# Package 'forrel'

July 22, 2025

**Title** Forensic Pedigree Analysis and Relatedness Inference Version 1.8.1 **Description** Forensic applications of pedigree analysis, including likelihood ratios for relationship testing, general relatedness inference, marker simulation, and power analysis. 'forrel' is part of the 'pedsuite', a collection of packages for pedigree analysis, further described in the book 'Pedigree Analysis in R' (Vigeland, 2021, ISBN:9780128244302). Several functions deal specifically with power analysis in missing person cases, implementing methods described in Vigeland et al. (2020) <doi:10.1016/j.fsigen.2020.102376>. Data import from the 'Familias' software (Egeland et al. (2000) <doi:10.1016/S0379-0738(00)00147-X>) is supported through the 'pedFamilias' package. License GPL (>= 2)URL https://github.com/magnusdv/forrel BugReports https://github.com/magnusdv/forrel/issues **Depends** pedtools (>= 2.8.1), R (>= 4.2.0) **Imports** glue, phapply, pedprobr (>= 1.0.0), ribd (>= 1.7.1), verbalisr (>=0.7.1)Suggests ggplot2, ggrepel, ibdsim2, plotly, poibin, scales, testthat **Encoding** UTF-8 Language en-GB LazyData true RoxygenNote 7.3.2 NeedsCompilation no **Author** Magnus Dehli Vigeland [aut, cre] (ORCID: <a href="https://orcid.org/0000-0002-9134-4962">https://orcid.org/0000-0002-9134-4962</a>), Thore Egeland [ctb] Maintainer Magnus Dehli Vigeland <m.d.vigeland@medisin.uio.no> **Repository** CRAN **Date/Publication** 2025-07-21 20:00:12 UTC

Type Package

2 checkPairwise

# **Contents**

check	Pairwise Check pedigree data for relationship errors	
muca		7
Index		49
	simpleSim	47
	showInTriangle	46
	rankProfiles	45
	randomPersonEP	44
	quickLR	43
	profileSimParametric	42
	profileSim	41
	powerPlot	38
	NorwegianFrequencies	36
	MPPsims	34
	missingPersonPlot	32
	missingPersonLR	30
	missingPersonIP	28
	missingPersonEP	27
	markerSimParametric	25
	markerSim	23
	LRpower	21
	kinshipLR	18
	ibdLoglik	17
	ibdEstimate	14
	ibdBootstrap	12
	FORCE	11
	findExclusions	10
	familias	10
	exclusionPower	5
	checkPairwise	2

# Description

The checkPairwise() function provides a convenient way to check for pedigree errors, given the available marker data. The function calls <code>ibdEstimate()</code> to estimate IBD coefficients for all pairs of typed pedigree members, and uses the estimates to test for potential errors. By default, the results are shown in a colour-coded plot (based on <code>ribd::ibdTriangle())</code> where unlikely relationships are easy to spot.

checkPairwise 3

#### Usage

```
checkPairwise(
  ids = typedMembers(x),
  includeInbred = FALSE,
  acrossComps = TRUE,
  plotType = c("base", "ggplot2", "plotly", "none"),
 GLRthreshold = 1000,
  pvalThreshold = NULL,
 nsim = 0,
  seed = NULL,
  legendData = NULL,
 plot = TRUE,
 verbose = TRUE,
  excludeInbred = NULL,
)
plotCP(
  cpRes = NULL,
  plotType = c("base", "ggplot2", "plotly"),
  labels = FALSE,
  legendData = NULL,
  errtxt = "Potential error",
  seed = NULL,
)
```

# **Arguments**

X	A ped object or a list of such.

ids A vector of ID labels; the individuals to include in the check. Default: All typed

members of x.

includeInbred A logical, by default FALSE, indicating if inbred individuals should be excluded

from the analysis.

acrossComps A logical indicating if pairs of individuals in different components should be

considered. Default: TRUE.

plotType Either "base" (default), "ggplot2", "plotly" or "none". Abbreviations are al-

lowed.

GLRthreshold A positive number, by default 1000. Threshold for the generalised likelihood

ratio (see Details). Scores exceeding this are flagged as potential errors in the

output table and encircled in the plot.

pvalThreshold A positive number, or NULL (default). If given, this is used instead of GLRthreshold

to identify potential errors. Ignored if nsim = 0.

nsim A nonnegative number; the number of simulations used to estimate p-values. If

0 (default), this step is skipped.

4 checkPairwise

seed An integer seed for the random number generator (optional, and only relevant if

nsim > 0).

legendData A data frame with columns k0, k2, lab, col and shape.

plot Deprecated. To suppress the triangle plot, use plotType = "none".

verbose A logical.

excludeInbred Deprecated; renamed to 'includeInbred'.

... Further parameters passed on to ribd::ibdTriangle().

cpRes A data frame: the output from checkPairwise().

labels A logical (default: FALSE). If TRUE, labels are included in the IBD triangle

plot.

errtxt A character string to use for the error legend.

#### **Details**

To identify potential pedigree errors, the function calculates the *generalised likelihood ratio* (GLR) for each pairwise relationship, as explained by Egeland & Vigeland (2025). This compares the likelihood of the estimated coefficients with that of the coefficients implied by the pedigree. By default, relationships whose GLR exceed 1000 are flagged as errors and shown with a circle in the plot. Alternatively, if arguments nsim and pvalThreshold are supplied, the p-value of each score is estimated by simulation, and used as threshold for calling errors.

By default, inbred individuals are excluded from the analysis, since pairwise relationships involving inbred individuals have undefined kappa coefficients (and therefore no position in the triangle). In some cases it may still be informative to include their estimates; set includeInbred = TRUE to enforce this.

## Value

If plotType is "none" or "base": A data frame containing both the estimated and pedigree-based IBD coefficients for each pair of typed individuals. The last columns (GLR, pval and err) contain test results using the GLR scores to identify potential pedigree errors.

If plotType is "ggplot2" or "plotly", the plot objects are returned.

#### References

T. Egeland and M.D. Vigeland, *Kinship cases with partially specified hypotheses*. Forensic Science International: Genetics 78 (2025). doi:10.1016/j.fsigen.2025.103270

#### See Also

```
ibdEstimate().
```

```
### Example with realistic data
x = avuncularPed() |>
profileSim(markers = NorwegianFrequencies, seed = 1729)
```

exclusionPower 5

```
checkPairwise(x)
### Create an error: sample swap 1 <-> 3
als = getAlleles(x)
als[c(1,3), ] = als[c(3,1), ]
y = setAlleles(x, alleles = als)
checkPairwise(y)
# Using p-values instead of GLR
nsim = 10 # increase!
checkPairwise(y, nsim = nsim, pvalThreshold = 0.05)
# Plot can be done separately
res = checkPairwise(y, nsim = nsim, pvalThreshold = 0.05, plotType = "none")
plotCP(res, plotType = "base", errtxt = "Not good!")
# Combined plot of pedigree and check results
dev.new(height = 5, width = 8, noRStudioGD = TRUE)
layout(rbind(1:2), widths = 2:3)
plot(y, margins = 2, title = "Swapped 1 - 3")
plotCP(res, labels = TRUE)
```

exclusionPower

Power of exclusion

# Description

Computes the power (of a single marker, or for a collection of markers) of excluding a claimed relationship, given the true relationship.

#### Usage

```
exclusionPower(
  claimPed,
  truePed,
  ids,
  markers = NULL,
  source = "claim",
  disableMutations = NA,
  exactMaxL = Inf,
  nsim = 1000,
  seed = NULL,
  alleles = NULL,
  afreq = NULL,
```

6 exclusionPower

```
knownGenotypes = NULL,
  Xchrom = FALSE,
  plot = FALSE,
 plotMarkers = NULL,
  verbose = TRUE
)
```

#### **Arguments**

claimPed A ped object (or a list of such), describing the claimed relationship. If a list, the

sets of ID labels must be disjoint, that is, all ID labels must be unique.

truePed A ped object (or a list of such), describing the true relationship. ID labels must

be consistent with claimPed.

ids Individuals available for genotyping.

markers A vector indicating the names or indices of markers attached to the source pedi-

gree. If NULL (default), then all markers attached to the source pedigree are used. If alleles or afreq is non-NULL, then this parameter is ignored.

Either "claim" (default) or "true", deciding which pedigree is used as source for source

marker data.

disableMutations

This parameter determines how mutation models are treated. Possible values are as follows:

- NA (the default): Mutations are disabled only for those markers whose known genotypes are compatible with both claimPed and truePed. This is determined by temporarily removing all mutation models and checking which markers have nonzero likelihood in both alternatives.
- TRUE: Mutations are disabled for all markers.
- FALSE: No action is done to disable mutations.
- A vector containing the names or indices of those markers for which mutations should be disabled.

exactMaxL

A positive integer, or Inf (default). Exact EPs are calculated for markers whose number of alleles is less or equal to exactMaxL; remaining markers are handled by simulation.

A positive integer; the number of simulations used for markers whose number nsim

of alleles exceeds exactMaxL.

