Package 'motifbreakR'

April 12, 2022

Title A Package For Predicting The Disruptiveness Of Single Nucleotide Polymorphisms On Transcription Factor Binding Sites

Version 2.8.0

Description We introduce motifbreakR, which allows the biologist to judge in the first place whether the sequence surrounding the polymorphism is a good match, and in the second place how much information is gained or lost in one allele of the polymorphism relative to another. MotifbreakR is both flexible and extensible over previous offerings; giving a choice of algorithms for interrogation of genomes with motifs from public sources that users can choose from; these are 1) a weighted-sum probability matrix, 2) log-probabilities, and 3) weighted by relative entropy. MotifbreakR can predict effects for novel or previously described variants in public databases, making it suitable for tasks beyond the scope of its original design.

Lastly, it can be used to interrogate any genome curated within Bioconductor (currently there are 22).

Depends R (>= 3.5.0), grid, MotifDb

Imports methods, compiler, grDevices, grImport, stringr, BiocGenerics, S4Vectors (>= 0.9.25), IRanges, GenomeInfoDb, GenomicRanges, Biostrings, BSgenome, rtracklayer, VariantAnnotation, BiocParallel, motifStack, Gviz, matrixStats, TFMPvalue, SummarizedExperiment

Suggests BSgenome. Hsapiens. UCSC. hg19, SNPlocs. Hsapiens. dbSNP.20120608, SNPlocs. Hsapiens. dbSNP142. GRCh37, knitr, rmarkdown, BSgenome. Drerio. UCSC. dan Rer7, BiocStyle

VignetteBuilder knitr

License GPL-2 LazyData true

BugReports https://github.com/Simon-Coetzee/motifbreakR/issues

biocViews ChIPSeq, Visualization, MotifAnnotation

NeedsCompilation no

2 calculatePvalue

R topics documented:

omer		
notifbreakR		
lotMB		
nps.from.file		17

calculatePvalue

Calculate the significance of the matches for the reference and alternate alleles for the for their PWM $\,$

Description

Calculate the significance of the matches for the reference and alternate alleles for the for their PWM

Usage

```
calculatePvalue(
  results,
  background = c(A = 0.25, C = 0.25, G = 0.25, T = 0.25)
)
```

encodemotif 3

Arguments

results The output of motifbreakR that was run with filterp=TRUE background Numeric Vector; the background probabilities of the nucleotides

Details

This function is intended to be used on a selection of results produced by motifbreakR, and this can be (although not always) a very memory and time intensive process if the algorithm doesn't converge rapidly.

Value

a GRanges object. The same Granges object that was input as results, but with Refpvalue and Altpvalue columns in the output modified from NA to the p-value calculated by TFMsc2pv.

Source

H\'el\'ene Touzet and Jean-St\'ephane Varr\'e (2007) Efficient and accurate P-value computation for Position Weight Matrices. Algorithms for Molecular Biology, **2: 15**.

See Also

See TFMsc2pv from the **TFMPvalue** package for information about how the p-values are calculated.

Examples

```
data(example.results)
rs2661839 <- example.results[names(example.results) %in% "rs2661839"]
rs2661839 <- calculatePvalue(rs2661839)</pre>
```

encodemotif MotifDb object containing motif information from the known and discovered motifs for the ENCODE TF ChIP-seq datasets.

Description

From the abstract: "Recent advances in technology have led to a dramatic increase in the number of available transcription factor ChIP-seq and ChIP-chip data sets. Understanding the motif content of these data sets is an important step in understanding the underlying mechanisms of regulation. Here we provide a systematic motif analysis for 427 human ChIP-seq data sets using motifs curated from the literature and also discovered de novo using five established motif discovery tools. We use a systematic pipeline for calculating motif enrichment in each data set, providing a principled way for choosing between motif variants found in the literature and for flagging potentially problematic data sets. Our analysis confirms the known specificity of 41 of the 56 analyzed factor groups and reveals motifs of potential cofactors. We also use cell type-specific binding to find factors active

4 encodemotif

in specific conditions. The resource we provide is accessible both for browsing a small number of factors and for performing large-scale systematic analyses. We provide motif matrices, instances and enrichments in each of the ENCODE data sets. The motifs discovered here have been used in parallel studies to validate the specificity of antibodies, understand cooperativity between data sets and measure the variation of motif binding across individuals and species."

