Package 'STRMPS'

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Type Package

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

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Author Søren B. Vilsen

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

Description Loading, identifying, aggregating, manipulating, and analysing short tandem repeat regions of massively parallel sequencing data in forensic genetics. 'STRMPS' can work with the package 'STRaitRazoR' (an R interface to the 'STRaitRazor' commandline tool) for added speed. 'STRaitRazoR' only works on linux and can found at <https://github.com/svilsen/STRaitRazoR>. The analyses and framework implemented in this package relies on the papers 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, speedups will only be seen on UNIX based systems.

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LazyData TRUE

Depends R (>= 3.1.0), Biostrings, ShortRead

Imports methods, utils, IRanges, tidyr, tibble, dplyr, stringr, purrr, parallel

Suggests STRaitRazoR

biocViews Biostrings, ShortRead, IRanges

RoxygenNote 6.0.1

NeedsCompilation no

Repository CRAN

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Contents

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'.

Examples

```
# 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.

`extractedReadsListReverseComplement-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` *Find neighbours*

Description

Generic function for finding neighbouring strings, given identified alleles.

Usage

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

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.

`findNeighbours,stringCoverageGenotypeList-method` *Find neighbours*

Description

Generic function for finding neighbouring strings, given identified alleles.

Usage

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

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

`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
```

findStutter,stringCoverageGenotypeList-method *Find 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
```

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.

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>

`genotypeIdentifiedList-class` *Genotype list*

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 *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

```
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.

Examples

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

getGenotype(stringCoverageList)
```

getGenotype,stringCoverageList-method
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.

Examples

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

getGenotype(stringCoverageList)
```

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.

Examples

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

identifyNoise(stringCoverageList, thresholdSignal = 0.03)
```

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.

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)
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,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')

# 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.

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)
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.

---

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.
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

`mergeGenotypeStringCoverage` merges genotypeIdentifiedList-objects and stringCoverageList-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.

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,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)
```

neighbourList-class *A neighbour list*

Description

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

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.

Value

`phredQualityScore(q_phred)` and `solexaQualityScore(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

`p`	Probability of error.

Value

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

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

Examples

```
p <- 1e-3
phredQualityScore(p)
solexaQualityScore(p)
```

`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.

Examples

```
# 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,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

`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.

Examples

```
# 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

`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.

Examples

```
# 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,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.

Examples

```
# 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,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.

Examples

```
# 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.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.

stringCoverageList *Aggregated string coverage.*

Description

The aggregated string coverage of the identifiedSTRs data, created by the stringCoverage 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.

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.

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

```
readPath <- system.file('extdata', 'sampleSequences.fastq', package = 'STRMPS')

STRMPSWorkflow(readPath,
               control = workflow.control(restrictType = "Autosomal",
                                          numberOfThreads = 1)
               )
```

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.
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.

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.

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)
```

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 identiication.
internalTrace	Show trace.
simpleReturn	TRUE/FALSE: Should the regions be aggregated without including flanking regions?
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 implemented) 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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