

Package ‘varitas’

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Type Package

Title Variant Calling in Targeted Analysis Sequencing Data

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Description Multi-caller variant analysis pipeline for targeted analysis sequencing (TAS) data. Features a modular, automated workflow that can start with raw reads and produces a user-friendly PDF summary and a spreadsheet containing consensus variant information.

SystemRequirements perl, bedtools (>=2.27.1), bwa

License GPL-2

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| | |
|------------|-------------------|
| add.option | <i>add.option</i> |
|------------|-------------------|

Description

Add option to nested list of options. Applied recursively

Usage

```
add.option(name, value, old.options, nesting.character = "\\.")
```

Arguments

| | |
|-------------------|--|
| name | Option name. Nesting is indicated by character specified in nesting.character. |
| value | New value of option |
| old.options | Nested list the option should be added to |
| nesting.character | String giving Regex pattern of nesting indication string. Defaults to '\.' |

Value

Nested list with updated options

| | |
|---------------------|----------------------------|
| alternate.gene.sort | <i>alternate.gene.sort</i> |
|---------------------|----------------------------|

Description

Given a data frame containing coverage statistics and gene information, returns that frame with the rows sorted by alternating gene size (for plotting)

Usage

```
alternate.gene.sort(coverage.statistics)
```

Arguments

| | |
|---------------------|-----------------------------------|
| coverage.statistics | Data frame of coverage statistics |
|---------------------|-----------------------------------|

Details

Genes have varying numbers of associated amplicons and when plotting coverage statistics, if two genes with very low numbers of amplicons are next to each other, the labels will overlap. This function sorts the coverage statistics data frame in a way that places the genes with the most amplicons (largest) next to those with the least (smallest).

Value

Coverage statistics data frame sorted by alternating gene size

```
build.variant.specification
      build.variant.specification
```

Description

Build data frame with paths to variant files.

Usage

```
build.variant.specification(sample.ids, project.directory)
```

Arguments

`sample.ids` Vector of sample IDs. Must match subdirectories in `project.directory`.
`project.directory` Path to directory where sample subdirectories

Details

Parses through sample IDs in a project directory and returns paths to variant files based on (theoretical) file name patterns. Useful for testing, or for entering the pipeline at non-traditional stages.

Value

Data frame with paths to variant files.

```
caller.overlap.venn.diagram
      . Make Venn diagram of variant caller overlap
```

Description

. Make Venn diagram of variant caller overlap

Usage

```
caller.overlap.venn.diagram(variants, file.name)
```

Arguments

`variants` Data frame containing variants, typically from `merge.variants` function
`file.name` Name of output file

capitalize.caller *capitalize.caller*

Description

Capitalize variant caller name

Usage

```
capitalize.caller(caller)
```

```
capitalise.caller(caller)
```

Arguments

caller Character vector of callers to be capitalized

Value

Vector of same length as caller where eligible callers have been capitalized

classify.variant *classify.variant*

Description

Classify a variant as SNV, MNV, or indel based on the reference and alternative alleles

Usage

```
classify.variant(ref, alt)
```

Arguments

ref Vector of reference bases

alt Vector of alternate bases

Value

Character vector giving type of variant.

convert.ides.output *Convert output of iDES step 1 to variant call format*

Description

Convert output of iDES step 1 to variant call format

Usage

```
convert.ides.output(filename, output = TRUE,  
  output.suffix = ".calls.txt", minreads = 5, mindepth = 50)
```

Arguments

| | |
|---------------|--|
| filename | Path to file |
| output | Logical indicating whether output should be saved to file. Defaults to true. |
| output.suffix | Suffix to be appended to input filename if saving results to file |
| minreads | Minimum numbers of reads |
| mindepth | Minimum depth |

Value

potential.calls Data frame of converted iDES calls

create.directories *create.directories*

Description

Create directories in a given path

Usage

```
create.directories(directory.names, path)
```

Arguments

| | |
|-----------------|--|
| directory.names | Vector of names of directories to be created |
| path | Path where directories should be created |

date.stamp.file.name *date.stamp.file.name*

Description

Prefix file name with a date-stamp.

Usage

```
date.stamp.file.name(file.name, date = Sys.Date(), separator = "_")
```

Arguments

| | |
|-----------|---|
| file.name | File name to be date-stamped |
| date | Date to be added. Defaults to current date. |
| separator | String that should separate the date from the file name. Defaults to a single underscore. |

Value

String giving the datestamped file name

Examples

```
date.stamp.file.name('plot.png');  
date.stamp.file.name('yesterdays_plot.png', date = Sys.Date() - 1);
```

extract.sample.ids *Extract sample IDs from file paths*

Description

Extract sample IDs from a set of paths to files in sample-specific subfolders

Usage

```
extract.sample.ids(paths, from.filename = FALSE)
```

Arguments

| | |
|---------------|---|
| paths | vector of file paths |
| from.filename | Logical indicating whether sample ID should be extracted from filename rather than path |

Value

vector of extracted sample IDs

filter.variant.file *Filter variants in file.*

Description

Filter variants from file, and save to output. Wrapper function that opens the variant file, calls filter.variants, and saves the result to file

Usage

```
filter.variant.file(variant.file, output.file, config.file = NULL,
  caller = c("vardict", "ides", "mutect", "pgm", "consensus"))
```

Arguments

| | |
|--------------|---|
| variant.file | Path to variant file |
| output.file | Path to output file |
| config.file | Path to config file to be used. If not supplied, will use the pre-existing VariTAS options. |
| caller | Name of caller used (needed to match appropriate filters from settings) |

Value

None

filter.variants *Filter variant calls*

Description

Filter data frame of variant calls based on thresholds specified in settings.

Usage

```
filter.variants(variants, caller = c("vardict", "ides", "mutect", "pgm",
  "consensus", "isis", "varscan", "lofreq"), config.file = NULL,
  verbose = FALSE)
```

Arguments

| | |
|-------------|--|
| variants | Data frame of variant calls with ANNOVAR annotation, or path to variant file. |
| caller | Name of caller used (needed to match appropriate filters from settings) |
| config.file | Path to config file to be used. If not supplied, will use the pre-existing VariTAS options. |
| verbose | Logical indicating whether to output descriptions of filtering steps. Defaults to False, useful for debugging. |

Value

filtered.variants Data frame of filtered variants

| | |
|---------------|----------------------|
| fix.lofreq.af | <i>fix.lofreq.af</i> |
|---------------|----------------------|

Description

LoFreq also does not output allele frequencies, so this script calculates them from the DP (depth) and AD (variant allele depth) values—which are also not output nicely— and adds them to the annotated vcf.

Usage

```
fix.lofreq.af(variant.specification)
```

Arguments

```
variant.specification
    Data frame of variant file information
```

| | |
|-----------|--------------------------------------|
| fix.names | <i>Fix variant call column names</i> |
|-----------|--------------------------------------|

Description

Fix headers of variant calls to prepare for merging. This mostly consists in making sure the column headers will be unique by prefixing the variant caller in question.

Usage

```
fix.names(column.names, variant.caller, sample.id = NULL)
```

Arguments

```
column.names    Character vector of column names
variant.caller  String giving name of variant caller
sample.id       Optional sample ID. Used to fix headers.
```

Value

```
new.column.names Vector of column names after fixing]
```

| | |
|----------------|-----------------------|
| fix.varscan.af | <i>fix.varscan.af</i> |
|----------------|-----------------------|

Description

VarScan does not output allele frequencies, so this script calculates them from the DP (depth) and AD (variant allele depth) values and adds them to the annotated vcf.