Usage

encodemotif

Format

```
MotifDb object of length 2064; to access metadata use mcols(encodemotif)
providerName Name provided by ENCODE
providerId Same as providerName
dataSource "ENCODE-motif"
geneSymbol Gene symbol for the transcription factor
geneId Entrez gene id for the transcription factor
geneIdType "ENTREZ"
proteinId UNIPROT id for the transcription factor
proteinIdType "UNIPROT"
organism "Hsapiens"
sequenceCount NA not available
bindingSequence Consensus sequence for the motif
bindingDomain NA incomplete
tfFamily NA incomplete
experimentType occurs in two forms:
    For motifs that were discovered in this study, the format is cellType_source-LabMetadata: MotifFinder#Location
    for example H1-hESC_encode-Myers_seq_hsa_v041610.2_r1:MEME#2#Intergenic.
    For motifs that were "known" the format tends to be TF_source_sourceId for example
    AP1_jaspar_MA0099.2.
pubmedID "24335146" see Source for more details
```

Details

Load with data(encodemotif)

Value

MotifList-class object

Source

Pouya Kheradpour and Manolis Kellis (2013 December 13) Systematic discovery and characterization of regulatory motifs in ENCODE TF binding experiments. Nucleic Acids Research, doi:10.1093/nar/gkt1249

example.results 5

See Also

```
http://compbio.mit.edu/encode-motifs/
```

Examples

```
data(encodemotif)
encodemotif
```

example.results

Example Results from motifbreakR

Description

This contains example results from motifbreaker for use in examples from the help docs

Usage

```
example.results
```

Format

GRanges output from motifbreakR

Value

GRanges object. See motifbreakR for information on it's structure.

Examples

```
data(example.results)
example.results
```

factorbook

MotifDb object containing motif information from around the genomic regions bound by 119 human transcription factors in Factorbook.

6 factorbook

Description

From the abstract: "Chromatin immunoprecipitation coupled with high-throughput sequencing (ChIP-seq) has become the dominant technique for mapping transcription factor (TF) binding regions genome-wide. We performed an integrative analysis centered around 457 ChIP-seq data sets on 119 human TFs generated by the ENCODE Consortium. We identified highly enriched sequence motifs in most data sets, revealing new motifs and validating known ones. The motif sites (TF binding sites) are highly conserved evolutionarily and show distinct footprints upon DNase I digestion. We frequently detected secondary motifs in addition to the canonical motifs of the TFs, indicating tethered binding and cobinding between multiple TFs. We observed significant position and orientation preferences between many cobinding TFs. Genes specifically expressed in a cell line are often associated with a greater occurrence of nearby TF binding in that cell line. We observed cell-line-specific secondary motifs that mediate the binding of the histone deacetylase HDAC2 and the enhancer-binding protein EP300. TF binding sites are located in GC-rich, nucleosome-depleted, and DNase I sensitive regions, flanked by well-positioned nucleosomes, and many of these features show cell type specificity. The GC-richness may be beneficial for regulating TF binding because, when unoccupied by a TF, these regions are occupied by nucleosomes in vivo. We present the results of our analysis in a TF-centric web repository Factorbook (http://factorbook.org) and will continually update this repository as more ENCODE data are generated."

