Package: branchpointer (via r-universe)

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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, Seqinfo, IRanges, S4Vectors, data.table

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VignetteBuilder knitr

biocViews Software, GenomeAnnotation, GenomicVariation, MotifAnnotation

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`gtfToExons` *Convert GTF file to exon location file*

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("ENSE00003518965.1", "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 predicitons 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 probaility 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 proability 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 occuring 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 *Read a query file*

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)
```

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