypes,</pre>
                  LifeHistData = LH_HSg5,
                  Err=5e-3, Module="ped",
                  quiet=TRUE, Plot=FALSE)
conf.B <- EstConf(Pedigree = SegOUT$Pedigree,</pre>
              LifeHistData = LH_HSg5,
               args.sim = list(nSnp = 100, SnpError = 5e-3,
                                ParMis=c(0.2, 0.5)),
              args.seq = list(Err=5e-3, Module="ped"),
              nSim = 2, nCores=2)
conf.B$ConfProb
Ped.withConf <- getAssignCat(Pedigree = SeqOUT$Pedigree,</pre>
                              SNPd = rownames(RealGenotypes))
Ped.withConf <- merge(Ped.withConf, conf.B$ConfProb, all.x=TRUE, sort=FALSE)</pre>
Ped.withConf <- Ped.withConf[, c("id","dam","sire", "dam.conf", "sire.conf",</pre>
                                  "id.cat", "dam.cat", "sire.cat")]
head(Ped.withConf[Ped.withConf$dam.cat=="G", ])
head(Ped.withConf[Ped.withConf$dam.cat=="D", ])
## P(actual FS | inferred as FS) etc.
PairL <- list()</pre>
for (i in 1:length(conf.A$Pedigree.inferred)) { # nSim
  cat(i, "\t")
  PairL[[i]] <- ComparePairs(conf.A$Pedigree.reference,</pre>
                              conf.A$Pedigree.inferred[[i]],
                              GenBack=1, patmat=TRUE, ExcludeDummies = TRUE,
                              Return="Counts")
```

FieldMums\_griffin 25

```
# P(actual relationship (Ped1) | inferred relationship (Ped2))
PairA <- plyr::laply(PairL, function(M) sweep(M, 2, colSums(M), "/"))
PairRel.prop <- apply(PairA, 2:3, mean, na.rm=TRUE) # mean across simulations
round(PairRel.prop, 2)
#' # or: P(inferred relationship | actual relationship)
PairA2 <- plyr::laply(PairL, function(M) sweep(M, 1, rowSums(M), "/"))</pre>
```

FieldMums\_griffin

Example field-observed mothers: griffins

# **Description**

Example field pedigree used in vignette for PedCompare example. Non-genotyped females have IDs 'BlueRed', 'YellowPink', etc.

# Usage

```
data(FieldMums_griffin)
```

## **Format**

A data frame with 144 rows and 2 variables (id, mum)

#### Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

# See Also

SeqOUT\_griffin for a sequoia run on simulated genotype data, Ped\_griffin for the 'true' pedigree.

FindFamilies

Assign Family IDs

# **Description**

Add a column with family IDs (FIDs) to a pedigree, with each number denoting a cluster of connected individuals.

## Usage

```
FindFamilies(Ped = NULL, SeqList = NULL, UseMaybeRel = FALSE)
```

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# **Arguments**

Ped	dataframe with columns id - parent1 - parent2; only the first 3 columns will be used.
SeqList	list as returned by sequoia. If 'Ped' is not provided, the element 'Pedigree' from this list will be used if present, and element 'Pedigreepar' otherwise.
UseMaybeRel	use SeqList\$MaybeRel, the dataframe with probable but non-assigned relatives, to assign additional family IDs?

# **Details**

This function repeatedly finds all ancestors and all descendants of each individual in turn, and ensures they all have the same Family ID. Not all connected individuals are related, e.g. all grand-parents of an individual will have the same FID, but will typically be unrelated.

When UseMaybeRel = TRUE, probable relatives are added to existing family clusters, or existing family clusters may be linked together. Currently no additional family clusters are created.

#### Value

A dataframe with the provided pedigree, with a column 'FID' added.

GenoConvert Convert Genotype Data

# Description

Convert genotype data in various formats to sequoia's 1-column-per-marker format or Colony's 2-columns-per-marker format.

# Usage

```
GenoConvert(
    InData = NULL,
    InFile = NULL,
    InFormat = "raw",
    OutFile = NA,
    OutFormat = "seq",
    Missing = c("-9", "??", "?", "NA", "NULL", c("0")[InFormat %in% c("col", "ped")]),
    sep = c(" ", "\t", ",", ";"),
    header = NA,
    IDcol = NA,
    FIDcol = NA,
    FIDsep = "__",
    dropcol = NA,
    quiet = FALSE
)
```

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#### **Arguments**

InData dataframe or matrix with genotypes to be converted. InFile character string with name of genotype file to be converted. One of 'single', 'double', 'col', 'ped', 'raw', or 'seq', see Details. InFormat OutFile character string with name of converted file. If NA, return matrix with genotypes in console (default); if NULL, write to 'GenoForSequoia.txt' in current working directory. OutFormat as InFormat; only 'seq' and 'col' are implemented. Missing vector with symbols interpreted as missing data. '0' is missing data for InFormats 'col' and 'ped' only. vector with field separator strings that will be tried on InFile. The OutFile sep separator uses the write. table default, i.e. one blank space. header a logical value indicating whether the file contains a header as its first line. If NA (default), set to TRUE for 'raw', and FALSE otherwise. **IDcol** number giving the column with individual IDs; 0 indicates the rownames (for InData only). If NA (default), set to 2 for InFormat 'raw' and 'ped', and otherwise to 1 for InFile and 0 (rownames) for InData, except when InData has a column labeled 'ID'. **FIDcol** column with the family IDs, if any are wished to be used. This is column 1 for InFormat 'raw' and 'seq', but those are by default not used. string used to paste FID and IID together into a composite-ID (value passed to **FIDsep** paste's collapse). This joining can be reversed using PedStripFID. dropcol columns to exclude from the output data, on top of IDcol and FIDcol (which become rownames). When NA, defaults to columns 3-6 for InFormat 'raw' and 'seq'. Can also be used to drop some SNPs, see example below on how to do this for the 2-columns-per-SNP input formats. quiet suppress messages and warnings.

#### **Details**

The first two arguments are interchangeable, and can be given unnamed. The first argument is assumed to be a file name if it is of class 'character' and length 1, and to be the genetic data if it is a matrix or dataframe.

#### Value

A genotype matrix in the specified output format. If 'OutFile' is specified, the matrix is written to this file and nothing is returned inside R. When converting to 0/1/2 format, 2 is the homozygote for the minor allele, and 0 the homozygote for the major allele.

# **Input formats**

The following formats can be specified by InFormat:

**seq** (sequoia) genotypes are coded as 0, 1, 2, missing as -9, in 1 column per marker. Column 1 contains IDs, there is no header row.

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**raw** (PLINK) genotypes are coded as 0, 1, 2, missing as NA, in 1 column per marker. The first 6 columns are descriptive (1:FID, 2:IID, 3 to 6 ignored), and there is a header row. This is produced by PLINK's option –recodeA

**ped** (PLINK) genotypes are coded as A, C, T, G, missing as 0, in 2 columns per marker. The first 6 columns are descriptive (1:FID, 2:IID, 3 to 6 ignored).

col (Colony) genotypes are coded as numeric values, missing as 0, in 2 columns per marker. Column 1 contains IDs.

single 1 column per marker, otherwise unspecified

double 2 columns per marker, otherwise unspecified

For each InFormat, its default values for Missing, header, IDcol, FIDcol, and dropcol can be overruled by specifying the corresponding input parameters.

## Error messages

Occasionally when reading in a file GenoConvert may give an error that 'rows have unequal length'. GenoConvert makes use of readLines and strsplit, which is much faster than read.table for large datafiles, but also more sensitive to unusual line endings, unusual end-of-file characters, or invisible characters (spaces or tabs) after the end of some lines. In these cases, try to read the data from file using read.table or read.csv, and then use GenoConvert on this dataframe or matrix, see example.

## Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

#### See Also

CheckGeno, SnpStats, LHConvert.

getAssignCat 29

```
dropcol = 1 + c(2*dropSNP-1, 2*dropSNP) )

# circumvent a 'rows have unequal length' error:
GenoTmp <- as.matrix(read.table("mydata.txt", header=TRUE, row.names=1))
GenoM <- GenoConvert(InData=GenoTmp, InFormat="single", IDcol=0)

## End(Not run)</pre>
```

getAssignCat

Assignability of Reference Pedigree

# **Description**

Identify which individuals are SNP genotyped, and which can potentially be substituted by a dummy individual ('Dummifiable').

# Usage

```
getAssignCat(Pedigree, SNPd, minSibSize = "1sib1GP")
```

# **Arguments**

Pedigree dataframe with columns id-dam-sire. Reference pedigree.

SNPd character vector with ids of genotyped individuals.

minSibSize minimum requirements to be considered 'dummifiable':

- '1sib': sibship of size 1, i.e. the non-genotyped individual has at least 1 genotyped offspring. If there is no sibship-grandparent this isn't really a sibship, but can be useful in some situations. Used by CalcoHLLR.
- '1sib1GP': sibship of size 1 with at least 1 genotyped grandparent. The minimum to be potentially assignable by sequoia.
- '2sib': at least 2 siblings, with or without grandparents. Used by PedCompare.

#### **Details**

It is assumed that all individuals in SNPd have been genotyped for a sufficient number of SNPs. To identify samples with a too-low call rate, use CheckGeno. To calculate the call rate for all samples, see the examples below.

Some parents indicated here as assignable may never be assigned by sequoia, for example parentoffspring pairs where it cannot be determined which is the older of the two, or grandparents that are indistinguishable from full avuncular (i.e. genetics inconclusive because the candidate has no parent assigned, and ageprior inconclusive). 30 getGenerations

#### Value

The Pedigree dataframe with 3 additional columns, id.cat, dam.cat and sire.cat, with coding similar to that used by PedCompare:

G Genotyped

D Dummy or 'dummifiable'

X Not genotyped and not dummifiable, or no parent in pedigree

# **Examples**

 ${\tt getGenerations}$ 

Count Generations

## **Description**

For each individual in a pedigree, count the number of generations since its most distant pedigree founder.

## Usage