Usage

```
fix.varscan.af(variant.specification)
```

Arguments

```
variant.specification  
    Data frame of variant file information
```

| | |
|-----------------------|------------------------------|
| get.base.substitution | <i>Get base substitution</i> |
|-----------------------|------------------------------|

Description

Get base substitution represented by pyrimidine in base pair. If more than one base in REF/ALT (i.e. MNV or indel rather than SNV), NA will be returned

Usage

```
get.base.substitution(ref, alt)
```

Arguments

```
ref          Vector of reference bases  
alt         Vector of alternate bases
```

Value

```
base.substitutions
```

`get.bed.chromosomes` *get.bed.chromosomes*

Description

Extract chromosomes from bed file

Usage

`get.bed.chromosomes`(bed)

Arguments

bed Path to BED file

Value

Vector containing all chromosomes in BED file

`get.buildver` *get.buildver*

Description

Get build version (hg19/hg38) based on settings.

Parses VariTAS pipeline settings to get the build version. When this function was first developed, the idea was to be able to explicitly set ANNOVAR filenames based on the build version.

Usage

`get.buildver`()

Value

String giving reference genome build version (hg19 or hg38)

| | |
|-------------|---------------------------------|
| get.colours | <i>Generate a colour scheme</i> |
|-------------|---------------------------------|

Description

Generate a colour scheme

Usage

```
get.colours(n)
```

Arguments

| | |
|---|---------------------------|
| n | Number of colours desired |
|---|---------------------------|

Value

Colour.scheme generated colours

| | |
|--------------------------|--|
| get.coverage.by.amplicon | <i>Process sample coverage per amplicon data</i> |
|--------------------------|--|

Description

Parse coverageBed output to get coverage by amplicon

Usage

```
get.coverage.by.amplicon(project.directory)
```

Arguments

| | |
|-------------------|---|
| project.directory | Path to project directory. Each sample should have its own subdirectory |
|-------------------|---|

Value

combined.data Data frame giving coverage per amplicon per sample.

References

<http://bedtools.readthedocs.io/en/latest/content/tools/coverage.html>

```
get.coverage.by.sample.statistics
```

Get statistics about coverage per sample

Description

Get statistics about coverage per sample

Usage

```
get.coverage.by.sample.statistics(project.directory)
```

Arguments

```
project.directory
```

Path to project directory. Each sample should have its own subdirectory

Value

`coverage.by.sample.statistics` Data frame with coverage statistics per sample

```
get.fasta.chromosomes get.fasta.chromosomes
```

Description

Extract chromosomes from fasta headers.

Usage

```
get.fasta.chromosomes(fasta)
```

Arguments

```
fasta
```

Path to reference fasta

Value

Vector containing all chromosomes in fasta file.

| | |
|---------------|----------------------|
| get.file.path | <i>get.file.path</i> |
|---------------|----------------------|

Description

Get absolute path to sample-specific file for one or more samples

Usage

```
get.file.path(sample.ids, directory, extension = NULL,
              allow.multiple = FALSE, allow.none = FALSE)
```

Arguments

| | |
|----------------|--|
| sample.ids | Vector of sample IDs to match filename on |
| directory | Path to directory containing files |
| extension | String giving extension of file |
| allow.multiple | Boolean indicating whether to allow multiple matching files. Defaults to false, which throws an error if the query matches more than one file. |
| allow.none | Boolean indicating whether to allow no matching files. Defaults to false, which throws an error if the query does not match any files. |

Value

Paths to matched files

| | |
|-------------|--------------------|
| get.filters | <i>get.filters</i> |
|-------------|--------------------|

Description

Determine filters per caller, given default and caller-specific values.

Usage

```
get.filters(filters)
```

Arguments

| | |
|---------|--|
| filters | List of filter values. These will be updated to use default as the baseline, with caller-specific filters taking precedence if supplied. |
|---------|--|

Value

A list with updated filters

| | |
|-----------------------|-----------------|
| <code>get.gene</code> | <i>get.gene</i> |
|-----------------------|-----------------|

Description

Use guesswork to extract gene from data frame of targeted panel data. The panel designer output can change, so try to guess what the format is.

Usage

```
get.gene(bed.data)
```

Arguments

| | |
|-----------------------|--|
| <code>bed.data</code> | Data frame containing data from bed file |
|-----------------------|--|

Value

vector of gene names, one entry for each row of `bed.data`

| | |
|---------------------------------------|---------------------------------|
| <code>get.miniseq.sample.files</code> | <i>get.miniseq.sample.files</i> |
|---------------------------------------|---------------------------------|

Description

Get files for a sample in a directory, ensuring there's only a single match per sample ID.

Usage

```
get.miniseq.sample.files(sample.ids, directory,
  file.suffix = "_S\\d{1,2}_.*")
```

Arguments

| | |
|--------------------------|--|
| <code>sample.ids</code> | Vector of sample ids. Should form first part of file name |
| <code>directory</code> | Directory where files can be found |
| <code>file.suffix</code> | Regex expression for end of file name. For example, <code>'file.suffix = '_S\\d1,2_.*_R1_.*'</code> will match R1 files.1 files. |

Value

Character vector of file paths

| | |
|------------|---|
| get.option | <i>Helper function to recursively get an VariTAS option</i> |
|------------|---|

Description

Helper function to recursively get an VariTAS option

Usage

```
get.option(name, varitas.options = NULL, nesting.character = "\\.")
```

Arguments

| | |
|-------------------|---|
| name | Option name |
| varitas.options | Optional list of options to search in |
| nesting.character | String giving Regex pattern of nesting indication string. Defaults to '\\.' |

Value

value Requested option

| | |
|----------------------------|---|
| get.panel.coverage.by.gene | <i>Summarise panel coverage by gene</i> |
|----------------------------|---|

Description

Summarise panel coverage by gene

Usage

```
get.panel.coverage.by.gene(panel.file, gene.col = 5)
```

Arguments

| | |
|------------|--------------------------------------|
| panel.file | path to panel |
| gene.col | index of column containing gene name |

Value

panel.coverage.by.gene data frame giving the number of amplicons and their total length by gene

```
get.pool.from.panel.data
```

Get pool corresponding to each amplicon

Description

The bed files are not consistent, so it's not clear where the pool will appear. This function parses through the columns to identify where the pool

Usage

```
get.pool.from.panel.data(panel.data)
```

Arguments

panel.data data frame pool should be extracted from

Value

pools vector of pool information

```
get.varitas.options      Return VariTAS settings
```

Description

Return VariTAS settings

Usage

```
get.varitas.options(option.name = NULL, nesting.character = "\\.")
```

Arguments

option.name Optional name of option. If no name is supplied, the full list of VariTAS options will be provided.

nesting.character String giving Regex pattern of nesting indication string. Defaults to '\\.'

Value

varitas.options list specifying VariTAS options

Examples

```
reference.build <- get.varitas.options('reference_build');
mutect.filters <- get.varitas.options('filters.mutect');
```

logical.to.character *logical.to.character*

Description

Convert a logical vector to a T/F coded character vector. Useful for preventing unwanted T->TRUE nucleotide conversions

Usage

```
logical.to.character(x)
```

Arguments

x Vector to be converted

Value

Character vector after converting TRUE/FALSE

make.command.line.call

Make string with command line call from its individual components

Description

Make string with command line call from its individual components

Usage

```
make.command.line.call(main.command, options = NULL, flags = NULL,
  option.prefix = "--", option.separator = " ", flag.prefix = "--")
```

Arguments

main.command String or vector of strings giving main part of command (e.g. "python test.py" or c("python", "test.py"))

options Named vector or list giving options

flags Vector giving flags to include.

option.prefix String to preface all options. Defaults to "-"

option.separator String to separate options from their values. Defaults to a single space.

flag.prefix String to preface all flags. Defaults to "-"

Value

command string giving command line call

| | |
|------------------|-------------------------|
| mean.field.value | <i>mean.field.value</i> |
|------------------|-------------------------|

Description

Get mean value of a variant annotation field

Usage

```
## S3 method for class 'field.value'
mean(variants, field = c("TUMOUR.DP", "NORMAL.DP",
  "NORMAL.AF", "TUMOUR.AF", "QUAL"), caller = c("consensus", "vardict",
  "pgm", "mutect", "isis", "varscan", "lofreq"))
```

Arguments

| | |
|----------|---|
| variants | Data frame with variants |
| field | String giving field of interest. |
| caller | String giving caller to calculate values from |

Details

As part of the variant merging process, annotated variant data frames are merged into one, with the value from each caller prefixed by CALLER. For example, the VarDict normal allele frequency will have header VARDICT.NORMAL.AF. This function takes the average of all callers' value for a given field, removing NA's. If only a single caller is present in the data frame, that value is returned.

