

Package ‘VariantExperiment’

December 14, 2024

Title A RangedSummarizedExperiment Container for VCF/GDS Data with GDS Backend

Version 1.21.0

Description VariantExperiment is a Bioconductor package for saving data in VCF/GDS format into RangedSummarizedExperiment object. The high-throughput genetic/genomic data are saved in GDSArray objects. The annotation data for features/samples are saved in DelayedDataFrame format with mono-dimensional GDSArray in each column. The on-disk representation of both assay data and annotation data achieves on-disk reading and processing and saves memory space significantly. The interface of RangedSummarizedExperiment data format enables easy and common manipulations for high-throughput genetic/genomic data with common SummarizedExperiment metaphor in R and Bioconductor.

biocViews Infrastructure, DataRepresentation, Sequencing, Annotation, GenomeAnnotation, GenotypingArray

Depends R (>= 3.6.0), S4Vectors (>= 0.21.24), SummarizedExperiment (>= 1.13.0), GenomicRanges,

License GPL-3

Encoding UTF-8

URL <https://github.com/Bioconductor/VariantExperiment>

BugReports <https://github.com/Bioconductor/VariantExperiment/issues>

Imports GDSArray (>= 1.11.1), DelayedDataFrame (>= 1.6.0), tools, utils, stats, methods, gdsfmt, SNPRelate, SeqArray, DelayedArray, Biostrings, IRanges

RoxygenNote 7.3.1

Suggests testthat, knitr, rmarkdown, markdown, BiocStyle

VignetteBuilder knitr

git_url <https://git.bioconductor.org/packages/VariantExperiment>

git_branch devel

git_last_commit f3af19f

git_last_commit_date 2024-10-29

Repository Bioconductor 3.21

Date/Publication 2024-12-13

Author Qian Liu [aut, cre],
Hervé Pagès [aut],
Martin Morgan [aut]

Maintainer Qian Liu <Qian.Liu@roswellpark.org>

Contents

VariantExperiment-package	2
loadVariantExperiment	3
makeVariantExperimentFromGDS	4
makeVariantExperimentFromVCF	6
saveVariantExperiment	8
showAvailable	9
VariantExperiment-class	10

Index	12
--------------	-----------

VariantExperiment-package

VariantExperiment: A package to represent VCF / GDS files using standard SummarizedExperiment metaphor with on-disk representation.

Description

The package VariantExperiment takes GDS file or VCF file as input, and save them in VariantExperiment object. Assay data are saved in GDSArray objects and annotation data are saved in DelayedDataFrame format, both of which remain on-disk until needed. Common manipulations like subsetting, mathematical transformation and statistical analysis are done easily and quickly in `_R_`.

Author(s)

Maintainer: Qian Liu <Qian.Liu@roswellpark.org>

Authors:

- Hervé Pagès
- Martin Morgan

See Also

Useful links:

- <https://github.com/Bioconductor/VariantExperiment>
- Report bugs at <https://github.com/Bioconductor/VariantExperiment/issues>

loadVariantExperiment *loadVariantExperiment to load the GDS back-end SummarizedExperiment object into R console.*

Description

loadVariantExperiment to load the GDS back-end SummarizedExperiment object into R console.

Usage

```
loadVariantExperiment(dir = tempdir())
```

Arguments

dir The directory to save the gds format of the array data, and the newly generated SummarizedExperiment object with array data in GDSArray format.

Value

An VariantExperiment object.

Examples

```
gds <- SeqArray::seqExampleFileName("gds")
## ve <- makeVariantExperimentFromGDS(gds)
## ve1 <- subsetByOverlaps(ve, GRanges("22:1-48958933"))
aa <- tempfile()
## saveVariantExperiment(ve1, dir=aa, replace=TRUE)
## loadVariantExperiment(dir = aa)
```

```
makeVariantExperimentFromGDS  
    makeVariantExperimentFromGDS
```

Description

Conversion of gds files into SummarizedExperiment object.

Usage

```
makeVariantExperimentFromGDS(  
  file,  
  ftnode,  
  smpname,  
  assayNames = NULL,  
  rowDataColumns = NULL,  
  colDataColumns = NULL,  
  rowDataOnDisk = TRUE,  
  colDataOnDisk = TRUE,  
  infoColumns = NULL  
)  
  
makeVariantExperimentFromSEQGDS(  
  file,  
  ftnode = "variant.id",  
  smpname = "sample.id",  
  assayNames = NULL,  
  rowDataColumns = NULL,  
  colDataColumns = NULL,  
  infoColumns = NULL,  
  rowDataOnDisk = TRUE,  
  colDataOnDisk = TRUE  
)  
  
makeVariantExperimentFromSNPGDS(  
  file,  
  ftnode = "snp.id",  
  smpname = "sample.id",  
  assayNames = NULL,  
  rowDataColumns = NULL,  
  colDataColumns = NULL,  
  rowDataOnDisk = TRUE,  
  colDataOnDisk = TRUE  
)
```

