Package 'STRAH'

April 3, 2019

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Type F	Package
Title S	Short Tandem Repeats Analysis of Hotspot Zones
Version	n 1.0
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Mainta	ainer Philipp Hermann <philipp.hermann@jku.at></philipp.hermann@jku.at>
s ie fi b o fi	ption Searches for short tandem repeats (STR) in a specified region of any genome. This analysis can be expanded such that several regions (chromosomes) are studed. These STRs can be grouped into hotspot as well as flanking regions of user specified width. Hotspots are defined by the double strand break maps from Pratto et al. (2014) <doi:10.1126 science.1256442="">. Moreover, the user can also search for a specified motif in a DNAStringSet-object, or a fastable, or a specified region of any genome. For an application of STR detections please see Heissl et al. (2018) <doi:10.1101 431841="">.</doi:10.1101></doi:10.1126>
Import	ts Biostrings (>= 2.38.4), BSgenome, BiocManager
	sts BSgenome.Hsapiens.UCSC.hg19, BSgenome.Ptroglodytes.UCSC.panTro5
Depend	ds R (>= 2.10)
Licenso	e MIT + file LICENSE
Encodi	ing UTF-8
LazyDa	ata TRUE
	eports https://github.com/PhHermann/STRAH/issues enNote 6.1.1
URL b	https://github.com/PhHermann/STRAH
Needs(Compilation no
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chr6_1580213_1582559 Sequence of human chromosome 6 from 1,580,213 until 1,582,559

Description

A DNAStringSet object containing the data of human chromosome 6 starting with position 1,580,213 and ending at 1,582,559.

Usage

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```
data (chr6_1580213_1582559)
```

Format

The data set is a DNAStringSet object containing one sequence of length 2346 nucleotides.

See Also

motif_detection

 ${\it chr6_1581473_1586032} \quad \textit{Sequence of human chromosome 6 from 1,} 581,473 \textit{ until 1,} 586,032$

Description

A DNAStringSet object containing the data of human chromosome 6 starting with position 1,581,473 and ending at 1,586,032.

Usage

```
data (chr6_1581473_1586032)
```

Format

The data set is a DNAStringSet object containing one sequence of length 4559 nucleotides.

See Also

```
motif_detection
```

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dsb_map

Data of the DsbMap for humans

Description

A dataset containing the PRDM9-A type hotspots of Pratto et al. 2014.

Usage

```
data (dsb_map)
```

Format

The data set contains 37527 rows and 27 columns. We provide information on the most important columns (column nr, column name) hereafter:

- **1, chrom** The chromosome under study
- 2, start Start coordinates of the hotspot
- 3, end End coordinates of the hotspot
- 4-8, (AA1,AA2,AB1,AB2,AC)_strength Strength of the corresponding PRDM9-type hotspot.
- **9-15**, (AA1,AA2,AB1,AB2,AC)_hotspots Dummy coding whether these positions (start/end) define a hotspot of given PRDM9-type

References

Heissl, A., et al. (2018) Length asymmetry and heterozygosity strongly influences the evolution of poly-A microsatellites at meiotic recombination hotspots. doi: https://doi.org/10.1101/431841 Pratto, F., et al. (2014). Recombination initiation maps of individual human genomes. Science, 346(6211).

See Also

STR_analysis, STR_detection

dsb_map_chimp_full

Data of the DsbMap for chimpanzees

Description

A dataset containing all translated PRDM9-A type hotspots of Pratto et al. (2014) for chimpanzees. The translation was performed using liftOver of the UCSC Genome Browser from the human genome (hg19) to the chimpanzees genome (panTro5).

Usage

```
data (dsb_map_chimp_full)
```

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Format

The data set contains 64078 rows and 3 columns. We provide information on the columns (column nr, column name) hereafter:

- 1, chrom The chromosome under study
- 2, start Start coordinates of the hotspot
- 3, end End coordinates of the hotspot

References

Heissl, A., et al. (2018) Length asymmetry and heterozygosity strongly influences the evolution of poly-A microsatellites at meiotic recombination hotspots. doi: https://doi.org/10.1101/431841 Pratto, F., et al. (2014). Recombination initiation maps of individual human genomes. Science, 346(6211). Kuhn RM, et al. (2013) The UCSC genome browser and associated tools, Brief. Bioinform., 14, 144-161.

See Also

STR_analysis, STR_detection

getflank2

Extract a specified region of the (human) genome

Description

This function extracts a specified region of the human genome with corresponding start and end position of the region under study.

Usage

