# Package 'PopGenome'

July 4, 2017

Type Package				
Title An Efficient Swiss Army Knife for Population Genomic Analyses				
Version 2.2.4				
<b>Date</b> 2017-07-3				
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<b>Depends</b> R (>= 2.14.2), ff				
Imports methods				
Suggests parallel, bigmemory, BASIX, WhopGenome				
<b>Description</b> Provides efficient tools for population genomics data analysis, able to process individual loci, large sets of loci, or whole genomes. PopGenome not only implements a wide range of population genetics statistics, but also facilitates the easy implementation of new algorithms by other researchers. PopGenome is optimized for speed via the seamless integration of C code.				
License GPL-3				
<pre>URL http://popgenome.weebly.com</pre>				
LazyLoad yes				
Copyright inst/COPYRIGHTS				
SystemRequirements zlib headers and library.				
Repository CRAN				
NeedsCompilation yes				
<b>Date/Publication</b> 2017-07-03 22:17:58 UTC				
R topics documented:				
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# Description

Achaz statistic

# Usage

```
## S4 method for signature 'GENOME'
Achaz.stats(object,new.populations=FALSE,new.outgroup=FALSE,subsites=FALSE)
```

# Arguments

```
an object of class "GENOME"
object
new.populations
                 list of populations. default:FALSE
new.outgroup
                 outgroup vector. default:FALSE
                  "transitions": SNPs that are transitions.
subsites
                  "transversions": SNPs that are transversions.
                  "syn": synonymous sites.
                  "nonsyn": nonsynonymous sites.
                  "exon": SNPs in exon regions.
                  "intron": SNPs in intron regions.
                  "coding": SNPs in coding regions (CDS).
                  "utr": SNPs in UTR regions.
                  "gene": SNPs in genes.
                 default:FALSE
```

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#### Value

```
returned value is a modified object of class "GENOME"
```

The following Slots will be modified in the "GENOME" object

Achaz Y statistic

#### References

Yach

Achaz G.,2008 Testing for neutrality in samples with sequencing errors. Genetics 179: 1409.

## **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# GENOME.class
# GENOME.class <- Achaz.stats(GENOME.class)
# GENOME.class <- Achaz.stats(GENOME.class,list(1:7,8:12))
# show the result:
# GENOME.class@Yach</pre>
```

BayeScanR

An R implementation of BayeScan (Foll \& Gagiotti 2008)

## **Description**

BayeScanR is an R implementation of BayeScan for analysis of codominant markers.

# Usage

```
BayeScanR(input,nb.pilot=10,pilot.runtime=2500,main.runtime=100000, discard=50000)
```

#### **Arguments**

input textfile or an R-object returned by getBayes()

nb.pilot number of pilot runs
pilot.runtime length of pilot runs
main.runtime length of main runs

discard how many runs in the main.loop should be discarded?

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## Value

returned value is an object of class "BAYESRETURN"

The following Slots will be filled

alpha alpha effects beta beta effects

var\_alpha variance of alpha values

a\_inc which alpha is included in the model

fst FST values
P P-value

#### References

[1] Foll M and OE Gaggiotti (2008). A genome scan method to identify selected loci appropriate for both dominant and codominant markers: A Bayesian perspective. Genetics 180: 977-993

# **Examples**

```
# GENOME.class <- readData("...\Alignments")
# GENOME.class <- F_ST.stats(GENOME.class,list(1:5,6:10))
# Bayes.input <- getBayes(GENOME.class)
# BAYES.class <- BayeScanR(Bayes.input)
# BAYES.class</pre>
```

 ${\tt calc.fixed.shared-methods}$ 

Fixed and shared polymorphisms

# Description

A generic function to calculate the number of fixed and shared polymorphisms.

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### Usage

```
## S4 method for signature 'GENOME'
calc.fixed.shared(object,
subsites=FALSE,
new.populations=FALSE,
fixed.threshold=1,
fixed.threshold.fst=1)
```

## **Arguments**

An object of class "GENOME" object

subsites "transitions": SNPs that are transitions.

"transversions": SNPs that are transversions.

"syn": synonymous sites.

"nonsyn": nonsynonymous sites. "exon": SNPs in exon regions. "intron": SNPs in intron regions.

"coding": SNPs in coding regions (CDS).

"utr": SNPs in UTR regions. "gene": SNPs in genes.

"intergenic": SNPs in intergenic regions.

new.populations

list of populations. default=FALSE

fixed.threshold

Polymorphisms are considered as fixed >= threshold value

fixed.threshold.fst

Polymorphisms are considered as fixed >= threshold value

#### **Details**

The nucleotide diversities have to be devided by GENOME.class@n.sites to give diversities per site.

# Value

Returned value is a modified object of class "GENOME"

The following slots will be modified in the "GENOME" object

Slot Description Reference n.fixed.sites Number of fixed sites [x] Number of shared sites n.shared.sites [x]

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3. n.monomorphic.sites [x] Number of monomorphic sites

#### References

[x]

#### **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# set.populations
# GENOME.class <- calc.fixed.shared(GENOME.class)</pre>
```

calc.R2-methods

Linkage statistics (R2, P-value, Distance)

# **Description**

This generic function calculates some linkage disequilibrium statistics.

# Usage

```
## S4 method for signature 'GENOME'
calc.R2(object,subsites=FALSE,lower.bound=0,upper.bound=1)
```

#### **Arguments**

object an object of class "GENOME" subsites same as in the other modules

lower.bound sites with minor-allele-frequency>=lower.bound are considered upper.bound sites with minor-allele-frequency<=upper.bound are considered

#### **Details**

Note, the pairwise comparisons are computed via combn(n.snps, 2).

# Value

The slot GENOME.class@region.stats@linkage.disequilibrium will be filled. (R2,P-value,Distance)

Fisher's Exact Test is used for the P-values.

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# **Examples**

```
# GENOME.class <- readData("...\Alignments")
# GENOME.class
# GENOME.class <- calc.R2(GENOME.class)
# show the result:
# GENOME.class@region.stats@linkage.disequilibrium
# [[x]][[y]] x:region, y:population</pre>
```

codontable

Prints the codon table which is used in the PopGenome framework

## **Description**

This functions prints the nucleotide triplets (as numerical values) and the corresponding protein character strings.

## Usage

codontable()

# Arguments

no arguments

# **Details**

The returned value is a list including two matrices.

The first matrix contains the amino acids and the second matrix the corresponding nucleotide triplets. In the PopGenome Vignette you can see how to manipulate these tables to use alternative genetic codes.

# **Examples**

```
# table <- codontable()</pre>
```

# table\$Proteins

# table\$Triplets

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concatenate.classes

Concatenate GENOME classes

# Description

This function concatenates objects of class GENOME, allowing to stitch together larger datasets from smaller objects.

## Usage

```
concatenate.classes(classlist)
```

# **Arguments**

```
classlist a list of GENOME objects
```

#### Value

The function creates an object of class "GENOME".

# **Examples**

```
# a <- readData("Three_Alignments/")
# b <- readData("Two_Alignments/")
# ab <- concatenate.classes(list(a,b))
# ab <- neutrality.stats(ab)
# ab@Tajima.D
# ab@region.names</pre>
```

 ${\tt concatenate.regions}$ 

Concatenate regions

# Description

This function concatenates the regions/chunks contained in one GENOME object.

# Usage

```
concatenate.regions(object)
```

10 count.unknowns-methods

### **Arguments**

object of class GENOME

# Value

The function creates an object of class "GENOME".

## **Examples**

count.unknowns-methods

Calculate missing nucleotide frequencies

# Description

A generic function to calculate the missing nucleotide frequencies.

# Usage

```
## S4 method for signature 'GENOME'
count.unknowns(object)
```

# **Arguments**

object An object of class "GENOME"

#### Value

Returned value is a modified object of class "GENOME"

The slot GENOME.class@missing.freqs for the missing frequencies for the whole region. The slot GENOME.class@region.stats@missing.freqs for the missing frequencies for each SNP in a given region

## **Examples**

```
# GENOME.class <- readData("VCF", format="VCF", include.unknown=TRUE)
# GENOME.class@region.stats
# GENOME.class <- count.unknowns(GENOME.class)
# GENOME.class@missing.freqs
# GENOME.class@region.stats@missing.freqs</pre>
```

create.PopGenome.method

Integration of own functions into the PopGenome-framework

#### **Description**

This function generates a skeleton for a PopGenome function. It thereby facilitates the effortless integration of new methods into the PopGenome framework.

## Usage

```
\verb|create.PopGenome.method(function.name,population.specific=TRUE)| \\
```

#### **Arguments**

```
function.name name of your function
population.specific
```

TRUE:function returns one value per population.FALSE:function returns one value calculated across all populations (as in the case of FST measurements)

#### **Details**

This mechanism enables you to use your own functions in the PopGenome environment. The functions can also be applied to sliding windows or subsites.

Please look at the generated function, which documents where to place your own function in detail.

## **Examples**

```
# GENOME.class <- readData(".../Alignments")
# create.PopGenome.method("myFunction")
# edit myFunction.R
# source("myFunction")
# value <- myFunction(test)
# value</pre>
```

12 detail.stats-methods

```
detail.stats-methods Several statistics
```

# **Description**

This generic function calculates some mixed statistics.

# Usage

# **Arguments**

```
object
                 an object of class "GENOME"
new.populations
                 list of populations.
                 outgroup sequences.
new.outgroup
subsites
                  "transitions": SNPs that are transitions.
                  "transversions": SNPs that are transversions.
                  "syn": synonymous sites.
                  "nonsyn": nonsynonymous sites.
                  "exon": SNPs in exon regions.
                  "intron": SNPs in intron regions.
                  "coding": SNPs in coding regions (CDS).
                  "utr": SNPs in UTR regions.
                  "gene": SNPs in genes.
biallelic.structure
                 fixed and shared polymorphisms (stored in GENOME.class@region.stats).
```

detail.stats-methods

```
mismatch.distribution
statistics based on mismatch distribution
site.spectrum minor allele frequency of each SNP
site.FST computes FST for each SNP
```

#### Value

The return value is a modified object of class "GENOME"

The following Slots will be modified in the "GENOME" object

MDSD ...
MDG1 ...
MDG2 ...

region.stats the slot biallelic.structure and minor.allele.freqs will be filled

The function get.detail(GENOME.class, biallelic.structure=TRUE) returns a matrix for each region, where

0	population is polymorphic, the remaining individuals are polymorphic
1	population is polymorphic, the remaining individuals are monomorphic
2	population is monomorphic, the remaining individuals are polymorphic
3	population is monomorphic, the remaining individuals are monomorphic with the same value
4	population is monomorphic, the remaining individuals are monomorphic with different values

# **Examples**

14 diversity.stats-methods

```
# GENOME.class@region.stats@biallelic.structure[[1]]
```

```
diversity.stats-methods
```

Diversities

# Description

A generic function to calculate nucleotide & haplotype diversities.

