# Package 'STRMPS'

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Type Package

**Title** Analysis of Short Tandem Repeat (STR) Massively Parallel Sequencing (MPS) Data

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**Description** Loading, identifying, aggregating, manipulating, and analysing short tandem repeat regions of massively parallel sequencing data in forensic genet

gions of massively parallel sequencing data in forensic genet-

ics. 'STRMPS' can work with the package 'STRaitRazoR' (an R interface to the 'STRaitRazor' commandline tool) for added speed. 'STRaitRa-

zoR' only works on linux and can found at <a href="https://doi.org/10.1007/j.je/10.2007/j.je/10.

//github.com/svilsen/STRaitRazoR>. The analyses and framework imple-

mented in this package relies on the pa-

pers of Vilsen et al. (2017) <doi:10.1016/j.fsigen.2017.01.017> and Vilsen et al. (2018) <doi:10.1016/j.fsigen.2018.04.003>

allelisation in the package relies on mclapply() and, thus, speed-

ups will only be seen on UNIX based systems.

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LazyData TRUE

**Depends** R (>= 3.1.0), Biostrings, ShortRead

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BLMM 3

**BLMM** 

Block length of the missing motif.

# **Description**

Given a motif length and a string it finds the blocks of the string.

# Usage

```
BLMM(s, motifLength = 4, returnType = "numeric")
```

### **Arguments**

s a string of either class: 'character' or 'DNAString'.

motifLength the known motif length of the STR region.

returnType the type of return wanted. Takes three values 'numeric', 'string', or 'fullList' (or any other combination cased letters).

#### **Details**

If returnType is 'numeric', the function returns the numeric value of the LUS. If returnType is instead chosen as 'string', the function returns "[AATG]x" i.e. the motif, AATG, is repeated 'x' times. Lastly if the returnType is set to fullList, the function returns a list of data.frames containing every possible repeat structure their start and the numeric value of the repeat unit length.

# Value

Depending on returnType it return an object of class 'numeric', 'string', or 'fulllist'.

```
# Creating compound string 's'
stretch1 = paste0(rep("AATG", 10), collapse = "")
stretch2 = paste0(rep("ATCG", 4), collapse = "")
s = paste0(stretch1, stretch2)

# Return BLMM only
BLMM(s, motifLength = 4, returnType = "numeric")

# Return BLMM and motif of stretch
BLMM(s, motifLength = 4, returnType = "string")

# Return all blocks of 's'
BLMM(s, motifLength = 4, returnType = "fulllist")
```

extractedReadsList-class

Extract STR region information

### **Description**

Identifies the marker of the read using flanking regions and trims the read to include what is between the flanking regions.

extractedReadsListCombined-class

Combined extract STR region information.

# **Description**

Identifies the marker of the read for both the provided and reverse complement flanking regions. The resulting lists are then combined into a single list.

extractedReadsListNonCombined-class

Combined extract STR region information.

# **Description**

Identifies the marker of the read for both the provided and reverse complement flanking regions.

extracted Reads List Reverse Complement-class

Extract STR region information of the reverse complement DNA strand.

# **Description**

Identifies the marker of the read using reverse complement flanking regions and trims the read to include what is between the flanking regions.

findNeighbours 5

findNeighbours

Find neighbours

# Description

Generic function for finding neighbouring strings, given identified alleles.

# Usage

```
findNeighbours(stringCoverageGenotypeListObject, searchDirection,
    trace = FALSE)
```

# Arguments

 ${\it string} Coverage Genotype List Object$ 

A stringCoverageGenotypeList-class object.

searchDirection

The direction to search for neighbouring strings. Default is -1, indicating a

search for '-1' stutters.

trace

Should a trace be shown?

### Value

A 'neighbourList' with the neighbouring strings, in the specified direction, for the identified allele regions.

```
\label{lem:coverageGenotypeList-method} Find \textit{Neighbours}, \textit{stringCoverageGenotypeList-method}
```

### **Description**

Generic function for finding neighbouring strings, given identified alleles.

# Usage

```
## S4 method for signature 'stringCoverageGenotypeList'
findNeighbours(stringCoverageGenotypeListObject,
   searchDirection = -1, trace = FALSE)
```

6 findStutter

### **Arguments**

stringCoverageGenotypeListObject

A stringCoverageGenotypeList-class object.

searchDirection

The direction to search for neighbouring strings. Default is -1, indicating a

search for '-1' stutters.

trace Should a trace be shown?