```
getflank2(species, chrs, start.position, end.position)
```

Arguments

species The human genome (version 19) is default but an alternative genome can be pro-

 $vided.\ For\ chimpanzees\ the\ parameter\ has\ to\ be\ BS genome. Ptroglodytes. UCSC. pan Tro 5$

(given that the data is installed).

chrs A string reflecting the chromosome under study (starting with "chr" and adding

either the integers from 1-22 or "X" respectively "Y"). This argument can also

be a vector of strings to study several chromosomes.

start.position An integer value reflecting the start position of the region to be analyzed. If set

to NA the analysis starts from the beginning of the chromosome.

end.position An integer value reflecting the end position of the region to be analyzed. If set

to NA the analysis is performed until the end of the chromosome.

Value

The DNA-sequence of the region under study (defined by the chromosome, start position, and end position) is returned.

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References

Heissl, A., et al. (2018) Length asymmetry and heterozygosity strongly influences the evolution of poly-A microsatellites at meiotic recombination hotspots. doi: https://doi.org/10.1101/431841

See Also

```
STR_analysis
```

Examples

```
getflank2(BSgenome.Hsapiens.UCSC.hg19::Hsapiens, "chr1", 100000, 100010)
```

 $\verb|motif_detection||$

DNA-motif detection in a given DNAStringSet, a given DNA-sequence in fasta-format, or a specified region of any genome

Description

This function searches for a given "DNA-motif" in a DNA-sequence. The argument seqName can be either a DNAStringSet object or it refers to a fasta-file. Additionally, we provide the option to specify a species, a chromosome, a start, and a stop position for a region of any reference genome to be analyzed. By default a region of the human genome is analyzed. Optionally, one can also specify the number of mismatches of the DNA-motif and whether the reverse complement has to be searched.

Usage

```
motif_detection(seqName, chrs, start.position, end.position, motif,
    nr.mismatch = 0, reverse.comp = F, print.status = T,
    species = BSgenome.Hsapiens.UCSC.hg19::Hsapiens)
```

Arguments

seqName A character string which can either be the name of a DNAStringSet object or a sequence name referring to a fasta-file to be analyzed. This argument can only

be ignored if chr and start.position and end.position are specified.

chrs A character string reflecting the chromosome under study (starting with "chr"

and adding either the integers from 1-22 or "X" respectively "Y" for the human chromosome). This argument can also be a vector of strings to study several

chromosomes.

start.position An integer value reflecting the start position of the region to be analyzed. If set

to NA the analysis starts from the beginning of the chromosome.

end.position An integer value reflecting the end position of the region to be analyzed. If set

to NA the analysis is performed until the end of the chromosome.

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motif A character string reflecting the specified DNA-motif to be searched for in the

DNA-sequence.

nr.mismatch This integer specifies the number of allowed mismatches when searching for the

specified DNA-motif.

reverse.comp A logical value, by default FALSE, which enables to search the reverse comple-

ment of the sequence if set to TRUE.

print.status A logical value reflecting whether the current status of the worked sequence

(relative to the sequence length) is printed (TRUE) or not (FALSE).

species The human genome (version 19) is default but an alternative genome can be pro-

vided. For chimpanzees the parameter has to be BSgenome.Ptroglodytes.UCSC.panTro5

(given that the data is installed).

Value

The output of the function is a list with the following content:

Species The name of the species under study
Sequence Name The name of the region under study

Reverse Complement

Indicator whether the reverse complement was searched

Number of Matches

The frequency of found DNA-motifs in the region under study

Start Positions of Matches

The start positions of the found DNA-motifs

Number of allowed Mismatches

The number of allowed mismatches when searching for the DNA-motif

Matched Segments

The list of the segments containing the DNA-motif

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References

Heissl, A., et al. (2018) Length asymmetry and heterozygosity strongly influences the evolution of poly-A microsatellites at meiotic recombination hotspots. doi: https://doi.org/10.1101/431841

See Also