```
getGenerations(Ped, StopIfInvalid = TRUE)
```

# **Arguments**

Ped dataframe, pedigree with the first three columns being id - dam - sire. Column

names are ignored, as are additional columns.

StopIfInvalid if a pedigree loop is detected, stop with an error (TRUE, default) or return the

Pedigree, to see where the problem(s) occur.

GetLLRAge 31

# Value

A vector with the generation number for each individual, starting at 0 for founders. NA indicates a pedigree loop where an individual is its own ancestor (or that the pedigree has >1000 generations). Returned invisibly to be a part of QC.

GetLLRAge

LLR-age from Ageprior Matrix

# Description

Get log10-likelihood ratios for a specific age difference from matrix AgePriorExtra.

# Usage

```
GetLLRAge(AgePriorExtra, agedif, patmat)
```

# Arguments

AgePriorExtra matrix in sequoia output

agedif vector with age differences, in whole numbers. Must occur in rownames of

AgePriorExtra.

patmat numeric vector; choose maternal (1), paternal (2) relatives, or for each relation-

ship the most-likely alternative (3).

# Value

A matrix with nrow equal to the length of agedif, and 7 columns: PO-FS-HS-GP-FA-HA-U.

32 GetMaybeRel

GetMaybeRel

Find Putative Relatives

## Description

Identify pairs of individuals likely to be related, but not assigned as such in the provided pedigree.

# Usage

```
GetMaybeRel(
  GenoM = NULL,
  SeqList = NULL,
 Pedigree = NULL,
 LifeHistData = NULL,
  AgePrior = NULL,
  ParSib = NULL,
 Module = "par"
 Complex = "full",
 Herm = "no",
 Err = 1e-04,
 ErrFlavour = "version2.0",
 MaxMismatch = NA,
 Tassign = 0.5,
  Tfilter = -2,
 MaxPairs = 7 * nrow(GenoM),
  quiet = FALSE
)
```

# **Arguments**

GenoM

numeric matrix with genotype data: One row per individual, and one column per SNP, coded as 0, 1, 2 or -9 (missing). See also GenoConvert.

SeqList

list with output from sequoia. SeqList\$Pedigree is used if present, and SeqList\$PedigreePar otherwise, and overrides the input parameter Pedigree. If 'Specs' is present, its elements override all input parameters with the same name. The list elements 'LifeHist', 'AgePriors', and 'ErrM' are also used if present, and similarly override the corresponding input parameters.

Pedigree

dataframe with id - dam - sire in columns 1-3. May include non-genotyped individuals, which will be treated as dummy individuals. When provided, all likelihoods (and thus all maybe-relatives) are conditional on this pedigree. Note: SeqList\$Pedigree or SeqList\$PedigreePar take precedent (for this function only).

LifeHistData

dataframe with 3 columns (optionally 5):

**ID** max. 30 characters long

**Sex** 1 = female, 2 = male, 3 = unknown, 4 = hermaphrodite, other numbers or NA = unknown

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**BirthYear** birth or hatching year, integer, with missing values as NA or any negative value.

**BY.min** minimum birth year, only used if BirthYear is missing **BY.max** maximum birth year, only used if BirthYear is missing

If the species has multiple generations per year, use an integer coding such that the candidate parents' 'Birth year' is at least one smaller than their putative offspring's. Column names are ignored, so ensure column order is ID - sex - birth year (- BY.min - BY.max). Individuals do not need to be in the same order as in 'GenoM', nor do all genotyped individuals need to be included.

AgePrior Agepriors matrix, as generated by MakeAgePrior and included in the sequoia

output. Affects which relationships are considered possible (only those where

P(A|R)/P(A) > 0).

ParSib either 'par' to check for putative parent-offspring pairs only, or 'sib' to check for

all types of first and second degree relatives. This argument will be deprecated,

please use Module.

Module type of relatives to check for. One of

par parent - offspring pairs

**ped** all first and second degree relatives

When 'par', all pairs are returned that are more likely parent-offspring than unrelated, potentially including pairs that are even more likely to be otherwise

related.

Complex Breeding system complexity. Either "full" (default), "simp" (simplified, no ex-

plicit consideration of inbred relationships), "mono" (monogamous).

Herm Hermaphrodites, either "no", "A" (distinguish between dam and sire role, default

if at least 1 individual with sex=4), or "B" (no distinction between dam and sire

role). Both of the latter deal with selfing.

Err estimated genotyping error rate, as a single number or 3x3 matrix. Details below.

The error rate is presumed constant across SNPs, and missingness is presumed

random with respect to actual genotype.

ErrFlavour function that takes Err (single number) as input, and returns a 3x3 matrix of

observed (columns) conditional on actual (rows) genotypes, or choose from inbuilt options 'version2.0', 'version1.3', or 'version1.1', referring to the sequoia

version in which they were the default. Ignored if Err is a matrix. See ErrToM.

MaxMismatch DEPRECATED AND IGNORED. Now calculated automatically using CalcMaxMismatch.

minimum LLR required for acceptance of proposed relationship, relative to next most likely relationship. Higher values result in more conservative assignments.

Must be zero or positive.

Tfilter threshold log10-likelihood ratio (LLR) between a proposed relationship versus

unrelated, to select candidate relatives. Typically a negative value, related to the fact that unconditional likelihoods are calculated during the filtering steps. More negative values may decrease non-assignment, but will increase computational

time.

Tassign

MaxPairs the maximum number of putative pairs to return.

quiet logical, suppress messages.

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## **Details**

When Module="par", the age difference of the putative pair is temporarily set to NA so that genetic parent-offspring pairs declared to be born in the same year may be discovered. When Module="ped", only relationships possible given the age difference, if known from the LifeHistData, are considered.

#### Value

A list with

MaybePar or MaybeRel

A dataframe with non-assigned likely relatives, with columns ID1 - ID2 - TopRel - LLR - OH - BirthYear1 - BirthYear2 - AgeDif - Sex1 - Sex2 - SNPdBoth

MaybeTrio A dataframe with non-assigned parent-parent-offspring trios, with columns id -

parent1 - parent2 - LLRparent1 - LLRparent2 - LLRpair - OHparent1 - OHpar-

ent2 - MEpair - SNPd.id.parent1 - SNPd.id.parent2

The following categories are used in column 'TopRel', indicating the most likely relationship category:

PO	Parent-Offspring
FS	Full Siblings
HS	Half Siblings
GP	GrandParent - grand-offspring
FA	Full Avuncular (aunt/uncle)
2nd	$2nd$ degree relatives, not enough information to distinguish between HS,GP and $F\!A$
Q	Unclear, but probably 1st, 2nd or 3rd degree relatives

#### See Also

sequoia to identify likely pairs of duplicate genotypes and for pedigree reconstruction; GetRelM to identify all pairs of relatives in a pedigree; CalcPairLL for the likelihoods underlying the LLR.

GetRelM 35

```
LifeHistData = LH_HSg5,

Err=0.0001, Complex = "full",

Module = "ped")

head(Maybe$MaybeRel)

# visualise results, turn dataframe into matrix first:

MaybeM <- GetRelM(Pairs=Maybe$MaybeRel)

PlotRelPairs(MaybeM)
```

GetRelM

Matrix with Pairwise Relationships

## **Description**

Generate a matrix or 3D array with all pairwise relationships from a pedigree or dataframe with pairs.

## Usage

```
GetRelM(
  Pedigree = NULL,
  Pairs = NULL,
  GenBack = 1,
  patmat = FALSE,
  Return = "Matrix"
)
```

# Arguments

Pedigree dataframe with columns id - dam - sire.

Pairs dataframe with columns ID1 - ID2 - Rel.

GenBack number of generations back to consider; 1 returns parent-offspring and sibling

relationships, 2 also returns grand-parental, avuncular and first cousins.

patmat logical, distinguish between paternal versus maternal relative pairs? For avun-

cular pairs, the distinction is never made.

Return 'Matrix' or 'Array'. The former returns an N x N matrix with the closest rela-

tionship between each pair, the latter an N x N x R array with for each of the R considered relationships whether it exists between the pair (1) or not (0). See

Details below.

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#### **Details**

Double relationships are ignored when Return='Matrix', but not when Return='Array'. For example, when A and B are both mother-offspring and paternal siblings (A mated with her father to produce B), only the mother-offspring relationship will be indicated when Return='Matrix'.

Note that full siblings are the exception to this rule: in the Array they will be indicated as 'FS' only, and not as 'MHS' or 'PHS'. Similarly, full avuncular pairs are not indicated as 'HA'.

When Pairs is provided, GenBack and patmat are ignored, and no check is performed if the abbreviations are compatible with other functions.

#### Value

If Return='Matrix', an N x N square matrix, with N equal to the number of rows in Pedigree (after running PedPolish) or the number of unique individuals in Pairs. If Return='Array', an N x N x R array is returned, with R, the number of different relationships, determined by GenBack and patmat.

The following abbreviations are used within the returned Matrix, or as names of the 3rd dimension in the Array:

S	Self
М	Mother
Р	Father

MP Mother or Father (patmat=FALSE)

O OffspringFS Full sibling

MHS Maternal half-sibling
PHS Paternal half-sibling

XHS other half-sibling (hermaphrodites)
HS half-sibling (patmat=FALSE)
MGM Maternal grandmother

MGM Maternal grandmother
MGF Maternal grandfather
PGM Paternal grandmother
PGF Paternal grandfather

GP Grandparent (patmat=FALSE)

GO Grand-offspring

FA Full avuncular; maternal or paternal aunt or uncle

HA Half avuncular

FN Full nephew/niece

HN Half nephew/niece

FC1 Full first cousin

DFC1 Double full first cousin

U Unrelated (or otherwise related)

Inherit 37

#### See Also

ComparePairs for comparing pairwise relationships between two pedigrees; PlotRelPairs.

# Examples

```
data(Ped_griffin)
Rel.griffin <- GetRelM(Ped_griffin, patmat=TRUE, GenBack=2)
table(c(Rel.griffin))
# turning matrix into vector first makes table() much faster
PlotRelPairs(Rel.griffin)</pre>
```

Inherit

Inheritance patterns

#### **Description**

Inheritance patterns used by SimGeno for non-autosomal SNPs, identical to those in Inherit.xlsx

#### Usage