An integer seed for the random number generator (optional). seed

alleles, afreq, Xchrom

If these are given, they are used (together with knownGenotypes) to create a marker object on the fly.

knownGenotypes A list of triplets (a, b, c), indicating that individual a has genotype b/c.

Ignored unless alleles or afreq is non-NULL.

plot Either a logical or the character "plotOnly". If the latter, a plot is drawn, but no

further computations are done.

plotMarkers A vector of marker names or indices whose genotypes are to be included in the

plot.

A logical. verbose

exclusionPower 7

#### **Details**

This function implements the formula for exclusion power as defined and discussed in (Egeland et al., 2014).

It should be noted that claimPed and truePed may be any (lists of) pedigrees, as long as they both contain the individuals specified by ids. In particular, either alternative may have inbred founders (with the same or different coefficients), but this must be set individually for each.

#### Value

If plot = "plot0nly", the function returns NULL after producing the plot.

Otherwise, the function returns an EPresult object, which is essentially a list with the following entries:

- EPperMarker: A numeric vector containing the exclusion power of each marker. If the known genotypes of a marker are incompatible with the true pedigree, the corresponding entry is NA.
- EPtotal: The total exclusion power, computed as 1 prod(1 EPperMarker, na.rm = TRUE).
- expectedMismatch: The expected number of markers giving exclusion, computed as sum(EPperMarker, na.rm = TRUE).
- distribMismatch: The probability distribution of the number of markers giving exclusion. This is given as a numeric vector of length n+1, where n is the number of nonzero elements of EPperMarker. The vector has names 0:n.
- time: The total computation time.
- params: A list containing the (processed) parameters ids, markers and disableMutations.

#### Author(s)

Magnus Dehli Vigeland

# References

T. Egeland, N. Pinto and M.D. Vigeland, *A general approach to power calculation for relationship testing*. Forensic Science International: Genetics 9 (2014). doi:10.1016/j.fsigen.2013.05.001

8 expectedLR

```
# Truth: 'AF' and 'CH' are unrelated
true = singletons(c("AF", "CH"))
# EP when both are available for genotyping
exclusionPower(claim, true, ids = c("AF", "CH"))
# EP when the child is typed; homozygous 1/1 at both markers
claim2 = claim |>
 setGenotype(marker = 1:2, id = "CH", geno = "1/1")
exclusionPower(claim2, true, ids = "AF")
### Two females claim to be mother and daughter, but are in reality sisters.
### We compute the power of various markers to reject the claim.
ids = c("A", "B")
claim = nuclearPed(father = "NN", mother = "A", children = "B", sex = 2)
true = nuclearPed(children = ids, sex = 2)
# SNP with MAF = 0.1:
PE1 = exclusionPower(claimPed = claim, truePed = true, ids = ids,
                    alleles = 1:2, afreq = c(0.9, 0.1))
stopifnot(round(PE1$EPtotal, 5) == 0.00405)
# Tetra-allelic marker with one major allele:
PE2 = exclusionPower(claimPed = claim, truePed = true, ids = ids,
                    alleles = 1:4, afreq = c(0.7, 0.1, 0.1, 0.1)
stopifnot(round(PE2$EPtotal, 5) == 0.03090)
### How does the power change if the true pedigree is inbred?
trueLOOP = halfSibPed(sex2 = 2) |> addChildren(4, 5, ids = ids)
# SNP with MAF = 0.1:
PE3 = exclusionPower(claimPed = claim, truePed = trueLOOP, ids = ids,
                    alleles = 1:2, afreq = c(0.9, 0.1))
# Power almost doubled compared with PE1
stopifnot(round(PE3$EPtotal, 5) == 0.00765)
```

expectedLR 9

## **Description**

This function computes the expected LR for a single marker, in a kinship test comparing two hypothesised relationships between a set of individuals. The true relationship may differ from both hypotheses. Some individuals may already be genotyped, while others are available for typing. The implementation uses oneMarkerDistribution() to find the joint genotype distribution for the available individuals, conditional on the known data, in each pedigree.

## Usage

```
expectedLR(numeratorPed, denominatorPed, truePed = numeratorPed, ids, marker)
```

### **Arguments**

numeratorPed A ped object.
denominatorPed A ped object.
truePed A ped object.

ids A vector of ID labels corresponding to untyped pedigree members. (These must

be members of all three input pedigrees).

marker either a marker object compatible with numeratorPed, or the name or index of

a marker attached to numeratorPed.

#### Value

A positive number.

```
#------
# Curious example showing that ELR may decrease
# by typing additional reference individuals
#------
# Numerator ped
numPed = nuclearPed(father = "fa", mother = "mo", child = "ch")
# Denominator ped: fa, mo, ch are unrelated. (Hack!)
denomPed = halfSibPed() |> relabel(old = 1:3, new = c("mo", "fa", "ch"))
# Scenario 1: Only mother is typed; genotype 1/2
p = 0.9
m1 = marker(numPed, mo = "1/2", afreq = c("1" = p, "2" = 1-p))
expectedLR(numPed, denomPed, ids = "ch", marker = m1)

1/(8*p*(1-p)) + 1/2 # exact formula
# Scenario 2: Include father, with genotype 1/1
m2 = m1
genotype(m2, id = "fa") = "1/1"
expectedLR(numPed, denomPed, ids = "ch", marker = m2)
```

10 findExclusions

```
1/(8*p*(1-p)) + 1/(4*p^2) # exact formula
```

familias

Import/export from Familias

### **Description**

Functions for reading .fam files associated with the Familias software for forensic kinship computations.

**Deprecated** These functions have been moved to a separate package, pedFamilias, and will be removed from forrel in a future version.

#### Usage

```
readFam(...)
```

## **Arguments**

... Arguments passed on to the respective pedFamilias function.

findExclusions

Find markers excluding an identification

# Description

Find markers for which the genotypes of a candidate individual is incompatible with a pedigree

# Usage

```
findExclusions(x, id, candidate, removeMut = TRUE)
```

## **Arguments**

x A ped object or a list of such.

id A character of length 1; the name of an untyped member of x.

candidate A singleton pedigree, with genotypes for the same markers as x.

removeMut A logical. If TRUE (default), all mutations models are stripped.

### Value

A character vector containing the names of incompatible markers.

FORCE 11

#### **Examples**

```
db = NorwegianFrequencies[1:5]

# Pedigree with 3 siblings; simulate data for first two
x = nuclearPed(3) |>
    profileSim(ids = 3:4, markers = db, seed = 1)

# Simulate random person
poi = singleton("POI") |>
    profileSim(markers = db, seed = 1)

# Identify incompatible markers
findExclusions(x, id = 5, candidate = poi) # D21S11

# Inspect
plotPedList(list(x, poi), marker = "D21S11", hatched = typedMembers)
```

**FORCE** 

FORCE panel SNP data

# **Description**

Data frames describing the FORCE panel of SNPs for forensic genetics (Tillmar et al., 2021). We provide here two subsets of the complete panel: the autosomal kinship SNPs (FORCE, n = 3930) and the X-chromosomal SNPs (XFORCE, n = 246). To attach the markers to a pedigree, use pedtools::setSNPs() (see Examples).

#### Usage

**FORCE** 

**XFORCE** 

# **Format**

Both FORCE and XFORCE are data frames with the following columns:

- CHROM: Chromosome
- MARKER: Marker name (rs number)
- MB: Physical position in megabases (GRCh38)
- A1: First allele
- A2: Second allele
- FREQ1: Allele frequency of A1

12 ibdBootstrap

#### **Details**

Allele frequencies were retrieved from Ensembl using the REST API, with the population 1000GENOMES: phase\_3: ALL as primary source. For 9 SNPs where this was unavailable, gnomADg: ALL was used instead.

One SNP - rs2323964 - was excluded due to lack of Ensembl/dbSNP support.

For details, the code used to download and process the data is available in the data-raw folder on GitHub: https://github.com/magnusdv/forrel/tree/master/data-raw

*Note*: The autosomal dataset (FORCE) was updated in version 1.8.1, adding 15 markers that were previously missing and revising some frequencies. The previous version is available via:

```
pth = system.file("extdata/FORCE_old.txt", package = "forrel")
oldforce = read.table(pth, header = TRUE)
```

#### Source

Tillmar et al. The FORCE Panel: An All-in-One SNP Marker Set for Confirming Investigative Genetic Genealogy Leads and for General Forensic Applications. Genes. (2021)

# **Examples**

```
x = setSNPs(nuclearPed(), snpData = FORCE)
summary(x)
getMap(x, markers = 1:3)
getFreqDatabase(x, markers = 1:3)
```

ibdBootstrap

Bootstrap estimation of IBD coefficients

# **Description**

This function produces (parametric or nonparametric) bootstrap estimates of the IBD coefficients between two individuals; either the three  $\kappa$  -coefficients or the nine condensed identity coefficients  $\Delta$  (see ibdEstimate()).

## Usage

```
ibdBootstrap(
  x = NULL,
  ids = NULL,
  param = NULL,
  kappa = NULL,
  delta = NULL,
  N,
  method = "parametric",
  freqList = NULL,
```

ibdBootstrap 13

```
plot = TRUE,
  seed = NULL
)
```

#### **Arguments**

Χ

extract the allele frequencies, and can be skipped if freqList is provided.

