Package 'simDNAmixtures'

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Title Simulate Forensic DNA Mixtures

Version 1.0.1

Description Mixed DNA profiles can be sampled according to models for probabilistic genotyping. Peak height variability is modelled using a log normal distribution or a gamma distribution. Sample contributors may be related according to a pedigree.

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allele_specific_stutter_model

Stutter model where the expected stutter rate depends on the allele and locus

Description

Stutter model where the expected stutter rate depends on the allele and locus

Usage

Index

allele_specific_stutter_model(stutter_types, size_regression)

Arguments

stutter_types List. See stutter_type. size_regression Function, see read_size_regression.

Details

When a pg_model is constructed (see gamma_model), a stutter model can optionally be applied. The allele specific stutter model is commonly used with a log normal model. The expected stutter ratio for a parent allele at a locus is obtained from a linear regression of observed stutter ratios against allele length. For some loci or alleles the linear model may not be satisfactory. To override the expected stutter rates for specific alleles, a list of exceptions can be used. See stutter_type for more detail.

Value

Object of class stutter_model to be used by e.g. log_normal_model.

gamma_model

See Also

global_stutter_model for a stutter model where the expected stutter ratio does not depend on the locus or parent allele.

Examples

we will define an allele specific stutter model for back stutter only

```
# prepare stutter regression
filename_bs_regression <- system.file("extdata",</pre>
"GlobalFiler_Stutter_3500.txt",package = "simDNAmixtures")
bs_regression <- read_stutter_regression(filename_bs_regression)</pre>
# prepare exceptions, i.e. where does the regression not apply?
filename_bs_exceptions <- system.file("extdata",</pre>
"GlobalFiler_Stutter_Exceptions_3500.csv",package = "simDNAmixtures")
bs_exceptions <- read_stutter_exceptions(filename_bs_exceptions)</pre>
# prepare a stutter type
backstutter <- stutter_type(name = "BackStutter", delta = -1,</pre>
                             stutter_regression = bs_regression,
                             stutter_exceptions = bs_exceptions)
# assign stutter model
size_regression <- read_size_regression(system.file("extdata",</pre>
"GlobalFiler_SizeRegression.csv", package = "simDNAmixtures"))
bs_model <- allele_specific_stutter_model(list(backstutter), size_regression)</pre>
bs_model
```

```
gamma_model
```

```
Defines a gamma model for peak height variability
```

Description

Defines a gamma model for peak height variability

Usage

```
gamma_model(
    mixture_proportions,
    mu,
    cv,
    degradation_beta = rep(1, length(mixture_proportions)),
    LSAE = stats::setNames(rep(1, length(model_settings$locus_names)),
    model_settings$locus_names),
    model_settings
)
```

Arguments

mixture_proportions		
	Numeric vector with the mixture proportion for each contributor.	
mu	Numeric. Expectation of a full heterozygote contributing allele peak height.	
CV	Numeric. Coefficient of variation of a full heterozygote contributing allele peak height	
degradation_beta		
	Numeric Vector of same length as mixture_proportions. Degradation slope parameters for each contributor. Defaults to 1 for each contributor (i.e. not degraded)	
LSAE	Numeric vector (named) with Locus Specific Amplification Efficiencies. See sample_LSAE. Defaults to 1 for each locus.	
<pre>model_settings</pre>	List. Possible parameters:	
	locus_names. Character vector.	
	• detection_threshold. Numeric vector (named) with Detection Thresholds.	
	• size_regression. Function, see read_size_regression.	

• stutter_model. Optionally a stutter_model object that gives expected stutter heights. See global_stutter_model.

Details

Define a gamma model for peak height variability with the parametrisation as described by Bleka et al. The model may then be used to sample DNA profiles using the sample_mixture_from_genotypes function. Alternatively, to sample many models and profiles in one go with parameters according to a specified distribution, the sample_mixtures function can be used.

Value

Object of class pg_model.

References

Bleka, Ø., Storvik, G., & Gill, P. (2016). EuroForMix: An open source software based on a continuous model to evaluate STR DNA profiles from a mixture of contributors with artefacts. Forensic Science International: Genetics, 21, 35-44. doi: 10.1016/j.fsigen.2015.11.008

See Also

log_normal_model.

Examples

data(gf)

gf

Stutters and size regressions for a GlobalFiler 3500 kit

Description

A dataset containing default parameters and settings for a GlobalFiler 3500 kit.

