

Package ‘branchpointer’

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Type Package

Title Prediction of intronic splicing branchpoints

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Description Predicts branchpoint probability for sites in intronic branchpoint windows. Queries can be supplied as intronic regions; or to evaluate the effects of mutations, SNPs.

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LazyData FALSE

Depends caret, R(>= 3.4)

Imports plyr, kernlab, gbm, stringr, cowplot, ggplot2, biomaRt, Biostrings, parallel, utils, stats, BSgenome.Hsapiens.UCSC.hg38, rtracklayer, GenomicRanges, GenomeInfoDb, IRanges, S4Vectors, data.table

Suggests knitr, BiocStyle

RoxygenNote 6.0.1

VignetteBuilder knitr

biocViews Software, GenomeAnnotation, GenomicVariation, MotifAnnotation

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| | |
|------------|---|
| gtfToExons | <i>Convert GTF file to exon location file</i> |
|------------|---|

Description

Converts a GTF annotation to exon locations

Usage

```
gtfToExons(gtf)
```

Arguments

gtf file containing the gtf annotation.

Value

exon annotation GRanges

Author(s)

Beth Signal

Examples

```
smallExons <- system.file("extdata", "gencode.v26.annotation.small.gtf",
package = "branchpointer")
exons <- gtfToExons(smallExons)
```

| | |
|-------------------------------|--|
| makeBranchpointWindowForExons | <i>Make branchpoint window regions</i> |
|-------------------------------|--|

Description

Generate branchpoint window regions corresponding to annotated exon(s) within a queried gene, transcript or exon id

Usage

```
makeBranchpointWindowForExons(id, idType, exons, forceClosestExon = FALSE)
```

Arguments

| | |
|------------------|--|
| id | identifier(s) for the query gene/transcript/exon id |
| idType | type of id to match in the exon annotation file ("gene_id", "transcript_id", or "exon_id") |
| exons | GRanges containing exon co-ordinates. |
| forceClosestExon | Force branchpointer to find the closest exon and not the exon annotated as 5' to the query |

Value

Granges with formatted query

Author(s)

Beth Signal

Examples

```
smallExons <- system.file("extdata", "gencode.v26.annotation.small.gtf", package = "branchpointer")
exons <- gtfToExons(smallExons)
windowquery <- makeBranchpointWindowForExons("ENSG00000139618.14", "gene_id", exons)
windowquery <- makeBranchpointWindowForExons("ENST00000357654.7", "transcript_id", exons)
windowquery <- makeBranchpointWindowForExons("ENSE000003518965.1", "exon_id", exons)
```

```
makeBranchpointWindowForSNP
```

Makes a branchpointer formatted GRanges object from refsnp ids

Description

Searches Biomart for refsnp ids, and pulls genomic location and sequence identity information
Reformats alleles so each query has only one alternative allele

Usage

```
makeBranchpointWindowForSNP(refsnp, mart.snp, exons, maxDist = 50,
  filter = TRUE)
```

Arguments

| | |
|----------|--|
| refsnp | Vector of refsnp ids |
| mart.snp | biomaRt mart object specifying the BioMart database and dataset to be used |
| exons | GRanges containing exon co-ordinates. Should be produced by gtfToExons() |
| maxDist | maximum distance a SNP can be from an annotated 3' exon. |
| filter | remove SNP queries prior to finding finding nearest exons? |

Value

formatted SNP query GRanges

Author(s)

Beth Signal

Examples

```
smallExons <- system.file("extdata", "gencode.v26.annotation.small.gtf", package = "branchpointer")
exons <- gtfToExons(smallExons)

mart.snp <- biomaRt::useMart("ENSEMBL_MART_SNP", dataset="hsapiens_snp", host="www.ensembl.org")
query <- makeBranchpointWindowForSNP("rs587776767", mart.snp, exons)
```

plotBranchpointWindow *Plots branchpointer predictions*

Description

Plots branchpointer predictions

Usage

```
plotBranchpointWindow(queryName, predictions, probabilityCutoff = 0.52,  
  plotMutated = FALSE, plotStructure = TRUE, exons)
```

Arguments

| | |
|-------------------|---|
| queryName | query id used to identify the SNP or region |
| predictions | Granges object generated by predictBranchpoints() |
| probabilityCutoff | probability score cutoff value for displaying U2 binding energy |
| plotMutated | plot alternative sequence predicitions alongside reference sequence predictions |
| plotStructure | plot structures for gene and 3' exon containing and skipping isoforms |
| exons | Granges containing exon co-ordinates. Should be produced by gtfToExons() |

Value

ggplot2 plot with branchpoint features in the specified intronic region

Author(s)

Beth Signal

Examples

```
smallExons <- system.file("extdata", "gencode.v26.annotation.small.gtf",  
  package = "branchpointer")  
exons <- gtfToExons(smallExons)  
g <- BSgenome.Hsapiens.UCSC.hg38::BSgenome.Hsapiens.UCSC.hg38  
  
querySNPFile <- system.file("extdata", "SNP_example.txt", package = "branchpointer")  
querySNP <- readQueryFile(querySNPFile, queryType = "SNP", exons = exons, filter = FALSE)  
predictionsSNP <- predictBranchpoints(querySNP, queryType = "SNP", BSgenome = g)  
plotBranchpointWindow(querySNP$id[1], predictionsSNP,  
  plotMutated = TRUE, exons = exons)
```

predictBranchpoints *Predict branchpoint probability scores*

Description

predicts branchpoint probability scores for each query site.