# Usage

```
## S4 method for signature 'GENOME'
diversity.stats(object,new.populations=FALSE,subsites=FALSE,pi=FALSE)
```

#### **Arguments**

```
object An object of class "GENOME"

new.populations
list of populations. default=FALSE

subsites "transitions": SNPs that are transitions.
    "transversions": SNPs that are transversions.
    "syn": synonymous sites.
    "nonsyn": nonsynonymous sites.
    "exon": SNPs in exon regions.
    "intron": SNPs in intron regions.
    "coding": SNPs in coding regions (CDS).
    "utr": SNPs in UTR regions.
    "gene": SNPs in genes.
    "intergenic": SNPs in intergenic regions.
```

pi Nei's calculation of pi

#### **Details**

The nucleotide diversities have to be devided by GENOME.class@n.sites to give diversities per site.

#### Value

Returned value is a modified object of class "GENOME"

The following slots will be modified in the "GENOME" object

Slot Reference Description
1. nuc.diversity.within [1,3] Nucleotide diversity (within the population)
2. Pi [2] Diversity from Nei (within the population)
3. hap.diversity.within [1] Haplotype diversity (within the population)

#### References

- [1] Hudson, R. R., M. Slatkin, and W.P. Maddison (1992). *Estimating of levels of gene flow from DNA sequence data*. Gentics 13(2),583-589
- [2] Nei, M. (1987). Molecular Evolutionary Genetics. Columbia Univ. Press, New York.
- [3] Wakeley, J. (1996). The Variance of Pairwise Nucleotide Differences in Two Populations with Migration. THEORETICAL POPULATION BIOLOGY. 49, 39-57.

#### **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# GENOME.class
# GENOME.class <- diversity.stats(GENOME.class)
# GENOME.class <- diversity.stats(GENOME.class,list(1:4,5:10))
# GENOME.class <- diversity.stats(GENOME.class,
# list(c("seq1","seq5","seq3"),c("seq2","seq8")))
# show the result:
# GENOME.class@nuc.diversity.within</pre>
```

```
diversity.stats.between-methods

Diversities
```

# Description

A generic function to calculate nucleotide & haplotype diversities between populations (dxy).

#### Usage

```
## S4 method for signature 'GENOME'
diversity.stats.between(object,new.populations=FALSE,subsites=FALSE,keep.site.info=FALSE,
haplotype.mode=FALSE, nucleotide.mode=TRUE)
```

## **Arguments**

object An object of class "GENOME"

new.populations

list of populations. default=FALSE

subsites "transitions": SNPs that are transitions.

"transversions": SNPs that are transversions.

"syn": synonymous sites.

"nonsyn": nonsynonymous sites.
"exon": SNPs in exon regions.
"intron": SNPs in intron regions.

"coding": SNPs in coding regions (CDS).

"utr": SNPs in UTR regions. "gene": SNPs in genes.

"intergenic": SNPs in intergenic regions.

keep.site.info Store SNP specific values in the region.stats

haplotype.mode Haplotype Diversities

nucleotide.mode

**Nucleotide Diversities** 

#### **Details**

The nucleotide diversities have to be devided by GENOME.class@n.sites to give diversities per site.

#### Value

Returned value is a modified object of class "GENOME"

\_\_\_\_\_

The following slots will be modified in the "GENOME" object

Slot Reference Description

nuc.diversity.between [1,3]
 hap.diversity.between [1]
 Nucleotide diversity (between the population)
 Haplotype diversity (between the population)

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#### References

[1] Hudson, R. R., M. Slatkin, and W.P. Maddison (1992). Estimating of levels of gene flow from DNA sequence data. Gentics 13(2),583-589

[2] Wakeley, J. (1996). The Variance of Pairwise Nucleotide Differences in Two Populations with Migration. THEORETICAL POPULATION BIOLOGY. 49, 39-57.

## **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# GENOME.class
# GENOME.class <- diversity.stats.between(GENOME.class)
# GENOME.class <- set.populations(GENOME.class, list(...))
# GENOME.class <- diversity.stats.between(GENOME.class)
# GENOME.class <- diversity.stats(GENOME.class,
# list(c("seq1","seq5","seq3"),c("seq2","seq8")))
# show the result:
# GENOME.class@nuc.diversity.within</pre>
```

fasta\_file

FASTA file (subdirectory "data")

#### **Description**

The FASTA files (unpacked) in the subdirectory "data" of the PopGenome package have to be stored in a folder (multiple files can be stored in this folder). The folder name is then used as the input for the readData function.

 $F\_ST.stats-methods$ 

Fixation Index

#### **Description**

A generic function to calculate some F-statistics and nucleotide/haplotype diversities.

# Usage

```
## S4 method for signature 'GENOME'
F_ST.stats(
object,
new.populations=FALSE,
subsites=FALSE,
```

F\_ST.stats-methods

```
detail=TRUE,
mode="ALL",
only.haplotype.counts=FALSE,
FAST=FALSE
)

## $4 method for signature 'GENOME'
get.diversity(object,between=FALSE)
## $4 method for signature 'GENOME'
get.F_ST(object,mode=FALSE,pairwise=FALSE)
```

# **Arguments**

object An object of class "GENOME"

new.populations

list of populations. default:FALSE

subsites "transitions": SNPs that are transitions.

"transversions": SNPs that are transversions.

"syn": synonymous sites.

"nonsyn": nonsynonymous sites.
"exon": SNPs in exon regions.
"intron": SNPs in intron regions.

"coding": SNPs in coding regions (CDS).

"utr": SNPs in UTR regions. "gene": SNPs in genes.

"intergenic": SNPs in intergenic regions.

detail detail statistics. Note: slower!

between TRUE: show between-diversities. FALSE: show within-diversities

mode mode="haplotype" or mode="nucleotide"

only.haplotype.counts

only calculate the haplotype counts

FAST if TRUE only calculate a subset of statistics. see details!

pairwise show paiwise comparisons. default:FALSE

## Details

If FAST is switched on, this module only calculates nuc. diversity. within, hap. diversity. within, haplotype.  $F_ST$ , nucleotide.  $F_ST$  and pi.

#### Note:

- 1) The nucleotide diversities have to be devided by the size of region considered (e.g. GENOME@n.sites) to give diversities per site.
- 2) When missing or unknown nucleotides are included (include.unknown=TRUE) those sites are completely deleted in case of haplotype based statistics.
- 3) The function detail.stats(...,site.FST=TRUE) will compute SNP specific FST values which are then stored in the slot GENOME.class@region.stats@site.FST.

F\_ST.stats-methods

4) We recommend to use mode="nucleotide" in case you have many unknowns included in your dataset.

## Value

	Slot	Reference	Description
1.	haplotype.F_ST	[1]	Fixation Index based on haplotype frequencies
2.	nucleotide.F_ST	[1]	Fixation Index based on minor allele frequencies
3.	Nei.G_ST	[2]	Nei's Fixation Index
4.	Hudson.G_ST	[3]	see reference
5.	Hudson.H_ST	[3]	see reference
6.	Hudson.K_ST	[3]	see reference
7.	nuc.diversity.within	[1,5]	Nucleotide diversity (within the population)
8.	hap.diversity.within	[1]	Haplotype diversity (within the population)
9.	Pi	[4]	Nei's diversity (within the population)
10.	hap.F_ST.vs.all	[1]	Fixation Index for each population against all other individuals (haplotype)
11.	nuc.F_ST.vs.all	[1]	Fixation Index for each population against tall other individuals (nucleotide)
12.	hap.diversity.between	[1]	Haplotype diversities between populations
13.	nuc.diversity.between	[1,5]	Nucleotide diversities between populations
14.	nuc.F_ST.pairwise	[1]	Fixation Index for every pair of populations (nucleotide)
15.	hap.F_ST.pairwise	[1]	Fixation Index for every pair of populations (haplotype)
16.	Nei.G_ST.pairwise	[2]	Fixation Index for every pair of populations (Nei)
17.	region.stats		an object of class "region.stats" for detailed statistics

#### References

- [1] Hudson, R. R., M. Slatkin, and W.P. Maddison (1992). *Estimating levels of gene flow from DNA sequence data*. Gentics 13(2),583-589
- [2] Nei, M. (1973). Analysis of gene diversity in subdivided populations. Proc.Natl. Acad. Sci. USA 70: 3321-3323
- [3] Hudson, R. R., Boos, D.D. and N. L. Kaplan (1992). A statistical test for detecting population subdivison. Mol. Biol. Evol. 9: 138-151.
- [4] Nei, M. (1987). Molecular Evolutionary Genetics. Columbia Univ. Press, New York.
- [5] Wakeley, J. (1996). The Variance of Pairwise Nucleotide Differences in Two Populations with Migration. THEORETICAL POPULATION BIOLOGY. 49, 39-57.

# See Also

# methods?F\_ST.stats.2 #F\_ST.stats.2

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#### **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# GENOME.class <- F_ST.stats(GENOME.class)
# GENOME.class <- F_ST.stats(GENOME.class,list(1:4,5:10),subsites="syn")
# GENOME.class <- F_ST.stats(GENOME.class,list(c("seq1","seq5","seq3"),c("seq2","seq8")))
# show the result:
# get.F_ST(GENOME.class)
# get.F_ST(GENOME.class, pairwise=TRUE)
# get.diversity(GENOME.class, between=TRUE)
# GENOME.class@Pi --> population specific view
# GENOME.class@region.stats
```

F\_ST.stats.2-methods Fixation Index (2)

## **Description**

A generic function to calculate some FST measurenments.

## Usage

```
## S4 method for signature 'GENOME'
F_ST.stats.2(object,new.populations="list",subsites=FALSE,snn=TRUE,Phi_ST=FALSE)
```

## Arguments

Phi\_ST

```
object
                  An object of class "GENOME"
new.populations
                  list of populations. default=FALSE
                  "transitions": SNPs that are transitions.
subsites
                  "transversions": SNPs that are transversions.
                  "syn": synonymous sites.
                  "nonsyn": nonsynonymous sites.
                  "exon": SNPs in exon regions.
                  "intron": SNPs in intron regions.
                  "coding": SNPs in coding regions (CDS).
                  "utr": SNPs in UTR regions.
                  "gene": SNPs in genes.
                  "intergenic": SNPs in intergenic regions.
                  Snn statistic from Hudson
snn
```

Statistic from Excoffier et al.

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#### Value

Returned value is an modified object of class "GENOME"

Following slots will be modified in the "GENOME" object

Slot Reference Description

1. Hudson.Snn Snn statistic from Hudson (2000) [1] Phi\_ST from Excoffier (1992) 2. Phi\_ST [2]

#### References

[1] Hudson, R. R. (2000). A new statistic for detecting genetic differentiation. Genetics 155: 2011-2014.

[2] Excoffier, L., Smouse, P., Quattro, J. (1992), Analysis of molecular variance inferred from metric distances among DNA haplotypes: application to human mitochondrial DNA restriction data. Genetics 131: 479-91

# **Examples**

