Package: seqminer (via r-universe)

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Type Package

Title Efficiently Read Sequence Data (VCF Format, BCF Format, METAL Format and BGEN Format) into R

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Description Integrate sequencing data (Variant call format, e.g. VCF or BCF) or meta-analysis results in R. This package can help you (1) read VCF/BCF/BGEN files by chromosomal ranges (e.g. 1:100-200); (2) read 'RareMETAL' summary statistics files; (3) read tables from a 'tabix'-indexed files; (4) annotate VCF/BCF files; (5) create customized workflow based on Makefile.

Copyright We have used the following software and made minimal necessary changes: tabix, Heng Li <lh3@live.co.uk> (MIT license), SQLite (Public Domain), Zstandard (BSD license). For tabix, we removed standard IO related functions, e.g. printf, fprintf ; also changed its un-safe pointer arithmetics. For zstandard, we removed compiler (clang, MSVC) specific preprocessing flags.

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URL http://zhanxw.github.io/seqminer/

BugReports https://github.com/zhanxw/seqminer/issues

Suggests testthat, SKAT

SystemRequirements zlib headers and libraries, GNU make, optionally also bzip2 and POSIX-compliant regex functions.

NeedsCompilation yes

RoxygenNote 7.2.3

Encoding UTF-8

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license). We removed standard IO related functions, e.g. printf, fprintf ; also changed its un-safe pointer arithmetics.), Broad Institute / Massachusetts Institute of Technology [cph], Genome Research Ltd (GRL) [cph], Facebook, Inc [cph], D. Richard Hipp [cph]

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`[.PlinkFile` *Read from binary PLINK file and return a genotype matrix*

Description

Read from binary PLINK file and return a genotype matrix

Usage

```
## S3 method for class 'PlinkFile'
plinkFileObject[sampleIndex, markerIndex]
```

Arguments

`plinkFileObject`	a PlinkFileObject obtained by openPlink()
`sampleIndex`	integer, 1-basd, index of samples to be extracted
`markerIndex`	integer, 1-basd, index of markers to be extracted

Value

genotype matrix, marker by sample

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
## these indice are nonsynonymous markers for 1:196621007-196716634",
## refer to the readVCFToMatrixByRange()
fileName = system.file("plink/all.anno.filtered.extract.bed", package = "seqminer")
filePrefix = sub(fileName, pattern = ".bed", replacement = "")
plinkObj = openPlink(filePrefix)
sampleIndex = seq(3)
markerIndex =c(14, 36)
cfh <- plinkObj[sampleIndex, markerIndex]
```

---

addJob *Add a job to a workflow*

Description

Add a job to a workflow

Usage

```
addJob(wf, job)
```

Arguments

wf	a variable of workflow class
job	a variable of job class

Examples

```
j1 <- newJob('id1', 'cmd out1', 'out1')
j2 <- newJob('id2', 'cmd out2', 'out2', depend = 'out1')
w <- newWorkflow("wf")
w <- addJob(w, j1)
w <- addJob(w, j2)

outFile <- file.path(tempdir(), "Makefile")
writeWorkflow(w, outFile)
cat('Outputted Makefile file are in the temp directory:', outFile, '\n')
```

annotateGene *Annotate a test variant*

Description

Annotate a test variant

Usage

```
annotateGene(param, chrom, position, ref, alt)
```

Arguments

param	a list of annotation configuaration (e.g. reference file, gene definition)
chrom	a vector of chromosome names
position	a vector of chromosome positions
ref	a vector of reference alleles
alt	a vector of alternative alleles

Value

annotated results in a data frame structure

See Also

makeAnnotationParameter

Examples

```
if (.Platform$endian == "little") {
  param <- list(reference = system.file("tabanno/test.fa", package = "seqminer"),
                geneFile = system.file("tabanno/test.gene.txt", package = "seqminer"))
  param <- makeAnnotationParameter(param)
  print(param)
  annotateGene(param, c("1", "1"), c(3, 5) , c("A", "C"), c("G", "C"))
} else {
  message("Tabix does not work well for big endian for now")
}
```

annotatePlain *Annotate a plain text file*

Description

Annotate a plain text file

Usage

