## **BSgenome**

April 20, 2011

available.genomes Find available/installed genomes

#### **Description**

available.genomes gets the list of BSgenome data packages that are currently available on the Bioconductor repositories for your version of R/Bioconductor. installed.genomes gets the list of BSgenome data packages that are already installed on your machine.

#### Usage

```
available.genomes(type=getOption("pkgType"))
installed.genomes()
```

## Arguments

type Character string indicating the type of package ("source", "mac.binary" or "win.binary") to look for.

## **Details**

A BSgenome data package contains the full genome for a given organism. Its name has 4 parts separated by a dot (e.g. BSgenome.Celegans.UCSC.ce2). The 1st part is always BSgenome, the 2nd part is the name of the organism (abbreviated), the 3rd part is the name of the organisation who assembled the genome and the 4th part is the release string or number used by this organisation for this genome. A BSgenome data package contains a single top-level object (a BSgenome object) named like the second part of the package name (e.g. Celegans in the case of BSgenome.Celegans.UCSC.ce2) where all the sequences for this genome are stored.

#### Value

A character vector containing the names of the BSgenome data packages that are currently available (for available.genomes), or already installed (for installed.genomes).

#### Author(s)

H. Pages

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#### See Also

BSgenome-class, available.packages

## **Examples**

```
# What genomes are already installed:
installed.genomes()

# What genomes are available:
available.genomes()

# Make your choice and install with:
source("http://bioconductor.org/biocLite.R")
biocLite("BSgenome.Scerevisiae.UCSC.sacCerl")

# Have a coffee ;-)

# Load the package and display the index of sequences for this genome:
library(BSgenome.Scerevisiae.UCSC.sacCerl)
Scerevisiae
```

bsapply

bsapply

## **Description**

Apply a function to each chromosome in a genome.

## Usage

```
bsapply(BSParams, ...)
```

#### **Arguments**

a BSParams object that holds the various parameters needed to configure the bsapply function

optional arguments to 'FUN'.

#### **Details**

By default the exclude parameter is set to not exclude anything. A popular option will probably be to set this to "rand" so that random bits of unassigned contigs are filtered out.

## Value

If BSParams sets simplify = FALSE, a GenomeData object is returned containing the results generated using the remaining BSParams specifications. If BSParams sets simplify = TRUE, an sapply-like simplification is used on the results.

#### Author(s)

Marc Carlson

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#### See Also

BSParams-class, BSgenome-class, BSgenome-utils, GenomeData-class

```
## Load the Worm genome:
library("BSgenome.Celegans.UCSC.ce2")
\#\# Count the alphabet frequencies for every chromosome but exclude
## mitochrondrial ones:
params <- new("BSParams", X = Celegans, FUN = alphabetFrequency,
exclude = "M")
bsapply(params)
## Or we can do this same function with simplify = TRUE:
params <- new("BSParams", X = Celegans, FUN = alphabetFrequency,</pre>
exclude = "M", simplify = TRUE)
bsapply(params)
## Examples to show how we might look for a string (in this case an
## ebox motif) across the whole genome.
Ebox <- DNAStringSet("CACGTG")</pre>
pdict0 <- PDict(Ebox)</pre>
params <- new("BSParams", X = Celegans, FUN = countPDict, simplify = TRUE)
bsapply(params, pdict = pdict0)
params@FUN <- matchPDict
bsapply(params, pdict = pdict0)
## And since its really overkill to use matchPDict to find a single pattern:
params@FUN <- matchPattern</pre>
bsapply(params, pattern = "CACGTG")
## Examples on how to use the masks
library("BSgenome.Hsapiens.UCSC.hg19")
\#\# I can make things verbose if I want to see the chromosomes getting processed.
options(verbose=TRUE)
## For the 1st example, lets use default masks
params <- new("BSParams", X = Hsapiens, FUN = alphabetFrequency,
exclude = c(1:8, "M", "X", "random", "hap"), simplify = TRUE)
bsapply(params)
if (interactive()) {
  ## Set up the motifList to filter out all double T's and all double C's
  params@motifList <-c("TT","CC")</pre>
  bsapply(params)
  ## Get rid of the motifList
  params@motifList=as.character()
##Enable all standard masks
params@maskList <- c("RM"=TRUE, "TRF"=TRUE)</pre>
```