Value

Vector of mean values.

| | |
|-----------------------|--|
| merge.ides.annotation | <i>Merge potential iDES calls with variant annotation.</i> |
|-----------------------|--|

Description

Merge potential iDES calls with variant annotation.

Usage

```
## S3 method for class 'ides.annotation'
merge(ides.filename, output = TRUE,
  output.suffix = ".ann.txt",
  annovar.suffix.pattern = ".annovar.hg(\\d{2})_multianno.txt")
```

Arguments

`ides.filename` Path to formatted iDES output (typically from `convert.ides.output` file)
`output` Logical indicating whether output should be saved to file. Defaults to true.
`output.suffix` Suffix to be appended to input filename if saving results to file
`annovar.suffix.pattern` Suffix to match ANNOAR file

Details

The VarDict variant calling includes a GATK call merging the call vcf file (allele frequency information etc.) with the ANNOVAR annotation, and saving the result as a table. This function is an attempt to emulate that step for the iDES calls.

Value

`annotated.calls` Data frame of annotations and iDES output.

| | |
|-----------------------------|-----------------------|
| <code>merge.variants</code> | <i>Merge variants</i> |
|-----------------------------|-----------------------|

Description

Merge variants from multiple callers and return a data frame of merged calls. By default filtering is also applied, although this behaviour can be turned off by setting `apply.filters` to FALSE.

Usage

```
## S3 method for class 'variants'
merge(variant.specification, apply.filters = TRUE,
      remove.structural.variants = TRUE,
      separate.consensus.filters = FALSE, verbose = FALSE)
```

Arguments

`variant.specification` Data frame containing details of file paths, sample IDs, and caller.
`apply.filters` Logical indicating whether to apply filters. Defaults to TRUE.
`remove.structural.variants` Logical indicating whether structural variants (including CNVs) should be removed. Defaults to TRUE.
`separate.consensus.filters` Logical indicating whether to apply different thresholds to variants called by more than one caller (specified under `consensus` in config file). Defaults to FALSE.
`verbose` Logical indicating whether to print information to screen

Value

Data frame

```
overwrite.varitas.options
      overwrite.varitas.options
```

Description

Overwrite VariTAS options with options provided in config file.

Usage

```
overwrite.varitas.options(config.file)
```

Arguments

`config.file` Path to config file that should be used to overwrite options

Value

None

Examples

```
## Not run:
config <- file.path(path.package('varitas'), 'config.yaml')
overwrite.varitas.options(config)

## End(Not run)
```

```
parse.job.dependencies
      Parse job dependencies
```

Description

Parse job dependencies to make the functions more robust to alternate inputs (e.g. people writing alignment instead of bwa)

Usage

```
parse.job.dependencies(dependencies)
```

Arguments

`dependencies` Job dependency strings to be parsed.

Value

`parsed.dependencies` Vector of job dependencies after reformatting.

```
plot.amplicon.coverage.per.sample
plot.amplicon.coverage.per.sample
```

Description

Create one scatterplot per sample, showing coverage per amplicon, and an additional plot giving the median

Usage

```
## S3 method for class 'amplicon.coverage.per.sample'
plot(coverage.statistics,
     output.directory)
```

Arguments

`coverage.statistics`
Data frame containing coverage per amplicon per sample, typically from `get.coverage.by.amplicon`.

`output.directory`
Directory where per sample plots should be saved

Value

None

```
plot.coverage.by.genome.order
Plot amplicon coverage by genome order
```

Description

Use values obtained by `bedtools coverage` to make a plot of coverage by genome order

Usage

```
## S3 method for class 'coverage.by.genome.order'
plot(coverage.data)
```

Arguments

coverage.data data frame with results from bedtools coverage command

plot.coverage.by.sample
plot.coverage.by.sample

Description

Make a barplot of coverage per sample

Usage

```
## S3 method for class 'coverage.by.sample'  
plot(coverage.sample, file.name,  
      statistic = c("mean", "median"))
```

Arguments

coverage.sample Data frame of coverage data, typically from `get.coverage.by.sample.statistics`
file.name Name of output file
statistic Statistic to be plotted (mean or median)

Value

None

plot.ontarget.percent *plot.ontarget.percent*

Description

Make a scatterplot of ontarget percent per sample

Usage

```
## S3 method for class 'ontarget.percent'  
plot(coverage.sample, file.name)
```

Arguments

coverage.sample Data frame of coverage data, typically from `get.coverage.by.sample.statistics`
file.name Name of output file

Value

None

| | |
|---------------------|----------------------------|
| plot.paired.percent | <i>plot.paired.percent</i> |
|---------------------|----------------------------|

Description

Make a barplot of percent paired reads per sample

Usage

```
## S3 method for class 'paired.percent'
plot(coverage.sample, file.name)
```

Arguments

| | |
|-----------------|--|
| coverage.sample | Data frame of coverage data, typically from <code>get.coverage.by.sample.statistics</code> |
| file.name | Name of output file |

Value

None

| | |
|-----------------|--|
| post.processing | <i>Post-processing of variants to generate outputs</i> |
|-----------------|--|

Description

Post-processing of variants to generate outputs

Usage

```
post.processing(variant.specification, project.directory,
  config.file = NULL, variant callers = NULL,
  remove.structural.variants = TRUE,
  separate.consensus.filters = FALSE, sleep = FALSE, verbose = FALSE)
```

Arguments

| | |
|----------------------------|--|
| variant.specification | Data frame specifying variants to be processed, or path to data frame (useful if calling from Perl) |
| project.directory | Directory where output should be stored. Output files will be saved to a date-stamped subdirectory |
| config.file | Path to config file specifying post-processing options. If not provided, the current options are used (i.e. from <code>get.varitas.options()</code>) |
| variant callers | Optional vector of variant callers for which filters should be included in Excel file |
| remove.structural.variants | Logical indicating whether structural variants (including CNVs) should be removed. Defaults to TRUE. |
| separate.consensus.filters | Logical indicating whether to apply different thresholds to variants called by more than one caller (specified under consensus in config file). Defaults to FALSE. |
| sleep | Logical indicating whether script should sleep for 60 seconds before starting. |
| verbose | Logical indicating whether to print verbose output |

Value

None

```
prepare.bam.specification
```

Prepare BAM specification data frame to standardized format for downstream analyses.

Description

This function prepares a data frame that can be used to run variant callers. For matched normal variant calling, this data frame will contain three columns with names: `sample.id`, `tumour.bam`, `normal.bam`. For unpaired variant calling, the data frame will contain two columns with names: `sample.id`, `tumour.bam`.

Usage

```
prepare.bam.specification(sample.details, paired = TRUE,
  sample.id.column = 1, tumour.bam.column = 2, normal.bam.column = 3)
```

Arguments

| | |
|--------------------------------|--|
| <code>sample.details</code> | Data frame where each row represents a sample to be run. Must contain sample ID, path to tumour BAM, and path to normal BAM. |
| <code>paired</code> | Logical indicating whether the sample specification is for a paired analysis. |
| <code>sample.id.column</code> | Index or string giving column of <code>sample.details</code> that contains the sample ID |
| <code>tumour.bam.column</code> | Index or string giving column of <code>sample.details</code> that contains the path to the tumour BAM |
| <code>normal.bam.column</code> | Index or string giving column of <code>sample.details</code> that contains the path to the normal BAM |

Value

`bam.specification` Data frame with one row per sample to be run

`prepare.fastq.specification`
prepare.fastq.specification

Description

Prepare FASTQ specification data frame to standardized format for downstream analyses.