```
annotatePlain(inFile, outFile, params)
```

Arguments

inFile	input file name
outFile	output file name
params	parameters

Value

0 if succeed

Examples

```
param <- list(reference = system.file("tabanno/test.fa", package = "seqminer"),
              geneFile = system.file("tabanno/test.gene.txt", package = "seqminer"),
              inputFormat = "plain")
param <- makeAnnotationParameter(param)
inFile <- system.file("tabanno/input.test.plain.txt", package = "seqminer")
outFile <- file.path(tempdir(), "out.annotated.txt")
annotatePlain(inFile, outFile, param)
cat('Outputted annotation results are in the temp directory:', outFile, '\n')
```

annotateVcf *Annotate a VCF file*

Description

Annotate a VCF file

Usage

```
annotateVcf(inVcf, outVcf, params)
```

Arguments

inVcf	input VCF file name
outVcf	output VCF file name
params	parameters

Value

0 if succeed

Examples

```
param <- list(reference = system.file("tabanno/test.fa", package = "seqminer"),
              geneFile = system.file("tabanno/test.gene.txt", package = "seqminer"))
param <- makeAnnotationParameter(param)
inVcf <- system.file("tabanno/input.test.vcf", package = "seqminer")
outVcf <- file.path(tempdir(), "/", "out.vcf")
annotateVcf (inVcf, outVcf, param)
cat('Annotated VCF files are in the temp directory:', outVcf, '\n')
```

---

createSingleChromosomeBCFIndex

Create a single chromosome index

Description

Create a single chromosome index

Usage

```
createSingleChromosomeBCFIndex(fileName, indexFileName = NULL)
```

Arguments

fileName	character, represents an input BCF file (Bgzipped, with Tabix index)
indexFileName	character, by default, create 'fileName'.scIdx

Value

indexFileName if success, or NULL is failed

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.headerFixed.bcf.gz", package = "seqminer")
cfh <- createSingleChromosomeBCFIndex(fileName)
```

createSingleChromosomeVCFIndex
Create a single chromosome index

Description

Create a single chromosome index

Usage

```
createSingleChromosomeVCFIndex(fileName, indexFileName = NULL)
```

Arguments

fileName	character, represents an input VCF file (Bgzipped, with Tabix index)
indexFileName	character, by default, create 'fileName'.scIdx

Value

indexFileName if success, or NULL is failed

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.vcf.gz", package = "seqminer")
cfh <- createSingleChromosomeVCFIndex(fileName)
```

download.annotation.resource
Download annotation resources to a directory

Description

Download annotation resources to a directory

Usage

```
download.annotation.resource(outputDirectory)
```

Arguments

outputDirectory	the directory to store annotation resources

Value

will not return anything

Examples

```
## Not run:
download.annotation.resource("/tmp")

## End(Not run)
```

`getCovPair` *Extract pair of positions by ranges*

Description

Extract pair of positions by ranges

Usage

```
getCovPair(covData, rangeList1, rangeList2)
```

Arguments

`covData`	a covariance matrix with positions as dimnames
`rangeList1`	character specify a range, 1-based index
`rangeList2`	character specify a range, 1-based index

Value

a covariance matrix covFileName = system.file("rvtests/rvtest.MetaCov.assoc.gz", package = "seqminer") cfh <- rvmeta.readCovByRange(covFileName, "1:196621007-196716634") rangeList1 <- "1:196621007-196700000" rangeList2 <- "1:196700000-196716634" getCovPair(cfh, rangeList1, rangeList2)

`getRefBase` *Annotate a test variant*

Description

Annotate a test variant

Usage

```
getRefBase(reference, chrom, position, len = NULL)
```

Arguments

reference	path to the reference genome file (.fa file)
chrom	a vector of chromosome names
position	a vector of chromosome positions
len	a vector of length

Value

based extracted from the reference genome

---

isDirWritable *Test whether directory is writable*

Description

Test whether directory is writable

Usage

```
isDirWritable(outDir)
```

Arguments

outDir	the name of the directory

Value

TRUE if the file is writable isDirWritable("~")

---

isInRange *Test whether a vector of positions are inside given ranges*

Description

Test whether a vector of positions are inside given ranges

Usage

