Package 'HTSeqGenie'

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

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Type Package

LazyLoad yes

Author Gregoire Pau, Jens Reeder

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`alignReads` *Align reads against genome*

Description

Align reads against genome

Usage

```
alignReads()
```

Value

Nothing

Author(s)

Gregoire Pau

`alignReadsChunk` *Genomic alignment*

Description

Genomic alignment using gsnap.

Usage

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

Arguments

`fp1`	Path to FastQ file
`fp2`	Path to second FastQ file if paired end data, NULL if single ended
`save_dir`	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

save_dir	Save directory of a pipeline run
paired_ends	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` *Calculate read coverage*

Description

Calculate read coverage

Usage

```
calculateCoverage()
```

Value

Nothing

Author(s)

Jens Reeder

`calculateTargetLengths` *Plot target length for paired end*

Description

Calculate and plot a histogram of mapped target lengths after trimmming 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 piplein 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/TargetLenghts.[pdf|png]"

Author(s)

Jens Reeder, Melanie Huntley

callVariantsGATK *Variant calling via GATK*

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 *Check configuration*

Description

Performs all configuration checks

Usage

```
checkConfig()
```

Value

Nothing. Individual checks will throw error instead.

checkGATKJar *Check for the GATK jar file*

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 *checkPicardJar*

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 *Count RNA-Seq Pipeline Genomic Features*

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 contaminatino

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

`prefix`	A string that will preceed the directory name
`dir`	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

`lreads`	List of reads as ShortRead objects
`save_dir`	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

`variants`	Variants as Vranges, GRanges or VCF object
`mask`	region to mask, given as GRanges

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_nbreads = 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_nbreads	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` *Filter reads by length*

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` *Filter reads by quality*

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 *Get a vcf filename given a HTSeqGenie directory*

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 *gatk*

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 *Read list of Illumina adapter seqs from package data*

Description

Read list of Illumina adapter seqs from package data

Usage

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

Arguments

paired_ends	Dow 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 *Get bam files of a pipeline run*

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 *Get the list of chunk directories*

Description

Get the list of chunk directories

Usage

```
getChunkDirs()
```

Value

List of chunk directories

Author(s)

Gregoire Pau

getConfig *Get a configuration parameter*

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.
...	Additinal 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 *Load tabular data from the NGS pipeline result directory*

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 ontaining the regular expression matching a filename in dir_path

Value

A data frame.

getTraceback *Get traceback from tryKeepTraceback()*

Description

Get traceback from tryKeepTraceback()

Usage

```
getTraceback(mto)
```

Arguments

mto	An object of the try-error class

Value

Traceback as a string

hashCoverage *Hashing function for coverage*

Description

Hashing function for coverage

Usage

```
hashCoverage(cov)
```

Arguments

cov	A SimpleRleList object

Value

A numeric

Author(s)

Gregoire Pau

hashVariants *Hashing function for variants*

Description

Hashing function for variants

Usage

```
hashVariants(var)
```

Arguments

var	A GRanges object

Value

A numeric

Author(s)

Gregoire Pau

hashVector *Hashing function for vector*

Description

Hashing function for vector

Usage

```
hashVector(x)
```

Arguments

x	A vector

Value

A numeric

Author(s)

Gregoire Pau

HTSeqGenie *Package overview*

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 minquality.

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 *Test the presence of the parameter in the current config*

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 *Does a SAM flag indicate the first fragment*

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

`cov`	A cov object as SimpleRleList
`threshold`	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 *Load configuration file*

Description

Loads the indicated configuration file. Creates and installs a global variable that should be accesssed 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 *Log debug using the logging package*

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 *Log warning using the logging package*

Description

Log warning (with a try statement)

Usage

```
logwarn(msg)
```

Arguments

...	Arguments passed to logging::logwarn

Value

Nothing

Author(s)

Gregoire Pau

makeDir *Make a directory after performing an existence check*

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` *Generate a couple if random ShortReadQ, intended for testing*

Description

Generate a couple if random ShortReadQ, intended for testing

Usage

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

Arguments

`num`	an integer
`len`	an integer

Value

a DNAStringSet

Author(s)

Gregoire Pau

`markDuplicates` *markDuplicates*

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 *Merge coverage files*

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 *Merge input lanes*

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 *Merge after preprocessReads*

Description

Merge detectAdapterContam, merge preprocessed reads, create summary preprocess, build shortReadReport, remove processed

Usage

```
mergePreprocessReads(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

Value

Nothing

Author(s)

Gregoire Pau

---

mergeSummaryAlignment *Merge summary alignments*

Description

Merge summary alignments

Usage

```
mergeSummaryAlignment(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

Value

Nothing

Author(s)

Gregoire Pau

parseDCF *Read and parse a configuration file*

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 *parse summary files from save dirs*

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 *picard*

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 *Make continuous plots of distribution function*

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 containg the path to the preproceesed FastQ files and a few other statistics

---

preprocessReadsChunk *Preprocess a chunk*

Description

Preprocess a chunk

Usage

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

Arguments

lreads	A list of GRanges objects, containing the reads
save_dir	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 *Read FastQ input files*

Description

Uses the global config to find input files

Usage

```
readInputFiles()
```

Value

Reads as list of ShortRead objects

readRNASeqEnds *Read single/paired End Bam Files*

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` *realignIndels*

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` *Realign indels via GATK*

Description

Realing 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` *Make relative bar plots*

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` *Remove chunk directories*

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` *Reload package source code*

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` *Calculate RPKM*

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 perfomred on
nbreads	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` *Run the preprocesing steps of the pipeline*

Description

Runs the preprocesing 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` *Overloaded yield(...) method catching truncated exceptions for FastqStreamer*

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 *Execute function in try catch with trace function*

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 *Safely load a R data file*

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 *safeUnlink*

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 *Save an R object*

Description

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

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 *Scheduled parallel processing*

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 intervall between calls to tracefun

Value

Return value of applied function

---

setChunkDir *Set the base directory for the chunks*

Description

Set the base directory for the chunks

Usage

```
setChunkDir()
```

Value

path to chunk dir

Author(s)

Jens Reeder

setUpDirs — *Create output directory and subdirectories for sequencing pipeline analysis outputs*

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 — *setup test framework*

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` *Compute statistics on count features*

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` *Demo genomic features around the TP53 gene*

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 *Show memory usage*

Description

For debugging purposes only. Show memory usage if config variable

Usage

```
traceMem()
```

Value

Nothing

---

trimReads *Trim/truncate a set of reads*

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` *Trim off low quality tail*

Description

The illuminsa 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 chhose as default here. For illumina 1.5 make sure to set the minqual to 'B'

Value

A list of quality trimmed ShortReadQ objects

`truncateReads` *Trim/truncate a set of reads*

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` *Wrapper around try-catch*

Description

Wrapper around try-catch

Usage

```
tryKeepTraceback(expr)
```

Arguments

`expr`	Expression to evaluate

Value

Result of expression or error if thrown

`updateConfig` *Update the existing config*

Description

Update the existing config

Usage

```
updateConfig(tconfig)
```

Arguments

`tconfig`	List of configuration name value pairs

Value

Nothing.

vcfStat *Compute stats on a VCF file*

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 *Variant calling*

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 *Write Session information*

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 *Write a config file*

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` *Write reads to file*

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` *writeFeatureCountsHTML*

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` *Write HTML summary*

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` *writeVCF*

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

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