```
data(Inherit)
```

## **Format**

An array with the following dimensions:

- d1 type: autosomal, x-chromosome, y-chromosome, or mtDNA
- d2 offspring sex: female, male, or unknown
- **d3** offspring genotype: aa (0), aA (1), Aa (1), or AA (2)
- d4 mother genotype
- d5 father genotype

# Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

### See Also

SimGeno

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LHConvert

Extract Sex and Birth Year from PLINK File

#### Description

Convert the first six columns of a PLINK .fam, .ped or .raw file into a three-column lifehistory file for sequoia. Optionally FID and IID are combined.

#### Usage

```
LHConvert(
  PlinkFile = NULL,
  UseFID = FALSE,
  SwapSex = TRUE,
  FIDsep = "__",
  LifeHistData = NULL
)
```

# Arguments

PlinkFile character string with name of genotype file to be converted.

UseFID use the family ID column. The resulting ids (rownames of GenoM) will be in

the form FID\_\_IID.

SwapSex change the coding from PLINK default (1=male, 2=female) to sequoia default

(1=female, 2=male); any other numbers are set to NA.

FIDsep characters inbetween FID and IID in composite-ID. By default a double under-

score is used, to avoid problems when some IIDs contain an underscore. Only

used when UseFID=TRUE.

LifeHistData dataframe with additional sex and birth year info. In case of conflicts, LifeHist-

Data takes priority, with a warning. If UseFID=TRUE, IDs in LifeHistData are

assumed to be already as FID\_\_IID.

# Details

The first 6 columns of PLINK .fam, .ped and .raw files are by default FID - IID - father ID (ignored) - mother ID (ignored) - sex - phenotype.

### Value

A dataframe with id, sex and birth year, which can be used as input for sequoia.

#### See Also

GenoConvert, PedStripFID to reverse UseFID.

LH\_griffin 39

# **Examples**

LH\_griffin

Example lifehistory data: griffins

# Description

Example lifehistory data for griffin pedigree

# Usage

```
data(LH_griffin)
```

# **Format**

A data frame with 200 rows and 3 variables (ID, Sex, BirthYear)

#### Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

# See Also

```
Ped_griffin, SeqOUT_griffin
```

LH\_HSg5

Example life history file

# Description

This is the lifehistory file associated with Ped\_HSg5, which is **Pedigree II** in the paper.

# Usage

```
data(LH_HSg5)
```

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#### **Format**

A data frame with 1000 rows and 3 variables:

**ID** Female IDs start with 'a', males with 'b'; the next 2 numbers give the generation number (00 – 05), the last 3 numbers the individual ID number (runs continuously across all generations)

```
Sex 1 = \text{female}, 2 = \text{male}
```

BirthYear from 2000 (generation 0, founders) to 2005

# Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

#### References

Huisman, J. (2017) Pedigree reconstruction from SNP data: Parentage assignment, sibship clustering, and beyond. Molecular Ecology Resources 17:1009–1024.

#### See Also

Ped\_HSg5 sequoia

MakeAgePrior

Age Priors

# **Description**

Estimate probability ratios P(R|A)/P(R) for age differences A and five categories of parent-offspring and sibling relationships R.

#### Usage

```
MakeAgePrior(
  Pedigree = NULL,
  LifeHistData = NULL,
  MaxAgeParent = NULL,
  Discrete = NULL,
  Flatten = NULL,
  lambdaNW = -log(0.5)/100,
  Smooth = TRUE,
  Plot = TRUE,
  Return = "LR",
  quiet = FALSE
)
```

MakeAgePrior 41

#### **Arguments**

Pedigree dataframe wit

dataframe with id - dam - sire in columns 1-3, and optional column with birth years. Other columns are ignored.

LifeHistData

dataframe with 3 or 5 columns: id - sex (not used) - birth year (- BY.min - BY.max), with unknown birth years coded as negative numbers or NA. Column names are ignored, so the column order is important. "Birth year" may be in any arbitrary discrete time unit relevant to the species (day, month, decade), as long as parents are never born in the same time unit as their offspring. It may include individuals not in the pedigree, and not all individuals in the pedigree need to be in LifeHistData.

MaxAgeParent

maximum age of a parent, a single number (max across dams and sires) or a vector of length two (dams, sires). If NULL, it will be estimated from the pedigree. See details below.

Discrete

discrete generations? By default (NULL), discrete generations are assumed if all parent-offspring pairs have an age difference of 1, and all siblings an age difference of 0, and there are at least 20 pairs of each category (mother, father, maternal sibling, paternal sibling). Otherwise, overlapping generations are presumed. When Discrete=TRUE (explicitly or deduced), Smooth and Flatten are always automatically set to FALSE. Use Discrete=FALSE to enforce (potential for) overlapping generations.

Flatten

logical. To deal with small sample sizes for some or all relationships, calculate weighed average between the observed age difference distribution among relatives and a flat (0/1) distribution. When Flatten=NULL (the default) automatically set to TRUE when there are fewer than 20 parents with known age of either sex assigned, or fewer than 20 maternal or paternal siblings with known age difference. Also advisable if the sampled relative pairs with known age difference are non-typical of the pedigree as a whole.

1ambdaNW

control weighing factors when Flatten=TRUE. Weights are calculated as W(R)=1-exp(-lambdaNW\*N(R)), where N(R) is the number of pairs with relationship R for which the age difference is known. Large values (>0.2) put strong emphasis on the pedigree, small values (<0.0001) cause the pedigree to be ignored. Default results in W=0.5 for N=100.

Smooth

smooth the tails of and any dips in the distribution? Sets dips (<10% of average of neighbouring ages) to the average of the neighbouring ages, sets the age after the end (oldest observed age) to LR(end)/2, and assigns a small value (0.001) to the ages before the front (youngest observed age) and after the new end. Peaks are not smoothed out, as these are less likely to cause problems than dips, and are more likely to be genuine characteristics of the species. Is set to FALSE when generations do not overlap (Discrete=TRUE).

Plot plot a heatmap of the results?

Return return only a matrix with the likelihood-ratio P(A|R)/P(A) ("LR") or a list

including also various intermediate statistics ("all")?

quiet suppress messages.

#### **Details**

 $\alpha_{A,R}$  is the ratio between the observed counts of pairs with age difference A and relationship R  $(N_{A,R})$ , and the expected counts if age and relationship were independent  $(N_{...} * p_A * p_R)$ .

During pedigree reconstruction,  $\alpha_{A,R}$  are multiplied by the genetic-only P(R|G) to obtain a probability that the pair are relatives of type R conditional on both their age difference and their genotypes.

The age-difference prior is used for pairs of genotyped individuals, as well as for dummy individuals. This assumes that the propensity for a pair with a given age difference to both be sampled does not depend on their relationship, so that the ratio P(A|R)/P(A) does not differ between sampled and unsampled pairs.

For further details, see the vignette.

#### Value

A matrix with the probability ratio of the age difference between two individuals conditional on them being a certain type of relative (P(A|R)) versus being a random draw from the sample (P(A)). Assuming conditional independence, this equals the probability ratio of being a certain type of relative conditional on the age difference, versus being a random draw.

The matrix has one row per age difference (0 - nAgeClasses) and five columns, one for each relationship type, with abbreviations:

М	Mothers
Р	Fathers
FS	Full siblings
MS	Maternal half-siblings
PS	Paternal half-siblings

When Return='all', a list is returned with the following elements:

and Flatten.

BirthYearRange	vector length 2
MaxAgeParent	vector length 2, see details
tblA.R	matrix with the counts per age difference (rows) / relationship (columns) combination, plus a column 'X' with age differences across all pairs of individuals
PA.R	Proportions, i.e. tblA.R divided by its colSums, with full-sibling correction applied if necessary (see vignette).
LR.RU.A.raw	Proportions PA.R standardised by global age difference distribution (column 'X'); LR.RU.A prior to flattening and smoothing
Weights	vector length 4, the weights used to flatten the distributions
LR.RU.A	the ageprior, flattend and/or smoothed
Specs.AP	the names of the input Pedigree and LifeHistData (or NULL), lambdaNW, and the 'effective' settings (i.e. after any automatic update) of Discrete, Smooth,

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#### **CAUTION**

The small sample correction with Smooth and/or Flatten prevents errors in one dataset, but may introduce errors in another; a single solution that fits to the wide variety of life histories and datasets is impossible. Please do inspect the matrix, e.g. with PlotAgePrior, and adjust the input parameters and/or the output matrix as necessary.

#### Single cohort

When all individuals in LifeHistData have the same birth year, it is assumed that Discrete=TRUE and MaxAgeParent=1. Consequently, it is assumed there are no avuncular pairs present in the sample; cousins are considered as alternative. To enforce overlapping generations, and thereby the consideration of full- and half- avuncular relationships, set MaxAgeParent to some value greater than 1.

When no birth year information is given at all, a single cohort is assumed, and the same rules apply.

#### Other time units

"Birth year" may be in any arbitrary time unit relevant to the species (day, month, decade), as long as parents are always born before their putative offspring, and never in the same time unit (e.g. parent's BirthYear=1 (or 2001) and offspring BirthYear=5 (or 2005)). Negative numbers and NA's are interpreted as unknown, and fractional numbers are not allowed.

#### MaxAgeParent

The maximum parental age for each sex equals the maximum of:

- the maximum age of parents in Pedigree,
- the input parameter MaxAgeParent,
- the maximum range of birth years in LifeHistData (including BY.min and BY.max). Only used if both of the previous are NA, or if there are fewer than 20 parents of either sex assigned.
- 1, if Discrete=TRUE or the previous three are all NA

If the age distribution of assigned parents does not capture the maximum possible age of parents, it is advised to specify MaxAgeParent for one or both sexes. Not doing so may hinder subsequent assignment of both dummy parents and grandparents.

@section grandparents & avuncular The agepriors for grand-parental and avuncular pairs is calculated from these by sequoia, and included in its output as 'AgePriorExtra'.

#### See Also

sequoia and its argument args.AP, PlotAgePrior for visualisation. The age vignette gives further details, mathematical justification, and some examples.

#### **Examples**

```
# without pedigree or lifehistdata:
MakeAgePrior()
MakeAgePrior(MaxAgeParent = c(2,3))
```

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```
MakeAgePrior(Discrete=TRUE)
# single cohort:
MakeAgePrior(LifeHistData = data.frame(ID = letters[1:5], Sex=3,
  BirthYear=1984))
# overlapping generations:
data(Ped_griffin, SeqOUT_griffin, package="sequoia")
# without pedigree: MaxAgeParent = max age difference between any pair +1
MakeAgePrior(LifeHistData = SeqOUT_griffin$LifeHist)
# with pedigree:
MakeAgePrior(Pedigree=Ped_griffin,
             LifeHistData=SeqOUT_griffin$LifeHist,
             Smooth=FALSE, Flatten=FALSE)
# with small-sample correction:
MakeAgePrior(Pedigree=Ped_griffin,
             {\tt LifeHistData=SeqOUT\_griffin\$LifeHist},
             Smooth=TRUE, Flatten=TRUE)
```

MkGenoErrors

Simulate Genotyping Errors

#### **Description**

Generate errors and missing values in a (simulated) genotype matrix.

