

Package ‘diemr’

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Title Diagnostic Index Expectation Maximisation in R

Version 1.4.3

Description Likelihood-based genome polarisation finds which alleles of genomic markers belong to which side of the barrier.

Co-estimates which individuals belong to either side of the barrier and barrier strength. Uses expectation maximisation in likelihood framework. The method is described in Baird et al. (2023) <[doi:10.1111/2041-210X.14010](https://doi.org/10.1111/2041-210X.14010)>.

BugReports <https://github.com/StuartJEBaird/diem/issues>

License GPL (>= 3)

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brenthis	<i>Dataset of Butterfly Genotypes</i>
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Description

A subset of single nucleotide polymorphisms in butterflies of the genus *Brenthis*.

Format

vcf file with 13 individuals and 4 markers.

Details

The data is used to test conversion of genotype data from vcf to diem format with function `vcf2diem`.

Examples

```
filename <- system.file("extdata", "brenthis.vcf", package = "diemr")
```

CheckDiemFormat	<i>diem input file checker</i>
-----------------	--------------------------------

Description

Checks format of files with genotype data.

Usage

```
CheckDiemFormat(files, ChosenInds, ploidy)
```

Arguments

files	A character vector with paths to files with genotypes.
ChosenInds	A numeric or logical vector of indices of individuals to be included in the analysis.
ploidy	A logical or a list of length equal to length of files. Each element of the list contains a numeric vector with ploidy numbers for all individuals specified in the files.

Details

The input file must have genotypes of one marker for all individuals on one line. The line must start with a letter "S" and contain only characters "_" or "U" for unknown genotypes or a third/fourth allele, "0" for homozygots for allele 1, "1" for heterozygots, and "2" for homozygots for allele 2. Check the vignette with `browseVignettes(package = "diemr")` for the example of the input format.

Ploidies must be given as a list with each element corresponding to a genomic compartment (aka a file). For each compartment, the numeric vector specifying ploidies of all individuals chosen for the specific analysis must be given.

Value

Returns invisible TRUE if all files are executable by diem. Exits with informative error messages otherwise, specifying file names and lines with potential problems. When too many lines contain problems, the first six are given.

Examples

```
# set up input genotypes file names, ploidies and selection of individual samples
inputFile <- system.file("extdata", "data7x3.txt", package = "diemr")
ploidies <- list(c(2, 1, 2, 2, 2, 1, 2))
inds <- 1:7

# check input data
CheckDiemFormat(files = inputFile, ploidy = ploidies, ChosenInds = inds)
# File check passed: TRUE
# Ploidy check passed: TRUE
```

diem

Diagnostic Index Expectation Maximisation

Description

Estimates how to assign alleles in a genome to maximise the distinction between two unknown groups of individuals. Using expectation maximisation (EM) in likelihood framework, diem provides marker polarities for importing data, their likelihood-based diagnostic index and its support for all markers, and hybrid indices for all individuals.

Usage

```

diem(
  files,
  ploidy = FALSE,
  markerPolarity = FALSE,
  ChosenInds,
  ChosenSites = "all",
  epsilon = 0.99999,
  verbose = FALSE,
  nCores = parallel::detectCores() - 1,
  maxIterations = 50,
  ...
)

```

Arguments

<code>files</code>	A character vector with paths to files with genotypes.
<code>ploidy</code>	A logical or a list of length equal to length of <code>files</code> . Each element of the list contains a numeric vector with ploidy numbers for all individuals specified in the files.
<code>markerPolarity</code>	FALSE or a list of logical vectors.
<code>ChosenInds</code>	A numeric or logical vector of indices of individuals to be included in the analysis.
<code>ChosenSites</code>	A logical vector indicating which sites are to be included in the analysis.
<code>epsilon</code>	A numeric, specifying how much the hypothetical diagnostic markers should contribute to the likelihood calculations. Must be in $[\emptyset, 1)$, keeping tolerance setting of the R session in mind.
<code>verbose</code>	Logical or character with path to directory where run diagnostics will be saved.
<code>nCores</code>	A numeric number of cores to be used for parallelisation. Must be <code>nCores = 1</code> on Windows.
<code>maxIterations</code>	A numeric.
<code>...</code>	additional arguments.