Usage

factorbook

Format

```
MotifDb object of length 79; to access metadata use mcols(factorbook)
```

```
providerName Name listed in meme output of 'Supp TableS2.pdf' for the citation indicated below providerId Same as providerName dataSource "FactorBook" geneSymbol NA these motifs don't have a direct 1 to 1 relationship with a transcription factor geneId NA geneIdType NA proteinId NA proteinId NA proteinIdType NA organism "Hsapiens" sequenceCount NA bindingSequence Consensus sequence for the motif bindingDomain NA tfFamily NA experimentType NA pubmedID "22955990" see Source for more details
```

Details

Load with data(factorbook)

hocomoco 7

Value

MotifList-class object

Source

J Wang, J Zhuang, S Iyer, XY Lin, et al. (2012) Sequence features and chromatin structure around the genomic regions bound by 119 human transcription factors. Genome Research, **22** (**9**), 1798-1812, doi:10.1101/gr.139105.112

See Also

http://factorbook.org

Examples

data(factorbook) factorbook

hocomoco

MotifDb object containing motif information from Homo Sapiens Comprehensive Model Collection (HOCOMOCO) of transcription factor (TF) binding models

Description

From the abstract: "We present the Homo sapiens comprehensive model collection (HOCOMOCO, http://autosome.ru/HOCOMOCO/, http://cbrc.kaust.edu.sa/hocomoco/) containing carefully hand-curated TFBS models constructed by integration of binding sequences obtained by both lowand high-throughput methods. To construct position weight matrices to represent these TFBS models, we used ChIPMunk software in four computational modes, including newly developed periodic positional prior mode associated with DNA helix pitch. We selected only one TFBS model per TF, unless there was a clear experimental evidence for two rather distinct TFBS models. We assigned a quality rating to each model. HOCOMOCO contains 426 systematically curated TFBS models for 401 human TFs, where 172 models are based on more than one data source."

Usage

hocomoco

Format

MotifDb object of length 426; to access metadata use mcols(hocomoco)

providerName Name provided by HOCOMOCO
providerId ID provided by HOCOMOCO including experiment type

dataSource "HOCOMOCO"

geneSymbol Gene symbol for the transcription factor

8 hocomoco

```
geneId Entrez gene id for the transcription factor
geneIdType "ENTREZ"
proteinId UNIPROT id for the transcription factor
proteinIdType "UNIPROT"
organism "Hsapiens"
sequenceCount Number of sequences evaluated for producing the PWM
bindingSequence Consensus sequence for the motif
bindingDomain NA incomplete
tfFamily NA incomplete
experimentType from http://autosome.ru/HOCOMOCO/Details.php#200 quoted here:
```

"TFBS model identification modes

To construct TFBS models ChIPMunk was run four times: two times (f1) and (f2) with uniform model positional prior and two times (si) and (do) with informative model positional prior.

The min-to-max (f1) model length estimation mode was used with the min length of 7 bp and increasing it by 1 bp until the default max length of 25 bp was reached following the optimal length selection procedure as in Kulakovskiy and Makeev, Biophysics, 2009. For max-to-min (f2) model length estimation mode we started from 25 bp and searched for the best alignment decreasing the length by 1 bp until the minimal length of 7 bp. We also used the single (si) and double box (do) model positional priors in order to simulate DNA helix turn. For a single box, the positional weights are to be distributed as $\cos 2(pi \text{ n} / \text{T})$, where T=10.5 is the DNA helix pitch, n is the coordinate within the alignment, and the center of the alignment of the length L is at n=0. During the internal cycle of PWM optimization the PWM column scores are multiplied by prior values so the columns closer to the center of the alignment (n=0) receive no score penalty while the columns around (n = 5,6,-5,-6) contribute much less to the score of the PWM under optimization. The single box model prior was used along with the min-to-max length estimation mode (si). We also used the double box model prior with a shape prior equal to $\sin 2(pin / T)$, which was used to search for possibly longer double box models in the max-to-min length estimation mode (do).