```
# GENOME.class <- readData("\home\Alignments")</pre>
# GENOME.class
# GENOME.class <- F_ST.stats.2(GENOME.class)</pre>
# GENOME.class <- F_ST.stats.2(GENOME.class,list(1:4,5:10))</pre>
# GENOME.class <- F_ST.stats.2(GENOME.class,
# list(c("seq1", "seq5", "seq3"), c("seq2", "seq8")))
# show the result:
# GENOME.class@Hudson.Snn
```

GENOME-class

Class "GENOME"

#### **Description**

A class where all data and calculated values are stored

#### **Slots**

```
BIG.BIAL: Biallelic matrix as an ff-object
```

SLIDE.POS: Positions of biallelic sites (Sliding window mode)

big.data: ff-package?

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```
gff.info: Gff information?
snp.data: SNP data?
basepath: The basepath of the data
project: ---
populations: Populations definded before reading data
poppairs: —
outgroup: A vector of outgroup sequences
region.names: Names/identifier of each region
feature.names: Feature attributes of a given region
genelength: Number of regions
keep.start.pos: Start positions for sliding window
n.sites: Total number of sites
n.sites2: Total number of sites
n.biallelic.sites: Number of biallelic sites (SNPs)
n.gaps: Number of gaps observed in the data
n.unknowns: Number of unknown.positions
n.valid.sites: Sites without gaps
n.polyallelic.sites: Sites with more than two variants
trans.transv.ratio: Transition-transversion ratio
Coding. region: Number of nucleotides in CDS regions
UTR. region: Number of nucleotides in UTR regions
Intron.region: Number of nucleotides in Intron regions
Exon.region: Number of nucleotides in Exon regions
Gene.region: Number of nucleotides in Gene regions
Pop_Neutrality: Populations defined in the neutrality module
Pop_FSTN: Populations defined in the FST (nucleotide) module
Pop_FSTH: Populations defined in the FST (haplotype) module
Pop_Linkage: Populations defined in the Linkage module
Pop_Slide: —
Pop_MK: Populations defined in the MK module
Pop_Detail: Populations defined in the Detail module
Pop_Recomb: Populations defined in the Recombination module
Pop_Sweeps: Populations defined in the Selective sweeps module
FSTNLISTE: —
nucleotide.F_ST: Nucleotide FST
nucleotide.F_ST2: —
nuc.diversity.between: Nucleotide diversity between the populations
```

nuc.diversity.within: Nucleotide diversity within the populations nuc.F\_ST.pairwise: FST for each pair of populations nuc.F\_ST.vs.all: FST for one population vs. all other individuals n.haplotypes: hap.diversity.within: Haplotype diversity withing the populations hap.diversity.between: Haplotype diversity between the populations Pi: Pi from Nei PIA\_nei: Pi between the populations haplotype.counts: Counts of the haplotypes observed haplotype.F\_ST: Haplotype FST hap.F\_ST.pairwise: Haplotype diversity for each pair of populations Nei.G\_ST.pairwise: Haplotype diversity for each pair of populations hap.F\_ST.vs.all: FST for one population vs. all other individuals Nei.G\_ST: GST from Nei Hudson. G ST: GST from Hudson Hudson, H ST: HST from Hudson Hudson.K\_ST: KST from Hudson Hudson. Snn: Snn from Hudson Phi\_ST: Fixation index from Excoffier hap.pair.F\_ST: — MKT: Mcdonald-Kreitman values Tajima.D: Tajima's D SLIDE: — Fay.Wu.H: Zeng.E: theta\_Tajima: theta\_Watterson: theta\_Fu.Li: theta\_Achaz.Watterson: theta\_Achaz.Tajima: theta\_Fay.Wu: theta\_Zeng: Fu.Li.F: Fu.Li.D: Yach: n.segregating.sites: Total number of segregating sites Rozas.R\_2:

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```
Fu.F_S:
Strobeck.S:
Kelly.Z_nS:
Rozas.ZZ:
Rozas.ZA:
Wall.B:
Wall.Q:
mult.Linkage: Linkage disequilibrium between regions
RM: Minimum number of recombination events (Hudson)
CL: Composite likelihood of SNPs (Nielsen et. al)
CLmax: Max. composite likelihood of SNPs (Nielsen et.al)
CLR: Composite likelihood ratio test (Nielsen et. al)
MDSD:
MDG1:
MDG2:
genes:
region.data: Detailed information about the data
region.stats: Detailed (site-specific) statistics
D Pattersons D statistic
f the fraction of the genome that is admixed
jack.knife jacknife mode
missing.freqs: Missing nucleotide frequency
n.fixed.sites: ...
n.shared.sites: ...
n.monomorphic.sites: ...
BD: ...
BDF: ...
BDF_bayes: ...
alpha_ABBA: ...
alpha_BABA: ...
beta_BBAA: ...
Bd_clr: ...
Bd_dir: ...
D.pval: ...
D.z: ...
P.Bd_clr: ...
RNDmin: ...
```

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#### Methods

detail.stats Several misc. statistics **diversity.stats** Haplotype and nucleotide diversities diversity.between Haplotype and nucleotide diversities F\_ST.stats.2 Snn from Hudson **F\_ST.stats** Fixation index getBayes Get the input for BayeScanR get.detail Get the results from the Detail module get.codons Get information about the nature of codon changes get.diversity Get diversities from the FST module get.F ST Get FST values from the FST module get.linkage Get the values from the Linkage module get.MKT Get Mcdonald-Kreitman values getMS get.neutrality Get the values from the Neutrality module get.status Status of calculations **get.sum.data** Get some data observed from the alignments linkage.stats Linkage disequilibrium calc.R2 Linkage disequilibrium mult.linkage.stats Linkage disequilibrium between regions recomb.stats Recombination statistics sweeps.stats Selective sweeps Achaz.stats Achaz's statistics **get.recomb** Get the values from the Recombination module **get.sweeps** Get the values frome the Selective Sweep module **set.ref.positions** Set the SNP positions set.synnonsyn Verify synonymous positions **splitting.data** Split the data into subsites MKT MKT Test neutrality.stats Neutrality statistics popFSTN Internal function **get.biallelic.matrix** Print the biallelic.matrix **set.populations** Define the populations set.outgroup Define the outgroup **get.individuals** get the names/IDs of individuals region.as.fasta Extract the region as a fasta file show —

show.slots Show slots of the class GENOME

**sliding.window.transform** Transform a GENOME object into a new object suitable for sliding window analysis

```
usage —
```

PG\_plot.biallelic.matrix Plot the biallelic matrix introgression.stats Methods to measure archaic admixture count.unknowns Calculates the frequencies of missing nucleotides calc.fixed.shared Calculates the frequencies of missing nucleotides

set.filter SNP Filtering

## Author(s)

**Bastian Pfeifer** 

#### References

See the documentation for each module

# Examples

```
#GENOME.class <- readData("Alignments")
#GENOME.class@n.sites
#GENOME.class@region.names</pre>
```

```
get.biallelic.matrix-methods
```

Get the biallelic matrix

# Description

This function returns the biallelic matrix of a specific region.

# Usage

```
## S4 method for signature 'GENOME'
get.biallelic.matrix(object,region)
```

# Arguments

object An object of class "GENOME"

region ID of the region

get.codons-methods 27

#### Value

Biallelic matrix

rows: names of individuals columns: biallelic sites

## **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# get.biallelic.matrix(GENOME.class,7) # biallelic matrix of the 7th alignment</pre>
```

get.codons-methods

Detailed information about the nature of codon changes

## **Description**

This generic function returns some information about the codon changes resulting from the observed SNPs.

## Usage

```
## S4 method for signature 'GENOME'
get.codons(object, regionID)
```

# Arguments

object an object of class "GENOME"

regionID what region/alignment should be analyzed?

## **Details**

The slot GENOME.class@region.data@synonymous and GENOME.class@region.data@codons have to be set.

The data have to be read in with the correponding GFF file.

The function set.synnonsyn(..., save.codons=TRUE) sets the syn/nonsny sites in case of SNP data and stores the corresponding codon changes.

#### Value

The function get.codons returns a data.frame with the following information

- 1 Position of the SNPs
- 2 Major Codon
- 3 Minor Codon

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4	Major amino acid
5	Minor amino acid
6	synonymous (TRUE/FALSE)
7	Polarity of the major amino acid
8	Polarity of the minor amino acid

#### **Examples**

```
# Alignments
# GENOME.class <- readData("FASTA",gffpath="GFF")
# get.codons(GENOME.class,1)
# SNP data
# GENOME.class <- readData("VCF",gffpath="GFF")
# GENOME.class <- set.synnonsyn(GENOME.class, ref.chr="ref.fas",save.codons=TRUE)
# get.codons(GENOME.class,1)</pre>
```

get.feature.names

Feature informations and GFF-attributes

#### **Description**

Returns feature names and additional attributes for a given region

# Usage

```
get.feature.names(object, gff.file, chr)
```

#### **Arguments**

object An object of class GENOME gff.file The corresponding GFF file

chr The chromosome/scaffold identifier

#### **Details**

The algorithm uses the information stored in GENOME.class.split@region.names to iterate over the GFF file and returns attribute plus feature informations for each given region. Note, the functions splitting.data, split\_data\_into\_GFF\_attributes or sliding.window.transform should be performed prior to that.

The slot region.names must have the following form: "pos1 - pos2".

get.individuals-methods 29

## Value

The returned value is a character vector of length length(GENOME.class.split@region.names)

#### **Examples**

```
# GENOME.class <- readVCF("chr1.vcf.gz",1000,"1",1,100000)
# GENOME.class.split <- split_data_into_GFF_attributes(GENOME.class,"Homo_sapiens.GRCh37.73.gtf",
# "1", "gene_name")
# GENOME.class.split@region.names
# info <- get.feature.names(GENOME.class.split, gff.file="Homo_sapiens.GRCh37.73.gtf", chr="1")
# GENOME.class.split <- splitting.data(GENOME.class, subsites="gene")
# GENOME.class.split@region.names
# info <- get.feature.names(GENOME.class.split, gff.file="Homo_sapiens.GRCh37.73.gtf", chr="1")</pre>
```

get.individuals-methods

Print the names/IDs of individuals

# Description

Extract the names/IDs of individuals.