### Value

A 'neighbourList' with the neighbouring strings, in the specified direction, for the identified allele regions.

findStutter

Find stutters

# **Description**

Given identified alleles it search for '-1' stutters of the alleles.

### **Usage**

```
findStutter(stringCoverageGenotypeListObject, trace = FALSE)
```

### **Arguments**

```
{\it string} Coverage Genotype List Object
```

A stringCoverageGenotypeList-class object.

trace Should a trace be shown?

### Value

A 'neighbourList' with the stutter strings for the identified allele regions.

```
# The object returned by merging a stringCoverageList-Object
# and a genotypeList-Object.
data("stringCoverageGenotypeList")

stutterList <- findStutter(stringCoverageGenotypeList)
stutterTibble <- subset(do.call("rbind", stutterList), !is.na(Genotype))

stutterTibble$BlockLengthMissingMotif
stutterTibble$NeighbourRatio</pre>
```

 $find {\tt Stutter}, {\tt string Coverage Genotype List-method} \\ Find {\tt stutters}$ 

# **Description**

Given identified alleles it search for '-1' stutters of the alleles.

# Usage

```
## S4 method for signature 'stringCoverageGenotypeList'
findStutter(stringCoverageGenotypeListObject,
    trace = FALSE)
```

# **Arguments**

```
stringCoverageGenotypeListObject
A stringCoverageGenotypeList-class object.

trace Should a trace be shown?
```

#### Value

A 'neighbourList' with the stutter strings for the identified allele regions.

## **Examples**

```
# The object returned by merging a stringCoverageList-Object
# and a genotypeList-Object.
data("stringCoverageGenotypeList")

stutterList <- findStutter(stringCoverageGenotypeList)
stutterTibble <- subset(do.call("rbind", stutterList), !is.na(Genotype))

stutterTibble$BlockLengthMissingMotif
stutterTibble$NeighbourRatio</pre>
```

flankingRegions

Flanking regions

# **Description**

The flanking regions searched for to identify the markers and STR regions of all autosomal/X/Y STR's in the Illumina ForenSeq prep kit.

8 genotypeList

### **Usage**

```
data("flankingRegions")
```

# **Format**

A tibble containing the flanks (forward and reverse), motif, motif length, adjustment need to make it compatible with CE, and the shifts needed for further trimming, for each marker

# Author(s)

```
Søren B. Vilsen <svilsen@math.aau.dk>
```

```
\begin{tabular}{ll} genotype I dentified List-class \\ Genotype \ list \\ \end{tabular}
```

# Description

A reduced stringCoverageList restricted to the identified genotypes.

genotypeList

Genotype list

# **Description**

The identified genotypes of the stringCoverageList data, created by the getGenotype function.

# Usage

```
data("genotypeList")
```

### **Format**

A list of tibble's one for each of the 10 markers, showing which strings are the potential alleles based on the 'Coverage'.

# Author(s)

Søren B. Vilsen <svilsen@math.aau.dk>

getGenotype 9

### **Description**

getGenotype takes an stringCoverageList-object, assumes the sample is a reference file and assings a genotype, based on a heterozygote threshold, for every marker in the provided list.

# Usage

```
getGenotype(stringCoverageListObject, colBelief = "Coverage",
    thresholdSignal = 0, thresholdHeterozygosity = 0.35,
    thresholdAbsoluteLowerLimit = 1)
```

# **Arguments**

```
stringCoverageListObject
an stringCoverageList-object, created using the stringCoverage-function.

colBelief the name of the coloumn used for identification.

thresholdSignal
threshold applied to the signal (generally the coverage) of every string.

thresholdHeterozygosity
threshold used to determine whether a marker is hetero- or homozygous.

thresholdAbsoluteLowerLimit
a lower limit on the coverage for it to be called as an allele.
```

## Value

Returns a list, with an element for every marker in stringCoverageList-object, each element contains the genotype for a given marker.

```
# Strings aggregated by 'stringCoverage()'
data("stringCoverageList")
getGenotype(stringCoverageList)
```

```
\label{eq:coverageList-method} {\it Assigns genotype}.
```

# Description

getGenotype takes an stringCoverageList-object, assumes the sample is a reference file and assings a genotype, based on a heterozygote threshold, for every marker in the provided list.