A pair of ID labels.

param Either NULL (default), "kappa" or "delta". (See below.)

kappa, delta Probability vectors of length 3 (kappa) or 9 (delta). Exactly one of param, kappa and delta must be non-NULL. If kappa and delta are both NULL, the appropriate set of coefficients is computed as ibdEstimate(x, ids, param).

N The number of simulations.

method Either "parametric" (default) or "nonparametric". Abbreviations are allowed. see Details for more information about each method.

A ped object, or a list of such. If method = "parametric", this is only used to

freqList A list of probability vectors: The allele frequencies for each marker.

plot A logical, only relevant for bootstraps of kappa. If TRUE, the bootstrap esti-

mates are plotted in the IBD triangle.

seed An integer seed for the random number generator (optional).

#### **Details**

The parameter method controls how bootstrap estimates are obtained in each replication:

- "parametric": new profiles for two individuals are simulated from the input coefficients, followed by a re-estimation of the coefficients.
- "nonparametric": the original markers are sampled with replacement, before the coefficients are re-estimated.

Note that the pedigree itself does not affect the output of this function; the role of x is simply to carry the marker data.

#### Value

A data frame with N rows containing the bootstrap estimates. The last column, dist, gives the Euclidean distance to the original coefficients (either specified by the user or estimated from the data), viewed as a point in  $R^3$  (kappa) or  $R^9$  (delta).

#### See Also

```
ibdEstimate()
```

14 ibdEstimate

#### **Examples**

```
# Frequency list of 15 standard STR markers
freqList = NorwegianFrequencies[1:15]
# Number of bootstrap simulations (increase!)
N = 5
# Bootstrap estimates for kappa of full siblings
boot1 = ibdBootstrap(kappa = c(0.25, .5, .25), N = N, freqList = freqList)
boot1
# Mean deviation
mean(boot1$dist)
# Same, but with the 9 identity coefficients.
delta = c(0, 0, 0, 0, 0, 0, .25, .5, .25)
boot2 = ibdBootstrap(delta = delta, N = N, freqList = freqList)
# Mean deviation
mean(boot2$dist)
#### Non-parametric bootstrap.
# Requires `x` and `ids` to be provided
x = nuclearPed(2)
x = markerSim(x, ids = 3:4, N = 50, alleles = 1:10, seed = 123)
bootNP = ibdBootstrap(x, ids = 3:4, param = "kappa", method = "non", N = N)
# Parametric bootstrap can also be done with this syntax
bootP = ibdBootstrap(x, ids = 3:4, param = "kappa", method = "par", N = N)
```

ibdEstimate

Pairwise relatedness estimation

## **Description**

Estimate the IBD coefficients  $\kappa = (\kappa_0, \kappa_1, \kappa_2)$  or the condensed identity coefficients  $\Delta = (\Delta_1, ..., \Delta_9)$  between a pair (or several pairs) of pedigree members, using maximum likelihood methods. Estimates of  $\kappa$  may be visualised with showInTriangle().

# Usage

```
ibdEstimate(
    x,
    ids = typedMembers(x),
    param = c("kappa", "delta"),
    acrossComps = TRUE,
```

ibdEstimate 15

```
markers = NULL,
start = NULL,
tol = sqrt(.Machine$double.eps),
beta = 0.5,
sigma = 0.5,
contourPlot = FALSE,
levels = NULL,
maxval = FALSE,
verbose = TRUE
)
```

# **Arguments**

X	A ped object or a list of such.
ids	Either a vector with ID labels, or a data frame/matrix with two columns, each row indicating a pair of individuals. The entries are coerced to characters, and must match uniquely against the ID labels of x. By default, all pairs of genotyped members of x are included.
param	Either "kappa" (default) or "delta"; indicating which set of coefficients should be estimated.
acrossComps	A logical indicating if pairs of individuals in different components should be included. Default: TRUE.
markers	A vector with names or indices of markers attached to x, indicating which markers to include. By default, all markers are used.
start	A probability vector (i.e., with nonnegative entries and sum 1) of length 3 (if param = "kappa") or 9 (if param = "delta"), indicating the initial value of for the optimisation. By default, start is set to (1/3, 1/3, 1/3) if param = "kappa" and (1/9,, 1/9) if param = "delta".
tol, beta, sigma	Control parameters for the optimisation routine; can usually be left untouched.
contourPlot	A logical. If TRUE, contours of the log-likelihood function are plotted overlaying the IBD triangle.
levels	(Only relevant if contourPlot = TRUE.) A numeric vector of levels at which to draw contour lines. If NULL (default), the levels are chosen automatically.
maxval	A logical. If TRUE, the maximum log-likelihood value is included in the output. Default: FALSE
verbose	A logical.

#### **Details**

It should be noted that this procedure estimates the *realised* identity coefficients of each pair, i.e., the actual fractions of the autosomes in each IBD state. These may deviate substantially from the theoretical pedigree coefficients.

Maximum likelihood estimation of relatedness coefficients originates with Thompson (1975). Optimisation of  $\kappa$  is done in the  $(\kappa_0, \kappa_2)$ -plane and restricted to the triangle defined by

$$\kappa_0 \ge 0, \kappa_2 \ge 0, \kappa_0 + \kappa_2 \le 1$$

16 ibdEstimate

Optimisation of  $\Delta$  is done in unit simplex of  $\mathbb{R}^8$ , using the first 8 coefficients.

The implementation optimises the log-likelihood using a projected gradient descent algorithm, combined with a version of Armijo line search.

When param = "kappa", the output may be fed directly to showInTriangle() for visualisation.

#### Value

An object of class ibdEst, which is basically a data frame with either 6 columns (if param = "kappa") or 12 columns (if param = "delta"). The first three columns are id1 (label of first individual), id2 (label of second individual) and N (the number of markers with no missing alleles). The remaining columns contain the coefficient estimates. If maxval = T, a column named maxloglik is added at the end.

#### Author(s)

Magnus Dehli Vigeland

#### References

- E. A. Thompson (1975). *The estimation of pairwise relationships*. Annals of Human Genetics 39.
- E. A. Thompson (2000). *Statistical Inference from Genetic Data on Pedigrees*. NSF-CBMS Regional Conference Series in Probability and Statistics. Volume 6.

#### See Also

```
ibdBootstrap()
```

```
### Example 1: Siblings

# Create pedigree and simulate 100 markers
x = nuclearPed(2) |> markerSim(N = 100, alleles = 1:4, seed = 123)
x

# Estimate kappa (expectation: (0.25, 0.5, 0.25)
k = ibdEstimate(x, ids = 3:4)
k

# Visualise estimate
showInTriangle(k, labels = TRUE)

# Contour plot of the log-likelihood function
ibdEstimate(x, ids = 3:4, contourPlot = TRUE)

### Example 2: Full sib mating
y = fullSibMating(1) |>
markerSim(ids = 5:6, N = 1000, alleles = 1:10, seed = 123)
```

ibdLoglik 17

```
# Estimate the condensed identity coefficients
ibdEstimate(y, param = "delta")

# Exact coefficient by `ribd`:
ribd::condensedIdentity(y, 5:6, simplify = FALSE)
```

ibdLoglik

Pairwise IBD likelihood

#### **Description**

Given genotype data from two individuals, computes the log-likelihood of a single set of IBD coefficients, either kappa =  $(\kappa_0, \kappa_1, \kappa_2)$  or the Jacquard coefficients delta =  $(\Delta_1, ..., \Delta_9)$ . The ibdLoglikFUN version returns an efficient *function* for computing such likelihoods, suitable for optimisations such as in ibdEstimate().

## Usage

```
ibdLoglik(x = NULL, ids = NULL, kappa = NULL, delta = NULL)
ibdLoglikFUN(x, ids, input = c("kappa", "kappa02", "delta"))
```

# Arguments

Χ	A ped object or a list of such.
ids	A vector of ID labels.
kappa	A probability vector of length 3.
delta	A probability vector of length 9.
input	Either "kappa", "kappa02" or "delta". See Value.

#### Value

ibdLoglik() returns a single number; the total log-likelihood over all markers included.

ibdLoglikFUN() returns a function for computing such log-likelihoods. The function takes a single input vector p, whose interpretation depends on the input parameter:

- "kappa": p is expected to be a set of kappa coefficients  $(\kappa_0, \kappa_1, \kappa_2)$ .
- "kappa02": p should be a vector of length 2 containing the coefficients  $\kappa_0$  and  $\kappa_2$ . This is sometimes a convenient shortcut when working in the IBD triangle.
- "delta": Expects p to be a set of condensed Jacquard coefficients  $(\Delta_1, ..., \Delta_9)$ .