Details

Given two alleles of a marker, one allele can belong to one side of a barrier to geneflow and the other to the other side. Which allele belongs where is a non-trivial matter. A marker state in an individual can be encoded as 0 if the individual is homozygous for the first allele, and 2 if the individual is homozygous for the second allele. Marker polarity determines how the marker will be imported. Marker polarity equal to FALSE means that the marker will be imported as-is. A marker with polarity equal to TRUE will be imported with states 0 mapped as 2 and states 2 mapped as 0, in effect switching which allele belongs to which side of a barrier to geneflow.

When `markerPolarity = FALSE`, `diem` uses random null polarities to initiate the EM algorithm. To fix the null polarities, `markerPolarity` must be a list of length equal to the length of the `files` argument, where each element in the list is a logical vector of length equal to the number of markers (rows) in the specific file.

Ploidy needs to be given for each compartment and for each individual. For example, for a dataset of three diploid mammal males consisting of an autosomal compartment, an X chromosome compartment and a Y chromosome compartment, the ploidy list would be `ploidy = list(rep(2, 3), rep(1, 3), rep(1, 3))`. If the dataset consisted of one male and two females, ploidy for the sex chromosomes should be vectors reflecting that females have two X chromosomes, but males only one, and females have no Y chromosomes: `ploidy = list(rep(2, 3), c(1, 2, 2), c(1, 0, 0))`.

When a subset of individuals is used to inform the genome polarisation in the `ChosenInds` argument, ploidy must still be provided for all individuals included in the files.

`ChosenInds` should preferably be numeric values within the range from 1 to the number of individuals in the files. Logical vectors must have a length equal to the number of individuals in the files.

When `verbose = TRUE`, `diem` will output multiple files with information on the iterations of the EM algorithm, including tracking marker polarities and the respective likelihood-based diagnostics. See vignette `vignette("Understanding-genome-polarisation-output-files", package = "diemr")` for a detailed explanation of the individual output files.

Value

A list including suggested marker polarities, diagnostic indices and support for all markers, four genomic state counts matrix for all individuals, and polarity changes for the EM iterations.

Note

To ensure that the data input format of the genotype files, ploidies and individual selection are readable for `diem`, first use [CheckDiemFormat](#). Fix all errors, and run `diem` only once the checks all passed.

The working directory or a folder optionally specified in the `verbose` argument must have write permissions. `diem` will store temporary files in the location and output results files.

The grain for parallelisation is the compartment files.

See Also

[CheckDiemFormat](#)

Examples

```
# set up input genotypes file names, ploidies and selection of individual samples
inputFile <- system.file("extdata", "data7x3.txt", package = "diemr")
ploidies <- list(c(2, 1, 2, 2, 2, 1, 2))
inds <- 1:6

# check input data
CheckDiemFormat(files = inputFile, ploidy = ploidies, ChosenInds = inds)
# File check passed: TRUE
# Ploidy check passed: TRUE

# run diem
## Not run:
# diem will write temporal files during EM iterations
```

```
# prior to running diem, set the working directory to a location with write permission
fit <- diem(files = inputFile, ChosenInds = inds, ploidy = ploidies, nCores = 1)

# run diem with fixed null polarities
fit2 <- diem(
  files = inputFile, ChosenInds = inds, ploidy = ploidies, nCores = 1,
  markerPolarity = list(c(TRUE, FALSE, TRUE))
)

## End(Not run)
```

emPolarise

Polarises a marker

Description

Changes encodings of genomic markers according to user specification.

Usage

```
emPolarise(origM, changePolarity = TRUE)
```

Arguments

origM A character vector of genotypes comprising of _012 encodings.
changePolarity A logical scalar, indicating whether to leave the marker as is (FALSE) or whether to change its polarity (TRUE).

Value

Returns a character vector with polarised markers.

Note

Note that [diem](#) and [importPolarized](#) accept also a U encoding for an unknown or third allele, but emPolarise requires all U to be replaced with _.

See Also

[diem](#) for determining appropriate marker polarity with respect to a barrier to gene flow.

Examples

```
emPolarise(c("0", "0", "1", "2", "2"), TRUE)
# [1] "2" "2" "1" "0" "0"

emPolarise(c("0", "_", "2", "2", "1"), FALSE)
# [1] "0" "_" "2" "2" "1"
```

importPolarized	<i>Imports genomic data polarized according to the specification</i>
-----------------	--

Description

Reads genotypes from a file and changes marker polarity.