## Usage

```
## S4 method for signature 'stringCoverageList'
getGenotype(stringCoverageListObject,
  colBelief = "Coverage", thresholdSignal = 0,
  thresholdHeterozygosity = 0.35, thresholdAbsoluteLowerLimit = 1)
```

# **Arguments**

```
stringCoverageListObject
an stringCoverageList-object, created using the stringCoverage-function.

colBelief the name of the coloumn used for identification.

thresholdSignal
threshold applied to the signal (generally the coverage) of every string.

thresholdHeterozygosity
threshold used to determine whether a marker is hetero- or homozygous.

thresholdAbsoluteLowerLimit
a lower limit on the coverage for it to be called as an allele.
```

## Value

Returns a list, with an element for every marker in stringCoverageList-object, each element contains the genotype for a given marker.

```
# Strings aggregated by 'stringCoverage()'
data("stringCoverageList")
getGenotype(stringCoverageList)
```

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identifiedSTRs

Identified STR regions

### Description

The identified STR regions of the sampleSequences.fastq file, created by the identifySTRRegions function.

### Usage

```
data("identifiedSTRs")
```

#### **Format**

A list with an element for each of the 10 identified markers indicating which sequences were identified for each marker.

# Author(s)

Søren B. Vilsen <svilsen@math.aau.dk>

identifyNoise

Idenfities the noise.

# **Description**

identifyNoise takes an stringCoverageList-object and identifies the noise based on a signal threshold for every marker in the provided list.

# Usage

```
identifyNoise(stringCoverageListObject, colBelief = "Coverage",
    thresholdSignal = 0.01)
```

### **Arguments**

stringCoverageListObject

an stringCoverageList-object, created using the stringCoverage-function.

colBelief the name of the coloumn used for identification.

thresholdSignal

threshold applied to the signal (generally the coverage) of every string.

### Value

Returns a list, with an element for every marker in stringCoverageList-object, each element contains the genotype for a given marker.

### **Examples**

```
# Strings aggregated by 'stringCoverage()'
data("stringCoverageList")
identifyNoise(stringCoverageList, thresholdSignal = 0.03)
```

```
identifyNoise, stringCoverageList-method Idenfities the noise.
```

# **Description**

identifyNoise takes an stringCoverageList-object and identifies the noise based on a signal threshold for every marker in the provided list.

### Usage

```
## S4 method for signature 'stringCoverageList'
identifyNoise(stringCoverageListObject,
  colBelief = "Coverage", thresholdSignal = 0.01)
```

### **Arguments**

```
stringCoverageListObject
an stringCoverageList-object, created using the stringCoverage-function.

colBelief the name of the coloumn used for identification.

thresholdSignal
threshold applied to the signal (generally the coverage) of every string.
```

# Value

Returns a list, with an element for every marker in stringCoverageList-object, each element contains the genotype for a given marker.

```
# Strings aggregated by 'stringCoverage()'
data("stringCoverageList")
identifyNoise(stringCoverageList, thresholdSignal = 0.03)
```

identifySTRRegions 13

identifySTRRegions

*Identify the STR regions of a fastq-file or ShortReadQ-object.* 

### Description

identifySTRRegions takes a fastq-file location or a ShortReadQ-object and identifies the STR regions based on a directly adjacent flanking regions. The function allows for mutation in the flanking regions through the numberOfMutation argument.

# Usage

```
identifySTRRegions(reads, flankingRegions, numberOfMutation, control)
```

### **Arguments**

reads either a fastq-file location or a ShortReadQ-object

flankingRegions

containing marker ID/name, the directly adjacent forward and reverse flanking

regions, used for identification.

numberOfMutation

the maximum number of mutations (base-calling errors) allowed during flanking

region identification.

control an identifySTRRegions.control-object.

### Value

The returned object is a list of lists. If the reverse complement strings are not included or if the control\$combineLists == TRUE, a list, contains lists of untrimmed and trimmed strings for each row in flankingRegions. If control\$combineLists == FALSE, the function returns a list of two such lists, one for forward strings and one for the reverse complement strings.

```
library("Biostrings")
library("ShortRead")

# Path to file
readPath <- system.file('extdata', "sampleSequences.fastq", package = 'STRMPS')

# Flanking regions
data("flankingRegions")

# Read the file into memory
readFile <- readFastq(readPath)
sread(readFile)
quality(readFile)

# Identify the STR's of the file, both readPath and readFile can be used.</pre>
```

identifySTRRegions, character-method

Identify the STR regions of a fastq-file or ShortReadQ-object.