Usage

gf

Format

A list of:

autosomal_markers Names of autosomal markers in the GlobalFiler kit

repeat_length_by_marker Named numeric with STR repeat length by locus name

size_regression See read_size_regression

stutters List of 4 stutter types, to be used with allele_specific_stutter_model

stutter_model For convenience, a pre-defined allele_specific_stutter_model

log_normal_settings Settings corresponding to a log normal model with all stutter types

log_normal_settings_fwbw Settings corresponding to a log normal model with backward and forward stutter only

gamma_settings Settings corresponding to a gamma model with all stutter types

gamma_settings_no_stutter Settings for a gamma model without stutter

global_stutter_model Global stutter model where the expected stutter rate is constant across alleles and loci

Description

Global stutter model where the expected stutter rate is constant across alleles and loci

Usage

```
global_stutter_model(back_stutter_rate, forward_stutter_rate, size_regression)
```

Arguments

back_stutter_rate

Numeric. (Optional)

forward_stutter_rate

Numeric. (Optional)

size_regression

Function, see read_size_regression.

Details

When a pg_model is constructed (see gamma_model), a stutter model can optionally be applied. In the global stutter model, the expected stutter rate is constant across all loci and for all parent alleles.

Value

Object of class stutter_model to be used by e.g. gamma_model.

See Also

allele_specific_stutter_model for a stutter model where the expected stutter rate depends on the allele and locus.

Examples

stutter_model

kits

Description

A dataset containing the properties of forensic DNA kits.

Usage

kits

Format

A list of data frames containing variables such as:

Panel Kit name Marker Allele Size Fragment length Color Dye colour

Source

https://github.com/oyvble/euroformix/blob/master/inst/extdata/kit.txt

log_normal_model Defines a log normal model for peak height variability

Description

Defines a log normal model for peak height variability

Usage

```
log_normal_model(
  template,
  degradation = rep(0, length(template)),
  LSAE = stats::setNames(rep(1, length(model_settings$locus_names)),
    model_settings$locus_names),
  c2,
  k2,
  model_settings
)
```

Arguments

template	Numeric vector
degradation	Numeric vector of same length as template. Degradation parameters for each contributor.
LSAE	Numeric vector (named) with Locus Specific Amplification Efficiencies. See sample_LSAE. Defaults to 1 for each locus.
c2	Numeric. Allele variance parameter.
k2	Optionally a numeric vector with stutter variance parameters. See sample_log_normal_stutter_variance .
model_settings	List. Possible parameters:
	locus_names. Character vector.
	 degradation_parameter_cap. Numeric.
	• c2_prior. Numeric of length two with shape and scale.

- LSAE_variance_prior. Numeric of length one.
- detection_threshold. Numeric vector (named) with Detection Thresholds. Defaults to 50 for each locus.
- size_regression. Function, see read_size_regression.
- stutter_model. Optionally a stutter_model object that gives expected stutter heights. See global_stutter_model.
- stutter_variability. Optionally peak height variability parameters for stutters. Required when stutter_model is supplied.

Details

Define a log normal model for peak height variability with the parametrisation as described by Bright et al. The model may then be used to sample DNA profiles using the sample_mixture_from_genotypes function. Alternatively, to sample many models and profiles in one go with parameters according to a specified distribution, the sample_mixtures function can be used.

Value

Object of class pg_model.

References

Bright, J.A. et al. (2016). Developmental validation of STRmixTM, expert software for the interpretation of forensic DNA profiles. Forensic Science International: Genetics, 23, 226-239. doi: 10.1016/j.fsigen.2016.05.007

See Also

gamma_model.

read_allele_freqs

Examples

read_allele_freqs Read allele frequencies in FSIgen format (.csv)

Description

Read allele frequencies in FSIgen format (.csv)

Usage

```
read_allele_freqs(filename, remove_zeroes = TRUE, normalise = TRUE)
```

Arguments

filename	Path to csv file.
remove_zeroes	Logical. Should frequencies of 0 be removed from the return value? Default is TRUE.
normalise	Logical. Should frequencies be normalised to sum to 1? Default is TRUE.

Details

Reads allele frequencies from a .csv file. The file should be in FSIgen format, i.e. comma separated with the first column specifying the allele labels and one column per locus. The last row should be the number of observations. No error checking is done since the file format is only loosely defined, e.g. we do not restrict the first column name or the last row name.

Value

Named list with frequencies by locus. The frequencies at a locus are returned as a named numeric vector with names corresponding to alleles.

