# Package ‘segregatr’

## April 15, 2021

**Title**  Segregation Analysis for Variant Interpretation

**Version**  0.2.0

**Description**  An implementation of the full-likelihood Bayes factor (FLB) for evaluating segregation evidence in clinical medical genetics. The method was introduced by Thompson et al. (2003) <doi:10.1086/378100>, and further popularised by Bayrak-Toydemir et al. (2008) <doi:10.1016/j.yexmp.2008.03.006>. This implementation allows custom penetrance values and liability classes, and includes specialised pedigree visualisations.

**License**  GPL-3

**URL**  https://github.com/magnusdv/segregatr

**BugReports**  https://github.com/magnusdv/segregatr/issues

**Encoding**  UTF-8

**Language**  en-GB

**Depends**  pedtools

**Imports**  pedprobr

**Suggests**  testthat (>= 2.1.0)

**RoxygenNote**  7.1.1

**NeedsCompilation**  no

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Description

Computes the Bayes factor for co-segregation, as described by Thompson et al. (2003).

Usage

```r
FLB(
  x,
  carriers,
  noncarriers = NULL,
  freq,
  affected,
  unknown = NULL,
  proband,
  penetrances,
  liability = NULL,
  details = FALSE,
  plot = FALSE,
  ...
)
```

Arguments

- **x**: A `pedtools::ped()` object.
- **carriers**: A character vector (or coercible to such), containing the ID labels of pedigree members known to carry the variant in question.
- **noncarriers**: A character vector (or coercible to such), containing the ID labels of pedigree members known not to carry the variant in question.
- **freq**: A single number strictly between 0 and 1: the population frequency of the observed allele.
- **affected**: The affected pedigree members.
- **unknown**: Pedigree members with unknown affection status.
- **proband**: The ID label of the proband. This person must also be in both carriers and affected.
- **penetrances**: Either a numeric vector of length 3, corresponding to (f0, f1, f2) or a matrix or data frame with 3 columns. Each row contains the penetrance values of a liability class.
- **liability**: A vector of length `pedsize(x)`, containing for each pedigree member the row number of penetrances which should be used for that individual. (If `penetrances` is just a vector, it will be used for all classes.) If `liability` is NULL (the default), it is set to 1 for all individuals.
plotSegregation

Plot Pedigree for Segregation Analysis

**Usage**

```r
plotSegregation(
  x,
  affected = NULL,
  unknown = NULL,
  proband = NULL,
  carriers = NULL,
  noncarriers = NULL,
  cex = 1,
  margins = rep(1, 4),
  ...,
)
```

**Details**

- `details` A logical, indicating if detailed output should be returned (for debugging purposes).
- `plot` A logical.
- `...` Optional plot parameters passed on to `pedtools::plot.ped()`.

**Value**

A positive number. If `details = TRUE`, a list of intermediate results is returned.

**References**


**Examples**

```r
x = nuclearPed(2)

FLB(x, carriers = 3:4, aff = 3:4, unknown = 1:2,
    freq = 0.0001, penetrances = c(0, 1, 1), proband = 3)
```
Arguments

x A `pedtools::ped()` object.
affected The affected pedigree members.
unknown Pedigree members with unknown affection status.
proband The ID label of the proband. This person must also be in both carriers and affected.
carriers A character vector (or coercible to such), containing the ID labels of pedigree members known to carry the variant in question.
noncarriers A character vector (or coercible to such), containing the ID labels of pedigree members known not to carry the variant in question.
cex, margins Arguments passed on to `pedtools::plot.ped()`.
... Optional plot parameters passed on to `pedtools::plot.ped()`.

Examples

```r
x = nuclearPed(2)
plotSegregation(x, affected = 3:4, unknown = 1:2, proband = 3,
carriers = 3:4, margins = c(1,3,1,1))
```

Description

An implementation of the full-likelihood Bayes factor (FLB) for evaluating segregation evidence in clinical medical genetics. The method was introduced by Thompson et al. (2003), and further popularised by Bayrak-Toydemir et al. (2008). This implementation allows custom penetrance values and liability classes, and includes specialised pedigree visualisations.

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