# **Description**

identifySTRRegions takes a fastq-file location or a ShortReadQ-object and identifies the STR regions based on a directly adjacent flanking regions. The function allows for mutation in the flanking regions through the numberOfMutation argument.

# Usage

```
## S4 method for signature 'character'
identifySTRRegions(reads, flankingRegions,
   numberOfMutation = 1, control = identifySTRRegions.control())
```

## **Arguments**

reads path to fastq-file.

flankingRegions

containing marker ID/name, the directly adjacent forward and reverse flanking regions, used for identification.

numberOfMutation

the maximum number of mutations (base-calling errors) allowed during flanking region identification.

control an identifySTRRegions.control-object.

### Value

The returned object is a list of lists. If the reverse complement strings are not included or if the control\$combineLists == TRUE, a list, contains lists of untrimmed and trimmed strings for each row in flankingRegions. If control\$combineLists == FALSE, the function returns a list of two such lists, one for forward strings and one for the reverse complement strings.

### **Examples**

```
library("Biostrings")
library("ShortRead")
# Path to file
readPath <- system.file('extdata', "sampleSequences.fastq", package = 'STRMPS')</pre>
# Flanking regions
data("flankingRegions")
# Read the file into memory
readFile <- readFastq(readPath)</pre>
sread(readFile)
quality(readFile)
# Identify the STR's of the file, both readPath and readFile can be used.
identifySTRRegions(reads = readFile, flankingRegions = flankingRegions,
                   numberOfMutation = 1,
                   control = identifySTRRegions.control(
                       numberOfThreads = 1,
                       includeReverseComplement = FALSE)
                   )
```

identifySTRRegions,ShortReadQ-method

Identify the STR regions of a fastq-file or ShortReadQ-object.

## Description

identifySTRRegions takes a fastq-file location or a ShortReadQ-object and identifies the STR regions based on a directly adjacent flanking regions. The function allows for mutation in the flanking regions through the numberOfMutation argument.

# Usage

```
## S4 method for signature 'ShortReadQ'
identifySTRRegions(reads, flankingRegions,
   numberOfMutation = 1, control = identifySTRRegions.control())
```

# **Arguments**

```
reads a ShortReadQ-object flankingRegions
```

containing marker ID/name, the directly adjacent forward and reverse flanking regions, used for identification.

numberOfMutation

the maximum number of mutations (base-calling errors) allowed during flanking region identification.

control

an identifySTRRegions.control-object.

### Value

The returned object is a list of lists. If the reverse complement strings are not included or if the control\$combineLists == TRUE, a list, contains lists of untrimmed and trimmed strings for each row in flankingRegions. If control\$combineLists == FALSE, the function returns a list of two such lists, one for forward strings and one for the reverse complement strings.

# **Examples**

```
library("Biostrings")
library("ShortRead")
# Path to file
readPath <- system.file('extdata', "sampleSequences.fastq", package = 'STRMPS')
# Flanking regions
data("flankingRegions")
# Read the file into memory
readFile <- readFastq(readPath)</pre>
sread(readFile)
quality(readFile)
# Identify the STR's of the file, both readPath and readFile can be used.
identifySTRRegions(reads = readFile, flankingRegions = flankingRegions,
                   numberOfMutation = 1,
                   control = identifySTRRegions.control(
                       numberOfThreads = 1,
                       includeReverseComplement = FALSE)
                   )
```

identifySTRRegions.control

Control function for identifySTRRegions

## **Description**

A list containing default parameters passed to the identifySTRRegions function.

# Usage

```
identifySTRRegions.control(colList = NULL, numberOfThreads = 4L,
  reversed = TRUE, includeReverseComplement = TRUE, combineLists = TRUE,
  removeEmptyMarkers = TRUE, matchPatternMethod = "mclapply")
```

### **Arguments**

colList The position of the forward, reverse, and motifLength columns in the flanking

region tibble/data.frame. If 'NULL' a function searches for the words 'forward',

'reverse', and 'motif' ot identify the columns.

numberOfThreads

The number of threads used by mclapply (stuck at '2' on windows).

reversed TRUE/FALSE: In a revrse complementary run, should the strings/quality be

reversed (recommended)?

includeReverseComplement

TRUE/FALSE: Should the function also search for the reverse complement

DNA strand (recommended)?

combineLists TRUE/FALSE: If 'includeReverseComplement' is TRUE, should the sets be

combined?

removeEmptyMarkers

TRUE/FALSE: Should markers returning no identified regions be removed?

matchPatternMethod

Which method should be used to identify the flanking regions (only 'mclapply'

implemented at the moment)?