Examples

```
# below we read an allele freqs file that comes with the package
filename <- system.file("extdata","FBI_extended_Cauc.csv",package = "simDNAmixtures")
freqs <- read_allele_freqs(filename)
freqs # the output is a list with an attribute named \code{N} giving the sample size.
```

Description

Reads a size regression file

Usage

read_size_regression(filename)

Arguments

filename Path to file (character).

Details

Read a regression file from disk and returns a function that provides the fragment length (bp) for a given locus and allele.

DNA profiles consist of the observed peaks (alleles or stutter products) at several loci as well as the peak heights and sizes. The size refers to the fragment length (bp). A linear relationship exists between the size of a peak and the size. When peaks are sampled in the sample_mixture_from_genotypes function, a size is assigned using a size regression. The read_size_regression function reads such a regression from disk.

Value

A function that takes a locus name and allele as arguments and returns the size.

Examples

obtain size for the 12 allele at the vWA locus regression("vWA", 12)

read_stutter_exceptions

Reads a stutter exceptions file with overrides for expected stutter ratios

Description

Reads a stutter exceptions file with overrides for expected stutter ratios

Usage

read_stutter_exceptions(filename)

Arguments

filename Character. Path to file.

Details

Reads the file from disk and returns a named numeric vector with stutter ratio exceptions for a given locus and allele.

Value

A named list with the stutter exceptions by locus. For each loucs, the exceptions are given as a named numeric with the names corresponding to the parent alleles and the expected stutter rates given as the values.

Examples

read_stutter_regression

Reads a stutter regression file

Description

Reads a stutter regression file

Usage

```
read_stutter_regression(filename, min_stutter_ratio = 0.001)
```

Arguments

filename Character. Path to file. min_stutter_ratio Numeric.

Details

Reads the file from disk and returns a function that provides the expected stutter ratio for a given locus and allele.

Value

A function that takes a locus name and allele as arguments and returns the expected stutter ratio.

Examples

```
\# obtain the expected stutter ratio for a 12 parent allele at the vWA locus regression("vWA", 12)
```

read_wide_table	Read wide table	(.txt) with Allele1, Allele2,	columns as is
-----------------	-----------------	-------------------------------	---------------

Description

Read wide table (.txt) with Allele1, Allele2, ... columns as is

Usage

```
read_wide_table(filename)
```

Arguments

filename Path to txt file.

Value

Dataframe

sample_contributor_genotypes

Sample genotypes for mixture contributors according to allele frequencies

Description

Sample genotypes for mixture contributors according to allele frequencies

Usage

```
sample_contributor_genotypes(
   contributors,
   freqs,
   pedigree,
   loci = names(freqs),
   return_non_contributors = FALSE
)
```

Arguments

contributors	Character vector with unique names of contributors. Valid names are "U1", "U2", for unrelated contributors or the names of pedigree members for related contributors.	
freqs	Allele frequencies (see read_allele_freqs)	
pedigree	(optionally) ped object	
loci	Character vector of locus names (defaults to names attribute of freqs)	
return_non_cont	ributors	
	Logical. Should genotypes of non-contributing pedigree members also be re- turned?	

Details

For each founder or unrelated person, a genotype is sampled randomly by drawing two alleles from allele frequencies. The non-founders get genotypes by allele dropping, see sample_pedigree_genotypes for details.

Value

List of DataFrames with genotypes for each pedigree member. See sample_genotype for the DataFrame format.

Examples

sample_gamma_model Sample gamma model(s) with parameters according to priors

Description

Sample gamma model(s) with parameters according to priors

Usage

```
sample_gamma_model(number_of_contributors, sampling_parameters, model_settings)
```

Arguments

List. Needs to contain:

- min_mu. Numeric of length one.
- max_mu. Numeric of length one.
- min_cv. Numeric of length one.
- max_cv. Numeric of length one.
- degradation_shape1. Numeric of length one.
- degradation_shape2. Numeric of length one.

model_settings List. See gamma_model.

Details

In simulation studies involving many mixed DNA profiles, one often needs to generate various samples with different model parameters. This function samples a gamma model with parameters according to prior distributions. The mean peak height parameter mu is sampled uniformly between min_mu and max_mu. Likewise, the variability parameter cv is sampled uniformly between min_cv and max_cv. The degradation slope parameter beta is sampled according to a Beta distribution with parameters degradation_shape1 and degradation_shape2.

