

# Package ‘HTSeqGenie’

October 17, 2024

**Imports** BiocGenerics (>= 0.2.0), S4Vectors (>= 0.9.25), IRanges (>= 1.21.39), GenomicRanges (>= 1.23.21), Rsamtools (>= 1.8.5), Biostrings (>= 2.24.1), palign, chipseq (>= 1.6.1), hwriter (>= 1.3.0), Cairo (>= 1.5.5), GenomicFeatures (>= 1.9.31), BiocParallel, parallel, tools, rtracklayer (>= 1.17.19), GenomicAlignments, VariantTools (>= 1.7.7), GenomeInfoDb, SummarizedExperiment, methods

**Maintainer** Jens Reeder <reeder.jens@gene.com>

**License** Artistic-2.0

**Title** A NGS analysis pipeline.

**Type** Package

**LazyLoad** yes

**Author** Gregoire Pau, Jens Reeder

**Description** Libraries to perform NGS analysis.

**Version** 4.34.0

**Depends** R (>= 3.0.0), gmapR (>= 1.8.0), ShortRead (>= 1.19.13), VariantAnnotation (>= 1.8.3)

**Suggests** TxDb.Hsapiens.UCSC.hg19.knownGene, LungCancerLines, org.Hs.eg.db, RUnit

**RoxygenNote** 5.0.1

**git\_url** <https://git.bioconductor.org/packages/HTSeqGenie>

**git\_branch** RELEASE\_3\_19

**git\_last\_commit** e25c9ee

**git\_last\_commit\_date** 2024-04-30

**Repository** Bioconductor 3.19

**Date/Publication** 2024-10-16

## Contents

alignReads . . . . .	4
alignReadsChunk . . . . .	5
analyzeVariants . . . . .	6
annotateVariants . . . . .	6
bamCountUniqueReads . . . . .	7
buildConfig . . . . .	7
buildGenomicFeaturesFromTxDb . . . . .	8
buildShortReadReports . . . . .	9
buildTallyParam . . . . .	9
buildTP53FastaGenome . . . . .	10
buildTP53GenomeTemplate . . . . .	10
calculateCoverage . . . . .	11
calculateTargetLengths . . . . .	11
callVariantsGATK . . . . .	12
checkConfig . . . . .	12
checkGATKJar . . . . .	13
checkPicardJar . . . . .	13
computeBamStats . . . . .	14
computeCoverage . . . . .	14
countFeatures . . . . .	15
countGenomicFeatures . . . . .	16
countGenomicFeaturesChunk . . . . .	16
createTmpDir . . . . .	17
detectAdapterContam . . . . .	17
detectQualityInFASTQFile . . . . .	18
detectRRNA . . . . .	18
excludeVariantsByRegions . . . . .	19
FastQStreamer.getReads . . . . .	20
FastQStreamer.init . . . . .	20
FastQStreamer.release . . . . .	21
filterByLength . . . . .	22
filterQuality . . . . .	22
findVariantFile . . . . .	23
gatk . . . . .	23
generateSingleGeneDETs . . . . .	24
getAdapterSeqs . . . . .	25
getBams . . . . .	25
getChunkDirs . . . . .	26
getConfig . . . . .	26
getConfig.integer . . . . .	27
getConfig.logical . . . . .	27
getConfig.numeric . . . . .	28
getConfig.vector . . . . .	28
getEndNumber . . . . .	29
getMemoryUsage . . . . .	29
getNumberOfReadsInFASTQFile . . . . .	30

getNumericVectorDataFromFile . . . . .	30
getObjectFilename . . . . .	31
getPackageFile . . . . .	31
getRandomAlignCutoff . . . . .	32
getRRNAIds . . . . .	32
getTabDataFromFile . . . . .	33
getTraceback . . . . .	33
hashCoverage . . . . .	34
hashVariants . . . . .	34
hashVector . . . . .	35
HTSeqGenie . . . . .	35
initDirs . . . . .	36
initLog . . . . .	37
initLogger . . . . .	37
initPipelineFromConfig . . . . .	38
initPipelineFromSaveDir . . . . .	38
isAboveQualityThresh . . . . .	39
isAdapter . . . . .	39
isConfig . . . . .	40
isFirstFragment . . . . .	40
isSparse . . . . .	41
listIterator.init . . . . .	42
listIterator.next . . . . .	42
loadConfig . . . . .	43
logdebug . . . . .	43
logerror . . . . .	44
loginfo . . . . .	44
logwarn . . . . .	45
makeDir . . . . .	45
makeRandomSreads . . . . .	46
markDuplicates . . . . .	46
markDups . . . . .	47
mergeAlignReads . . . . .	47
mergeCoverage . . . . .	48
mergeLanes . . . . .	49
mergePreprocessReads . . . . .	49
mergeSummaryAlignment . . . . .	50
parseDCF . . . . .	51
parseSummaries . . . . .	51
picard . . . . .	52
plotDF . . . . .	52
preprocessReads . . . . .	53
preprocessReadsChunk . . . . .	53
processChunks . . . . .	54
readInputFiles . . . . .	55
readRNASeqEnds . . . . .	55
realnIndels . . . . .	56
realnIndelsGATK . . . . .	56

relativeBarPlot . . . . .	57
removeChunkDir . . . . .	57
resource . . . . .	58
rpkm . . . . .	58
runAlignment . . . . .	59
runPipeline . . . . .	60
runPipelineConfig . . . . .	61
runPreprocessReads . . . . .	62
safe.yield . . . . .	62
safeExecute . . . . .	63
safeGetObject . . . . .	63
safeUnlink . . . . .	64
saveWithID . . . . .	65
sclapply . . . . .	65
setChunkDir . . . . .	66
setUpDirs . . . . .	67
setUpTestFramework . . . . .	67
statCountFeatures . . . . .	68
TP53GenomicFeatures . . . . .	68
traceMem . . . . .	69
trimReads . . . . .	69
trimTailsByQuality . . . . .	70
truncateReads . . . . .	70
tryKeepTraceback . . . . .	71
updateConfig . . . . .	71
vcfStat . . . . .	72
wrap.callVariants . . . . .	72
writeAudit . . . . .	73
writeConfig . . . . .	73
writeFastQFiles . . . . .	74
writeFeatureCountsHTML . . . . .	74
writeGenomicFeaturesReport . . . . .	75
writePreprocessAlignHTML . . . . .	75
writePreprocessAlignReport . . . . .	76
writeSummary . . . . .	77
writeVCF . . . . .	77
<b>Index</b>	<b>79</b>

---

alignReads

*Align reads against genome*


---

**Description**

Align reads against genome

**Usage**

```
alignReads()
```

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

<code>alignReadsChunk</code>	<i>Genomic alignment</i>
------------------------------	--------------------------

---

**Description**

Genomic alignment using gsnap.

**Usage**

```
alignReadsChunk(fp1, fp2 = NULL, save_dir = NULL)
```

**Arguments**

<code>fp1</code>	Path to FastQ file
<code>fp2</code>	Path to second FastQ file if paired end data, NULL if single ended
<code>save_dir</code>	Save directory

**Details**

Aligns reads in `fp1` and `fp2` to genome specified via global config variable `alignReads.genome`. Gsnap output is converted into BAM files and sorted + indexed.

**Value**

List of alignment files in BAM format

analyzeVariants      *Calculate and process Variants*

---

**Description**

Calculate and process Variants

**Usage**

```
analyzeVariants()
```

**Value**

Nothing

**Author(s)**

Jens Reeder

---

annotateVariants      *Annotate variants via vep*

---

**Description**

Annotate variants via vep

**Usage**

```
annotateVariants(vcf.file)
```

**Arguments**

vcf.file      A character vector pointing to a VCF (or gzipped VCF) file

**Value**

Path to a vcf file with variant annotations

**Author(s)**

Jens Reeder

---

`bamCountUniqueReads`      *Uniquely count number of reads in bam file*

---

**Description**

Uniquely count number of reads in bam file

**Usage**

`bamCountUniqueReads(bam)`

**Arguments**

`bam`                      Name of bam file

**Value**

number of reads

**Author(s)**

Jens Reeder

---

`buildConfig`              *Build a configuration file based on a list of parameters*

---

**Description**

Build a configuration file based on a list of parameters

**Usage**

`buildConfig(config_filename, ...)`

**Arguments**

`config_filename`              The path of a configuration filename.  
`...`                              A list of named value pairs.

**Value**

Nothing.

**Author(s)**

Gregoire Pau

**See Also**

runPipeline

---

buildGenomicFeaturesFromTxDb

*Build genomic features from a TxDb object*

---

**Description**

Build genomic features from a TxDb object

**Usage**

```
buildGenomicFeaturesFromTxDb(txdb)
```

**Arguments**

txdb            A TxDb object.

