

# 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

**Author** Magnus Dehli Vigeland [aut, cre]  
(<<https://orcid.org/0000-0002-9134-4962>>)

**Maintainer** Magnus Dehli Vigeland <m.d.vigeland@medisin.uio.no>

**Repository** CRAN

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FLB

*Full-likelihood Bayes factor***Description**

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

**Usage**

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

**Arguments**

<code>x</code>	A <code>pedtools::ped()</code> object.
<code>carriers</code>	A character vector (or coercible to such), containing the ID labels of pedigree members known to carry the variant in question.
<code>noncarriers</code>	A character vector (or coercible to such), containing the ID labels of pedigree members known <i>not</i> to carry the variant in question.
<code>freq</code>	A single number strictly between 0 and 1: the population frequency of the observed allele.
<code>affected</code>	The affected pedigree members.
<code>unknown</code>	Pedigree members with unknown affection status.
<code>proband</code>	The ID label of the proband. This person must also be in both <code>carriers</code> and <code>affected</code> .
<code>penetrances</code>	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.
<code>liability</code>	A vector of length <code>pedsize(x)</code> , containing for each pedigree member the row number of <code>penetrances</code> which should be used for that individual. (If <code>penetrances</code> is just a vector, it will be used for all classes.) If <code>liability</code> is <code>NULL</code> (the default), it is set to 1 for all individuals.

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

### Value

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

### References

Thompson D, Easton DF, Goldgar DE. *A full-likelihood method for the evaluation of causality of sequence variants from family data*. Am J Hum Genet, 2003. doi: [10.1086/378100](https://doi.org/10.1086/378100).

### Examples

```
x = nuclearPed(2)

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

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plotSegregation	<i>Pedigree plot for segregation analysis</i>
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### Description

Plots a pedigree showing the segregation of a variant.

### Usage

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

**Arguments**

x	A <code>pedtools::ped()</code> 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 <i>not</i> to carry the variant in question.
cex, margins	Arguments passed on to <code>pedtools::plot.ped()</code> .
...	Optional plot parameters passed on to <code>pedtools::plot.ped()</code> .

**Examples**

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

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 segregatr

*segregatr: Segregation Analysis for Identifying Pathogenic Variants*


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**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.

**References**

- Thompson D, Easton DF, Goldgar DE. *A full-likelihood method for the evaluation of causality of sequence variants from family data*. Am J Hum Genet, 2003. doi: [10.1086/378100](https://doi.org/10.1086/378100).
- Bayrak-Toydemir et al. *Likelihood ratios to assess genetic evidence for clinical significance of uncertain variants: Hereditary hemorrhagic telangiectasia as a model*. Exp Mol Pathol, 2008. doi: [10.1016/j.yexmp.2008.03.006](https://doi.org/10.1016/j.yexmp.2008.03.006).

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