# Package 'bamUtils'

January 23, 2018

Title Utility functions for manipulating bams

**Version** 0.0.0.9000

| <b>Description</b> Utility functions for manipulating BAMs   |
|--|
| biocViews  |
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|  |
| R topics documented:   |
| bam.cov.gr       2         bam.cov.tile       3         bamflag       4         bamtag       4         chunk       5         count.clips       5         countCigar       6         get.mate.gr       6         get.pairs.grl       7         hets       7         is.paired.end       8         mafcount       8         oneoffs       9         read.bam       10         splice.cigar       11         varbase       12         varcount       12 |

2 bam.cov.gr

bam.cov.gr

Get coverage as GRanges from BAM on custom set of GRanges

#### **Description**

Gets coverage from BAM in supplied GRanges using 'countBam()', returning GRanges with coverage counts in each of the provided GRanges (different from 'bamUtils::bam.cov()') specified as the columns \$file, \$records, and \$nucleotides in the values field

Basically a wrapper for 'Rsamtools::countBam()' with some standard settings for 'Rsamtools::ScanBamParams()'

#### Usage

```
bam.cov.gr(bam, bai = NULL, intervals = NULL, all = FALSE,
  count.all = FALSE, isPaired = TRUE, isProperPair = TRUE,
  isUnmappedQuery = FALSE, hasUnmappedMate = FALSE,
  isNotPassingQualityControls = FALSE, isDuplicate = FALSE, mc.cores = 1,
  chunksize = 10, verbose = FALSE, ...)
```

#### **Arguments**

bam string Input BAM file. Advisable to make the input BAM a BamFile instance

instead of a plain string, so that the index does not have to be reloaded.

bai string Input BAM index file

intervals GRanges of intervals to retrieve

all boolean Flag to read in all of BAM as a GRanges via 'si2gr(seqinfo())' (default

= FALSE)

isPaired boolean Flag indicates whether unpaired (FALSE), paired (TRUE), or any (NA)

read should be returned. See documentation for Rsamtools::scanBamFlag().

(default == NA)

isProperPair boolean Flag indicates whether improperly paired (FALSE), properly paired

(TRUE), or any (NA) read should be returned. A properly paired read is defined by the alignment algorithm and might, e.g., represent reads aligning to identical reference sequences and with a specified distance. See documentation

for Rsamtools::scanBamFlag(). (default == NA)

isUnmappedQuery

boolean Flag indicates whether unmapped (TRUE), mapped (FALSE), or any (NA) read should be returned. See documentation for Rsamtools::scanBamFlag().

(default == NA)

hasUnmappedMate

boolean Flag indicates whether reads with mapped (FALSE), unmapped (TRUE), or any (NA) mate should be returned. See documentation for Rsamtools::scanBamFlag(). (default == NA)

 $is {\tt NotPassingQualityControls}$ 

boolean Flag indicates whether reads passing quality controls (FALSE), reads not passing quality controls (TRUE), or any (NA) read should be returned. See documentation for Rsamtools::scanBamFlag(). (default == NA)

bam.cov.tile 3

| isDuplicate | boolean Flag indicates that un-duplicated (FALSE), duplicated (TRUE), or any   |
|-------------|--|
|             | (NA) reads should be returned. 'Duplicated' reads may represent PCR or optical duplicates. See documentation for Rsamtools::scanBamFlag(). (default == |
|             | FALSE)   |
| mc.cores    | integer Number of cores in mclapply call   |
| chunksize   | integer How many intervals to process per core (default == 10)   |
| verbose     | boolean "verbose" flag (default == FALSE)  |
|             | futher arguments passed into Rsamtools::scanBamFlag()  |

#### Value

GRanges parallel to input GRanges, but with metadata filled in.

| bam.cov.tile | Get coverage as GRanges from BAM on genome tiles across se- |
|--------------|---|
|              | qlengths of genome  |

## **Description**

Quick way to get tiled coverage via piping to samtools (e.g. ~10 CPU-hours for 100bp tiles, 5e8 read pairs)