```
isInRange(positions, rangeList)
```

Arguments

positions	characters, positions. e.g. c("1:2-3", "1:4")
rangeList	character, ranges, e.g. "1:1-3,1:2-4", 1-based index

Value

logical vector, TRUE/FALSE/NA

Examples

```
positions <- c("1:2-3", "1:4", "XX")
ranges <- "1:1-3,1:2-4,1:5-10"
isInRange(positions, ranges)
```

---

isTabixRange *Check if the inputs are valid tabix range such as chr1:2-300*

Description

Check if the inputs are valid tabix range such as chr1:2-300

Usage

```
isTabixRange(range)
```

Arguments

range	characer vector

Examples

```
valid <- isTabixRange(c("chr1:1-200", "X:1", "1:100-100", "chr1", "1:1-20,1:30-40"))
stopifnot(all(valid))
invalid <- isTabixRange(c(":1", "chr1::", ":-"))
stopifnot(all(!invalid))
```

---

makeAnnotationParameter *Construct a usable set of annotation parameters*

Description

Construct a usable set of annotation parameters

Usage

```
makeAnnotationParameter(param = NULL)
```

Arguments

param	a list of annotation elements

Value

list, a complete list of supported parameters

---

newJob *Create a new job*

Description

Create a new job

Usage

```
newJob(id, cmd, outFile, depend = NULL)
```

Arguments

id	character, job ids.
cmd	character, commands to run
outFile	character, the output file names after command are run successfully
depend	character vector, specify the prerequisite files (e.g. outFile from other jobs)

Examples

```
j1 <- newJob('id1', 'cmd out1', 'out1')
j2 <- newJob('id2', 'cmd out2', 'out2', depend = 'out1')
```

---

newWorkflow *Create a new workflow*

Description

Create a new workflow

Usage

```
newWorkflow(name)
```

Arguments

name	character, specify the name of the workflow

Examples

```
w <- newWorkflow("wf")
```

openPlink *Open binary PLINK files*

Description

Open binary PLINK files

Usage

```
openPlink(fileName)
```

Arguments

fileName	character, represents the prefix of PLINK input file

Value

an PLINK file object with class name ("PlinkFile")

Examples

```
fileName = system.file("plink/all.anno.filtered.extract.bed", package = "seqminer")
fileName = sub(fileName, pattern = ".bed", replacement = "")
plinkObj <- openPlink(fileName)
str(plinkObj)
```

readBGENToListByGene *Read information from BGEN file in a given range and return a list*

Description

Read information from BGEN file in a given range and return a list

Usage

```
readBGENToListByGene(fileName, geneFile, geneName)
```

Arguments

fileName	character, represents an input BGEN file (Bgzipped, with Tabix index)
geneFile	character, a text file listing all genes in refFlat format
geneName	character vector, which gene(s) to be extracted

Value

a list of chrom, pos, varid, rsid, alleles, isPhased, probability, sampleId

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("bgen/all.anno.filtered.extract.bgen", package = "seqminer")
geneFile = system.file("vcf/refFlat_hg19_6col.txt.gz", package = "seqminer")
cfh <- readBGENToListByGene(fileName, geneFile, "CFH")
```

---

readBGENToListByRange *Read information from BGEN file in a given range and return a list*

Description

Read information from BGEN file in a given range and return a list

Usage

```
readBGENToListByRange(fileName, range)
```

Arguments

fileName	character, represents an input BGEN file (Bgzipped, with Tabix index)
range	character, a text indicating which range in the BGEN file to extract. e.g. 1:100-200, 1-based index

Value

a list of chrom, pos, varid, rsid, alleles, isPhased, probability, sampleId

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("bgen/all.anno.filtered.extract.bgen", package = "seqminer")
cfh <- readBGENToListByRange(fileName, "1:196621007-196716634")
```

readBGENToMatrixByGene *Read a gene from BGEN file and return a genotype matrix*

Description

Read a gene from BGEN file and return a genotype matrix

Usage