```
getflank2
```

Examples

```
data(chr6_1580213_1582559)
motif_detection(seqName = chr6_1580213_1582559, chrs="",
start.position = NA, end.position = NA, motif = "CCNCCNTNNCCNC",
nr.mismatch = 1, reverse.comp = FALSE, print.status = FALSE)
motif_detection(seqName = "", chrs = "chr6",
```

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```
start.position = 1580213, end.position = 1582559,
motif = "CCNCCNTNNCCNC", nr.mismatch = 1, reverse.comp = FALSE, print.status = FALSE)
# If you want to use the function with a different reference genome
# make your choice and install it before:
BiocManager::install("BSgenome.Ptroglodytes.UCSC.panTro5")
library(BSgenome.Ptroglodytes.UCSC.panTro5)
motif_detection(seqName = "", chrs = "chr1", start.position =222339618, end.position = 222339660, motif = "A", nr.mismatch = 0, reverse.comp = FALSE,
print.status = FALSE, species = BSgenome.Ptroglodytes.UCSC.panTro5)
```

STRAH

STRAH: A package to detect DNA-motifs or short tandem repeats (STRs) in reference genomes

Description

The STRAH package provides functions to extract DNA sequence data of reference genomes, functions to search for DNA-motifs in a defined DNA-sequence or to detect short tandem repeats (STRs) of specified length, and to analyze detected STRs in the human (or chimpanzees) genome by comparing them with (translated) double strand break hotspots.

STRAH functions:

getflank2 Extract a specified region of any reference genome (e.g. humans, chimpanzees, ...)

motif_detection DNA-motif detection of a given DNAStringSet, a given DNA-sequence in fastaformat, or a specified region of any genome

STR_detection Detection of short tandem repeats (STRs) in either one or several regions of a given species

STR_analysis Analysis of detected short tandem repeats (STRs) in either one or several regions of a given species

STRAH data sets:

chr6_1580213_1582559 DNA-sequence of human chromosome 6 from 1,580,213 until 1,582,559 chr6_1581473_1586032 DNA-sequence of human chromosome 6 from 1,581,473 until 1,586,032 dsb_map Data of the double strand break map of Pratto et al. (2014).

dsb_map_chimp_full Translated data of the double strand break map of Pratto et al. (2014) for the chimpanzees genome.

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References

Heissl, A., et al. (2018) Length asymmetry and heterozygosity strongly influences the evolution of poly-A microsatellites at meiotic recombination hotspots. doi: https://doi.org/10.1101/431841 Pratto, F., et al. (2014). Recombination initiation maps of individual human genomes. Science, 346(6211). Kuhn RM, et al. (2013) The UCSC genome browser and associated tools, Brief. Bioinform., 14, 144-161.

STR_analysis

STR_analysis	Analysis of short tandem repeats (STRs) in a given region of any reference genome

Description

This function separates detected short tandem repeats (STRs) into different zones. These zones are either the hotspot zone defined by the double strand break maps of Pratto et al. (2014) or adjacent flanking zones (greyzones) left and right of the hotspots of user specified lengths. The parameters of the regions under study can be directly given in the function arguments or read in via either a BED-file or a position matrix.

Usage

```
STR_analysis(seqName = "", nr.STRs = 10, nr.mismatch = 0,
  chrs = "", STR = "A", lens.grey = 0:5 * 1000,
  start.position = NA, end.position = NA, reverse.comp = FALSE,
  bed_file = "", pos_matrix = "", output_file = "",
  species = BSgenome.Hsapiens.UCSC.hg19::Hsapiens,
  dsb_map = STRAH::dsb_map)
```

Arguments

seqName	A character string which is the name of the given sequence file under study. Can also be set to "" in order to analyze a defined sequence from any reference genome such as the package BSgenome.Hsapiens.UCSC.hg19 for humans.
nr.STRs	An integer value reflecting the minimum length of STRs to be searched for.
nr.mismatch	An integer value reflecting the allowed number of mismatches of the short tandem repeats. By default it set to 0 .
chrs	A string reflecting the chromosome under study (starting with "chr" and adding either the integers from 1-22 or "X" respectively "Y" for the human genome). This argument can also be a vector of strings to study several chromosomes.
STR	A character string for the nucleotide to be searched for. By default one searches for poly-As, hence set to "A".
lens.grey	An integer value which is by default a vector of 6 integer values. These values represent the greyzones to be studied left and right from the hotspot regions.
start.position	An integer value reflecting the start position of the region to be analyzed. If set to NA the analysis starts from the beginning of the chromosome.
end.position	An integer value reflecting the end position of the region to be analyzed. If set to NA the analysis is performed until the end of the chromosome.
reverse.comp	A logical value by default FALSE. If set to TRUE then the reverse complement of the sequence is analyzed.
bed_file	A bed file containing the chromosomes, start, and end positions of the region(s) that should be analyzed.
pos_matrix	A matrix or dataframe containing the chromosomes, start, and end positions of the region(s) that should be analyzed.