**Value**

A list named list of GRanges objects containing the biological entities to account for.

**Author(s)**

Gregoire Pau

**Examples**

```
## Not run:
library("TxDb.Hsapiens.UCSC.hg19.knownGene")
txdb <- TxDb.Hsapiens.UCSC.hg19.knownGene
genomic_features <- buildGenomicFeaturesFromTxDb(txdb)

## End(Not run)
```

---

`buildShortReadReports` *Build a ShortRead report*

---

**Description**

Build a ShortRead report

**Usage**

```
buildShortReadReports(save_dir, paired_ends)
```

**Arguments**

<code>save_dir</code>	Save directory of a pipeline run
<code>paired_ends</code>	A logical, indicating whether reads are paired

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

`buildTallyParam` *Build tally parameters*

---

**Description**

Build tally parameters

**Usage**

```
buildTallyParam()
```

**Value**

a VariantTallyParam object

**Author(s)**

Gregoire Pau

---

`buildTP53FastaGenome`    *buildTP53FastaGenome*

---

**Description**

create fasta genome file of TP53 genome

**Usage**

`buildTP53FastaGenome()`

**Value**

Path to tp53 genome directory

**Author(s)**

Jens Reeder

---

`buildTP53GenomeTemplate`  
*buildTP53GenomeTemplate*

---

**Description**

Create a tp53 config template

**Usage**

`buildTP53GenomeTemplate()`

**Value**

Path to tp53 template file

**Author(s)**

Jens Reeder

---

calculateCoverage	<i>Calculate read coverage</i>
-------------------	--------------------------------

---

**Description**

Calculate read coverage

**Usage**

```
calculateCoverage()
```

**Value**

Nothing

**Author(s)**

Jens Reeder

---

calculateTargetLengths	<i>Plot target length for paired end</i>
------------------------	--

---

**Description**

Calculate and plot a histogram of mapped target lengths after trimmimg of trim/2 of the data points at the lower and upper end of the distribution

**Usage**

```
calculateTargetLengths(bamfile, save_dir, trim = 0.4)
```

**Arguments**

bamfile	Path to a bam file
save_dir	Path to a pipeline results dir
trim	Amount of data to be trimmed at the edges

**Value**

Target length table and writes two files in save\_dir/reports/images/TargetLengths.[pdf/png]"

**Author(s)**

Jens Reeder, Melanie Huntley

---

callVariantsGATK	<i>Variant calling via GATK</i>
------------------	---------------------------------

---

**Description**

Call variants via GATK using the pipeline framework. Requires a GATK compatible genome with a name matching the alignment genome to be installed in 'path.gatk\_genome'

**Usage**

```
callVariantsGATK(bam.file)
```

**Arguments**

bam.file	Path to bam.file
----------	------------------

**Value**

Path to variant file

**Author(s)**

Jens Reeder

---

checkConfig	<i>Check configuration</i>
-------------	----------------------------

---

**Description**

Performs all configuration checks

**Usage**

```
checkConfig()
```

**Value**

Nothing. Individual checks will throw error instead.

---

checkGATKJar	<i>Check for the GATK jar file</i>
--------------	------------------------------------

---

**Description**

Check for the GATK jar file

**Usage**

```
checkGATKJar(path = getOption("gatk.path"))
```

**Arguments**

path	Path to the GATK jar file
------	---------------------------

**Value**

TRUE if tool can be called, FALSE otherwise

---

checkPicardJar	<i>checkPicardJar</i>
----------------	-----------------------

---

**Description**

Check for a jar file from picard tools

**Usage**

```
checkPicardJar(toolname, path = getOption("picard.path"))
```

**Arguments**

toolname	Name of the Picard Tool, e.g. MarkDuplicates
path	Path to folder containing picard jars

**Details**

Call a tool from picard and see if it responds.

**Value**

TRUE if tool can be called, FALSE otherwise

**Author(s)**

Jens Reeder

---

computeBamStats      *Compute record statistics from a bam file*

---

**Description**

Compute record statistics from a bam file

**Usage**

```
computeBamStats(bam)
```

**Arguments**

bam                    A character string containing an existing bam file

**Details**

The statistics are additive over chunks/lanes.

**Value**

A numeric vector

**Author(s)**

Gregoire Pau

---

computeCoverage      *Compute the coverage vector given a bamfile*

---

**Description**

Compute the coverage vector given a bamfile

**Usage**

```
computeCoverage(bamfile, extendReads = FALSE, paired_ends = FALSE,  
                  fragmentLength = NULL, maxFragmentLength = NULL)
```

**Arguments**

bamfile	A character string indicating the path of bam file
extendReads	A logical, indicating whether reads should be extended
paired_ends	A logical, indicating whether reads are paired
fragmentLength	An integer, indicating the new size of reads when extendReads is TRUE and paired_ends is FALSE. If NULL, read size is estimated using estimate.mean.fraglen from the chipseq package.
maxFragmentLength	An optional integer, specifying the maximal size of fragments. Longer fragments will be disregarded when computing coverage.

**Value**

A SimpleRleList object containing the coverage

**Author(s)**

Gregoire Pau

---

countFeatures	<i>Count RNA-Seq Pipeline Genomic Features</i>
---------------	--

---

**Description**

Given GRanges, counts number of hits by gene, exon, intergenic, etc

**Usage**

```
countFeatures(reads, features)
```

**Arguments**

reads	GRangesList object of interval, usually where reads aligned
features	A list of genome annotations as GRangesList

**Details**

Given a GRanges object, this function performs an overlap against a previously created set of genomic regions. These genomic regions include genes, coding portions of genes (CDS), exons, intergenic regions, and exon groups (which contain two or more exons)

**Value**

A list of counts by feature

**Author(s)**

Cory Barr

countGenomicFeatures *Count overlaps with genomic features*

---

**Description**

Count overlaps with genomic features

**Usage**

```
countGenomicFeatures()
```

**Value**

Nothing

**Author(s)**

Gegoire Pau

---

countGenomicFeaturesChunk  
*Count reads by genomic Feature*

---

**Description**

Count reads by genomic Feature

**Usage**

```
countGenomicFeaturesChunk(save_dir, genomic_features)
```

**Arguments**

save\_dir            Path to a pipeline run's save dir  
genomic\_features    A list of genomic features to tally

**Details**

given a BAM-file output from gsnap (with the MD tag), count hits to exons, genes, ncRNAs, etc. and quantify miRNA/ncRNA contaminatio

**Value**

Nothing

**Author(s)**

Cory Barr

---

`createTmpDir`      *Create a random directory with prefix in R temp dir*

---

**Description**

Especially for testing code it is very helpful to have a temp directory with a defined prefix, so one knows which test produced which directory.

**Usage**

```
createTmpDir(prefix = NULL, dir = tempdir())
```

**Arguments**

<code>prefix</code>	A string that will precede the directory name
<code>dir</code>	Directory where the random dir will be created under. Defaults to tempdir()

**Value**

Name of temporary directory

---

`detectAdapterContam`      *Detect sequencing adapter contamination*

---

**Description**

For each read or pair of read, search for specific Illumina adapter sequences in the read. Flag if at least one read has significant overlap with adapter.

**Usage**

```
detectAdapterContam(lreads, save_dir = NULL)
```

**Arguments**

<code>lreads</code>	List of reads as ShortRead objects
<code>save_dir</code>	Save directory of a pipeline run

**Value**

Boolean vector indicating vector contamination for each read

---

detectQualityInFASTQFile

*Detect quality protocol from a FASTQ file*

---

### Description

Detect quality protocol from a FASTQ file

### Usage

```
detectQualityInFASTQFile(filename, nreads = 5000)
```

### Arguments

filename	Path to a FASTQ or gzipped-FASTQ file
nreads	Number of reads to test quality on. Default is 5000.

### Value

A character vector containing the compatible qualities. NULL if none.

### Author(s)

Jens Reeder

---

detectRRNA

*Detect rRNA Contamination in Reads*

---

### Description

Returns a named vector indicating if a read ID has rRNA contamination or not

### Usage

```
detectRRNA(lreads, remove_tmp_dir = TRUE, save_dir = NULL)
```

### Arguments

lreads	A list of ShortReadQ objects
remove_tmp_dir	boolean indicating whether or not to delete temp directory of gsnap results
save_dir	Save directory

### Details

Given a genome and fastq data, each read in the fastq data is aligned against the rRNA sequences for that genome

**Value**

a named logical vector indicating if a read has rRNA contamination

**Author(s)**

Cory Barr

---

`excludeVariantsByRegions`

*Filter variants by regions*

---

**Description**

Filter variants by regions

**Usage**

```
excludeVariantsByRegions(variants, mask)
```

**Arguments**

<code>variants</code>	Variants as <code>Vranges</code> , <code>GRanges</code> or VCF object
<code>mask</code>	region to mask, given as <code>GRanges</code>

**Details**

This function can be used to filter variants in a given region, e.g. low complexity and repeat regions

**Value**

The filtered variants

**Author(s)**

Jens Reeder

FastQStreamer.getReads

*Get FastQ reads from the FastQ streamer*

---

### **Description**

Get FastQ reads from the FastQ streamer

### **Usage**

```
FastQStreamer.getReads()
```

### **Value**

A list of ShortRead object containing reads. NULL if there are no more reads to read.