#### Usage

# **Arguments**

SGeno

matrix with genotype data in Sequoia's format: 1 row per individual, 1 column per SNP, and genotypes coded as 0/1/2.

CallRate

either a single number for the mean call rate (genotyping success), OR a vector with the call rate at each SNP, OR a named vector with the call rate for each individual. In the third case, ParMis is ignored, and individuals in the pedigree (as id or parent) not included in this vector are presumed non-genotyped.

SnpError

mean per-locus genotyping error rate across SNPs, and a beta-distribution will be used to simulate the number of missing cases per SNP, OR a vector with the genotyping error for each SNP.

ErrorFM

function taking the error rate (scalar) as argument and returning a 4x4 or 3x3 matrix with probabilities that actual genotype i (rows) is observed as genotype j (columns).

Error.shape

first shape parameter (alpha) of beta-distribution of per-SNP error rates. A higher value results in a flatter distribution.

CallRate.shape

serior.shape, for per-SNP call rates.

#### Value

The input genotype matrix, with some genotypes replaced, and some set to missing (-9).

## **Examples**

PedCompare

Compare Two Pedigrees

# Description

Compare an inferred pedigree (Ped2) to a previous or simulated pedigree (Ped1), including comparison of sibship clusters and sibship grandparents.

## Usage

```
PedCompare(
   Ped1 = NULL,
   Ped2 = NULL,
   DumPrefix = c("F0", "M0"),
   SNPd = NULL,
   Symmetrical = TRUE,
   minSibSize = "2sib",
   Plot = TRUE
)
```

#### **Arguments**

Ped1 first (e.g. original) pedigree, dataframe with columns id-dam-sire; only the first

3 columns will be used.

Ped2 second pedigree, e.g. newly inferred SeqOUT\$Pedigree or SeqOUT\$PedigreePar,

with columns id-dam-sire.

DumPrefix character vector with the prefixes identifying dummy individuals in Ped2. Use

'F0' ('M0') to avoid matching to regular individuals with IDs starting with 'F'

('M'), provided Ped2 has fewer than 999 dummy females (males).

SNPd character vector with IDs of genotyped individuals. If NULL, defaults to the IDs

occurring in both Ped1 and Ped2 and not starting with any of the prefixes in

DumPrefix.

Symmetrical when determining the category of individuals (Genotyped/Dummy/X), use the

'highest' category across the two pedigrees (TRUE, default) or only consider

Ped1 (Symmetrical = FALSE).

minSibSize minimum requirements to be considered 'dummifiable', passed to getAssignCat:

• '1sib': sibship of size 1, with or without grandparents. The latter aren't

really a sibship, but can be useful in some situations.

• '1sib1GP': sibship of size 1 with at least 1 grandparent

• '2sib': at least 2 siblings, with or without grandparents (default)

Plot show square Venn diagrams of counts?

#### **Details**

The comparison is divided into different classes of 'assignable' parents (getAssignCat). This includes cases where the focal individual and parent according to Ped1 are both Genotyped (G-G), as well as cases where the non-genotyped parent according to Ped1 can be lined up with a sibship Dummy parent in Ped2 (G-D), or where the non-genotyped focal individual in Ped1 can be matched to a dummy individual in Ped2 (D-G and D-D). If SNPd is NULL (the default), and DumPrefix is set to NULL, the intersect between the IDs in Pedigrees 1 and 2 is taken as the vector of genotyped individuals.

#### Value

A list with

Counts A 7 x 5 x 2 named numeric array with the number of matches and mismatches,

see below

Counts.detail a large numeric array with number of matches and mismatches, with more detail

for all possible combination of categories

MergedPed A dataframe with side-by-side comparison of the two pedigrees

ConsensusPed A consensus pedigree, with Pedigree 2 taking priority over Pedigree 1

DummyMatch Dataframe with all dummy IDs in Pedigree 2 (id.2), and the best-matching indi-

vidual in Pedigree 1 (id.1). Also includes the class of the dam & sire, as well as

counts of offspring per outcome class (off.Match, off.Mismatch, etc.)

Mismatch A subset of MergedPed with mismatches between Ped1 and Ped2, as defined

below

Ped1only as Mismatches, with parents in Ped1 that were not assigned in Ped2
Ped2only as Mismatches, with parents in Ped2 that were missing in Ped1

'MergedPed', 'Mismatch', 'Ped1only' and 'Ped2only' provide the following columns:

id All ids in both Pedigree 1 and 2. For dummy individuals, this is the id in pedi-

gree 2

dam.1, sire.1 parents in Pedigree 1 dam.2, sire.2 parents in Pedigree 2

id.r, dam.r, sire.r

The *real* id of dummy individuals or parents in Pedigree 2, i.e. the best-matching non-genotyped individual in Pedigree 1, or "nomatch". If a sibship in Pedigree 1 is divided over 2 sibships in Pedigree 2, the smaller one will be denoted as "nomatch"

id.dam.cat, id.sire.cat

the category of the individual (first letter) and *highest category* of the dam (sire) in Pedigree 1 or 2: G=Genotyped, D=(potential) dummy, X=none. Individual, one-letter categories are generated by getAssignCat. Using the 'best' category from both pedigrees makes comparison between two inferred pedigrees symmetrical and more intuitive.

dam.class, sire.class

classification of dam and sire: Match, Mismatch, Plonly, P2only, or '\_' when no parent is assigned in either pedigree

The first dimension of Counts denotes the following categories:

GG Genotyped individual, assigned a genotyped parent in either pedigree

Genotyped individual, assigned a dummy parent, or at least 1 genotyped sibling

or a genotyped grandparent in Pedigree 1)

GT Genotyped individual, total

DG Dummy individual, assigned a genotyped parent (i.e., grandparent of the sibship

in Pedigree 2)

DD Dummy individual, assigned a dummy parent (i.e., avuncular relationship be-

tween sibships in Pedigree 2)

DT Dummy total

TT Total total, includes all genotyped individuals, plus non-genotyped individuals

in Pedigree 1, plus non-replaced dummy individuals (see below) in Pedigree 2

The second dimension of Counts gives the outcomes:

Total The total number of individuals with a parent assigned in either or both pedigrees

Match The same parent is assigned in both pedigrees (non-missing). For dummy parents, it is considered a match if the inferred sibship which contains the most

ents, it is considered a match if the inferred sibship which contains the most offspring of a non-genotyped parent, consists for more than half of this individ-

ual's offspring.

Mismatch

Different parents assigned in the two pedigrees. When a sibship according to Pedigree 1 is split over two sibships in Pedigree 2, the smaller fraction is included in the count here.

Plonly

Parent in Pedigree 1 but not 2; includes non-assignable parents (e.g. not genotyped and no genotyped offspring).

Parent in Pedigree 2 but not 1.

The third dimension Counts separates between maternal and paternal assignments, where e.g. paternal 'DT' is the assignment of fathers to both maternal and paternal sibships (i.e., to dummies of both sexes).

In 'ConsensusPed', the priority used is parent.r (if not "nomatch") > parent.2 > parent.1. The columns 'id.cat', dam.cat' and 'sire.cat' have two additional levels compared to 'MergedPed':

G	Genotyped
D	Dummy individual (in Pedigree 2)
R	Dummy individual in pedigree 2 replaced by best matching non-genotyped individual in pedigree 1
U	Ungenotyped, Unconfirmed (parent in Pedigree 1, with no dummy match in Pedigree 2)
Χ	No parent in either pedigree

# Assignable

P2only

Note that 'assignable' may be overly optimistic. Some parents from Ped1 indicated as assignable may never be assigned by sequoia, for example parent-offspring pairs where it cannot be determined which is the older of the two, or grandparents that are indistinguishable from full avuncular (i.e. genetics inconclusive because the candidate has no parent assigned, and ageprior inconclusive).

#### **Dummifiable**

Considered as potential dummy individuals are all non-genotyped individuals in Pedigree 1 who have, according to either pedigree, at least 2 genotyped offspring, or at least one genotyped offspring and a genotyped parent.

#### Mismatches

Perhaps unexpectedly, cases where all siblings are correct but a dummy parent rather than the genotyped Ped1-parent are assigned, are classified as a mismatch (for each of the siblings). These are typically due to a too low assumed genotyping error rate, a wrong parental birth year, or some other issue that requires user inspection. To identify these cases, ComparePairs may be of help.

# Genotyped 'mystery samples'

If Pedigree 2 includes samples for which the ID is unknown, the behaviour of PedCompare depends on whether the temporary IDs for these samples are included in SNPd. If they are included, matching (actual) IDs in Pedigree 1 will be flagged as mismatches (because the IDs differ). If they are not included in SNPd, or SNPd is not explicitly provided, matches are accepted, as the situation is indistinguishable from comparing dummy parents across pedigrees.

This is of course all conditional on relatives of the mystery sample being assigned in Pedigree 2.

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#### Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

#### See Also

ComparePairs for comparison of all pairwise relationships in 2 pedigrees, EstConf for repeated simulate-reconstruct-compare, sequoia for the main pedigree reconstruction function, getAssignCat for all parents in the reference pedigree that could have been assigned.

