

Package ‘svaNUMT’

September 25, 2022

Type Package

Title NUMT detection from structural variant calls

Version 1.2.0

Date 2022-03-07

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, Biostings, R (>= 4.0)

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

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>

git_url <https://git.bioconductor.org/packages/svaNUMT>

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numtDetect	<i>Detecting nuclear mitochondria fusion events.</i>
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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 between 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)
```

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 break-end 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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