### **Author(s)**

Gregoire Pau

### **See Also**

FastQStreamer.init

---

FastQStreamer.init

*Open a streaming connection to a FastQ file*

---

### **Description**

Open a streaming connection to a FastQ file

### **Usage**

```
FastQStreamer.init(input_file, input_file2 = NULL, chunk_size,  
  subsample_nbread = NULL, max_nbchunks = NULL)
```

### **Arguments**

input_file	Path to a FastQ file
input_file2	Optional path to a FastQ file. Default is NULL.
chunk_size	Number of reads per chunk
subsample_nbread	Optional number of reads to subsample (deterministic) from the input files. Default is NULL.
max_nbchunks	Optional maximal number of chunks to read

**Details**

Only one FastQStreamer object can be open at any time.

**Value**

Nothing.

**Author(s)**

Gregoire Pau

**See Also**

FastQStreamer.getReads

---

FastQStreamer.release *Close the FastQStreamer*

---

**Description**

Close the FastQStreamer

**Usage**

FastQStreamer.release()

**Value**

Nothing

**Author(s)**

Gregoire Pau

**See Also**

FastQStreamer.init

---

filterByLength	<i>Filter reads by length</i>
----------------	-------------------------------

---

**Description**

Checks whether reads have at least a length of minlength. Useful values are zero the rid of empty reads or 12 to match the gsnap k-mer size.

**Usage**

```
filterByLength(lreads, minlength = 12, paired = FALSE)
```

**Arguments**

lreads	A set of reads as ShortReadQ object
minlength	Minimum length
paired	Indicates whether lreads has one of two elements

**Value**

A boolean vector indicating whether read passes filter

---

filterQuality	<i>Filter reads by quality</i>
---------------	--------------------------------

---

**Description**

Filtering reads by quality score. Discards reads that have more than a fraction of X nucleotides with a score below Y.

**Usage**

```
filterQuality(lreads)
```

**Arguments**

lreads	A list of ShortReadQ objects
--------	------------------------------

**Details**

X and Y are controlled by global config variables X: filterQuality.minFrac Y: filterQuality.minQuality

**Value**

A list of quality filterered ShortReadQ objects

---

findVariantFile	<i>Get a vcf filename given a HTSeqGenie directory</i>
-----------------	--

---

**Description**

Get the filename of the variant file

**Usage**

```
findVariantFile(save_dir)
```

**Arguments**

dir_path	A character string containing a dir path
----------	--

**Details**

Depending on the variant caller used and the version of VariantAnotation used to create the file a file might have the ending vcf.gz, vcf.bgz. To function hides all this mess.

**Value**

A character vector containing an existing filename, stops if 0 or more than 1

**Author(s)**

Jens Reeder

---

gatk	<i>gatk</i>
------	-------------

---

**Description**

Run a command from the GATK

**Usage**

```
gatk(gatk.jar.path = getOption("gatk.path"), method, args, maxheap = "4g")
```

**Arguments**

gatk.jar.path	Path to the gatk jar file
method	Name of the gatk method, e.g. UnifiedGenotyper
args	additional args passed to gatk
maxheap	Maximal heap space allocated for java, GATK recommends 4G heap for most of its apps

**Details**

Execute the GATK jar file using the method specified as arg. Stops if the command executed fails.

**Value**

0 for success, stops otherwise

**Author(s)**

Jens Reeder

---

`generateSingleGeneDERs`

*generateSingleGeneDERs*

---

**Description**

Generate DEXSeq-ready exons

**Usage**

`generateSingleGeneDERs(txdb)`

**Arguments**

`txdb`            A transcript DB object

**Details**

`generateSingleGeneDERs()` generates exons by: 1) disjoining the whole exon set 2) keeping only the exons of coding regions 3) keeping only the exons that belong to unique genes

**Value**

single gene DERs

---

getAdapterSeqs	<i>Read list of Illumina adapter seqs from package data</i>
----------------	---

---

**Description**

Read list of Illumina adapter seqs from package data

**Usage**

```
getAdapterSeqs(paired_ends, force_paired_end_adapter, pair_num = 1)
```

**Arguments**

paired_ends	Do we have paired ends reads?
force_paired_end_adapter	Force paired end adapters for single end reads?
pair_num	1 for forward read, 2 for reverse read

**Value**

The adapter seq as string

---

getBams	<i>Get bam files of a pipeline run</i>
---------	--

---

**Description**

Get bam files of a pipeline run

**Usage**

```
getBams(save_dir)
```

**Arguments**

save_dir	Save directory of a pipeline run
----------	----------------------------------

**Value**

named list of bam files

**Author(s)**

Gregoire Pau

---

getChunkDirs	<i>Get the list of chunk directories</i>
--------------	--

---

**Description**

Get the list of chunk directories

**Usage**

```
getChunkDirs()
```

**Value**

List of chunk directories

**Author(s)**

Gregoire Pau

---

getConfig	<i>Get a configuration parameter</i>
-----------	--------------------------------------

---

**Description**

Get a configuration parameter

**Usage**

```
getConfig(p, stop.ifempty = FALSE)
```

**Arguments**

p	Name of parameter
stop.ifempty	throw error if value is not set, otherwise returns NULL

**Value**

If parameter is missing, return the config list otherwise return the value of the parameter name as a character string throws an exception if the parameter is not present in the config

---

`getConfig.integer`      *Check if a config parameter is an integer*

---

**Description**

Throws exception if value is no integer

**Usage**

`getConfig.integer(p, tol = 1e-08, ...)`

**Arguments**

`p`                      Name of parameter  
`tol`                     Tolerance that controls how far a value can be from the next integer.  
`...`                    Additional parameters passed to `getConfig()`

**Value**

Value of parameter as integer

---

`getConfig.logical`      *Check if a config parameter has a logical value*

---

**Description**

Throws exception if value is not logical

**Usage**

`getConfig.logical(p, ...)`

**Arguments**

`p`                      Name of parameter  
`...`                    extra params passed to `getConfig`

**Value**

Logical value of parameter

getConfig.numeric      *Check if a config parameter is a numeric*

---

**Description**

Throws exception if value can't be cast into numeric

**Usage**

```
getConfig.numeric(p, ...)
```

**Arguments**

p	Name of parameter
...	Extra params passed to getConfig

**Value**

Value of parameter as numeric

---

getConfig.vector      *Return values of a config variable as vector*

---

**Description**

Return values of a config variable as vector

**Usage**

```
getConfig.vector(p, ...)
```

**Arguments**

p	Name of parameter
...	extra params passed to getConfig

**Value**

value of config param as vector

---

*getEndNumber*                      *Get Read End Number*

---

**Description**

Returns the end number of an end from a paired-end read

**Usage**

`getEndNumber(int)`

**Arguments**

`int`                      an int from a SAM flag

**Details**

Given an integer from the BAM flag field, tells which end it is in a read

**Value**

1,2

**Author(s)**

Cory Barr

---

*getMemoryUsage*                      *Returns memory usage in bytes*

---

**Description**

For debugging.

**Usage**

`getMemoryUsage()`

**Value**

Memory usage in bytes

getNumberOfReadsInFASTQFile  
*Count reads in Fastq file*

---

**Description**

Count reads in Fastq file

**Usage**

```
getNumberOfReadsInFASTQFile(filename)
```

**Arguments**

filename          Name of FastQ file

**Value**

Number of reads

**Author(s)**

Gregoire Pau

---

getNumericVectorDataFromFile  
*Load data as numerical values*

---

**Description**

Load data as numerical values

**Usage**

```
getNumericVectorDataFromFile(dir_path, object_name)
```

**Arguments**

dir\_path          Save dir of a pipeline run  
object\_name      Object name

**Value**

loaded data as table of numbers

**Author(s)**

Jens Reeder

---

getObjectFilename      *Get a filename given a directory and the object name*

---

**Description**

Get a filename given a directory and the object name

**Usage**

```
getObjectFilename(dir_path, object_name)
```

**Arguments**

dir_path	A character string containing a dir path
object_name	A character string containing the regular expression matching a filename in dir_path

**Value**

A character vector containing an existing filename, stops if 0 or more than 1

**Author(s)**

Gregoire Pau

---

getPackageFile      *Get a package file*

---

**Description**

Magically get package files from the inst directory, which will be in different location, depending on whether we run in: - local mode: if interactive() is TRUE - package mode: if interactive() is FALSE

**Usage**

```
getPackageFile(filename, package = "HTSeqGenie", mustWork = TRUE)
```

**Arguments**

filename	Name of package file
package	Name of the package the file is coming from
mustWork	Boolean, will stop the code if set tot TRUE and file not found otherwise returns Nothing.

