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| Contents   |
| CalcBYprobs  |

2 Contents

| CalcPairLL        | <br>9  |
|-------------------|--------|
| CalcRped          | <br>14 |
| CheckGeno         | <br>15 |
| ComparePairs      | <br>17 |
| Conf_griffin      | <br>20 |
| DyadCompare       | <br>21 |
| ErrToM            | <br>22 |
| EstConf           | 25     |
| EstEr             | <br>29 |
| FieldMums_griffin | <br>30 |
| FindFamilies      | 31     |
| GenoConvert       | 32     |
| Geno_griffin      | 35     |
| Geno_HSg5         | 36     |
| GetAncestors      | 36     |
| getAssignCat      | 37     |
| GetDescendants    | 38     |
| getGenerations    | 39     |
| GetLLRAge         | 40     |
| GetMaybeRel       | 41     |
| GetRelM           | 45     |
| Inherit_patterns  | 47     |
| LHConvert         | 48     |
| LH_griffin        | 49     |
| LH_HSg5           | 50     |
| MakeAgePrior      | 50     |
| MaybeRel_griffin  | 54     |
| MkGenoErrors      | 55     |
| PedCompare        | 56     |
| PedPolish         | 60     |
| PedStripFID       | 62     |
| Ped_griffin       | 63     |
| Ped_HSg5          | 63     |
| PlotAgePrior      | 64     |
| PlotPairLL        | 65     |
| PlotPedComp       | 66     |
| PlotRelPairs      |        |
| PlotSeqSum        |        |
| •                 |        |
| SeqOUT_griffin    | 69     |
| SeqOUT_HSg5       | 70     |
| sequoia           | 70     |
| SimGeno           | 78     |
| SimGeno_example   | 81     |
| SnpStats          | 82     |
| SummarySeq        | 83     |
| writeColumns      | 85     |
| writeSeq          | <br>86 |
|                   |        |

88

Index

CalcBYprobs 3

| CalcBYprobs | Birth year probabilities |  |
|-------------|--------------------------|--|
|             |                          |  |

# Description

Estimate the probability that an individual with unknown birth year is born in year y, based on BirthYears or BY.min and/or BY.max of its parents, offspring, and siblings, combined with AgePrior (the age distribution of other parent-offspring pairs), and/or Year.last of its parents.

## Usage

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

# Arguments

Pedigree dataframe with columns id-dam-sire. LifeHistData data.frame with up to 6 columns:

**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 number.

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

**Year.last** Last year in which individual could have had offspring. Can e.g. in mammals be the year before death for females, and year after death for males.

"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, and only integers are used. Individuals do not need to be in the same order as in 'GenoM', nor do all genotyped individuals 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.

## 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.

4 CalcMaxMismatch

## 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.

# **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 = "version2.9", 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.

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

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

 $Q^{(1/N)}$ , 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        |
|-----|---|
| ОН  | 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

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#### 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 for a pedigree

# **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).

## Usage

```
CalcOHLLR(
  Pedigree = NULL,
  GenoM = NULL,
  CalcLLR = TRUE,
  LifeHistData = NULL,
  AgePrior = FALSE,
  SeqList = NULL,
  Err = 1e-04,
  ErrFlavour = "version2.9",
  Tassign = 0.5,
  Tfilter = -2,
  Complex = "full",
 Herm = "no",
  quiet = FALSE
)
```

## **Arguments**

Pedigree

dataframe with columns id-dam-sire. May include non-genotyped individuals, 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, one column per SNP, coded as 0, 1, 2, missing values as a negative number or NA. You can reformat data with GenoConvert, or use other packages to get it into a genlight object and then use as.matrix.

CalcLLR

calculate log-likelihood ratios for all assigned parents (genotyped + dummy/nongenotyped; 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

data.frame with up to 6 columns:

**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 number.

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

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

**Year.last** Last year in which individual could have had offspring. Can e.g. in mammals be the year before death for females, and year after death for

"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, and only integers are used. 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 parameters. Err estimated genotyping error rate, as a single number, or a length 3 vector with P(homlhom), P(hetlhom), P(homlhet), or a 3x3 matrix. See details below. The error rate is presumed constant across SNPs, and missingness is presumed random with respect to actual genotype. Using Err >5% is not recommended, and Err >10% strongly discouraged. ErrFlavour function that takes Err (single number) as input, and returns a length 3 vector or 3x3 matrix, or choose from inbuilt options 'version2.9', 'version2.0', 'version1.3', or 'version1.1', referring to the sequoia version in which they were the default. Ignored if Err is a vector or matrix. See ErrToM for details. 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  |
| LLRpair      | 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.

## **Examples**

```
# count Mendelian errors in an existing pedigree
Ped.OH <- CalcOHLLR(Pedigree = Ped_HSg5, GenoM = SimGeno_example,
                    CalcLLR = FALSE)
Ped. OHΓ50:55.1
# 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")
## Not run:
# 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:
SeqOUT <- sequoia(Geno_griffin, 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"))
## End(Not run)
```

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 = "version2.9",
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

GenoM

numeric matrix with genotype data: One row per individual, one column per SNP, coded as 0, 1, 2, missing values as a negative number or NA. You can reformat data with GenoConvert, or use other packages to get it into a genlight object and then use as.matrix.

Pedigree

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.

LifeHistData

data.frame with up to 6 columns:

**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 number.

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

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

Year.last Last year in which individual could have had offspring. Can e.g. in mammals be the year before death for females, and year after death for males.

"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, and only integers are used. 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. The list elements 'LifeHist', 'AgePriors', and 'ErrM' are also used if present, and

override the corresponding input parameters.

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 a length 3 vector with

P(homlhom), P(hetlhom), P(homlhet), or a 3x3 matrix. See details below. The error rate is presumed constant across SNPs, and missingness is presumed random with respect to actual genotype. Using Err >5% is not recommended, and

Err >10% strongly discouraged.

ErrFlavour function that takes Err (single number) as input, and returns a length 3 vector

> or 3x3 matrix, or choose from inbuilt options 'version2.9', 'version2.0', 'version 1.3', or 'version 1.1', referring to the sequoia version in which they were the

default. Ignored if Err is a vector or matrix. See ErrToM for details.

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

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:

xx Log10-Likelihood of this pair having relationship xx, with xx being the relation-

ship abbreviations listed below.

TopRel Abbreviation of most likely relationship

LLR Log10-Likelihood ratio between most-likely and second most likely relation-

ships. Other LLRs, e.g. between most-likely and unrelated, can easily be com-

puted.

#### **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   |

## Why does it say 777 (impossible)?

This function uses the same machinery as sequoia, which will to save time not calculate the likelihood when it is quickly obvious that the pair cannot be related in the specified manner.

For PO (putative parent-offspring pairs) this is the case when:

- the sex of the candidate parent, via Pairs\$Sex2 or LifeHistData, does not match Pairs\$patmat, which defaults to 1 (maternal relatives, i.e. dam)
- a dam is already assigned via Pedigree and Pairs\$dropPar1 = 'none', and Pairs\$patmat
   = 1
- Pairs\$focal is not 'U' (the default), and the OH count between the two individuals exceeds MaxMismatchOH. This value can be found in SeqList\$Specs), and is calculated by CalcMaxMismatch
- the age difference, either calculated from LifeHistData or specified via Pairs\$AgeDif, is impossible for a parent-offspring pair according to the age prior. The latter can be specified via AgePrior, or is taken from SeqList, or is calculated when both Pedigree and LifeHistData are provided.

For FS (putative full siblings) this happens when e.g. ID1 has a dam assigned which is not dropped (Pairs\$dropPar1='none' or 'sire'), and the OH count between ID1's dam and ID2 exceeds MaxMismatchOH. The easiest way to 'fix' this is by increasing the presumed genotyping error rate.

