

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)

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Biostrings, parallel, utils, stats,
BSgenome.Hsapiens.UCSC.hg38, rtracklayer, GenomicRanges,
GenomeInfoDb, IRanges, S4Vectors, data.table

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VignetteBuilder knitr

biocViews Software, GenomeAnnotation, GenomicVariation,
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Contents

| | |
|----------------------------------|---|
| exonsToIntrons | 2 |
| getBranchpointSequence | 2 |
| getCanonical3SS | 3 |
| getExonDists | 4 |

| | |
|---|----|
| getPPT | 4 |
| getQueryLoc | 5 |
| gtfToExons | 5 |
| makeBranchpointWindowForExons | 6 |
| makeBranchpointWindowForSNP | 7 |
| plotBranchpointWindow | 7 |
| plotStructure | 8 |
| predictBranchpoints | 9 |
| predictionsToSummary | 10 |
| readQueryFile | 11 |

| | |
|--------------|-----------|
| Index | 12 |
|--------------|-----------|

| | |
|----------------|--|
| exonsToIntrons | <i>Convert exon annotation GRanges to intron locations</i> |
|----------------|--|

Description

Converts exon annotation to intron locations overlapping the branchpoint region for exculsion of non-branchpoint region SNPs Returns a character vector of chromosome locations

Usage

```
exonsToIntrons(exons, maxDist = 50)
```

Arguments

| | |
|---------|--|
| exons | GRanges containing exon co-ordinates. Should be produced by gtfToExons() |
| maxDist | Maximum distance from the 3' exon to create the branchpoint region. |

Value

GRanges containing intron window co-ordinates

Author(s)

Beth Signal

| | |
|------------------------|--|
| getBranchpointSequence | <i>Get branchpoint sequence features Gets intronic sequence covering the branchpoint window and extracts predictive features</i> |
|------------------------|--|

Description

Get branchpoint sequence features Gets intronic sequence covering the branchpoint window and extracts predictive features

Usage

```
getBranchpointSequence(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 with all features required to predict branchpoint probability scores

Author(s)

Beth Signal

| | |
|-----------------|---|
| getCanonical3SS | <i>Get locations of the first five AG 3' splice site motifs</i> |
|-----------------|---|

Description

Takes a variable length vector of distances to the AG motif, sorts and returns the first five. If there are less than five elements in the vector, returns the sorted vector and fills the remainder of the values with 300.

Usage

```
getCanonical3SS(ag)
```

Arguments

| | |
|----|--|
| ag | Vector of distances to the AG splice site motif. |
|----|--|

Value

Locations of the first five AG dinucleotides

Author(s)

Beth Signal

`getExonDists`*Get the closest 3' and 5' exons*

Description

Finds the closest annotated exons from a genomic co-ordinate. Returns the distance to the 3' exon, distance to the 5' exon, ids of the 3' and 5' exon, and if the exons are from the same parent gene

Usage

```
getExonDists(query, exons, queryType)
```

Arguments

| | |
|-----------|---|
| query | GenomicRangesquery |
| exons | GenomicRanges containing exon co-ordinates. Should be produced by <code>gtfToExons()</code> |
| queryType | type of query. "SNP" or "region" |

Value

GenomicRanges with distance to the closest 3' and 5' exons, whether these exons are part of the same gene (i.e is the location intronic), and the identifiers for the 3' and 5' exons.

Author(s)

Beth Signal

`getPPT`*Get the best polypyrimidine tract*

Description

Takes a query genomic sequence, finds all potential polypyrimidine tracts (PPTs) between the test site and the annotated 3' exon. Returns the distance to the start of the longest PPT, and its length.

Usage

```
getPPT(attributes)
```

Arguments

| | |
|------------|--------------------------------|
| attributes | query attributes GenomicRanges |
|------------|--------------------------------|

Value

distance to the start of the longest PPT, and its length

Author(s)

Beth Signal

| | |
|-------------|--|
| getQueryLoc | <i>Find the closest 3' and 5' exons to a branchpointer query</i> |
|-------------|--|

Description

Finds the closest annotated exons from genomic co-ordinates in a branchpointer query GRanges

Usage

```
getQueryLoc(query, queryType, maxDist = 50, filter = TRUE, exons)
```

Arguments

| | |
|-----------|--|
| query | branchpointer query GenomicRanges must have chromosome at position 2, genomic co-ordinate at position 3, and strand at position 4. |
| queryType | type of query file ("SNP" or "region") |
| maxDist | maximum distance a SNP can be from an annotated 3' exon. |
| filter | remove SNP queries prior to finding finding nearest exons. |
| exons | data.frame containing exon co-ordinates. Should be produced by gtfToExons() |

Value

GenomicRanges with the query and its location relative to the 3' and 5' exons

Author(s)

Beth Signal

| | |
|------------|---|
| 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

Make branchpoint window regions

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 refsnps ids*

Description

Searches Biomart for refsnps 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

| | |
|-----------------------|---|
| <code>refSNP</code> | Vector of refsnps ids |
| <code>mart.snp</code> | biomaRt mart object specifying the BioMart database and dataset to be used |
| <code>exons</code> | GRanges containing exon co-ordinates. Should be produced by <code>gtfToExons()</code> |
| <code>maxDist</code> | maximum distance a SNP can be from an annotated 3' exon. |
| <code>filter</code> | remove SNP queries prior to 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", "encode.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)
```

plotStructure *Plots transcript structures*

Description

Plots transcript structures

Usage

```
plotStructure(exonID, exons, keepTranscripts = "overlapping")
```

Arguments

| | |
|-----------------|---|
| exonID | id of the exon to plot |
| exons | Granges containing exon co-ordinates. |
| keepTranscripts | which transcripts to plot ("overlapping" or "withExon") "overlapping" will plot all transcripts overlapping the exon, whereas "withExon" will plot all transcripts containing the exon. |

Value

ggplot2 plot transcript structures

Author(s)

Beth Signal

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

| | |
|--------------------------------|--|
| <code>query</code> | query GRanges containing all SNP ids to be summarised |
| <code>predictions</code> | site-wide branchpoint probability predictions produced from <code>predictBranchpoints()</code> |
| <code>probabilityCutoff</code> | Value to be used as the cutoff for discriminating branchpoint sites from non-branchpoint sites (default = 0.52) |
| <code>probabilityChange</code> | 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 realtive 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 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)
```

Index

* internal

- exonsToIntrons, [2](#)
- getBranchpointSequence, [2](#)
- getCanonical3SS, [3](#)
- getExonDists, [4](#)
- getPPT, [4](#)
- getQueryLoc, [5](#)
- plotStructure, [8](#)

exonsToIntrons, [2](#)

- getBranchpointSequence, [2](#)
- getCanonical3SS, [3](#)
- getExonDists, [4](#)
- getPPT, [4](#)
- getQueryLoc, [5](#)
- gtfToExons, [5](#)

- makeBranchpointWindowForExons, [6](#)
- makeBranchpointWindowForSNP, [7](#)

- plotBranchpointWindow, [7](#)
- plotStructure, [8](#)
- predictBranchpoints, [9](#)
- predictionsToSummary, [10](#)

readQueryFile, [11](#)