

# Package ‘atSNP’

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**Type** Package

**Title** Affinity test for identifying regulatory SNPs

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**Description**

atSNP performs affinity tests of motif matches with the SNP or the reference genomes and SNP-led changes in motif matches.

**License** GPL-2

**LinkingTo** Rcpp

**Depends** R (>= 3.6)

**Suggests** BiocStyle, knitr, rmarkdown

**Imports** BSgenome, BiocFileCache, BiocParallel, Rcpp, data.table,  
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**biocViews** Software, ChIPSeq, GenomeAnnotation, MotifAnnotation,  
Visualization

**URL** <https://github.com/sunyoungshin/atSNP>

**BugReports** <https://github.com/sunyoungshin/atSNP/issues>

**VignetteBuilder** knitr

**git\_url** <https://git.bioconductor.org/packages/atSNP>

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atSNP-package

*atSNP: affinity tests for regulatory SNP detection*


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## Description

atSNP implements the affinity test for large sets of SNP-motif interactions using the importance sampling algorithm. Users may identify SNPs that potentially may affect binding affinity of transcription factors. Given a set of SNPs and a library of motif position weight matrices (PWMs), atSNP provides two main functions for analyzing SNP effects: (i) the binding affinity score for each allele and each PWM and the p-values for allele-specific binding affinity scores (ii) the p-values for affinity score changes between the two alleles for each SNP. Compared to other bioinformatics tools that provide similar functionalities, atSNP is highly scalable.

The atSNP main functions are:

1. [LoadMotifLibrary](#) - Load position weight matrices
2. [LoadSNPData](#) - Load the SNP information and code the genome sequences around the SNP locations
3. [LoadFastaData](#) - Load the SNP data from fasta files
4. [ComputeMotifScore](#) - Compute the scores for SNP effects on motifs
5. [ComputePValues](#) - Compute p-values for affinity scores

Some helper functions are:

1. [MatchSubsequence](#) - Compute the matching subsequence
2. [GetIUPACSequence](#) - Get the IUPAC sequence of a motif
3. [dtMotifMatch](#) - Compute the augmented matching subsequence on SNP and reference alleles

The composite logo plotting function is:

1. [plotMotifMatch](#) - Plot sequence logos of the position weight matrix of the motif and sequences of its corresponding best matching augmented subsequence on the reference and SNP allele

### Author(s)

Chandler Zuo Sunyoung Shin <sunyoung.shin@utdallas.edu>

### References

Zuo, Chandler, Shin, Sunyoung, and Keles, Sunduz. (2015). atSNP: Transcription factor binding affinity testing for regulatory SNP detection. *Bioinformatics* 31 (20): 3353-5.

### See Also

atSNP vignette for more information

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|                   |                                                      |
|-------------------|------------------------------------------------------|
| ComputeMotifScore | <i>Compute the scores for SNP effects on motifs.</i> |
|-------------------|------------------------------------------------------|

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### Description

Compute the log-likelihood scores for motifs.

### Usage

```
ComputeMotifScore(motif.lib, snp.info, ncores = 1)
```

### Arguments

|           |                                                                                     |
|-----------|-------------------------------------------------------------------------------------|
| motif.lib | A list object with the output format of function <a href="#">LoadMotifLibrary</a> . |
| snp.info  | A list object with the output format of function <a href="#">LoadSNPData</a> .      |
| ncores    | An integer for the number of parallel process. Default: 1.                          |

### Details

This function computes the binding affinity scores for both alleles at each SNP window. For each pair of SNP and motif, it finds the subsequence from both strand that maximizes the affinity binding score. It returns both the matching positions and the maximized affinity scores.

Value

A list of two data.frame's. Field `snp.tbl` contains:

|                          |                                                             |
|--------------------------|-------------------------------------------------------------|
| <code>snpid</code>       | SNP id.                                                     |
| <code>ref_seq</code>     | Reference allele nucleotide sequence.                       |
| <code>snp_seq</code>     | SNP allele nucleotide sequence.                             |
| <code>ref_seq_rev</code> | Reference allele nucleotide sequence on the reverse strand. |
| <code>snp_seq_rev</code> | SNP allele nucleotide sequence on the reverse strand.       |