# 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.

# **Examples**

14 CalcRped

```
tail(SeqOUT_griffin$PedigreePar, n=3)
# set up dataframe with these pairs. LLRdam & LLRsire ignore any co-parent
Pairs_d <- data.frame(ID1 = SeqOUT_griffin$PedigreePar$id[140:142],</pre>
                      ID2 = SeqOUT_griffin$PedigreePar$dam[140:142],
                      focal = "PO",
                      dropPar1 = 'both')
# Calculate LL's, conditional on the rest of the pedigree + age differences
CalcPairLL(Pairs_d, GenoM = Geno_griffin, Err = 1e-04,
           LifeHistData = LH_griffin, Pedigree = SeqOUT_griffin$PedigreePar)
# LLR changes when ignoring age and/or pedigree, as different relationships
# become (im)possible
CalcPairLL(Pairs_d, GenoM = Geno_griffin, Err = 1e-04)
# LLRpair is calculated conditional on co-parent, and min. of dam & sire LLR
Pairs_d$dropPar1 <- 'dam'
Pairs_s <- data.frame(ID1 = SeqOUT_griffin$PedigreePar$id[141:142],</pre>
                      ID2 = SeqOUT_griffin$PedigreePar$sire[141:142],
                      focal = "P0",
                      dropPar1 = 'sire')
CalcPairLL(rbind(Pairs_d, Pairs_s), GenoM = Geno_griffin, Err = 1e-04,
           LifeHistData = LH_griffin, Pedigree = SeqOUT_griffin$PedigreePar)
## likelihoods underlying LLR in getMaybeRel output:
MaybeRel_griffin$MaybePar[1:5, ]
FivePairs <- MaybeRel_griffin$MaybePar[1:5, c("ID1", "ID2", "Sex1", "Sex2")]
PairLL <- CalcPairLL(Pairs = rbind( cbind(FivePairs, focal = "PO"),</pre>
                                    cbind(FivePairs, focal = "HS"),
                                     cbind(FivePairs, focal = "GP")),
                     GenoM = Geno_griffin, Plot=FALSE)
PairLL[PairLL$ID1=="i121_2007_M", ]
# 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")
```

CheckGeno 15

# **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.

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",
   Strict = TRUE,
   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).

Strict Exclude any individuals genotyped for <5 genotyped for <5 up to version 2.4.1.

Otherwise only excluded are (very nearly) monomorphic SNPs, SNPs scored for

fewer than 2 individuals, and individuals scored for fewer than 2 SNPs.

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

16 CheckGeno

#### Value

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**

```
GenoM <- SimGeno(Ped_HSg5, nSnp=400, CallRate = runif(400, 0.2, 0.8))</pre>
# the quick way:
GenoM.checked <- CheckGeno(GenoM, Return="GenoM")</pre>
# the user supervised way:
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 ("ExcludedSnps-mono" %in% names(Excl))
  GenoM <- GenoM[, -Excl[["ExcludedSnps-mono"]]]</pre>
if ("ExcludedIndiv" %in% names(Excl))
  GenoM <- GenoM[!rownames(GenoM) %in% Excl[["ExcludedIndiv"]], ]</pre>
# warning about SNPs scored for <50% of individuals ?
# note: this is not necessarily a problem, and sometimes unavoidable.
SnpCallRate <- apply(GenoM, MARGIN=2,</pre>
                      FUN = function(x) sum(x!=-9)) / nrow(GenoM)
```

ComparePairs 17

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 as first three columns: ID1-ID2- relationship, e.g. as

returned by GetMaybeRel. Column names and any additional columns are ig-

nored. May be provided in addition to, or instead of Ped2.

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.

18 ComparePairs

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

 $(\mbox{'M0'})$  to avoid matching to regular individuals with IDs starting with 'F'  $(\mbox{'M'}),$ 

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

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

 ${f hi}$  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 unre-

lated in Ped2

Array 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 pair

id.B Second individual of the pair

RC1 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.

ComparePairs 19

# 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 subclasses. When a pair is related via multiple paths, the first-listed relationship is returned. To get all the different paths between a pair, use GetRelM with Return='Array'.

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

20 Conf\_griffin

# 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.

## **Examples**

Conf\_griffin

Example output from estimating confidence probabilities: griffins

# **Description**

Example output of EstConf, with the inferred pedigree in SeqOUT\_griffin used as reference pedigree.

## Usage

```
data(Conf_griffin)
```

## Format

```
a list, see sequoia
```

#### Author(s)

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

## See Also

```
Ped_griffin, Geno_griffin,
```

DyadCompare 21

## **Examples**

DyadCompare

Compare Dyads (DEPRECATED)

# **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

# **Examples**

```
## Not run:
DyadCompare(Ped1=Ped_HSg5, Ped2=SeqOUT_HSg5$Pedigree)
## End(Not run)
```

22 ErrToM

**ErrToM** 

Generate Genotyping Error Matrix

# Description

Make a vector or matrix specifying the genotyping error pattern, or a function to generate such a vector/matrix from a single value Err.

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 = "version2.9", Return = "matrix")
```

# **Arguments**

| Err | estimated genotyping error rate, as a single number, or 3x3 or 4x4 matrix, or      |
|-----|--|
|     | length 3 vector. 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. If a vector,    |
|     | these are the probabilities (observed given actual) homlother hom, hetlhom, and    |

homlhet.

flavour vector-generating or matrix-generating function, or one of 'version2.9', 'ver-

sion2.0', 'version1.3' (='SNPchip'), 'version1.1' (='version111'), referring to the sequoia version in which it was used as default. Only used if Err is a single

number.

Return output, 'matrix' (default), 'vector', 'function' (matrix-generating), or 'v\_function'

(vector-generating)

#### **Details**

By default (flavour = "version2.9"), 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 one homozygote is observed as the other is  $(Err/2)^2$ .

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.

This function, and throughout the package, it is assumed that the two alleles A and a are equivalent. Thus, using notation P(observed genotype | lactual genotype), that P(AA|aa) = P(aa|AA), P(aa|Aa) = P(AA|Aa), and P(aA|aa) = P(aA|AA).

version hom/hom het/hom hom/het

ErrToM 23

or in matrix form, Pr(observed genotype (columns) | actual genotype (rows)): *version2.9:* 

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

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

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

version1.1

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

version0.9 (not recommended)

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

24 ErrToM

When Err is a length 3 vector, or if Return = 'vector' these are the following probabilities:

- homlhom: an actual homozygote is observed as the other homozygote  $(E_1)$
- hetlhom: an actual homozygote is observed as heterozygote  $(E_2)$
- homlhet: an actual heterozygote is observed as homozygote  $(E_3)$

and Pr(observed genotype (columns) | actual genotype (rows)) is then:

When the SNPs are scored via sequencing (e.g. RADseq or DArTseq), the 3rd error rate (homlhet) is typically considerably higher than the other two, while for SNP arrays it tends to be similar to P(hetlhom).

#### Value

Depending on Return, either:

- 'matrix': a 3x3 matrix, with probabilities of observed genotypes (columns) conditional on actual (rows)
- 'function': a function taking a single value Err as input, and generating a 3x3 matrix
- 'vector': a length 3 vector, with the probabilities (observed given actual) homlother hom, hetlhom, and homlhet.

# **Examples**

| 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), birth year, and optionally BY.min - BY.max - YearLast.  |
| 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 (assumed 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 <b>parallel</b> 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). With large datasets, the amount of computer memory may be the limiting factor for the number of cores you can use. |
| quiet        | suppress messages. TRUE runs SimGeno and sequoia quietly, 'very' also suppresses 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 array with Counts from the 'forward' PedCompare, from which PedErrors is

calculated

RunParams a list with the call to EstConf as a semi-nested list (args.sim, args.seq, nSim,

nCores), 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:

• 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.

An experimental version offering more fine-grained control is available at https://github.com/JiscaH/sequoiaExtra

# 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.

#### **Errors**

If you have a large pedigree and try to run this function on multiple cores, you may run into "Cannot allocate vector of size ..." errors or even unexpected crashes: there is not enough computer memory for each separate run. Try reducing 'nCores'.

## See Also