Usage

```
importPolarized(  
  files,  
  changePolarity,  
  ChosenInds,  
  ChosenSites = "all",  
  nCores = 1,  
  verbose = FALSE,  
  ...  
)
```

Arguments

files	A character vector with paths to files with genotypes.
changePolarity	A logical vector with length equal to the number of markers.
ChosenInds	A numeric or logical vector of indices of individuals to be included in the analysis.
ChosenSites	A logical vector indicating which sites are to be included in the analysis.
nCores	A numeric number of cores to be used for parallelisation. Must be nCores = 1 on Windows.
verbose	Logical whether to show messages on import progress.
...	Optional numeric vector of compartmentSizes.

Details

For details on the input data format, check the file with [CheckDiemFormat](#).

The changePolarity argument influences how each marker is imported. Value FALSE means that the marker will be imported as it is saved in the file. Value TRUE means that the genotypes encoded as 0 will be imported as 2, and genotypes encoded in the file as 2 will be imported as 0.

Value

Returns a character matrix with rows containing individual genotypes and columns containing markers.

See Also

[diem](#) for determining appropriate marker polarity with respect to a barrier to gene flow.

Examples

```

dat <- importPolarized(
  files = system.file("extdata", "data7x3.txt", package = "diemr"),
  changePolarity = c(FALSE, TRUE, TRUE),
  ChosenInds = 1:6,
  ChosenSites = "all"
)
dat
#   m1 m2 m3
# 1 "0" "1" "2"
# 2 "0" "0" "0"
# 3 "1" "1" "0"
# 4 "1" "2" "0"
# 5 "2" "2" "1"
# 6 "2" "2" "-"

```

ModelOfDiagnostic

Model of Diagnostic Marker Based on All Individual State Counts

Description

Estimates a diagnostic marker for the state counts of all genomic markers for all individuals. Using the hypothetical, diagnostic marker, calculates individual state counts with respect to their weighted similarity to the diagnostic marker states.

Usage

```

ModelOfDiagnostic(
  I4,
  OriginalHI,
  epsilon = 0.99,
  verbose = FALSE,
  folder = "likelihood",
  ...
)

```

Arguments

I4	a matrix or data.frame with 4 numeric columns representing character state counts for missing data, homozygots for allele 1, heterozygots, and homozygots for allele 2. Individuals in rows.
OriginalHI	numeric vector of length equal to number of rows in I4, representing hybrid indices of individuals.
epsilon	A numeric, specifying how much the hypothetical diagnostic markers should contribute to the likelihood calculations. Must be in $[\emptyset, 1)$, keeping tolerance setting of the R session in mind.
verbose	Logical or character with path to directory where run diagnostics will be saved.

folder character specifying path to a folder for the verbose output.
... additional arguments.

Details

The OriginalHI can be calculated with [pHetErrOnStateCount](#).

Value

Matrix with dimensions of I4.

See Also

[diem](#) for utilising the model to determine appropriate marker polarisation in estimating barriers to gene flow.

myotis

Dataset of Modified Genotypes of Bats

Description

A subset of single nucleotide polymorphisms in *Myotis myotis* from Harazim et al. (2021). The genotypes were modified for testing purposes in such a way that markers 15 and 17 now include additional indel and substitution alleles. Eight markers used in the dataset are monomorphic.

Format

vcf file with 14 individuals and 20 markers.

Details

The data is used to test conversion of genotype data from vcf to diem format with function `vcf2diem`.

Source

Harazim M., Pialek L., Pikula J., Seidlova V., Zukal J., Bachorec E., Bartonicka T., Kokurewicz T., Martinkova N. (2021) Associating physiological functions with genomic variability in hibernating bats. *Evolutionary Ecology*, 35, 291-308, doi: 10.1007/s10682-020-10096-4.

Examples

```
filename <- system.file("extdata", "myotis.vcf", package = "diemr")
```

pHetErrOnStateCount *Hybrid index, heterozygosity, error rate*

Description

Using genotype allele counts, calculates the hybrid index, heterozygosity and error rate in a single individual.

Usage

```
pHetErrOnStateCount(sCount)
```

Arguments

sCount A numeric vector of length 4 with allele counts for missing data, homozygots for allele 1, heterozygots, and homozygots for allele 2.

