

Package ‘seqminer’

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Type Package

Title Efficiently Read Sequence Data (VCF Format, BCF Format and METAL 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 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.

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URL <http://seqminer.genomic.codes>

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

Repository CRAN

Suggests testthat, SKAT

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

NeedsCompilation yes

RoxygenNote 6.0.1

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addJob	<i>Add a job to a workflow</i>
--------	--------------------------------

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)
writeWorkflow(w, "Makefile")
```

annotateGene	<i>Annotate a test variant</i>
--------------	--------------------------------

Description

Annotate a test variant

Usage

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

Arguments

param	a list of annotation configuration (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

```
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"))
```

annotatePlain	<i>Annotate a plain text file</i>
---------------	-----------------------------------

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 <- paste0(getwd(), "/", "out.annotated.txt")
annotatePlain(inFile, outFile, param)
```

annotateVcf	<i>Annotate a VCF file</i>
-------------	----------------------------

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 <- paste0(getwd(), "/", "out.vcf")
annotateVcf (inVcf, outVcf, param)
```

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	<i>Extract pair of positions by ranges</i>
------------	--

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
rangeList2	character specify a range

Value

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

getRefBase	<i>Annotate a test variant</i>
------------	--------------------------------

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	<i>Test whether directory is writable</i>
---------------	---

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	<i>Test whether a vector of positions are inside given ranges</i>
-----------	---

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"

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	<i>Create a new job</i>
--------	-------------------------

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	<i>Create a new workflow</i>
-------------	------------------------------

Description

Create a new workflow

Usage

```
newWorkflow(name)
```

Arguments

name	character, specify the name of the workflow
------	---

Examples

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

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, vcffIndv

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
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 extract. 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
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 <code>-meta cov</code>)
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, covariate 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, covariate files (rvtests outputs using `-meta cov`)
`ranges` character, a text indicating which range in the VCF file to extract. e.g. 1:100-200
`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 <code>-meta skew</code>)
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")
rvmeta.writeCovData(cfh, "cfh.MetaCov.assoc.gz")
```

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")
rvmeta.writeScoreData(cfh, "cfh.MetaScore.assoc")
```

SeqMiner	<i>Efficiently Read Sequencing Data (VCF format, METAL format) into R</i>
----------	---

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 contains annotation by TabAnno (), it is possible to extract information at the unit of genes.

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	<i>Read tabix file, similar to running tabix in command line.</i>
------------------	---

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	<i>validate the inVcf can be created, and outVcf can be write to. will stop if any error occurs</i>
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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	<i>Export workflow to Makefile</i>
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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)
writeWorkflow(w, "Makefile")
```

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