#### Value

A control list setting default behaviour.

 ${\tt mergeGenotypeStringCoverage}$ 

Merge genotypeIdentifiedList and stringCoverageList.

#### **Description**

mergeGenotypeStringCoverage merges genotypeIdentifiedList-objects and stringCoverageList-objects.

# Usage

```
mergeGenotypeStringCoverage(stringCoverageListObject,
  noiseGenotypeIdentifiedListObject)
```

#### **Arguments**

stringCoverageListObject

a stringCoverageList-object, created using the stringCoverage-function.

 ${\tt noiseGenotypeIdentifiedListObject}$ 

a noiseGenotypeIdentifiedList-object, created using the getGenotype-function.

#### Value

Returns a list, with an element for every marker in extractedReadsList-object, each element contains the string coverage of all unique strings of a given marker.

### **Examples**

```
# Strings aggregated by 'stringCoverage()'
data("stringCoverageList")
# Genotypes identified by 'getGenotype()'
data("genotypeList")
# Noise identified by 'identifyNoise()'
data("noiseList")

mergeGenotypeStringCoverage(stringCoverageList, genotypeList)
mergeNoiseStringCoverage(stringCoverageList, noiseList)
```

mergeGenotypeStringCoverage,genotypeIdentifiedList-method

Merge genotypeIdentifiedList and stringCoverageList.

# **Description**

 $merge Genotype String Coverage \ merges \ genotype Identified List-objects \ and \ string Coverage List-objects.$ 

# Usage

```
## S4 method for signature 'genotypeIdentifiedList'
mergeGenotypeStringCoverage(stringCoverageListObject,
    noiseGenotypeIdentifiedListObject)
```

### **Arguments**

```
stringCoverageListObject
a stringCoverageList-object, created using the stringCoverage-function.
noiseGenotypeIdentifiedListObject
a noiseGenotypeIdentifiedList-object, created using the getGenotype-function.
```

#### Value

Returns a list, with an element for every marker in extractedReadsList-object, each element contains the string coverage of all unique strings of a given marker.

### **Examples**

```
# Strings aggregated by 'stringCoverage()'
data("stringCoverageList")
# Genotypes identified by 'getGenotype()'
data("genotypeList")
# Noise identified by 'identifyNoise()'
data("noiseList")

mergeGenotypeStringCoverage(stringCoverageList, genotypeList)
mergeNoiseStringCoverage(stringCoverageList, noiseList)
```

mergeNoiseStringCoverage

Merge noiseIdentifiedList and stringCoverageList.

# **Description**

mergeNoiseStringCoverage merges noiseIdentifiedList-objects and stringCoverageList-objects.

### Usage

```
mergeNoiseStringCoverage(stringCoverageListObject,
    noiseGenotypeIdentifiedListObject)
```

#### **Arguments**

```
stringCoverageListObject
a stringCoverageList-object, created using the stringCoverage-function.
noiseGenotypeIdentifiedListObject
a noiseGenotypeIdentifiedList-object, created using the identifyNoise-function.
```

### Value

Returns a list, with an element for every marker in extractedReadsList-object, each element contains the string coverage of all unique strings of a given marker.

```
# Strings aggregated by 'stringCoverage()'
data("stringCoverageList")
# Genotypes identified by 'getGenotype()'
data("genotypeList")
# Noise identified by 'identifyNoise()'
data("noiseList")

mergeGenotypeStringCoverage(stringCoverageList, genotypeList)
mergeNoiseStringCoverage(stringCoverageList, noiseList)
```

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```
\label{lem:mergeNoiseStringCoverage,noiseIdentifiedList-method} Merge\ noiseIdentifiedList\ and\ stringCoverageList.
```

### **Description**

mergeNoiseStringCoverage merges noiseIdentifiedList-objects and stringCoverageList-objects.

### **Usage**

```
## S4 method for signature 'noiseIdentifiedList'
mergeNoiseStringCoverage(stringCoverageListObject,
    noiseGenotypeIdentifiedListObject)
```

# Arguments