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```
bsapply(params)
##Disable all standard masks
params@maskList <- c("AGAPS"=FALSE,"AMB"=FALSE)
bsapply(params)</pre>
```

BSgenome-class

BSgenome objects

#### **Description**

The BSgenome class is a container for the complete genome sequence of a given organism.

#### Accessor methods

In the code snippets below, x is a BSgenome object and name is the name of a sequence (characterstring). Note that, because the BSgenome class contains the GenomeDescription class, then all the accessor methods for GenomeDescription objects can also be used on x.

- sourceUrl(x) Return the source URL i.e. the permanent URL to the place where the FASTA files used to produce the sequences contained in x can be found (and downloaded).
- segnames (x) Return the index of the single sequences contained in x. Each single sequence is stored in a DNAString or MaskedDNAString object and typically comes from a source file (FASTA) with a single record. The names returned by segnames (x) usually reflect the names of those source files but a common prefix or suffix was eventually removed in order to keep them as short as possible.
- seqlengths (x) Return the lengths of the single sequences contained in x.
  - See ¿length, XVector-method 'and ¿length, MaskedXString-method 'for the definition of the length of a DNAString or MaskedDNAString object. Note that the length of a masked sequence (MaskedXString object) is not affected by the current set of active masks but the nchar method for MaskedXString objects is.
  - names (seqlengths (x)) is guaranteed to be identical to seqnames (x).
- mseqnames (x) Return the index of the multiple sequences contained in x. Each multiple sequence is stored in a DNAStringSet object and typically comes from a source file (FASTA) with multiple records. The names returned by mseqnames (x) usually reflect the names of those source files but a common prefix or suffix was eventually removed in order to keep them as short as possible.
- names (x) Return the index of all sequences contained in x. This is the same as c (seqnames (x), mseqnames (x)).
- length (x) Return the length of x, i.e., the number of all sequences that it contains. This is the same as length (names (x)).
- x[[name]] Return sequence (single or multiple) named name. No sequence is actually loaded
  into memory until this is explicitly requested with a call to x[[name]] or x\$name. When
  loaded, a sequence is kept in a cache. It will be automatically removed from the cache at
  garbage collection if it's not in use anymore i.e. if there are no reference to it (other than
  the reference stored in the cache). With options (verbose=TRUE), a message is printed
  each time a sequence is removed from the cache.

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x\$name Same as x[[name]] but name is not evaluated and therefore must be a literal character string or a name (possibly backtick quoted).

masknames (x) The names of the built-in masks that are defined for all the single sequences. There can be up to 4 built-in masks per sequence. These will always be (in this order): (1) the mask of assembly gaps, aka "the AGAPS mask";

- (2) the mask of intra-contig ambiguities, aka "the AMB mask";
- (3) the mask of repeat regions that were determined by the RepeatMasker software, aka "the RM mask";
- (4) the mask of repeat regions that were determined by the Tandem Repeats Finder software (where only repeats with period less than or equal to 12 were kept), aka "the TRF mask".

All the single sequences in a given package are guaranteed to have the same collection of built-in masks (same number of masks and in the same order).

masknames (x) gives the names of the masks in this collection. Therefore the value returned by masknames (x) is a character vector made of the first N elements of c("AGAPS", "AMB", "RM", "TRF"), where N depends only on the BSgenome data package being looked at  $(0 \le N \le 4)$ . The man page for most BSgenome data packages should provide the exact list and permanent URLs of the source data files that were used to extract the built-in masks. For example, if you've installed the BSgenome.Hsapiens.UCSC.hg19 package, load it and see the Note section in ¿BSgenome.Hsapiens.UCSC.hg19'.

#### Author(s)

H. Pages

#### See Also

available.genomes, GenomeDescription-class, BSgenome-utils, DNAString-class, DNAStringSet-class, MaskedDNAString-class, getSeq, injectSNPs, subseq,XVector-method, rm, gc

```
## Loading a BSgenome data package doesn't load its sequences
## into memory:
library(BSgenome.Celegans.UCSC.ce2)
## Number of sequences in this genome:
length(Celegans)
## Display a summary of the sequences:
Celegans
## Index of single sequences:
seqnames (Celegans)
## Lengths (i.e. number of nucleotides) of the sequences:
seqlengths (Celegans)
## Load chromosome I from disk to memory (hence takes some time)
## and keep a reference to it:
chrI <- Celegans[["chrI"]] # equivalent to Celegans$chrI</pre>
chrI
class(chrI)
              # a DNAString instance
```

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```
length(chrI) # with 15080483 nucleotides
## Multiple sequences:
msegnames (Celegans)
upstream1000 <- Celegans$upstream1000</pre>
upstream1000
class(upstream1000) # a DNAStringSet instance
## Character vector containing the description lines of the first
## 4 sequences in the original FASTA file:
names(upstream1000)[1:4]
## PASS-BY-ADDRESS SEMANTIC, CACHING AND MEMORY USAGE
## -----
## We want a message to be printed each time a sequence is removed
## from the cache:
options(verbose=TRUE)
gc() # nothing seems to be removed from the cache
rm(chrI, upstream1000)
qc() # chrI and upstream1000 are removed from the cache (they are
     # not in use anymore)
options(verbose=FALSE)
## Get the current amount of data in memory (in Mb):
mem0 <- gc()["Vcells", "(Mb)"]</pre>
system.time(chrV <- Celegans[["chrV"]]) # read from disk</pre>
qc()["Vcells", "(Mb)"] - mem0 # chrV occupies 20Mb in memory
system.time(tmp <- Celegans[["chrV"]]) # much faster! (sequence</pre>
                                       # is in the cache)
gc()["Vcells", "(Mb)"] - mem0 # we're still using 20Mb (sequences
                              # have a pass-by-address semantic
                              # i.e. the sequence data are not
                              # duplicated)
## subseq() doesn't copy the sequence data either, hence it is very
## fast and memory efficient (but the returned object will hold a
## reference to chrV):
y <- subseq(chrV, 10, 8000000)
gc()["Vcells", "(Mb)"] - mem0
## We must remove all references to chrV before it can be removed from
## the cache (so the 20Mb of memory used by this sequence are freed).
options(verbose=TRUE)
rm(chrV, tmp)
gc()
## Remember that 'y' holds a reference to chrV too:
rm(y)
gc()
```