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will be saved if the parameter is changed to a user defined string excluding the

extension (by default .bed).

species The human genome (version 19) is default but an alternative genome can be pro-

vided. For chimpanzees the parameter has to be BSgenome.Ptroglodytes.UCSC.panTro5

(given that the data is installed).

dsb_map The DSB map of the human genome (version 19) is default but an alternative

DSB map from a different genome can be provided. This parameter needs to be a data frame with at least 3 columns that contains the chromosome, start, and end position of the DSB. The DSB map for chimpanzees is included in the

package.

Value

The output of the function is a list with the following content:

Sequence Name The chromosome with the starting and end position of the region under study is provided.

Reverse Complement

An indicator whether the reverse complement was considered

Number of allowed Mismatches

The number of allowed mismatches is provided.

Minimum Length The minimum length of the STR to be extracted is provided.

Number of Matches

The total number of STR matches of the region is provided.

Length of STR stretch in bp

A vector containing the length of STRs per match is provided.

Start positions

The starting positions of the STRs are provided.

Zone

The zones where the STR is found are provided. 1 reflects within a hotspot, the last integer reflects that it is outside, and the integers between these two reflect the given flanking regions starting with 2 as the next closest region to the hotspot.

A BED file with the chromosomes, start, and end position of the STRs, length of the STR stretch, the zone where the STR was found, and the specified region that was analyzed are given as columns.

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References

Heissl, A., et al. (2018) Length asymmetry and heterozygosity strongly influences the evolution of poly-A microsatellites at meiotic recombination hotspots. doi: https://doi.org/10.1101/431841 Pratto, F., et al. (2014). Recombination initiation maps of individual human genomes. Science, 346(6211). Kuhn RM, et al. (2013) The UCSC genome browser and associated tools, Brief. Bioinform., 14, 144-161.

See Also

getflank2, STR_detection

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Examples

```
data(chr6_1580213_1582559)
STR_analysis(seqName = chr6_1580213_1582559, nr.STRs = 10, nr.mismatch = 0, chrs = "chr6",
STR = "A", \#' lens.grey = 0:1*100, start.position = 1580213, end.position = 1582559,
reverse.comp = FALSE, bed_file = "", pos_matrix = "", output_file = "",
species = BSgenome.Hsapiens.UCSC.hg19::Hsapiens, dsb_map = STRAH::dsb_map)
STR\_analysis(nr.STRs = 10, nr.mismatch = 0, chrs = "chr22", STR = "A", lens.grey = 0:1*100, lens.grey = 0:1*1000, lens.grey = 0:
start.position = 30000000, end.position = 31000000, reverse.comp = FALSE,
bed_file = "", pos_matrix = "", output_file = "",
species = BSgenome.Hsapiens.UCSC.hg19::Hsapiens, dsb_map = STRAH::dsb_map)
# If you want to use the function with a different reference genome
# make your choice and install it before:
BiocManager::install("BSgenome.Ptroglodytes.UCSC.panTro5")
library(BSgenome.Ptroglodytes.UCSC.panTro5)
STR_analysis(nr.STRs = 10, nr.mismatch = 0, chrs = "chr22", STR = "A", lens.grey = 0:5*1000,
start.position = 30000000, end.position = 31000000, reverse.comp = FALSE, bed_file = "",
pos_matrix = "", output_file = "", species = BSgenome.Ptroglodytes.UCSC.panTro5,
dsb_map = STRAH::dsb_map_chimp_full)
```

STR_detection

Detection of short tandem repeats (STRs) in a given region of any reference genome

Description

This function searches for short tandem repeats (STRs) in a specified region of any reference genome. The parameters of the regions under study can be directly given in the function arguments or read in via either a BED-file or a position matrix. We recommend to search for STRs of minimum length 6. Options to save the output or usage of any reference genome are provided.