18 kinshipLR

## **Examples**

```
# Siblings typed with 10 markers
x = nuclearPed(2) |> markerSim(N = 10, alleles = 1:4)
# Calculate log-likelihood at a single point
k = c(0.25, 0.5, 0.25)
ibdLoglik(x, ids = 3:4, kappa = k)
# Or first get a function, and then apply it
llFun = ibdLoglikFUN(x, ids = 3:4, input = "kappa")
llFun(k)
```

kinshipLR

Likelihood ratios for kinship testing

# **Description**

This function computes likelihood ratios (LRs) for a list of pedigrees. One of the pedigrees (the last one, by default) is designated as 'reference', to be used in the denominator in all LR calculations. To ensure that all pedigrees use the same data set, one of the pedigrees may be chosen as 'source', from which data is transferred to all the other pedigrees.

# Usage

```
kinshipLR(
    ...,
    ref = NULL,
    source = NULL,
    markers = NULL,
    likArgs = NULL,
    linkageMap = NULL,
    keepMerlin = NULL,
    verbose = FALSE
)
```

# Arguments

Pedigree alternatives. Each argument should be either a single ped object or a list of such. The pedigrees may be named; otherwise they are assigned names "H1", "H2", ... automatically.

It is also possible to pass a single list containing all the pedigrees.

ref

An index or name indicating which of the input pedigrees should be used as "reference pedigree", i.e., used in the denominator of each LR. If NULL (the default), the last pedigree is used as reference.

kinshipLR 19

An index or name designating one of the input pedigrees as source for marker data. If given, marker data is transferred from this to all the other pedigrees (replacing any existing markers). The default action (source = NULL) is as follows: If all pedigree have attached markers, no transfers are done. If exactly one of the pedigrees have attached markers, these are transferred to the others. all other cases give an error.

A vector of marker names or indices indicating which markers should be in-

cluded. If NULL (the default) all markers are used.

likArgs An optional list of arguments to be passed to pedprobr::likelihood(), e.g.

likArgs = list(special = TRUE).

linkageMap If this is non-NULL, the markers are interpreted as being linked, and likelihoods

will be computed by an external call to MERLIN.

The supplied object should be either:

• a data frame, whose first three columns must be (i) chromosome (ii) marker name (iii) centiMorgan position, or

• a map object created with ibdsim2::uniformMap() or ibdsim2::loadMap(). This will internally be applied to the attached markers to produce a suitable data frame as above.

keepMerlin Either NULL (default) or the path to an existing folder. If given, MERLIN files

are stored here, typically for debugging purposes.

verbose A logical.

#### **Details**

markers

By default, all markers are assumed to be unlinked. To accommodate linkage, a genetic map may be supplied with the argument linkageMap. This requires the software MERLIN to be installed.

#### Value

A LRresult object, which is essentially a list with entries

- LRtotal: A vector of length L, where L is the number of input pedigrees. The i'th entry is the total LR (i.e., the product over all markers) comparing pedigree i to the reference pedigree. The entry corresponding to the reference will always be 1.
- LRperMarker: A numerical matrix, where the i'th column contains the marker-wise LR values comparing pedigree i to the reference. The product of all entries in a column should equal the corresponding entry in LRtotal.
- likelihoodsPerMarker: A numerical matrix of the same dimensions as LRperMarker, but where the entries are likelihood of each pedigree for each marker.
- time: Elapsed time

#### Author(s)

Magnus Dehli Vigeland and Thore Egeland

### See Also

LRpower(), pedprobr::likelihood(), pedprobr::likelihoodMerlin()

20 kinshipLR

```
### Example 1: Full vs half sibs
# Simulate 5 markers for a pair of full sibs
ids = c("A", "B")
sibs = nuclearPed(children = ids)
sibs = simpleSim(sibs, N = 5, alleles = 1:4, ids = ids, seed = 123)
# Create two alternative hypotheses
halfsibs = relabel(halfSibPed(), old = 4:5, new = ids)
unrel = singletons(c("A", "B"))
# Compute LRs. By default, the last ped is used as reference
kinshipLR(sibs, halfsibs, unrel)
# Input pedigrees can be named, reflected in the output
kinshipLR(S = sibs, H = halfsibs, U = unrel)
# Select non-default reference (by index or name)
kinshipLR(S = sibs, H = halfsibs, U = unrel, ref = "H")
# Alternative syntax: List input
peds = list(S = sibs, H = halfsibs, U = unrel)
kinshipLR(peds, ref = "H", source = "S", verbose = TRUE)
# Detailed results
res = kinshipLR(peds)
res$LRperMarker
res$likelihoodsPerMarker
### Example 2: Separating grandparent/halfsib/uncle-nephew
# Requires ibdsim2 and MERLIN
if(requireNamespace("ibdsim2", quietly = TRUE) && pedprobr::checkMerlin()) {
  # Load recombination map
  map = ibdsim2::loadMap("decode19", uniform = TRUE) # unif for speed
  # Define pedigrees
  ids = c("A", "B")
  H = relabel(halfSibPed(), old = c(4,5), new = ids)
  U = relabel(avuncularPed(), old = c(3,6), new = ids)
  G = relabel(linearPed(2), old = c(1,5), new = ids)
  # Attach FORCE panel of SNPs to G
  G = setSNPs(G, FORCE[1:10, ]) # use all for better results
  # Simulate recombination pattern in G
  ibd = ibdsim2::ibdsim(G, N = 1, ids = ids, map = map)
  # Simulate genotypes conditional on pattern
```

LRpower 21

```
G = ibdsim2::profileSimIBD(G, ibdpattern = ibd)

# Compute LR (genotypes are automatically transferred to H and U)
kinshipLR(H, U, G, linkageMap = map)
}
```

LRpower

Power simulation for kinship LR

# **Description**

This function uses simulations to estimate the likelihood ratio (LR) distribution in a given kinship testing scenario. In the most general setting, three pedigrees are involved: the two pedigrees being compared, and the true relationship (which may differ from the other two). A subset of individuals are available for genotyping. Some individuals may already be genotyped; all simulations are then conditional on these.

# Usage

```
LRpower(
  numeratorPed,
  denominatorPed,
  truePed = numeratorPed,
  ids,
  markers = NULL,
  source = "true",
  nsim = 1,
  threshold = NULL,
  disableMutations = NA,
  alleles = NULL,
  afreq = NULL,
  Xchrom = FALSE,
  knownGenotypes = NULL,
  plot = FALSE,
  plotMarkers = NULL,
  seed = NULL,
  verbose = TRUE
)
```

#### **Arguments**

numeratorPed, denominatorPed

ped objects (or lists of such), describing the two relationships under comparison.

truePed A ped object (or a list of such), describing the true relationship. By default equal

 $to \ numerator {\tt Ped}.$ 

ids Individuals available for genotyping.

LRpower LRpower

markers A vector indicating the names or indices of markers attached to the source pedi-

gree. If NULL (default), then all markers attached to the source pedigree are

used. If alleles or afreq is non-NULL, then this parameter is ignored.

source Either "true" (default), "numerator" or "denominator", indicating which pedi-

gree is used as source for marker data.

nsim A positive integer: the number of simulations.

threshold A numeric vector with one or more positive numbers used as LR thresholds.

disableMutations

Not implemented yet.

alleles, afreq, Xchrom

If these are given, they are used (together with knownGenotypes) to create a

marker object on the fly.

knownGenotypes A list of triplets (a, b, c), indicating that individual a has genotype b/c.

Ignored unless alleles or afreq is non-NULL.

plot Either a logical or the character "plotOnly". If the latter, a plot is drawn, but no

further computations are done.

plotMarkers A vector of marker names or indices whose genotypes are to be included in the

plot.

seed An integer seed for the random number generator (optional).

verbose A logical.

#### Value

A LRpowerResult object, which is essentially a list with the following entries:

- LRperSim: A numeric vector of length nsim containing the total LR for each simulation.
- meanLRperMarker: The mean LR per marker, over all simulations.
- meanLR: The mean total LR over all simulations.
- meanLogLR: The mean total log10(LR) over all simulations.
- IP: A named numeric of the same length as threshold. For each element of threshold, the fraction of simulations resulting in a LR exceeding the given number.
- time: The total computation time.
- params: A list containing the input parameters missing, markers, nsim, threshold and disableMutations

```
# Paternity LR of siblings
ids = c("A", "B")
truth = nuclearPed(children = ids)
claim = nuclearPed(fa = "A", mo = "NN", children = "B")
unrel = singletons(ids)

# Simulation parameters
nsim = 10  # increase!
```

markerSim 23

```
thresh = 1
# Simulation 1:
als = 1:5
afr = runif(5)
afr = afr/sum(afr)
pow1 = LRpower(claim, unrel, truth, ids = ids, nsim = nsim,
               threshold = thresh, alleles = als, afreq = afr,
               seed = 123)
pow1
# Simulation 2: Same, but using an attached marker
truth = addMarker(truth, alleles = als, afreq = afr)
pow2 = LRpower(claim, unrel, truth, ids = ids, nsim = nsim,
               threshold = thresh, markers = 1, seed = 123)
stopifnot(identical(pow1$LRperSim, pow2$LRperSim))
# True pedigree has inbred founders
truth2 = setFounderInbreeding(truth, value = 0.5)
pow3 = LRpower(claim, unrel, truth2, ids = ids, nsim = nsim,
               threshold = thresh, markers = 1, seed = 123) # plot = TRUE
pow3
```

markerSim

Marker simulation

#### **Description**

Simulates marker genotypes conditional on the pedigree structure and known genotypes. Note: This function simulates independent realisations at a single locus. Equivalently, it can be thought of as independent simulations of identical, unlinked markers. For simulating profiles for a set of different markers, see profileSim().