Details

Allele counts are genomic state counts multiplied by ploidy. As different compartments might have different ploidies (e.g. autosomal markers, sex chromosomes, mitochondrial markers), allele counts should be calculated per compartment and then summarised to obtain the correct genomic allele counts. When all individuals in each compartment have the same ploidy, state counts do not need to be corrected.

Value

Returns a named numeric vector with three values: p - hybrid index, Het - heterozygosity, Err - error rate.

Examples

```
pHetErrOnStateCount(sCount = c(2, 4, 2, 6))
#        p        Het        Err
# 0.5833333 0.1666667 0.1428571
```

plotDeFinetti *Plot the De Finetti Diagram for Polarized Genotypes*

Description

This function calculates genotype frequencies from polarized genotypes, ideally imported using the importPolarized function. It plots individuals onto a ternary De Finetti diagram and includes a curve indicating Hardy-Weinberg equilibrium if specified.

Usage

```
plotDeFinetti(
  genotypes,
  cols,
  HWE = TRUE,
  tipLabels = c("Homozygous 0", "Heterozygous 1", "Homozygous 2"),
  ...
)
```

Arguments

genotypes	A character matrix comprising of _012 encodings.
cols	A character vector of colors with a length equal to the number of individuals (rows) in genotypes.
HWE	Logical indicating whether to plot the curve for Hardy-Weinberg Equilibrium.
tipLabels	A character vector of length 3 with names for the ternary plot vertices.
...	additional graphical parameters (see plot.default).

Details

To import and polarize genotypes, use the [importPolarized](#) function. Alternatively, the I4 matrix can be used as input for genotypes.

Value

No return value; the function is called for its side effects.

Examples

```
gen <- importPolarized(
  file = system.file("extdata", "data7x10.txt", package = "diemr"),
  changePolarity = c(TRUE, FALSE, TRUE, TRUE, FALSE, FALSE, TRUE, FALSE, FALSE, TRUE),
  ChosenInds = 1:7
)

plotDeFinetti(gen, cols = palette.colors(nrow(gen), "Accent"), pch = 19)
```

plotMarkerAxis	<i>Add a Marker Axis with Chromosome Names to a Plot of Polarized Genotypes</i>
----------------	---

Description

This function adds a marker axis with chromosome names to an existing plot of polarized genotypes. It requires that the plot is already created using [plotPolarized](#).

Usage

```
plotMarkerAxis(
  includedSites,
  ChosenSites,
  tickDist = 1e+06,
  axisInfo = NULL,
  ...
)
```

Arguments

`includedSites` A character path to a file with columns CHROM and POS.

`ChosenSites` A logical vector indicating which sites are to be included in the analysis.

`tickDist` A numeric indicating the spacing of physical tick marks along a chromosome.

`axisInfo` A list with user-defined tick positions and labels for marker axis. See Details.

... additional arguments.

Details

The `includedSites` file should ideally be generated by [vcf2diem](#) to ensure congruence between the plotted genotypes and the respective metadata.

Tick mark distances within a chromosome are located at `tickDist` and formatted to multiples of millions.

The optional `axisInfo` argument must have five named elements with the following information:

- `CHROMbreaks`: Numeric vector with positions defining ticks separating chromosomes. The metric for all positions is in the number of markers.
- `CHROMnamesPos`: Numeric vector with positions to place the chromosome labels.
- `CHROMnames`: Character vector with the names of the chromosomes. Must be the same length as `CHROMnamesPos`.
- `ticksPos`: Numeric vector with positions of ticks within chromosomes.
- `ticksNames`: Character vector with the names to be displayed at `ticksPos`.

When `axisInfo = NULL`, the function extracts the necessary information from the `includedSites` file.

Value

Returns an invisible `axisInfo` list with the tick positions and labels for the marker axis.

Examples

```
## Not run:
# Run this example in a working directory with write permissions
myo <- system.file("extdata", "myotis.vcf", package = "diemr")
vcf2diem(myo, "myo")
inds <- 1:14
```

```

fit <- diem("myo-001.txt", ChosenInds = inds, ploidy = FALSE)
gen <- importPolarized("myo-001.txt", fit$markerPolarity, inds)
h <- apply(gen, 1, function(x) pHetErrOnStateCount(sStateCount(x)))[1, ]
plotPolarized(gen, h, xlab = "")
plotMarkerAxis("myo-includedSites.txt", rep(TRUE, 11), tickDist = 100)

## End(Not run)

```

plotPolarized

Plot Polarized Genotypes

Description

Plots genotypes that can be optionally polarized.