**Value**

relative path to requested file

---

getRandomAlignCutoff    *Estimate an adapter alignment cutoff score*

---

**Description**

Empirically estimate a threshold that discriminates random reads from reads with adapter contamination

**Usage**

```
getRandomAlignCutoff(read_len, n)
```

**Arguments**

read_len	The read length
n	Number of samples

---

getRRNAIds                    *Detect reads that look like rRNA*

---

**Description**

Detect reads that look like rRNA

**Usage**

```
getRRNAIds(file1, file2 = NULL, tmp_dir, rRNADb)
```

**Arguments**

file1	FastQ file of forward reads
file2	FastQ of reverse reads in paired-end sequencing, NULL otherwise
tmp_dir	temporary directory used for storing the gsnap results
rRNADb	Name of the rRNA sequence database. Must exist in the gsnap genome directory

**Value**

IDs of reads flagged as rRNA

---

getTabDataFromFile	<i>Load tabular data from the NGS pipeline result directory</i>
--------------------	---

---

**Description**

Load tabular data from the NGS pipeline result directory

**Usage**

```
getTabDataFromFile(save_dir, object_name)
```

**Arguments**

save_dir	A character string containing an NGS pipeline output directory.
object_name	A character string containing the regular expression matching a filename in dir_path

**Value**

A data frame.

---

getTraceback	<i>Get traceback from tryKeepTraceback()</i>
--------------	--

---

**Description**

Get traceback from tryKeepTraceback()

**Usage**

```
getTraceback(mto)
```

**Arguments**

mto	An object of the try-error class
-----	----------------------------------

**Value**

Traceback as a string

---

hashCoverage	<i>Hashing function for coverage</i>
--------------	--------------------------------------

---

**Description**

Hashing function for coverage

**Usage**

```
hashCoverage(cov)
```

**Arguments**

cov	A SimpleRleList object
-----	------------------------

**Value**

A numeric

**Author(s)**

Gregoire Pau

---

hashVariants	<i>Hashing function for variants</i>
--------------	--------------------------------------

---

**Description**

Hashing function for variants

**Usage**

```
hashVariants(var)
```

**Arguments**

var	A GRanges object
-----	------------------

**Value**

A numeric

**Author(s)**

Gregoire Pau

---

hashVector	<i>Hashing function for vector</i>
------------	------------------------------------

---

**Description**

Hashing function for vector

**Usage**

```
hashVector(x)
```

**Arguments**

x                    A vector

**Value**

A numeric

**Author(s)**

Gregoire Pau

---

HTSeqGenie	<i>Package overview</i>
------------	-------------------------

---

**Description**

The HTSeqGenie package is a robust and efficient software to analyze high-throughput sequencing experiments in a reproducible manner. It supports the RNA-Seq and Exome-Seq protocols and provides: quality control reporting (using the ShortRead package), detection of adapter contamination, read alignment versus a reference genome (using the gmapR package), counting reads in genomic regions (using the GenomicRanges package), and read-depth coverage computation.

**Package content**

To run the pipeline:

- runPipeline

To access the pipeline output data:

- getTabDataFromFile

To build the genomic features object:

- buildGenomicFeaturesFromTxDb
- TP53GenomicFeatures

## Examples

```
## Not run:
## build genome and genomic features
tp53Genome <- TP53Genome()
tp53GenomicFeatures <- TP53GenomicFeatures()

## get the FASTQ files
fastq1 <- system.file("extdata/H1993_TP53_subset2500_1.fastq.gz", package="HTSeqGenie")
fastq2 <- system.file("extdata/H1993_TP53_subset2500_2.fastq.gz", package="HTSeqGenie")

## run the pipeline
save_dir <- runPipeline(
  ## input
  input_file=fastq1,
  input_file2=fastq2,
  paired_ends=TRUE,
  quality_encoding="illumina1.8",

  ## output
  save_dir="test",
  prepend_str="test",
  overwrite_save_dir="erase",

  ## aligner
  path.gsnap_genomes=path(directory(tp53Genome)),
  alignReads.genome=genome(tp53Genome),
  alignReads.additional_parameters="--indel-penalty=1 --novelsplicing=1 --distant-splice-penalty=1",

  ## gene model
  path.genomic_features=dirname(tp53GenomicFeatures),
  countGenomicFeatures.gfeatures=basename(tp53GenomicFeatures)
)

## End(Not run)
```

---

initDirs

*Set up NGS output dir*

---

## Description

Set up NGS output dir (using save\_dir from getConfig)

## Usage

```
initDirs()
```

## Value

Nothing

**Author(s)**

Gregoire Pau

---

*initLog*                      *Initialize the logger*

---

**Description**

Setup logging file in save\_dir/progress.log and log sessionInfo and configuration

**Usage**

```
initLog(save_dir, debug_level = "INFO")
```

**Arguments**

save\_dir            Save dir of a pipeline run  
debug\_level        One of INFO, WARN, ERROR, FATAL

**Value**

Log file name

---

*initLogger*                      *Init loggers*

---

**Description**

Init loggers (output dir log, using save\_dir from getConfig, and console log)

**Usage**

```
initLogger()
```

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

`initPipelineFromConfig`*Init pipeline environment*

---

**Description**

Init pipeline environment

**Usage**

```
initPipelineFromConfig(config_filename, config_update)
```

**Arguments**

`config_filename`

Name of config file

`config_update` List of name value pairs that will update the config parameters

**Value**

Nothing

**Author(s)**

Jens Reeder

---

`initPipelineFromSaveDir`*Init Pipeline environment from previous run*

---

**Description**

Init Pipeline environment from previous run

**Usage**

```
initPipelineFromSaveDir(save_dir, config_update)
```

**Arguments**

`save_dir` Save dir of a previous pipeline run

`config_update` List of name value pairs that will update the config parameters

**Details**

Loads the config file from a previous run stored in `[save_dir]/logs/config.txt`

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

 isAboveQualityThresh *Check for high quality reads*


---

**Description**

Checks whether reads have more than a fraction of minFrac nucleotides with a score below min-quality.

**Usage**

```
isAboveQualityThresh(reads, minquality, minfrac)
```

**Arguments**

reads	A set of reads as ShortReadQ object
minquality	Minimal quality score
minfrac	Fraction of positions that need to be over minquality to be considered a good read.

**Value**

A boolean vector indicating whether read is considered high quality.

---

 isAdapter *Detect adapter contamination*


---

**Description**

Does a Needleman-Wunsch like small-in-large alignment of the adapter vs each read. Flag read if score exceeds threshold

**Usage**

```
isAdapter(reads, score_cutoff, adapter_seqs)
```

**Arguments**

reads	Set of reads as ShortRead object
score_cutoff	Alignment score threshold that needs to be exceeded to be flagged as adapter. Usually this value is determined empirically by getAdpaterThreshold()
adapter_seqs	One or more adapter sequences

**Value**

boolean vector indicating adapter contamination

---

isConfig	<i>Test the presence of the parameter in the current config</i>
----------	---

---

**Description**

Test the presence of the parameter in the current config

**Usage**

isConfig(parameter)

**Arguments**

parameter	Name of parameter
-----------	-------------------

**Value**

TRUE if present, FALSE otherwise

---

isFirstFragment	<i>Does a SAM flag indicate the first fragment</i>
-----------------	--

---

**Description**

Compute whether a SAM/BAM flag indicates a first fragment. Method is not foolproof, as it ignores a lot of SAM semantics. E.g the SAM spec says: "If 0x1 is unset, no assumptions can be made about 0x2, 0x8, 0x20, 0x40 and 0x80". For our purpose this should be enough, but we should keep an open eye for a more robust implementation in Rsamtools.

**Usage**

isFirstFragment(flag)

**Arguments**

flag	A flag from the BAM/SAM file
------	------------------------------

**Value**

Logical

---

`isSparse`*isSparse*

---

**Description**

Check coverage for sparseness

**Usage**`isSparse(cov, threshold = 0.1)`**Arguments**

<code>cov</code>	A cov object as SimpleRleList
<code>threshold</code>	Fraction of number of runs over total length

**Details**

Some Rle related operations become very slow when they are dealing with data that violates their sparseness assumption. This method provides an estimate about whether the data is dense or sparse. More precicely it checks if the fraction of the number of runs over the total length is smaller than a threshold

**Value**

Boolean whether this object is dense or sparse

**Author(s)**

Jens Reeder

---

`listIterator.init`      *Create a iterator on a list*

---

**Description**

Create a iterator on a list

**Usage**

```
listIterator.init(x)
```

**Arguments**

x                      A list.