## Usage

```
## S4 method for signature 'GENOME'
get.individuals(object,region=FALSE)
```

## **Arguments**

```
object an object of class "GENOME"
region a vector of regions. Default: ALL
```

## **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# get.individuals(GENOME.class)</pre>
```

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get.status-methods

State of calculations

# Description

Some information about the definitions of populations and subsites.

# Usage

```
## S4 method for signature 'GENOME'
get.status(object)
```

# Arguments

object

An object of class "GENOME"

## **Examples**

```
# get.status(GENOME.class)
```

getBayes-methods

Get values for BayeScanR

# **Description**

This function returns the values that are necessary to run BayeScanR.

# Usage

```
## S4 method for signature 'GENOME'
getBayes(object,snps=FALSE)
```

# Arguments

object An object of class "GENOME" snps SNPs are considered seperately

# Value

coming soon!

get\_gff\_info 31

#### References

Foll M and OE Gaggiotti (2008). A genome scan method to identify selected loci appropriate for both dominant and codominant markers: A Bayesian perspective. Genetics 180: 977-993

# Examples

```
# GENOME.class <- readData("\home\Alignments")
# GENOME.class <- F_ST.stats(GENOME.class,list(1:4,5:10))
# Bayes.input <- getBayes(GENOME.class)
# Bayes.class <- BayeScanR(Bayes.input)</pre>
```

get\_gff\_info

Annotation info

# **Description**

This function extracts annotation information from a GTF/GFF file.

#### Usage

```
get_gff_info(object=FALSE,gff.file,chr,position,feature=FALSE,extract.gene.names=FALSE)
```

# Arguments

object of class GENOME
gff.file basepath of the GTF/GFF file

chr the chromosome

position reference positions or region id (when object is specified)
feature feature to search for in the gff-file. returns a list of positions

extract.gene.names

returns the gene names of the chromosome

#### **Details**

This function extracts annotation information from a GTF/GFF file.

# Examples

```
# get_gff_info("Arabidopsis.gff",chr=1,200202)
# get_gff_info(GENOME.class,"Arabidopsis.gff",chr=1,position=3)
```

gff\_file

GFF file (subdirectory "data")

#### **Description**

A typical GFF file which should be stored in a folder (for example in "GFF"). This folder is the input for the readData(...,gffpath="GFF") function. The corresponding FASTA file is stored in the "data" subdirectory of the PopGenome package. It has to be stored in a folder with the SAME NAME as the GFF file (for example in "FASTA"). readData("FASTA", gffpath="GFF")

```
GFF_split_into_scaffolds
```

Split a GFF file into multiple scaffold-GFFs

# **Description**

This function splits a GFF file into multiple GFFs including data for exactly one scaffold each.

#### Usage

```
GFF_split_into_scaffolds(GFF.file, output.folder)
```

#### Arguments

```
GFF.file the basepath of the GFF file
```

output.folder name of the folder where the GFFs should be stored

#### **Details**

The algorithm splits the GFF into multiple scaffold based GFFs and stores the files in a given folder. This folder can be used as an input for readData(,gffpath="")

#### Value

**TRUE** 

#### **Examples**

```
# GFF_split_into_scaffolds("GFFfile.gff","scaffoldGFFs")
# test <- readData("scaffoldVCFs", format="VCF", gffpath="scaffoldGFFs")</pre>
```

```
introgression.stats-methods
```

Introgression statistics

## **Description**

A generic function to estimate archaic admixture.

#### Usage

```
## S4 method for signature 'GENOME'
introgression.stats(object,
subsites=FALSE,
do.D=FALSE,
do.BD=FALSE,
do.BDF=FALSE,
keep.site.info=TRUE,
block.size=FALSE,
dxy.table=FALSE,
D.global=FALSE,
do.CLR=FALSE,
do.CLR=FALSE,
dd=2,
do.RNDmin=FALSE,
lambda=1)
```

#### **Arguments**

```
object
                  An object of class "GENOME"
                  "transitions": SNPs that are transitions.
subsites
                  "transversions": SNPs that are transversions.
                  "syn": synonymous sites.
                  "nonsyn": nonsynonymous sites.
                  "exon": SNPs in exon regions.
                  "intron": SNPs in intron regions.
                  "coding": SNPs in coding regions (CDS).
                  "utr": SNPs in UTR regions.
                  "gene": SNPs in genes.
                  "intergenic": SNPs in intergenic regions.
do.D
                 Pattersons D and Martin's f statistic
do.BD
do.BDF
                 Bd-fraction
keep.site.info Keep site specific values
```

jacknife block.size dxy.table ... not used ... D.global The global set of D,Bd values do.CLR Composite Likelihood approach dgt Digit for the CLR test RNDmin (Rosenzweig, 2016) do.RNDmin

lambda Scale ... not used ...

#### **Details**

To perform the D and f statistic one needs to define 3 populations via the function set.populations, where the third population represent the archaic population. In addition, an outgroup is required and have to be set via the function set.outgroup. Here, only SNPs where the outgroup is monomorphic are considered. f is the fraction of the genome that is admixed [2].

#### Value

Returned value is a modified object of class "GENOME"

The following slots will be modified in the "GENOME" object

	Slot	Reference	Description
1.	D	[1;eq. 2]	Pattersons D statistic
2.	f	[2]	f statistic
3.	BDF	[x]	Bd-fraction
4.	RNDmin	[x]	RNDmin

#### References

[1] Durand, E. Y., Patterson, N. J., Reich, D., & Slatkin, M. (2011). Testing for ancient admixture between closely related populations.

Molecular Biology and Evolution, 28(8), 2239–2252. doi:10.1093/molbev/msr048

[2] Simon H Martin, Kanchon K Dasmahapatra, Nicola J Nadeau, et al. (2013). Genome-wide evidence for speciation with gene flow in Heliconius butterflies.

Genome Res. doi:10.1101/gr.159426.113

#### **Examples**

```
# GENOME.class <- readData("\home\Alignments")</pre>
# GENOME.class <- set.populations(GENOME.class, list(1:3,4:8,9:12))
# GENOME.class <- set.outgroup(GENOME.class,13)</pre>
# GENOME.class <- introgression.stats(GENOME.class, do.D=TRUE)</pre>
# GENOME.class <- introgression.stats(GENOME.class, do.BDF=TRUE)</pre>
# show the result:
```

jack.knife.transform 35

```
# GENOME.class@D
# GENOME.class@f
# GENOME.class@BDF
```

```
jack.knife.transform Jacknife Transformation
```

# Description

This generic function transforms an existing object of class "GENOME" into another object of class "GENOME", in which each region corresponds to the (JACKNIFE!) window. Each jacknife window will be excluded from the analyses and the calculation will be applied to the union of all other windows.

#### Usage

```
## S4 method for signature 'GENOME'
jack.knife.transform(object,
width=7, jump=5,
type=1,
start.pos=FALSE,end.pos=FALSE
)
```

### **Arguments**

```
object an object of class "GENOME"
width window size. default:711
jump jump size. default:5
type 1 scan only biallelic positions (SNPs), 2 scan the genome. default:1
start.pos start position
end.pos end position
```

# Value

The function creates a transformed object of class "GENOME".

# Note

This function currently is only available for SNP data formats. PopGenome will scan the data from position 1 to the last observed SNP if start or end-positions are not specified. This mechanism can also be applied to the splitting.data() function. Just set split.GENOME.class@jack.knife <- TRUE after splitting the data.

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#### **Examples**

```
# GENOME.class <- readData("...", format="VCF")
# jack.GENOME.class <- jack.knife.transform(GENOME.class,100,100)
# jack.GENOME.class <- neutrality.stats(jack.GENOME.class)
# jack.GENOME.class@Tajima.D</pre>
```

linkage.stats-methods Linkage Disequilibrium

## Description

A generic function to calculate some linkage disequilibrium statistics.

## Usage

```
## S4 method for signature 'GENOME'
linkage.stats(object,new.populations=FALSE,subsites=FALSE,detail=FALSE,
do.ZnS,do.WALL=TRUE)
## S4 method for signature 'GENOME'
get.linkage(object)
```

#### **Arguments**

```
object
                  An object of class "GENOME"
new.populations
                 list of populations. default=FALSE
                  "transitions": SNPs that are transitions.
subsites
                  "transversions": SNPs that are transversions.
                  "syn": synonymous sites.
                  "nonsyn": nonsynonymous sites.
                  "exon": SNPs in exon regions.
                  "intron": SNPs in intron regions.
                  "coding": SNPs in coding regions (CDS).
                  "utr": SNPs in UTR regions.
                  "gene": SNPs in genes.
                  default:FALSE
detail
                 if you want to calculate some detailed statistics. This can be considerably
                  slower! default:FALSE
do.ZnS
                  calculate ZnS, ZA and ZZ
do.WALL
                 calculate Wall's B/Q
```

#### **Details**

Note, the pairwise comparisons are computed via combn(n.snps, 2).

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# Value

The return value is a modified object of class "GENOME"

The following slots will be modified in the "GENOME" object

	Slot	Reference	Description
1.	Wall.B	[2]	Wall \$B\$ statistic (only adjacent positions are considered)
2.	Wall.Q	[2]	Wall \$Q\$ statistic (only adjacent positions are considered)
3.	Kelly.Z_nS	[3]	Kelly \$Z_nS\$ statistic (if detail==TRUE)
4.	Rozas.ZA	[1]	Rozas \$ZA\$ statistic (adjacent positions, if detail==TRUE)
5.	Rozas.ZZ	[1]	Rozas \$ZZ\$ statistic (\$ZZ=ZA-Z_nS\$, if detail==TRUE)

#### References

- [1] Rozas, J., M.Gullaud, G.Blandin, and M.Aguade(2001). DNA variation at the rp49 gene region of Drosophila simulans: evolutionary inferences from an unusual haplotype structure. Genetics 158(3),1147-1155
- [2] Wall, J.(1999). Recombination and the power of statistical tests of neutrality. Genet Res 74, 65-79
- [3] Kelly, J.K. (1997). A test of neutrality based on interlocus associations. Genetics 146: 1197-

```
# GENOME.class <- readData("\home\Alignments")</pre>
# GENOME.class
# GENOME.class <- linkage.stats(GENOME.class)</pre>
# GENOME.class <- linkage.stats(GENOME.class,list(1:4,5:10),subsites="syn")</pre>
# GENOME.class <- linkage.stats(GENOME.class,list(c("seq1","seq5","seq3"),</pre>
# c("seq2", "seq8")))
# GENOME.class <- linkage.stats(GENOME.class, detail=TRUE)</pre>
# show the result:
# get.linkage(GENOME.class)
# GENOME.class@Wall.B --> population specific view
# GENOME.class@region.stats
```

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load.session

Loading a PopGenome session

# Description

This function loads a PopGenome session (more precisely: the corresponding "GENOME" object) from the current workspace.

# Usage

```
load.session(folder)
```

# Arguments

folder

name of the folder/object

#### **Details**

This function has to be used in the same workspace (folder) where the object of class "GENOME" was saved.

#### Value

An object of class "GENOME".

# **Examples**

```
# GENOME.class <- readData("...\Alignments")
# save.session(GENOME.class,folder="GENOME.class")
# q()
# R
# library(PopGenome)
# load.session("GENOME.class")</pre>
```

MKT-methods

McDonald-Kreitman Test (McDonald \& Kreitman 1991)

# Description

This generic function calculates an approximate version of the McDonald-Kreitman Test.

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#### Usage