Usage

```
prepare.fastq.specification(sample.details, sample.id.column = 1,
  fastq.columns = c(2, 3), patient.id.column = NA,
  tissue.column = NA)
```

Arguments

| | |
|--------------------------------|--|
| <code>sample.details</code> | Data frame where each row represents a sample to be run. Must contain sample ID, path to tumour BAM, and path to normal BAM. |
| <code>sample.id.column</code> | Index or string giving column of <code>sample.details</code> that contains the sample ID |
| <code>fastq.columns</code> | Index or string giving column(s) of <code>sample.details</code> that contain path to FASTQ files |
| <code>patient.id.column</code> | Index or string giving column of <code>sample.details</code> that contains the patient ID |
| <code>tissue.column</code> | Index or string giving column of <code>sample.details</code> that contains information on tissue (tumour/ normal) |

Details

This function prepares a data frame that can be used to run alignment. For paired-end reads, this data frame will contain three columns with names: sample.id, reads, mates For single-end reads, the data frame will contain two columns with names: sample.id, reads

Value

Data frame with one row per sample to be run

```
prepare.miniseq.specifications  
  prepare.miniseq.specifications
```

Description

Process a MiniSeq directory and sample sheet to get specification data frames that can be used to run the VariTAS pipeline.

Note: This assumes normal samples are not available.

Usage

```
prepare.miniseq.specifications(sample.sheet, miniseq.directory)
```

Arguments

sample.sheet Data frame containing sample information, or path to a MiniSeq sample sheet
miniseq.directory Path to directory with MiniSeq files

Value

A list with specification data frames 'fastq', 'bam', and 'vcf' (as applicable)

Examples

```
miniseq.sheet <- file.path(path.package('varitas'), 'extdata/miniseq/Example_template.csv')  
miniseq.directory <- file.path(path.package('varitas'), 'extdata/miniseq')  
miniseq.info <- prepare.miniseq.specifications(miniseq.sheet, miniseq.directory)
```

```
prepare.vcf.specification
```

```
prepare.vcf.specification
```

Description

Prepare VCF specification data frame for annotation

Usage

```
prepare.vcf.specification(vcf.details, sample.id.column = 1,
  vcf.column = 2, job.dependency.column = NA, caller.column = NA)
```

Arguments

| | |
|-----------------------|---|
| vcf.details | Data frame containing details of VCF files |
| sample.id.column | Identifier of column in vcf.details containing sample IDs (index or name) |
| vcf.column | Identifier of column in vcf.details containing VCF file (index or name) |
| job.dependency.column | Identifier of column in vcf.details containing job dependency (index or name) |
| caller.column | Identifier of column in vcf.details containing caller (index or name) |

Value

Properly formatted VCF details

```
process.coverage.reports
```

```
Process coverageBed reports
```

Description

Process the coverage reports generated by bedtools coverage tool.

Usage

```
process.coverage.reports(project.directory)
```

Arguments

| | |
|-------------------|---|
| project.directory | Path to project directory. Each sample should have its own subdirectory |
|-------------------|---|

Value

final.statistics data frame of coverage statistics generated by parsing through coverage reports

`process.sample.contamination.checks`*Process sample contamination checks*

Description

Takes *selfSM reports generated by VerifyBamID during alignment, and returns a vector of freemix scores. The freemix score is a sequence only estimate of sample contamination that ranges from 0 to 1.

Note: Targeted panels are often too small for this step to work properly.

Usage

```
process.sample.contamination.checks(project.directory)
```

Arguments

```
project.directory
```

Path to project directory. Each sample should have its own subdirectory

Value

freemix.scores Data frame giving sample contamination (column freemix) score per sample.

References

<https://genome.sph.umich.edu/wiki/VerifyBamID>

`process.total.coverage.statistics`*Process total coverage statistics*

Description

Process reports generated by flagstat. Assumes reports for before and after off-target filtering have been written to the same file, with separating headers

Usage

```
process.total.coverage.statistics(project.directory)
```

Arguments

```
project.directory
```

Path to project directory. Each sample should have its own subdirectory

Value

data frame with extracted statistics

| | |
|----------------|-----------------------|
| read.all.calls | <i>read.all.calls</i> |
|----------------|-----------------------|

Description

Read all calls made with a certain caller

Usage

```
read.all.calls(sample.ids, caller = c("vardict", "mutect", "pgm"),
  project.directory, patient.ids = NULL, apply.filters = TRUE,
  variant.file.pattern = NULL)
```

Arguments

| | |
|----------------------|--|
| sample.ids | Vector giving sample IDs to process |
| caller | String indicating which caller was used |
| project.directory | Path to project directory |
| patient.ids | Optional vector giving patient ID (or other group) corresponding to each sample |
| apply.filters | Logical indicating whether filters specified in VariTAS options should be applied. Defaults to TRUE. ! |
| variant.file.pattern | Pattern indicating where the variant file can be found. Sample ID should be indicated by SAMPLE_ID |

Value

combined.variant.calls Data frame with variant calls from all patients

| | |
|----------------|-------------------------|
| read.ides.file | <i>Read iDES output</i> |
|----------------|-------------------------|

Description

Read output from iDES_step1.pl and return data frame

Usage

```
read.ides.file(filename)
```

Arguments

filename path to file

Value

ides.data data frame read from iDES output

read.variant.calls *Read variant calls from file and format for ease of downstream analyses.*

Description

Read variant calls from file and format for ease of downstream analyses.

Usage

```
read.variant.calls(variant.file, variant.caller)
```

Arguments

variant.file Path to variant file.
variant.caller String indicating which variant caller was used. Needed to format the headers.

Value

variant.calls Data frame of variant calls

read.yaml *read.yaml*

Description

Read a yaml file

Usage

```
read.yaml(file.name)
```

Arguments

file.name Path to yaml file

Value

list containing contents of yaml file

Examples

```
read.yaml(file.path(path.package('varitas'), 'config.yaml'))
```

| | |
|---------------|----------------------|
| run.alignment | <i>Run alignment</i> |
|---------------|----------------------|

Description

Run alignment

Usage

```
run.alignment(fastq.specification, output.directory, paired.end = FALSE,
  sample.directories = TRUE, output.subdirectory = FALSE,
  job.name.prefix = NULL, job.group = "alignment", quiet = FALSE,
  verify.options = !quiet)
```

Arguments

| | |
|---------------------|---|
| fastq.specification | Data frame detailing FASTQ files to be processed, typically from prepare.fastq.specification |
| output.directory | Path to project directory |
| paired.end | Logical indicating whether paired-end sequencing was performed |
| sample.directories | Logical indicating whether all sample files should be saved to sample-specific subdirectories (will be created) |
| output.subdirectory | If further nesting is required, name of subdirectory. If no further nesting, set to FALSE |
| job.name.prefix | Prefix for job names on the cluster |
| job.group | Group job should be associated with on cluster |
| quiet | Logical indicating whether to print commands to screen rather than submit them |
| verify.options | Logical indicating whether to run verify.varitas.options |

Details

Runs alignment (and related processing steps) on each sample.