Usage

```
STR_detection(seqName = "", chrs = "", start.position = NA,
  end.position = NA, bed_file = "", pos_matrix = "", nr.STRs,
  nr.mismatch = 0, reverse.comp = F, STR = "A",
  species = BSgenome.Hsapiens.UCSC.hg19::Hsapiens,
  translated_regions = F, output_file = "")
```

Arguments

seqName	A character string which is the name of the given sequence file under study. Can also be set to "" in order to analyze a defined sequence from any reference genome such as the package BSgenome.Hsapiens.UCSC.hg19 for humans.
chrs	A string reflecting the chromosome under study (starting with "chr" and adding either the integers from 1-22 or "X" respectively "Y" for the human genome). This argument can also be a vector of strings to study several chromosomes.
start.position	An integer value reflecting the start position of the region to be analyzed. If set to NA the analysis starts from the beginning of the chromosome.
end.position	An integer value reflecting the end position of the region to be analyzed. If set to NA the analysis is performed until the end of the chromosome.

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bed_file A bed file containing the chromosomes, start, and end positions of the region(s)

that should be analyzed.

pos_matrix A matrix or dataframe containing the chromosomes, start, and end positions of

the region(s) that should be analyzed.

nr. STRs An integer value as the minimum length of STRs to be detected.

nr.mismatch An integer value reflecting the allowed number of mismatches of the short tan-

dem repeats. By defaults set to 0.

reverse.comp A logical value by default FALSE. If set to TRUE then the reverse complement of

the sequence is analyzed.

STR A character string for the nucleotide to be searched for. By default one searches

for poly-As, hence set to "A".

species The human genome (version 19) is default but an alternative genome can be pro-

vided. For chimpanzees the parameter has to be BSgenome.Ptroglodytes.UCSC.panTro5

(given that the data is installed).

translated_regions

A logical value by default FALSE. If set to TRUE then the function assumes that the parameters start.position and end.position were translated by some tool (e.g. liftOver) from one species to another. The untranslated and translated positions

are included in the output.

will be saved if the parameter is changed to a user defined string excluding the

extension (by default .bed).

Value

The output of the function is a list with the following content:

Sequence name The chromosome with the start and end position of the region under study is pro-

 $vided. \ If \ translated_region \ is \ set \ to \ TRUE, \ the \ interval \ will \ be \ the \ translated$

region.

Sequence name (untranslated)

Only if translated_region is set to TRUE, then the untranslated region (chro-

mosome with the starting and end position) is provided.

Reverse complement

An indicator whether the reverse complement was considered

Number of allowed mismatches

The number of allowed mismatches is provided.

Minimum length The minimum length of the STR to be extracted is provided.

Number of matches

The total number of STR matches of the region is provided.

Length of STR stretch in bp

A vector containing the length of STRs per match is provided.

Start positions

The starting positions of the STRs are provided.

Matched segments

The matched segments of the STRs are provided.

A BED file with chromosomes, start, and end position of the STRs, length of the STR stretch, the matched segments, and the specified region (untranslated and translated) that was analyzed are given as columns.

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Author(s)

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References

Heissl, A., et al. (2018) Length asymmetry and heterozygosity strongly influences the evolution of poly-A microsatellites at meiotic recombination hotspots. doi: https://doi.org/10.1101/431841 Pratto, F., et al. (2014). Recombination initiation maps of individual human genomes. Science, 346(6211). Kuhn RM, et al. (2013) The UCSC genome browser and associated tools, Brief. Bioinform., 14, 144-161.

See Also

```
getflank2, STR_analysis
```

Examples

```
data(chr6_1580213_1582559)
STR_detection(seqName = chr6_1580213_1582559, chrs = "chr6", start.position = 1580213,
end.position = 1582559, nr.STRs = 10, nr.mismatch = 0, reverse.comp = FALSE, STR = "A",
species = BSgenome.Hsapiens.UCSC.hg19::Hsapiens, translated_regions=FALSE, output_file = "")
STR_detection(seqName = "", chrs = "chr22", start.position = 30000000, end.position = 31000000,
nr.STRs = 10, nr.mismatch = 0, reverse.comp = FALSE, STR = "A",
species=BSgenome.Hsapiens.UCSC.hg19::Hsapiens, translated_regions=FALSE, output_file = "")
# If you want to use the function with a different reference genome
# make your choice and install it before:
BiocManager::install("BSgenome.Ptroglodytes.UCSC.panTro5")
library(BSgenome.Ptroglodytes.UCSC.panTro5)
STR_detection(seqName = "", chrs = "chr1", start.position =222339618, end.position = 222339660,
nr.STRs = 10, nr.mismatch = 0, reverse.comp = FALSE, STR = "A",
species = BSgenome.Ptroglodytes.UCSC.panTro5)
```

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