```
stringCoverageListObject
a stringCoverageList-object, created using the stringCoverage-function.
noiseGenotypeIdentifiedListObject
a noiseGenotypeIdentifiedList-object, created using the identifyNoise-function.
```

#### Value

Returns a list, with an element for every marker in extractedReadsList-object, each element contains the string coverage of all unique strings of a given marker.

## **Examples**

```
# Strings aggregated by 'stringCoverage()'
data("stringCoverageList")
# Genotypes identified by 'getGenotype()'
data("genotypeList")
# Noise identified by 'identifyNoise()'
data("noiseList")

mergeGenotypeStringCoverage(stringCoverageList, genotypeList)
mergeNoiseStringCoverage(stringCoverageList, noiseList)
```

 ${\tt neighbourList-class} \qquad \textit{A neighbour list}$ 

# **Description**

A list of the identified neightbours of the called alleles in a stringCoverageGenotypeList

noiseIdentifiedList-class 21

noiseIdentifiedList-class

Noise list

# Description

Creates a flag to the sequences in a stringCoverageList which cloud be classified as noise.

noiseList

Noise list

# **Description**

The identified noise of the stringCoverageList data, created by the identifyNoise function.

# Usage

```
data("noiseList")
```

#### **Format**

A list of tibble's one for each of the 10 markers, showing which strings can be safely classified as noise based on the 'Coverage'.

# Author(s)

Søren B. Vilsen <svilsen@math.aau.dk>

```
phredQualityProbability
```

Quality score to probability

# Description

Converts a quality score (Phred or Solexa) to a probability of error.

# Usage

```
phredQualityProbability(q)
solexaQualityProbability(q)
```

### **Arguments**

q Quality score.

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# Value

 $phred Quality Score (q\_phred) \ and \ solexa Quality Score (q\_solexa) \ returns \ a \ probability \ of \ error.$ 

# **Examples**

```
q_phred = phredQualityScore(1e-3)
q_solexa = solexaQualityScore(1e-3)
phredQualityProbability(q_phred)
solexaQualityProbability(q_solexa)
```

phredQualityScore

Convert probability to quality score

# Description

Calculates the quality score (Phred or Solexa) given a probability of error.

### Usage

```
phredQualityScore(p)
solexaQualityScore(p)
```

# Arguments

р

Probability of error.

# Value

```
phredQualityScore(p) returns a Phred quality score.
solexaQualityScore(p) returns a Solexa quality score.
```

```
p <- 1e-3
phredQualityScore(p)
solexaQualityScore(p)</pre>
```

stringCoverage 23

stringCoverage

Get string coverage STR identified objects.

# **Description**

stringCoverage takes an extractedReadsList-object and finds the coverage of every unique string for every marker in the provided list.

# Usage

```
stringCoverage(extractedReadsListObject, control = stringCoverage.control())
```

# **Arguments**

```
extractedReadsListObject an extractedReadsList-object, created using the identifySTRRegions-function.

control an stringCoverage.control-object.
```

#### Value

Returns a list, with an element for every marker in extractedReadsList-object, each element contains the string coverage of all unique strings of a given marker.

```
stringCoverage, extractedReadsList-method
```

Get string coverage STR identified objects.

### **Description**

stringCoverage takes an extractedReadsList-object and finds the coverage of every unique string for every marker in the provided list.

# Usage

```
## S4 method for signature 'extractedReadsList'
stringCoverage(extractedReadsListObject,
  control = stringCoverage.control())
```

### **Arguments**

control

```
extractedReadsListObject
                  an extractedReadsList-object, created using the identifySTRRegions-function.
                  an stringCoverage.control-object.
```

Value

Returns a list, with an element for every marker in extractedReadsList-object, each element contains the string coverage of all unique strings of a given marker.