Gets coverage for window size "window" [bp], pulling "chunksize" records at a time and incrementing bin corresponding to midpoint or overlaps of corresponding (proper pair) fragment (uses TLEN and POS for positive strand reads that are part of a proper pair)

#### Usage

```
bam.cov.tile(bam.file, window = 100, chunksize = 1e+05, min.mapq = 30,
  verbose = TRUE, max.tlen = 10000, st.flag = "-f 0x02 -F 0x10",
  fragments = TRUE, region = NULL, do.gc = FALSE, midpoint = TRUE)
```

#### **Arguments**

```
string Input BAM file
bam.file
                  integer Window size (in bp) (default == 1e2)
window
                  integer Size of window (default == 1e5)
chunksize
                  integer Minimim map quality reads to consider for counts (default == 30)
min.mapq
verbose
                  boolean "verbose" flag (default == TRUE)
                  max paired-read insert size to consider
max.tlen
                  boolean Samtools flag to filter reads on (default == '-f 0x02 -F 0x10')
st.flag
fragments
                  boolean Flag (default == FALSE)
                  um? (default == NULL)
region
do.gc
                  boolean Flag to execute garbage collection via 'gc()' (default == FALSE)
                  boolean Flag if TRUE will only use the fragment midpoint, if FALSE will count
midpoint
                  all bins that overlap the fragment
```

#### Value

GRanges of "window" bp tiles across seqlengths of bam.file with meta data field \$counts specifying fragment counts centered in the given bin.

4 bamtag

| bamflag Returns matrix of bits from BAM fla |
|---|
|---|

# Description

Shortcut function: assumes reads are GappedAlignments with flag variable or actual integers representing BAM flag

#### Usage

```
bamflag(reads)
```

# Arguments

reads

GenomicRanges or 'GappedAlignments' or data.table holding the reads

# Value

matrix of bits from BAM flags

| bamtag | Outputs a tag to identify duplicate reads in GRanges input |
|--------|--|
|--------|--|

# Description

Outputs a tag that cats 'qname', first vs first second mate +/- secondary alignment +/- gr.string to give an identifier for determine duplicates in a read pile

# Usage

```
bamtag(reads, secondary = FALSE, gr.string = FALSE)
```

# Arguments

| reads     | GenomicRanges or GappedAlignments or data.frame holding the reads |
|-----------|---|
| secondary | boolean including secondary alignment(s) (default == FALSE)       |
| gr.string | boolean input reads into gr.string() (default == FALSE)           |

chunk 5

# Description

Internal function takes same input as seq (from, to, by, length.out) and outputs a 2 column matrix of indices corresponding to "chunks"

#### Usage

```
chunk(from, to = NULL, by = 1, length.out = NULL)
```

#### **Arguments**

from integer where to begin sequence

to integer to end sequence by interval to space sequence

length.out number of desired chunks, i.e. nrows of output matrix

#### Value

2 column matrix of indices, each row representing chunk

#### Author(s)

Marcin Imielinski

count.clips

Return data.frame with fields of "right" soft clips and "left" soft clips

#### **Description**

Takes GRanges or GappedAlignments object and uses cigar field (or takes character vector of cigar strings) and returns data.frame with fields (for character input) \$right.clips number of "right" soft clips (e.g. cigar 89M12S) \$left.clips number of "left" soft clips (e.g. cigar 12S89M), or appends these fields to the reads object

# Usage

```
count.clips(reads)
```

#### **Arguments**

reads GenomicRanges or GappedAlignments or data.frame or data.table holding the

reads

# Value

GRanges with 'right.clips' and 'left.clips' columns added

6 get.mate.gr

countCigar

Count bases in cigar string

# Description

Counts the total number of bases, per cigar, that fall into D, I, M, S categories. countCigar makes no distinction between, for instance 1S2M2S, 2S2M1S, or 3S2M

# Usage

```
countCigar(cigar)
```

#### **Arguments**

cigar

character vector of cigar strings

#### Value

matrix of dimensions (4-column, length(cigar)) with the total counts for each type

get.mate.gr

returns GRanges corresponding to mates of reads

# Description

Inputs GRanges or data.frame/data.table of reads. Outputs GRanges corresponding to mates of reads.