**Details**

Only one listIterator object can be open at any time.

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

`listIterator.next`      *Get reads from the listIterator*

---

**Description**

Get reads from the listIterator

**Usage**

```
listIterator.next()
```

**Value**

An object. NULL if there are no more objects in the listIterator.

**Author(s)**

Gregoire Pau

**See Also**

listIterator.init

---

loadConfig	<i>Load configuration file</i>
------------	--------------------------------

---

**Description**

Loads the indicated configuration file. Creates and installs a global variable that should be accessed only via getConfig().

**Usage**

```
loadConfig(filename)
```

**Arguments**

filename	Path to configuration file
----------	----------------------------

**Value**

Nothing. Called for its side effect, which is setting the global config variable.

---

logdebug	<i>Log debug using the logging package</i>
----------	--

---

**Description**

Log debug (with a try statement)

**Usage**

```
logdebug(msg)
```

**Arguments**

...	Arguments passed to logging::logdebug
-----	---------------------------------------

**Value**

Nothing

**Author(s)**

Gregoire Pau

logerror *Log info using the logging package*

---

**Description**

Log error (with a try statement)

**Usage**

logerror(msg)

**Arguments**

... Arguments passed to logging::loginfo

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

loginfo *Log info using the logging package*

---

**Description**

Log info (with a try statement)

**Usage**

loginfo(msg)

**Arguments**

... Arguments passed to logging::loginfo

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

logwarn	<i>Log warning using the logging package</i>
---------	--

---

**Description**

Log warning (with a try statement)

**Usage**

```
logwarn(msg)
```

**Arguments**

... Arguments passed to logging::logwarn

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

makeDir	<i>Make a directory after performing an existence check</i>
---------	---

---

**Description**

Throws an exception if file or directory with same name exist and overwrite is TRUE.

**Usage**

```
makeDir(dir, overwrite = "never")
```

**Arguments**

dir Name of directory to create  
overwrite A character string: never (default), erase, overwrite

**Value**

Path to created directory

---

makeRandomSreads	<i>Generate a couple if random ShortReadQ, intended for testing</i>
------------------	---

---

**Description**

Generate a couple if random ShortReadQ, intended for testing

**Usage**

```
makeRandomSreads(num, len)
```

**Arguments**

num	an integer
len	an integer

**Value**

a DNAStrngSet

**Author(s)**

Gregoire Pau

---

markDuplicates	<i>markDuplicates</i>
----------------	-----------------------

---

**Description**

Mark duplicates in bam

**Usage**

```
markDuplicates(bamfile, outfile = NULL, path = getOption("picard.path"))
```

**Arguments**

bamfile	Name of input bam file
outfile	Name of output bam file
path	Full path to MarkDuplicates jar

**Details**

Use MarkDuplicates from PicardTools to mark duplicate alignments in bam file.

**Value**

Path to output bam file

**Author(s)**

Jens Reeder

---

markDups

*markDups*

---

**Description**

Mark duplicates in pipeline context

**Usage**

markDups()

**Details**

High level function call to mark duplicates in the analyzed.bam file of a pipelin run.

**Value**

Nothing

**Author(s)**

Jens Reeder

---

mergeAlignReads

*Merge after alignReads*

---

**Description**

Merge BAMs and create summary alignment file

**Usage**

mergeAlignReads(indirs, outdir, prepend\_str, num\_cores)

**Arguments**

indirs	A character vector, indicating which directories have to be merged
outdir	A character string indicating the output directory (which must exist)
prepend_str	A character string, containing a prefix going to be appended on all output result files
num_cores	Number of cores available for parallel processing (for the merge bam step)

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

mergeCoverage	<i>Merge coverage files</i>
---------------	-----------------------------

---

**Description**

Merge coverage files

**Usage**

```
mergeCoverage(indirs, outdir, prepend_str)
```

**Arguments**

indirs	A character vector, indicating which directories have to be merged
outdir	A character string indicating the output directory (which must exist)
prepend_str	A character string, containing a prefix going to be appended on all output result files

**Details**

Merges coverage objects, usually SipleRleLists, in a tree-reduce fashion. The coverage object dynamically switches to a SimpleIntegerList, once the data becomes too dense.

**Value**

Nothing

**Author(s)**

Jens Reeder

---

mergeLanes	<i>Merge input lanes</i>
------------	--------------------------

---

**Description**

Merge input lanes built by the NGS pipeline

**Usage**

```
mergeLanes(indirs, outdir, prepend_str, num_cores, config_update,
           preMergeChecks.do = TRUE, ignoreConfigParameters)
```

**Arguments**

indirs	A character vector of directory paths containing NGS pipeline output
outdir	A character string pointing to a non-existing output directory
prepend_str	A character string, containing a prefix going to be appended on all output result files
num_cores	Number of cores available for parallel processing (for the merge bam step)
config_update	List of name value pairs that will update the config parameters
preMergeChecks.do	A logical, indicating whether to perform pre merge checks
ignoreConfigParameters	A character vector containing the configuration parameters that are not required to be identical

**Value**

Nothing

**Author(s)**

greg

---

mergePreprocessReads	<i>Merge after preprocessReads</i>
----------------------	------------------------------------

---

**Description**

Merge detectAdapterContam, merge preprocessed reads, create summary preprocess, build short-ReadReport, remove processed

**Usage**

```
mergePreprocessReads(indirs, outdir, prepend_str)
```

**Arguments**

<code>indirs</code>	A character vector, indicating which directories have to be merged
<code>outdir</code>	A character string indicating the output directory (which must exist)
<code>prepend_str</code>	A character string, containing a prefix going to be appended on all output result files

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

`mergeSummaryAlignment` *Merge summary alignments*

---

**Description**

Merge summary alignments

**Usage**

```
mergeSummaryAlignment(indirs, outdir, prepend_str)
```

**Arguments**

<code>indirs</code>	A character vector, indicating which directories have to be merged
<code>outdir</code>	A character string indicating the output directory (which must exist)
<code>prepend_str</code>	A character string, containing a prefix going to be appended on all output result files

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

parseDCF	<i>Read and parse a configuration file</i>
----------	--

---

**Description**

From a file like x1: y1 x2: y2 extract field, using the rules: - split on ':' - first element of split id name of parameter, second is value - trailing whitespaces (tabs and spaces) are removed - comments (text flow starting with #) are removed

**Usage**

```
parseDCF(filename)
```

**Arguments**

filename	File name
----------	-----------

**Value**

Named list

---

parseSummaries	<i>parse summary files from save dirs</i>
----------------	---

---

**Description**

Parse a summary from a list of save\_dirs

**Usage**

```
parseSummaries(save_dirs, summary.name)
```

**Arguments**

save_dirs	list of result dirs
summary.name	name of summary file e.g. summary_counts

**Details**

This function allows to parse a given summary from a list of pipeline results save\_dirs

**Value**

data frame with summaries

**Author(s)**

Jens Reeder

---

picard	<i>picard</i>
--------	---------------

---

**Description**

Generic function to call all picard command line java tools

**Usage**

```
picard(tool, ..., path = getOption("picard.path"))
```

**Arguments**

tool	Name of the Picard Tool, e.g. MarkDuplicates
...	Arguments forwarded to the picard tool
path	full path to the picard tool jar file.

**Value**

Nothing

**Author(s)**

Jens Reeder, Michael Lawrence

---

plotDF	<i>Make continuous plots of distribution function</i>
--------	---

---

**Description**

Make continuous plots of distribution function

**Usage**

```
plotDF(df, ylab, xlab, filename)
```

**Arguments**

df	distribution function, given as absolute count and percent
ylab	label of y axis
xlab	label of x axis
filename	plots will be saved under [filename].png and [filename].pdf

**Value**

Nothing, creates two files instead

**Author(s)**

Jens Reeder

---

```
preprocessReads      Pipeline preprocessing
```

---

**Description**

The preprocessing for our NGS pipelines consists of : - quality filtering - check for adapter contamination - filtering of rRNA reads - read trimming - shortRead report generation of surviving reads

**Usage**

```
preprocessReads()
```

**Details**

These steps are mostly controlled by the global config.