# **Examples**

```
data(Ped_HSg5, SimGeno_example, LH_HSg5, package="sequoia")
SeqOUT <- sequoia(GenoM = SimGeno_example, LifeHistData = LH_HSg5,</pre>
 Err=0.0001, quiet=TRUE, Plot=FALSE)
# (Performance is better when using Err=0.001, but this makes for a more
# interesting example)
compare <- PedCompare(Ped1=Ped_HSg5, Ped2=SeqOUT$Pedigree)</pre>
compare$Counts["TT",,] # totals only
compare$Counts[,,"dam"] # dams only
# 2 mismatch & 3+1 non-assigned, due to simulated genotyping errors
# inspect 'assignable but non-assigned in Ped2', id + dam both genotyped:
compare$P1only[compare$P1only$id.dam.cat=="GG", ]
# further inspection:
compare$MergedPed[which(compare$MergedPed$dam.1=="a00013"), ]
# overview of all non-genotyped -- dummy matches
head(compare$DummyMatch)
# success of paternity assignment, if genotyped mother correctly assigned
dimnames(compare$Counts.detail)
compare$Counts.detail["G","G",,"Match",]
```

PedPolish

Fix Pedigree

# Description

Ensure all parents & all genotyped individuals are included, remove duplicates, rename columns, and replace 0 by NA or v.v..

#### Usage

```
PedPolish(
  Pedigree,
  gID = NULL,
```

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```
ZeroToNA = TRUE,
NAToZero = FALSE,
DropNonSNPd = TRUE,
FillParents = FALSE,
NullOK = FALSE,
LoopCheck = TRUE,
StopIfInvalid = TRUE)
```

#### **Arguments**

Pedigree dataframe where the first 3 columns are id, dam, sire.

gID character vector with ids of genotyped individuals (rownames of genotype ma-

trix).

ZeroToNA logical, replace 0's for missing values by NA's (defaults to TRUE).

NAToZero logical, replace NA's for missing values by 0's. If TRUE, ZeroToNA is automati-

cally set to FALSE.

DropNonSNPd logical, remove any non-genotyped individuals (but keep non-genotyped par-

ents), & sort pedigree in order of gID.

FillParents logical, for individuals with only 1 parent assigned, set the other parent to a

dummy (without assigning siblings or grandparents). Makes the pedigree compatible with R packages and software that requires individuals to have either 2

or 0 parents, such as kinship.

NullOK logical, is it OK for Ped to be NULL? Then NULL will be returned.

LoopCheck logical, check for invalid pedigree loops by calling getGenerations.

StopIfInvalid if a pedigree loop is detected, stop with an error (TRUE, default).

### **Details**

Recognized column names are any that contain:

```
dam "dam", "mother", "mot", "mom", "mum", "mat"
sire "sire", "father", "fat", "dad", "pat"
```

sequoia requires the column order id - dam - sire; columns 2 and 3 are swapped if necessary.

#### **Examples**

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```
Ped.k <- with(Ped.fix, kinship2::pedigree(id, dadid, momid, sex, missid=0))
## End(Not run)</pre>
```

PedStripFID

Back-transform IDs

# Description

Reverse the joining of FID and IID in GenoConvert and LHConvert

# Usage

```
PedStripFID(Ped, FIDsep = "__")
```

# **Arguments**

Ped pedigree as returned by sequoia (e.g. SeqOUT\$Pedigree).

FIDsep characters inbetween FID and IID in composite-ID.

# **Details**

Note that the family IDs are the ones provided, and not automatically updated. New, numeric ones can be obtained with FindFamilies.

# Value

A pedigree with 6 columns

FID family ID of focal individual (offspring).

id within-family of focal individual dam.FID original family ID of assigned dam

dam within-family of dam

sire.FID original family ID of assigned sire

sire within-family of sire

52 Ped\_HSg5

Ped\_griffin

Example pedigree: griffins

# Description

Example pedigree used in the ageprior vignette, with overlapping generations.

#### Usage

```
data(Ped_griffin)
```

#### **Format**

A data frame with 200 rows and 4 variables (id, dam, sire, birthyear)

#### Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

#### See Also

LH\_griffin; SeqOUT\_griffin for a sequoia run on simulated genotype data based on this pedigree; Ped\_HSg5 for another pedigree, sequoia

Ped\_HSg5

Example pedigree

# **Description**

This is **Pedigree II** in the paper, with discrete generations and considerable inbreeding

# Usage

```
data(Ped_HSg5)
```

# **Format**

A data frame with 1000 rows and 3 variables (id, dam, sire)

#### Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

## References

Huisman, J. (2017) Pedigree reconstruction from SNP data: Parentage assignment, sibship clustering, and beyond. Molecular Ecology Resources 17:1009–1024.

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## See Also

LH\_HSg5 SimGeno\_example sequoia

PlotAgePrior

Plot Age Priors

# **Description**

Visualise the age-difference based prior probability ratios as a heatmap.

# Usage

```
PlotAgePrior(AP = NULL, legend = TRUE)
```

## **Arguments**

AΡ

matrix with age priors (P(A|R)/P(A)) with age differences in rows and relationships in columns; by default M: maternal parent (mother), P: paternal parent (father), FS: full siblings, MS: maternal siblings (full + half), PS: paternal siblings.

legend

if TRUE, a new plotting window is started and layout is used to plot a legend next to the main plot. Set to FALSE if you want to add it as panel to an existing plot (e.g. with par(mfcol=c(2,2))).

#### Value

A heatmap.

# See Also

MakeAgePrior, SummarySeq.

# **Examples**

```
data(SeqOUT_griffin, package="sequoia")
PlotAgePrior(SeqOUT_griffin$AgePriorExtra)
PlotAgePrior(SeqOUT_griffin$AgePriorExtra)
```

54 PlotPairLL

PlotPairLL

Plot Pair Log10-Likelihoods

# **Description**

Colour-coded scatter plots of e.g. LLR(PO/U) against LLR(FS/U), for various relationship combinations.

# Usage

```
PlotPairLL(
   PairLL,
   combo = list(c("FS", "PO"), c("HS", "FS"), c("GP", "HS"), c("FA", "HS")),
   nrows = NULL,
   ncols = NULL,
   bgcol = TRUE,
   Tassign = 0.5,
   Tfilter = -2
)
```

# **Arguments**

PairLL	dataframe, output from CalcPairLL.
combo	list with length-2 character vectors, specifying which likelihoods to plot against each other. Choose from 'PO', 'FS', 'HS', 'GP', 'FA', and 'HA'. The first one gets plotted on the x-axis, the second on the y-axis. Subsequent figures will be drawn row-wise.
nrows	$number of rows in the figure layout. If \verb+NULL+, set to \verb+ceiling(length(combo)/ncols).$
ncols	number of columns in the figure layout. If both nrows and ncols are NULL, ncols is set to ceiling(sqrt(length(combo))), and nrows will be equal to ncols or one less.
bgcol	logical, colour the upper and lower triangle background of each figure to match the specified relationship combo.
Tassign	assignment threshold, shown as grey square in bottom-left corner and a band along the diagonal.
Tfilter	filter threshold, shown as dark grey square in bottom-left.

#### **Details**

The colour of each point is determined by columns focal (outer circle) and TopRel (inner filling) of PairLL.

Impossible relationships (LL > 0 in PairLL) are shown as -Inf on the axes, if any are present.

# See Also

CalcPairLL.

PlotPedComp 55

#### **Examples**

PlotPedComp

Visualise PedCompare Output

# **Description**

square Venn diagrams with PedCompare Counts.

# Usage

```
PlotPedComp(Counts, sameSize = FALSE)
```

## **Arguments**

Counts a 7x5x2 array with counts of matches and mismatches per category (genotyped

vs dummy), as returned by PedCompare.

sameSize logical, make all per-category Venn diagrams the same size TRUE, or make their

size proportional to the counts (FALSE, the default). If TRUE, a warning is printed

at the bottom.

#### See Also

PedCompare

#### **Examples**

56 PlotRelPairs

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Plot Pairwise Relationships

# Description

Plot pairwise 1st and 2nd degree relationships between individuals, similar to Colony's dyad plot.

### Usage

```
PlotRelPairs(
  RelM = NULL,
  subset.x = NULL,
  subset.y = NULL,
  drop.U = TRUE,
  pch.symbols = FALSE,
  cex.axis = 0.7,
  mar = c(5, 5, 1, 8)
)
```

## **Arguments**

RelM	square matrix with relationships between all pairs of individuals, as generated by GetRelM. Row and column names should be individual IDs.
subset.x	vector with IDs to show on the x-axis; the y-axis will include all siblings, parents and grandparents of these individuals.
subset.y	vector with IDs to show on the y-axis; the x-axis will include all siblings, off-spring and grandoffspring of these individuals. Specify either subset.x or subset.y (or neither), not both.
drop.U	logical: omit individuals without relatives from the plot, and omit individuals without parents from the x-axis. Ignored if subset.x or subset.y is specified.
pch.symbols	logical: use different symbols for the different relationships (TRUE) or only colours in a heatmap-like fashion (FALSE). Question marks in the plot indicate that one or more of the symbols are not supported on your machine.
cex.axis	the magnification to be used for axis annotation. Decrease this value if $R$ is dropping axis labels to prevent them from overlapping.
mar	A numerical vector of the form c(bottom, left, top, right) which gives the number of lines of margin to be specified on the four sides of the plot.

# **Details**

Parents are shown above the diagonal (y-axis is parent of x-axis), siblings below the diagonal. If present, grandparents and full aunts/uncles are also shown above the diagonal. Individuals are sorted by dam ID and sire ID so that siblings are grouped together, and then by generation (getGenerations) so that later generations are closer to the origin.

SeqOUT\_griffin 57

If RelM is based on a dataframe with pairs rather than a pedigree, parents and grandparents are similarly only displayed above the diagonal, but the order of individuals is arbitrary and the ID on the x-axis is as likely to be the grandparent of the one on the y-axis as vice versa. Second degree relatives of unknown classification ('2nd', may be HS, GP or FA) are only shown below the diagonal. The switch between pedigree-based versus pairs-based is made on whether parent-offspring pairs are coded as 'M','P', 'MP', 'O' (unidirectional, from pedigree) or as 'PO' (bidirectional, from pairs).