# Usage

```
markerSim(
    x,
    N = 1,
    ids = NULL,
    alleles = NULL,
    afreq = NULL,
    mutmod = NULL,
```

24 markerSim

```
rate = NULL,
partialmarker = NULL,
loopBreakers = NULL,
seed = NULL,
verbose = TRUE
)
```

#### **Arguments**

x A ped object or a list of such.

N A positive integer: the number of markers to be simulated.

ids A vector indicating the pedigree members whose genotypes should be simu-

lated. Alternatively, a function taking x as input and returning a character vector

of ID labels. Default: All individuals.

alleles (Only if partialmarker is NULL.) A vector with allele labels. If NULL, the

following are tried in order:

• names(afreq)

'seq\_along(afreq)'

• 1:2 (Fallback if both alleles and afreq are NULL.)

afreq (Only if partialmarker is NULL.) A numeric vector with allele frequencies,

possibly named with allele labels.

mutmod, rate Arguments specifying a mutation model, passed on to pedtools::marker()

(see there for explanations).

partialmarker Either NULL (resulting in unconditional simulation), a marker object (on which

the simulation should be conditioned) or the name (or index) of a marker at-

tached to x.

loopBreakers A numeric containing IDs of individuals to be used as loop breakers. Relevant

only if the pedigree has loops, and only if partialmarker is non-NULL. See

pedtools::breakLoops().

seed An integer seed for the random number generator (optional).

verbose A logical.

#### **Details**

This implements (with various time savers) the algorithm used in SLINK of the LINKAGE/FASTLINK suite. If partialmarker is NULL, genotypes are simulated by simple gene dropping, using simpleSim().

### Value

A ped object equal to x except its MARKERS entry, which consists of the N simulated markers.

#### Author(s)

Magnus Dehli Vigeland

markerSimParametric 25

#### References

G. M. Lathrop, J.-M. Lalouel, C. Julier, and J. Ott, *Strategies for Multilocus Analysis in Humans*, PNAS 81(1984), pp. 3443-3446.

#### See Also

```
profileSim(), simpleSim()
```

## **Examples**

```
x = nuclearPed(2)
# Unconditional simulation
markerSim(x, N = 2, alleles = 1:3)
# Conditional on one child being homozygous 1/1
x = addMarker(x, "3" = "1/1", alleles = 1:3)
markerSim(x, N = 2, partialmarker = 1)
markerSim(x, N = 1, ids = 4, partialmarker = 1, verbose = FALSE)
```

markerSimParametric

Simulate marker data given IBD coefficients

### **Description**

This function simulates genotypes for two individuals given their IBD distribution, for N identical markers.

## Usage

```
markerSimParametric(
   kappa = NULL,
   delta = NULL,
   states = NULL,
   N = 1,
   alleles = NULL,
   afreq = NULL,
   seed = NULL,
   returnValue = c("singletons", "alleles", "genotypes", "internal")
)
```

# **Arguments**

kappa A probability vector of length 3, giving a set of realised kappa coefficients (be-

tween two noninbred individuals).

delta A probability vector of length 9, giving a set of condensed identity coefficients

(Jacquard coefficients).

26 markerSimParametric

states	An integer vector of length N, with entries in 1-9. Each entry gives the identity state of the corresponding marker. (See details.)
N	A positive integer: the number of independent markers to be simulated.
alleles	A vector with allele labels. If NULL, the following are tried in order:
	• names(afreq)
	• 'seq_along(afreq)'
	• 1:2 (fallback if both alleles and afreq are NULL)
afreq	A numeric vector with allele frequencies, possibly named with allele labels.
seed	An integer seed for the random number generator (optional).
returnValue	Either "singleton" (default) or "alleles". (see Value).

#### **Details**

Exactly one of kappa, delta and states must be given; the other two should remain NULL.

If states is given, it explicitly determines the condensed identity state at each marker. The states are described by integers 1-9, using the tradition order introduced by Jacquard.

If kappa is given, the states are generated by the command states = sample(9:7, size = N, replace = TRUE, prob = kappa). (Note that identity states 9, 8, 7 correspond to IBD status 0, 1, 2, respectively.)

If delta is given, the states are generated by the command states = sample(1:9, size = N, replace = TRUE, prob = delta).

#### Value

The output depends on the value of the returnValue parameter:

- "singletons": a list of two singletons with the simulated marker data attached.
- "alleles": a list of four vectors of length N, named a, b, c and d. These contain the simulated alleles, where a/b and c/d are the genotypes of the to individuals.
- "genotypes": a list of two vectors of length N, containing the simulated genotypes. Identical to paste(a, b, sep = "/") and paste(c, d, sep = "/"), where a, b, c, d are the vectors returned when returnValue == "alleles".
- "internal": similar to "alleles", but using the index integer of each allele. (This option is mostly for internal use.)

```
# MZ twins
markerSimParametric(kappa = c(0,0,1), N = 5, alleles = 1:10)

# Equal distribution of states 1 and 2
markerSimParametric(delta = c(.5,.5,0,0,0,0,0,0), N = 5, alleles = 1:10)

# Force a specific sequence of states
markerSimParametric(states = c(1,2,7,8,9), N = 5, alleles = 1:10)
```

missingPersonEP 27

missingPersonEP

Exclusion power for missing person cases

# Description

This is a special case of exclusionPower() for use in missing person cases. The function computes the probability that a random person is genetically incompatible with the typed relatives of the missing person.

# Usage

```
missingPersonEP(
  reference,
  missing,
  markers = NULL,
  disableMutations = NA,
  verbose = TRUE
)
```

#### Arguments

reference A ped object with attached markers.

missing The ID label of the missing pedigree member.

markers A vector indicating the names or indices of mark

A vector indicating the names or indices of markers attached to the source pedigree. If NULL (default), then all markers attached to the source pedigree are used. If alleles or afreq is non-NULL, then this parameter is ignored.

disableMutations

This parameter determines how mutation models are treated. Possible values are as follows:

- NA (the default): Mutations are disabled only for those markers whose known genotypes are consistent with the pedigree. This is determined by temporarily removing all mutation models and checking which markers have nonzero likelihood.
- TRUE: Mutations are disabled for all markers. This will result in an error if any markers are inconsistent.
- FALSE: No action is done to disable mutations.
- A vector containing the names or indices of those markers for which mutations should be disabled.

verbose A logical.

## **Details**

This function is identical to randomPersonEP(), but with different argument names. This makes it consistent with missingPersonIP() and the other 'missing person' functions.

28 missingPersonIP

#### Value

The EPresult object returned by exclusionPower().

#### See Also

```
randomPersonEP(), exclusionPower()
```

#### **Examples**

```
# Four siblings; the fourth is missing
x = nuclearPed(4)

# Remaining sibs typed with 4 triallelic markers
x = markerSim(x, N = 4, ids = 3:5, alleles = 1:3, seed = 577, verbose = FALSE)

# Add marker with inconsistency in reference genotypes
# (by default this is ignored by `missingPersonEP()`)
x = addMarker(x, "3" = "1/1", "4" = "2/2", "5" = "3/3")

# Compute exclusion power statistics
missingPersonEP(x, missing = 6)
```

missingPersonIP

Inclusion power for missing person cases

# **Description**

This function simulates the LR distribution for the true missing person in a reference family. The output contains both the total and marker-wise LR of each simulation, as well as various summary statistics. If a specific LR threshold is given, the *inclusion power* is computed as the probability that LR exceeds the threshold.

# Usage

```
missingPersonIP(
  reference,
  missing,
  markers,
  nsim = 1,
  threshold = NULL,
  disableMutations = NA,
  seed = NULL,
  verbose = TRUE
)
```

missingPersonIP 29

#### **Arguments**

reference A ped object with attached markers.

missing The ID label of the missing pedigree member.

markers A vector indicating the names or indices of markers attached to the source pedi-

gree. If NULL (default), then all markers attached to the source pedigree are used. If alleles or afreq is non-NULL, then this parameter is ignored.

nsim A positive integer: the number of simulations

threshold A numeric vector with one or more positive numbers used as the likelihood ratio

thresholds for inclusion

disableMutations

This parameter determines how mutation models are treated. Possible values are as follows:

- NA (the default): Mutations are disabled only for those markers whose known genotypes are consistent with the pedigree. This is determined by temporarily removing all mutation models and checking which markers have nonzero likelihood.
- TRUE: Mutations are disabled for all markers. This will result in an error if any markers are inconsistent.
- FALSE: No action is done to disable mutations.
- A vector containing the names or indices of those markers for which mutations should be disabled.

seed An integer seed for the random number generator (optional).

verbose A logical.