**Value**

A named vector containing the path to the preprocessed FastQ files and a few other statistics

---

```
preprocessReadsChunk  Preprocess a chunk
```

---

**Description**

Preprocess a chunk

**Usage**

```
preprocessReadsChunk(lreads, save_dir = NULL)
```

**Arguments**

<code>lreads</code>	A list of GRanges objects, containing the reads
<code>save_dir</code>	Save directory of a pipeline run

**Value**

save\_dir Save directory of a pipeline run

**Author(s)**

Gregoire Pau

---

processChunks      *Process chunk in the pipeline framework*

---

**Description**

Process chunk in the pipeline framework

**Usage**

```
processChunks(inext, fun, nb.parallel.jobs)
```

**Arguments**

inext	A function (without argument) returning an object to process; NULL if none left; this function is run in the main thread
fun	Function to process the object returned by inext; this function is run in children threadfunction to apply to a chunk
nb.parallel.jobs	number of parallel jobs

**Details**

High-level pipeline-specific version of sclapply, with chunk loggers and safeExecute

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

readInputFiles	<i>Read FastQ input files</i>
----------------	-------------------------------

---

**Description**

Uses the global config to find input files

**Usage**

```
readInputFiles()
```

**Value**

Reads as list of ShortRead objects

---

readRNASeqEnds	<i>Read single/paired End Bam Files</i>
----------------	---

---

**Description**

Read single/paired end BAM files with requested columns from the BAM

**Usage**

```
readRNASeqEnds(filename, paired_ends, remove.strandness = TRUE)
```

**Arguments**

filename	Path to a bam file
paired_ends	A logical indicating whether the reads are paired
remove.strandness	A logical indicating whether read strands should be set to "*".

**Value**

GRangesList

**Author(s)**

Cory Barr

---

realignIndels	<i>realignIndels</i>
---------------	----------------------

---

**Description**

Realign indels in pipeline context

**Usage**

```
realignIndels()
```

**Details**

High level function call to realign indels in the analyzed.bam file using GATK

**Value**

Nothing

**Author(s)**

Jens Reeder

---

realignIndelsGATK	<i>Realign indels via GATK</i>
-------------------	--------------------------------

---

**Description**

Realigning indels using the GATK tools RealignerTargetCreator and IndelRealigner. Requires a GATK compatible genome with a name matching the alignment genome to be installed in 'path.gatk\_genome'

**Usage**

```
realignIndelsGATK(bam.file)
```

**Arguments**

bam.file	Path to bam.file
----------	------------------

**Details**

Since GATKs IndelRealigner is not parallelized, we run it in parallel per chromosome.

**Value**

Path to realigned bam file

**Author(s)**

Jens Reeder

---

relativeBarPlot	<i>Make relative bar plots</i>
-----------------	--------------------------------

---

**Description**

Make relative bar plots

**Usage**

```
relativeBarPlot(data, total, labels, title, filename, ylab = "Percent",  
                cex.names = 0.9, ymax = 100)
```

**Arguments**

data	vector of raw, absolute counts
total	number to normalize by, can be vector of same length as data
labels	x-axes labels, category labels for data
title	Title of the plot
filename	plots will be saved under [filename].png and [filename].pdf
ylab	label of y axis
cex.names	scaling param of lables, passed to plot
ymax	extent of y-axis

**Value**

Nothing, creates two files instead

---

removeChunkDir	<i>Remove chunk directories</i>
----------------	---------------------------------

---

**Description**

Remove chunk directories

**Usage**

```
removeChunkDir()
```

**Details**

A pipeline run processes the data in small chunks, which are eventually combined into the final result. Afterwards, this function can be called to remove the temporary results per chunk.

**Value**

Nothing

**Author(s)**

Jens Reeder

---

resource	<i>Reload package source code</i>
----------	-----------------------------------

---

**Description**

When developing code this function can be used to quickly reload all of the packages code, without installing it.

**Usage**

```
resource(dirname = ".")
```

**Arguments**

dirname            Directore with files to source

**Value**

Nothing

---

rpkm	<i>Calculate RPKM</i>
------	-----------------------

---

**Description**

Calculate RPKM

**Usage**

```
rpkm(counts, widths, nbreads)
```

**Arguments**

counts            A vector of counts  
widths            vector of the width of each bin the counts were performed on  
nreads            vector containing number of reads mapped to each bin

**Value**

vector of RPKMs

**Author(s)**

Gregoire Pau

---

runAlignment            *Runs the read alignment step of the pipeline*

---

**Description**

Runs the read alignment step of the pipeline

**Usage**

```
runAlignment(config_filename, config_update)
```

**Arguments**

config\_filename            Path to configuration file  
config\_update            List of name value pairs that will update the config parameters

**Value**

Nothing

**Author(s)**

Jens Reeder

---

`runPipeline`*Run the NGS analysis pipeline*

---

**Description**

Run the NGS analysis pipeline

**Usage**

```
runPipeline(...)
```

**Arguments**

...                   A list of parameters. See the vignette for details.

**Details**

This function starts the pipeline. It first preprocesses the input FASTQ reads, align them, count the read overlaps with genomic features and compute the coverage. See the vignette for details.

**Value**

The path to the NGS output directory.

**Author(s)**

Jens Reeder, Gregoire Pau

**See Also**

TP53Genome, TP53GenomicFeatures

**Examples**

```
## Not run:
## build genome and genomic features
tp53Genome <- TP53Genome()
tp53GenomicFeatures <- TP53GenomicFeatures()

## get the FASTQ files
fastq1 <- system.file("extdata/H1993_TP53_subset2500_1.fastq.gz", package="HTSeqGenie")
fastq2 <- system.file("extdata/H1993_TP53_subset2500_2.fastq.gz", package="HTSeqGenie")

## run the pipeline
save_dir <- runPipeline(
  ## input
  input_file=fastq1,
  input_file2=fastq2,
  paired_ends=TRUE,
```

```
quality_encoding="illumina1.8",

## output
save_dir="test",
prepend_str="test",
overwrite_save_dir="erase",

## aligner
path.gsnap_genomes=path(directory(tp53Genome)),
alignReads.genome=genome(tp53Genome),
alignReads.additional_parameters="--indel-penalty=1 --novelsplicing=1 --distant-splice-penalty=1",

## gene model
path.genomic_features=dirname(tp53GenomicFeatures),
countGenomicFeatures.gfeatures=basename(tp53GenomicFeatures)
)

## End(Not run)
```

---

runPipelineConfig      *Run the NGS analysis pipeline*

---

## Description

Run the NGS analysis pipeline from a configuration file

## Usage

```
runPipelineConfig(config_filename, config_update)
```

## Arguments

config\_filename      Path to a pipeline configuration file

config\_update      A list of name value pairs that will update the config parameters

## Details

This is the launcher function for all pipeline runs. It will do some preprocessing steps, then aligns the reads, counts overlap with genomic Features such as genes, exons etc and applies a variant caller.

## Value

Nothing

**Author(s)**

Jens Reeder, Gregoire Pau

---

runPreprocessReads	<i>Run the preprocessing steps of the pipeline</i>
--------------------	--

---

**Description**

Runs the preprocessing steps of the pipeline

**Usage**

```
runPreprocessReads(config_filename, config_update)
```

**Arguments**

config_filename	Path to configuration file
config_update	List of name value pairs that will update the config parameters

**Value**

Nothing

**Author(s)**

Jens Reeder

---

safe.yield	<i>Overloaded yield(...) method catching truncated exceptions for FastqStreamer</i>
------------	---

---

**Description**

Overloaded yield(...) method catching truncated exceptions for FastqStreamer

**Usage**

```
safe.yield(fqs)
```

**Arguments**

fqs	An instance from the FastqSampler or FastqStreamer class.
-----	---

**Value**

Same as FastqStreamer::yield

**Author(s)**

Gregoire Pau

---

safeExecute	<i>Execute function in try catch with trace function</i>
-------------	--

---

**Description**

Requires the logger to be set

**Usage**

```
safeExecute(expr, memtracer = TRUE, newthread = TRUE)
```

**Arguments**

expr	Expression to safely execute
memtracer	A boolean, to enable/disable a periodic memory tracer. Default is TRUE.
newthread	A boolean, indicating if a new thread should be used (to save memory from the main thread)

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

safeGetObject	<i>Safely load a R data file</i>
---------------	----------------------------------

---

**Description**

Attempts to load a file given by object\_name. Bails out if none or more than one files match the object name.

**Usage**

```
safeGetObject(dir_path, object_name)
```

**Arguments**

dir_path	Save dir of a pipeline run
object_name	object name, can be a regexp

**Value**

loaded object

---

safeUnlink	<i>safeUnlink</i>
------------	-------------------

---

**Description**

Symlink-safe file/directory delete function

**Usage**

safeUnlink(path)

**Arguments**

path	A character string indicating which file/directory to delete.
------	---

**Details**

Unlike unlink(), safeUnlink() does not follow symlink directories for deletion.