Field `motif.score` contains:

|                                             |                                                                                                                    |
|---------------------------------------------|--------------------------------------------------------------------------------------------------------------------|
| <code>motif</code>                          | Name of the motif.                                                                                                 |
| <code>motif_len</code>                      | Length of the motif.                                                                                               |
| <code>ref_start, ref_end, ref_strand</code> | Location of the best matching subsequence on the reference allele.                                                 |
| <code>snp_start, snp_end, snp_strand</code> | Location of the best matching subsequence on the SNP allele.                                                       |
| <code>log_lik_ref</code>                    | Log-likelihood score for the reference allele.                                                                     |
| <code>log_lik_snp</code>                    | Log-likelihood score for the SNP allele.                                                                           |
| <code>log_lik_ratio</code>                  | The log-likelihood ratio.                                                                                          |
| <code>log_enhance_odds</code>               | Difference in log-likelihood ratio between SNP allele and reference allele based on the best matching subsequence. |
| <code>log_reduce_odds</code>                | Difference in log-likelihood ratio between reference allele and SNP allele based on the best matching subsequence. |

Author(s)

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

Examples

```
data(example)
ComputeMotifScore(motif_library, snpInfo, ncores = 2)
```

---

|                |                                              |
|----------------|----------------------------------------------|
| ComputePValues | <i>Compute p-values for affinity scores.</i> |
|----------------|----------------------------------------------|

---

Description

This function computes the p-values for allele-specific affinity scores and between-allele affinity score changes using the importance sampling technique.

Usage

```
ComputePValues(motif.lib, snp.info, motif.scores, ncores = 1,
testing.mc = FALSE, figdir = NULL)
```

**Arguments**

|              |                                                                                                                                                                                                                                                                                                                                                    |       |                        |             |                                                    |             |                                              |
|--------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------|------------------------|-------------|----------------------------------------------------|-------------|----------------------------------------------|
| motif.lib    | A list object with the output format of function <a href="#">LoadMotifLibrary</a> .                                                                                                                                                                                                                                                                |       |                        |             |                                                    |             |                                              |
| snp.info     | A list object with the output format of function <a href="#">LoadSNPData</a> .                                                                                                                                                                                                                                                                     |       |                        |             |                                                    |             |                                              |
| motif.scores | A data.frame object containing at least the following columns: <table data-bbox="467 457 1149 554"> <tr> <td>motif</td><td>The name of the motif.</td></tr> <tr> <td>log_lik_ref</td><td>The log-likelihood score for the reference allele.</td></tr> <tr> <td>log_lik_snp</td><td>The log-likelihood score for the SNP allele.</td></tr> </table> | motif | The name of the motif. | log_lik_ref | The log-likelihood score for the reference allele. | log_lik_snp | The log-likelihood score for the SNP allele. |
| motif        | The name of the motif.                                                                                                                                                                                                                                                                                                                             |       |                        |             |                                                    |             |                                              |
| log_lik_ref  | The log-likelihood score for the reference allele.                                                                                                                                                                                                                                                                                                 |       |                        |             |                                                    |             |                                              |
| log_lik_snp  | The log-likelihood score for the SNP allele.                                                                                                                                                                                                                                                                                                       |       |                        |             |                                                    |             |                                              |
| ncores       | An integer for the number of parallel process. Default: 1.                                                                                                                                                                                                                                                                                         |       |                        |             |                                                    |             |                                              |
| testing.mc   | Monte Carlo sample size of 200 is considered. Do not change the default unless conducting a quick test. Default: FALSE                                                                                                                                                                                                                             |       |                        |             |                                                    |             |                                              |
| figdir       | A string for the path to print p-value plots for monitoring results. Default: NULL (no figure).                                                                                                                                                                                                                                                    |       |                        |             |                                                    |             |                                              |

**Value**

A data.frame extending motif.scores by the following additional columns:

|               |                                                                                  |
|---------------|----------------------------------------------------------------------------------|
| pval_ref      | P-values for scores on the reference allele.                                     |
| pval_snp      | P-values for scores on the SNP allele.                                           |
| pval_cond_ref | Conditional p-values for scores on the reference allele.                         |
| pval_cond_snp | Conditional p-values for scores on the SNP allele.                               |
| pval_diff     | P-values for the difference in scores between the reference and the SNP alleles. |
| pval_rank     | P-values for the log rank ratio between the reference and the SNP alleles.       |

**Author(s)**

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

**Examples**

```
data(example)
ComputePValues(motif_library, snpInfo, motif_scores$motif.scores, ncores = 2, testing.mc=TRUE)
```

---

|              |                                                                                  |
|--------------|----------------------------------------------------------------------------------|
| dtMotifMatch | <i>Compute the augmented matching subsequence on SNP and reference allele s.</i> |
|--------------|----------------------------------------------------------------------------------|

---

**Description**

Calculate the best matching augmented subsequences on both SNP and reference alleles for motifs. Obtain extra unmatching position on the best matching augmented subsequence of the reference and SNP alleles.