```
## S4 method for signature 'GENOME'
MKT(object,
new.populations=FALSE,
do.fisher.test=FALSE,
fixed.threshold.fst=FALSE,
subsites=FALSE)
## S4 method for signature 'GENOME'
get.MKT(object)
```

# **Arguments**

object an object of class "GENOME"

new.populations

list of populations. default:FALSE

do.fisher.test P-value calculation out of the Dn,Ds,Pn,Ps table fixed.threshold.fst

Fixed threshold

subsites Subsites

#### **Details**

This approximate version of the McDonald-Kreitman test assumes that the probability that two single nucleotide polymorphisms (SNPs) occur in the same codon is very small. Thus, only codons with a single SNP are examined.

If no gff-file was specified when the data was read in, it is assumed that the alignment is in the correct reading frame (starting at a first codon position). The outgroup has to be defined as a population!

#### Value

The return value is a modified object of class "GENOME"

The following slots will be modified in the "GENOME" object

MKT a matrix which includes the following values:

Columns Description
1. P\_nonsyn nonsynonymous sites
2. P\_syn synonymous sites
3. D\_nonsyn fixed nonsynonymous sites

4. D\_syn fixed synonymous sites

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```
5. neutrality.index $(P_nonsyn/P_syn)/(D_nonsyn/D_syn)$6. alpha 1-neutrality.index
```

#### References

McDonald, J. H.; Kreitman, M. (1991). *Adaptive protein evolution at the Adh locus in Drosophila*. Nature 351 (6328): 652-654

# **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# GENOME.class
# GENOME.class <- MKT(GENOME.class)
# GENOME.class <- MKT(GENOME.class,list(1:7,8:12))
# show the result:
# get.MKT(GENOME.class)</pre>
```

MS

Coalescent simulation with or without selection

# **Description**

This function uses Hudson's MS and Ewing's MSMS to compare simulated data with the observed data.

# Usage

```
MS(GENO, niter=10, thetaID="user", params=FALSE, detail=FALSE, neutrality=FALSE, linkage=FALSE, F_ST=FALSE, MSMS=FALSE, big.data=FALSE)
```

# **Arguments**

GENO	an object of class "GENOME"
niter	number of samples per locus
thetaID	"Tajima","Watterson" or "user". default:"user"
neutrality	Calculate neutrality tests. default=FALSE
linkage	Calculate linkage disequilibrium. default=FALSE
F_ST	Calculate fixation index. default=FALSE
params	an object of class "test.params". see ?test.params
detail	detailed statistics. Note: slower computations! default=FALSE

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MSMS specify parameter for MSMS simulation with selection (has to be specified as a

string)

big.data if TRUE the ff-package is used

#### **Details**

You can choose different mutation rate estimators to generate simulation data. When thetaID="user", you have to define the theta values in an object of class "test.params". The "test.params" class can also be used to specify some additional parameter like migration and/or recombination rates... (?test.params).

Please read the MSMS documentation for the correct use of coalescent simulations to assess statistical significance.

#### Value

The function creates an object of class "cs.stats"

#### Note

The executable file ms has to be stored in the current workspace.

If you want to use the MSMS application, put the msms folder including the corresponding executable files in the current workspace.

Both programs can be obtained from their websites (see references).

#### References

Hudson, R. R. (2002). Generating samples under a Wright-Fisher neutral model of genetic variation. Bioinformatics 18: 337-338

Gregory Ewing and Joachim Hermisson, MSMS: A Coalescent Simulation Program Including Recombination, Demographic Structure, and Selection at a Single Locus. Bioinformatics 2010, doi: 10.1093/bioinformatics/btq322

```
# GENOME.class <- readData("...\Alignments")
# GENOME.class <- neutrality.stats(GENOME.class,list(1:6))
# MS.class <- MS(GENOME.class,thetaID="Tajima",neutrality=TRUE)
# MS.class <- MS(GENOME.class,thetaID="Tajima",neutrality=TRUE,
# MSMS="-N 1000 -SAA 200 -SaA 100 -SF 1e-2")
# MS.class
# MS.class@obs.val
# MS.class@locus[[1]]</pre>
```

MS\_getStats

Get the simulated MS/MSMS statistics

#### **Description**

This function extracts the simulated values from the class cs.stats

# Usage

```
MS_getStats(object,locus=1,population=1)
```

# Arguments

object of class "cs.stats"

locus the locus ID population the population ID

#### Value

The return value is a matrix containing the simulation results of different statistical tests. (see MS())

# **Examples**

```
# GENOME.class <- readData("...\Alignments")
# GENOME.class <- neutrality.stats(GENOME.class)
# ms <- MS(GENOME.class,thetaID="Tajima",neutrality=TRUE)
# MS_getStats(ms)</pre>
```

```
mult.linkage.stats-methods
```

Multilocus linkage statistics

# Description

This generic function calculates the linkage disequilibrium between regions.

```
## S4 method for signature 'GENOME'
mult.linkage.stats(object,lower.bound=0,upper.bound=1,pairs=FALSE)
```

# **Arguments**

object	an object of class "GENOME"
lower.bound	sites with minor-allele-frequency>=lower.bound are considered
upper.bound	sites with minor-allele-frequency<=upper.bound are considered
pairs	permutation matrix of pairwise comparisons

# **Details**

pairs is a matrix. Each column contains the pairwise comparison region IDs.

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compares region 1 with 2, and region 1 with 3.

# Value

The return value is a modified object of class "GENOME"

The following slots will be modified in the "GENOME" object

Some linkage statistics for each pair of regions (R2, P-value, Distance) mult.Linkage

The Fisher-Exact-Test is used to calculate the P-values.

```
# GENOME.class <- readData("...\Alignments")</pre>
# GENOME.class
# GENOME.class <- mult.linkage.stats(GENOME.class)</pre>
# show the result:
# GENOME.class@mult.Linkage
```

neutrality.stats-methods

Neutrality Statistics

# **Description**

This generic function calculates some neutrality statistics.

# Usage

```
## S4 method for signature 'GENOME'
neutrality.stats(object,new.populations=FALSE,new.outgroup=FALSE,
subsites=FALSE,detail=FALSE, FAST=FALSE, do.R2=FALSE)
## S4 method for signature 'GENOME'
get.neutrality(object,theta=FALSE,stats=TRUE)
```

# Arguments

_	
object	an object of class "GENOME"
new.population	S
	list of populations. default:FALSE
new.outgroup	vector of outgroup sequences. default:FALSE
subsites	"transitions": SNPs that are transitions.
	"transversions": SNPs that are transversions.
	"syn": synonymous sites.
	"nonsyn": non-synonymous sites.
	"exon": SNPs in exon regions.
	"intron": SNPs in intron regions.
	"coding": SNPs in coding regions (CDS).
	"utr": SNPs in UTR regions.
	"gene": SNPs in genes.
	default:FALSE
detail	default: FALSE, TRUE for some detailed statistics. Note: slows down calculations!
FAST	Fast computation. only works if there is no outgroup defined.
do.R2	Ramos-Onsins' & Rozas' R2
stats	show the results of each statistic. default:TRUE
theta	show the theta values. default:FALSE

# Value

The return value is a modified object of class "GENOME"

The following slots will be modified in the "GENOME" object

	Slot	Reference	Description
1.	n.segregating.sites		Total number of segregating sites
2.	Tajima.D	[1]	Tajima's' D statistic 1989
3.	Fu.Li.F	[3]	Fu & Li's' F* statistic 1993
4.	Fu.Li.D	[3]	Fu & Li's D* statistic 1993
5.	Fay.Wu.H	[6]	Fay & Wu's H statistic 2000
6.	Zeng.E	[7]	Zeng's E statistic 2006
7.	Strobeck.S	[5]	Strobeck's S statistic 1987 (if detail==TRUE)
8.	Fu.F_S	[4]	Fu's F\$_S\$ statistic 1997 (if detail==TRUE)
9.	Rozas.R_2	[2]	Ramos-Onsins' & Rozas' \$R_2\$ statistic 2002
10.	theta_Tajima	[1]	
11.	theta_Watterson		
12.	theta_Fu.Li	[3]	
13.	theta_Achaz.Watterson		
14.	theta_Achaz.Tajima		
15.	theta_Fay.Wu [6]		
16.	theta_Zeng	[7]	

# References

- [1] Tajima, F.(1989) Statistical Method for Testing the Neutral Mutation Hypothesis by DNA Polymorphism. Genetics, 123(3): 585-595.
- [2] Ramos-Onsins, S.E. and J.Rozas (2002). *Statistical Properties of New Neutrality Tests Against Population Growth*. Mol.Biol.Evol.19(12),2092-2100
- [3] Fu, Y.X. and W.H.Li (1993). Statistical Tests of Neutrality of Mutations. Genetics 133(3),693-709
- [4] Fu, Y.-X.(1997). Statistical Tests of Neutrality of mutations against population growth, hitch-hiking and background selection. Genetics 147(2),915-925.
- [5] Strobeck, C. (1987). Average number of nucleotide differences in a sample from a single sub-population: a test for population subdivision. Genetics 117, 149-153
- [6] Fay, J.C. and C.-I. Wu (2000). *Hitchhiking under positive Darwinian selection*. Genetics 155 (3),1405-1413
- [7] Zeng, K., Y.-X. Fu, S. Shi, and C.-I. Wu (2006). Statistical tests for detecting positive selection by utilizing high-frequency variants. Genetics 174, 1431-1439

## **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# GENOME.class
# GENOME.class <- neutrality.stats(GENOME.class, FAST=TRUE)
# GENOME.class <- neutrality.stats(GENOME.class,list(1:4,5:10),subsites="syn")
# GENOME.class <- neutrality.stats(GENOME.class,list(c("seq1","seq5","seq3"),
# c("seq2","seq8")))
# GENOME.class <- neutrality.stats(GENOME.class,detail=TRUE)
# show the result:
# get.neutrality(GENOME.class)
# GENOME.class@Tajima.D --> population specific view
# detail = TRUE
# GENOME.class@region.stats
```

```
PG_plot.biallelic.matrix-methods

Plot the biallelic matrix
```

# **Description**

This function plots the biallelic matrix of a specific region.

# Usage

```
## S4 method for signature 'GENOME'
PG_plot.biallelic.matrix(object,region, ind.names = FALSE , cex.axis = 0.5,
title="")
```

# **Arguments**

```
object of class "GENOME"

region the region ID

ind.names individual names/IDs. default:ALL

cex.axis size of text (y-axis)

title Title of the plot
```

```
# GENOME.class <- readData("...\Alignments")
# PG_plot.biallelic.matrix(GENOME.class, region = 1)</pre>
```

PopGenome 47

## Description

R-package for Population genetic & genomic analyses

#### **Details**

Index:

F\_ST.stats Fixation index diversity.stats Diversities MKT McDonald & Kreitman test

MS Coalescent simulations detail.stats Several misc. statistics linkage.stats Linkage disequilibrium neutrality.stats Neutrality statistics

readData Reading alignments and calculating summary data

readSNP Read data in .SNP format (e.g., from the 1001 Arabidopsis Genomes project)

readVCF Read data in VCF format (e.g., from the 1000 human Genomes project)

readHapMap Read data in HapMap format sliding.window.transform Sliding window transformation splitting.data Split data into subsites

test.params Set parameters for coalescent simulations.