Usage

```

plotPolarized(
  genotypes,
  HI,
  cols = c("#FFFFFF", "#800080", "#FFE500", "#008080"),
  ...
)

```

Arguments

genotypes	A character matrix comprising of <code>_012</code> encodings.
HI	A numeric vector of individual hybrid indices with length equal to number of rows in genotypes.
cols	A vector of four colors, representing missing data, homozygotes for genotype 0, heterozygotes and homozygotes for genotype 2.
...	Additional selected arguments passed to image and axis .

Details

To import and polarize genotypes, use the [importPolarized](#) function.

When using [diem](#), hybrid indices, HI, can be found in the file 'HIwithOptimalPolarities.txt'. Alternatively, calculate HI from the polarized genotypes as shown in the examples.

By default, the function plots colored tick marks for individuals, changing the color at the steepest change in sorted HI. The second and fourth colors in `cols` are used for the tick marks.

- To turn off this feature, use the argument `tick = FALSE`.
- To use custom tick mark colors, provide a vector of colors for all individuals (equal to the number of rows in genotypes). The **vector of colors must be ordered** according to order (HI).
- To include individual labels (e.g., accession numbers), provide a character vector with the **names in the same order as they are** in the genotypes.

Value

No return value, called for side effects. In the default plot, purple and green represent sides of the barrier to gene flow encoded as 0 and 2, respectively, yellow shows heterozygotes and white represents missing or undetermined genotypes. Individuals are ordered according to the sorted HI.

See Also

[plotMarkerAxis](#) to add chromosome information to the x axis.

Examples

```
gen <- importPolarized(
  file = system.file("extdata", "data7x10.txt", package = "diemr"),
  changePolarity = c(TRUE, FALSE, TRUE, TRUE, FALSE, FALSE, TRUE, FALSE, FALSE, TRUE),
  ChosenInds = 1:7
)

h <- apply(gen, 1, FUN = function(x) pHetErrOnStateCount(sStateCount(x)))[1, ]

plotPolarized(genotypes = gen, HI = h)

# Incorrect tick color order
plotPolarized(gen, h, col.ticks = c(rep("purple", 5), "green", "purple"), lwd = 3)

# Correct tick color order
plotPolarized(gen, h, col.ticks = c(rep("purple", 5), "green", "purple")[order(h)], lwd = 3)

# Correct individual label order
plotPolarized(gen, h, labels = c(paste("purple", 1:5), "green 1", "purple 6"), ylab = "")
```

rank2map

Convert SNP Ranks To Windows Corresponding to Mapping Distance

Description

This function estimates positions of ordered single nucleotide polymorphisms (SNPs) that correspond to a window spanning a user-defined distance in the SNP positions mapped to a reference. Each window is centered at the SNP mapped position. Conversion of a SNP rank position metric to a mapped position metric is useful for kernel smoothing of the diem output state along a genomic sequence.

Usage

```
rank2map(includedSites, ChosenSites = "all", windowSize = 1e+07, nCores = 1)
```

Arguments

includedSites	A character path to a file with columns CHROM and POS.
ChosenSites	A logical vector indicating which sites are to be included in the analysis.
windowSize	A numeric window size for metric conversion in base-pairs.
nCores	A numeric number of cores to be used for parallelisation. Must be nCores = 1 on Windows.

Details

Single nucleotide polymorphisms (SNPs) tend to be spread across a genome randomly. To facilitate interpretation of the diem output, the marker states should be assessed on the metric of their position along chromosomes (contigs). The windows for kernel smoothing might contain a variable number of markers. This function estimates which markers should be assessed together given their proximity on a chromosome.

Values in includedSites are in essence SNP positions in BED format with a header. The includedSites file should ideally be generated by [vcf2diem](#) to ensure congruence across all analyses.

The function reads SNP positions from the specified BED-like file and divides the genome into segments based on chromosomes. Each segment is then processed to identify genomic windows encompassing each SNP, considering the specified window size. This process is parallelized to enhance performance, and each SNP is considered within its chromosomal context to ensure accurate window placement.