Model quality assignment

The resulting models were rated (from A to F) according to their quality. Model quality rates from A-to-D were assigned to proteins known to be TFs, including those listed in Schaefer et al., Nucleic Acids Research, 2011 with addition of a number of proteins having relevant models and sufficient evidence to be TFs. The ratings were assigned by human curation according to the following criteria:

Relevant distribution of position-specific information content over alignment columns, which means a model LOGO representation displaying well formed core positions with a high information content surrounded by flanking letters with lower information content; the information content at flanking positions decreasing with the distance from the model core.

"Stability", which means that in more than one of the ChIPMunk modes we obtained models with a similar length, consensus, and comparable number of aligned binding sites, along with a similar shape of model LOGO representation. "Similarity" of the model to the binding sequence consensus for this TF given in the UniProt or other databases, which means similarity of the shape of the model LOGO and TFBS lengths to those of other TFs from the same TF family. "A total number of binding sites" was also considered as a quality measure, as a large

homer 9

set of binding regions (mostly but not limited to ChIP-Seq and parallel SELEX) implies that there are many observations of each letter in any position of the alignment, particularly many observations of non-consensus letters in core positions. In positions with low information content, where there is no strong consensus, all variants have many observations, and thus the observed letter frequencies are less dependent on statistical fluctuations.

Quality A was assigned to high confidence models complying with all four criteria listed in the section above. Quality B was assigned to models built from large sequence sets that failed no more than one out of the three remaining criteria. Quality C was assigned to models built from small sequence sets but (with a number of specifically marked exceptions) complying with the three remaining criteria. Quality D models missed part of the known consensus sequence or had no clearly significant core positions in the TFBS model. Quality E (error) was assigned to models for proteins not convincingly shown to be TFs or to models exhibiting an irrelevant LOGO shape or a wrong consensus sequence. Quality F (failure) was assigned to TFs for which there was no reliable model identified."

pubmedID "23175603" see Source for more details

Details

Load with data(hocomoco)

Value

MotifList-class object

Source

Kulakovskiy,I.V., Medvedeva,Y.A., Schaefer,U., Kasianov,A.S., Vorontsov,I.E., Bajic,V.B. and Makeev,V.J. (2013) HOCOMOCO: a comprehensive collection of human transcription factor binding sites models. Nucleic Acids Research, **41**, D195–D202.

See Also

http://autosome.ru/HOCOMOCO/http://cbrc.kaust.edu.sa/hocomoco/

Examples

data(hocomoco) hocomoco

homer

MotifDb object containing motif information from motif databases included in HOMER. 10 homer

Description

From the website: "Homer includes several motif databases that are used to help annotate results and conduct searches for known motifs. HOMER contains a custom motif database based on independent analysis of mostly ChIP-Seq data sets which is heavily utilized in the software." See http://homer.salk.edu/homer/motif/motifDatabase.html for more information on how these files were generated, and Homer's sources.

Usage

homer

Format

```
MotifDb object of length 247; to access metadata use mcols(homer)
providerName Name provided HOMER
providerId Factor Name provided by HOMER
dataSource "HOMER"
geneSymbol Symbol provided by HOMER
geneId Entrez gene id for the transcription factor
geneIdType "ENTREZ"
proteinId UNIPROT id for the transcription factor
proteinIdType "UNIPROT"
organism "Hsapiens"
sequenceCount NA
bindingSequence Consensus sequence for the motif
bindingDomain DBD provided by HOMER
tfFamily NA
experimentType The Celltype, IP, Assay, and GEO id if applicable for the motif
pubmedID "20513432" see Source for more details
```

Details

Load with data(homer)

Value

MotifList-class object

Source

Heinz S, Benner C, Spann N, Bertolino E et al. (2010 May 28) Simple Combinations of Lineage-Determining Transcription Factors Prime cis-Regulatory Elements Required for Macrophage and B Cell Identities. Mol Cell, **38(4):576-589**. PMID: 20513432

See Also

http://homer.salk.edu/homer/index.html http://homer.salk.edu/homer/motif/motifDatabase.html http://homer.salk.edu/homer/motif/HomerMotifDB/homerResults.html

Examples

```
data(homer)
homer
```

motifbreakR

Predict The Disruptiveness Of Single Nucleotide Polymorphisms On Transcription Factor Binding Sites.