#### Author(s)

Bastian Pfeifer Maintainer: Bastian Pfeifer <Bastian.Pfeifer@uni-duesseldorf.de>

#### See Also

?readData readData

```
# GENOME.class <- readData("...\Alignments")
# GENOME.class <- neutrality.stats(GENOME.class)
# values <- get.neutrality(GENOME.class)
# GENOME.class <- F_ST.stats(GENOME.class,list(1:5,6:10))
# values <- get.F_ST(GENOME.class)</pre>
```

48 read.big.fasta

PopGplot	Smoothed line-plot for multiple populations	

# Description

This function plots values with smoothed lines using spline interpolation.

# Usage

```
PopGplot(values,colors=FALSE,span=0.1,ylab="",xlab="",
ylim=c(min(values,na.rm=TRUE),max(values,na.rm=TRUE)))
```

# **Arguments**

values	the statistical values (matrix); columns=populations
colors	the colors for each population (character vector)
span	the degree of smoothing
ylab	a title for the y axis
xlab	a title for the x axis
ylim	ranges for the y axis

# **Examples**

```
# GENOME.class <- readSNP("Arabidopsis",CHR=1)
# GENOME.class.slide <- sliding.window.transform(test,1000,1000)
# GENOME.class.slide <- diversity.stats(GENOME.class.slide)
# values <- GENOME.class.slide@nuc.divesity.within
# PopGplot(values)</pre>
```

read.big.fasta

Reading large FASTA alignments

# Description

This function splits FASTA alignments that are too large to fit into the computer memory into chunks.

read.big.fasta 49

#### Usage

```
read.big.fasta(filename,populations=FALSE,outgroup=FALSE,window=2000,
               SNP.DATA=FALSE, include.unknown=FALSE,
               parallized=FALSE,FAST=FALSE,big.data=TRUE)
```

# **Arguments**

filename the basepath of the FASTA alignment

outgroup vector of outgroup sequences

list of populations populations

window chunk size: number of columns/nucleotide sites

SNP.DATA should be switched to TRUE if you use SNP data in alignment format

include.unknown

include unknown positions in the biallelic.matrix

parallized Use parallel computations to speed up the reading - works only on UNIX sys-

**FAST** Fast computation. see readData()

big.data use the ff-package

#### **Details**

The algorithm reads the data for each individual and stores the information on disk. The data can be analyzed as regions of the defined window size, or can be concatenated in the PopGenome framework via the function concatenate.regions. This function should only be used when the FASTA file does not fit into the RAM; else, use the function readData.

#### Value

The function creates an object of class "GENOME"

The following slots will be filled in the "GENOME" object

Slot Description

total number of sites 1. n.sites n.biallelic.sites number of biallelic sites

3. region.names names of regions

4. region.data some detailed information about the data 50 readData

#### **Examples**

```
# GENOME.class <- read.big.fasta("Alignment.fas", big.data=TRUE)
# GENOME.class
# GENOME.class@region.names
# CON <- concatenate.regions(GENOME.class)
# CON@region.data@biallelic.sites
# GENOME.class.slide <- sliding.window.transform(GENOME.class,100,100)
# GENOME.class <- neutrality.stats(GENOME.class,FAST=TRUE)
# show the result:
# get.sum.data(GENOME.class)
# GENOME.class@region.data</pre>
```

readData

Read alignments and calculate summary data

## Description

This function reads alignments/SNP data in several formats and calculates some summary data.

#### Usage

# **Arguments**

object of class "GENOME"

path the basepath (folder) of the alignments

outgroup vector of outgroup sequences

include.unknown

if positions with unknown nucleotides should be considered.

populations list of populations. default:FALSE

gffpath the basepath (folder) of the corresponding GFF-files. default:FALSE

format data formats. "fasta" is default. See details!

parallized parallel processing to accelerate the reading process. See details!

readData 51

progress\_bar\_switch

progress\_bar

FAST fast computation. See details!

big.data use the ff-package

SNP. DATA important for reference positions; should be TRUE if you use SNP-data in align-

ment format

#### **Details**

All data (alignments or SNP-files) have to be stored in one folder. The folder is the input of this function. If no GFF file (which also have to be stored in a folder) is specified, an alignment in the correct reading frame (starting at a first codon position) is expected.

Otherwise synonymous and non-synonymous positions are not identified correctly.

#### Note:

The GFF-files have to be EXACTLY the same names (without any extensions like .fas or .gff) as the files storing the nucleotide data to ensure correct matching

#### format:

```
"fasta","nexus","phylip",
"MAF","MEGA"
"HapMap","VCF"
"RData"
```

Valid nucleotides are T,t,U,u,G,g,A,a,C,c,N,n,-

#### parallized:

- will speed up calculations if you use a very large amount of alignments

#### FAST:

- will not classify synonymous/non-synonymous SNPs directly
- fast computation (via compiled C code) of biallelic matrix, biallelic sites, transversions/transitions and biallelic substitutions
- can be switched to TRUE in case of SNP data without loss of information

## big.data:

- use the ff-package
- ff mechanism is used for biallelic.matrix and GFF/GTF information
- is automatically activated for readVCF or readSNP
- Note! you should set this to TRUE if you use big chunks of data and you want to later concatenate them in the PopGenome framework (for example: sliding windows of the whole dataset).

52 readHapMap

## SNP.DATA:

- should be switched to TRUE if you use SNP-data in alignment format.
- the corresponding SNP positions can be set via set.ref.positions

#### Value

The function creates an object of class "GENOME"

The following slots will be filled in the "GENOME" object

	Slot	Description
1.	n.sites	total number of sites
2.	n.biallelic.sites	number of biallelic sites
3.	n.gaps	number of sites with gaps
4.	n.unknowns	number of sites with unknown nucleotides
5.	n.valid.sites	number of valid sites
6.	n.polyallelic.sites	number of sites with >2 nucleotides
7.	trans.transv.ratio	transition/transversion ratio of biallelic sites
8.	region.names	names of regions
9.	region.data	some detailed information about the data read

# **Examples**

```
# GENOME.class <- readData("...\Alignments", FAST=TRUE)</pre>
# GENOME.class <- readData("VCF", format="VCF")</pre>
# Note, "Alignments" and "VCF" are folders!
# GENOME.class@region.names
# GENOME.class <- readData("...\Alignments", big.data=TRUE)</pre>
# object.size(GENOME.class)
# GENOME.class <- readData("...\Alignments",gffpath="...\Alignments_GFF")</pre>
# GENOME.class
# show the result:
# get.sum.data(GENOME.class)
# GENOME.class@region.data
```

readHapMap

Read SNP data from the HapMap consortium

# Description

This function reads HapMap data.

readHapMap 53

#### Usage

readHapMap(folder,hap\_gffpath,populations=FALSE,outgroup=FALSE)

#### Arguments

folder the basepath of the variant\_calls

hap\_gffpath the basepath of the corresponding GFF files. Note! The HapMap GFF file does

not contain information about subsites. see details!

populations list of populations

outgroup vector of outgroup sequences

#### **Details**

PopGenome reads the GFF file distributed on the HapMap plattform only to verify the reference positions of the chromosomes. In the next release, this function will also handle GFF/GTF files to get information about subsites (exons, introns, ...). The input folder should include the files of different individuals for one chromosome. This facilitates FST calculations of the HapMap data. See also readData("...", format="HapMap") which can read the files of single populations directly.

#### Value

The function creates an object of class "GENOME"

The following slots will be filled in the "GENOME" object

Slot Description

1. n.sites total number of sites 2. n.biallelic.sites number of biallelic sites

region.data some detailed information about the data read

# **Examples**

```
# GENOME.class <- readHapMap("...\HapMapData")</pre>
```

# GENOME.class

# show the result:

# get.sum.data(GENOME.class)

# GENOME.class@region.data

54 readMS

readMS

Read output data from MS and MSMS

# Description

This function reads data produced from the coalescent simulation programs MS (Hudson, 2002) and MSMS (Greg, 2010)

# Usage

```
readMS(file,big.data=FALSE)
```

# Arguments

file the basepath of the MS/MSMS output

big.data The ff package is used

# Value

An object of class GENOME

## References

Hudson, R. R. (2002). *Generating samples under a Wright-Fisher neutral model of genetic variation*. Bioinformatics 18: 337-338

Gregory Ewing and Joachim Hermisson, MSMS: A Coalescent Simulation Program Including Recombination, Demographic Structure, and Selection at a Single Locus. Bioinformatics 2010, doi: 10.1093/bioinformatics/btq322

# **Examples**

```
# GENOME.class <- readMS("ms.output.txt")</pre>
```

# GENOME.class@region.names

readSNP 55

readSNP	Read data in .SNP format	
---------	--------------------------	--

# **Description**

This function reads data in .SNP (quality\_variant) format, as distributed by the 1001 Genomes project (Arabidopsis).

# Usage

```
readSNP(folder,populations=FALSE,outgroup=FALSE,gffpath=FALSE,
CHR=FALSE,ref.chr=FALSE,snp.window.size=FALSE,
parallized=FALSE,ffpackagebool=TRUE,
include.unknown=FALSE
)
```

## **Arguments**

folder the basepath of the variant\_calls outgroup vector of outgroup sequences populations list of populations gffpath the corresponding GFF file CHR which chromosome?, default: all chromosomes ref.chr reference chromosome (to classify synonymous/non-synonymous positions) snp.window.size scan SNP chunks parallized multicore computation ffpackagebool use the ff-package to save memory space. (slower) include.unknown include positions with unknown nucleotides

## **Details**

The ff-package we use to store the SNP information limits the data size to individuals \* (number of SNPs) <= .Machine\$integer.max

The text files containing the SNP information of each individual have to be stored in one folder. The slots transitions, biallelic.sites, and biallelic.substitutions of the class "regions.data" will be filled.

At this time, if a GFF/GTF is used the data should be organized in a way that

the "CHR" is a numerical value. The prefix "Chr" or "chr" is also supported.

56 readVCF

#### Value

The function creates an object of class "GENOME"

Following Slots will be filled in the "GENOME" object

Slot Description total number of sites 1. n.sites 2. n.biallelic.sites number of biallelic sites some detailed information about the data read 3. region.data 4. region.names names of regions

### **Examples**

```
# GENOME.class <- readSNP("...\SNPData")</pre>
# GENOME.class <- readSNP("...\SNPData", CHR=1)</pre>
# GENOME.class <- readSNP("...\SNPData", CHR=1, gffpath="Gff_file.gff")
# GENOME.class
# GENOME.class <- neutrality.stats(GENOME.class,FAST=TRUE)</pre>
# show the result:
# get.sum.data(GENOME.class)
# GENOME.class@region.data
```

readVCF

Read SNP data in tabixed VCF format

# **Description**

This function reads tabixed VCF-files, as distributed from the 1000 Genomes project (human).