Arguments

file	the GDS file name to be converted.
ftnode	the node name for feature id (e.g., "variant.id", "snp.id", etc.).
smnnode	the node name for sample id (e.g., "sample.id").
assayNames	the gds node name that will be read into the assays slot and be represented as DelayedArray object.
rowDataColumns	which columns of rowData to import. The default is NULL to read in all variant annotation info.
colDataColumns	which columns of colData to import. The default is NULL to read in all sample related annotation info.
rowDataOnDisk	whether to save the rowData as DelayedArray object. The default is TRUE.
colDataOnDisk	whether to save the colData as DelayedArray object. The default is TRUE.
infoColumns	which columns of infoColumns to import for "SEQ_ARRAY" ("SeqVarGDSClass" gds class). The default is NULL to read in all available info columns.

Value

An VariantExperiment object.

Examples

```
## gds file from DNA-seq data

seqfile <- SeqArray::seqExampleFileName(type="gds")
ve <- makeVariantExperimentFromGDS(seqfile)
## all assay data
names(assays(ve))
showAvailable(seqfile)

## only read specific columns for feature / sample annotation.

assayNames <- showAvailable(seqfile)$assayNames
rowDatacols <- showAvailable(seqfile)$rowDataColumns
coldatacols <- showAvailable(seqfile)$colDataColumns
infocols <- showAvailable(seqfile)$infoColumns
ve1 <- makeVariantExperimentFromGDS(
  seqfile,
  assayNames = assayNames[2],
  rowDataColumns = rowDatacols[1:3],
  colDataColumns = coldatacols[1],
  infoColumns = infocols[c(1,3,5,7)],
  rowDataOnDisk = FALSE,
  colDataOnDisk = FALSE)
assay(ve1)

## the rowData(ve1) and colData(ve1) are now in DataFrame format

rowData(ve1)
```

```

colData(ve1)

## gds file from genotyping data

snpfile <- SNPRelate::snpgdsExampleFileName()
ve <- makeVariantExperimentFromGDS(snpfile)
rowData(ve)
colData(ve)
metadata(ve)

## Only read specific columns for feature annotation.

showAvailable(snpfile)
ve1 <- makeVariantExperimentFromGDS(snpfile, rowDataColumns=c("snp.allele"))
rowRanges(ve1)

## use specific conversion functions for certain gds types

veseq <- makeVariantExperimentFromSEQGDS(seqfile)
vesnp <- makeVariantExperimentFromSNPGDS(snpfile)

```

```
makeVariantExperimentFromVCF
```

The function to convert VCF files directly into VariantExperiment object.

Description

makeVariantExperimentFromVCF is the function to convert a vcf file into VariantExperiment object. The genotype data will be written as GDSArray format, which is saved in the assays slot. The annotation info for variants or samples will be written as DelayedDataFrame object, and saved in the rowData or colData slot.

Usage

```

makeVariantExperimentFromVCF(
  vcf.fn,
  out.dir = tempfile(),
  replace = FALSE,
  header = NULL,
  info.import = NULL,
  fmt.import = NULL,
  sample.info = NULL,
  ignore.chr.prefix = "chr",
  reference = NULL,
  start = 1L,
  count = -1L,
  parallel = FALSE,
  verbose = FALSE
)

```



```

ve
## convert without the INFO and FORMAT fields
ve <- makeVariantExperimentFromVCF(vcf, out.dir = tempfile(),
                                  info.import=character(0),
                                  fmt.import=character(0))

ve
## now the assay data does not include the
##"annotation/format/DP/data", and the rowData(ve) does not include
##any info columns.

```

saveVariantExperiment *saveVariantExperiment* Save all the assays in GDS format, including in-memory assays. Delayed assays with delayed operations on them are realized while they are written to disk.

Description

saveVariantExperiment Save all the assays in GDS format, including in-memory assays. Delayed assays with delayed operations on them are realized while they are written to disk.

