Package 'svaNUMT'

April 12, 2022

Type Package

Title NUMT detection from structural variant calls

Version 1.0.0

Date 2021-03-09

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, R (>= 4.0)

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

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

RoxygenNote 7.1.1

Encoding UTF-8

VignetteBuilder knitr

biocViews DataImport, Sequencing, Annotation, Genetics, VariantAnnotation

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

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R topics documented:

`numtDetect` *Detecting nuclear mitochondria fusion events.*

Description

Detecting nuclear mitochondria fusion events.

Usage

```
numtDetect(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

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)
numt.gr <- numtDetect(gr, 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 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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