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sample_genotype

Value

When length(number_of_contributors)==1, a single gamma_model of class pg_model. Otherwise, a list of these.

Examples

data(gf)

```
sampling_parameters = sampling_parameters,
model_settings = gf$gamma_settings_no_stutter)
```

model_no_stutter\$parameters

sample_genotype Sample a genotype according to allele frequencies

Description

Sample a genotype according to allele frequencies

Usage

```
sample_genotype(freqs, loci = names(freqs), label = "U")
```

Arguments

freqs	Allele frequencies (see read_allele_freqs)
loci	$Character \ vector \ of \ locus \ names \ (defaults \ to \ names \ attribute \ of \ freqs)$
label	Sample name

Details

A genotype is sampled randomly by drawing two alleles from allele frequencies for each locus.

Value

DataFrame with columns Sample Name, Locus, Allele1 and Allele2.

Examples

sample_log_normal_model

Sample log normal model(s) with parameters according to priors

Description

Sample log normal model(s) with parameters according to priors

Usage

```
sample_log_normal_model(
    number_of_contributors,
    sampling_parameters,
    model_settings
)
```

Arguments

```
number_of_contributors
Integer
sampling_parameters
List. Needs to contain:
```

- min_template. Numeric of length one.
- max_template. Numeric of length one.
- degradation_shape. Numeric of length one.
- degradation_scale. Numeric of length one.

model_settings List. See log_normal_model.

Details

In simulation studies involving many mixed DNA profiles, one often needs to generate various samples with different model parameters. This function samples a log normal model with parameters according to prior distributions. The template parameter for each contributor is sampled uniformly between min_template and max_template. The degradation parameter for each contributor is sampled from a gamma distribution with parameters degradation_shape and degradation_scale.

Value

When length(number_of_contributors)==1, a single log_normal_model of class pg_model. Otherwise, a list of these.

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Examples

Description

Sample log normal stutter variance parameters according to priors

Usage

sample_log_normal_stutter_variance(log_normal_stutter_variability)

Arguments

```
log_normal_stutter_variability
List of variability parameters. See gf for an example.
```

Value

Named numeric with stutter variance parameter for all stutter types. Names are k2 concatenated with the name of the stuter type. See example.

Examples

```
data(gf)
log_normal_stutter_variability <- gf$log_normal_settings$stutter_variability
k2 <- sample_log_normal_stutter_variance(log_normal_stutter_variability)</pre>
```

sample_LSAE

Description

Sample Locus Specific Amplification Efficiency (LSAE) according to prior

Usage

sample_LSAE(LSAE_variance, locus_names)

Arguments

LSAE_variance Numeric. See gf for an example. locus_names Character vector.

Details

In the Bright et al. log normal model, the expected peak height includes a multiplicative factor for the locus (marker). These factors are called the LSAEs (Locus Specific Amplification Efficiencies). In the model, the prior for the log10 of LSAE is normal with mean 0. The variance can be specified.

Value

Named numeric with LSAEs for each locus (names).

Examples

sample_mixtures	Sample mixtures with random genotypes and random parameters ac-
	cording to priors

Description

Sample mixtures with random genotypes and random parameters according to priors

sample_mixtures

Usage

```
sample_mixtures(
    n,
    contributors,
    freqs,
    sampling_parameters,
    model_settings,
    sample_model,
    pedigree,
    results_directory,
    seed,
    write_non_contributors = FALSE,
    tag = "simulation"
)
```

Arguments

n	Integer. Number of samples.	
contributors	Character vector with unique names of contributors. Valid names are "U1", "U2", \dots for unrelated contributors or the names of pedigree members for related contributors.	
freqs	Allele frequencies (see read_allele_freqs)	
<pre>sampling_parame</pre>	ters	
	List. Passed to the sample_model function.	
model_settings	List. Passed to the sample_model function.	
sample_model	Function such as sample_log_normal_model.	
pedigree	(optionally) ped object. Contributors can be named pedigree members.	
results_directory		
	(optionally) Character with path to directory where results are written to disk.	
seed	(optionally) Integer seed value that can be used to get reproducible runs. If results are written to disk, the 'Run details.txt' file will contain a seed that can be used for reproducing the result.	
write_non_contributors		
	Logical. If TRUE, sampled genotypes for non-contributing pedigree members will also be written to disk. Defaults to FALSE.	
tag	Character. Used for sub directory name when results_directory is provided.	

