Package 'VariantExperiment'

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

Version 1.16.0

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

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Suggests testthat, knitr, rmarkdown, markdown, BiocStyle

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R topics documented:

`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_.

`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,
  smpnode,
  assayNames = NULL,
  rowDataColumns = NULL,
  colDataColumns = NULL,
  rowDataOnDisk = TRUE,
  colDataOnDisk = TRUE,
  infoColumns = NULL
)

makeVariantExperimentFromSEQGDS(
  file,
  ftnode = "variant.id",
  smpnode = "sample.id",
  assayNames = NULL,
  rowDataColumns = NULL,
  colDataColumns = NULL,
  infoColumns = NULL,
```

```
  rowDataOnDisk = TRUE,
  colDataOnDisk = TRUE
)

makeVariantExperimentFromSNPGDS(
  file,
  ftnode = "snp.id",
  smpnode = "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.).
smpnode	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.

assayNamess <- showAvailable(seqfile)$assayNames
```

```
rowdatacols <- showAvailable(seqfile)$rowDataColumns
coldatacols <- showAvailable(seqfile)$colDataColumns
infocols <- showAvailable(seqfile)$infoColumns
ve1 <- makeVariantExperimentFromGDS(
seqfile,
assayNames = assayNamess[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
)
```

Arguments

vcf.fn	the file name(s) of (compressed) VCF format; or a 'connection' object.
out.dir	The directory to save the gds format of the vcf data, and the newly generated VariantExperiment object with array data in GDSArray format and annotation data in DelayedDataFrame format. The default is a temporary folder.
replace	Whether to replace the directory if it already exists. The default is FALSE.
header	if NULL, 'header' is set to be 'seqVCF_Header(vcf.fn)', which is a list (with a class name "SeqVCFHeaderClass", S3 object).
info.import	characters, the variable name(s) in the INFO field for import; default is 'NULL' for all variables.
fmt.import	characters, the variable name(s) in the FORMAT field for import; default is 'NULL' for all variables.
sample.info	characters (with) file path for the sample info data. The data must have colnames (for phenotypes), rownames (sample ID's). No blank line allowed. The default is 'NULL' for no sample info.
ignore.chr.prefix	a vector of character, indicating the prefix of chromosome which should be ignored, like "chr"; it is not case-sensitive.
reference	genome reference, like "hg19", "GRCh37"; if the genome reference is not available in VCF files, users could specify the reference here.
start	the starting variant if importing part of VCF files.
count	the maximum count of variant if importing part of VCF files, -1 indicates importing to the end.
parallel	'FALSE' (serial processing), 'TRUE' (parallel processing), a numeric value indicating the number of cores, or a cluster object for parallel processing; 'parallel' is passed to the argument 'cl' in 'seqParallel', see '?SeqArray::seqParallel' for more details. The default is "FALSE".
verbose	whether to print the process messages. The default is FALSE.

Value

An VariantExperiment object.

Examples

```
## the vcf file
vcf <- SeqArray::seqExampleFileName("vcf")
## conversion
ve <- makeVariantExperimentFromVCF(vcf)
ve
## the filepath to the gds file.
gdsfile(ve)

## only read in specific info columns
ve <- makeVariantExperimentFromVCF(vcf, out.dir = tempfile(),
                                   info.import=c("OR", "GP"))
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 ve 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 rowData as DelayedArray object. The default is TRUE.
colDataOnDisk	whether to save the colData as DelayedArray object. The default is TRUE.
verbose	whether to print the process messages. The default is FALSE.

Details

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

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.
smpnode	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)

## S4 replacement method for signature 'VariantExperiment'
gdsfile(object) <- value
```

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

Details

VariantExperiment class and slot getters and setters.

check "?RangedSummarizedExperiment" for more details.

Value

a `VariantExperiment` object.

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