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```
options(verbose=FALSE)
gc()["Vcells", "(Mb)"] - mem0
```

BSgenomeForge

The BSgenomeForge functions

#### **Description**

A set of functions for making a BSgenome data package.

#### Usage

#### **Arguments**

x A BSgenomeDataPkgSeed object or the name of a BSgenome data package seed file. See the BSgenomeForge vignette in this package for more information.

seqs\_srcdir, masks\_srcdir

Single strings indicating the path to the source directories i.e. to the directories containing the source data files. Only read access to these directories is needed. See the BSgenomeForge vignette in this package for more information.

A single string indicating the path to the directory where the source tree of the target package should be created. This directory must already exist. See the BSgenomeForge vignette in this package for more information.

verbose TRUE or FALSE. seqnames, mseqnames

A character vector containing the names of the single (for seqnames) and multiple (for mseqnames) sequences to forge. See the BSgenomeForge vignette in this package for more information.

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```
prefix, suffix
```

See the BSgenomeForge vignette in this package for more information, in particular the description of the seqfiles\_prefix and seqfiles\_suffix fields of a BSgenome data package seed file.

```
seqs_destdir, masks_destdir
```

During the forging process the source data files are converted into serialized Biostrings objects. seqs\_destdir and masks\_destdir must be single strings indicating the path to the directories where these serialized objects should be saved. These directories must already exist.

forgeSeqlengthsFile will produce a single .rda file. Both forgeSeqFiles and forgeMasksFiles will produce one .rda file per sequence.

```
nmask_per_seq
```

A single integer indicating the desired number of masks per sequence. See the BSgenomeForge vignette in this package for more information.

```
AGAPSfiles_type, AGAPSfiles_name, AGAPSfiles_prefix, AGAPSfiles_suffix, RMfiles_
These arguments are named accordingly to the corresponding fields of a BSgenome
```

These arguments are named accordingly to the corresponding fields of a BSgenome data package seed file. See the BSgenomeForge vignette in this package for more information.

#### **Details**

These functions are intended for Bioconductor users who want to make a new BSgenome data package, not for regular users of these packages. See the BSgenomeForge vignette in this package (vignette ("BSgenomeForge")) for an extensive coverage of this topic.

#### Author(s)

H. Pages

#### **Examples**

BSgenome-utils

BSgenome utilities

## Description

Utilities for BSgenome objects.

## Usage

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```
maskList = logical(0))
  ## S4 method for signature 'BSgenome':
vmatchPattern(pattern, subject, max.mismatch = 0, min.mismatch = 0,
              with.indels = FALSE, fixed = TRUE, algorithm = "auto",
              exclude = "", maskList = logical(0), userMask =
                 RangesList(), invertUserMask = FALSE, asRangedData = TRUE)
  ## S4 method for signature 'BSgenome':
vcountPattern(pattern, subject, max.mismatch = 0, min.mismatch = 0,
              with.indels = FALSE, fixed = TRUE, algorithm = "auto",
              exclude = "", maskList = logical(0), userMask =
                 RangesList(), invertUserMask = FALSE)
  ## S4 method for signature 'BSgenome':
vmatchPDict(pdict, subject, max.mismatch = 0, min.mismatch = 0,
            fixed = TRUE, algorithm = "auto", verbose = FALSE,
            exclude = "", maskList = logical(0), asRangedData = TRUE)
  ## S4 method for signature 'BSgenome':
vcountPDict(pdict, subject, max.mismatch = 0, min.mismatch = 0,
            fixed = TRUE, algorithm = "auto", collapse = FALSE,
            weight = 1L, verbose = FALSE, exclude = "", maskList = logical(0))
```

#### **Arguments**

pwm A numeric matrix with row names A, C, G and T representing a Position Weight

Matrix.

pattern A DNAString object containing the pattern sequence.

pdict A DNAStringSet object containing the pattern sequences.

subject A BSgenome object containing the subject sequences.

min.score The minimum score for counting a match. Can be given as a character string

containing a percentage (e.g. "85%") of the highest possible score or as a single

number.

max.mismatch, min.mismatch

The maximum and minimum number of mismatching letters allowed (see <code>clowlevel-matching</code> ' for the details). If non-zero, an inexact matching algorithm is

used

with.indels

If TRUE then indels are allowed. In that case, min.mismatch must be 0 and max.mismatch is interpreted as the maximum "edit distance" allowed between the pattern and a match. Note that in order to avoid pollution by redundant matches, only the "best local matches" are returned. Roughly speaking, a "best local match" is a match that is locally both the closest (to the pattern P) and the shortest. More precisely, a substring S' of the subject S is a "best local match" iff:

```
(a) nedit(P, S') <= max.mismatch</pre>
```

- (b) for every substring S1 of S':
   nedit(P, S1) > nedit(P, S')
- (c) for every substring S2 of S that contains S':
   nedit(P, S2) <= nedit(P, S')</pre>

One nice property of "best local matches" is that their first and last letters are guaranteed to be aligned with letters in P (i.e. they match letters in P).

If FALSE then IUPAC extended letters are interpreted as ambiguities (see ¿lowlevel-matching 'for the details).

fixed

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algorithm For vmatchPattern and vcountPattern one of the following: "auto", "naive-exact", "naive-inexact", "boyer-moore", "shift-or", or "indels". For vmatchPDict and vcountPDict one of the following: "auto", "naiveexact", "naive-inexact", "boyer-moore", or "shift-or". collapse, weight ignored arguments. verbose TRUE or FALSE. exclude A character vector with strings that will be used to filter out chromosomes whose names match these strings. A named logical vector of maskStates preferred when used with a BSGenome maskList object. When using the bsapply function, the masks will be set to the states in this vector. userMask A RangesList, containing a mask to be applied to each chromosome. See bsapply. invertUserMask Whether the userMask should be inverted.

asRangedData A logical value to assist in migrating output type from RangedData (deprecated) to GRanges. Should be FALSE. If TRUE, a warning message is issued and a RangedData object is returned.

#### Value

A GRanges object for matchPWM with two elementMetadata columns: "score" (numeric), and "string" (DNAStringSet).

A GRanges object for vmatchPattern.

A GRanges object for vmatchPDict with one elementMetadata column: "index", which represents a mapping to a position in the original pattern dictionary.

A data.frame object for countPWM and vcountPattern with three columns: "seqname" (factor), "strand" (factor), and "count" (integer).

A DataFrame object for vcountPDict with four columns: "seqname" ('factor' Rle), "strand" ('factor' Rle), "index" (integer) and "count" ('integer' Rle). As with vmatchPDict the index column represents a mapping to a position in the original pattern dictionary.

## Author(s)

P. Aboyoun

#### See Also

matchPWM, matchPattern, matchPDict, bsapply

```
library(BSgenome.Celegans.UCSC.ce2)
data(HNF4alpha)

pwm <- PWM(HNF4alpha)
matchPWM(pwm, Celegans, asRangedData = FALSE)
countPWM(pwm, Celegans)

pattern <- consensusString(HNF4alpha)</pre>
```

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```
vmatchPattern(pattern, Celegans, fixed = "subject", asRangedData = FALSE)
vcountPattern(pattern, Celegans, fixed = "subject")

vmatchPDict(HNF4alpha[1:10], Celegans, asRangedData = FALSE)
vcountPDict(HNF4alpha[1:10], Celegans)
```

BSParams-class

Class "BSParams"

#### **Description**

A parameter class for representing all parameters needed for running the bsapply method.

#### **Objects from the Class**

Objects can be created by calls of the form new ("BSParams", ...).

#### **Slots**

X: a BSgenome object that contains chromosomes that you wish to apply FUN on

FUN: the function to apply to each chromosome in the BSgenome object 'X'

exclude: this is a character vector with strings that will be used to filter out chromosomes whose names match these strings.

simplify: TRUE/FALSE value to indicate whether or not the function should try to simplify the output for you.

maskList: A named logical vector of maskStates preferred when used with a BSGenome object. When using the bsapply function, the masks will be set to the states in this vector.

motifList: A character vector which should contain motifs that the user wishes to mask from the sequence.

userMask: A RangesList object, where each element masks the corresponding chromosome in X. This allows the user to conveniently apply masks besides those included in X.

invertUserMask: A logical indicating whether to invert each mask in userMask.

#### Methods

bsapply (p) Performs the function FUN using the parameters contained within BSParams.

#### Author(s)

Marc Carlson

#### See Also

bsapply

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Applies a function to elements of a GenomeData

## **Description**

Returns a list of values obtained by applying a function to elements of a GenomeData or GenomeDataList object.

## Usage

```
gdapply(X, FUN, ...)
```

## Arguments

X	An object of class "GenomeData" or "GenomeDataList"
FUN	A function to be applied to each chromosome-level sub-element of X.
	Further arguments; passed to FUN

## Value

Typically an object of the same class as X.

## Author(s)

Deepayan Sarkar

gdreduce

Reduces arguments to a single GenomeData instance

## Description

This function accepts one or more objects that are reduced, with a user-specified function, to a single GenomeData instance.

#### Usage

```
gdreduce(f, ..., init, right = FALSE, accumulate = FALSE, gdArgs = list())
```

## Arguments

f	An object of class "function", accepting two instances of classes appropriate for the arguments, and returning an object suitable for subsequent use in f and incorporation into GenomeData.
	Objects to be reduced. All objects should be of the same class, as dictated by methods defined on gdreduce A function to be applied to each chromosomelevel sub-element of X.
init	An R object of the same kind as the elements of
right	A logical indicating whether to proceed from left to right (default) or right to left.