```
# Regions identified using 'identifySTRs()'
data("identifiedSTRs")
# Limiting and restructuring
sortedIncludedMarkers <- sapply(names(identifiedSTRs$identifiedMarkersSequencesUniquelyAssigned),
                                function(m) which(m == flankingRegions$Marker))
# Aggregate the strings
stringCoverage(extractedReadsListObject = identifiedSTRs,
               control = stringCoverage.control(
                   motifLength = flankingRegions$MotifLength[sortedIncludedMarkers],
                   Type = flankingRegions$Type[sortedIncludedMarkers],
                   numberOfThreads = 1,
                   trace = FALSE,
                   simpleReturn = TRUE))
```

```
stringCoverage, extractedReadsListCombined-method

Get string coverage STR identified objects.
```

### **Description**

stringCoverage takes an extractedReadsList-object and finds the coverage of every unique string for every marker in the provided list.

# Usage

```
## S4 method for signature 'extractedReadsListCombined'
stringCoverage(extractedReadsListObject,
   control = stringCoverage.control())
```

### **Arguments**

```
extracted Reads List Object \\ an extracted Reads List - object, created using the identify STR Regions-function. \\ control an string Coverage.control-object.
```

### Value

Returns a list, with an element for every marker in extractedReadsList-object, each element contains the string coverage of all unique strings of a given marker.

stringCoverage, extractedReadsListNonCombined-method

Get string coverage STR identified objects.

# **Description**

stringCoverage takes an extractedReadsList-object and finds the coverage of every unique string for every marker in the provided list.

# Usage

```
## S4 method for signature 'extractedReadsListNonCombined'
stringCoverage(extractedReadsListObject,
  control = stringCoverage.control())
```

### **Arguments**

```
extractedReadsListObject an extractedReadsList-object, created using the identifySTRRegions-function. control an stringCoverage.control-object.
```

### Value

Returns a list, with an element for every marker in extractedReadsList-object, each element contains the string coverage of all unique strings of a given marker.

stringCoverage, extractedReadsListReverseComplement-method

Get string coverage STR identified objects.

### **Description**

stringCoverage takes an extractedReadsList-object and finds the coverage of every unique string for every marker in the provided list.

### Usage

```
## S4 method for signature 'extractedReadsListReverseComplement'
stringCoverage(extractedReadsListObject,
  control = stringCoverage.control())
```

### **Arguments**

```
extractedReadsListObject an extractedReadsList-object, created using the identifySTRRegions-function. control an stringCoverage.control-object.
```

### Value

Returns a list, with an element for every marker in extractedReadsList-object, each element contains the string coverage of all unique strings of a given marker.

```
stringCoverage.control
```

String coverage coontrol object

# **Description**

String coverage coontrol object

### Usage

```
stringCoverage.control(motifLength = 4, Type = "AUTOSOMAL",
    simpleReturn = TRUE, includeLUS = FALSE, numberOfThreads = 4L,
    meanFunction = mean, includeAverageBaseQuality = FALSE, trace = FALSE,
    uniquelyAssigned = TRUE)
```

# **Arguments**

motifLength The motif lengths of each marker.

Type The chromosome type of each marker (autosomal, X, or Y).

simpleReturn TRUE/FALSE: Should the returned object be simplified?

includeLUS TRUE/FALSE: Should the LUS of each region be calculated?

numberOfThreads

The number of cores used for parallelisation.

meanFunction The function used to average the base qualities.

includeAverageBaseQuality

Should the average base quality of the region be included?

trace TRUE/FALSE: Show trace?

uniquelyAssigned

TRUE/FALSE: Should regions not uniquely assigned be removed?

### **Details**

Control function for the 'stringCoverage' function. Sets default values for the parameters.

### Value

List of parameters used for the 'stringCoverage' function.

stringCoverageGenotypeList

Combined string coverage and genotype information

# **Description**

A merge of the stringCoverageList and genotypeList data.

#### **Usage**

```
data("stringCoverageGenotypeList")
```

### **Format**

A list of tibble's one for each of the 10 markers containing the combined string coverage and genotypic information.

### Author(s)

```
Søren B. Vilsen <svilsen@math.aau.dk>
```

```
stringCoverageGenotypeList-class
```

Combined stringCoverage- and genotypeIdentifiedList

# **Description**

Merges a stringCoverageList with a genotypeIdentifiedList.

 ${\it string} Coverage List$ 

Aggregated string coverage.

# **Description**

The aggregated string coverage of the identified STRs data, created by the string Coverage function.

### **Usage**

