Package 'svaNUMT'

May 30, 2024

Type Package

Title NUMT detection from structural variant calls

Version 1.10.0

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Description svaNUMT contains functions for detecting NUMT events from structural variant calls. It takes structural variant calls in GRanges of breakend notation and identifies NUMTs by nuclear-mitochondrial breakend junctions. The main function reports candidate NUMTs if there is a pair of valid insertion sites found on the nuclear genome within a certain distance threshold. The candidate NUMTs are reported by events.

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Depends GenomicRanges, rtracklayer, VariantAnnotation, StructuralVariantAnnotation, BiocGenerics, Biostrings, R (>= 4.0)

Imports assertthat, stringr, dplyr, methods, rlang, GenomeInfoDb, S4Vectors, GenomicFeatures, pwalign

Suggests TxDb.Hsapiens.UCSC.hg19.knownGene, BSgenome.Hsapiens.UCSC.hg19, ggplot2, devtools, testthat (>= 2.1.0), roxygen2, knitr, readr, plyranges, circlize, IRanges, SummarizedExperiment, rmarkdown

RoxygenNote 7.1.2

Encoding UTF-8

VignetteBuilder knitr

biocViews DataImport, Sequencing, Annotation, Genetics, VariantAnnotation

BugReports https://github.com/PapenfussLab/svaNUMT/issues

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Contents

`.mtLen` *Calculating MT sequence length.*

Description

Calculating MT sequence length.

Usage

```
.mtLen(bnd.start, bnd.end, chrM.len)
```

Arguments

`bnd.start`	starting breakend of the MT sequence.
`bnd.end`	ending breakend of the MT sequence.
`chrM.len`	length of the reference MT genome.

Details

This function calculate the length of MT sequence length with BND notations.

Value

The length of the MT sequence. When the candidate MT BNDs can't be linked as one sequence, the returned value is NA.

numtDetect *Detecting nuclear mitochondria fusion events.*

Description

Detecting nuclear mitochondria fusion events.

Usage

```
numtDetect(
  gr,
  numtS,
  genomeMT,
  max_ins_dist = 10,
  maxgap_numtS = 10,
  min_len = 20,
  min.Align = 0.8
)
```

Arguments

gr	A GRanges object
numtS	A GRanges object of known NUMT sites.
genomeMT	A genome object of the mitochondria.
max_ins_dist	The maximum distance allowed on the reference genome between the paired insertion sites. Only intra-chromosomal NUMT events are supported. Default value is 10.
maxgap_numtS	The maximum distance allowed betweeen the insertion sequence loci and known NUMTs.
min_len	The minimum length allowed of the insertion sequences. Default value is 20.
min.Align	The minimum alignment score allowed between the insertion sequence and MT genome.

Details

Nuclear mitochondrial fusion (NUMT) is a common event found in human genomes. This function searches for NUMT events by identifying breakpoints supporting the fusion of nuclear chromosome and mitochondrial genome. Only BND notations are supported at the current stage. Possible linked nuclear insertion sites are reported by chromosome in GRanges format.

Value

A nested list of GRanges objects of candidate NUMTs.

Examples

```
vcf.file <- system.file("extdata", "MT.vcf", package = "svaNUMT")
vcf <- VariantAnnotation::readVcf(vcf.file, "hg19")
gr <- breakpointRanges(vcf, nominalPosition=TRUE)
numtS <- readr::read_table(system.file("extdata", "numtS.txt", package = "svaNUMT"), col_names = FALSE)
colnames(numtS) <- c("bin", "seqnames", "start", "end", "name", "score", "strand")
numtS <- `seqlevelsStyle<-`(GRanges(numtS), "NCBI")
genome <- BSgenome.Hsapiens.UCSC.hg19::BSgenome.Hsapiens.UCSC.hg19
genomeMT <- genome$chrMT
numt.gr <- numtDetect(gr, numtS, genomeMT, max_ins_dist=20)
```

`numtDetect_insseq` *Detecting nuclear mitochondria fusion events from unmapped insertion sequences.*

Description

Detecting nuclear mitochondria fusion events from unmapped insertion sequences.

Usage

```
numtDetect_insseq(gr, genomeMT, min_len = 20, min.Align = 0.8)
```

Arguments

`gr`	A GRanges object
`genomeMT`	A genome object of the mitochondria.
`min_len`	The minimum length allowed of the insertion sequences. Default value is 20.
`min.Align`	The minimum alignment score allowed between the insertion sequence and MT genome.

Details

This function looks for NUMTs which the insertion MT sequences come from insertion sequences reported by SV callers.

Value

A nested list of GRanges objects of candidate NUMTs.

numtDetect_known *Detecting nuclear mitochondria fusion events from known NUMT sites.*

Description

Detecting nuclear mitochondria fusion events from known NUMT sites.

Usage

```
numtDetect_known(gr, numtS, max_ins_dist = 10, maxgap_numtS = 10)
```

Arguments

gr	A GRanges object
numtS	A GRanges object of known NUMT sites.
max_ins_dist	The maximum distance allowed on the reference genome between the paired insertion sites. Only intra-chromosomal NUMT events are supported. Default value is 10.
maxgap_numtS	The maximum distance allowed betweeen the insertion sequence loci and known NUMTs.

Details

This function looks for NUMTs which the insertion MT sequences come from known NUMT sites.

Value

A nested list of GRanges objects of candidate NUMTs.

numtDetect_MT *Detecting nuclear mitochondria fusion events from breakpoints connected to MT reference genome.*

Description

Detecting nuclear mitochondria fusion events from breakpoints connected to MT reference genome.

Usage

```
numtDetect_MT(gr, max_ins_dist = 10)
```

Arguments

gr	A GRanges object
max_ins_dist	The maximum distance allowed on the reference genome between the paired insertion sites. Only intra-chromosomal NUMT events are supported. Default value is 10.

Details

This function looks for NUMTs which the insertion MT sequences come from known NUMT sites.

Value

A nested list of GRanges objects of candidate NUMTs.

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seqAlignment.score *Calculating the alignment score between a DNA sequence and target genome.*

Description

Calculating the alignment score between a DNA sequence and target genome.

Usage

```
seqAlignment.score(seq, genome)
```

Arguments

seq	A string of DNA sequence.
genome	An XString of the target genome.

Details

This function calculates the alignment score between a DNA sequence and target genome.

Value

A alignment score between a DNA sequence and target genome.

svaNUMT *svaNUMT: a package for NUMT detection*

Description

svaNUMT contains functions for detecting NUMT events from structural variant calls. svaNUMT contains functions for detecting NUMT events from structural variant calls. It takes structural variant calls in GRanges of breakend notation and identifies NUMTs by nuclear-mitochondrial breakend junctions. The main function reports candidate NUMTs if there is a pair of valid insertion sites found on the nuclear genome within a certain distance threshold. The candidate NUMTs are reported by events.

Details

For more details on the features of StructuralVariantAnnotation, read the vignette: ‘browseVignettes(package = "svaNUMT")‘

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