Note that half-avuncular and (double) full cousin pairs are ignored.

#### Value

The subsetted, rearranged RelM is returned invisible.

The numbers of unique pairs of each relationship type are given in the figure legend. The number of 'self' pairs refers to the number of individuals on the x-axis, not all of whom may occur on the y-axis when drop. U=TRUE or a subset is specified.

#### See Also

GetRelM; SummarySeq for individual-wise graphical pedigree summaries.

## **Examples**

SeqOUT\_griffin

Example sequoia output (griffins)

#### **Description**

Example output of a sequoia run including sibship clustering, based on the griffin pedigree.

## Usage

```
data(SeqOUT_griffin)
```

#### **Format**

```
a list, see sequoia
```

#### Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

#### See Also

```
Ped_griffin, sequoia
```

# **Examples**

sequoia

Pedigree Reconstruction

# Description

Perform pedigree reconstruction based on SNP data, including parentage assignment and sibship clustering.

# Usage

```
sequoia(
   GenoM = NULL,
   LifeHistData = NULL,
   SeqList = NULL,
   Module = "ped",
   MaxSibIter = 42,
   Err = 1e-04,
   ErrFlavour = "version2.0",
   MaxMismatch = NA,
   Tfilter = -2,
   Tassign = 0.5,
   MaxSibshipSize = 100,
```

```
DummyPrefix = c("F", "M"),
  Complex = "full",
 Herm = "no",
 UseAge = "yes"
  args.AP = list(Flatten = NULL, Smooth = TRUE),
 FindMaybeRel = FALSE,
 CalcLLR = TRUE,
  quiet = FALSE,
 Plot = NULL
)
```

## **Arguments**

GenoM

numeric matrix with genotype data: One row per individual, and one column per SNP, coded as 0, 1, 2 or -9 (missing). See also GenoConvert.

LifeHistData

dataframe with 3 columns (optionally 5):

**ID** max. 30 characters long

**Sex** 1 = female, 2 = male, 3 = unknown, 4 = hermaphrodite, other numbers orNA = unknown

BirthYear birth or hatching year, integer, with missing values as NA or any negative value.

**BY.min** minimum birth year, only used if BirthYear is missing

**BY.max** maximum birth year, only used if BirthYear is missing

If the species has multiple generations per year, use an integer coding such that the candidate parents' 'Birth year' is at least one smaller than their putative offspring's. Column names are ignored, so ensure column order is ID - sex birth year (- BY.min - BY.max). Individuals do not need to be in the same order as in 'GenoM', nor do all genotyped individuals need to be included.

SeqList

list with output from a previous run, to be re-used in the current run. Used are elements 'PedigreePar', 'LifeHist', 'AgePriors', 'Specs', and 'ErrM', and these override the corresponding input parameters. Not all of these elements need to be present, and all other elements are ignored. If SeqList\$Specs is provided, all input parameters with the same name as its items are ignored, except Module/MaxSibIter.

Module

one of

pre Only input check, return SeqList\$Specs

**dup** Also check for duplicate genotypes

par Also perform parentage assignment (genotyped parents to genotyped off-

**ped** (Also) perform full pedigree reconstruction, including sibship clustering and grandparent assignment. By far the most time consuming, and may take several hours for large datasets.

NOTE: Until 'MaxSibIter' is fully deprecated: if 'MaxSibIter' differs from the default (42), and 'Module' equals the default ('ped'), MaxSibIter overrides 'Module'.

MaxSibIter [will be deprecated] number of iterations of sibship clustering, including as-

signment of grandparents to sibships and avuncular relationships between sibships. Clustering continues until convergence or until MaxSibIter is reached.

Set to 0 for parentage assignment only.

Err estimated genotyping error rate, as a single number or 3x3 matrix. Details below.

The error rate is presumed constant across SNPs, and missingness is presumed

random with respect to actual genotype.

ErrFlavour function that takes Err (single number) as input, and returns a 3x3 matrix of

observed (columns) conditional on actual (rows) genotypes, or choose from inbuilt options 'version2.0', 'version1.3', or 'version1.1', referring to the sequoia version in which they were the default. Ignored if Err is a matrix. See ErrToM.

MaxMismatch DEPRECATED AND IGNORED. Now calculated automatically using CalcMaxMismatch.

Tfilter threshold log10-likelihood ratio (LLR) between a proposed relationship versus unrelated, to select candidate relatives. Typically a negative value, related to the

fact that unconditional likelihoods are calculated during the filtering steps. More negative values may decrease non-assignment, but will increase computational

time.

Tassign minimum LLR required for acceptance of proposed relationship, relative to next

most likely relationship. Higher values result in more conservative assignments.

Must be zero or positive.

MaxSibshipSize maximum number of offspring for a single individual (a generous safety margin

is advised).

DummyPrefix character vector of length 2 with prefixes for dummy dams (mothers) and sires

(fathers); maximum 20 characters each. Length 3 vector in case of hermaphrodites

(or default prefix 'H').

Complex Breeding system complexity. Either "full" (default), "simp" (simplified, no ex-

plicit consideration of inbred relationships), "mono" (monogamous).

Herm Hermaphrodites, either "no", "A" (distinguish between dam and sire role, default

if at least 1 individual with sex=4), or "B" (no distinction between dam and sire

role). Both of the latter deal with selfing.

UseAge either "yes" (default), "no", or "extra" (additional rounds with extra reliance on

ageprior, may boost assignments but increased risk of erroneous assignments);

used during full reconstruction only.

args.AP list with arguments to be passed on to MakeAgePrior.

FindMaybeRel **DEPRECATED AND IGNORED**, advised to run GetMaybeRel separately.

TRUE/FALSE to identify pairs of non-assigned likely relatives after pedigree

reconstruction. Can be time-consuming in large datasets.

CalcLLR TRUE/FALSE; calculate log-likelihood ratios for all assigned parents (geno-

typed + dummy; parent vs. otherwise related). Time-consuming in large datasets.

Can be done separately with CalcOHLLR.

quiet suppress messages: TRUE/FALSE/"verbose".

Plot display plots from SnpStats, MakeAgePrior, and SummarySeq. Defaults (NULL)

to TRUE when quiet=FALSE or "verbose", and FALSE when quiet=TRUE. If you get error 'figure margins too large', enlarge the plotting area (drag with mouse). Error 'invalid graphics state' can be dealt with by clearing the plotting

area with dev.off().

#### **Details**

For each pair of candidate relatives, the likelihoods are calculated of them being parent-offspring (PO), full siblings (FS), half siblings (HS), grandparent-grandoffspring (GG), full avuncular (niece/nephew - aunt/uncle; FA), half avuncular/great-grandparental/cousins (HA), or unrelated (U). Assignments are made if the likelihood ratio (LLR) between the focal relationship and the most likely alternative exceed the threshold Tassign.

Dummy parents of sibships are denoted by F0001, F0002, ... (mothers) and M0001, M0002, ... (fathers), are appended to the bottom of the pedigree, and may have been assigned real or dummy parents themselves (i.e. sibship-grandparents). A dummy parent is not assigned to singletons.

The genotyping error rate 'Err' is by default at locus level, not allele level: the probability to observe true homozygote aa as heterozygote Aa is  $\approx E$ , and as alternate homozygote AA  $(E/2)^2$ ; the probability to observe a true heterozygote as aa = the probability to observe it as AA = E/2. This error structure can be fully customised by providing a 3x3 matrix of observed genotype (columns) conditional on actual genotype (rows) instead.

Full explanation of the various options and interpretation of the output is provided in the vignette.

#### Value

A list with some or all of the following components:

AgePriors	Matrix with age-difference based probability ratios for each relationship, used for full pedigree reconstruction; see MakeAgePrior for details. When running only parentage assignment (Module="par") the returned AgePriors has been updated to incorporate the information of the assigned parents, and is ready for use during full pedigree reconstruction.
DummyIDs	Dataframe with pedigree for dummy individuals, as well as their sex, estimated birth year (point estimate, upper and lower bound of 95% confidence interval; see also CalcBYprobs), number of offspring, and offspring IDs. From version 2.1 onwards, this includes dummy offspring.
DupGenotype	Dataframe, duplicated genotypes (with different IDs, duplicate IDs are not allowed). The specified number of maximum mismatches is used here too. Note that this dataframe may include pairs of closely related individuals, and monozygotic twins.
DupLifeHistID	Dataframe, row numbers of duplicated IDs in life history dataframe. For convenience only, but may signal a problem. The first entry is used.
ErrM	Error matrix; probability of observed genotype (columns) conditional on actual genotype (rows)
ExcludedInd	Individuals in GenoM which were excluded because of a too low genotyping success rate ( $<50\%$ ).
ExcludedSNPs	Column numbers of SNPs in GenoM which were excluded because of a too low genotyping success rate (<10%).
LifeHist	Provided dataframe with sex and birth year data.
LifeHistPar	LifeHist with additional columns 'Sexx' (inferred Sex when assigned as part of parent-pair), 'BY.est' (mode of birth year probability distribution), 'BY.lo' (lower limit of 95% highest density region), 'BY.hi' (higher limit), inferred after

parentage assignment. 'BY.est' is NA when the probability distribution is flat

between 'BY.lo' and 'BY.hi'.

LifeHistSib as LifeHistPar, but estimated after full pedigree reconstruction

MaybeParent Dataframe with pairs of individuals who are more likely parent-offspring than

unrelated, but which could not be phased due to unknown age difference or sex,

or for whom LLR did not pass Tassign.

MaybeRel Dataframe with pairs of individuals who are more likely to be first or second

degree relatives than unrelated, but which could not be assigned.

MaybeTrio Dataframe with non-assigned parent-parent-offspring trios (both parents are of

unknown sex), with similar columns as the pedigree

NoLH Vector, IDs in genotype data for which no life history data is provided.

Pedigree Dataframe with assigned genotyped and dummy parents from Sibship step; en-

tries for dummy individuals are added at the bottom.

PedigreePar Dataframe with assigned parents from Parentage step.

Specs Named vector with parameter values.

TotLikParents Numeric vector, Total likelihood of the genotype data at initiation and after each

iteration during Parentage.