Description

Predict The Disruptiveness Of Single Nucleotide Polymorphisms On Transcription Factor Binding Sites.

Usage

```
motifbreakR(
   snpList,
   pwmList,
   threshold = 0.85,
   filterp = FALSE,
   method = "default",
   show.neutral = FALSE,
   verbose = FALSE,
   bkg = c(A = 0.25, C = 0.25, G = 0.25, T = 0.25),
   legacy.score = TRUE,
   BPPARAM = bpparam()
)
```

Arguments

snpList	The output of snps.from.rsid or snps.from.file
pwmList	An object of class MotifList containing the motifs that you wish to interrogate
threshold	Numeric; the maximum p-value for a match to be called or a minimum score threshold
filterp	Logical; filter by p-value instead of by pct score.
method	Character; one of default, log, ic, or notrans; see details.
show.neutral	Logical; include neutral changes in the output
verbose	Logical; if running serially, show verbose messages
bkg	Numeric Vector; the background probabilites of the nucleotides used with method=log method=ic

BPPARAM

a BiocParallel object see register and see getClass("BiocParallelParam") for additional parameter classes. Try BiocParallel::registered() to see what's availible and for example BiocParallel::bpparam("SerialParam") would allow serial evaluation.

Details

motifbreakR works with position probability matrices (PPM). PPM are derived as the fractional occurrence of nucleotides A,C,G, and T at each position of a position frequency matrix (PFM). PFM are simply the tally of each nucleotide at each position across a set of aligned sequences. With a PPM, one can generate probabilities based on the genome, or more practically, create any number of position specific scoring matrices (PSSM) based on the principle that the PPM contains information about the likelihood of observing a particular nucleotide at a particular position of a true transcription factor binding site. What follows is a discussion of the three different algorithms that may be employed in calls to the **motifbreakR** function via the method argument.

Suppose we have a frequency matrix M of width n (i.e. a PPM as described above). Furthermore, we have a sequence s also of length n, such that $s_i \in \{A, T, C, G\}, i = 1, \dots n$. Each column of M contains the frequencies of each letter in each position.

Commonly in the literature sequences are scored as the sum of log probabilities:

Equation 1

$$F(s, M) = \sum_{i=1}^{n} \log(\frac{M_{s_i, i}}{b_{s_i}})$$

where b_{s_i} is the background frequency of letter s_i in the genome of interest. This method can be specified by the user as method='log'.

As an alternative to this method, we introduced a scoring method to directly weight the score by the importance of the position within the match sequence. This method of weighting is accessed by specifying method='ic' (information content). A general representation of this scoring method is given by:

Equation 2

$$F(s, M) = p_s \cdot \omega_M$$

where p_s is the scoring vector derived from sequence s and matrix M, and w_M is a weight vector derived from M. First, we compute the scoring vector of position scores p

Equation 3

$$p_s = (M_{s_i,i}) \quad \text{where} \quad \frac{i = 1, \dots n}{s_i \in \{A,C,G,T\}}$$

and second, for each M a constant vector of weights $\omega_M = (\omega_1, \omega_2, \dots, \omega_n)$.

There are two methods for producing ω_M . The first, which we call weighted sum, is the difference in the probabilities for the two letters of the polymorphism (or variant), *i.e.* Δp_{s_i} , or the difference of the maximum and minimum values for each column of M:

Equation 4.1

$$\omega_i = \max\{M_i\} - \min\{M_i\}$$
 where $i = 1, \dots n$

The second variation of this theme is to weight by relative entropy. Thus the relative entropy weight for each column i of the matrix is given by:

Equation 4.2

$$\omega_i = \sum_{j \in \{A,C,G,T\}} M_{j,i} \log_2(\frac{M_{j,i}}{b_i}) \quad \text{ where } \quad i = 1, \dots n$$

where b_i is again the background frequency of the letter i.

