# Package 'heteroplasmy'

June 1, 2021

Title Calculation of the standard error of the variance for heteroplasmy data

Version 0.0.0.9000

**Description** The package offers different methods to quantify uncertainty by calculating the standard errors of the variance of given data. The included functions are primarily aimed to heteroplasmy data, which are not assumed to follow a predefined distribution and the sample size is usually low. Included, there is a set of synthetic datasets to use, as well as real heteroplasmy data from mouse specimen, found in (xxx Iain's paper xxx) The code and the methods described in the package are used in the (xxx our report xxx), so please cite that work in case you use the heteroplasmy package.

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LazyData true

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Analytic calculation of the standard error of the variance

#### **Description**

This function calculates analytically the standard error of the variance.

## Usage

```
analyticVar(data, normal = FALSE)
```

### **Arguments**

data The input data in the form of a dataframe or matrix (which will be transformed

into a dataframe). NA values are omitted.

normal Parameter that indicates if the normal approximation should be used instead of

the general formula from (Wilks, S. S. (1962). Mathematical Statistics). Default

is FALSE.

### Value

The analytically derived standard error of the variance of data.

### **Examples**

```
# size of the sample
n=50
#generate a random sample of size n from a normal distribution
data_ex=rnorm(n,0.5,0.1)
analyticVar(data)

mouseData=readHeteroplasmyData("HB")
mouseData1 = mouseData[which(!is.na(mouseData[,1])),1]
analyticVar(mouseData1)
```

bootstrapVar

A boostap method to calculate the standard error of the variance

#### **Description**

This function uses the bootrstap method to calculate the uncertainty of the variance of a given sample based on random resampling. The number of the resamples is a parameter (default is 1000) and along with the the "vanilla" version, we offer an optimized variation (using the **sigmaOpt** parameter) which has been seen to improve the precision of the calculation (see our report/paper). Given that the resampling methods underestimate the uncertainty and thus provide a biased estimation, we offer the the unbiased method as a default, although the user may change this option through the biased parameter for experimental purposes (they are strongly advised not to do for real problems with small samples).

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#### **Usage**

```
bootstrapVar(
  data,
  nrep = 1000,
  biased = FALSE,
  corrected = FALSE,
  sigmaOpt = FALSE
)
```

## **Arguments**

data The input data in the form of a dataframe or matrix (which will be transformed

into a dataframe). NA values are omitted.

nrep The number of bootstrap resamples. Default is 1000. The higher the number of

the samples, the better the bootstrap outcome.

biased A logical parameter to indicate if the user wants the biased version. Resampling

techniques always underestimate statistics like the variance or the standard error

of it for small samples.

corrected Simle correction with a factor of 2.61 that was experimentally found. It is in-

cluded also in the case of sigmaOpt=TRUE

sigmaOpt The outcome of the bootstrap resampling with a fitted sigmoid function g(x)

with four parameters. Derived through simulations on both real heteroplasmy data and various synthetic ones. Try the plotStdErrVar function in this package

to observe it.

#### Value

The standard error of the variance of data.

## **Examples**

```
# size of the sample
n=50
#generate a random sample of size n from a normal distribution
data_ex=rnorm(n,0.5,0.1)
bootstrapVar(data)

mouseData=readHeteroplasmyData("HB")
mouseData1 = mouseData[which(!is.na(mouseData[,1])),1]
bootstrapVar(mouseData1, sigmaOpt=TRUE)
```

bootstrapVarCor

The bootstrapVar function with default correction

## Description

This function is simply the bootstrapVar with the correction argument being TRUE. it is provided as a seperate function for usability.

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#### Usage

```
bootstrapVarCor(data, nrep = 1000)
```

## **Arguments**

data The input data in the form of a dataframe or matrix (which will be transformed

into a dataframe). NA values are omitted.

nrep The number of bootstrap resamples. Default is 1000. The higher the number of

the samples, the better the bootstrap outcome (see par).

#### Value

The analytically derived standard error of the variance of data.

### Author(s)

Kostas and Iain, <us@example.com>

### References

```
https://en.wikipedia.org/
```

#### See Also

bootstrapVar

## **Examples**

```
# size of the sample
n=50
#generate a random sample of size n from a normal distribution
data_ex=rnorm(n,0.5,0.1)
bootstrapVarCor(data)

mouseData=readHeteroplasmyData("HB")
mouseData1 = mouseData[which(!is.na(mouseData[,1])),1]
bootstrapVarCor(mouseData1,nrep=10000)
```

heteroplasmyShift

Transformed heteroplasmy shift

## Description

A numerical transformation of the heteroplasmy samples in order to work with the heteroplasmy shifts across diverse samples (e.g., due to time or different tissue samples). This transformation is used for comparing a heteroplasmy observation h to a reference value  $h_0$ . It corresponds to the formula:

$$\Delta h = \ln \left( \frac{h(h_0 - 1)}{h_0(h - 1)} \right)$$

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### Usage

```
heteroplasmyShift(h, h0)
```

## **Arguments**

h The heteroplasmy observation.h0 The reference heteroplasmy value.