Usage

```
predictBranchpoints(query, uniqueId = "test", queryType,
  workingDirectory = ".", genome = NA, bedtoolsLocation = NA,
  BSgenome = NULL, useParallel = FALSE, cores = 1, rmChr = FALSE)
```

Arguments

| | |
|------------------|--|
| query | branchpointer query GenomicRanges |
| uniqueId | unique string identifier for intermediate .bed and .fa files. |
| queryType | type of branchpointer query. "SNP" or "region". |
| workingDirectory | directory where intermediate .bed and .fa are located |
| genome | .fa genome file location |
| bedtoolsLocation | bedtools binary location (which bedtools) |
| BSgenome | BSgenome object |
| useParallel | use parallelisation to speed up code? |
| cores | number of cores to use in parallelisation (default = 1) |
| rmChr | remove "chr" before chromosome names before writing bed file. Required if genome sequence names do not contain "chr" |

Value

GenomicRanges object with branchpoint probability scores for each site in query

Author(s)

Beth Signal

Examples

```
smallExons <- system.file("extdata", "gencode.v26.annotation.small.gtf",
  package = "branchpointer")
exons <- gtfToExons(smallExons)
g <- BSgenome.Hsapiens.UCSC.hg38::BSgenome.Hsapiens.UCSC.hg38

querySNPFile <- system.file("extdata", "SNP_example.txt", package = "branchpointer")
querySNP <- readQueryFile(querySNPFile, queryType = "SNP", exons = exons, filter = FALSE)
predictionsSNP <- predictBranchpoints(querySNP, queryType = "SNP", BSgenome = g)
```

predictionsToSummary *Convert SNP branchpoint predictions across the branchpoint window to an intronic summary*

Description

Takes predictions of branchpoint probabilities from a reference and alternative SNP and summarises the effect(s) of the SNP.

Usage

```
predictionsToSummary(query, predictions, probabilityCutoff = 0.52,
  probabilityChange = 0.15)
```

Arguments

query query GRanges containing all SNP ids to be summarised

predictions site-wide branchpoint probability predictions produced from predictBranchpoints()

probabilityCutoff Value to be used as the cutoff for discriminating branchpoint sites from non-branchpoint sites (default = 0.52)

probabilityChange Minimum probability score change required to call a branchpoint site as deleted or created by a SNP (default = 0.15)

Value

GRanges with summarised branchpoint changes occurring within the intron due to a SNP.

Author(s)

Beth Signal

Examples

```
smallExons <- system.file("extdata", "gencode.v26.annotation.small.gtf", package = "branchpointer")
exons <- gtfToExons(smallExons)
g <- BSgenome.Hsapiens.UCSC.hg38::BSgenome.Hsapiens.UCSC.hg38

querySNPFile <- system.file("extdata", "SNP_example.txt", package = "branchpointer")
querySNP <- readQueryFile(querySNPFile, queryType = "SNP", exons = exons, filter = FALSE)
predictionsSNP <- predictBranchpoints(querySNP, queryType = "SNP", BSgenome = g)

summarySNP <- predictionsToSummary(querySNP, predictionsSNP)
```

| | |
|---------------|--------------------------|
| readQueryFile | <i>Read a query file</i> |
|---------------|--------------------------|

Description

Reads and formats a manually generated query file, and finds relative locations of the closest annotated exons. Converts unstranded SNPs to two entries for each strand. Checks for duplicate names and replaces if found.

Usage

```
readQueryFile(queryFile, queryType, exons, maxDist = 50, filter = TRUE)
```

Arguments

| | |
|-----------|--|
| queryFile | tab delimited file containing query information. For intronic regions should be in the format: region id, chromosome name, region start, region end, strand. For SNP variants should be in the format: SNP id, chromosome name, SNP position, strand, reference allele (A/T/C/G), alternative allele (A/T/C/G) |
| queryType | type of query file ("SNP" or "region") |
| exons | GRanges containing exon co-ordinates. Should be produced by gtfToExons() |
| maxDist | maximum distance a SNP can be from an annotated 3' exon. |
| filter | remove SNP queries prior to finding nearest exons. |

Value

Formatted query GRanges

Author(s)

Beth Signal

Examples

```
smallExons <- system.file("extdata", "gencode.v26.annotation.small.gtf", package = "branchpointer")
exons <- gtfToExons(smallExons)
```

```
querySNPFile <- system.file("extdata", "SNP_example.txt", package = "branchpointer")
querySNP <- readQueryFile(querySNPFile, queryType = "SNP", exons)
```

```
queryIntronFile <- system.file("extdata", "intron_example.txt", package = "branchpointer")
queryIntron <- readQueryFile(queryIntronFile, queryType = "region", exons)
```

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