Value

None

Examples

```
run.alignment(
  fastq.specification = data.frame(
    sample.id = c('1', '2'),
    reads = c('1-R1.fastq.gz', '2-R1.fastq.gz'),
    mates = c('1-R2.fastq.gz', '2-R2.fastq.gz'),
    patient.id = c('P1', 'P1'),
    tissue = c('tumour', 'normal')
  ),
  output.directory = '.',
  quiet = TRUE,
  paired.end = TRUE
)
```

run.alignment.sample *Run alignment for a single sample*

Description

Run alignment for a single sample

Usage

```
run.alignment.sample(fastq.files, sample.id, output.directory = NULL,
  output.filename = NULL, code.directory = NULL,
  log.directory = NULL, config.file = NULL, job.dependencies = NULL,
  job.name = NULL, job.group = NULL, quiet = FALSE,
  verify.options = !quiet)
```

Arguments

| | |
|------------------|--|
| fastq.files | Paths to FASTQ files (one file if single-end reads, two files if paired-end) |
| sample.id | Sample ID for labelling |
| output.directory | Path to output directory |
| output.filename | Name of resulting VCF file (defaults to SAMPLE_ID.vcf) |
| code.directory | Path to directory where code should be stored |
| log.directory | Path to directory where log files should be stored |
| config.file | Path to config file |
| job.dependencies | Vector with names of job dependencies |
| job.name | Name of job to be submitted |
| job.group | Group job should belong to |

quiet Logical indicating whether to print command to screen rather than submit it to the system. Defaults to false, useful for debugging.

verify.options Logical indicating whether to run verify.varitas.options

run.all.scripts *Run all the generated bash scripts without HPC commands*

Description

Run all the scripts generated by previous parts of the pipeline, without using HPC commands

Usage

```
run.all.scripts(output.directory, stages.to.run = c("alignment", "qc",
  "calling", "annotation", "merging"), variant.callers = NULL,
  quiet = FALSE)
```

Arguments

output.directory Main directory where all files should be saved

stages.to.run A character vector of all stages that need running

variant.callers A character vector of variant callers to run

quiet Logical indicating whether to print commands to screen rather than submit jobs. Defaults to FALSE, can be useful to set to TRUE for testing.

Value

None

run.annotation *Run annotation on a set of VCF files*

Description

Takes a data frame with paths to VCF files, and runs ANNOVAR annotation on each file. To allow for smooth connections with downstream pipeline steps, the function returns a variant specification data frame that can be used as input to merging steps.

Usage

```
run.annotation(vcf.specification, output.directory = NULL,
  job.name.prefix = NULL, job.group = NULL, quiet = FALSE,
  verify.options = !quiet)
```

Arguments

| | |
|-------------------|---|
| vcf.specification | Data frame detailing VCF files to be processed, from prepare.vcf.specification. |
| output.directory | Path to folder where code and log files should be stored in their respective sub-directories. If not supplied, code and log files will be stored in the directory with each VCF file. |
| job.name.prefix | Prefix to be added before VCF name in job name. Defaults to 'annotate', but should be changed if running multiple callers to avoid |
| job.group | Group job should be associated with on cluster |
| quiet | Logical indicating whether to print commands to screen rather than submit them |
| verify.options | Logical indicating whether to run verify.varitas.options |

Value

Data frame with details of variant files

Examples

```
run.annotation(
  data.frame(
    sample.id = c('a', 'b'),
    vcf = c('a.vcf', 'b.vcf'),
    caller = c('mutect', 'mutect')
  ),
  output.directory = '.',
  quiet = TRUE
)
```

| | |
|-----------------|----------------------------------|
| run.annovar.vcf | <i>Run ANNOVAR on a VCF file</i> |
|-----------------|----------------------------------|

Description

Run ANNOVAR on a VCF file

Usage

```
run.annovar.vcf(vcf.file, output.directory = NULL,
  output.filename = NULL, code.directory = NULL,
  log.directory = NULL, config.file = NULL, job.dependencies = NULL,
  job.group = NULL, job.name = NULL, isis = FALSE, quiet = FALSE,
  verify.options = !quiet)
```

Arguments

| | |
|------------------|---|
| vcf.file | Path to VCF file |
| output.directory | Path to output directory |
| output.filename | Name of resulting VCF file (defaults to SAMPLE_ID.vcf) |
| code.directory | Path to directory where code should be stored |
| log.directory | Path to directory where log files should be stored |
| config.file | Path to config file |
| job.dependencies | Vector with names of job dependencies |
| job.group | Group job should belong to |
| job.name | Name of job to be submitted |
| isis | Logical indicating whether VCF files are from the isis (MiniSeq) variant caller |
| quiet | Logical indicating whether to print command to screen rather than submit it to the system. Defaults to false, useful for debugging. |
| verify.options | Logical indicating whether to run verify.varitas.options |

Value

None

| | |
|-------------------|---|
| run.filtering.txt | <i>Run filtering on an ANNOVAR-annotated txt file</i> |
|-------------------|---|

Description

Run filtering on an ANNOVAR-annotated txt file

Usage

```
run.filtering.txt(variant.file, caller = c("consensus", "vardict",
    "ides", "mutect"), output.directory = NULL, output.filename = NULL,
    code.directory = NULL, log.directory = NULL, config.file = NULL,
    job.dependencies = NULL, job.group = NULL, quiet = FALSE)
```

Arguments

| | |
|------------------|---|
| variant.file | Path to variant file |
| caller | String giving variant caller that was used (affects which filters were applied. |
| output.directory | Path to output directory |

| | |
|------------------|---|
| output.filename | Name of resulting VCF file (defaults to SAMPLE_ID.vcf) |
| code.directory | Path to directory where code should be stored |
| log.directory | Path to directory where log files should be stored |
| config.file | Path to config file |
| job.dependencies | Vector with names of job dependencies |
| job.group | Group job should belong to |
| quiet | Logical indicating whether to print command to screen rather than submit it to the system. Defaults to false, useful for debugging. |

run.ides

Run iDES

Description

Run iDES

Usage

```
run.ides(project.directory, sample.id.pattern = "_S\\d+$",
         sample.ids = NULL, job.dependencies = NULL)
```

Arguments

| | |
|-------------------|-----------------------------------|
| project.directory | Directory containing files |
| sample.id.pattern | Regex pattern to match sample IDs |
| sample.ids | Vector of sample IDs |
| job.dependencies | Vector of job dependencies |

Details

Run iDES step 1 on each sample, to tally up calls by strand. Files are output to a the sample subdirectory

Value

None

Note

Deprecated function for running iDES. Follows previous development package without specification data frames

References

<https://cappseq.stanford.edu/ides/>

| | |
|-------------------|--------------------------------|
| run.lofreq.sample | <i>Run LoFreq for a sample</i> |
|-------------------|--------------------------------|

Description

Run LoFreq for a sample

Usage

```
run.lofreq.sample(tumour.bam, sample.id, paired, normal.bam = NULL,
  output.directory = NULL, output.filename = NULL,
  code.directory = NULL, log.directory = NULL, config.file = NULL,
  job.dependencies = NULL, quiet = FALSE, job.name = NULL,
  verify.options = !quiet, job.group = NULL)
```

Arguments

| | |
|------------------|---|
| tumour.bam | Path to tumour sample BAM file. |
| sample.id | Sample ID for labelling |
| paired | Logical indicating whether to do variant calling with a matched normal. |
| normal.bam | Path to normal BAM file if paired = TRUE |
| output.directory | Path to output directory |
| output.filename | Name of resulting VCF file (defaults to SAMPLE_ID.vcf) |
| code.directory | Path to directory where code should be stored |
| log.directory | Path to directory where log files should be stored |
| config.file | Path to config file |
| job.dependencies | Vector with names of job dependencies |
| quiet | Logical indicating whether to print command to screen rather than submit it to the system. Defaults to false, useful for debugging. |
| job.name | Name of job to be submitted |
| verify.options | Logical indicating whether to run verify.varitas.options |
| job.group | Group job should belong to |

| | |
|-----------------|------------------------------|
| run.muse.sample | <i>Run MuSE for a sample</i> |
|-----------------|------------------------------|