#### Usage

```
readVCF(filename, numcols, tid, frompos, topos,
        samplenames=NA, gffpath = FALSE, include.unknown=FALSE, approx=FALSE,
out="", parallel=FALSE)
```

# **Arguments**

the corresponding tabixed VCF-file filename

numcols number of SNPs that should be read in as a chunk readVCF 57

tid which chromosome ? (character)

frompos start of the region
topos end of the region
samplenames a vector of individuals
gffpath the corresponding GFF file

include.unknown

includ positions with unknown/missing nucleotides

approx see details!

out a folder suffix where the temporary files should be saved

parallel parallel computation using mclapply

#### **Details**

The readVCF function expects a tabixed VCF file with a diploid GT field. In case of haploid data, the GT field has to be transformed to a pseudo-diploid field (such as 0 -> 0|0). An alternative is to use readData(..., format="VCF"), which can read non-tabixed haploid and any kind of polyploid VCFs directly. When approx=TRUE, the algorithm will apply a logical OR to the GT-field: (0|0=0,1|0=1,0|1=1,1|1=1). Note, this is an approximation for diploid data, which will speed up calculations. In case of haploid data, approx should be switched to TRUE. If approx=FALSE, the full diploid information will be considered. The ff-package PopGenome uses to store the SNP information limits total data size to individuals \* (number of SNPs) <= .Machine\$integer.max In case of very large data sets, the bigmemory package will be used; this will slow down calculations (e.g. this package have to be installed first !!!). Use the function vcf\_handle <-.Call("VCF\_open", filename) to open a VCF-file and .Call("VCF\_getSampleNames",vcf\_handle) to get and define the individuals which should be considered in the analysis. See also readData(..., format="VCF")!

## Value

The function creates an object of class "GENOME"

The following slots will be filled in the "GENOME" object

The following slots will be filled in the GENORE Obje

Slot Description
1. n.sites total number of sites

1. n.sites total number of sites
2. n.biallelic.sites number of biallelic sites

3. region.data some detailed information about the data read

4. region.names names of regions

58 recomb.stats-methods

#### **Examples**

```
# GENOME.class <- readVCF("...\chr1.vcf.gz", 1000, "1", 1, 100000)
# GENOME.class
# GENOME.class@region.names
# GENOME.class <- neutrality.stats(GENOME.class,FAST=TRUE)
# show the result:
# get.sum.data(GENOME.class)
# GENOME.class@region.data</pre>
```

recomb.stats-methods Recombination statistics

# **Description**

This generic function calculates the Four-Gamete test (Hudson 1985).

## Usage

```
## S4 method for signature 'GENOME'
recomb.stats(object,new.populations=FALSE,subsites=FALSE)
## S4 method for signature 'GENOME'
get.recomb(object)
```

# **Arguments**

```
object an object of class "GENOME"

new.populations
list of populations. default:FALSE

subsites "transitions": SNPs that are transitions.
    "transversions": SNPs that are transversions.
    "syn": synonymous sites.
    "nonsyn": nonsynonymous sites.
    "exon": SNPs in exon regions.
    "intron": SNPs in intron regions.
    "coding": SNPs in coding regions (CDS).
    "utr": SNPs in UTR regions.
    "gene": SNPs in genes.
    default:FALSE
```

region.as.fasta-methods 59

# Value

```
The return value is a modified object of class "GENOME"

The following slots will be modified in the "GENOME" object
```

Hudson.RM Four-gamete test

## References

Hudson, R. K. (1985). Statistical Properties of the Number of Recombination Events in the History of a Sample of DNA Sequences Genetics 111 (1): 147-164.

# **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# GENOME.class
# GENOME.class <- recomb.stats(GENOME.class)
# GENOME.class <- recomb.stats(GENOME.class,list(1:7,8:12))
# show the result:
# recomb.values <- get.recomb(GENOME.class)
# recomb.values[[1]] # first population !
# GENOME.class@region.stats@Hudson.RM</pre>
```

```
region.as.fasta-methods
```

Extract a region and write it to a FASTA file

# **Description**

This generic function writes a FASTA file of the observed biallelic positions to the current workspace.

```
## S4 method for signature 'GENOME'
region.as.fasta(object,region.id=FALSE,filename=FALSE,type=1,ref.chr=FALSE)
```

60 save.session

# **Arguments**

object an object of class "GENOME"

region.id region of the genome filename name of the FASTA file

type 1: extract SNPs; 2: extract all nucleotides

ref.chr reference sequence

#### **Details**

In case of type=2 we recommend to use the function splitting.data(,positions=list( ... ), type=2) before and apply the region.as.fasta() to this splitted object afterwards. The type=1 method will write a FASTA file including only the biallelic.sites.

region. id is the the region number specified in the PopGenome class GENOME.

### **Examples**

```
#GENOME.class <- readSNP("Arabidopsis",CHR=1)
# split the data into the genmic positions 100 to 2000
#GENOME.class.split <- splitting.data(GENOME.class, positions=list(100:2000),type=2)
#GENOME.class.split@region.names
#region.as.fasta(GENOME.class.split,1,"my_fasta_file.fas",type=2, ref.chr="chrom1.fas")</pre>
```

save.session

Save the "GENOME" object of a PopGenome session

# Description

This function saves the "GENOME" object of a PopGenome session to the current workspace. The object can be loaded again with load.session().

#### Usage

```
save.session(object,folder)
```

# Arguments

object of class "GENOME" folder name of the folder/object

## **Details**

Saving R and ff-objects created by the ff-package in a folder.

set.filter-methods 61

## **Examples**

```
# GENOME.class <- readData("...\Alignments")
# save.session(GENOME.class,"GENOME.class")
# load.session("GENOME.class")</pre>
```

set.filter-methods

*Setting filter to the analysis* 

# Description

A generic function to set filter regarding e.g missing data.

# Usage

```
## S4 method for signature 'GENOME'
set.filter(object,
missing.freqs=TRUE,
minor.freqs=FALSE,
maf.lower.bound=0,
maf.upper.bound=1,
miss.lower.bound=0,
miss.upper.bound=1)
```

#### **Arguments**

```
object An object of class "GENOME"
missing.freqs Set filter for missing data
minor.freqs Set filter for the MAF
maf.lower.bound
frequency of the MAF
maf.upper.bound
...
miss.lower.bound
frequency of the missing freq.
miss.upper.bound
```

# **Details**

This function sets the slot region.data@included.

62 set.outgroup-methods

# Value

Returned value is a modified object of class "GENOME"

The following slots will be modified in the "GENOME" object

\_\_\_\_\_

Slot Reference Description 1. region.data@included [x] ...

#### References

[x]

# **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# GENOME.class <- set.filter(GENOME.class, missing.freqs=TRUE,
# miss.lower.bound=0, miss.upper.bound=0.2)
# now apply any statistic to the filtered data set.
# GENOME.class <- calc.fixed.shared(GENOME.class, subsites="included")</pre>
```

# **Description**

This generic function defines the outgroup by matching the specified vector against each region.

# Usage

```
## S4 method for signature 'GENOME'
set.outgroup(object,new.outgroup=FALSE, diploid=FALSE)
```

# **Arguments**

object an object of class "GENOME"

new.outgroup a vector of outgroup individuals
diploid if diploid data is present

set.populations-methods

#### **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# outgroup <- c("seq1","seq2")
# GENOME.class <- set.outgroup(GENOME.class,new.outgroup=outgroup)
# GENOME.class <- neutrality.stats(GENOME.class)</pre>
```

```
set.populations-methods
```

Define populations

# **Description**

This generic function defines the populations.

Using this function obviates the need to specify the populations for each calculation separately. The populations can be set differently for different PopGenome modules by applying the function between module calls.

# Usage

```
## S4 method for signature 'GENOME'
set.populations(object,new.populations=FALSE, diploid=FALSE,
triploid=FALSE,tetraploid=FALSE)
```

# **Arguments**

```
object an object of class "GENOME"

new.populations
list of populations. default:FALSE

diploid if diploid data is present

triploid if triploid data is present

tetraploid if tetraploid data is present
```

```
# GENOME.class <- readData("\home\Alignments")
# pop.1 <- c("seq1","seq2")
# pop.2 <- c("seq3","seq4","seq1")
# GENOME.class <- set.populations(GENOME.class,list(pop.1,pop.2))
# GENOME.class@region.data@populations2
# GENOME.class <- neutrality.stats(GENOME.class)</pre>
```

```
set.ref.positions-methods

Set reference positions for SNP data
```

# Description

This generic function sets the positions of the SNP data. Should be used if you use alignment formats to store SNP data (i.e., data restricted to the polymorphic positions).

# Usage

```
## S4 method for signature 'GENOME'
set.ref.positions(object, positions)
```

# Arguments

object an object of class "GENOME" positions a list of reference positions

## Value

returned value is a modified object of class "GENOME"

# **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# GENOME.class
# GENOME.class <- set.ref.positions(GENOME.class,list(c(1000,2001,3000),
# c(3200,12000)))</pre>
```

set.synnonsyn-methods Set synonymous positions for SNP data

# Description

This generic function classifies the observed biallelic positions read from SNP data files into synonymous and non-synonymous SNPs.

show.slots-methods 65

## Usage

```
## S4 method for signature 'GENOME'
set.synnonsyn(object,ref.chr,save.codons=FALSE)
```

# **Arguments**

object an object of class "GENOME"

ref.chr the reference chromosome in FASTA format

save.codons save codon changes

#### Value

The return value is a modified object of class "GENOME" storing syn/nonsyn informations in the slot GENOME.class@region.data@synonymous for each SNP. (1=synonymous,0=non-synonymous)
When save.codons is TRUE the SNP related codon changes are saved in the corresponding slot GENOME.class@region.data@codons. (see also get.codons(), codontable() and codonise64())

#### Note

The data has to be read in with a corresponding GFF/GTF file (CDS fields must be specified); otherwise a correct classification is not possible. The set.synnonsyn() function does not work for splitted objects e.g produced via sliding.window.transform() or splitting.data(). Note, transcripts which are in the same CDS region but have different reading frames are are not specified correctly. PopGenome can also handle coding regions on reverse strands. We have used the program SNPeff to validate our results.

## **Examples**

```
# GENOME.class <- readData("VCF",format="VCF",gffpath="GFF.Folder")
# GENOME.class <- set.synnonsyn(GENOME.class,ref.chr="ref.fas")
# GENOME.class@region.data@synonymous</pre>
```

show.slots-methods

Show Slots of class GENOME

# Description

```
coming soon ...
```

#### Methods

```
object = "GENOME" coming soon ...
```

#### **Examples**

# show.slots(GENOME.class)

```
sliding.window.transform-methods
```

Sliding Window Transformation

# Description

This generic function transforms an existing object of class "GENOME" into another object of class "GENOME", in which each region corresponds to one window. This allows to apply the full spectrum of PopGenome methods to sliding window data.

# Usage

```
## S4 method for signature 'GENOME'
sliding.window.transform(object,
width=7, jump=5,
type=1,
start.pos=FALSE,end.pos=FALSE,
whole.data=TRUE
)
```

#### **Arguments**

object an object of class "GENOME" width window size. default:7 jump jump size. default:5

type 1 scan only biallelic positions (SNPs), 2 scan the genome. default:1

start.pos start position end.pos end position

whole.data scan the complete data by concatenating the regions in "object". If FALSE, each

region is scanned seperately.