Usage

```

saveVariantExperiment(
  ve,
  dir = tempdir(),
  replace = FALSE,
  fileFormat = NULL,
  compress = "LZMA_RA",
  chunk_size = 1000,
  rowDataOnDisk = TRUE,
  colDataOnDisk = TRUE,
  verbose = FALSE
)

```

Arguments

ve	A SummarizedExperiment object, with the array data being ordinary array structure.
dir	The directory to save the gds format of the array data, and the newly generated SummarizedExperiment object with array data in GDSArray format. The default is temporary directory within the R session.
replace	Whether to replace the directory if it already exists. The default is FALSE.
fileFormat	File format for the output gds file. See details.
compress	the compression method for writing the gds file. The default is "LZMA_RA".

chunk_size	The chunk size (number of columns) when reading GDSArray-based assays from input <code>ve</code> into memory and then write into a new gds file. Default is 1000. Can be modified to smaller value if chunk data is too big (e.g., when number of rows are large).
rowDataOnDisk	whether to save the <code>rowData</code> as <code>DelayedArray</code> object. The default is <code>TRUE</code> .
colDataOnDisk	whether to save the <code>colData</code> as <code>DelayedArray</code> object. The default is <code>TRUE</code> .
verbose	whether to print the process messages. The default is <code>FALSE</code> .

Details

If the input `SummarizedExperiment` object has `GDSArray`-based assay data, there is no need to specify the argument `fileFormat`. Otherwise, it takes values of `SEQ_ARRAY` for sequencing data or `SNP_ARRAY` for SNP array data.

Value

An `VariantExperiment` object with the new `gdsfile()` `ve.gds` as specified in `dir` argument.

Examples

```
gds <- SeqArray::seqExampleFileName("gds")
ve <- makeVariantExperimentFromGDS(gds)
gdsfile(ve)
ve1 <- subsetByOverlaps(ve, GRanges("22:1-48958933"))
ve1
gdsfile(ve1)
aa <- tempfile()
obj <- saveVariantExperiment(ve1, dir=aa, replace=TRUE)
obj
gdsfile(obj)
```

showAvailable

ShowAvailable

Description

The function to show the available entries for the arguments within `makeVariantExperimentFromGDS`

Usage

```
showAvailable(
  file,
  args = c("assayNames", "rowDataColumns", "colDataColumns", "infoColumns"),
  ftnode,
  smpname
)
```

Arguments

file	the path to the gds.class file.
args	the arguments in makeVariantExperimentFromGDS.
ftnode	the node name for feature id (e.g., "variant.id", "snp.id", etc.). Must be provided if the file format is not SNP_ARRAY or SEQ_ARRAY.
smnode	the node name for sample id (e.g., "sample.id"). Must be provided if the file format is not SNP_ARRAY or SEQ_ARRAY.

Examples

```
## snp gds file
gds <- SNPRelate::snpgdsExampleFileName()
showAvailable(gds)

## sequencing gds file
gds <- SeqArray::seqExampleFileName("gds")
showAvailable(gds)
```

VariantExperiment-class

VariantExperiment-class

Description

VariantExperiment could represent big genomic data in RangedSummarizedExperiment object, with on-disk GDS back-end data. The assays are represented by DelayedArray objects; rowData and colData could be represented by DelayedDataFrame or DataFrame objects.

Usage

```
VariantExperiment(
  assays,
  rowRanges = GRangesList(),
  colData = DelayedDataFrame(),
  metadata = list()
)

## S4 method for signature 'VariantExperiment'
gdsfile(object)
```

Arguments

assays	A 'list' or 'SimpleList' of matrix-like elements, or a matrix-like object. All elements of the list must have the same dimensions, and dimension names (if present) must be consistent across elements and with the row names of 'rowRanges' and 'colData'.
--------	---

<code>rowRanges</code>	A <code>GRanges</code> or <code>GRangesList</code> object describing the ranges of interest. Names, if present, become the row names of the <code>SummarizedExperiment</code> object. The length of the <code>GRanges</code> or <code>GRangesList</code> must equal the number of rows of the matrices in 'assays'.
<code>colData</code>	An optional <code>DataFrame</code> describing the samples. Row names, if present, become the column names of the <code>VariantExperiment</code> .
<code>metadata</code>	An optional 'list' of arbitrary content describing the overall experiment.
<code>object</code>	a <code>VariantExperiment</code> object.

Details

`VariantExperiment` class and slot getters and setters.
check "`?RangedSummarizedExperiment`" for more details.

Value

a `VariantExperiment` object.

Index

`gdsfile`, `VariantExperiment`-method
(`VariantExperiment`-class), [10](#)

`loadVariantExperiment`, [3](#)

`makeVariantExperimentFromGDS`, [4](#)
`makeVariantExperimentFromSEQGDS`
(`makeVariantExperimentFromGDS`),
[4](#)

`makeVariantExperimentFromSNPGDS`
(`makeVariantExperimentFromGDS`),
[4](#)

`makeVariantExperimentFromVCF`, [6](#)

`saveVariantExperiment`, [8](#)
`showAvailable`, [9](#)

`VariantExperiment`
(`VariantExperiment`-class), [10](#)

`VariantExperiment`-class, [10](#)

`VariantExperiment`-package, [2](#)