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

saveWithID	<i>Save an R object</i>
------------	-------------------------

---

**Description**

Exists so objects can be serialized and reloaded with the a unique identifier in the symbol. Stores the data object with a new name

**Usage**

```
saveWithID(data, orig_name, id, save_dir, compress = TRUE, format = "RData")
```

**Arguments**

data	The data to store
orig_name	The original name of the data
id	A meaningful id the is prepended to the stored objects name
save_dir	The directory where the data should be saved in
compress	Save the data compressed or not
format	Choice of 'RData' or 'tab'(ular)

**Value**

Name of the stored file

---

sclapply	<i>Scheduled parallel processing</i>
----------	--------------------------------------

---

**Description**

Scheduled parallel processing

**Usage**

```
sclapply(inext, fun, max.parallel.jobs, ..., stop.onfail = TRUE,  
         tracefun = NULL, tracefun.period = 60)
```

**Arguments**

inext	A function (without argument) returning an object to process; NULL if none left; this function is run in the main thread
fun	Function to process the object returned by inext; this function is run in children thread
max.parallel.jobs	Number of jobs to start in parallel
...	Further arguments passed to fun
stop.onfail	Throw error if one
tracefun	Callback function that will be executed in a separate thread
tracefun.period	Time interval between calls to tracefun

**Value**

Return value of applied function

---

setChunkDir	<i>Set the base directory for the chunks</i>
-------------	--

---

**Description**

Set the base directory for the chunks

**Usage**

```
setChunkDir()
```

**Value**

path to chunk dir

**Author(s)**

Jens Reeder

---

setUpDirs	<i>Create output directory and subdirectories for sequencing pipeline analysis outputs</i>
-----------	--

---

**Description**

Creates a directory with all needed subdirectories for pipeline outputs

**Usage**

```
setUpDirs(save_dir, overwrite = "never")
```

**Arguments**

save_dir	path to the directory that will contain all needed subdirectories
overwrite	A character string: never (default), erase, overwrite

**Value**

Nothing. Called for its side effects

**Author(s)**

Cory Barr, Jens Reeder

---

setupTestFramework	<i>setup test framework</i>
--------------------	-----------------------------

---

**Description**

setup test framework

**Usage**

```
setupTestFramework(config.filename, config.update = list(),
  testname = "test", package = "HTSeqGenie", use.TP53Genome = TRUE)
```

**Arguments**

config.filename	configuration file
config.update	update list of config values
testname	name of test case
package	name of package
use.TP53Genome	Boolean indicating the use of the TP53 genome as template config

**Value**

the created temp directory

---

statCountFeatures	<i>Compute statistics on count features</i>
-------------------	---

---

**Description**

Compute statistics on count features

**Usage**

```
statCountFeatures(save_dir, feature = "counts_gene")
```

**Arguments**

save_dir	A character string containing a NGS analysis directory
feature	A character string containing a features name. Default is "counts_gene".

**Value**

A numeric vector containing statistics about features.

**Author(s)**

Gregoire Pau

---

TP53GenomicFeatures	<i>Demo genomic features around the TP53 gene</i>
---------------------	---

---

**Description**

Build the genomic features of the TP53 demo region

**Usage**

```
TP53GenomicFeatures()
```

**Details**

Returns a list of genomic features (gene, exons, transcripts) annotating a region of UCSC hg19 sequence centered on the region of the TP53 gene, with 1 Mb flanking sequence on each side. This is intended as a test/demonstration to run the NGS pipeline in conjunction with the LungCancerLines data package.

**Value**

A list of GRanges objects containing the genomic features

**Author(s)**

Gregoire Pau

**See Also**

TP53Genome, buildGenomicFeaturesFromTxDb, runPipeline

---

traceMem	<i>Show memory usage</i>
----------	--------------------------

---

**Description**

For debugging purposes only. Show memory usage if config variable

**Usage**

```
traceMem()
```

**Value**

Nothing

---

trimReads	<i>Trim/truncate a set of reads</i>
-----------	-------------------------------------

---

**Description**

Trim/truncate a set of reads

**Usage**

```
trimReads(lreads, trim_len = NULL, trim5 = 0)
```

**Arguments**

lreads	A list of ShortReadQ objects
trim_len	The length reads will be truncated to; default is NULL (no length truncation)
trim5	The number of nucleotides to trim from the 5'-end; default is 0

**Value**

A list of truncated ShortReadQ objects

---

trimTailsByQuality	<i>Trim off low quality tail</i>
--------------------	----------------------------------

---

### Description

The illumina manuals states: If a read ends with a segment of mostly low quality (Q15 or below), then all of the quality values in the segment are replaced with a value of 2( encoded as the letter B in Illumina's text-based encoding of quality scores)... This Q2 indicator does not predict a specific error rate, but rather indicates that a specific final portion of the read should not be used in further analyses.

### Usage

```
trimTailsByQuality(lreads, minqual = "#")
```

### Arguments

lreads	A list (usually a pair) of ShortReadQ object
minqual	An ascii encoded quality score

### Details

For illumina 1.8 the special char is encoded as '#', which we chose as default here. For illumina 1.5 make sure to set the minqual to 'B'

### Value

A list of quality trimmed ShortReadQ objects

---

truncateReads	<i>Trim/truncate a set of reads</i>
---------------	-------------------------------------

---

### Description

Trim/truncate a set of reads

### Usage

```
truncateReads(reads, trim_len = NULL, trim5 = 0)
```

### Arguments

reads	A set of reads as ShortReadQ object
trim_len	The length reads will be truncated to; default is NULL (no length truncation)
trim5	The number of nucleotides to trim from the 5'-end; default is 0

**Value**

A truncated ShortReadQ object

---

tryKeepTraceback	<i>Wrapper around try-catch</i>
------------------	---------------------------------

---

**Description**

Wrapper around try-catch

**Usage**

```
tryKeepTraceback(expr)
```

**Arguments**

expr	Expression to evaluate
------	------------------------

**Value**

Result of expression or error if thrown

---

updateConfig	<i>Update the existing config</i>
--------------	-----------------------------------

---

**Description**

Update the existing config

**Usage**

```
updateConfig(tconfig)
```

**Arguments**

tconfig	List of configuration name value pairs
---------	--

**Value**

Nothing.

---

vcfStat	<i>Compute stats on a VCF file</i>
---------	------------------------------------

---

**Description**

Compute stats on a VCF file

**Usage**

```
vcfStat(vcf.filename)
```

**Arguments**

vcf.filename     A character pointing to a VCF (or gzipped VCF) file

**Value**

A numeric vector

**Author(s)**

Gregoire Pau

---

wrap.callVariants	<i>Variant calling</i>
-------------------	------------------------

---

**Description**

Call Variants in the pipeline framework

**Usage**

```
wrap.callVariants(bam.file)
```

**Arguments**

bam.file         Aligned reads as bam file

**Details**

A wrapper around VariantTools callVariant framework.

**Value**

Variants as Vranges

**Author(s)**

Jens Reeder

---

writeAudit	<i>Write Session information</i>
------------	----------------------------------

---

**Description**

Write Session information

**Usage**

```
writeAudit(filename)
```

**Arguments**

filename	Optional name of file. If missing, prints session information on the standard output.
----------	---

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

writeConfig	<i>Write a config file</i>
-------------	----------------------------

---

**Description**

Writes the currently active configuration to file

**Usage**

```
writeConfig(config.filename)
```

**Arguments**

config.filename	Optional name of output file. If missing, print the config file on the standard output.
-----------------	---

**Value**

Name of saved file

---

writeFastQFiles	<i>Write reads to file</i>
-----------------	----------------------------

---

**Description**

Write reads to file

**Usage**

```
writeFastQFiles(lreads, dir, filename1, filename2)
```

**Arguments**

lreads	List of reads as ShortRead objects
dir	Save directory
filename1	Name of file 1
filename2	Name of file 2

**Value**

Named list of filepaths

---

writeFeatureCountsHTML	<i>writeFeatureCountsHTML</i>
------------------------	-------------------------------

---

**Description**

writeFeatureCountsHTML

**Usage**

```
writeFeatureCountsHTML(outfile, dirPath, ExonsCoveredTable,
  GenomicFeaturesTable, GenomicFeaturesDetectedTable)
```

**Arguments**

outfile	a path
dirPath	a path
ExonsCoveredTable	a table
GenomicFeaturesTable	a table
GenomicFeaturesDetectedTable	a table

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

writeGenomicFeaturesReport  
*Generate pipeline report*

---

**Description**

Generates a summary HTML for the Genomic Feature counting step

**Usage**

writeGenomicFeaturesReport()

**Value**

Name of created HTML file

**Author(s)**

Melanie Huntley, Cory Barr, Jens Reeder

---

writePreprocessAlignHTML  
*writePreprocessAlignHTML*

---

**Description**

writePreprocessAlignHTML

**Usage**

writePreprocessAlignHTML(outfile, dirPath, sanity\_check, readFilteringTable,  
ReadMappingsTable, targetLengthTable)

**Arguments**

outfile            a path  
dirPath            a path  
sanity\_check      a logical  
readFilteringTable  
                  a table  
ReadMappingsTable  
                  a table  
targetLengthTable  
                  a table

**Value**

Nothing

**Author(s)**

Gregoire Pau

---

writePreprocessAlignReport

*Generate Pipeline Report*

---

**Description**

Generates a summary HTML for the preprocess and align step

**Usage**

```
writePreprocessAlignReport()
```

**Value**

Name of created HTML file

**Author(s)**

Melanie Huntley, Cory Barr, Jens Reeder

---

writeSummary	<i>Write HTML summary</i>
--------------	---------------------------

---

**Description**

Write html Summary for list of runs

**Usage**

```
writeSummary(dirs, cutoffs, outdir = "./")
```

**Arguments**

dirs	List of pipeline result dirs
cutoffs	list, cutoffs for each plotting/QA function
outdir	Path to output directory. Does not create dir.