#### Value

A mpIP object, which is essentially a list with the following entries:

- LRperSim: A numeric vector of length nsim containing the total LR for each simulation.
- meanLRperMarker: The mean LR per marker, over all simulations.
- meanLR: The mean total LR over all simulations.
- meanLogLR: The mean total log10(LR) over all simulations.
- IP: A named numeric of the same length as threshold. For each element of threshold, the fraction of simulations resulting in a LR exceeding the given number.
- time: The total computation time.
- params: A list containing the input parameters missing, markers, nsim, threshold and disableMutations

```
# Four siblings; the fourth is missing
x = nuclearPed(4)

# Remaining sibs typed with 5 triallelic markers
x = markerSim(x, N = 5, ids = 3:5, alleles = 1:3, seed = 123, verbose = FALSE)
```

30 missingPersonLR

```
# Compute inclusion power statistics
ip = missingPersonIP(x, missing = 6, nsim = 5, threshold = c(10, 100))
ip
# LRs from each simulation
ip$LRperSim
```

missingPersonLR

Likelihood ratio calculation for missing person identification

# **Description**

This is a wrapper function for kinshipLR() for the special case of missing person identification. A person of interest (POI) is matched against a reference dataset containing genotypes of relatives of the missing person.

# Usage

```
missingPersonLR(reference, missing, poi = NULL, verbose = TRUE, ...)
```

#### **Arguments**

reference A ped object with attached markers.

missing The ID label of the missing member of reference.

poi A singleton object, or NULL. If NULL, and missing is genotyped, this data

is extracted and used as poi.

verbose A logical.

... Optional parameters to be passed on to kinshipLR().

#### **Details**

Note that this function accepts two forms of input:

- 1. With poi a typed singleton. This is the typical use case, when you want to compute the LR for some person of interest.
- 2. With poi = NULL, but missing being genotyped. The data for missing is then extracted as a singleton POI. This is especially useful in simulation procedures, e.g., for simulating the LR distribution of the true missing person.

See Examples for illustrations of both cases.

#### Value

The LRresult object returned by kinshipLR(), but without the trivial H2:H2 comparison.

missingPersonLR 31

```
#-----
# Example: Identification of a missing grandchild
# Database with 5 STR markers (increase to make more realistic)
db = NorwegianFrequencies[1:5]
# Pedigree with missing person (MP); grandmother is genotyped
x = linearPed(2) >
 relabel(old = 5, new = "MP") |>
 profileSim(markers = db, ids = "2", seed = 123)
### Scenario 1: Unrelated POI -----
# Generate random unrelated profile
poi = singleton("POI") |>
 profileSim(markers = db, seed = 1234)
# Compute LR
lr = missingPersonLR(x, missing = "MP", poi = poi)
lr
1r$LRperMarker
### Scenario 2: POI is the missing person -----
# A small simulation example
# Simulate profiles for MP conditional on the grandmother
y = profileSim(x, N = N, ids = "MP", seed = 12345)
# Compute LRs for each sim
LRsims = lapply(y, missingPersonLR, missing = "MP", verbose = FALSE)
# Plot distribution
LRtotal = sapply(LRsims, function(a) a$LRtotal)
plot(density(LRtotal))
# LRs for each marker
LRperMarker = sapply(LRsims, function(a) a$LRperMarker)
LRperMarker
# Overlaying marker-wise density plots (requires tidyverse)
# library(tidyverse)
# t(LRperMarker) |> as_tibble() |> pivot_longer(everything()) |>
   ggplot() + geom_density(aes(value, fill = name), alpha = 0.6)
```

32 missingPersonPlot

missingPersonPlot Missing person plot

### **Description**

Visualises the competing hypotheses of a family reunion case. A plot with two panels is generated. The left panel shows a pedigree in which the *person of interest* (POI) is identical to the *missing person* (MP). The right panel shows the situation where these two are unrelated. See Details for further explanations.

# Usage

```
missingPersonPlot(
  reference,
 missing,
  labs = labels(reference),
 marker = NULL,
 hatched = typedMembers(reference),
 MP.label = "MP",
 POI.label = "POI"
 MP.col = "#FF9999",
 POI.col = "lightgreen",
 POI.sex = getSex(reference, missing),
 POI.hatched = NULL,
 titles = c(expression(H[1] * ": POI = MP"), expression(H[2] * ": POI unrelated")),
 width = NULL,
  cex = 1.2,
)
```

# **Arguments**

reference	A pedtools::ped() object.	
missing	The ID label of the missing pedigree member.	
labs	A character vector with labels for the pedigree members. See $pedtools::plot.ped()$ .	
marker	Optional vector of marker indices to be included in the plot.	
hatched	A vector of ID labels indicating who should appear with hatched symbols in the plot. By default, all typed members.	
MP.label, POI.label		
	Custom labels of the missing person and the POI. Default: "MP" and "POI".	
MP.col,POI.col	Fill colours for MP and POI.	
POI.sex	The sex of POI. This defaults to that of the missing person, but may be set explicitly. This is particularly useful when the missing person has unknown sex.	
POI.hatched	Deprecated (ignored).	

missingPersonPlot 33

titles	A character of length 2, with subtitles for the two frames.
width	A positive number controlling the width of the plot. More specifically this number is the relative width of the reference pedigree, compared to a singleton.
cex	Expansion factor for pedigree symbols and font size.
	Extra parameters passed on to pedtools::plotPedList().

#### **Details**

A standard family reunification case involves the following ingredients:

- A reference family with a single missing person ("MP").
- Some of the family members have been genotyped
- A person of interest ("POI") is to be matched against the reference family

After genotyping of POI, the genetic evidence is typically assessed by computing the likelihood ratio of the following hypotheses:

- H1: POI is MP
- H2: POI is unrelated to the family

The goal of this function is to illustrate the above hypotheses, using labels, colours and shading to visualise the different aspects of the situation.

This function cannot handle cases with more complicated hypotheses (e.g. multiple missing persons, or where H2 specifies a different relationship). However, as it is basically a wrapper of pedtools::plotPedList(), an interested user should be able to extend the source code to such cases without too much trouble.

#### Value

None

34 MPPsims

**MPPsims** 

Missing person power simulations

#### **Description**

Estimate the exclusion/inclusion power for various selections of available individuals.

## Usage

```
MPPsims(
  reference,
  missing = "MP",
  selections,
  ep = TRUE,
  ip = TRUE,
  addBaseline = TRUE,
  nProfiles = 1,
  lrSims = 1,
  thresholdIP = NULL,
  disableMutations = NA,
  numCores = 1,
  seed = NULL,
  verbose = TRUE
)
```

# **Arguments**

reference A connected ped object, or a list of pedigrees. In the latter case, the list must

have the same length as selections.

missing The ID label of the missing pedigree member.

selections A list of pedigree member subsets. In the special case that all subsets consist of

a single individual, selections can be given as a simple vector.

ep A logical: Estimate the exclusion power? (Default: TRUE)

ip A logical: Estimate the inclusion power? (Default: TRUE)

addBaseline A logical. If TRUE (default) an *empty* selection, named "Baseline", is added as

the first element of selection.

nProfiles The number of profile simulations for each selection.

1rSims, thresholdIP

Parameters passed onto missingPersonIP().

disableMutations

This parameter determines how mutation models are treated. Possible values are as follows:

 NA (the default): Mutations are disabled only for those markers whose known genotypes are consistent with the pedigree. This is determined by temporarily removing all mutation models and checking which markers have nonzero likelihood. MPPsims 35

• TRUE: Mutations are disabled for all markers. This will result in an error if any markers are inconsistent.

- FALSE: No action is done to disable mutations.
- A vector containing the names or indices of those markers for which mutations should be disabled.

numCores The number of cores used for parallelisation, by default 1.

seed An integer seed for the random number generator (optional).

verbose A logical.

#### Value

An object of class "MPPsim", which is basically a list with one entry for each element of selections. Each entry has elements ep and ip, each of which is a list of length nProfiles.

The output object has various attributes reflecting the input. Note that reference and selection may differ slightly from the original input, since they may be modified during the function run. (For instance, a "Baseline" entry is added to selection if addBaseline is TRUE.) The crucial point is that the output attributes correspond exactly to the output data.

- reference (always a list, of the same length as the selections attribute
- selections
- nProfiles,1rSims,thresholdIP,seed (as in the input)
- totalTime (the total time used)

NorwegianFrequencies Norwegian STR frequencies

## **Description**

A database of Norwegian allele frequencies for 35 STR markers. NOTE: An updated database of these and other markers, are available in the norSTR package. The NorwegianFrequencies object is kept here for backward compatibility.