Description

Run MuSE for a sample

Usage

```
run.muse.sample(tumour.bam, sample.id, paired, normal.bam = NULL,  
  output.directory = NULL, output.filename = NULL,  
  code.directory = NULL, log.directory = NULL, config.file = NULL,  
  job.dependencies = NULL, quiet = FALSE, job.name = NULL,  
  verify.options = !quiet, job.group = NULL)
```

Arguments

| | |
|------------------|---|
| tumour.bam | Path to tumour sample BAM file. |
| sample.id | Sample ID for labelling |
| paired | Logical indicating whether to do variant calling with a matched normal. |
| normal.bam | Path to normal BAM file if paired = TRUE |
| output.directory | Path to output directory |
| output.filename | Name of resulting VCF file (defaults to SAMPLE_ID.vcf) |
| code.directory | Path to directory where code should be stored |
| log.directory | Path to directory where log files should be stored |
| config.file | Path to config file |
| job.dependencies | Vector with names of job dependencies |
| quiet | Logical indicating whether to print command to screen rather than submit it to the system. Defaults to false, useful for debugging. |
| job.name | Name of job to be submitted |
| verify.options | Logical indicating whether to run verify.varitas.options |
| job.group | Group job should belong to |

run.mutect.sample *Run MuTect for a sample*

Description

Run MuTect for a sample

Usage

```
run.mutect.sample(tumour.bam, sample.id, paired, normal.bam = NULL,
  output.directory = NULL, output.filename = NULL,
  code.directory = NULL, log.directory = NULL, config.file = NULL,
  job.dependencies = NULL, quiet = FALSE, job.name = NULL,
  verify.options = !quiet, job.group = NULL)
```

Arguments

| | |
|------------------|---|
| tumour.bam | Path to tumour sample BAM file. |
| sample.id | Sample ID for labelling |
| paired | Logical indicating whether to do variant calling with a matched normal. |
| normal.bam | Path to normal BAM file if paired = TRUE |
| output.directory | Path to output directory |
| output.filename | Name of resulting VCF file (defaults to SAMPLE_ID.vcf) |
| code.directory | Path to directory where code should be stored |
| log.directory | Path to directory where log files should be stored |
| config.file | Path to config file |
| job.dependencies | Vector with names of job dependencies |
| quiet | Logical indicating whether to print command to screen rather than submit it to the system. Defaults to false, useful for debugging. |
| job.name | Name of job to be submitted |
| verify.options | Logical indicating whether to run verify.varitas.options |
| job.group | Group job should belong to |

run.post.processing *run.post.processing*

Description

Submit post-processing job to the cluster with appropriate job dependencies

Usage

```
run.post.processing(variant.specification, output.directory,  
  code.directory = NULL, log.directory = NULL, config.file = NULL,  
  job.name.prefix = NULL, quiet = FALSE, email = NULL,  
  verify.options = !quiet)
```

Arguments

| | |
|-----------------------|---|
| variant.specification | Data frame specifying files to be processed |
| output.directory | Path to directory where output should be saved |
| code.directory | Directory where code should be saved |
| log.directory | Directory where log files should be saved |
| config.file | Path to config file |
| job.name.prefix | Prefix for job names on the cluster |
| quiet | Logical indicating whether to print commands to screen rather than submit the job |
| email | Email address that should be notified when job finishes. If NULL or FALSE, no email is sent |
| verify.options | Logical indicating whether <code>verify.varitas.options()</code> should be run. |

Value

None

Examples

```
run.post.processing(  
  variant.specification = data.frame(  
    sample.id = c('a', 'b'),  
    vcf = c('a.vcf', 'b.vcf'),  
    caller = c('mutect', 'mutect'),  
    job.dependency = c('example1', 'example2')  
  ),  
  output.directory = '.',  
  quiet = TRUE  
)
```

| | |
|---------------|---|
| run.target.qc | <i>Perform sample QC by looking at target coverage.</i> |
|---------------|---|

Description

Perform sample QC by looking at target coverage.

Usage

```
run.target.qc(bam.specification, project.directory,  
             sample.directories = TRUE, paired = FALSE,  
             output.subdirectory = FALSE, quiet = FALSE, job.name.prefix = NULL,  
             verify.options = FALSE, job.group = "target_qc")
```

Arguments

| | |
|---------------------|--|
| bam.specification | Data frame containing details of BAM files to be processed, typically from prepare.bam.specification. |
| project.directory | Path to project directory where code and log files should be saved |
| sample.directories | Logical indicating whether output for each sample should be put in its own directory (within output.directory) |
| paired | Logical indicating whether the analysis is paired. This does not affect QC directly, but means normal samples get nested |
| output.subdirectory | If further nesting is required, name of subdirectory. If no further nesting, set to FALSE |
| quiet | Logical indicating whether to print commands to screen rather than submit the job |
| job.name.prefix | Prefix for job names on the cluster |
| verify.options | Logical indicating whether to run verify.varitas.options |
| job.group | Group job should be associated with on cluster |

run.target.qc.sample *Get ontarget reads and run coverage quality control*

Description

Get ontarget reads and run coverage quality control

Usage

```
run.target.qc.sample(bam.file, sample.id, output.directory = NULL,
  code.directory = NULL, log.directory = NULL, config.file = NULL,
  job.dependencies = NULL, job.name = NULL, job.group = NULL,
  quiet = FALSE)
```

Arguments

| | |
|------------------|---|
| bam.file | Path to BAM file |
| sample.id | Sample ID for labelling |
| output.directory | Path to output directory |
| code.directory | Path to directory where code should be stored |
| log.directory | Path to directory where log files should be stored |
| config.file | Path to config file |
| job.dependencies | Vector with names of job dependencies |
| job.name | Name of job to be submitted |
| job.group | Group job should belong to |
| quiet | Logical indicating whether to print command to screen rather than submit it to the system. Defaults to false, useful for debugging. |

run vardict.sample *run.vardict.sample*

Description

Run VarDict on a sample. Idea: have a low-level function that simply submits job to Perl, after BAM paths have been found. and output paths already have been decided upon

Usage

```
run.vardict.sample(tumour.bam, sample.id, paired, proton = FALSE,
  normal.bam = NULL, output.directory = NULL, output.filename = NULL,
  code.directory = NULL, log.directory = NULL, config.file = NULL,
  job.dependencies = NULL, job.name = NULL, job.group = NULL,
  quiet = FALSE, verify.options = !quiet)
```

Arguments

| | |
|-------------------------------|---|
| <code>tumour.bam</code> | Path to tumour sample BAM file. |
| <code>sample.id</code> | Sample ID for labelling |
| <code>paired</code> | Logical indicating whether to do variant calling with a matched normal. |
| <code>proton</code> | Logical indicating whether the data was generated by proton sequencing. Defaults to False (i.e. Illumina) |
| <code>normal.bam</code> | Path to normal BAM file if <code>paired = TRUE</code> |
| <code>output.directory</code> | Path to output directory |
| <code>output.filename</code> | Name of resulting VCF file (defaults to <code>SAMPLE_ID.vcf</code>) |
| <code>code.directory</code> | Path to directory where code should be stored |
| <code>log.directory</code> | Path to directory where log files should be stored |
| <code>config.file</code> | Path to config file |
| <code>job.dependencies</code> | Vector with names of job dependencies |
| <code>job.name</code> | Name of job to be submitted |
| <code>job.group</code> | Group job should belong to |
| <code>quiet</code> | Logical indicating whether to print command to screen rather than submit it to the system. Defaults to false, useful for debugging. |
| <code>verify.options</code> | Logical indicating whether to run <code>verify.varitas.options</code> |

`run.variant.calling` *run.variant.calling*

Description

Run variant calling for all samples

Usage

```
run.variant.calling(bam.specification, output.directory,
  variant callers = c("vardict", "mutect", "varscan", "lofreq", "muse"),
  paired = TRUE, proton = FALSE, sample.directories = TRUE,
  job.name.prefix = NULL, quiet = FALSE, verify.options = !quiet)
```

Arguments

| | |
|--------------------|--|
| bam.specification | Data frame containing details of BAM files to be processed, typically from prepare.bam.specification. |
| output.directory | Path to directory where output should be saved |
| variant callers | Character vector of variant callers to be used |
| paired | Logical indicating whether to do variant calling with a matched normal |
| proton | Logical indicating whether data was generated by proton sequencing (ignored if running MuTect) |
| sample.directories | Logical indicating whether output for each sample should be put in its own directory (within output.directory) |
| job.name.prefix | Prefix for job names on the cluster |
| quiet | Logical indicating whether to print commands to screen rather than submit the job |
| verify.options | Logical indicating whether to run verify.varitas.options |

Details

Run VarDict on each sample, and annotate the results with ANNOVAR. Files are output to a vardict/ subdirectory within each sample directory.