Thus, there are 3 possible algorithms to apply via the method argument. The first is the standard summation of log probabilities (method='log'). The second and third are the weighted sum and information content methods (method='default' and method='ic') specified by equations 4.1 and 4.2, respectively. **motifbreakR** assumes a uniform background nucleotide distribution (b) in equations 1 and 4.2 unless otherwise specified by the user. Since we are primarily interested in the difference between alleles, background frequency is not a major factor, although it can change the results. Additionally, inclusion of background frequency introduces potential bias when collections of motifs are employed, since motifs are themselves unbalanced with respect to nucleotide composition. With these cautions in mind, users may override the uniform distribution if so desired. For all three methods, **motifbreakR** scores and reports the reference and alternate alleles of the sequence ($F(s_{REF}, M)$) and $F(s_{ALT}, M)$), and provides the matrix scores $p_{s_{REF}}$ and $p_{s_{ALT}}$ of the SNP (or variant). The scores are scaled as a fraction of scoring range 0-1 of the motif matrix, M. If either of $F(s_{REF}, M)$ and $F(s_{ALT}, M)$ is greater than a user-specified threshold (default value of 0.85) the SNP is reported. By default **motifbreakR** does not display neutral effects, ($\Delta p_i < 0.4$) but this behaviour can be overridden.

Additionally, now, with the use of TFMPvalue-package, we may filter by p-value of the match. This is unfortunately a two step process. First, by invoking filterp=TRUE and setting a threshold at a desired p-value e.g 1e-4, we perform a rough filter on the results by rounding all values in the PWM to two decimal place, and calculating a scoring threshold based upon that. The second step is to use the function calculatePvalue() on a selection of results which will change the Refpvalue and Altpvalue columns in the output from NA to the p-value calculated by TFMsc2pv. This can be (although not always) a very memory and time intensive process if the algorithm doesn't converge rapidly.

Value

a GRanges object containing:

REF the reference allele for the SNP
ALT the alternate allele for the SNP
snpPos the coordinates of the SNP

motifPos the coordinates of the SNP within the TF binding motif

geneSymbol the geneSymbol corresponding to the TF of the TF binding motif

dataSource the source of the TF binding motif

providerName, providerId					
	the name and id provided by the source				
seqMatch	the sequence on the 5' -> 3' direction of the "+" strand that corresponds to DNA at the position that the TF binding motif was found.				
pctRef	The score as determined by the scoring method, when the sequence contains the reference SNP allele, normalized to a scale from $0 - 1$. If filterp = FALSE, this is the value that is thresholded.				
pctAlt	The score as determined by the scoring method, when the sequence contains the alternate SNP allele, normalized to a scale from $0 - 1$. If filterp = FALSE, this is the value that is thresholded.				
scoreRef	The score as determined by the scoring method, when the sequence contains the reference SNP allele				
scoreAlt	The score as determined by the scoring method, when the sequence contains the alternate SNP allele				
Refpvalue	p-value for the match for the pctRef score, initially set to NA. see calculatePvalue for more information $ \frac{1}{2} \int_{\mathbb{R}^{n}} \frac{1}{2} \left(\frac{1}{2} \int_{\mathbb{R}^{$				
Altpvalue	p-value for the match for the pctAlt score, initially set to NA. see calculatePvalue for more information				
alleleRef	The proportional frequency of the reference allele at position motifPos in the motif				
alleleAlt	The proportional frequency of the alternate allele at position motifPos in the motif				
effect	one of weak, strong, or neutral indicating the strength of the effect.				
each SNP in this object may be plotted with plotMB					

See Also

See snps.from.rsid and snps.from.file for information about how to generate the input to this function and plotMB for information on how to visualize it's output

Examples

motifbreakR_motif

motifbreakR_motif

MotifDb object containing motif information from the motif databases of HOCOMOCO, Homer, FactorBook and ENCODE

Description

This object contains all the MotifList-class objects that were generated for this package. See the individual help sections for hocomoco, homer, factorbook, and encodemotif, for how the data is formatted.