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accumulate A logical indicating whether the successive reduce combinations should be ac-

cumulated. By default, only the final combination is used.

qdArqs Additional arguments passed to the GenomeData constructor used to assemble

the final object.

#### Value

An object of class GenomeData, containing elements corresponding to the intersection of all named elements of . . . .

#### Author(s)

Martin Morgan

#### See Also

Reduce

## **Examples**

```
showMethods(gdreduce, where=getNamespace("BSgenome"))
```

GenomeData-class

Data on the genome

## **Description**

GenomeData formally represents genomic data as a list, with one element per chromosome in the genome.

#### **Details**

This class facilitates storing data on the genome by formalizing a set of metadata fields for storing the organism (e.g. Mmusculus), genome build provider (e.g. UCSC), and genome build version (e.g. mm9).

The data is represented as a list, with one element per chromosome (or really any sequence, like a gene). There are no constraints as to the data type of the elements.

Note that as a SimpleList, it is possible to store chromosome-level data (e.g. the lengths) in the elementMetadata slot. The organism, provider and providerVersion are all stored in the SimpleList metadata, so they may be retrieved in list form by calling metadata (x).

#### **Accessor methods**

In the code snippets below, x is a GenomeData object.

```
organism(x): Get the single string indicating the organism, if specified, otherwise NULL.
```

 $\label{eq:provider} {\tt provider}\,({\tt x})\,:\, \textbf{Get the single string indicating the genome build provider, if specified, otherwise} \\ {\tt NULL}.$ 

providerVersion(x): Get the single string indicating the genome build version, if specified, otherwise NULL.

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#### Constructor

```
GenomeData(listData = list(), providerVersion = metadata[["providerVersion"]], organism = metadata[["organism"]], provider = metadata[["provider"]], metadata = list(), elementMetadata = NULL, ...): Creates a GenomeData with the elements from the listData parameter, a list. The other arguments correspond to the metadata fields, and, with the exception of elementMetadata, should all be either single strings or NULL (unspecified). Additional global metadata elements may be passed in metadata, in list-form, and via . . . . The elements in metadata are always overridden by the explicit arguments, like organism and those in . . . . elementMetadata should be an DataTable or NULL.
```

#### Coercion

- as (from, "data.frame"): Coerces each subelement to a data frame, and binds them into a single data frame with an additional column indicating chromosome
- as (from, "RangesList"): Coerces each subelement to a Ranges and combines them into a RangesList with the same names. The "universe" metadata property is set to the providerVersion of from.
- as (from, "RangedData"): Coerces each subelement to a RangedData and combines them into a single RangedData with the same names. The "universe" metadata property is set to the providerVersion of from.

#### Reduction

```
gdreduce(f, ..., init, right=FALSE, accumulate=FALSE, gdArgs=list()):
    Successively combine GenomeData elements of ... using f; all arguments assigned to
    ... must be of class GenomeData. f is a function accepting two objects returned by "[["
    applied to the successive elements of ..., returning a single GenomeData object to be used
    in subsequent calls to f. init, right, and accumulate are as described for Reduce.
    gdArgs can be used to provide metadata information to the constructor used to create the
    final GenomeData object.
```

#### Author(s)

Michael Lawrence

#### See Also

GenomeDataList, a container of this class and useful for storing data on multiple samples. SimpleList, the base of this class.

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GenomeDataList-class

List of GenomeData objects

#### **Description**

GenomeDataList is a list of GenomeData objects. It could be useful for storing data on multiple experiments or samples.

#### **Details**

This class inherits from SimpleList and requires that all of its elements to be instances of GenomeData.

One should try to take advantage of the metadata storage facilities provided by SimpleList. The elementMetadata field, for example, could be used to store the experimental design, while the metadata field could store the experimental platform.

#### Constructor

```
GenomeDataList (listData = list(), metadata = list(), elementMetadata = NULL): Creates a GenomeDataList with the elements from the listData parameter, a list of GenomeData instances. The other arguments correspond to the optional metadata stored in SimpleList.
```

#### Coercion

as (from, "data.frame"): Coerces each subelement to a data frame, and binds them into a single data frame with an additional column indicating chromosome

## Reduction

```
gdreduce(f, ..., init, right=FALSE, accumulate=FALSE, gdArgs=list()):

Currently this method works when a single GenomeDataList is provided as .... It successively combines the GenomeData elements in the GenomeDataList using f. f is a function accepting two objects returned by "[[" applied to the successive GenomeData elements of ..., returning a single GenomeData object to be used in subsequent calls to f. init, right, and accumulate are as described for Reduce. gdArgs can be used to provide metadata information to the constructor used to create the final GenomeData object.
```

## Author(s)

Michael Lawrence

#### See Also

GenomeData, the type of elements stored in this class. SimpleList

#### **Examples**

GenomeDescription-class

GenomeDescription objects

## **Description**

A GenomeDescription object holds the meta information describing a given genome.

#### **Details**

In general the user will not need to manipulate directly a GenomeDescription instance but will manipulate instead a higher-level object that belongs to a class containing the GenomeDescription class. For example the top-level object defined in any BSgenome data package is a BSgenome object. But because the BSgenome class contains the GenomeDescription class, it is also a GenomeDescription object and can therefore be treated as such. In other words all the methods described below will work on it.