```
SimGeno, sequoia, PedCompare.
```

## **Examples**

```
args.seq = list(Err=5e-3, Module="par"), # as in real run
               nSim = 1, # try-out, proper run >=20 (10 if huge pedigree)
               nCores=1)
# parent-pair confidence, per category (Genotyped/Dummy/None)
conf_grif$ConfProb
# Proportion of true parents that was correctly assigned
1 - apply(conf_grif$PedErrors, MARGIN=c('cat', 'parent'), FUN=sum, na.rm=TRUE)
# add columns with confidence probabilities to pedigree
# first add columns with category (G/D/X)
Ped.withConf <- getAssignCat(Pedigree = SeqOUT_griffin$Pedigree,</pre>
                              SNPd = SeqOUT_griffin$PedigreePar$id)
Ped.withConf <- merge(Ped.withConf, conf_grif$ConfProb, all.x=TRUE,</pre>
                      sort=FALSE) # (note: merge() messes up column order)
head(Ped.withConf[Ped.withConf$dam.cat=="G", ])
# save output summary
## Not run:
conf_griff[['Note']] <- 'You could add a note'</pre>
saveRDS(conf_grif[c('ConfProb', 'PedComp.fwd', 'RunParams', 'RunTime', 'Note')],
   file = 'conf_200SNPs_Err005_Callrate80.RDS')
## End(Not run)
## P(actual FS | inferred as FS) etc.
## Not run:
PairL <- list()</pre>
for (i in 1:length(conf_grif$Pedigree.inferred)) {  # nSim
  cat(i, "\t")
  PairL[[i]] <- ComparePairs(conf_grif$Pedigree.reference,</pre>
                              conf_grif$Pedigree.inferred[[i]],
                              GenBack=1, patmat=TRUE, ExcludeDummies = TRUE,
                              Return="Counts")
# P(actual relationship (Ped1) | inferred relationship (Ped2))
PairRel.prop.A <- plyr::laply(PairL, function(M)
                     sweep(M, MARGIN='Ped2', STATS=colSums(M), FUN="/"))
PairRel.prop <- apply(PairRel.prop.A, 2:3, mean, na.rm=TRUE) #avg across sims
round(PairRel.prop, 3)
# or: P(inferred relationship | actual relationship)
PairRel.prop2 <- plyr::laply(PairL, function(M)</pre>
   sweep(M, MARGIN='Ped1', STATS=rowSums(M), FUN="/"))
## End(Not run)
## Not run:
# confidence probability vs. sibship size
source('https://raw.githubusercontent.com/JiscaH/sequoiaExtra/main/conf_vs_sibsize.R')
conf_grif_nOff <- Conf_by_nOff(conf_grif)</pre>
conf_grif_nOff['conf',,'GD',]
conf_grif_nOff['N',,'GD',]
```

EstEr 29

```
## End(Not run)
```

EstEr

Estimate genotyping error rate (REMOVED; will be re-implemented)

# **Description**

Estimate the genotyping error rates in SNP data, based on a pedigree and/or duplicates. Estimates probabilities (observed given actual) homlother hom, hetlhom, and homlhet. THESE ARE AP-PROXIMATE VALUES!

## Usage

```
EstEr(
   GenoM,
   Pedigree,
   Duplicates = NULL,
   Er_start = c(0.05, 0.05, 0.05),
   perSNP = FALSE
)
```

## **Arguments**

GenoM Genotype matrix

Pedigree data.frame with columns id - dam - sire

Duplicates matrix or data.frame with 2 columns, id1 & id2

Er\_start vector of length 3 with starting values for optim.

perSNP logical, estimate error rate per SNP. WARNING not very precise, use only as an

approximate indicator! Try on simulated data first, e.g. with SimGeno.

# **Details**

The result should be interpreted as approximate, ballpark estimates! The estimated error rates from a pedigree will not be as accurate as from duplicate samples. Errors in individuals without parents or offspring will not be counted, and errors in individuals with only few offspring may not be noted either. Deviation of genotype frequencies among founders from Hardy-Weinberg equilibrium may wrongly be attributed to genotyping errors. Last but not least, any pedigree errors will result in higher estimated genotyping errors.

30 FieldMums\_griffin

## Value

vector of length 3 with estimated genotyping error rates: the probabilities that

- · homlhom: an actual homozygote is observed as the other homozygote
- · hetlhom: an actual homozygote is observed as heterozygote
- homlhet: an actual heterozygote is observed as homozygote

These are three independent parameters, that define the genotyping error matrix (see ErrToM) as follows:

$$\begin{array}{ccccccc} & \mathbf{0} & \mathbf{1} & \mathbf{2} \\ \mathbf{0} & 1 - E_1 - E_2 & E_2 & E_1 \\ \mathbf{1} & E_3 & 1 - 2E_3 & E_3 \\ \mathbf{2} & E_1 & E_2 & 1 - E_1 - E_2 \end{array}$$

Note that for optim a lower bound of 1e-6 and upper bound of 0.499 are used; if these values are returned this should be interpreted as 'inestimably small' and 'inestimably large', respectively. PLEASE DO NOT USE THESE VALUES AS INPUT IN SUBSEQUENT ANALYSIS BUT SUBSITUTE BY A SENSIBLE VALUE!!

# **Examples**

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>

FindFamilies 31

# See Also

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

# Examples

FindFamilies

Assign Family IDs

## **Description**

Find clusters of connected individuals in a pedigree, and assign each cluster a unique family ID (FID).

## Usage

```
FindFamilies(Pedigree = NULL, SeqList = NULL, MaybeRel = NULL)
```

# **Arguments**

| Pedigree | dataframe with columns id - parent1 - parent2; only the first 3 columns will be used.   |
|----------|---|
| SeqList  | list as returned by sequoia. If Pedigree is not provided, the element Pedigree from this list will be used if present, and element Pedigreepar otherwise. |
| MavbeRel | Output from GetMaybeRel, a dataframe with probable but non-assigned rela-   |

tives.

## **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 numeric vector with length equal to the number of unique individuals in the pedigree (i.e. number of rows in pedigree after running PedPolish on Pedigree).

32 **GenoConvert** 

## See Also

GetAncestors, GetDescendants, getGenerations

## **Examples**

```
PedG <- SeqOUT_griffin$PedigreePar[,1:3]</pre>
FID_G <- FindFamilies(PedG)</pre>
PedG[FID_G==4,]
```

GenoConvert

Convert Genotype Data

# **Description**

Convert genotype data in various formats to sequoia's 1-column-per-marker format, PLINK's ped 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", "-1", <math>c("0")[InFormat \%in\% \ c("col", "number of "num
                                 "ped")]),
               sep = c("", "\t", ",", ";"),
              header = NA,
               IDcol = NA,
               FIDcol = NA,
              FIDsep = "_",
               dropcol = NA,
              quiet = FALSE
)
```

# **Arguments**

InData dataframe, matrix or genlight object with genotypes to be converted. InFile character string with name of genotype file to be converted. One of 'seq' (sequoia), 'ped' (PLINK .ped file), 'col' (COLONY), 'raw' (PLINK InFormat

-recodeA), 'vcf' (requires library {vcfR}), 'single' (1 column per SNP), or 'double' (2 columns per SNP); see Details.

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.

GenoConvert 33

| OutFormat | as InFormat; only 'seq', 'col', and 'ped' are implemented. For 'ped' also a sham .map file is created, so that the file can be read by PLINK. Only for 'ped' are extensions .ped & .map added to the specified OutFile filename.   |
|-----------|--|
| Missing   | vector with symbols interpreted as missing data. '0' is missing data for $InFormats$ 'col' and 'ped' only.   |
| sep       | vector with field separator strings that will be tried on InFile. Ignored if package <b>data.table</b> is present or if InFormat='vcf'. The OutFile 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.  |
| FIDsep    | string used to paste FID and IID together into a composite-ID (value passed to 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.

If package **data.table** is detected, fread is used to read in the data from file. Otherwise, a combination of readLines and strsplit is used.

## Value

A genotype matrix in the specified output format; the default sequoia format ('seq') has 1 column per SNP coded in 0/1/2 format (major homozygote /heterozygote /minor homozygote) with -9 for missing values, sample IDs in row names and SNP names in column names. If 'OutFile' is specified, the matrix is written to this file and nothing is returned inside R.