```
readBGENToMatrixByGene(fileName, geneFile, geneName)
```

Arguments

fileName	character, represents an input BGEN file (Bgzipped, with Tabix index)
geneFile	character, a text file listing all genes in refFlat format
geneName	character vector, which gene(s) to be extracted

Value

genotype matrix

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("bgen/all.anno.filtered.extract.bgen", package = "seqminer")
geneFile = system.file("vcf/refFlat_hg19_6col.txt.gz", package = "seqminer")
cfh <- readBGENToMatrixByGene(fileName, geneFile, "CFH")
```

readBGENToMatrixByRange *Read a gene from BGEN file and return a genotype matrix*

Description

Read a gene from BGEN file and return a genotype matrix

Usage

```
readBGENToMatrixByRange(fileName, range)
```

Arguments

fileName	character, represents an input BGEN file (Bgzipped, with Tabix index)
range	character, a text indicating which range in the BGEN file to extract. e.g. 1:100-200, 1-based index

Value

genotype matrix

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("bgen/all.anno.filtered.extract.bgen", package = "seqminer")
cfh <- readBGENToMatrixByRange(fileName, "1:196621007-196716634")
```

---

readPlinkToMatrixByIndex

Read from binary PLINK file and return a genotype matrix

Description

Read from binary PLINK file and return a genotype matrix

Usage

```
readPlinkToMatrixByIndex(plinkFilePrefix, sampleIndex, markerIndex)
```

Arguments

plinkFilePrefix	a PlinkFileObject obtained by openPlink()
sampleIndex	integer, 1-basd, index of samples to be extracted
markerIndex	integer, 1-basd, index of markers to be extracted

Value

genotype matrix, marker by sample

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
## these indice are nonsynonymous markers for 1:196621007-196716634",
## refer to the readVCFToMatrixByRange()
fileName = system.file("plink/all.anno.filtered.extract.bed", package = "seqminer")
fileName = sub(fileName, pattern = ".bed", replacement = "")
sampleIndex = seq(3)
markerIndex =c(14, 36)
cfh <- readPlinkToMatrixByIndex(fileName, sampleIndex, markerIndex)
```

`readSingleChromosomeBCFToMatrixByRange`
Read a range from BCF file and return a genotype matrix

Description

Read a range from BCF file and return a genotype matrix

Usage

```
readSingleChromosomeBCFToMatrixByRange(fileName, range, indexFileName = NULL)
```

Arguments

`fileName`	character, represents an input BCF file (Bgzipped, with Tabix index)
`range`	character, a text indicating which range in the BCF file to extract. e.g. 1:100-200, 1-based index
`indexFileName`	character, index file, by default, it s 'fileName'.scIdx

Value

genotype matrix

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.headerFixed.bcf.gz", package = "seqminer")
cfh <- readSingleChromosomeBCFToMatrixByRange(fileName, "1:196621007-196716634")
```

readSingleChromosomeVCFToMatrixByRange *Read a range from VCF file and return a genotype matrix*

Description

Read a range from VCF file and return a genotype matrix

Usage

```
readSingleChromosomeVCFToMatrixByRange(fileName, range, indexFileName = NULL)
```

Arguments

fileName	character, represents an input VCF file (Bgzipped, with Tabix index)
range	character, a text indicating which range in the VCF file to extract. e.g. 1:100-200, 1-based index
indexFileName	character, index file, by default, it s 'fileName'.scIdx

Value

genotype matrix

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.vcf.gz", package = "seqminer")
cfh <- readSingleChromosomeVCFToMatrixByRange(fileName, "1:196621007-196716634")
```

readVCFToListByGene *Read information from VCF file in a given range and return a list*

Description

Read information from VCF file in a given range and return a list

Usage

```
readVCFToListByGene(
  fileName,
  geneFile,
  geneName,
  annoType,
  vcfColumn,
  vcfInfo,
  vcfIndv
)
```

Arguments

fileName	character, represents an input VCF file (Bgzipped, with Tabix index)
geneFile	character, a text file listing all genes in refFlat format
geneName	character vector, which gene(s) to be extracted
annoType	character, annotated types you would like to extract, such as "Nonsynonymous", "Synonymous". This can be left empty.
vcfColumn	character vector, which vcf columns to extract. It can be chosen from CHROM, POS, ID, REF, ALT, QUAL, FILTER, INFO, FORMAT and etc.
vcfInfo	character vector, which should be tags in the INFO columns to extarct. Common choices include: DP, AC, AF, NS
vcfIndv	character vector, which values to extract at individual level. Common choices are: GT, GQ, GD