#### Accessor methods

In the code snippets below, x is a GenomeDescription object.

```
organism(x): Return the target organism for this genome e.g. "Homo sapiens", "Mus musculus", "Caenorhabditis elegans", etc...

species(x): Return the target species for this genome e.g. "Human", "Mouse", "Worm", etc...
```

provider(x): Return the provider of this genome e.g. "UCSC", "BDGP", "FlyBase", etc... providerVersion(x): Return the provider-side version of this genome. For example UCSC uses versions "hg18", "hg17", etc... for the different Builds of the Human genome.

releaseDate(x): Return the release date of this genome e.g. "Mar. 2006".

releaseName(x): Return the release name of this genome, which is generally made of the name of the organization who assembled it plus its Build version. For example, UCSC uses "hg18" for the version of the Human genome corresponding to the Build 36.1 from NCBI hence the release name for this genome is "NCBI Build 36.1".

bsgenomeName (x): Uses the meta information stored in x to make the name of the corresponding BSgenome data package (see available.genomes for details about the naming scheme used for those packages). Of course there is no guarantee that a package with that name actually exists.

#### Author(s)

H. Pages

#### See Also

```
available.genomes, BSgenome-class
```

#### **Examples**

```
library(BSgenome.Celegans.UCSC.ce2)
class(Celegans)
is(Celegans, "GenomeDescription")
provider(Celegans)
gendesc <- as(Celegans, "GenomeDescription")
class(gendesc)
gendesc
provider(gendesc)
bsgenomeName(gendesc)</pre>
```

getSeq

getSeq

## **Description**

A function for extracting a set of sequences (or subsequences) from a BSgenome object or other sequence container. This man page specifically documents the method for BSgenome objects.

## Usage

## **Arguments**

Х

A BSgenome object. See the available.genomes function for how to install a genome.

names

A character vector containing the names of the sequences in  $\times$  where to get the subsequences from, or a GRanges object, or a RangedData object, or a named RangesList object, or a named Ranges object. The RangesList or Ranges object must be named according to the sequences in  $\times$  where to get the subsequences from.

If names is missing, then seqnames (x) is used.

See ¿seqnames, BSgenome-method' and ¿mseqnames, BSgenome-method' to get the list of single sequences and multiple sequences (respectively) contained in x.

```
start, end, width
```

Vector of integers (eventually with NAs) specifying the locations of the subsequences to extract. These are not needed (and therefore it's an error to supply them) when names is a GRanges, RangedData, RangesList or Ranges object.

A vector containing "+"s or/and "-"s. This is not needed (and therefore it's an error to supply it) when names is a GRanges object or a RangedData object with a strand column.

as.character TRUE or FALSE. Should the extracted sequences be returned in a standard character vector?

Additional arguments. (Currently ignored.)

#### **Details**

L, the number of sequences to extract, is determined as follow:

- If names is a GRanges or Ranges object then L = length (names).
- If names is a RangedData object then L = nrow (names).
- If names is a RangesList object then L = length (unlist (names)).
- Otherwise, L is the length of the longest of names, start, end and width and all these arguments are recycled to this length. NAs and negative values in these 3 arguments are solved according to the rules of the SEW (Start/End/Width) interface (see ?solveUserSEW for the details).

If names is neither a GRanges object or a RangedData object with a strand column, then the strand argument is also recycled to length L.

Here is how the lookup between the names passed to the names argument and the sequences in x is performed. For each name in names:

- (1): If x contains a single sequence with that name then this sequence is used for extraction;
- (2): Otherwise the names of all the elements in all the multiple sequences are searched. If the names argument is a character vector then name is treated as a regular expression and grep is used for this search, otherwise (i.e. when the names are supplied via a higher level object like GRanges) name must match exactly the name of the sequence. If exactly one sequence is found, then it is used for extraction, otherwise an error is raised.

#### Value

A character vector of length L when as . character=TRUE.

A DNAString or DNAStringSet object when as .character=FALSE. More precisely the returned value is a DNAString object if L=1 and names is not a GRanges, RangedData, RangesList or Ranges object. Otherwise it's a DNAStringSet object.

#### Note

The default value for the as.character argument is TRUE even though getSeq is much more efficient (in terms of speed and memory usage) when used with as.character=FALSE.

Be aware that using as . character=TRUE can be very inefficient when extracting a high number of sequences (hundreds of thousands) or long sequences (> 1 million letters).

For this reason the plan is to change the default value to FALSE in the near future.

Note that the masks in x, if any, are always ignored. In other words, masked regions in the genome are extracted in the same way as unmasked regions (this is achieved by dropping the masks before extraction). See <code>:MaskedXString-class</code> ' for more information about masked sequences.