# **Input formats**

The following formats can be specified by InFormat:

seq (sequoia) genotypes are coded as 0, 1, 2, missing as -9 (in input any negative number or NA are OK), in 1 column per marker. Column 1 contains IDs, there is no header row.

34 GenoConvert

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). If an associated .map file exists, SNP names will be read from there.

- **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
- col (Colony) genotypes are coded as numeric values, missing as 0, in 2 columns per marker. Column 1 contains IDs.
- vcf (VCF) genotypes are coded as '0/0','0/1','1/1', variable number of header rows followed by 1 row per SNP, with various columns of metadata followed by 1 column per individual. Requires package vcfR.

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.

# **Examples**

Geno\_griffin 35

Geno\_griffin

Example genotype file: Griffins

# **Description**

Simulated genotype data from Pedigree Ped\_griffin

#### Usage

```
data(Geno_griffin)
```

# **Format**

A genotype matrix with 142 rows (individuals) 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

SimGeno

36 GetAncestors

Geno\_HSg5

Example genotype file: 'HSg5'

# Description

Simulated genotype data for all\* individuals in Pedigree Ped\_HSg5 (\*: with 40

# Usage

```
data(Geno_HSg5)
```

## **Format**

A genotype matrix with 920 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

```
LH_HSg5, SimGeno, SeqOUT_HSg5
```

# **Examples**

GetAncestors

Get ancestors

# Description

get all ancestors of an individual

# Usage

```
GetAncestors(id, Pedigree)
```

getAssignCat 37

# **Arguments**

id id of the individual

Pedigree dataframe with columns id - parent1 - parent2; only the first 3 columns will be

used.

#### Value

a list with as first element id, second parents, third grandparents, etc.. Each element is a vector with ids, the first three elements are named, the rest numbered. Ancestors are unsorted within each list element.

# Examples

```
Anc_i200 <- GetAncestors('i200_2010_F', Ped_griffin)</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.

.

38 GetDescendants

#### **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).

#### 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**

GetDescendants

Get descendants

## **Description**

get all descendants of an individual

## Usage

```
GetDescendants(id, Pedigree)
```

getGenerations 39

## **Arguments**

id id of the individual

Pedigree dataframe with columns id - parent1 - parent2; only the first 3 columns will be

used.

#### Value

a list with as first element id, second offspring, third grand-offspring, etc.. Each element is a vector with ids, the first three elements are named, the rest numbered.

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.

### Value

A vector with the generation number for each individual, starting at 0 for founders. Offspring of G0 X G0 are G1, offspring of G0 X G1 or G1 x G1 are G2, etc. NA indicates a pedigree loop where an individual is its own ancestor (or that the pedigree has >1000 generations).

If no output name is specified, no results are returned, only an error message when the pedigree contains a loop.

To get more details about a pedigree loop, you can use https://github.com/JiscaH/sequoiaExtra/blob/main/find\_pedigree\_loop

### See Also

GetAncestors, GetDescendants to get all ancestors resp. descendants of a specific individual (with a warning if it is its own ancestor); FindFamilies to find connected sub-pedigrees.

40 GetLLRAge

## **Examples**

```
# returns nothing if OK, else error:
getGenerations(SeqOUT_griffin$Pedigree)

# returns vector with generation numbers:
G <- getGenerations(SeqOUT_griffin$Pedigree, StopIfInvalid=FALSE)
table(G, useNA='ifany')
Ped_plus_G <- cbind(SeqOUT_griffin$Pedigree, G)</pre>
```

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.

## **Examples**

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,
 Module = "par",
  Complex = "full",
 Herm = "no",
 Err = 1e-04,
 ErrFlavour = "version2.9",
  Tassign = 0.5,
  Tfilter = -2,
 MaxPairs = 7 * nrow(GenoM),
  quiet = FALSE,
 ParSib = NULL,
 MaxMismatch = NA
)
```

### **Arguments**

GenoM

numeric matrix with genotype data: One row per individual, one column per SNP, coded as 0, 1, 2, missing values as a negative number or NA. You can reformat data with GenoConvert, or use other packages to get it into a genlight object and then use as .matrix.

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

data.frame with up to 6 columns:

**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 number.

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

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

**Year.last** Last year in which individual could have had offspring. Can e.g. in mammals be the year before death for females, and year after death for males.

"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, and only integers are used. 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).

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 explicit 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 a length 3 vector with P(homlhom), P(hetlhom), P(homlhet), or a 3x3 matrix. See details below. The error rate is presumed constant across SNPs, and missingness is presumed random with respect to actual genotype. Using Err >5% is not recommended, and Err >10% strongly discouraged.

ErrFlavour

function that takes Err (single number) as input, and returns a length 3 vector or 3x3 matrix, or choose from inbuilt options 'version2.9', 'version2.0', 'version1.3', or 'version1.1', referring to the sequoia version in which they were the default. Ignored if Err is a vector or matrix. See ErrToM for details.

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.

MaxPairs the maximum number of putative pairs to return.

quiet logical, suppress messages.

ParSib **DEPRECATED, use** Module either 'par' to check for putative parent-offspring

pairs only, or 'sib' to check for all types of first and second degree relatives.

MaxMismatch DEPRECATED AND IGNORED. Now calculated automatically using CalcMaxMismatch.

#### **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

A dataframe with non-assigned likely parent-offspring pairs, with columns:

- ID1
- ID2
- TopRel: the most likely relationship, using abbreviations listed below
- LLR: Log10-Likelihood Ratio between most likely and next most likely relationship
- OH: Number of loci at which the two individuals are opposite homozygotes
- BirthYear1: Birth year of ID1 (copied from LifeHistData)
- BirthYear2
- AgeDif: Age difference; BirthYear1 BirthYear2
- Sex1: Sex of ID1 (copied from LifeHistData)
- Sex2
- SnpdBoth: Number of loci at which the two individuals are both successfully genotyped

MaybeRe1

A dataframe with non-assigned likely pairs of relatives, with columns identical to MaybePar

MaybeTrio

A dataframe with non-assigned parent-parent-offspring trios, with columns:

- ID
- parent1
- parent2
- TopRel: the most likely relationship, using abbreviations listed below
- LLRparent1: Log10-Likelihood Ratio between parent1 being a parent of ID vs the next most likely relationship between the pair, ignoring parent2
- LLRparent2: as LLRparent1
- LLRpair: LLR for the parental pair, versus the next most likely configuration between the three individuals (with one or neither parent assigned)
- OHparent1: Number of loci at which ID and parent1 are opposite homozygotes

- OHparent2: as OHparent1
- 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.parent1: Number of loci at which ID and parent1 are both successfully genotyped
- SNPd.id.parent2: as SNPd.id.parent1

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 $FA$ |
| 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.

# **Examples**

```
## Not run:
# without conditioning on pedigree
MaybeRel_griffin <- GetMaybeRel(GenoM=Geno_griffin, Err=0.001, Module='par')</pre>
## End(Not run)
names(MaybeRel_griffin)
# conditioning on pedigree
MaybePO <- GetMaybeRel(GenoM = Geno_griffin, SeqList = SeqOUT_griffin,</pre>
                      Module = 'par')
head(MaybeP0$MaybePar)
# instead of providing the entire SeqList, one may specify the relevant
# elements separately
Maybe <- GetMaybeRel(GenoM = Geno_griffin,</pre>
                     Pedigree = SeqOUT_griffin$PedigreePar,
                     LifeHistData = LH_griffin,
                     Err=0.0001, Complex = "full",
                     Module = "ped")
head(Maybe$MaybeRel)
```

GetReIM 45

```
# visualise results, turn dataframe into matrix first:
MaybeM <- GetRelM(Pairs = Maybe$MaybeRel)
PlotRelPairs(MaybeM)
# or combine with pedigree (note suffix '?')
RelM <- GetRelM(Pedigree =SeqOUT_griffin$PedigreePar, Pairs = Maybe$MaybeRel)
PlotRelPairs(RelM)</pre>
```