#### Usage

NorwegianFrequencies

#### Format

A list of length 35. Each entry is a numerical vector summing to 1, named with allele labels. The following markers are included:

• D3S1358: 12 alleles

TH01: 10 allelesD21S11: 26 alleles

• D18S51 : 23 alleles

• PENTA\_E: 21 alleles

• D5S818: 9 alleles

• D13S317: 9 alleles

• D7S820: 19 alleles

• D16S539: 9 alleles

• CSF1P0: 11 alleles

- PENTA\_D: 24 alleles
- VWA: 12 alleles
- D8S1179: 12 alleles
- TPOX: 9 alleles
- FGA: 25 alleles
- D19S433: 17 alleles
- D2S1338: 13 alleles
- D10S1248: 9 alleles
- D1S1656: 17 alleles
- D22S1045: 9 alleles
- D2S441: 13 alleles
- D12S391: 23 alleles
- SE33: 55 alleles
- D7S1517: 11 alleles
- D3S1744: 8 alleles
- D2S1360: 10 alleles
- D6S474: 6 alleles
- D4S2366: 7 alleles
- D8S1132: 12 alleles
- D5S2500: 8 alleles
- D21S2055: 18 alleles
- D10S2325: 10 alleles
- D17S906: 78 alleles
- APOAI1: 41 alleles
- D11S554: 51 alleles

#### **Source**

Dupuy et al. (2013): Frequency data for 35 autosomal STR markers in a Norwegian, an East African, an East Asian and Middle Asian population and simulation of adequate database size. Forensic Science International: Genetics Supplement Series, Volume 4 (1).

38 powerPlot

powerPlot

Exclusion/inclusion power plots

## **Description**

This function offers four different visualisations of exclusion/inclusion powers, particularly for missing person cases. Output from MPPsims() may be fed directly as input to this function. The actual plotting is done with ggplot2.

## Usage

```
powerPlot(
  еp,
  ip = NULL,
  type = 1,
  majorpoints = TRUE,
 minorpoints = TRUE,
  ellipse = FALSE,
  col = NULL,
  labs = NULL,
  jitter = FALSE,
  alpha = 1,
  stroke = 1.5,
  shape = "circle",
  size = 1,
  hline = NULL,
  vline = NULL,
  xlim = NULL,
 ylim = NULL,
 xlab = NULL,
 ylab = NULL
)
```

## Arguments

ep, ip	Lists of equal length, with outputs from one or more runs of missingPersonEP() and missingPersonIP() respectively. Alternatively, ep can be a single output from MPPsims(), in which case ip should be NULL. See Examples.	
type	Plot type; either 1, 2, 3 or 4.	
majorpoints	A logical indicating whether "major" points should be drawn (see Details).	
minorpoints	A logical indicating whether "minor" points should be drawn (see Details).	
ellipse	A logical. If TRUE, data ellipsis are drawn for each group containing more than 1 element. NB: This fails with a warning if all points in a group fall on a line.	
col	A colour vector, recycle to match the top level length of ep.	
labs	A character of the same length as ep. If NULL, the names of ep are used, if present.	

powerPlot 39

jitter	A logical (default: FALSE). If TRUE, a small jitter is added to the major points.
alpha	Transparency for minor points (see Details).
stroke	Border width for major points (see Details).
shape	Either "circle", "square", "diamond", "triangleUp" or "triangleDown", determining the shapes of both minor and major points.
size	Point size.
hline, vline	Single numbers indicating positions for horizontal/vertical "threshold" lines. If NULL (default), no lines are drawn.
xlim, ylim	Axis limits; automatically chosen if NULL.
xlab, ylab	Axis labels; automatically chosen if NULL.

#### **Details**

The plot types are as follows:

```
type = 1: x = Exclusion power; y = Inclusion power
type = 2: x = Exclusion odds ratio; y = Inclusion odds ratio
type = 3: x = Expected number of exclusions; y = average log(LR)
type = 4: x = Exclusion power; y = average LR
```

In the most general case ep (and similarly for ip) can be a list of lists of EPresult objects. We refer to the inner lists as "groups". A group may consist of a single output, or several (typically many simulations of the same situation). Points within the same group are always drawn with the same colour and shape.

When plotting several groups, two sets of points are drawn by default:

- Major points: Group means.
- Minor points: Individual points in groups with more than one element.

The parameters majorpoints and minorpoints control which of the above points are included.

#### Value

A ggplot2 plot object.

#### See Also

```
MPPsims(), missingPersonEP(), missingPersonEP()
```

## **Examples**

```
### Example 1: Comparing the power of 3 reference families ###
# Frequencies for 2 STR markers
db = NorwegianFrequencies[1:2] # Increase!
# Define pedigrees and simulate data
PAR = nuclearPed(1, child = "MP") |>
```

40 powerPlot

```
profileSim(markers = db, ids = 1)
SIB = nuclearPed(2) |> relabel(old = 4, new = "MP") |>
  profileSim(markers = db, ids = 3)
GRA = linearPed(2) |> relabel(old = 5, new = "MP") |>
  profileSim(markers = db, ids = 1)
# Collect in list and plot
peds = list(PAR = PAR, SIB = SIB, GRA = GRA)
plotPedList(peds, marker = 1, hatched = typedMembers, frames = FALSE,
            col = list(red = "MP"))
# Compute exclusion/inclusion powers:
ep = lapply(peds, function(y)
 missingPersonEP(y, missing = "MP", verbose = FALSE))
ip = lapply(peds, function(y)
                               # increase nsim!
  missingPersonIP(y, missing = "MP", nsim = 5, threshold = 10, verbose = FALSE))
# Plot
powerPlot(ep, ip, size = 2)
# Different plot type, not dependent of `threshold`
powerPlot(ep, ip, size = 2, type = 3)
### Example 2: Exploring powers for different sets of available relatives
# Create trio pedigree
ref = nuclearPed(father = "fa", mother = "mo", child = "MP")
# Add empty marker with 5 alleles
ref = addMarker(ref, alleles = 1:5)
# Alternatives for genotyping
sel = list("fa", c("fa", "mo"))
# Simulate power for each selection
simData = MPPsims(ref, selections = sel, nProfiles = 3, lrSims = 5,
                  thresholdIP = 2, seed = 123, numCores = 1)
# Power plot 1: EP vs IP
powerPlot(simData, type = 1)
powerPlot(simData, type = 1, minorpoints = FALSE, hline = 0.8)
# Change shape, and modify legend order
powerPlot(simData[3:1], type = 1, shape = c("ci", "sq", "di"))
# Zoom in, and add threshold lines
powerPlot(simData, type = 1, xlim = c(0.2, 1), ylim = c(0.5, 1),
```

profileSim 41

```
hline = 0.8, vline = 0.8)

# Power plot 3: Expected number of exclusions vs E[log LR]
powerPlot(simData, type = 3)

# With horizontal/vertical lines
powerPlot(simData, type = 3, hline = log10(2), vline = 1)

# Plot 4: Illustrating the general inequality ELR > 1/(1-EP)
powerPlot(simData, type = 4)
```

profileSim

Simulation of complete DNA profiles

#### **Description**

Simulation of DNA profiles for specified pedigree members. Some pedigree members may already be genotyped; in that case the simulation is conditional on these. The main work of this function is done by markerSim().

#### Usage

```
profileSim(
    x,
    N = 1,
    ids = NULL,
    markers = NULL,
    seed = NULL,
    numCores = 1,
    simplify1 = TRUE,
    verbose = TRUE,
    ...
)
```

## **Arguments**

x A ped object or a list of such.

N The number of complete simulations to be performed.

ids A character (or coercible to character) of ID labels indicating whose genotypes should be simulated. Alternatively, a function taking x as input and returning a

character vector of ID labels.

markers Either a vector indicating a subset of markers attached to x, or a named list of frequency vectors. By default (NULL), all attached markers are used. If a

frequency list is given, marker objects are created and attached to x. Simulations are conditional on the locus attributes (allele frequencies, mutation models, etc)

and any existing genotypes in the indicated markers.

42 profileSimParametric

seed	An integer seed for the random number generator (optional).	
numCores	The number of cores to be used. The default is 1, i.e., no parallelisation.	
simplify1	A logical, by default TRUE, removing the outer list layer when N = 1. See Value.	
verbose	A logical, by default TRUE.	
	Further arguments passed on to markerSim().	

#### Value

A list of N objects similar to x, but with simulated genotypes. Any previously attached markers are replaced by the simulated profiles. If the indicated markers contained genotypes for some pedigree members, these are still present in the simulated profiles.

If N = 1 and simplify 1 = TRUE, the outer list layer is removed, i.e., profileSim(..., N = 1, simplify 1 = T) is equivalent to profileSim(..., N = 1, simplify 1 = T). This is usually the desired object in interactive use, and works well with piping.

When using profileSim() in other functions, it is recommended to add simplify1 = FALSE to safeguard against issues with N = 1.

#### **Examples**

```
# Example pedigree with two brothers
x = nuclearPed(children = c("B1", "B2"))
### Simulate profiles using built-in freq database
profileSim(x, markers = NorwegianFrequencies[1:3])
### Conditioning on known genotypes for one brother

# Attach two SNP markers with genotypes for B1
y = x |>
addMarker(B1 = "1/2", alleles = 1:2) |>
addMarker(B1 = "1", alleles = 1:2, chrom = "X")

# Simulate 2 profiles of B2 conditional on the above
profileSim(y, N = 2, ids = "B2", seed = 123)
```

profileSimParametric Simulate complete DNA profiles given IBD coefficients

## **Description**

This function generalises markerSimParametric() in the same way that profileSim() generalises markerSim().

quickLR 43

#### Usage

```
profileSimParametric(
   kappa = NULL,
   delta = NULL,
   states = NULL,
   N = 1,
   freqList = NULL,
   seed = NULL,
   returnValue = c("singletons", "alleles", "genotypes", "internal")
)
```

## Arguments

kappa	A probability vector of length 3, giving a set of realised kappa coefficients (between two noninbred individuals).
delta	A probability vector of length 9, giving a set of condensed identity coefficients (Jacquard coefficients).
states	An integer vector of length N, with entries in 1-9. Each entry gives the identity state of the corresponding marker. (See details.)
N	A positive integer: the number of complete profiles to be simulated
freqList	A list of numeric vectors. Each vector is the allele frequencies of a marker.
seed	An integer seed for the random number generator (optional).
returnValue	Either "singleton" (default) or "alleles". (see Value).