Usage

motifbreakR_motif

Format

MotifDb object of length 2816; to access metadata use mcols(motifbreakR motif)

Details

Load with data(motifbreakR_motif)

Value

MotifList-class object

Source

Kulakovskiy,I.V., Medvedeva,Y.A., Schaefer,U., Kasianov,A.S., Vorontsov,I.E., Bajic,V.B. and Makeev,V.J. (2013) HOCOMOCO: a comprehensive collection of human transcription factor binding sites models. Nucleic Acids Research, **41**, D195–D202.

Heinz S, Benner C, Spann N, Bertolino E et al. (2010 May 28) Simple Combinations of Lineage-Determining Transcription Factors Prime cis-Regulatory Elements Required for Macrophage and B Cell Identities. Mol Cell, **38**(4):**576-589**. PMID: 20513432

J Wang, J Zhuang, S Iyer, XY Lin, et al. (2012) Sequence features and chromatin structure around the genomic regions bound by 119 human transcription factors. Genome Research, **22** (**9**), 1798-1812, doi:10.1101/gr.139105.112

Pouya Kheradpour and Manolis Kellis (2013 December 13) Systematic discovery and characterization of regulatory motifs in ENCODE TF binding experiments. Nucleic Acids Research, doi:10.1093/nar/gkt1249

See Also

hocomoco, homer, factorbook, and encodemotif

16 plotMB

Examples

```
data(motifbreakR_motif)
motifbreakR_motif
```

plotMB Plot a genomic region surrounding a genomic variant, and potentially disrupted motifs

Description

Plot a genomic region surrounding a genomic variant, and potentially disrupted motifs

Usage

```
plotMB(results, rsid, reverseMotif = TRUE, effect = c("strong", "weak"))
```

Arguments

results The output of motifbreakR

rsid Character; the identifier of the variant to be visualized

reverseMotif Logical; if the motif is on the "-" strand show the the motifs as reversed FALSE

or reverse complement TRUE

effect Character; show motifs that are strongly effected c("strong"), weakly effected

c("weak"), or both c("strong", "weak")

Details

plotMB produces output showing the location of the SNP on the chromosome, the surrounding sequence of the + strand, the footprint of any motif that is disrupted by the SNP or SNV, and the DNA sequence motif(s)

Value

plots a figure representing the results of motifbreakR at the location of a single SNP, returns invisible NULL.

See Also

See motifbreakR for the function that produces output to be visualized here, also snps.from.rsid and snps.from.file for information about how to generate the input to motifbreakR function.

Examples

```
data(example.results)
example.results
## Not run:
plotMB(example.results, "rs2661839", effect = "strong")
## End(Not run)
```

scoreAllWindows 17

```
scoreAllWindows
```

 $scoreMotif.a <- function(snp.seq, ppm, len, offset = 1) \ snp.seq <- snp.seq[offset:(offset + len - 1)] \ \#\# \ diag \ code \ position.probs <- c(ppm[snp.seq,])[1L + 0L:(len - 1L) * (len + 1L)] \ return(position.probs) \#" @importFrom \ compiler \ cmpfun \ scoreMotif <- cmpfun(scoreMotif.a, options = list(optimize = 3))$

Description

scoreMotif.a <- function(snp.seq, ppm, len, offset = 1) snp.seq <- snp.seq[offset:(offset + len - 1)] ## diag code position.probs <- c(ppm[snp.seq,])[1L + 0L:(len - 1L) * (len + 1L)] return(position.probs) #' @importFrom compiler cmpfun scoreMotif <- cmpfun(scoreMotif.a, options = list(optimize = 3))