### Value

The Transformed heteroplasmy shift.

#### Author(s)

Kostas and Iain, <us@example.com>

#### References

Site or paper

### See Also

readHeteroplasmyData

## **Examples**

```
# size of the sample
n=50
#generate a random sample of size n from a normal distribution
data_ex=rnorm(n,0.5,0.1)
heteroplasmyShift(data)

mouseData=readHeteroplasmyData("HB")
mouseData1 = mouseData[which(!is.na(mouseData[,1])),1]
heteroplasmyShift(mouseData1,nrep=10000)
```

jackVar

A jackknife method to compute the uncertainty of heteroplasmy data

## Description

Similarly to the main bootstrapVar function that implements the bootstra method to measure the standard error of the variance, the jackknife technique is another resampling method that can be used for the same purpose. Unlike bootstrapVar, jackVar (and very jackknife method) is deterministic and doesn not rely on randomness, but instead it uses removals of the sample points, one each time to calculate different sub-samples of size (n-1). Note that the size of the input data should be strictly greater than 1.

### Usage

```
jackVar(data)
```

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### **Arguments**

data

The input data in the form of a dataframe or matrix (which will be transformed into a dataframe). Its size should be >=2. NA values are omitted.

### Value

The analytically derived standard error of the variance of data.

## **Examples**

```
# size of the sample
n=50
#generate a random sample of size n from a normal distribution
data_ex=rnorm(n,0.5,0.1)
jackVar(data)

mouseData=readHeteroplasmyData("HB")
mouseData1 = mouseData[which(!is.na(mouseData[,1])),1]
jackVar(mouseData1)
## Not run:
#input data of size 1 will fail
data_ex=rnorm(1,0.5,0.1)
jackVar(data)
## End(Not run)
```

mousedataHB

HB oocyte heteroplasmy data.

## **Description**

A dataset containing hetoplasmy values for the HB oocyte mouse lines. Each column corresponds to a different specimen. NA values have been added to make the number of rows equal. Please remove them after loading.

## Usage

mousedataHB

## Format

```
A data frame with 25 rows and 56 columns (TO FIX):

price price, in US dollars

carat weight of the diamond, in carats
```

## Source

```
http://www.example.info/
```

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mousedataLE

LE oocyte heteroplasmy data.

#### **Description**

A dataset containing hetoplasmy values for the HB oocyte mouse lines. Each column corresponds to a different specimen. NA values have been added to make the number of rows equal. Please remove them after loading.

### Usage

mousedataLE

### **Format**

```
A data frame with 20 rows and 43 columns (TO FIX): price price, in US dollars carat weight of the diamond, in carats
```

#### Source

```
http://www.example.info/
```

plotStdErrVar

An example plotting function

## **Description**

This function is used as a toy example on how to represent the data statistics regarding the variance of the sample. The mean variance and its standard error are depicted. Note that this is just an illustration to show that the analytic and the resampling approaches almost match each other.

### Usage

```
plotStdErrVar(
  data,
  functions = c("normalApr", "analytic", "bootstrap", "correctedBoot", "jackknife"),
   ...
)
```

## **Arguments**

data The input data in the form of a dataframe or matrix (which will be transformed

into a dataframe).

functions Choose the subset of the functions you wish use for the calculation and subse-

quent plot of the standard error of the variance. You can use one or a combination of "normalApr", "analytic", "bootstrap", "correctedBoot", and "jackknife".

For now, it outputs all of the aforementioned methods!

## Warning

This is a plotting function just for demonstration purposes.

## **Examples**

```
# size of the sample
#generate a random sample of size n from a normal distribution
data_ex=rnorm(n,0.5,0.1)
plotStdErrVar(data_ex)
```

## Description

This function allows you to read mouse heteroplasmy data from external files. Use with caution (for now).

## Usage

```
readHeteroplasmyData(nameD = "HB")
```

## Arguments

```
nameD
```

Either "HB" or "LE".

## Value

A dataframe containing mouse heteroplasmy data.

## **Examples**

```
readHeteroplasmyData(nameD="LE")
```

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