Value

a list of genes, and each elements has specified vcfColumn, vcfinfo, vcfIndv

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.vcf.gz", package = "seqminer")
geneFile = system.file("vcf/refFlat_hg19_6col.txt.gz", package = "seqminer")
cfh <- readVCFToListByGene(fileName, geneFile, "CFH", "Synonymous",
                           c("CHROM", "POS"), c("AF", "AC"), c("GT") )
```

`readVCFToListByRange` *Read information from VCF file in a given range and return a list*

Description

Read information from VCF file in a given range and return a list

Usage

```
readVCFToListByRange(fileName, range, annoType, vcfColumn, vcfInfo, vcfIndv)
```

Arguments

`fileName`	character, represents an input VCF file (Bgzipped, with Tabix index)
`range`	character, a text indicating which range in the VCF file to extract. e.g. 1:100-200, 1-based index
`annoType`	character, annotated types you would like to extract, such as "Nonsynonymous", "Synonymous". This can be left empty.
`vcfColumn`	character vector, which vcf columns to extract. It can be chosen from CHROM, POS, ID, REF, ALT, QUAL, FILTER, INFO, FORMAT and etc.
`vcfInfo`	character vector, which should be tags in the INFO columns to extarct. Common choices include: DP, AC, AF, NS
`vcfIndv`	character vector, which values to extract at individual level. Common choices are: GT, GQ, GD

Value

a list of genes, and each elements has specified vcfColumn, vcfinfo, vcfIndv

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.vcf.gz", package = "seqminer")
cfh <- readVCFToListByRange(fileName, "1:196621007-196716634", "Nonsynonymous",
                            c("CHROM", "POS"), c("AF", "AC"), c("GT") )
```

readVCFToMatrixByGene *Read a gene from VCF file and return a genotype matrix*

Description

Read a gene from VCF file and return a genotype matrix

Usage

```
readVCFToMatrixByGene(fileName, geneFile, geneName, annoType)
```

Arguments

fileName	character, represents an input VCF file (Bgzipped, with Tabix index)
geneFile	character, a text file listing all genes in refFlat format
geneName	character vector, which gene(s) to be extracted
annoType	character, annotated types you would like to extract, such as "Nonsynonymous", "Synonymous". This can be left empty.

Value

genotype matrix

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.vcf.gz", package = "seqminer")
geneFile = system.file("vcf/refFlat_hg19_6col.txt.gz", package = "seqminer")
cfh <- readVCFToMatrixByGene(fileName, geneFile, "CFH", "Synonymous")
```

readVCFToMatrixByRange *Read a gene from VCF file and return a genotype matrix*

Description

Read a gene from VCF file and return a genotype matrix

Usage

```
readVCFToMatrixByRange(fileName, range, annoType)
```

Arguments

fileName	character, represents an input VCF file (Bgzipped, with Tabix index)
range	character, a text indicating which range in the VCF file to extract. e.g. 1:100-200, 1-based index
annoType	character, annotated types you would like to extract, such as "Nonsynonymous", "Synonymous". This can be left empty.

Value

genotype matrix

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.vcf.gz", package = "seqminer")
cfh <- readVCFToMatrixByRange(fileName, "1:196621007-196716634", "Nonsynonymous")
```

---

rvmeta.readCovByRange *Read covariance by range from METAL-format files.*

Description

Read covariance by range from METAL-format files.