Value

None

Examples

```
run.variant.calling(  
  data.frame(sample.id = c('Z', 'Y'), tumour.bam = c('Z.bam', 'Y.bam')),  
  output.directory = '.',  
  variant.caller = c('lofreq', 'mutect'),  
  quiet = TRUE,  
  paired = FALSE  
)
```

run.varitas.pipeline *Run VariTAS pipeline in full.*

Description

Run all steps in VariTAS processing pipeline, with appropriate dependencies.

Usage

```
run.varitas.pipeline(file.details, output.directory, run.name = NULL,
  start.stage = c("alignment", "qc", "calling", "annotation", "merging"),
  variant callers = NULL, proton = FALSE, quiet = FALSE,
  email = NULL, verify.options = !quiet,
  save.specification.files = !quiet)
```

Arguments

| | |
|--------------------------|--|
| file.details | Data frame containing details of files to be used during first processing step. Depending on what you want to be the first step in the pipeline, this can either be FASTQ files, BAM files, VCF files, or variant (txt) files. |
| output.directory | Main directory where all files should be saved |
| run.name | Name of pipeline run. Will be added as a prefix to all LSF jobs. |
| start.stage | String indicating which stage pipeline should start at. If starting at a later stage of the pipeline, appropriate input files must be provided. For example, if starting with annotation, VCF files with variant calls must be provided. |
| variant.callers | Vector specifying which variant callers should be run. |
| proton | Logical indicating if data was generated by proton sequencing. Used to set base quality thresholds in variant calling steps. |
| quiet | Logical indicating whether to print commands to screen rather than submit jobs. Defaults to FALSE, can be useful to set to TRUE for testing. |
| email | Email address that should be notified when pipeline finishes. If NULL or FALSE, no email is sent. |
| verify.options | Logical indicating whether to run verify.varitas.options |
| save.specification.files | Logical indicating if specification files should be saved to project directory |

Value

None

Examples

```
run.varitas.pipeline(
  file.details = data.frame(
    sample.id = c('1', '2'),
    reads = c('1-R1.fastq.gz', '2-R1.fastq.gz'),
    mates = c('1-R2.fastq.gz', '2-R2.fastq.gz'),
    patient.id = c('P1', 'P1'),
    tissue = c('tumour', 'normal')
  ),
  output.directory = '.',
  quiet = TRUE,
  run.name = "Test",
  variant callers = c('mutect', 'varscan')
)
```

```
run.varitas.pipeline.hybrid
      run.varitas.pipeline.hybrid
```

Description

Run VariTAS pipeline starting from both VCF files and BAM/ FASTQ files. Useful for processing data from the Ion PGM or MiniSeq where variant calling has been done on the machine, but you are interested in running more variant callers.

Usage

```
run.varitas.pipeline.hybrid(vcf.specification, output.directory,
  run.name = NULL, fastq.specification = NULL,
  bam.specification = NULL, variant.callers = c("mutect", "vardict",
  "varscan", "lofreq", "muse"), proton = FALSE, quiet = FALSE,
  email = NULL, verify.options = !quiet,
  save.specification.files = !quiet)
```

Arguments

| | |
|---------------------|---|
| vcf.specification | Data frame containing details of vcf files to be processed. Must contain columns sample.id, vcf, and caller |
| output.directory | Main directory where all files should be saved |
| run.name | Name of pipeline run. Will be added as a prefix to all LSF jobs. |
| fastq.specification | Data frame containing details of FASTQ files to be processed |
| bam.specification | Data frame containing details of BAM files to be processed |

| | |
|--------------------------|--|
| variant.callers | Vector specifying which variant callers should be run. |
| proton | Logical indicating if data was generated by proton sequencing. Used to set base quality thresholds in variant calling steps. |
| quiet | Logical indicating whether to print commands to screen rather than submit jobs. Defaults to FALSE, can be useful to set to TRUE for testing. |
| email | Email address that should be notified when pipeline finishes. If NULL or FALSE, no email is sent. |
| verify.options | Logical indicating whether to run verify.varitas.options |
| save.specification.files | Logical indicating if specification files should be saved to project directory |

Value

None

Examples

```
run.varscan.pipeline.hybrid(
  bam.specification = data.frame(sample.id = c('Z', 'Y'), tumour.bam = c('Z.bam', 'Y.bam')),
  vcf.specification = data.frame(
    sample.id = c('a', 'b'),
    vcf = c('a.vcf', 'b.vcf'),
    caller = c('pgm', 'pgm')
  ),
  output.directory = '.',
  quiet = TRUE,
  run.name = "Test",
  variant.callers = c('mutect', 'varscan')
)
```

run.varscan.sample *Run VarScan for a sample*

Description

Run VarScan for a sample

Usage

```
run.varscan.sample(tumour.bam, sample.id, paired, normal.bam = NULL,
  output.directory = NULL, output.filename = NULL,
  code.directory = NULL, log.directory = NULL, config.file = NULL,
  job.dependencies = NULL, quiet = FALSE, job.name = NULL,
  verify.options = !quiet, job.group = NULL)
```

Arguments

| | |
|------------------|---|
| tumour.bam | Path to tumour sample BAM file. |
| sample.id | Sample ID for labelling |
| paired | Logical indicating whether to do variant calling with a matched normal. |
| normal.bam | Path to normal BAM file if paired = TRUE |
| output.directory | Path to output directory |
| output.filename | Name of resulting VCF file (defaults to SAMPLE_ID.vcf) |
| code.directory | Path to directory where code should be stored |
| log.directory | Path to directory where log files should be stored |
| config.file | Path to config file |
| job.dependencies | Vector with names of job dependencies |
| quiet | Logical indicating whether to print command to screen rather than submit it to the system. Defaults to false, useful for debugging. |
| job.name | Name of job to be submitted |
| verify.options | Logical indicating whether to run verify.varitas.options |
| job.group | Group job should belong to |

| | |
|-------------|--------------------|
| save.config | <i>save.config</i> |
|-------------|--------------------|

Description

Save current varitas config options to a temporary file, and return filename.

Usage

```
save.config(output.file = NULL)
```

Arguments

| | |
|-------------|--|
| output.file | Path to output file. If NULL (default), the config file will be saved as a temporary file. |
|-------------|--|

Value

Path to config file

save.coverage.excel *Save coverage statistics to multi-worksheet Excel file.*

Description

Save coverage statistics to multi-worksheet Excel file.

Usage

```
save.coverage.excel(project.directory, file.name, overwrite = TRUE)
```

Arguments

| | |
|-------------------|---|
| project.directory | Path to project directory |
| file.name | Name of output file |
| overwrite | Logical indicating whether to overwrite existing file if it exists. |

Value

None

save.variants.excel *Save variants to Excel.*

Description

Makes an Excel workbook with variant calls. If filters are provided, these will be saved to an additional worksheet within the same file.