```
data("stringCoverageList")
```

### **Format**

A list of tibble's one for each of the 10 markers, showing the aggregated information on a string-by-string basis.

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### Author(s)

Søren B. Vilsen <svilsen@math.aau.dk>

stringCoverageList-class

A string coverage list

# **Description**

A list of tibbles, one for every marker, used to contain the sequencing information of STR MPS data. The tibbles should include columns with the following names: "Marker", "BasePairs", "Allele", "Type", "MotifLength", "ForwardFlank", "Region", "ReverseFlank", "Coverage", "AggregateQuality", and "Quality".

stringCoverageNoiseList-class

Combined stringCoverage- and noiseIdentifiedList

### **Description**

Merges a stringCoverageList with a noiseIdentifiedList

STRMPSWorkflow

Workflow function

# **Description**

The function takes an input file and performs all preliminary analyses. The function creates a series of objects which can be further analysed. An output folder can be provided to store the objects as .RData-files.

# Usage

```
STRMPSWorkflow(input, output = NULL, continueCheckpoint = NULL,
  control = workflow.control())
```

### **Arguments**

input A path to a . fastq-file.

output A directory where output-files are stored.

continueCheckpoint

Choose a checkpoint to continue from in the workflow. If NULL the function

will run the entire workflow.

control Function controlling non-crucial parameters and other control functions.

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#### Value

If 'output' not provided the function simply returns the stringCoverageList-object. If an output is provided the function will store ALL created objects at the output-path, i.e. nothing is returned.

### **Examples**

STRMPSWorkflowBatch

Batch wrapper for the workflow function

### **Description**

The function takes an input directory and performs the entire analysis workflow described in (ADD REF). The function creates a series of objects needed for further analyses and stores them at the output location.

# Usage

```
STRMPSWorkflowBatch(input, output, ignorePattern = NULL,
  continueCheckpoint = NULL, control = workflow.control())
```

# **Arguments**

input A directory where fastq input-files are stored.

output A directory where output-files are stored.

ignorePattern A pattern parsed to grepl used to filter input strings.

continue Checkpoint

Choose a checkpoint to continue from in the workflow. If NULL the function

will run the entire workflow.

control Function controlling non-crucial parameters and other control functions.

### Value

If 'output' not provided the function simply returns the stringCoverageList-object. If an output is provided the function will store ALL created objects at the output-path, i.e. nothing is returned.

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```
STRMPSWorkflowCollectStutters
```

Collect stutters files

# **Description**

Collects all stutter files created by the batch version of the STRMPSWorkflow function.

# Usage

```
STRMPSWorkflowCollectStutters(stutterDirectory, storeCollection = TRUE)
```

### **Arguments**

```
stutterDirectory
```

The out most directory containing all stutter files to be collected.

storeCollection

TRUE/FALSE: Should the collected tibble be stored? If 'FALSE' the tibble is returned.

### Value

If 'storeCollection' is TRUE nothing is returned, else the stutter collection is returned.

workflow.control

Workflow default options

# Description

Control object for workflow function returning a list of default parameter options.

### Usage

```
workflow.control(numberOfMutations = 1, numberOfThreads = 4,
    createdThresholdSignal = 0.05, thresholdHomozygote = 0.4,
    internalTrace = FALSE, simpleReturn = TRUE, identifyNoise = FALSE,
    identifyStutter = FALSE, flankingRegions = NULL, useSTRaitRazor = FALSE,
    trimRegions = TRUE, restrictType = NULL, trace = TRUE,
    variantDatabase = NULL, reduceSize = FALSE)
```

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### **Arguments**

numberOfMutations

The maximum number of mutations (base-calling errors) allowed during flanking region identification.

numberOfThreads

The number of threads used by either the mclapply-function (stuck at '2' on windows) or STRaitRazor.

createdThresholdSignal

Noise threshold.

thresholdHomozygote

Homozygote threshold for genotype identification.

internalTrace Show trace.

simpleReturn TRUE/FALSE: Should the regions be aggregated without including flanking re-

gions?

identifyNoise TRUE/FALSE: Should noise be identified.

identifyStutter

TRUE/FALSE: Should stutters be identified.

flankingRegions

The flanking regions used to identify the STR regions. If 'NULL' a default set

is loaded and used.

useSTRaitRazor TRUE/FALSE: Should the STRaitRazor command line tool (only linux is im-

plemented) be used for flanking region identification.

trimRegions TRUE/FALSE: Should the identified regions be further trimmed.

restrictType A character vector specifying the marker 'Types' to be identified.

trace TRUE/FALSE: Should a trace be shown?

variantDatabase

A tibble of 'trusted' STR regions.

reduceSize TRUE/FALSE: Should the size of the data-set be reduced using the quality and

the variant database?

### Value

List of default of options.

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