# Usage

```
get.mate.gr(reads)
```

#### **Arguments**

reads

GRanges or data.table/data.frame Input reads

# Value

GRanges corresponding to mates of reads

get.pairs.grl 7

|               | C . CD               | C DA14            | COD                  |
|---------------|----------------------|-------------------|----------------------|
| get.pairs.grl | Get coverage as GRan | ges from BAM on c | ustom set of GRanges |

#### **Description**

Takes reads object and returns GRangesList with each read and its mate (if exists)

#### Usage

```
get.pairs.grl(reads, pairs.grl.split = TRUE, verbose = FALSE)
```

#### **Arguments**

hets Simple het "caller" meant to be used at validated het SNP sites for tumor / normal pairs

# Description

hets dumps a tsv file of alt (\$alt.count.t, \$alt.count.n) and ref (\$ref.count.t, \$ref.count.n) read counts to out.file for a tumor / normal pair across a set of sites specified by an input VCF

#### Usage

```
hets(tum.bam, norm.bam = NULL, out.file, vcf.file, chunk.size1 = 1000,
    chunk.size2 = 100, mc.cores = 14, verbose = T, na.rm = TRUE,
    filt.norm = T)
```

#### **Arguments**

| tum.bam     | character scalar or BamFile of tumor bam                                       |
|-------------|--|
| norm.bam    | character scalar or BamFile of normal bam (optional)                           |
| out.file    | path to TSV output file to be generated  |
| vcf.file    | VCF file of sites (eg hapmap or 1000G) at which to compute read counts         |
| chunk.size1 | number of variants to process from VCF file at a time                          |
| chunk.size2 | number of variants to access from BAM file in a single iteration               |
| mc.cores    | how many cores to parallelize  |
| verbose     | verbose logical flag   |
| na.rm       | logical flag to remove rows with NA counts                                     |
| filt.norm   | logical flag remove any sites that have allele fraction of 0 or 1 or NA in MAF |

8 mafcount

#### Value

nil

#### Author(s)

Marcin Imielinski

is.paired.end

Check if BAM file is paired end by using 0x1 flag

#### **Description**

Check if BAM file is paired-end by using 0x1 flag, pipes to 'samtools' via command line Create GRanges of read mates from reads

### Usage

```
is.paired.end(bams)
get.mate.gr(reads)
```

#### **Arguments**

bams

vector of input BAMs

#### Value

boolean returns TRUE if BAM file is paired-end, returns FALSE if BAM not paired-end GRanges corresponding to mates of reads

mafcount

Wrapper around varcount adapted to tumor and normal "paired" bams

#### **Description**

mafcount

Returns base counts for reference and alternative allele for an input tum and norm bam and maf data frame or GRAnges specifying substitutions

maf is a single width GRanges describing variants and field 'ref' (or 'Reference\_Allele'), 'alt' (or 'Tum\_Seq\_Allele1') specifying reference and alt allele. maf is assumed to have width 1 and strand is ignored.

# Usage

```
mafcount(tum.bam, norm.bam = NULL, maf, chunk.size = 100, verbose = T,
    mc.cores = 1, ...)
```

oneoffs 9

## **Arguments**

| tum.bam    | character scalar or BamFile specifying path to tumor sample                              |
|------------|--|
| norm.bam   | optional character scalar or BamFile specifying path to normal sample                    |
| maf        | GRanges or data.frame or data.table of imported maf (i.e. output of read.delim or fread) |
| chunk.size | integer number of variants to extract from bam file at each iteration                    |
| verbose    | logical flag whether to print verbose output   |
| mc.cores   | number of cores to parallelize   |
|            | additional params to pass to varcount  |

