

# 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

**Suggests** testthat, knitr, rmarkdown, futile.logger

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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');
```

---

`get.vcf.chromosomes`    *get.vcf.chromosomes*

---

**Description**

Extract chromosomes from a VCF file.

**Usage**

```
get.vcf.chromosomes(vcf)
```

**Arguments**

`vcf`                    Path to VCF file

**Value**

Vector containing all chromosomes in VCF

---

`in.varitas.options`    *Check if a key is in VariTAS options*

---

**Description**

Check if a key is in VariTAS options

**Usage**

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

**Arguments**

`option.name`            String giving name of option (with different levels joined by `nesting.character`)  
`varitas.options`                    Ampliseq options as a list. If missing, they will be obtained from `get.varitas.options()`  
`nesting.character`                    String giving Regex pattern of nesting indication string. Defaults to `'\.'`

**Value**

`in.options` Boolean indicating if the option name exists in the current varitas options

---

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

*Run alignment*


---

**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	<i>Run iDES</i>
----------	-----------------

---

## 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      *Run LoFreq for a sample*

---

**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.varitas.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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