Usage

```
rvmeta.readCovByRange(covFile, tabixRange)
```

Arguments

covFile	character, a covariance file (rvtests outputs using –meta cov)
tabixRange	character, a text indicating which range in the VCF file to extract. e.g. 1:100-200

Value

a matrix of covariance within given range

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
covFileName = system.file("rvtests/rvtest.MetaCov.assoc.gz", package = "seqminer")
cfh <- rvmeta.readCovByRange(covFileName, "1:196621007-196716634")
```

`rvmeta.readDataByGene` *Read association statistics by gene from METAL-format files. Both score statistics and covariance statistics will be extracted.*

Description

Read association statistics by gene from METAL-format files. Both score statistics and covariance statistics will be extracted.

Usage

```
rvmeta.readDataByGene(
  scoreTestFiles,
  covFiles,
  geneFile,
  geneName,
  multiAllelic = FALSE
)
```

Arguments

`scoreTestFiles`	character vector, score test output files (rvtests outputs using –meta score)
`covFiles`	character vector, covaraite files (rvtests outputs using –meta cov)
`geneFile`	character, a text file listing all genes in refFlat format
`geneName`	character vector, which gene(s) to be extracted
`multiAllelic`	boolean, whether to read multi-allelic sites as multiple variants or not

Value

a list of statistics including chromosome, position, allele frequency, score statistics, covariance and annotation(if input files are annotated).

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
scoreFileName = system.file("rvtests/rvtest.MetaScore.assoc.anno.gz", package = "seqminer")
covFileName = system.file("rvtests/rvtest.MetaCov.assoc.gz", package = "seqminer")
geneFile = system.file("vcf/refFlat_hg19_6col.txt.gz", package = "seqminer")
cfh <- rvmeta.readDataByGene(scoreFileName, covFileName, geneFile, "CFH")
```

`rvmeta.readDataByRange` *Read association statistics by range from METAL-format files. Both score statistics and covariance statistics will be extracted.*

Description

Read association statistics by range from METAL-format files. Both score statistics and covariance statistics will be extracted.

Usage

```
rvmeta.readDataByRange(scoreTestFiles, covFiles, ranges, multiAllelic = FALSE)
```

Arguments

`scoreTestFiles`	character vector, score test output files (rvtests outputs using –meta score)
`covFiles`	character vector, covaraite files (rvtests outputs using –meta cov)
`ranges`	character, a text indicating which range in the VCF file to extract. e.g. 1:100-200, 1-based index
`multiAllelic`	boolean, whether to read multi-allelic sites as multiple variants or not

Value

a list of statistics including chromosome, position, allele frequency, score statistics, covariance and annotation(if input files are annotated).

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
scoreFileName = system.file("rvtests/rvtest.MetaScore.assoc.anno.gz", package = "seqminer")
covFileName = system.file("rvtests/rvtest.MetaCov.assoc.gz", package = "seqminer")
geneFile = system.file("vcf/refFlat_hg19_6col.txt.gz", package = "seqminer")
cfh <- rvmeta.readDataByRange(scoreFileName, covFileName, "1:196621007-196716634")
```

rvmeta.readNullModel *Read null model statistics*

Description

Read null model statistics

Usage

```
rvmeta.readNullModel(scoreTestFiles)
```

Arguments

scoreTestFiles	character vector, score test output files (rvtests outputs using –meta score)

Value

a list of statistics fitted under the null mode (without genetic effects)

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
scoreFileName = system.file("rvtests/rvtest.MetaScore.assoc.anno.gz", package = "seqminer")
```

rvmeta.readScoreByRange *Read score test statistics by range from METAL-format files.*

Description

Read score test statistics by range from METAL-format files.

Usage

```
rvmeta.readScoreByRange(scoreTestFiles, tabixRange)
```

Arguments

scoreTestFiles	character vector, score test output files (rvtests outputs using –meta score)
tabixRange	character, a text indicating which range in the VCF file to extract. e.g. 1:100-200

Value

score test statistics within given range

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
scoreFileName = system.file("rvtests/rvtest.MetaScore.assoc.anno.gz", package = "seqminer")
cfh <- rvmeta.readScoreByRange(scoreFileName, "1:196621007-196716634")
```

`rvmeta.readSkewByRange`

Read skew by range from METAL-format files.

Description

Read skew by range from METAL-format files.

Usage

```
rvmeta.readSkewByRange(skewFile, tabixRange)
```

Arguments

`skewFile`	character, a skew file (rvtests outputs using –meta skew)
`tabixRange`	character, a text indicating which range in the VCF file to extract. e.g. 1:100-200

Value

an 3-dimensional array of skewness within given range

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
skewFileName = system.file("rvtests/rvtest.MetaSkew.assoc.gz", package = "seqminer")
cfh <- rvmeta.readSkewByRange(skewFileName, "1:196621007-196716634")
```

rvmeta.writeCovData *Write covariance association statistics files.*

Description

Write covariance association statistics files.

