Package: svaNUMT (via r-universe)

November 7, 2026

Type Package

Title NUMT detection from structural variant calls

Version 1.18.0

Date 2025-07-22

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, S4Vectors, Seqinfo, GenomeInfoDb, 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

Config/pak/sysreqs make libbz2-dev libicu-dev liblzma-dev libpng-dev libxml2-dev libssl-dev xz-utils zlib1g-dev

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`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 <- GRanges(numtS)
GenomeInfoDb::seqlevelsStyle(numtS) <- "NCBI"
genome <- BSgenome.Hsapiens.UCSC.hg19::BSgenome.Hsapiens.UCSC.hg19
genomeMT <- genome$chrMT
numt.gr <- numtDetect(gr, numtS, genomeMT, max_ins_dist=20)
```

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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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