#### Author(s)

H. Pages; improvements suggested by Matt Settles and others

#### See Also

available.genomes, BSgenome-class, seqnames, BSgenome-method, mseqnames, BSgenome-method, [[,BSgenome-method, DNAString-class, DNAStringSet-class, MaskedDNAString-class, GRanges-class, RangedData-class, RangesList-class, Ranges-class, subseq, XVector-method, grep

```
## A. SIMPLE EXAMPLES
 # Load the Caenorhabditis elegans genome (UCSC Release ce2):
 library(BSgenome.Celegans.UCSC.ce2)
 # Look at the index of sequences:
 Celegans
 # Get chromosome V as a DNAString object:
 getSeq(Celegans, "chrV", as.character=FALSE)
 # which is in fact the same as doing:
 Celegans$chrV
  ## Not run:
    # Never try this:
   getSeq(Celegans, "chrV")
   # or this (even worse):
   getSeq(Celegans)
## End(Not run)
 # Get the first 20 bases of each chromosome:
 getSeq(Celegans, end=20)
 # Get the last 20 bases of each chromosome:
 getSeq(Celegans, start=-20)
 \# Get the "NM_058280_up_1000" sequence (belongs to the upstream1000
 # multiple sequence) as a character string:
 s1 <- getSeq(Celegans, "NM_058280_up_1000")</pre>
 # or as a DNAString object (more efficient):
 s2 <- getSeq(Celegans, "NM_058280_up_1000", as.character=FALSE)
 stopifnot(identical(as.character(s2), s1))
 stopifnot(identical(getSeq(Celegans, "NM_058280_up_5000", start=-1000), s1))
 ## Not run:
   # Fails because there is more than one sequence across
   # Celegans$upstream1000, Celegans$upstream2000 and Celegans$upstream5000
   # with "NM_058280" in its name:
   getSeq(Celegans, "NM_058280")
   # Fails because there is no sequence named exactly "NM_058280":
   getSeg(Celegans, "^NM_058280$")
## End(Not run)
```

```
## B. EXTRACTING SMALL SEQUENCES FROM DIFFERENT CHROMOSOMES
## -----
myseqs <- data.frame(</pre>
 chr=c("chrI", "chrX", "chrM", "chrM", "chrX", "chrI", "chrI", "chrI"),
 start=c(NA, -40, 8510, 301, 30001, 9220500, -2804, -30),
 end=c(50, NA, 8522, 324, 30011, 9220555, -2801, -11),
 strand=c("+", "-", "+", "+", "-", "-", "+", "-")
getSeg(Celegans, mysegs$chr,
      start=myseqs$start, end=myseqs$end)
getSeq(Celegans, myseqs$chr,
      start=myseqs$start, end=myseqs$end, strand=myseqs$strand)
## C. USING A GRanges OBJECT
## -----
gr1 <- GRanges(seqnames=c("chrI", "chrI", "chrM"),</pre>
              ranges=IRanges(start=101:103, width=9))
gr1 # all strand values are "*"
getSeq(Celegans, gr1) # treats strand values as if they were "+"
strand(gr1)[] <- "-"
getSeq(Celegans, gr1)
strand(gr1)[1] <- "+"
getSeq(Celegans, gr1)
strand(gr1)[2] <- "*"
if (interactive())
 getSeq(Celegans, gr1) # Error: cannot mix "*" with other strand values
gr2 <- GRanges(seqnames=c("chrM", "NM_058280_up_1000"),</pre>
             ranges=IRanges(start=103:102, width=9))
gr2
if (interactive()) {
 ## Because the sequence names are supplied via a GRanges object, they
 ## are not treated as regular expressions:
 getSeq(Celegans, gr2) # Error: sequence NM_058280_up_1000 not found
## D. EXTRACTING A HIGH NUMBER OF RANDOM 40-MERS FROM A GENOME
## Note the use of 'as.character=FALSE'.
extractRandomReads <- function(x, density, readlength)</pre>
   if (!is.integer(readlength))
       readlength <- as.integer(readlength)</pre>
   start <- lapply(seqnames(x),
                   function (name)
                     seqlength <- seqlengths(x)[name]</pre>
                     sample(seqlength - readlength + 1L,
```

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seglength \* density,

```
replace=TRUE)
                    })
    names <- rep.int(seqnames(x), elementLengths(start))</pre>
    ranges <- IRanges(start=unlist(start), width=readlength)</pre>
    strand <- strand(sample(c("+", "-"), length(names), replace=TRUE))</pre>
   gr <- GRanges(seqnames=names, ranges=ranges, strand=strand)</pre>
    getSeq(x, gr, as.character=FALSE)
## With a density of 1 read every 100 genome bases, the total number of
## extracted 40-mers is about 1 million:
rndreads <- extractRandomReads(Celegans, 0.01, 40)</pre>
## Notes:
## - The short sequences in 'rndreads' can be seen as the result of a
##
    simulated high-throughput sequencing experiment. A non-realistic
##
    one though because:
##
       (a) It assumes that the underlying technology is perfect (the
##
           generated reads have no technology induced errors).
##
       (b) It assumes that the sequenced genome is exactly the same as
##
           the reference genome.
##
       (c) The simulated reads can contain IUPAC ambiguity letters only
##
           because the reference genome contains them. In a real
##
           high-throughput sequencing experiment, the sequenced genome
##
           of course doesn't contain those letters, but the sequencer
           can introduce them in the generated reads to indicate
##
##
           ambiguous base-calling.
## - Those reads are coming from the plus and minus strands of the
##
    chromosomes.
\#\# - With a density of 0.01 and the reads being only 40-base long, the
    average coverage of the genome is only 0.4 which is low. The total
    number of reads is about 1 million and it takes less than 10 sec.
##
##
    to generate them.
## - A higher coverage can be achieved by using a higher density and/or
##
    longer reads. For example, with a density of 0.1 and 100-base reads
##
    the average coverage is 10. The total number of reads is about 10
##
    millions and it takes less than 1 minute to generate them.
## - Those reads could easily be mapped back to the reference by using
##
    an efficient matching tool like matchPDict() for performing exact
##
    matching (see ?matchPDict for more information). Typically, a
    small percentage of the reads (4 to 5% in our case) will hit the
##
    reference at multiple locations. This is especially true for such
##
##
    short reads, and, in a lower proportion, is still true for longer
##
    reads, even for reads as long as 300 bases.
```