Minimum value of windowSize is equal to 3, but in genomic data evaluations, window size should be at least two orders of magnitude larger. A good approximation of a useful minimum window size is $\$(\text{genome size}) / ((\text{number of SNPs}) / 2)\$$. Throughout the diemr package, windowSize refers to the genomic context of the respective SNP that the user wishes to consider when smoothing over the polarized genomic states.

Value

A two-column matrix with the number of rows corresponding to the number of ChosenSites, indicating start and end indices of adjacent markers that are within an interval of length windowSize centered on the specific marker.

Note

The unit of parallelization when using nCores > 1 is set per chromosome. This may differ from the parallelization approach used in [diem](#), where processing of compartment files is parallelized. Note that while compartment files can correspond to chromosomes, this is not necessarily the case.

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See Also

[smoothPolarizedGenotypes](#)

Examples

```
## Not run:
# Run this example in a working directory with write permissions
myo <- system.file("extdata", "myotis.vcf", package = "diemr")
vcf2diem(myo, "myo")
rank2map("myo-includedSites.txt", windowSize = 50)

## End(Not run)
```

smoothPolarizedGenotypes

Smooth Polarized Genotype States

Description

This function smooths polarized genotype states using a Laplace kernel density estimation. It calculates a smoothed version of the genotype states over specified physical extent of genomic content around a site. At each genomic position, the function returns a weighted mode of the genomic state.

Usage

```
smoothPolarizedGenotypes(
  genotypes,
  includedSites,
  ChosenSites = "all",
  windows = NULL,
  windowSize = 250000,
  ...
)
```

Arguments

genotypes	A character matrix comprising of _012 encodings.
includedSites	A character path to a file with columns CHROM and POS.
ChosenSites	A logical vector indicating which sites are to be included in the analysis.
windows	A two-column numeric matrix with indices of start and end positions for windows for all markers indicated by ChosenSites. If windows = NULL, the function calculates the windows using rank2map .
windowSize	A numeric window size for metric conversion in base-pairs.
...	Additional arguments to be passed to rank2map if windows = NULL.

Details

Ensure that ChosenSites match those used to import polarized genotypes.

The function uses a truncated and scaled Laplace kernel to weight the genotype states within a window around each marker position, based on physical positions of the markers.

The Laplace kernel weights are calculated for physical positions of the sites centered at the site being smoothed as:

$$\frac{10}{19} \exp\left(\frac{-x}{b}\right),$$

when $x < 0$, meaning that the site x is upstream of the site being smoothed, and as:

$$\frac{10}{19} \exp\left(\frac{x}{b}\right),$$

when $x \geq 0$, meaning that the site is downstream. The value x is the position of a neighbouring site relative to the site being smoothed, and b is the scale parameter of the Laplace kernel. The scale parameter is equal to:

$$b = \frac{\text{windowSize}}{2 \ln(20)}.$$

See Also

[rank2map](#)

Examples

```
## Not run:
# Run this example in a working directory with write permissions
myo <- system.file("extdata", "myotis.vcf", package = "diemr")
vcf2diem(myo, "myo")
fit <- diem("myo-001.txt", ChosenInds = 1:14)
gen <- importPolarized("myo-001.txt", changePolarity = fit$markerPolarity, ChosenInds = 1:14)
h <- apply(gen, 1, \ (x) pHetErrOnStateCount(sStateCount(x)))[1, ]
gen2 <- smoothPolarizedGenotypes(genotypes = gen,
  includedSites = "myo-includedSites.txt", windowSize = 50)
plotPolarized(gen, h)
plotPolarized(gen2, h)

## End(Not run)
```

sStateCount

Count states in a vector

Description

Counts genomic states in one sample.

Usage

```
sStateCount(s)
```

Arguments

s A character vector with elements "_", "0", "1", "2" representing missing data, homozygots for allele 1, heterozygots, and homozygots for allele 2. The vector should represent a single individual.

Details

Summarizes the number of markers that are fixed for an allele in the genome of one individual. This is used to construct the I4 matrix in [diem](#).

Value

Numeric vector of length 4 with counts of "_", "0", "1", "2" respectively.

See Also

[emPolarise](#) for changing marker polarity.

Examples

```
genotype <- c("0", "0", "_", "2", "1", "0", "1")
sStateCount(genotype)
# [1] 1 3 2 1

# calculate state counts for a polarised genotype
sStateCount(emPolarise(genotype, TRUE))
# [1] 1 1 2 3
```

testdata

Dataset of Fish Genotypes

Description

A subset of single nucleotide polymorphisms in fish for testing purposes of multiallelic markers.