TotLikSib Numeric vector, Total likelihood of the genotype data at initiation and after each

iteration during Sibship clustering.

AgePriorExtra As AgePriors, but including columns for grandparents and avuncular pairs. NOT

updated after parentage assignment, but returned as used during the run.

DummyClones Hermaphrodites only: female-male dummy ID pairs that refer to the same non-

genotyped individual

List elements PedigreePar and Pedigree both have the following columns:

id Individual ID

dam Assigned mother, or NA sire Assigned father, or NA

LLRdam Log10-Likelihood Ratio (LLR) of this female being the mother, versus the next

most likely relationship between the focal individual and this female. See Details below for relationships considered, and see CalcPairLL for underlying

likelihood values and further details)

LLRsire idem, for male parent

LLR for the parental pair, versus the next most likely configuration between the

three individuals (with one or neither parent assigned)

OHdam Number of loci at which the offspring and mother are opposite homozygotes

OHsire idem, for father

MEpair Number of Mendelian errors between the offspring and the parent pair, includes

OH as well as e.g. parents being opposing homozygotes, but the offspring not being a heterozygote. The offspring being OH with both parents is counted as 2

errors.

#### Disclaimer

While every effort has been made to ensure that sequoia provides what it claims to do, there is absolutely no guarantee that the results provided are correct. Use of sequoia is entirely at your own risk.

#### Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

#### References

Huisman, J. (2017) Pedigree reconstruction from SNP data: Parentage assignment, sibship clustering, and beyond. Molecular Ecology Resources 17:1009–1024.

#### See Also

- GenoConvert to read in various data formats,
- CheckGeno, SnpStats to calculate missingness and allele frequencies,
- SimGeno to simulate SNP data from a pedigree
- MakeAgePrior to estimate effect of age on relationships,
- GetMaybeRel to find pairs of potential relatives,
- SummarySeq and PlotAgePrior to visualise results,
- GetRelM to turn a pedigree into pairwise relationships,
- CalcoHLLR to calculate Mendelian errors and LLR for any pedigree,
- CalcPairLL for likelihoods of various relationships between specific pairs,
- CalcBYprobs to estimate birth years,
- PedCompare and ComparePairs to compare to two pedigrees,
- EstConf to estimate assignment errors,
- writeSeq to save results,
- vignette("sequoia") for detailed manual & FAQ.

# **Examples**

```
# parentage assignment + full pedigree reconstruction:
SeqOUT2 <- sequoia(GenoM = SimGeno_example, Err = 0.005,</pre>
                  LifeHistData = LH_HSg5, Module="ped", quiet="verbose")
SeqOUT2$Pedigree[34:42, ]
PC2 <- PedCompare(Ped_HSg5, SeqOUT2$Pedigree)</pre>
PC2$Counts["GT",,]
PC2$Counts[,,"dam"]
# different kind of pedigree comparison:
ComparePairs(Ped1=Ped_HSg5, Ped2=SeqOUT$PedigreePar, patmat=TRUE)
# results overview:
SummarySeq(SeqOUT2)
# important to run with approx. correct genotyping error rate:
SeqOUT2.b <- sequoia(GenoM = SimGeno_example, # Err = 1e-4 by default
                  LifeHistData = LH_HSg5, Module="ped", Plot=FALSE)
PC2.b <- PedCompare(Ped_HSg5, SeqOUT2.b$Pedigree)</pre>
PC2.b$Counts["GT",,]
## Not run:
# === EXAMPLE 2: real data ===
# ideally, select 400-700 SNPs: high MAF & low LD
# save in 0/1/2/NA format (PLINK's --recodeA)
GenoM <- GenoConvert(InFile = "inputfile_for_sequoia.raw",</pre>
                     InFormat = "raw") # can also do Colony format
SNPSTATS <- SnpStats(GenoM)</pre>
# perhaps after some data-cleaning:
write.table(GenoM, file="MyGenoData.txt", row.names=T, col.names=F)
# later:
GenoM <- as.matrix(read.table("MyGenoData.txt", row.names=1, header=F))</pre>
LHdata <- read.table("LifeHistoryData.txt", header=T) # ID-Sex-birthyear
SeqOUT <- sequoia(GenoM, LHdata, Err=0.005)</pre>
SummarySeq(SeqOUT)
writeSeq(SeqOUT, folder="sequoia_output") # several text files
# runtime:
SeqOUT$Specs$TimeEnd - SeqOUT$Specs$TimeStart
## End(Not run)
```

## **Description**

Simulate SNP genotype data from a pedigree, with optional missingess and errors.

#### **Usage**

```
SimGeno(
  Pedigree,
  nSnp = 400,
  ParMis = 0.4,
  MAF = 0.3,
  CallRate = 0.99,
  SnpError = 5e-04,
  ErrorFM = "version2.0",
  ReturnStats = FALSE,
  OutFile = NA,
  Inherit = "autosomal",
  InheritFile = NA,
  quiet = FALSE
)
```

#### **Arguments**

Pedigree	dataframe, pedigr	ee with the first thre	e columns being id -	dam - sire. Column

names are ignored, as are additional columns, with the exception of a 'Sex'

column when Inherit is not 'autosomal'.

nSnp number of SNPs to simulate.

ParMis single number or vector length two with proportion of parents with fully missing

genotype. Ignored if CallRate is a named vector.

MAF minimum minor allele frequency, and allele frequencies will be sampled uni-

formly between this minimum and 0.5, OR a vector with minor allele frequency at each locus. In both cases, this is the MAF among pedigree founders, the MAF

in the sample will deviate due to drift.

CallRate either a single number for the mean call rate (genotyping success), OR a vector

with the call rate at each SNP, OR a named vector with the call rate for each individual. In the third case, ParMis is ignored, and individuals in the pedigree

(as id or parent) not included in this vector are presumed non-genotyped.

SnpError mean per-locus genotyping error rate across SNPs, and a beta-distribution will

be used to simulate the number of missing cases per SNP, OR a vector with the

genotyping error for each SNP.

ErrorFM function taking the error rate (scalar) as argument and returning a 3x3 ma-

trix with probabilities that actual genotype i (rows) is observed as genotype j (columns). Inbuilt ones are as used in sequoia 'version2.0', 'version1.3', or

'version1.1'. See details.

ReturnStats in addition to the genotype matrix, return the input parameters and mean &

quantiles of MAF, error rate and call rates.

OutFile file name for simulated genotypes. If NA (default), return results within R.

Inherit inheritance pattern, scalar or vector of length nSnp, Defaults to 'autosomal'. An

excel file included in the package has inheritance patterns for the X and Y chromosome and mtDNA, and allows custom inheritance patterns. Note that these are experimental, and NOT currently supported by the pedigree reconstruction

with sequoia!

InheritFile file name of file with inheritance patterns, with extension csv, txt, xls or xlsx

(the latter two require library xlsx).

quiet suppress messages.

#### **Details**

Please ensure the pedigree is a valid pedigree, for example by first running PedPolish. For founders, i.e. individuals with no known parents, genotypes are drawn according to the provided MAF and assuming Hardy-Weinberg equilibrium. Offspring genotypes are generated following Mendelian inheritance, assuming all loci are completely independent. Individuals with one known parent are allowed: at each locus, one allele is inherited from the known parent, and the other drawn from the genepool according to the provided MAF.

Genotyping errors are generated following a user-definable 3x3 matrix with probabilities that actual genotype i (rows) is observed as genotype j (columns). This is specified as ErrorFM, which is a function of SnpError. By default (ErrorFM = "version2.0"), SnpError is interpreted as a locuslevel error rate (rather than allele-level), and equals the probability that a homozygote is observed as heterozygote, and the probability that a heterozygote is observed as either homozygote (i.e., the probability that it is observed as AA = probability that observed as aa = SnpError/2). The probability that one homozygote is observed as the other is  $(SnpError/2)^2$ .

Note that this differs from versions up to 1.1.1, where a proportion of SnpError\*3/2 of genotypes were replaced with random genotypes. This corresponds to ErrorFM = "Version111".

Error rates differ between SNPs, but the same error pattern is used across all SNPs, even when inheritance patterns vary. When two or more different error patterns are required, SimGeno should be run on the different SNP subsets separately, and results combined.

Variation in call rates is assumed to follow a highly skewed (beta) distribution, with many samples having call rates close to 1, and a narrowing tail of lower call rates. The first shape parameter defaults to 1 (but see MkGenoErrors), and the second shape parameter is defined via the mean as CallRate. For 99.9% of SNPs to have a call rate of 0.8 (0.9; 0.95) or higher, use a mean call rate of 0.969 (0.985; 0.993).

Variation in call rate between samples can be specified by providing a named vector to CallRate, which supersedes PropLQ in versions up to 1.1.1. Otherwise, variation in call rate and error rate between samples occurs only as side-effect of the random nature of which individuals are hit by per-SNP errors and drop-outs. Finer control is possible by first generating an error-free genotype matrix, and then calling MkGenoErrors directly on subsets of the matrix.

#### Value

If ReturnStats=FALSE (the default), a matrix with genotype data in sequoia's input format, encoded as 0/1/2/-9.

If ReturnStats=TRUE, a named list with three elements: list 'ParamsIN', matrix 'SGeno', and list 'StatsOUT':

AF	Frequency in 'observed' genotypes of '1' allele
AF.act	Allele frequency in 'actual' (without genotyping errors & missingness)
SnpError	Error rate per SNP (actual /= observed AND observed /= missing)
SnpCallRate	Non-missing per SNP

IndivError Error rate per individual

IndivCallRate Non-missing per individual

#### Disclaimer

This simulation is highly simplistic and assumes that all SNPs segregate completely independently, that the SNPs are in Hardy-Weinberg equilibrium in the pedigree founders. It assumes that genotyping errors are not due to heritable mutations of the SNPs, and that missingness is random and not e.g. due to heritable mutations of SNP flanking regions. Results based on this simulated data will provide an minimum estimate of the number of SNPs required, and an optimistic estimate of pedigree reconstruction performance.

#### Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

#### See Also

The wrapper EstConf for repeated simulation and pedigree reconstruction; MkGenoErrors for fine control over the distribution of genotyping errors in simulated data.