# Value

GRanges of maf annotated with fields \$alt.count.t, \$ref.count.t, \$alt.count.n, \$ref.count.n

#### Author(s)

Marcin Imielinski

| oneoffs | Calls samtools mpileup to dump tsv of "one off" variants / sites (i.e. that are present in exactly one read per site) |
|---------|---|
|         |   |

#### **Description**

Calls samtools mpileup to dump tsv of "one off" variants / sites (i.e. that are present in exactly one read per site)

# Usage

```
oneoffs(out.file, bam, ref, min.bq = 30, min.mq = 60, indel = FALSE,
    chunksize = 10000, verbose = TRUE)
```

# **Arguments**

| out.file  | file to dump tsv to  |
|-----------|--|
| bam       | bam file path  |
| ref       | fasta path   |
| min.bq    | integer minimum base quality   |
| min.mq    | integer minimum mapping quality  |
| indel     | logical flag whether to collect one off indels (default is substitution) |
| chunksize | number of mpileup lines to put into memory                               |
| verbose   | logical flag   |

#### Note

The denominator (ie total reads) is just the sum of counts\$records

10 read.bam

read.bam

Read BAM file into GRanges or data.table

#### **Description**

Wrapper around Rsamtools bam scanning functions, by default, returns GRangesList of read pairs for which <at least one> read lies in the supplied interval

#### Usage

```
read.bam(bam, intervals = NULL, gr = intervals, all = FALSE, bai = NULL,
  pairs.grl = TRUE, stripstrand = TRUE, what = scanBamWhat(),
  unpack.flag = FALSE, verbose = FALSE, tag = NULL, isPaired = NA,
  isProperPair = NA, isUnmappedQuery = NA, hasUnmappedMate = NA,
  isNotPassingQualityControls = NA, isDuplicate = F,
  isValidVendorRead = TRUE, pairs.grl.split = TRUE, as.data.table = FALSE,
  ignore.indels = FALSE, ...)
```

#### **Arguments**

bam Input bam file. Advisable to make "bam" a BamFile instance instead of a plain

string, so that the index does not have to be reloaded.

intervals GRanges of intervals to retrieve gr GRanges of intervals to retrieve

stripstrand Flag to ignore strand information on the query intervals. Default TRUE what What fields to pull down from BAM. Default scanBamWhat ()

what helds to pair down from Britis. Belaut Scambanwilde

 $\verb"unpack.flag" Add features corresponding to read flags. Default FALSE$ 

verbose Increase verbosity

tag Additional tags to pull down from the BAM (e.g. 'R2') isPaired See documentation for scanBamFlag. Default NA isProperPair See documentation for scanBamFlag. Default NA

isUnmappedQuery

See documentation for scanBamFlag. Default NA

has Unmapped Mate

See documentation for  ${\tt scanBamFlag}.$  Default NA

isNotPassingQualityControls

See documentation for scanBamFlag. Default NA

 $\verb|isDuplicate| See documentation for \verb|scanBamFlag|. Default FALSE|$ 

isValidVendorRead

See documentation for scanBamFlag. Default TRUE

as.data.table

Return reads in the form of a data.table rather than GRanges/GRangesList

ignore.indels

messes with cigar to read BAM with indels removed. Useful for breakpoint

mapping on contigs

... passed to scanBamFlag

bami Input bam index file.

as.grl Return reads as GRangesList. Controls whether get.pairs.grl does split.

Default TRUE

splice.cigar 11

#### Value

Reads in one of GRanges, GRangesList or data.table

| splice.cigar Get coverage as GRanges from BAM on custom set of GRanges |
|--|
|--|

#### Description

Takes GRanges or GappedAlignments object "reads" and parses cigar fields to return GRanges or GRangesList corresponding to spliced alignments on the genome, which correspond to portions of the cigar

i.e. each outputted GRanges/GRangesList element contains the granges corresponding to all non-N portions of cigar string

If GRangesList provided as input (e.g. paired reads) then all of the spliced ranges resulting from each input GRangesList element will be put into the corresponding output GRangesList element

NOTE: does not update MD tag

If use.D = TRUE, then will treat "D" flags (deletion) in addition to "N" flags as indicative of deletion event.