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,
   directed = TRUE,
   Return = "Matrix",
   Pairs_suffix = "?"
)
```

# Arguments

Pedigree dataframe with columns id - dam - sire.

Pairs dataframe with columns ID1 - ID2 - Rel, e.g. as returned by GetMaybeRel.

Combining Pedigree and Pairs works best if the relationships are coded as

listed below.

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.

directed logical, distinguish between e.g. ID1=offspring, ID2=mother ('M') and ID1=mother,

ID2=offspring ('O')? Defaults to TRUE; if FALSE both are are scored as 'PO', as are father-offspring pairs, and all grandparent– grand-offspring pairs are scored as 'GPO', and avuncular pairs as 'FNA' and 'HNA'. Not (currently) compatible with patmat. When Return='List', each pair is included twice (as ID1-ID2 &

ID2-ID1)

46 GetReIM

Return 'Matrix', 'Array', or 'List'. 'Matrix' returns an N x N matrix with the closest

relationship between each pair. 'Array' returns 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. 'List' returns a list with for each of the R considered relationships a 2-column matrix with the IDs of the pairs having such a relationship. The size of the list (in Mb) is much smaller than for the matrix or array, and this is therefore the only format suitable for pedigrees with many thousands of individuals. If Pairs is specified, the only possible return type is 'Matrix'.

Pairs\_suffix symbol added to the relationship abbreviations derived from Pairs, when both

Pedigree and Pairs are provided. Can be an empty string.

#### **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'. Double half-avuncular relationships are indicated as both FA and 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 or of the List:

| S | Self   |
|---|--------|
| М | Mother |
| Р | Father |

MP Mother or Father (patmat=FALSE)

O Offspring
FS Full sibling

MHS Maternal half-sibling
PHS Paternal half-sibling

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

MGF Maternal grandfather
PGM Paternal grandmother

Inherit\_patterns 47

| PGF  | Paternal grandfather                                |
|------|---|
| GP   | Grandparent (patmat=FALSE)                          |
| GO   | Grand-offspring                                     |
| FA   | Full avuncular; maternal or paternal aunt or uncle. |
| FN   | Full nephew/niece                                   |
| HA   | Half avuncular                                      |
| HN   | Half nephew/niece                                   |
| DFC1 | Double full first cousin                            |
| FC1  | Full first cousin                                   |
| U    | Unrelated (or otherwise related)                    |

# See Also

ComparePairs for comparing pairwise relationships between two pedigrees; PlotRelPairs.

# **Examples**

```
Rel.griffin <- GetRelM(Ped_griffin, directed=FALSE) # few categories
Rel.griffin <- GetRelM(Ped_griffin, patmat=TRUE, GenBack=2) # many cat.
table(as.vector(Rel.griffin))
# turning matrix into vector first makes table() much faster
PlotRelPairs(Rel.griffin)</pre>
```

Inherit\_patterns

Inheritance patterns

# **Description**

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

## Usage

```
data(Inherit_patterns)
```

### **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

LHConvert

#### Author(s)

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

#### See Also

SimGeno

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.

LH\_griffin 49

## See Also

GenoConvert, PedStripFID to reverse UseFID.

# **Examples**

LH\_griffin

Example life history data: griffins

# **Description**

Example life history data associated with the 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
```

# **Examples**

LH\_HSg5

Example life history file: 'HSg5'

# Description

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

## Usage

```
data(LH_HSg5)
```

#### **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

 ${\tt MakeAgePrior}$ 

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

## **Arguments**

Pedigree

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) - birthyear (optional columns BY.min - BY.max - YearLast not used), with unknown birth years coded as negative numbers or NA. "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.

MinAgeParent

minimum age of a parent, a single number (min across dams and sires) or a vector of length two (dams, sires). Defaults to 1. When there is a conflict with the minimum age in the pedigree, the pedigree takes precedent.

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 set to latest - earliest birth year in LifeHistData, or estimated from the pedigree if one is provided. 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.

lambdaNW 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:

M Mothers
P Fathers
FS Full siblings

MS Maternal half-siblings
PS Paternal half-siblings

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

BirthYearRange vector length 2

MaxAgeParent vector length 2, see details

tblA.R matrix with the counts per age difference (rows) / relationship (columns) com-

bination, 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,

and Flatten.

#### **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

54 MaybeRel\_griffin

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. Not compatible with Smooth. If the largest age difference in the pedigree is larger than the specified MaxAgeParent, the pedigree takes precedent (i.e. the largest of the two is used).

@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(MaxAgeParent = c(2,3))
MakeAgePrior(Discrete=TRUE)
# single cohort:
MakeAgePrior(LifeHistData = data.frame(ID = letters[1:5], Sex=3,
 BirthYear=1984))
# overlapping generations:
# 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,
             LifeHistData=SeqOUT_griffin$LifeHist,
             Smooth=TRUE, Flatten=TRUE)
# Call from sequoia() via args.AP:
Seq_HSg5 <- sequoia(SimGeno_example, LH_HSg5, Module="par",</pre>
                args.AP=list(Discrete = TRUE), # non-overlapping generations
                CalcLLR = FALSE, # skip time-consuming calculation of LLR's
                Plot = FALSE)
                                   # no summary plots when finished
```

MaybeRel\_griffin

Example output from check for relatives: griffins

#### Description

Example output of a check for parent-offspring pairs and parent-parent-offspring trios with GetMaybeRel, with Geno\_griffin as input (simulated from Ped\_griffin).

MkGenoErrors 55

# Usage

```
data(MaybeRel_griffin)
```

#### **Format**

a list with 2 dataframes, 'MaybePar' and 'MaybeTrio'. See GetMaybeRel for further details.

## Author(s)

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

## See Also

```
SeqOUT_griffin
```

# **Examples**

MkGenoErrors

Simulate Genotyping Errors

# **Description**

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

# Usage

```
MkGenoErrors(
   SGeno,
   CallRate = 0.99,
   SnpError = 5e-04,
   ErrorFV = function(E) c((E/2)^2, E - (E/2)^2, E/2),
   ErrorFM = NULL,
   Error.shape = 0.5,
   CallRate.shape = 1,
   WithLog = FALSE
)
```

## **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 as parent) not included in this vector are presumed non-genotyped. |
| SnpError       | either a single value which will be combined with ErrorFV, or a length 3 vector with probabilities (observed given actual) homlother hom, hetlhom, and homlhet; OR a vector or 3XnSnp matrix with the genotyping error rate(s) for each SNP.   |
| ErrorFV        | function taking the error rate (scalar) as argument and returning a length 3 vector with hom->other hom, hom->het, het->hom. May be an 'ErrFlavour', e.g. 'version2.9'.  |
| ErrorFM        | function taking the error rate (scalar) as argument and returning a 3x3 matrix with probabilities that actual genotype i (rows) is observed as genotype j (columns). See below for details. To use, set ErrorFV = NULL   |
| Error.shape    | first shape parameter (alpha) of beta-distribution of per-SNP error rates. A higher value results in a flatter distribution.   |
| CallRate.shape | as Error.shape, for per-SNP call rates.  |
| WithLog        | Include dataframe in output with which datapoints have been edited, with columns id - $SNP$ - actual (original, input) - observed (edited, output).  |

# Value

The input genotype matrix, with some genotypes replaced, and some set to missing (-9). If WithLog=TRUE, a list with 3 elements: GenoM, Log, and Counts\_actual (genotype counts in input, to allow double checking of simulated genotyping error rate).

| 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 = "1sib1GP",
   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 Seq0UT\$Pedigree or Seq0UT\$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 (default)

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

version 2.4)

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 sibehin 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 in- |
|          | cluded in the count here.  |
| P1only   | Parent in Pedigree 1 but not 2; includes non-assignable parents (e.g. not geno-  |
|          | typed and no genotyped offspring).   |
| P2only   | 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

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.

60 PedPolish

### 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; getAssignCat for all parents in the reference pedigree that could have been assigned; CalcOHLLR to check how well an 'old' pedigree fits with the SNP data.

# **Examples**