Format

vcf file with 92 individuals and 6 markers.

Details

The data is used to test conversion of genotype data from vcf to diem format with the function `vcf2diem`.

Examples

```
filename <- system.file("extdata", "testdata.vcf", package = "diemr")
```

truncatedLaplace	<i>Truncated Laplace distribution to 95% of area under the curve, weights scaled to 10/19</i>
------------------	---

Description

Truncated Laplace distribution to 95% of area under the curve, weights scaled to 10/19

Usage

```
truncatedLaplace(x, laplaceScale)
```

Arguments

x	A vector of integers, representing centered positions of SNPs
laplaceScale	A numeric giving scale of the Laplace distribution.

Details

x must be within $\pm \text{laplaceScale} * \log(20)$

unbiasedWeightedStateChoice	<i>Selects weighted mode of the genomic state</i>
-----------------------------	---

Description

Selects weighted mode of the genomic state

Usage

```
unbiasedWeightedStateChoice(genomicStates, laplaceWeights)
```

Arguments

genomicStates	A character vector of genomic states in a given interval
laplaceWeights	A numeric vector of weights

vcf2diem

*Convert vcf files to diem format***Description**

Reads vcf files and writes genotypes of the most frequent alleles based on chromosome positions to diem format.

Usage

```
vcf2diem(
  SNP,
  filename,
  chunk = 1L,
  requireHomozygous = TRUE,
  ChosenInds = "all"
)
```

Arguments

SNP	A character vector with a path to the '.vcf' or '.vcf.gz' file, or an vcfR object. Diploid data are currently supported.
filename	A character vector with a path where to save the converted genotypes.
chunk	Numeric indicating by how many markers should the result be split into separate files.
requireHomozygous	A logical or numeric vector indicating whether to require the marker to have at least one or more homozygous individual(s) for each allele.
ChosenInds	A numeric or logical vector of indices of individuals to be included in the analysis.

Details

Importing vcf files larger than 1GB, and those containing multiallelic genotypes is not recommended. Instead, use the path to the vcf file in SNP. vcf2diem then reads the file line by line, which is a preferred solution for data conversion, especially for very large and complex genomic datasets.

The number of files vcf2diem creates depends on the chunk argument and class of the SNP object.

- Values of chunk < 100 are interpreted as the number of files into which to split data in SNP. For SNP object of class vcfR, the number of markers per file is calculated from the dimensions of SNP. When class of SNP is character, the number of markers per file is approximated from a model with a message. If this number of markers per file is inappropriate for the expected output, provide the intended number of markers per file in chunk greater than 100 (values greater than 10000 are recommended for genomic data). vcf2diem will scan the whole input specified in the SNP file, creating additional output files until the last line in SNP is reached.

- Values of chunk ≥ 100 mean that each output file in diem format will contain chunk number of lines with the data in SNP.

When the vcf file contains markers not informative for genome polarisation, those are removed and listed in a file ending with *omittedSites.txt* in the directory specified in the SNP argument or in the working directory. The omitted loci are identified by their information in the CHROM and POS columns, and include the QUAL column data. The last column is an integer specifying the reason why the respective marker was omitted. The reasons why markers are not informative for genome polarisation using diem are:

1. Marker has fewer than 2 alleles representing substitutions.
2. Required homozygous individuals for the 2 most frequent alleles are not present (optional, controlled by the requireHomozygous argument).
3. The second most frequent allele is found only in one heterozygous individual.
4. Dataset is invariant for the most frequent allele.
5. Dataset is invariant for the allele listed as the first ALT in the vcf input.

The CHROM, POS, and QUAL information for loci included in the converted files are listed in the file ending with *includedSites.txt*. Additional columns show which allele is encoded as 0 in its homozygous state and which is encoded as 2.

Value

No value returned, called for side effects.

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Examples

```
## Not run:
# vcf2diem will write files to a working directory or a specified folder
# make sure the working directory or the folder are at a location with write permission
myofile <- system.file("extdata", "myotis.vcf", package = "diemr")

vcf2diem(SNP = myofile, filename = "test1")
vcf2diem(SNP = myofile, filename = "test2", chunk = 3)

## End(Not run)
```

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