Usage

```
rvmeta.writeCovData(rvmetaData, outName)
```

Arguments

rvmetaData	a list vector. It's usually read by rvmeta.readDataByRange or rvmeta.readDataByGene function
outName	character, a text indicating output file prefix

Value

TRUE only if succeed

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
scoreFileName = system.file("rvtests/rvtest.MetaScore.assoc.anno.gz", package = "seqminer")
covFileName = system.file("rvtests/rvtest.MetaCov.assoc.gz", package = "seqminer")
geneFile = system.file("vcf/refFlat_hg19_6col.txt.gz", package = "seqminer")
cfh <- rvmeta.readDataByRange(scoreFileName, covFileName, "1:196621007-196716634")

outFile <- file.path(tempdir(), "cfh.MetaCov.assoc.gz")
rvmeta.writeCovData(cfh, outFile)
cat('Outputted MetaCov file are in the temp directory:', outFile, '\n')
```

rvmeta.writeScoreData *Write score-based association statistics files.*

Description

Write score-based association statistics files.

Usage

```
rvmeta.writeScoreData(rvmetaData, outName, createIndex = FALSE)
```

Arguments

rvmetaData	a list vector. It's usually read by rvmeta.readDataByRange or rvmeta.readDataByGene function
outName	character, a text indicating output file prefix
createIndex	boolean, (default FALSE), whether or not to create the index

Value

TRUE only if succeed

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
scoreFileName = system.file("rvtests/rvtest.MetaScore.assoc.anno.gz", package = "seqminer")
covFileName = system.file("rvtests/rvtest.MetaCov.assoc.gz", package = "seqminer")
geneFile = system.file("vcf/refFlat_hg19_6col.txt.gz", package = "seqminer")
cfh <- rvmeta.readDataByRange(scoreFileName, covFileName, "1:196621007-196716634")

outFile <- file.path(tempdir(), "cfh.MetaScore.assoc")
rvmeta.writeScoreData(cfh, outFile)
cat('Outputted MetaScore file are in the temp directory:', outFile, '\n')
```

SEQMINER *Efficiently Read Sequencing Data (VCF format, METAL format) into R*

Description

SeqMiner provides functions to easily load Variant Call Format (VCF) or METAL format into R

Details

The aim of this package is to save your time parsing large text file. That means data processing time can be saved for other researches. This packages requires Bgzip compressed and Tabix indexed files as input. If input files contaians annotation by TabAnno (), it is possible to extract information at the unit of genes.

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- Broad Institute / Massachusetts Institute of Technology [copyright holder]
- Genome Research Ltd (GRL) [copyright holder]
- Facebook, Inc [copyright holder]
- D. Richard Hipp [copyright holder]

See Also

Useful links:

- http://zhanxw.github.io/seqminer/
- Report bugs at https://github.com/zhanxw/seqminer/issues

`tabix.createIndex` *Create tabix index file, similar to running tabix in command line.*

Description

Create tabix index file, similar to running tabix in command line.

Usage

```
tabix.createIndex(
  bgzipFile,
  sequenceColumn = 1,
  startColumn = 4,
  endColumn = 5,
  metaChar = "#",
  skipLines = 0
)
```

Arguments

`bgzipFile`	character, an tabix indexed file
`sequenceColumn`	integer, sequence name column
`startColumn`	integer, start column
`endColumn`	integer, end column
`metaChar`	character, symbol for comment/meta lines
`skipLines`	integer, first this number of lines will be skipped

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.vcf.gz", package = "seqminer")
tabix.createIndex(fileName, 1, 2, 0, '#', 0)
```

`tabix.createIndex.meta` *Create tabix index for bgzipped MetaScore/MetaCov file*

Description

Create tabix index for bgzipped MetaScore/MetaCov file

Usage