```
compare <- PedCompare(Ped_griffin, SeqOUT_griffin$Pedigree)</pre>
compare$Counts["TT",,] # totals only; 45 dams & 47 sires non-assigned
compare$Counts[,,"dam"] # dams only
# inspect non-assigned in Ped2, id genotyped, dam might-be-dummy
PedM <- compare$MergedPed # for brevity
PedM[PedM$id.dam.cat=='GD' & PedM$dam.class=='P1only',]
# zoom in on specific dam
PedM[which(PedM$dam.1=="i011_2001_F"), ]
# no sire for 'i034_2002_F' -> impossible to tell if half-sibs or avuncular
# overview of all non-genotyped -- dummy matches
head(compare$DummyMatch)
# success of paternity assignment, if genotyped mother correctly assigned
dimnames(compare$Counts.detail)
compare$Counts.detail["G","G",,"Match",]
# default before version 3.5: minSibSize = '2sib'
compare_2s <- PedCompare(Ped_griffin, SeqOUT_griffin$Pedigree,</pre>
                         minSibSize = '2sib')
compare_2s$Counts[,,"dam"] # note decrease in Total 'dummies
with(compare_2s$MergedPed, table(id.dam.cat, dam.class))
# some with id.cat = 'X' or dam.cat='X' are nonetheless dam.class='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,
```

PedPolish 61

```
ZeroToNA = TRUE,
NAToZero = FALSE,
DropNonSNPd = TRUE,
FillParents = FALSE,
KeepAllColumns = TRUE,
KeepAllRows = 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.

KeepAllColumns Keep all columns in Pedigree (TRUE, default), or only id - dam - sire (FALSE).

Keep All rows in Pedigree (TRUE), or drop rows where id = NA (FALSE, de-

fault). Duplicated rows are always removed.

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 an exact or partial match with (case is ignored):

```
id "id", "iid", "off"
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 by this function if necessary.

62 PedStripFID

## **Examples**

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

Ped\_griffin 63

Ped\_griffin Example pedigree: griffins

# Description

Example pedigree with overlapping generations and polygamy.

## Usage

```
data(Ped_griffin)
```

## **Format**

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

#### Code

The R code used to create this pedigree can be found in /data-raw.

## 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: 'HSg5'

# Description

A pedigree with five non-overlapping generations and considerable inbreeding. Each female mated with two random males and each male with three random females, producing four full-sib offspring per mating. This is **Pedigree II** in the paper.

# Usage

```
data(Ped_HSg5)
```

## **Format**

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

PlotAgePrior

## 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

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**

AP matrix with age priors (P(A|R)/P(A)) with age differences in rows and rela-

tionships in columns; by default M: maternal parent (mother), P: paternal parent (father), FS: full siblings, MS: maternal siblings (full + half), PS: paternal sib-

lings.

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**

```
PlotAgePrior(SeqOUT_griffin$AgePriors)
PlotAgePrior(SeqOUT_griffin$AgePriorExtra)
```

PlotPairLL 65

|   | -   | _          |      |      |  |
|---|-----|------------|------|------|--|
| п | Ι ~ | <b>+</b> F | 'a i | <br> |  |
|   |     |            |      |      |  |
|   |     |            |      |      |  |

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   | $number of rows in the figure layout. \ If {\tt NULL}, set to {\tt ceiling(length(combo)/ncols)}.$  |
| 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.
```

66 PlotPedComp

## **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**

PlotRelPairs 67

|--|

# 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.

68 PlotSeqSum

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**

PlotSeqSum

Plot Summary Overview of sequoia Output

## **Description**

visualise the numbers of assigned parents, sibship sizes, and parental LLRs

## Usage

```
PlotSeqSum(SeqSum, Pedigree = NULL, Panels = "all", ask = TRUE)
```

SeqOUT\_griffin 69

## **Arguments**

SeqSum list output from SummarySeq.

Pedigree dataframe with at least id, dam and sire in columns 1-3, respectively. If columns

with parental LLRs and/or Mendelian errors are present, these will be plotted as

well.

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

genotyped individuals), 'D.parents' (parents of dummies), 'O.parents' (parents

of non-genotyped non-dummies), sibships', 'LLR', 'OH'.

ask ask for user key stroke before proceeding to next plot.

# **Examples**

```
sumry <- SummarySeq(SeqOUT_griffin, Plot=FALSE)
PlotSeqSum(sumry, SeqOUT_griffin$Pedigree, Panels='all', ask=FALSE)</pre>
```

SeqOUT\_griffin

Example output from pedigree inference: griffins

## **Description**

Example output of a sequoia run including sibship clustering, with Geno\_griffin as input (simulated from Ped\_griffin).

# Usage

```
data(SeqOUT_griffin)
```

## **Format**

```
a list, see sequoia
```

# Author(s)

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

#### See Also

sequoia

## **Examples**

70 sequoia

SeqOUT\_HSg5

Example output from pedigree inference: 'HSg5'

# **Description**

Example output of a sequoia run including sibship clustering, based on Pedigree Geno\_HSg5.

# Usage

```
data(SeqOUT_HSg5)
```

#### **Format**

```
a list, see sequoia
```

### Author(s)

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

#### See Also

```
Ped_HSg5, LH_HSg5
```

# **Examples**

sequoia

Pedigree Reconstruction

# Description

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

sequoia 71

## Usage

```
sequoia(
  GenoM = NULL,
  LifeHistData = NULL,
  SeqList = NULL,
 Module = "ped",
 Err = 1e-04,
 Tfilter = -2,
  Tassign = 0.5,
 MaxSibshipSize = 100,
 DummyPrefix = c("F", "M"),
  Complex = "full",
 Herm = "no",
 UseAge = "yes",
  args.AP = list(Flatten = NULL, Smooth = TRUE),
 mtSame = NULL,
 CalcLLR = TRUE,
  quiet = FALSE,
 Plot = NULL,
  StrictGenoCheck = TRUE,
 ErrFlavour = "version2.9",
 MaxSibIter = 42,
 MaxMismatch = NA,
 FindMaybeRel = FALSE
)
```

# **Arguments**

GenoM

numeric matrix with genotype data: One row per individual, one column per SNP, coded as 0, 1, 2, missing values as a negative number or NA. You can reformat data with GenoConvert, or use other packages to get it into a genlight object and then use as .matrix.

LifeHistData

data.frame with up to 6 columns:

**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 number.

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

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

**Year.last** Last year in which individual could have had offspring. Can e.g. in mammals be the year before death for females, and year after death for males.

"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, and only integers are used. Individuals do not need to be in the same order as in 'GenoM', nor do all genotyped individuals need to be included.

72 sequoia

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 offspring)

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'.

estimated genotyping error rate, as a single number, or a length 3 vector with P(homlhom), P(hetlhom), P(homlhet), or a 3x3 matrix. See details below. The error rate is presumed constant across SNPs, and missingness is presumed random with respect to actual genotype. Using Err >5% is not recommended, and Err >10% strongly discouraged.

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 explicit 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" (only use age differences for filtering), 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, e.g. 'Discrete' (nonoverlapping generations), 'MinAgeParent', 'MaxAgeParent'.

Err

mtSame NEW matrix indicating whether individuals (might) have the same mitochon-

drial haplotype (1), and may thus be matrilineal relatives, or not (0). Row names and column names should match IDs in 'GenoM'. Not all individuals need to be included and order is not important. Please report any issues. For details see

the mtDNA vignette.

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().

StrictGenoCheck

Automatically exclude any individuals genotyped for <5 the unavoidable default up to version 2.4.1. Otherwise only excluded are (very nearly) monomorphic SNPs, SNPs scored for fewer than 2 individuals, and individuals scored for

fewer than 2 SNPs.

ErrFlavour function that takes Err (single number) as input, and returns a length 3 vector

or 3x3 matrix, or choose from inbuilt options 'version2.9', 'version2.0', 'version1.3', or 'version1.1', referring to the sequoia version in which they were the

default. Ignored if Err is a vector or matrix. See ErrToM for details.

MaxSibIter **DEPRECATED, use** Module number of iterations of sibship clustering, includ-

ing assignment 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.

MaxMismatch DEPRECATED AND IGNORED. Now calculated automatically using CalcMaxMismatch.

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

#### **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.

Full explanation of the various options and interpretation of the output is provided in the vignettes and on the package website, https://jiscah.github.io/index.html .