injectSNPs

SNP injection

## **Description**

Inject SNPs from a SNPlocs data package into a genome.

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#### Usage

```
injectSNPs(x, SNPlocs_pkgname)

SNPlocs_pkgname(x)
SNPcount(x)
SNPlocs(x, seqname)

## Related utilities
available.SNPs(type=getOption("pkgType"))
installed.SNPs()
```

#### **Arguments**

x A BSgenome object.

SNPlocs\_pkgname

The name of a SNPlocs data package containing SNP information for the single sequences contained in x. This package must be already installed (inject SNPs

won't try to install it).

seqname The name of a single sequence in x.

type Character string indicating the type of package ("source", "mac.binary"

or "win.binary") to look for.

#### Value

injectSNPs returns a copy of the original genome x where some or all of the single sequences were altered by injecting the SNPs defined in the SNPlocs data package specified thru the SNPlocs\_pkgname argument. The SNPs in the altered genome are represented by an IUPAC ambiguity code at each SNP location.

SNPlocs\_pkgname, SNPcount and SNPlocs return NULL if no SNPs were injected in x (i.e. if x is not a BSgenome object returned by a previous call to <code>injectSNPs</code>). Otherwise SNPlocs\_pkgname returns the name of the package from which the SNPs were injected, SNPcount the number of SNPs for each altered sequence in x, and SNPlocs their locations in the sequence whose name is specified by <code>seqname</code>.

available. SNPs returns a character vector containing the names of the SNPlocs data packages that are currently available on the Bioconductor repositories for your version of R/Bioconductor. A SNPlocs data package contains basic SNP information (location and alleles) for a given organism.

installed. SNPs returns a character vector containing the names of the SNPlocs data packages that are already installed.

#### Note

inject SNPs, SNPlocs\_pkgname, SNPcount and SNPlocs have the side effect to try to load the SNPlocs data package if it's not already loaded.

#### Author(s)

H. Pages

## See Also

 $\label{eq:bounds} BS genome-class, \verb|IUPAC_CODE_MAP|, \verb|injectHardMask|, letterFrequencyInSlidingView|, \\ .inplaceReplaceLetterAt$ 

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```
## What SNPlocs data packages are already installed:
installed.SNPs()
## What SNPlocs data packages are available:
available.SNPs()
if (interactive()) {
  ## Make your choice and install with:
  source("http://bioconductor.org/biocLite.R")
  biocLite("SNPlocs.Hsapiens.dbSNP.20100427")
## Inject SNPs from dbSNP into the Human genome:
library(BSgenome.Hsapiens.UCSC.hg19)
Hsapiens
SNPlocs_pkgname(Hsapiens)
SNP_Hsapiens <- injectSNPs(Hsapiens, "SNPlocs.Hsapiens.dbSNP.20100427")
SNP_Hsapiens # note the extra "with SNPs injected from ..." line
SNPlocs_pkgname(SNP_Hsapiens)
SNPcount(SNP_Hsapiens)
head(SNPlocs(SNP_Hsapiens, "chr1"))
alphabetFrequency (Hsapiens$chr1)
alphabetFrequency(SNP_Hsapiens$chr1)
## Find runs of SNPs of length at least 25 in chr1. Might require
## more memory than some platforms can handle (e.g. 32-bit Windows
## and maybe some Mac OS X machines with little memory):
is_32bit_windows <- .Platform$0S.type == "windows" &&
                     .Platform$r_arch == "i386"
is_macosx <- substr(R.version$os, start=1, stop=6) == "darwin"</pre>
if (!is_32bit_windows && !is_macosx) {
    chr1 <- injectHardMask(SNP_Hsapiens$chr1)</pre>
    ambiguous_letters <- paste(DNA_ALPHABET[5:15], collapse="")</pre>
    lf <- letterFrequencyInSlidingView(chr1, 25, ambiguous_letters)</pre>
    sl <- slice(as.integer(lf), lower=25)</pre>
    v1 \leftarrow Views(chr1, start(sl), end(sl)+24)
    v1
    max(width(v1)) # length of longest SNP run
}
```

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