# **Examples**

```
data(Ped_HSg5)
GenoM <- SimGeno(Pedigree = Ped_HSg5, nSnp = 100, ParMis = c(0.2, 0.7))
## Not run:
# Alternative genotyping error model
EFM <- function(E) {  # Whalen, Gorjanc & Hickey 2018</pre>
matrix(c(1-E*3/4, E/4, E/4,
          E/4, 1/2-E/4, 1/2-E/4, E/4,
          E/4, E/4, 1-E*3/4),
          3,3, byrow=TRUE) }
EFM(0.01)
GenoM <- SimGeno(Pedigree = Ped_HSg5, nSnp = 100, ParMis = 0.2,
 SnpError = 5e-3, ErrorFM = EFM)
# combination of high & low quality SNPs
Geno.HQ <- SimGeno(Ped_HSg5, nSnp=50, MAF=0.3, CallRate=runif(50, 0.7, 1))
Geno.LQ <- SimGeno(Ped_HSg5, nSnp=20, MAF=0.1, CallRate=runif(20, 0.1, 5))</pre>
Geno.HQLQ <- merge(Geno.HQ, Geno.LQ, by="row.names")</pre>
## End(Not run)
```

SnpStats

SimGeno\_example

Example genotype file

## **Description**

Simulated genotype data for cohorts 1+2 in Pedigree Ped\_HSg5

# Usage

```
data(SimGeno_example)
```

#### **Format**

A genotype matrix with 214 rows (ids) and 200 columns (SNPs). Each SNP is coded as 0/1/2 copies of the reference allele, with -9 for missing values. Ids are stored as rownames.

#### Author(s)

Jisca Huisman, <jisca.huisman@gmail.com>

## See Also

Ped\_HSg5,SimGeno

SnpStats

SNP Summary Statistics

# **Description**

Estimate allele frequency (AF), missingness and Mendelian errors per SNP.

#### Usage

```
SnpStats(GenoM, Pedigree = NULL, ErrFlavour = "version2.0", Plot = TRUE)
```

# **Arguments**

Geno	ıΜ g	enotype matrix,	in sequoia's forma	at: 1 column per S	SNP, 1 row	per individual,
------	------	-----------------	--------------------	--------------------	------------	-----------------

genotypes coded as 0/1/2/-9, and rownames giving individual IDs.

Pedigree dataframe with 3 columns: ID - parent1 - parent2. Additional columns and

non-genotyped individuals are ignored. Used to estimate the error rate.

ErrFlavour function that takes the genotyping error rate Err as input, and returns a 3x3

matrix of observed (columns) conditional on actual (rows) genotypes, or choose from inbuilt ones as used in sequoia 'version2.0', 'version1.3', or 'version1.1'.

See ErrToM.

Plot show histograms of the results?

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#### **Details**

Calculation of these summary statistics can be done in PLINK, and SNPs with low minor allele frequency or high missingness should be filtered out prior to pedigree reconstruction. This function is provided as an aid to inspect the relationship between AF, missingness and genotyping error to find a suitable combination of SNP filtering thresholds to use.

For pedigree reconstruction, SNPs with zero or one copies of the alternate allele in the dataset (MAF  $\leq 1/2N$ ) are considered fixed, and excluded.

#### Value

A matrix with a number of rows equal to the number of SNPs (=number of columns of GenoM), and when no Pedigree is provided 2 columns:

AF Allele frequency of the 'second allele' (the one for which the homozygote is

coded 2)

Mis Proportion of missing calls

When a Pedigree is provided, there are 7 additional columns:

n.dam, n.sire, n.pair

Number of dams, sires, parent-pairs succesfully genotyped for the SNP

OHdam, OHsire Count of number of opposing homozygous cases

MEpair Count of Mendelian errors, includes opposing homozygous cases

Err. hat Error rate, as estimated from the joined offspring-parent (-parent) genotypes and

the presumed error structure (ErrFlavour)

# Estimated genotyping error

The error rate is estimated from the number of opposing homozygous cases (OH, parent is AA and offspring is aa) Mendelian errors (ME, e.g. parents AA and aa, but offspring not Aa) in parent-parent-offspring trios, and OH cases for offspring with a single genotyped parent.

The estimated error rates will not be as accurate as from duplicate samples. A single error in an individual with many offspring will be counted as many times, potentially resulting in non-sensical values of 'Err.hat' close to 1. On the other hand, errors in individuals without parents or offspring will not be counted at all. Moreover, a high error rate may interfere with pedigree reconstruction, and successful assignment will be biased towards parents with lower error count. Nonetheless, it may provide a ballpark estimate for the average error rate, which can be useful for subsequent (rerun of) pedigree reconstruction.

#### See Also

GenoConvert to convert from various data formats; CheckGeno to check the data is in valid format for sequoia and exclude monomorphic SNPs etc., CalcoHLLR to calculate OH & ME per individual.

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## **Examples**

```
data(Ped_HSg5)
Genotypes <- SimGeno(Ped_HSg5, nSnp=400, CallRate = runif(400, 0.2, 0.8),
    SnpError = 0.05)
SNPstats <- SnpStats(Genotypes, Pedigree=Ped_HSg5)</pre>
```

SummarySeq

Summarise Sequoia Output or Pedigree

# **Description**

Number of assigned parents and grandparents and sibship sizes, split by genotyped, dummy, and 'observed'.

# Usage

```
SummarySeq(
   SeqList = NULL,
   Pedigree = NULL,
   DumPrefix = c("F0", "M0"),
   SNPd = NULL,
   Plot = TRUE,
   Panels = "all"
)
```

# Arguments

SegList	the list returned by	seguoia.	Only elements	'Pedigree'	or 'PedigreePar'	and

'AgePriors' are used.

Pedigree dataframe, pedigree with the first three columns being id - dam - sire. Column

names are ignored, as are additional columns.

DumPrefix character vector of length 2 with prefixes for dummy dams (mothers) and sires

(fathers). Will be read from SeqList's 'Specs' if provided. Used to distinguish between dummies and non dummies. Length 3 in case of hermanhyrodites.

between dummies and non-dummies. Length 3 in case of hermaphrodites.

SNPd character vector with ids of SNP genotyped individuals. Only when Pedigree

is provided instead of SeqList, then used to distinguish between genetically assigned parents and 'observed' parents (e.g. observed in the field, or assigned previously using microsatellites). If SeqList's 'PedigreePar' is provided, all ids

in that dataframe will be presumed genotyped.

Plot show barplots and histograms of the results, as well as of the parental LLRs,

Mendelian errors, and agepriors, if present.

Panels character vector with panel(s) to plot. Choose from 'all', 'G.parents' (parents of

genotyped individuals), 'D.parents' (parents of dummy individuals), 'sibships' (distribution of sibship sizes), 'LLR' (log10-likelihood ratio parent/otherwise

related), 'OH' (count of opposite homozygote SNPs).

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#### Value

A list with the following elements:

PedSummary a 2-column matrix with basic summary statistics, similar to what used to be re-

turned by **Pedantics**' pedStatSummary (now archived on CRAN). First column refers to the complete pedigree, second column to SNP-genotyped individuals only. Maternal siblings sharing a dummy parent are counted in the 2nd column if both sibs are genotyped, but not if one of the sibs is a dummy individual.

ParentCount a 2x3x2x4 array with the number of assigned parents, split by D1: genotyped vs

dummy individuals; D2: female, male and unknown-sex individuals; D3: dams

vs sires; D4: genotyped, dummy, observed vs no parent

GPCount a 2x4x4 array with the number of assigned grandparents, split by D1: genotyped

vs dummy individuals; D2 Maternal grandmother (MGM), maternal grandfather (MGF), paternal grandmother (PGM), paternal grandfather (PGF); D3: geno-

typed, dummy, observed vs no grandparent

SibSize a list with as first element a table of maternal sibship sizes, and as second ele-

ment a table of paternal sibship sizes. Each table is a matrix with a number of rows equal to the maximum sibship size, and 3 columns, splitting by the type of

parent: genotyped, dummy, or observed.

#### See Also

sequoia for pedigree reconstruction and links to other functions.

#### **Examples**

writeColumns

Write Data to a File Column-wise

## **Description**

Write data.frame or matrix to a text file, using white space padding to keep columns aligned as in print.

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## Usage

```
writeColumns(x, file = "", row.names = TRUE, col.names = TRUE)
```

# **Arguments**

x the object to be written, preferably a matrix or data frame. If not, it is attempted

to coerce x to a matrix.

file a character string naming a file.

row.names a logical value indicating whether the row names of x are to be written along

with x.

col.names a logical value indicating whether the column names of x are to be written along

with x.

writeSeq

Write Sequoia Output to File

# Description

The various list elements returned by sequoia are each written to text files in the specified folder, or to separate sheets in a single excel file (requires library **xlsx**).

# Usage

```
writeSeq(
   SeqList,
   GenoM = NULL,
   MaybeRel = NULL,
   PedComp = NULL,
   OutFormat = "txt",
   folder = "Sequoia-OUT",
   file = "Sequoia-OUT.xlsx",
   ForVersion = 2,
   quiet = FALSE
)
```

#### **Arguments**

SeqList list returned by sequoia, to be written out.

GenoM matrix with genetic data (optional). Ignored if OutFormat='xls', as the resulting

file could become too large for excel.

MaybeRel list with results from GetMaybeRel (optional).

PedComp list with results from PedCompare (optional). SeqList\$DummyIDs is combined

with PedComp\$DummyMatch if both are provided.

OutFormat 'xls' or 'txt'.

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folder	the directory where the text files will be written; will be created if it does not
	already exists. Relative to the current working directory, or NULL for current
	working directory. Ignored if OutFormat='xls'.
file	the name of the excel file to write to, ignored if OutFormat='txt'.
ForVersion	choose '1' for back-compatibility with stand-alone sequoia versions 1.x
quiet	suppress messages.

# **Details**

The text files can be used as input for the stand-alone Fortran version of sequoia, e.g. when the genotype data is too large for R. See vignette('sequoia') for further details.

# See Also

writeColumns to write to a text file, using white space padding to keep columns aligned.

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