# Value

A list with some or all of the following components, depending on Module. All input except GenoM is included in the output.

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. (input) arguments used to specify age prior matrix. If a custom ageprior was args.AP provided via SeqList\$AgePrior, this matrix is returned instead 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. Dataframe, duplicated genotypes (with different IDs, duplicate IDs are not al-DupGenotype lowed). 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 (input) 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%). (input) Dataframe with sex and birth year data. All missing birth years are coded LifeHist as '-999', all missing sex as '3'. 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 Vector, IDs in genotype data for which no life history data is provided. NoLH Pedigree Dataframe with assigned genotyped and dummy parents from Sibship step; entries for dummy individuals are added at the bottom. Dataframe with assigned parents from Parentage step. PedigreePar 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. As AgePriors, but including columns for grandparents and avuncular pairs. NOT AgePriorExtra 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.

#### Genotyping error rate

The genotyping error rate Err can be specified three different ways:

- A single number, which is combined with ErrFlavour by ErrToM to create a length 3 vector (next item). By default (ErrFlavour = 'version2.9'), P(homlhom)=\$(E/2)^2\$, P(hetlhom)=\$E-(E/2)^2\$, P(homlhet)=\$E/2\$.
- a length 3 vector (NEW from version 2.6), with the probabilities to observe a actual homozygote as the other homozygote (homlhom), to observe a homozygote as heterozygote (hetlhom), and to observe an actual heterozygote as homozygote (homlhet). This assumes that the two alleles are equivalent with respect to genotyping errors, i.e. \$P(AAlaa) = P(aalAA)\$, \$P(aalAa)=P(AAlAa)\$, and \$P(aAlaa)=P(aAlAA)\$.
- a 3x3 matrix, with the probabilities of observed genotype (columns) conditional on actual genotype (rows). Only needed when the assumption in the previous item does not hold. See ErrToM for details.

#### (Too) Few Assignments?

Possibly Err is much lower than the actual genotyping error rate.

Alternatively, a true parent will not be assigned when it is:

- unclear who is the parent and who the offspring, due to unknown birth year for one or both individuals
- unclear whether the parent is the father or mother
- unclear if it is a parent or e.g. full sibling or grandparent, due to insufficient genetic data

And true half-siblings will not be clustered when it is:

- unclear if they are maternal or paternal half-siblings
- unclear if they are half-siblings, full avuncular, or grand-parental
- unclear what type of relatives they are due to insufficient genetic data

All pairs of non-assigned but likely/definitely relatives can be found with GetMaybeRel. For a method to do pairwise 'assignments', see https://jiscah.github.io/articles/pairLL\_classification.html; for further information, see the vignette.

#### 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.

#### Website

https://jiscah.github.io/

#### 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**

```
# === EXAMPLE 1: simulated data ===
head(SimGeno_example[,1:10])
head(LH_HSg5)
# parentage assignment:
SeqOUT <- sequoia(GenoM = SimGeno_example, Err = 0.005,</pre>
                  LifeHistData = LH_HSg5, Module="par", Plot=TRUE)
names(SeqOUT)
SeqOUT$PedigreePar[34:42, ]
# compare to true (or old) pedigree:
PC <- PedCompare(Ped_HSg5, SeqOUT$PedigreePar)</pre>
PC$Counts["GG",,]
# parentage assignment + full pedigree reconstruction:
# (note: this can be rather time consuming)
SeqOUT2 <- sequoia(GenoM = SimGeno_example, Err = 0.005,</pre>
                  LifeHistData = LH_HSg5, Module="ped", quiet="verbose")
SeqOUT2$Pedigree[34:42, ]
PC2 <- PedCompare(Ped_HSg5, SeqOUT2$Pedigree)</pre>
PC2$Counts["GT",,]
PC2$Counts[,,"dam"]
# different kind of pedigree comparison:
ComparePairs(Ped1=Ped_HSg5, Ped2=SegOUT$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</pre>
                  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>
```

78 SimGeno

```
SummarySeq(SeqOUT)
SeqOUT$notes <- "Trial run on cleaned data" # add notes for future reference
saveRDS(SeqOUT, file="sequoia_output_42.RDS") # save to R-specific file
writeSeq(SeqOUT, folder="sequoia_output") # save to several plain text files
# runtime:
SeqOUT$Specs$TimeEnd - SeqOUT$Specs$TimeStart
## End(Not run)</pre>
```

SimGeno

Simulate Genotypes

# Description

Simulate SNP genotype data from a pedigree, with optional missingness, genotyping errors, and non-genotyped parents.

# Usage

```
SimGeno(
   Pedigree,
   nSnp = 400,
   ParMis = c(0, 0),
   MAF = 0.3,
   CallRate = 0.99,
   SnpError = 5e-04,
   ErrorFV = function(E) c((E/2)^2, E - (E/2)^2, E/2),
   ErrorFM = NULL,
   ReturnStats = FALSE,
   quiet = FALSE
)
```

# Arguments

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

columns are ignored.

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. NOTE: default changed from

0.4 (up to version 2.8.5) to 0 (from version 2.9).

MAF either a single number with minimum minor allele frequency, and allele frequen

cies will be sampled uniformly between this minimum and 0.5, OR a vector with minor allele frequency at each locus. In both cases, this is the MAF among pedi-

gree founders, the MAF in the sample will deviate due to drift.

SimGeno 79

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

SnpError either a single value which will be combined with ErrorFV, or a length 3 vector

with probabilities (observed given actual) homlother hom, hetlhom, and homlhet; OR a vector or 3XnSnp matrix with the genotyping error rate(s) for each SNP.

ErrorFV function taking the error rate (scalar) as argument and returning a length 3 vec-

tor with hom->other hom, hom->het, het->hom. May be an 'ErrFlavour', e.g.

'version2.9'.

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). See below for details. To use, set ErrorFV = NULL

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

quantiles of MAF, error rate and call rates.

quiet suppress messages.

#### **Details**

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.

#### 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

# **Genotyping errors**

If SnpError is a length 3 vector, genotyping errors are generated following a length 3 vector with probabilities that 1) an actual homozygote is observed as the other homozygote, 2) an actual homozygote is observed as a heterozygote, and 3) an heterozygote is observed as an homozygote. The only assumption made is that the two alleles can be treated equally, i.e. observing actual allele \$A\$ as \$a\$ is as likely as observing actual \$a\$ as \$A\$.

80 SimGeno

If SnpError is a single value, by default this is interpreted as a locus-level 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 an = SnpError/2). The probability that one homozygote is observed as the other is  $(\text{SnpError}/2)^2$ . How this single value is rendered into a 3x3 error matrix is fully flexible and specified via ErrorFM; see link{ErrToM} for details.

The default values of SnpError=5e-4 and ErrorFM='version2.9' correspond to the length 3 vector  $c((5e-4/2)^2, 5e-4*(1-5e-4/2), 5e-4/2)$ .

A beta-distribution is used to simulate variation in the error rate between SNPs, the shape parameter of this distribution can be specified via MkGenoErrors. It is also possible to specify the error rate per SNP.

#### Call Rate

Variation in call rates across SNPs is assumed to follow a highly skewed (beta) distribution, with many SNPs 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. 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.

#### 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; ErrToM for more information about genotyping error patterns.

# **Examples**

SimGeno\_example 81

SimGeno\_example

Example genotype file: 'HSg5'

#### **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

```
LH_HSg5, SimGeno
```

82 SnpStats

SnpStats

SNP Summary Statistics

#### **Description**

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

#### Usage

```
SnpStats(
   GenoM,
   Pedigree = NULL,
   Duplicates = NULL,
   Plot = TRUE,
   quiet = TRUE,
   ErrFlavour
)
```

# **Arguments**

GenoM genotype matrix, in sequoia's format: 1 column per SNP, 1 row per individual,

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

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

genotyped individuals are ignored. Used to count Mendelian errors per SNP and

(poorly) estimate the error rate.