#### Value

A list of length N, whose entries are determined by returnValue, as explained in markerSimParametric().

## **Examples**

```
# A single profile with 9 markers, each with forced identity state
profileSimParametric(states = 1:9, freqList = NorwegianFrequencies[1:9])
```

quickLR

LR calculations for paternity and sibship

## **Description**

A thin wrapper around kinshipLR() for the common scenario of testing a pair of individuals for paternity and/or sibship, against being unrelated.

## Usage

```
quickLR(x, ids = typedMembers(x), test = c("pat", "sib", "half"))
```

44 randomPersonEP

#### **Arguments**

x A ped object or a list of such.

ids A vector of two typed members of x. If not given, the typed members of x

are selected by default, but note that this gives an error if the number of such

individuals is not 2.

test The hypotheses to be tested (against 'unrelatedness'). Allowed values are "pat"

(=paternity), "sib" (=full siblings), and "half" (=half siblings). By default, all

three are included.

#### Value

A (slightly simplified) LRresult object, as described in kinshipLR().

## **Examples**

```
# Simulate 100 markers for half siblings
x = halfSibPed() |> markerSim(N = 100, ids = 4:5, alleles = 1:3, seed = 1)
# Test paternity, full sib, half sib
quickLR(x)
```

randomPersonEP

Random person exclusion power

#### Description

This is a special case of exclusionPower(), computing the power to exclude a random person as a given pedigree member. More specifically, the function computes the probability of observing, in an individual unrelated to the family individual, a genotype incompatible with the typed family members.

## Usage

```
randomPersonEP(x, id, markers = NULL, disableMutations = NA, verbose = TRUE)
```

## Arguments

x A ped object with attached markers.

id The ID label of a single pedigree member.

markers A vector indicating the names or indices of markers attached to the source pedi-

gree. If NULL (default), then all markers attached to the source pedigree are

used. If alleles or afreq is non-NULL, then this parameter is ignored.

disableMutations

This parameter determines how mutation models are treated. Possible values are as follows:

rankProfiles 45

- NA (the default): Mutations are disabled only for those markers whose known genotypes are consistent with the pedigree. This is determined by temporarily removing all mutation models and checking which markers have nonzero likelihood.
- TRUE: Mutations are disabled for all markers. This will result in an error if any markers are inconsistent.
- FALSE: No action is done to disable mutations.
- A vector containing the names or indices of those markers for which mutations should be disabled.

verbose A logical.

#### Value

The EPresult object returned by exclusionPower().

#### **Examples**

```
# Four siblings:
x = nuclearPed(4)

# First 3 sibs typed with 4 triallelic markers
x = markerSim(x, N = 4, ids = 3:5, alleles = 1:3, seed = 577, verbose = FALSE)

# Probability that a random man is excluded as the fourth sibling
randomPersonEP(x, id = 6)
```

rankProfiles

Find the most likely profiles of a pedigree member

#### **Description**

Identify and rank the most likely DNA profiles of a pedigree member. For each marker, the possible genotypes of the indicated person are ranked by likelihood.

## Usage

```
rankProfiles(x, id, markers = NULL, maxPerMarker = Inf, verbose = FALSE)
```

#### **Arguments**

x A ped object with attached markers.

id The name of a single (typically untyped) pedigree member.

Mames or indices of the markers to be included. Default: all.

maxPerMarker A single number, limiting the number of top genotypes considered for each

marker. Default: Inf (no restriction).

verbose A logical, by default FALSE.

46 showInTriangle

#### **Details**

Note that this function assumes that all markers are independent.

If the marker data includes mutation models, it may be wise to try first with maxPerMarker = 1 to limit computation time.

#### Value

A list with the following components (N denotes the number of markers):

- profiles: A data frame with N+1 columns, containing the possible profiles, ranked by likelihood.
- marginal1: A numeric of length N, giving the marginal probability of the most likely genotype for each marker.
- marginal2: A numeric of length N, with marginals for the *second* most likely genotype for each marker, or NA if there is no second.
- best: A character of length N containing the most likely profile. This is the same as names (marginal1), and also as profiles[1, 1:N].

## **Examples**

```
x = nuclearPed(nch = 4) |>
    markerSim(N = 4, alleles = c("a", "b", "c"), seed = 1, verbose = FALSE)
x
# Remove data for father
y = setAlleles(x, ids = 1, alleles = 0)
# Most likely profiles of father
rankProfiles(y, id = 1)
# Compare with truth
getGenotypes(x, ids = 1)
# Same example with mutations allowed
z = setMutmod(y, model = "equal", rate = 0.01)
rankProfiles(z, id = 1)
```

showInTriangle

Add points to the IBD triangle

#### **Description**

This function is re-exported from the ribd package. For documentation see ribd::showInTriangle().

simpleSim 47

simpleSim	Unconditional marker simulation
-----------	---------------------------------

## **Description**

Unconditional simulation of unlinked markers

## Usage

```
simpleSim(
    x,
    N,
    alleles,
    afreq,
    ids,
    Xchrom = FALSE,
    mutmod = NULL,
    seed = NULL,
    verbose = TRUE
)
```

## Arguments

X	A ped object.
N	A positive integer: the number of markers to be simulated.
alleles	A vector with allele labels.
afreq	A numeric vector of allele frequencies. If missing, the alleles are assumed to be equifrequent.
ids	A vector containing ID labels of those pedigree members whose genotypes should be simulated.
Xchrom	A logical: X linked markers or not?
mutmod	A list of mutation matrices named 'female' and 'male'.
seed	An integer seed for the random number generator (optional).
verbose	A logical.

## **Details**

Simple genotype simulation, performed by first distributing alleles randomly to all founders, followed by Mendelian *gene dropping* down throughout the pedigree (i.e., for each non-founder a random allele is selected from each of the parents). Finally, genotypes of individuals not included in ids are removed.

## Value

A ped object equal to x except its MARKERS entry, which consists of the N simulated markers.

48 simpleSim

## See Also

```
markerSim()
```

## Examples

```
x = nuclearPed(1)
simpleSim(x, N = 3, afreq = c(0.5, 0.5))

y = cousinPed(1, child = TRUE)
simpleSim(y, N = 3, alleles = LETTERS[1:10])
```

# **Index**

* datasets	pedprobr::likelihood(), <i>19</i>
FORCE, 11	<pre>pedprobr::likelihoodMerlin(), 19</pre>
NorwegianFrequencies, 36	<pre>pedtools::breakLoops(), 24</pre>
	<pre>pedtools::marker(), 24</pre>
checkPairwise, 2	pedtools::ped(), 32
	<pre>pedtools::plot.ped(), 32</pre>
exclusionPower, 5	<pre>pedtools::plotPedList(), 33</pre>
exclusionPower(), 27, 28, 44, 45	<pre>pedtools::setSNPs(), 11</pre>
expectedLR, 8	<pre>plotCP (checkPairwise), 2</pre>
6 11 10	powerPlot, 38
familias, 10	profileSim,41
findExclusions, 10	profileSim(), 23, 25, 42
FORCE, 11	profileSimParametric, 42
ibdBootstrap, 12	
ibdBootstrap(), <i>16</i>	quickLR, 43
ibdEstimate, 14	nondemPensenED 44
ibdEstimate(), 2, 4, 12, 13, 17	randomPersonEP, 44
ibdLoglik, 17	randomPersonEP(), 27, 28 rankProfiles, 45
ibdLoglikFUN (ibdLoglik), 17	readFam (familias), 10
1201081111 011 (120108111), 17	
kinshipLR, 18	<pre>ribd::ibdTriangle(), 2, 4 ribd::showInTriangle(), 46</pre>
kinshipLR(), 30, 43, 44	ribu::Snowiniriangle(),40
	showInTriangle,46
LRpower, 21	showInTriangle(), <i>14</i> , <i>16</i>
LRpower(), <i>19</i>	simpleSim, 47
	simpleSim(), 24, 25
markerSim, 23	, , , , , , , , , , , , , , , , , , ,
markerSim(), 41, 42, 48	XFORCE (FORCE), 11
markerSimParametric, 25	
markerSimParametric(), 42, 43	
missingPersonEP, 27	
missingPersonEP(), 38, 39	
missingPersonIP, 28	
missingPersonIP(), 27, 34, 38	
missingPersonLR, 30	
missingPersonPlot, 32	
MPPsims, 34	
MPPsims(), 38, 39	
NorwegianFrequencies, 36	