Value

If results_directory is provided, this function has the side effect of writing results to disk. Return value is a list with simulation results:

- call matched call
- smash DataFrame with all samples in SMASH format (see SMASH_to_wide_table)
- samples Detailed results for each sample
- parameter_summary DataFrame with parameters for each sample

Examples

Description

Sample mixture profile with provided genotypes

Usage

```
sample_mixture_from_genotypes(genotypes, model, sample_name = "mixture")
```

Arguments

genotypes	List of contributor genotypes. See sample_contributor_genotypes.
model	pg_model object.
sample_name	Character. Defaults to "mixture".

Details

A mixture profile is sampled according to the provided pg_model (see gamma_model, log_normal_model and genotypes (see sample_contributor_genotypes).

Value

DataFrame with at least SMASH columns (see SMASH_to_wide_table). Depending on the chosen pg_model (e.g. gamma_model or log_normal_model), other columns with further details about the simulation are returned as well.

See Also

sample_mixtures for a function that samples many mixtures in one go.

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sample_offspring

Examples

sample_offspring Sample offspring from two parental genotypes

Description

Sample offspring from two parental genotypes

Usage

```
sample_offspring(father, mother, label = "Child")
```

Arguments

father	DataFrame (see sample_genotype)
mother	DataFrame (see sample_genotype)
label	SampleName of child (character)

Details

A genotype is sampled according to Mendelian inheritance. That is, one of two alleles of a parent is passed down to the offspring.

Value

DataFrame (see sample_genotype)

Examples

sample_pedigree_genotypes

Sample genotypes for pedigree according to allele frequencies by allele dropping.

Description

Sample genotypes for pedigree according to allele frequencies by allele dropping.

Usage

```
sample_pedigree_genotypes(pedigree, freqs, loci = names(freqs))
```

Arguments

pedigree	ped object
freqs	Allele frequencies (see read_allele_freqs)
loci	Character vector of locus names (defaults to names attribute of freqs)

Details

For each founder, a genotype is sampled randomly by drawing two alleles according to allele frequencies. Alleles for the rest of the pedigree are then obtained by allele dropping: sample_offspring is invoked for each non-founder.

Value

List of DataFrames with genotypes for each pedigree member. See sample_genotype for the DataFrame format.

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SMASH_to_wide_table

Examples

SMASH_to_wide_table Converts SMASH (SampleName, Marker, Allele, Size, Height) data to a wide table

Description

Converts SMASH (SampleName, Marker, Allele, Size, Height) data to a wide table

Usage

SMASH_to_wide_table(x)

Arguments

х

DataFrame with SampleName, Marker, Allele, Size, Height columns

Value

DataFrame with columns: Sample Name, Marker, Allele 1, Allele 2, ..., Size 1, Size 2, ..., Height 1, Height 2, ...

Examples

wide_table <- SMASH_to_wide_table(mixtures\$smash)</pre>

stutter_type

Description

Defines a stutter type to be used in the allele specific stutter model.

Usage

```
stutter_type(
  name,
  delta,
  applies_to_all_loci = TRUE,
  stutter_regression,
  stutter_exceptions,
  applies_to_loci,
  repeat_length_by_marker
)
```

Arguments

name	Character. Name of the stutter, e.g. "BackStutter"	
delta	Numeric. When length one, repeat units gained (lost when negative). When length two, the second element is the number of base pairs gained (lost).	
applies_to_all_loci		
	Logical. Defaults to TRUE.	
stutter_regression		
	Function. See read_stutter_regression.	
stutter_exceptions		
	Optionally a list. See read_stutter_exceptions.	
applies_to_loci		
	Optionally a character vector of locus names to which this stutter type applies.	
<pre>repeat_length_by_marker</pre>		
	Optionally a named integer vector with repeat lengths by marker. Only needed when delta is of length two.	

Details

When a pg_model is constructed (see log_normal_model), a stutter model can optionally be applied.

Value

Object of class stutter_type to be passed to allele_specific_stutter_model.

stutter_type

Examples

```
filename_bs_exceptions <- system.file("extdata",
 "GlobalFiler_Stutter_Exceptions_3500.csv",package = "simDNAmixtures")
bs_exceptions <- read_stutter_exceptions(filename_bs_exceptions)
filename_bs_regression <- system.file("extdata",
 "GlobalFiler_Stutter_3500.txt",package = "simDNAmixtures")
bs_regression <- read_stutter_regression(filename_bs_regression)</pre>
```

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