# Usage

```
splice.cigar(reads, verbose = TRUE, fast = TRUE, use.D = TRUE,
rem.soft = TRUE, get.seq = FALSE, return.grl = TRUE)
```

# Arguments

| reads      | GenomicRanges or GappedAlignments or data.frame input reads   |
|------------|---|
| verbose    | boolean verbose flag (default == TRUE)  |
| fast       | boolean Flag to use 'GenomicAlignments::cigarRangesAlongReferenceSpace()' to translate CIGAR to GRanges (default == TRUE) |
| use.D      | boolean Treats "D" tags as deletions, along with "N" tags (default == TRUE)   |
| rem.soft   | boolean Pick up splice 'S', soft-clipped (default == TRUE)  |
| get.seq    | boolean Get InDels (default == TRUE)  |
| return.grl | boolean Return as GRangesList (default == TRUE)   |

12 varcount

| varbase | Returns variant bases and ranges from GRanges or GappedAlignments input |
|---------|---|
|         |   |

## Description

Takes GRanges or GappedAlignments object "reads" and uses cigar, MD, seq fields to return variant bases and ranges

Teturns GRangesList (of same length as input) of variant base positions with character vector \$varbase field populated with variant bases for each GRanges item in grl[[k]], with the following handling for insertions, deletions, and substitution GRange's:

Substitutions: nchar(gr\$varbase) = width(gr) of the corresponding var Insertions: nchar(gr\$varbase)>=1, width(gr) ==0 Deletions: gr\$varbase = ", width(gr)>=1

Each GRanges also has type flag which shows the cigar string code for the event i.e. <math>S = soft clip -> varbase represents clipped bases I = insertion -> varbase represents inserted bases <math>D = deletion -> varbase is empty X = mismatch -> varbase represents mismatched bases

#### Usage

```
varbase(reads, soft = TRUE, verbose = TRUE)
```

#### **Arguments**

| reads   | GenomicRanges or GRangesList or GappedAlignments or data.frame/data.table reads to extract variants from |
|---------|--|
| soft    | boolean Flag to include soft-clipped matches (default == TRUE)   |
| verbose | boolean verbose flag (default == TRUE)   |

varcount

Wrapper around applyPileups

# Description

takes in vector of bam paths, GRanges corresponding to sites / territories to query, and outputs a list with fields \$counts = 3D matrix of base counts (A, C, G, T, N) x sites x bams subject to mapq and baseq thresholds #'

```
(uses varbase)
... = other args go to read.bam
```

## Usage

```
varcount(bams, gr, min.mapq = 0, min.baseq = 20, max.depth = 500,
indel = F, ...)
```

varcount 13

## **Arguments**

bams character vector of paths to bam files

granges of (width 1) sites ie intervals at which to compute base coujnts

 $\verb|min.mapq| \qquad \qquad \verb|minimal mapping quality at which to compute bases|$ 

max.depth max read depth to consider

indel logical flag whether to consider indels (default FALSE)

max.baseq minimal base qualitya t which to compute bases

#### Value

input GRanges gr annotated with fields \$alt.count.t, \$ref.count.t, \$alt.count.n, \$ref.count.n

# Author(s)

Marcin Imielinski

# **Index**

```
bam.cov.gr, 2
bam.cov.tile, 3
bamflag, 4
bamtag, 4
chunk, 5
count.clips,5
countCigar, 6
get.mate.gr, 6
get.mate.gr(is.paired.end), 8
get.pairs.grl,7
hets, 7
is.paired.end, 8
mafcount, 8
oneoffs, 9
read.bam, 10
splice.cigar, 11
varbase, 12
{\tt varcount}, {\color{red} 12}
```