Duplicates dataframe with pairs of duplicated samples

Plot show histograms of the results?

quiet suppress messages

ErrFlavour DEPRECATED AND IGNORED. Was used to estimate Err.hat

#### **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)

SummarySeq 83

Mis Proportion of missing calls

HWE.p p-value from chi-square test for Hardy-Weinberg equilibrium

When a Pedigree is provided, there are 8 additional columns:

n.dam, n.sire, n.pair

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

OHdam, OHsire Count of number of opposing homozygous cases

MEpair Count of Mendelian errors, includes opposing homozygous cases when only one

parent is genotyped

n.dups, n.diff Number of duplicate pairs successfully genotyped for the SNP; number of dif-

ferences. The latter does not count cases where one duplicate is not successfully

genotyped at the SNP

#### 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.

# **Examples**

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"
)
```

84 SummarySeq

#### **Arguments**

SeaList the list returned by sequoia. Only elements 'Pedigree' or 'PedigreePar' and

'AgePriors' are used. All ids in 'PedigreePar', and only those, are presumed

genotyped.

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

umn names are ignored, as are additional columns, except for columns OHdam,

OHsire, MEpair, LLRdam, LLRsire, LLRpair (plotting only).

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 hermaphrodites.

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

is provided instead of SeqList, to distinguish between genetically assigned parents and 'observed' parents (e.g. observed in the field, or assigned previously using microsatellites). If NULL (the default), all parents are presumed observed.

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).

#### 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 an array with the number of assigned parents, split by:

> • offspringCat: Genotyped, Dummy, or Observed\* (\*: only when Pedigree is provided rather than SegList, for ids which are not listed in SNPd and do not conform to DumPrefix + number (i.e. (almost) al individuals when SNPd = NULL. the default).

> • offspringSex: Female, Male, Unknown, or Herm\* (\*: hermaphrodite, only if any individuals occur as both dam and sire). Based only on whether an individual occurs as Dam or Sire.

• parentSex: Dam or Sire

• parentCat: Genotyped, Dummy, Observed\*, or None (\*: as for offspring-

Cat)

**GPCount** an array with the number of assigned grandparents, split by:

• offspringCat: Genotyped, Dummy, Observed\*, or All

• grandparent kind: maternal grandmothers (MGM), maternal grandfathers (MGF), paternal grandmothers (PGM), paternal grandfathers (PGF)

writeColumns 85

• grandparentCat: Genotyped, Dummy, Observed\*, or None

SibSize

a list with elements 'mat' (maternal half + full siblings), 'pat' (paternal half + full siblings), and 'full' (full siblings). Each 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

PlotSeqSum to plot the output of this function; sequoia for pedigree reconstruction and links to other functions.

# **Examples**

```
SummarySeq(Ped_griffin)
sumry_grif <- SummarySeq(SeqOUT_griffin, Panels=c("G.parents", "OH"))
sumry_grif$PedSummary</pre>
```

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.

# 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 $\boldsymbol{x}$ are to be written along with $\boldsymbol{x}$ . |
| col.names | a logical value indicating whether the column names of x are to be written along with x.                             |

86 writeSeq

| iteSeq 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 **openxlsx**).

# 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'.

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.

writeSeq 87

# **Examples**

# **Index**

| * datasets                                   | DyadCompare, 18, 21  |  |
|--|--|--|
| Conf_griffin, 20                             | by a de compare, 10, 21  |  |
| FieldMums_griffin, 30                        | ErrToM, 4, 7, 11, 22, 30, 42, 73, 75, 80                           |  |
| Geno_griffin, 35                             | EstConf, 20, 25, 60, 76, 80  |  |
| Geno_HSg5, 36                                | EstEr, 29  |  |
| Inherit_patterns, 47                         |  |  |
| LH_griffin, 49                               | FieldMums_griffin, 30  |  |
| LH_HSg5, 50                                  | FindFamilies, 31, 39, 62   |  |
| MaybeRel_griffin, 54                         | fread, <i>33</i>   |  |
| Ped_griffin, 63                              |  |  |
| Ped_HSg5, 63                                 | genlight, 32   |  |
| SeqOUT_griffin, 69                           | Geno_griffin, 20, 35, 54, 69                                       |  |
| SeqOUT_HSg5, 70                              | Geno_HSg5, 36, 70  |  |
| SimGeno_example, 81                          | GenoConvert, 6, 8, 10, 32, 41, 49, 62, 71, 76,                     |  |
| * inherit                                    | 83   |  |
| Inherit_patterns, 47                         | GetAncestors, 32, 36, 39   |  |
| * sequoia                                    | getAssignCat, 7, 8, 12, 37, 57, 58, 60                             |  |
| Conf_griffin, 20                             | GetDescendants, 32, 38, 39   |  |
| FieldMums_griffin, 30                        | getGenerations, 32, 39, 61, 67                                     |  |
| Geno_griffin, 35                             | GetLLRAge, 40  |  |
| Geno_HSg5, 36                                | GetMaybeRel, 13, 17, 18, 31, 41, 45, 54, 55,                       |  |
| Inherit_patterns, 47                         | 73, 76, 86<br>GetRelM, 13, 19, 20, 44, 45, 67, 68, 76              |  |
| LH_griffin, 49                               | GetReim, 13, 19, 20, 44, 43, 07, 08, 70                            |  |
| LH_HSg5, 50                                  | <pre>Inherit_patterns, 47</pre>                                    |  |
| MaybeRel_griffin, 54                         | invisible, 16, 68  |  |
| Ped_griffin, 63                              | 12020, 10, 00  |  |
| Ped_HSg5, 63                                 | kinship, <i>14</i> , <i>61</i>                                     |  |
| SeqOUT_griffin, 69                           |  |  |
| SeqOUT_HSg5, 70                              | layout, <i>64</i>  |  |
| SimGeno_example, 81                          | LH_griffin, 49, <i>63</i>  |  |
| ormoeno_exampre, or                          | LH_HSg5, <i>36</i> , <i>50</i> , <i>64</i> , <i>70</i> , <i>81</i> |  |
| CalcBYprobs, 3, 74, 76                       | LHConvert, <i>34</i> , 48, <i>62</i>                               |  |
| CalcMaxMismatch, 4, 13, 43, 73               |  |  |
| Calcohllr, 5, 12, 13, 16, 37, 60, 73, 76, 83 | MakeAgePrior, 3, 4, 7, 11, 42, 50, 64, 72–74,                      |  |
| CalcPairLL, 8, 9, 44, 65, 75, 76             | 76   |  |
| CalcRped, 14                                 | MaybeRel_griffin, 54   |  |
| CheckGeno, 8, 15, 34, 38, 76, 83             | MkGenoErrors, 55, 80   |  |
| ComparePairs, 17, 21, 47, 59, 60, 76         | paste, <i>33</i>   |  |
| Conf_griffin, 20                             | Ped_griffin, 20, 31, 35, 49, 54, 63, 69                            |  |
| COIII _BI 11 1 111, 20                       | 1 cu_g; 11 1111, 20, 31, 33, 43, 34, 03, 09                        |  |

INDEX 89

```
Ped_HSg5, 36, 50, 63, 63, 70, 81
PedCompare, 17, 20, 21, 25, 27, 30, 37, 38, 56,
         66, 76, 86
PedPolish, 8, 31, 46, 60
PedStripFID, 33, 49, 62
PlotAgePrior, 54, 64, 76
PlotPairLL, 11, 13, 65
PlotPedComp, 66
PlotRelPairs, 20, 47, 67
PlotSeqSum, 68, 85
read.table, 34
readLines, 33, 34
SeqOUT_griffin, 20, 31, 49, 55, 63, 69
Seq0UT_HSg5, 36, 70
sequoia, 5, 7, 8, 11, 16, 20, 25, 27, 31, 37,
         40-42, 44, 48, 50, 54, 63, 64, 69, 70,
         70, 84–86
SimGeno, 25, 27, 29, 35, 36, 48, 76, 78, 81
SimGeno_example, 64, 81
SnpStats, 5, 15, 16, 34, 73, 76, 82
strsplit, 33, 34
SummarySeq, 8, 64, 68, 69, 73, 76, 83
system.time, 26
write.table, 33
writeColumns, 85, 86
writeSeq, 76, 86
```