**Value**

Nothing, but writes file

**Author(s)**

Jens Reeder

---

writeVCF	<i>writeVCF</i>
----------	-----------------

---

**Description**

Write variants to VCF file

**Usage**

```
writeVCF(variants.vranges, filename)
```

**Arguments**

variants.vranges	Genomic Variants as VRanges object
filename	Name of vcf file to write

**Value**

VCF file name

**Author(s)**

Jens Reeder

# Index

## \* internal

- alignReads, [4](#)
- alignReadsChunk, [5](#)
- bamCountUniqueReads, [7](#)
- buildConfig, [7](#)
- buildShortReadReports, [9](#)
- buildTallyParam, [9](#)
- buildTP53FastaGenome, [10](#)
- buildTP53GenomeTemplate, [10](#)
- calculateCoverage, [11](#)
- calculateTargetLengths, [11](#)
- checkConfig, [12](#)
- checkPicardJar, [13](#)
- computeBamStats, [14](#)
- computeCoverage, [14](#)
- countFeatures, [15](#)
- countGenomicFeatures, [16](#)
- countGenomicFeaturesChunk, [16](#)
- createTmpDir, [17](#)
- detectAdapterContam, [17](#)
- detectQualityInFASTQFile, [18](#)
- FastQStreamer.getReads, [20](#)
- FastQStreamer.init, [20](#)
- FastQStreamer.release, [21](#)
- filterByLength, [22](#)
- filterQuality, [22](#)
- findVariantFile, [23](#)
- getAdapterSeqs, [25](#)
- getBams, [25](#)
- getChunkDirs, [26](#)
- getConfig, [26](#)
- getConfig.integer, [27](#)
- getConfig.logical, [27](#)
- getConfig.numeric, [28](#)
- getConfig.vector, [28](#)
- getEndNumber, [29](#)
- getMemoryUsage, [29](#)
- getNumberOfReadsInFASTQFile, [30](#)
- getNumericVectorDataFromFile, [30](#)
- getObjectFilename, [31](#)
- getPackageFile, [31](#)
- getRandomAlignCutoff, [32](#)
- getTraceback, [33](#)
- initDirs, [36](#)
- initLog, [37](#)
- initLogger, [37](#)
- initPipelineFromConfig, [38](#)
- initPipelineFromSaveDir, [38](#)
- isAboveQualityThresh, [39](#)
- isAdapter, [39](#)
- isConfig, [40](#)
- isFirstFragment, [40](#)
- listIterator.init, [42](#)
- listIterator.next, [42](#)
- loadConfig, [43](#)
- logdebug, [43](#)
- logerror, [44](#)
- loginfo, [44](#)
- logwarn, [45](#)
- makeDir, [45](#)
- makeRandomSreads, [46](#)
- mergeAlignReads, [47](#)
- mergeCoverage, [48](#)
- mergeLanes, [49](#)
- mergePreprocessReads, [49](#)
- mergeSummaryAlignment, [50](#)
- parseDCF, [51](#)
- parseSummaries, [51](#)
- picard, [52](#)
- plotDF, [52](#)
- preprocessReads, [53](#)
- preprocessReadsChunk, [53](#)
- processChunks, [54](#)
- readInputFiles, [55](#)
- readRNASeqEnds, [55](#)
- relativeBarPlot, [57](#)
- removeChunkDir, [57](#)
- resource, [58](#)

- rpkm, 58
- runAlignment, 59
- runPreprocessReads, 62
- safe.yield, 62
- safeExecute, 63
- safeGetObject, 63
- safeUnlink, 64
- saveWithID, 65
- sclapply, 65
- setChunkDir, 66
- setUpDirs, 67
- statCountFeatures, 68
- traceMem, 69
- trimReads, 69
- trimTailsByQuality, 70
- truncateReads, 70
- tryKeepTraceback, 71
- updateConfig, 71
- writeAudit, 73
- writeConfig, 73
- writeFastQFiles, 74
- writeFeatureCountsHTML, 74
- writeGenomicFeaturesReport, 75
- writePreprocessAlignHTML, 75
- writePreprocessAlignReport, 76
- writeSummary, 77
- \* **package**
  - HTSeqGenie, 35
- alignReads, 4
- alignReadsChunk, 5
- analyzeVariants, 6
- annotateVariants, 6
- bamCountUniqueReads, 7
- buildConfig, 7
- buildGenomicFeaturesFromTxDb, 8
- buildShortReadReports, 9
- buildTallyParam, 9
- buildTP53FastaGenome, 10
- buildTP53GenomeTemplate, 10
- calculateCoverage, 11
- calculateTargetLengths, 11
- callVariantsGATK, 12
- checkConfig, 12
- checkGATKJar, 13
- checkPicardJar, 13
- computeBamStats, 14
- computeCoverage, 14
- countFeatures, 15
- countGenomicFeatures, 16
- countGenomicFeaturesChunk, 16
- createTmpDir, 17
- detectAdapterContam, 17
- detectQualityInFASTQFile, 18
- detectRRNA, 18
- excludeVariantsByRegions, 19
- FastQStreamer.getReads, 20
- FastQStreamer.init, 20
- FastQStreamer.release, 21
- filterByLength, 22
- filterQuality, 22
- findVariantFile, 23
- gatk, 23
- generateSingleGeneDETs, 24
- getAdapterSeqs, 25
- getBams, 25
- getChunkDirs, 26
- getConfig, 26
- getConfig.integer, 27
- getConfig.logical, 27
- getConfig.numeric, 28
- getConfig.vector, 28
- getEndNumber, 29
- getMemoryUsage, 29
- getNumberOfReadsInFASTQFile, 30
- getNumericVectorDataFromFile, 30
- getObjectFilename, 31
- getPackageFile, 31
- getRandomAlignCutoff, 32
- getRRNAIds, 32
- getTabDataFromFile, 33
- getTraceback, 33
- hashCoverage, 34
- hashVariants, 34
- hashVector, 35
- HTSeqGenie, 35
- initDirs, 36
- initLog, 37
- initLogger, 37
- initPipelineFromConfig, 38
- initPipelineFromSaveDir, 38

- isAboveQualityThresh, 39
- isAdapter, 39
- isConfig, 40
- isFirstFragment, 40
- isSparse, 41
  
- listIterator.init, 42
- listIterator.next, 42
- loadConfig, 43
- logdebug, 43
- logerror, 44
- loginfo, 44
- logwarn, 45
  
- makeDir, 45
- makeRandomSreads, 46
- markDuplicates, 46
- markDups, 47
- mergeAlignReads, 47
- mergeCoverage, 48
- mergeLanes, 49
- mergePreprocessReads, 49
- mergeSummaryAlignment, 50
  
- parseDCF, 51
- parseSummaries, 51
- picard, 52
- plotDF, 52
- preprocessReads, 53
- preprocessReadsChunk, 53
- processChunks, 54
  
- readInputFiles, 55
- readRNASeqEnds, 55
- realignIndels, 56
- realignIndelsGATK, 56
- relativeBarPlot, 57
- removeChunkDir, 57
- resource, 58
- rpkm, 58
- runAlignment, 59
- runPipeline, 60
- runPipelineConfig, 61
- runPreprocessReads, 62
  
- safe.yield, 62
- safeExecute, 63
- safeGetObject, 63
- safeUnlink, 64
  
- saveWithID, 65
- sclapply, 65
- setChunkDir, 66
- setUpDirs, 67
- setUpTestFramework, 67
- statCountFeatures, 68
  
- TP53GenomicFeatures, 68
- traceMem, 69
- trimReads, 69
- trimTailsByQuality, 70
- truncateReads, 70
- tryKeepTraceback, 71
  
- updateConfig, 71
  
- vcfStat, 72
  
- wrap.callVariants, 72
- writeAudit, 73
- writeConfig, 73
- writeFastQFiles, 74
- writeFeatureCountsHTML, 74
- writeGenomicFeaturesReport, 75
- writePreprocessAlignHTML, 75
- writePreprocessAlignReport, 76
- writeSummary, 77
- writeVCF, 77