Usage

```
save.variants.excel(variants, file.name, filters = NULL,  
  overwrite = TRUE)
```

Arguments

| | |
|-----------|---|
| variants | Data frame containing variants |
| file.name | Name of output file |
| filters | Optional list of filters to be saved |
| overwrite | Logical indicating whether to overwrite exiting file if it exists. Defaults to TRUE for consistency with other R functions. |

set.varitas.options *Set options for varitas pipeline.*

Description

Set or overwrite options for the VariTAS pipeline. Nested options should be separated by a dot. For example, to update the reference genome for grch38, use reference_genome.grch38

Usage

```
set.varitas.options(...)
```

Arguments

```
...                    options to set
```

Value

None

Examples

```
## Not run:  
set.varitas.options(reference_build = 'grch38');  
set.varitas.options(  
  filters.mutect.min_normal_depth = 10,  
  filters.vardict.min_normal_depth = 10  
);  
  
## End(Not run)
```

split.on.column *split.on.column*

Description

Split data frame on a concatenated column.

Usage

```
## S3 method for class 'on.column'  
split(dat, column, split.character)
```

Arguments

| | |
|-----------------|---|
| dat | Data frame to be processed |
| column | Name of column to split on |
| split.character | Pattern giving character to split column on |

Value

Data frame after splitting on column

| | |
|---------|----------------|
| sum.dp4 | <i>sum.dp4</i> |
|---------|----------------|

Description

Simply calculates the depth of coverage of the variant allele given a string of DP4 values

Usage

```
## S3 method for class 'dp4'
sum(dp4.str)
```

Arguments

| | |
|---------|--|
| dp4.str | String of DP4 values in the form "1234,1234,1234,1234" |
|---------|--|

| | |
|-----------|-----------------------|
| system.ls | <i>Run ls command</i> |
|-----------|-----------------------|

Description

Runs ls command on system. This is a workaround since list.files can not match patterns based on subdirectory structure.

Usage

```
system.ls(pattern = "", directory = "", error = FALSE)
```

Arguments

| | |
|-----------|--|
| pattern | pattern to match files |
| directory | base directory command should be run from |
| error | logical indicating whether to throw an error if no matching founds found. Defaults to False. |

Value

paths returned by ls command

| | |
|--------------|---------------------|
| tabular.mean | <i>tabular.mean</i> |
|--------------|---------------------|

Description

Calculate the mean of data in tabular format

Usage

```
tabular.mean(values, frequencies, ...)
```

Arguments

| | |
|-------------|---------------------------------------|
| values | vector of values |
| frequencies | frequency corresponding to each value |
| ... | Additional parameters passed to sum |

Value

calculated mean

| | |
|----------------|-----------------------|
| tabular.median | <i>tabular.median</i> |
|----------------|-----------------------|

Description

Calculate the median of data in tabular format

Usage

```
tabular.median(values, frequencies, ...)
```

Arguments

| | |
|-------------|---------------------------------------|
| values | Vector of values |
| frequencies | Frequency corresponding to each value |
| ... | Additional parameters passed to sum |

Value

calculated median

trinucleotide.barplot *Make barplot of trinucleotide substitutions*

Description

Make barplot of trinucleotide substitutions

Usage

```
trinucleotide.barplot(variants, file.name)
```

Arguments

| | |
|-----------|--------------------------|
| variants | Data frame with variants |
| file.name | Name of output file |

Value

None

variant.recurrence.barplot
Make barplot of variants per caller

Description

Make barplot of variants per caller

Usage

```
variant.recurrence.barplot(variants, file.name)
```

Arguments

| | |
|-----------|--------------------------|
| variants | Data frame with variants |
| file.name | Name of output file |

Value

None

`variants.caller.barplot`*Make barplot of variants per caller*

Description

Make barplot of variants per caller

Usage

```
variants.caller.barplot(variants, file.name, group.by = NULL)
```

Arguments

| | |
|------------------------|--|
| <code>variants</code> | Data frame with variants |
| <code>file.name</code> | Name of output file |
| <code>group.by</code> | Optional grouping variable for barplot |

Value

None

`variants.sample.barplot`*Make barplot of variants per sample*

Description

Make barplot of variants per sample

Usage

```
variants.sample.barplot(variants, file.name)
```

Arguments

| | |
|------------------------|--------------------------|
| <code>variants</code> | Data frame with variants |
| <code>file.name</code> | Name of output file |

Value

None

```
verify.bam.specification
```

Check that sample specification data frame matches expected format, and that all files exist

Description

Check that sample specification data frame matches expected format, and that all files exist

Usage

```
verify.bam.specification(bam.specification)
```

Arguments

```
bam.specification
```

Data frame containing columns sample.id and tumour.bam, and optionally a column normal.bam.

Value

None

```
verify.bwa.index      verify.bwa.index
```

Description

Verify that bwa index files exist for a fasta file

Usage

```
verify.bwa.index(fasta.file, error = FALSE)
```

Arguments

```
fasta.file      Fasta file to check
```

```
error           Logical indicating whether to throw an (informative) error if verification fails
```

Value

index.files.exist Logical indicating if bwa index files were found (only returned if error set to FALSE)

| | |
|--------------------|---------------------------|
| verify.fasta.index | <i>verify.fasta.index</i> |
|--------------------|---------------------------|

Description

Verify that fasta index files exist for a given fasta file.

Usage

```
verify.fasta.index(fasta.file, error = FALSE)
```

Arguments

| | |
|------------|--|
| fasta.file | Fasta file to check |
| error | Logical indicating whether to throw an (informative) error if verification fails |

Value

faidx.exists Logical indicating if fasta index files were found (only returned if error set to FALSE)

| | |
|----------------------------|--|
| verify.fastq.specification | <i>Check that FASTQ specification data frame matches expected format, and that all files exist</i> |
|----------------------------|--|

Description

Check that FASTQ specification data frame matches expected format, and that all files exist

Usage

```
verify.fastq.specification(fastq.specification, paired.end = FALSE,
  files.ready = FALSE)
```

Arguments

| | |
|---------------------|--|
| fastq.specification | Data frame containing columns sample.id and reads, and optionally a column mates |
| paired.end | Logical indicating whether paired end reads are used |
| files.ready | Logical indicating if the files already exist on disk. If there are job dependencies, this should be set to FALSE. |

Value

None

```
verify.sequence.dictionary
    verify.sequence.dictionary
```

Description

Verify that sequence dictionary exists for a fasta file.

Usage

```
verify.sequence.dictionary(fasta.file, error = FALSE)
```

Arguments

| | |
|------------|--|
| fasta.file | Fasta file to check |
| error | Logical indicating whether to throw an (informative) error if verification fails |

Value

dict.exists Logical indicating if sequence dictionary files were found (only returned if error set to FALSE)

```
verify.varitas.options
    Check against common errors in the VariTAS options.
```

Description

Check against common errors in the VariTAS options before launching into pipeline

Usage

```
verify.varitas.options(stages.to.run = c("alignment", "qc", "calling",
    "annotation", "merging"), variant callers = c("mutect", "vardict",
    "ides", "varscan", "lofreq", "muse"), varitas.options = NULL)
```

Arguments

| | |
|-----------------|--|
| stages.to.run | Vector indicating which stages should be run. Defaults to all possible stages. If only running a subset of stages, only checks corresponding to the desired stages are run |
| variant.callers | Vector indicating which variant callers to run. Only used if calling is in stages.to.run. |
| varitas.options | Optional file path or list of VariTAS options. |

Value

None

`verify.vcf.specification`

verify.vcf.specification

Description

Verify that VCF specification data frame fits expected format

Usage

`verify.vcf.specification(vcf.specification)`

Arguments

`vcf.specification`

VCF specification data frame

Value

None

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