Usage

```
scoreAllWindows(
   snp.seq,
   snp.seq.rc,
   pwm,
   from = "default",
   to = "default",
   pwm.range = NULL,
   calcp = TRUE
)
```

snps.from.file

Import SNPs from a BED file or VCF file for use in motifbreakR

Description

Import SNPs from a BED file or VCF file for use in motifbreakR

Usage

```
snps.from.file(
  file = NULL,
  dbSNP = NULL,
  search.genome = NULL,
  format = "bed",
  indels = FALSE
)
```

18 snps.from.file

Arguments

file Character; a character containing the path to a bed file or a vcf file see Details

for a description of the required format

dbSNP OPTIONAL; an object of class SNPlocs to lookup rsids; see availible.SNPs

in injectSNPs to check for available SNPlocs

search.genome an object of class BSgenome for the species you are interrogating; see available.genomes

for a list of species

format Character; one of bed or vcf

Details

snps.from.file takes a character vector describing the file path to a bed file that contains the necissary information to generate the input for motifbreakR see http://www.genome.ucsc.edu/FAQ/FAQformat.html#format1 for a complete description of the BED format. Our convention deviates in that there is a required format for the name field. name is defined as chromosome:start:REF:ALT or the rsid from dbSNP (if you've included the optional SNPlocs argument). For example if you were to include rs123 in it's alternate format it would be entered as chr7:24966446:C:A

Value

a GRanges object containing:

SNP_id The rsid of the snp with the "rs" portion stripped

alleles_as_ambig

THE IUPAC ambiguity code between the reference and alternate allele for this

SNP

REF The reference allele for the SNP

ALT The alternate allele for the SNP

See Also

See motifbreakR for analysis; See snps.from.rsid for an alternate method for generating a list of variants.

Examples

snps.from.rsid

snps.from.rsid	Import SNPs from rsid for use in motifbreakR
onportromora	import sitt s from i sta for tise in mongoreanax

Description

Import SNPs from rsid for use in motifbreakR

Usage

```
snps.from.rsid(rsid = NULL, dbSNP = NULL, search.genome = NULL)
```

Arguments

rsid Character; a character vector of rsid values from dbSNP

dbSNP an object of class SNPlocs to lookup rsids; see availible . SNPs in injectSNPs

to check for availible SNPlocs

search.genome an object of class BSgenome for the species you are interrogating; see available.genomes

for a list of species

Details

snps.from.rsid take an rsid, or character vector of rsids and generates the required object to input into motifbreakR

Value

a GRanges object containing:

SNP_id The rsid of the snp with the "rs" portion stripped

alleles_as_ambig

THE IUPAC ambiguity code between the reference and alternate allele for this

SNP

REF The reference allele for the SNP

ALT The alternate allele for the SNP

See Also

See motifbreakR for analysis; See snps.from.file for an alternate method for generating a list of variants.

20 snps.from.rsid

Examples

Index

```
* datasets
    encodemotif, 3
    example.results, 5
    factorbook, 5
    hocomoco, 7
    homer, 9
    motifbreakR_motif, 15
available.genomes, 18, 19
calculatePvalue, 2, 13, 14
encodemotif, 3, 15
{\tt example.results}, {\tt 5}
factorbook, 5, 15
GRanges, 5
hocomoco, 7, 15
homer, 9, 15
injectSNPs, 18, 19
motifbreakR, 3, 5, 11, 16, 18, 19
motifbreakR\_motif, 15
MotifDb, 4, 6, 7, 10, 15
plotMB, 14, 16
register, 12
scoreAllWindows, 17
snps.from.file, 14, 16, 17, 19
snps.from.rsid, 14, 16, 18, 19
TFMsc2pv, 3, 13
```