```
tabix.createIndex.meta(bgzipFile)
```

Arguments

`bgzipFile`	character, input vcf file

See Also

http://zhanxw.com/seqminer/ for online manual and examples

http://zhanxw.github.io/rvtests/ for rvtests

Examples

```
fileName = system.file("rvtests/rvtest.MetaScore.assoc.anno.gz", package = "seqminer")
tabix.createIndex.meta(fileName)
```

`tabix.createIndex.vcf` *Create tabix index for bgzipped VCF file*

Description

Create tabix index for bgzipped VCF file

Usage

```
tabix.createIndex.vcf(bgzipVcfFile)
```

Arguments

`bgzipVcfFile`	character, input vcf file

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.vcf.gz", package = "seqminer")
tabix.createIndex.vcf(fileName)
```

`tabix.read` *Read tabix file, similar to running tabix in command line.*

Description

Read tabix file, similar to running tabix in command line.

Usage

```
tabix.read(tabixFile, tabixRange)
```

Arguments

`tabixFile`	character, an tabix indexed file
`tabixRange`	character, a text indicating which range in the VCF file to extract. e.g. 1:100-200

Value

character vector, each elements is an individual line

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
if (.Platform$endian == "little") {
  fileName = system.file("vcf/all.anno.filtered.extract.vcf.gz", package = "seqminer")
  snp <- tabix.read(fileName, "1:196623337-196632470")
} else {
  message("Tabix does not work well for big endian for now")
}
```

tabix.read.header *Read tabix file, similar to running tabix in command line.*

Description

Read tabix file, similar to running tabix in command line.

Usage

```
tabix.read.header(tabixFile, skippedLine = FALSE)
```

Arguments

tabixFile	character, an tabix indexed file
skippedLine	logical, whether to read tabix skipped lines (when used 'tabix -S NUM')

Value

a list

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.vcf.gz", package = "seqminer")
snp <- tabix.read.header(fileName)
```

tabix.read.table *Read tabix file, similar to running tabix in command line.*

Description

Read tabix file, similar to running tabix in command line.

Usage

```
tabix.read.table(
  tabixFile,
  tabixRange,
  col.names = TRUE,
  stringsAsFactors = FALSE
)
```

Arguments

tabixFile	character, an tabix indexed file
tabixRange	character, a text indicating which range in the VCF file to extract. e.g. 1:100-200
col.names	logical, use tabix file header as result headers (default: TRUE)
stringsAsFactors	logical, store loaded data as factors (default: FALSE)

Value

data frame, each elements is an individual line

See Also

http://zhanxw.com/seqminer/ for online manual and examples

Examples

```
fileName = system.file("vcf/all.anno.filtered.extract.vcf.gz", package = "seqminer")
snp <- tabix.read.table(fileName, "1:196623337-196632470")
```

---

validateAnnotationParameter

Validate annotate parameter is valid

Description

Validate annotate parameter is valid

Usage

```
validateAnnotationParameter(param, debug = FALSE)
```

Arguments

param	a list of annotation elements
debug	show extra debug information or not

Value

list, first element is TRUE/FALSE if parameter is valid/invalid;

`verifyFilename` *validate the inVcf can be created, and outVcf can be write to. will stop if any error occurs*

Description

validate the inVcf can be created, and outVcf can be write to. will stop if any error occurs

Usage

```
verifyFilename(inVcf, outVcf)
```

Arguments

`inVcf`	input file
`outVcf`	output file

`writeWorkflow` *Export workflow to Makefile*

Description

Export workflow to Makefile

Usage

```
writeWorkflow(wf, outFile)
```

Arguments

`wf`	a variable workflow class
`outFile`	character, typically named "Makefile"

Examples

```
j1 <- newJob('id1', 'cmd out1', 'out1')
j2 <- newJob('id2', 'cmd out2', 'out2', depend = 'out1')
w <- newWorkflow("wf")
w <- addJob(w, j1)
w <- addJob(w, j2)

outFile <-  file.path(tempdir(), "Makefile")
writeWorkflow(w, outFile)
cat('Outputted Makefile file are in the temp directory:', outFile, '\n')
```

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