#### Value

The function creates a transformed object of class "GENOME".

#### Note

If you want to scan regions seperately (whole.data=FALSE), you may not use the big.data option in the readData function. PopGenome will scan the data from position 1 to the last observed SNP if start or end-positions are not specified.

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#### **Examples**

snp\_file

.SNP file (variant call data from 1001 Arabidopsis Genomes project)

# Description

A .SNP file stored in the directory "data" of the PopGenome package. The file contains variant calls for exactly one individual. Put all files (individuals of interest) into one folder (for example "SNP"). readSNP("SNP", CHR=1)

```
splitting.data-methods
```

Split data into subsites

# Description

This generic function splits the data into subsites, if GFF/GTF information is present or if positions are defined accordingly.

#### **Arguments**

object an object of class "GENOME"

positions list of positions

subsites "exon": SNPs in exon regions.

"intron": SNPs in intron regions.

"coding": SNPs in coding regions (CDS).

"utr": SNPs in UTR regions. "gene": SNPs in genes.

type 1: SNP positions

2: Genome positions

whole.data Scan the whole data by concatenating the regions. If FALSE, the regions are

scanned seperately

#### **Details**

Note, if whole.data=FALSE data with n.biallelic.sites==0 should be removed.

#### Value

The return value is a modified object of class "GENOME".

# **Examples**

```
# GENOME.class <- readData("\home\Alignments")
# GENOME.class
# GENOME.class.split <- splitting.data(GENOME.class,subsites="exon")
# GENOME.class.split@region.names
# GENOME.class.split <- splitting.data(GENOME.class,positions=list(1:7,8:12))
# GENOME.class.split <- splitting.data(GENOME.class,
# positions=list(2000:3000,12000:13000),type=2)
# GENOME.class.split</pre>
```

```
{\tt split\_data\_into\_GFF\_attributes}
```

Split the data into GFF attributes

## **Description**

Splits the data into GFF attributes defined by the user.

```
split_data_into_GFF_attributes(object, gff.file, chr, attribute)
```

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## **Arguments**

object	An object of class GENOME
gff.file	The corresponding GFF file
chr	The chromosome/scaffold identifier

attribute The attribute to use for splitting

#### **Details**

```
The algorithm splits the data into attributes. An attribute can be "gene_name", "Parent" or just a single gene name like "geneXYZ".
```

#### Value

```
The returned value is an object of class "GENOME" See GENOME.class.split@region.names and GENOME.class.split@region.names after splitting the data.
```

# **Examples**

```
# GENOME.class <- readVCF("chr1.vcf.gz",1000,"1",1,100000)
# GENOME.class.split <- split_data_into_GFF_attributes(GENOME.class,"Homo_sapiens.GRCh37.73.gtf",
# "1", "gene_name")
# GENOME.class.split@region.names
# GENOME.class.split@feature.names</pre>
```

```
sweeps.stats-methods Selective Sweeps
```

# Description

This module calculates some statistics to detect selective sweeps.

```
## S4 method for signature 'GENOME'
sweeps.stats(object,new.populations=FALSE,subsites=FALSE,
freq.table=FALSE, FST=FALSE)
## S4 method for signature 'GENOME'
get.sweeps(object)
```

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## **Arguments**

object an object of class "GENOME"

new.populations

list of populations. default:FALSE

subsites "transitions": SNPs that are transitions.

"transversions": SNPs that are transversions.

"syn": synonymous sites.

"nonsyn": non-synonymous sites.
"exon": SNPs in exon regions.
"intron": SNPs in intron regions.

"coding": SNPs in coding regions (CDS).

"utr": SNPs in UTR regions. "gene": SNPs in genes.

default:FALSE

freq.table the frequency counts for the CLR test. "list"

FST use FST values instead of the minor allele frequencies

#### **Details**

The freq.table contains the global sets of frequency counts. It can be produced with the module detail.stats. The values in the slot GENOME.class@region.stats@minor.allele.frequencies can be used to create this global set. (use the R function table) freq.table is a list of length n.pops.

#### Value

The return value is a modified object of class "GENOME"

The following slots will be modified in the "GENOME" object

\_\_\_\_\_

CL Composite Likelihood of SNPs

CLR Nielsen's CLR test

#### References

Cai JJ (2008) *PGEToolbox: A Matlab toolbox for population genetics and evolution* Journal of Heredity Jul-Aug;99(4):438-40.doi:10.1093/jhered/esm127

Nielson, R. (2005). Genomic scans for selective sweeps using SNP data Genome Res. 2005 15: 1566-1575

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#### **Examples**

```
# Reading one alignment stored in the folder Aln
# GENOME.class <- readData("\home\Aln")</pre>
# CL
# GENOME.class <- sweeps.stats(GENOME.class)</pre>
# GENOME.class@CL
# CLR
# create global set
# GENOME.class <- detail.stats(GENOME.class)</pre>
# freq <- GENOME.class@region.stats@minor.allele.freqs[[1]]</pre>
# freq.table <- list()</pre>
# freq.table[[1]] <- table(freq)</pre>
# define the region of interest
# GENOME.class.split <- splitting.data(GENOME.class, positions= ...)</pre>
# calculate CLR
# GENOME.class.split <- sweeps.stats(GENOME.class.split, freq.table=freq.table)</pre>
# GENOME.class@CLR
```

test.params-class

Set parameters for coalescent simulations with Hudson's MS and Ewing's MSMS.

# **Description**

The object that contains the set parameter values can be passed to the function MS. This class simplifies the process of passing on all necessary values to the MS function.

# Arguments

theta	mutation parameter theta (4Nmu), where N is the diplod effective population size and mu the mutation rate per locus. It needs to be provided as a vector of length n.regions
seeds	specify 3 random number seeds. a vector of length 3 with positive integer values is expected
fixedSegsites	usually the number of segregating sites varies in each iteration. Please provide a single numeric value if the number of segregating sites needs to be fixed.
recombination	provide a vector of format: $c(p, nsites)$ , $p = cross$ -over parameter rate, nsites is the number of sites between which recombination occurs
geneConv	in addition to recombination, intra-locus non-cross-over exchange gene conversion can be included in the simulation; the expected format is c(f, gamma), where f denotes the ratio g/r (r is the probability per generation of crossing-over between adjacent sites (see Wiuf and Hein 2000), and gamma is the mean conversion tract length.

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growth

population size is assumed to be  $N(t) = N0 \exp^a h^*t$ . Provide alpha as an integer value. Negative values indicate that population was larger in the past than present, positive values indicate that it was smaller.

migration

specify the migration rate between populations. Please provide a single numeric value.

demography

vector of length 3 or 4 with first value denoted as 'type' valid 'types' for vectors of length 3 are as follows:
- 1 set a growth rate change alpha at a certain time t: c(1, t, alpha)

- 2 set all sub-populations to size  $x N_0$  and growth rate to zero: c(2, t, x)
- 3 set all elements of the migration matrix to x/(npop-1): c(3, t, x)

valid 'types' for vectors of length 4 are as follows:
- 4 set growth rate of sub-population i to alpha at time z: c(4, t, i, alpha)

- 5 set sub-population i size to  $x * N_0$  at time t and growth rate to zero: c(5, t, i, x)
- 6 split sub-population i into sub-population i and a new sub-population, labeled npop + 1. Each ancestral lineage in sub-population i is randomly assigned to sub-population i with probability p and sub-population npop + 1 with probability 1 p. The size of sub-population npop + 1 is set to \$N\_0\$. Migration rates to and from the new sub-population are assumed to be zero and the growth rate of the new sub-population is set to zero: c(6, t, i, p)
- 7 move all lineages in sub-population i to sub-population j at time t. Migration rates from sub-population i are set to zero: c(7,t,i,j)

#### Author(s)

Bastian Pfeifer

#### See Also

MS

```
# params
# params@theta <- new("test.params")
# params@theta <- rep(5,n.regions)</pre>
```

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```
# params@migration <- 3</pre>
```

vcf\_file

VCF file (subdirectory "data")

#### **Description**

A VCF file stored in the directory "data" of the PopGenome package. The file (unpacked) has to be stored in a folder (for example "VCF"). Note that many VCF-files can be stored in this folder and are read consecutively. If the VCF file is too large to fit into the computer's main memory, split it into chunks (by position)! PopGenome is able to concatenate these chunks afterwards. readData("VCF", format="VCF", FAST=TRUE)

```
VCF_split_into_scaffolds
```

Split a VCF file into multiple scaffold-VCFs

## **Description**

This function splits a VCF file into multiple VCFs including data for exactly one scaffold each.

#### **Usage**

```
VCF_split_into_scaffolds(VCF.file, output.folder)
```

#### **Arguments**

```
VCF.file the basepath of the VCF file output.folder name of the folder where the VCFs should be stored
```

# **Details**

The algorithm splits the VCF into multiple scaffold based VCFs and stores the files in a given folder. This folder can be used as an input for readData(,format="VCF")

#### Value

**TRUE** 

```
# VCF_split_into_scaffolds("VCFfile.vcf","scaffoldVCFs")
# test <- readData("scaffoldVCFs", format="VCF")</pre>
```

74 Whop\_readVCF

Whop_readVCF	Reading tabixed VCF files (an interface to WhopGenome)

# **Description**

This function provides an interface to the WhopGenome package which is specialized to read tabix-indexed VCF files.

## Usage

# Arguments

v a vcf\_handle returned from vcf\_open()

numcols number of SNPs that should be read in as one chunk

tid which chromosome ? (character)

frompos start of the region topos end of the region

samplenames a vector of individual names/IDs gffpath the corresponding GFF file

include.unknown

including positions with unknown nucleotides

## **Details**

WhopGenome is required! require(WhopGenome) WhopGenome provides some powerful filter meachanisms which can be applied to the VCF reading process. The filter rules can be set via WhopGenome functions. Whop\_readVCF expects a vcf\_handle returned from vcf\_open. The Whop\_readVCF function expects a tabixed VCF with a diploid GT-field.

In case of haploid data, the GT-field has to be transformed to a pseudo- diploid field (0 -> 010 etc.). An alternative is to use readData(..., format="VCFhap") which can read non-tabixed haploid VCFs directly.

The ff-package we use limits the data size to individuals \* (number of SNPs) <= .Machine\$integer.max In case of very large data sets, the bigmemory package will be used.

This may slow down calculations.

See also readData(..., format="VCF")!

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# Value

The function creates an object of class "GENOME"

The following slots will be filled in the "GENOME" object

Description Slot n.sites total number of sites 2. n.biallelic.sites number of biallelic sites region.data some detailed information on the data read 4. region.names names of regions

```
# require(WhopGenome)
# vcf_handle <- vcf_open("chr2.vcf.gz")</pre>
# GENOME.class <- Whop_readVCF(vcf_handle, 1000, "2", 1, 100000)</pre>
# GENOME.class
# GENOME.class@region.names
```

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