**Usage**

```
dtMotifMatch(snp.tbl, motif.scores, snpids = NULL, motifs = NULL,
             motif.lib, ncores = 2)
```

**Arguments**

`snp.tbl` A data.frame with the following information:

|                          |                                                             |
|--------------------------|-------------------------------------------------------------|
| <code>snpid</code>       | SNP id.                                                     |
| <code>ref_seq</code>     | Reference allele nucleobase sequence.                       |
| <code>snp_seq</code>     | SNP allele nucleobase sequence.                             |
| <code>ref_seq_rev</code> | Reference allele nucleobase sequence on the reverse strand. |
| <code>snp_seq_rev</code> | SNP allele nucleobase sequence on the reverse strand.       |

`motif.scores` A data.frame with the following information:

|                                             |                                                                                                                    |
|---------------------------------------------|--------------------------------------------------------------------------------------------------------------------|
| <code>motif</code>                          | Name of the motif.                                                                                                 |
| <code>motif_len</code>                      | Length of the motif.                                                                                               |
| <code>ref_start, ref_end, ref_strand</code> | Location of the best matching subsequence on the reference allele.                                                 |
| <code>snp_start, snp_end, snp_strand</code> | Location of the best matching subsequence on the SNP allele.                                                       |
| <code>log_lik_ref</code>                    | Log-likelihood score for the reference allele.                                                                     |
| <code>log_lik_snp</code>                    | Log-likelihood score for the SNP allele.                                                                           |
| <code>log_lik_ratio</code>                  | The log-likelihood ratio.                                                                                          |
| <code>log_enhance_odds</code>               | Difference in log-likelihood ratio between SNP allele and reference allele based on the best matching subsequence. |
| <code>log_reduce_odds</code>                | Difference in log-likelihood ratio between reference allele and SNP allele based on the best matching subsequence. |

`snpids` A subset of snpids to compute the subsequences. Default: NULL, when all snps are computed.

`motifs` A subset of motifs to compute the subsequences. Default: NULL, when all motifs are computed.

`motif.lib` A list of named position weight matrices.

`ncores` The number of cores used for parallel computing. Default: 10

**Value**

A data.frame containing all columns from the function, [MatchSubsequence](#). In addition, the following columns are added:

|                                                         |                                                                                                                  |
|---------------------------------------------------------|------------------------------------------------------------------------------------------------------------------|
| <code>snp_ref_start, snp_ref_end, snp_ref_length</code> | Location and Length of the best matching augmented subsequence on both the reference and SNP alleles.            |
| <code>ref_aug_match_seq_forward</code>                  | Best matching augmented subsequence or its corresponding sequence to the forward strand on the reference allele. |
| <code>snp_aug_match_seq_forward</code>                  | Best matching augmented subsequence or its corresponding sequence to the forward strand on the SNP allele.       |
| <code>ref_aug_match_seq_reverse</code>                  | Best matching augmented subsequence or its corresponding sequence to the reverse strand on the reference allele. |
| <code>snp_aug_match_seq_reverse</code>                  | Best matching augmented subsequence or its corresponding sequence to the reverse strand on the SNP allele.       |
| <code>ref_location</code>                               | SNP location of the best matching augmented subsequence on the reference allele.                                 |
| <code>snp_location</code>                               | SNP location of the best matching augmented subsequence on the SNP allele.                                       |

|                     |                                                                                |
|---------------------|--------------------------------------------------------------------------------|
| ref_extra_pwm_left  | Left extra unmatched position on the best matching augmented subsequence of t  |
| ref_extra_pwm_right | Right extra unmatched position on the best matching augmented subsequence of t |
| snp_extra_pwm_left  | Left extra unmatched position on the best matching augmented subsequence of t  |
| snp_extra_pwm_right | Right extra unmatched position on the best matching augmented subsequence of t |

**Author(s)**

Sunyoung Shin<sunyoung.shin@utdallas.edu>

**Examples**

```
data(example)
dtMotifMatch(motif_scores$snps.tbl, motif_scores$motif.scores,
motif.lib = motif_library)
```

---

|              |                                                                                                                                                                                  |
|--------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| encode_motif | <i>A motif library containing 2065 motifs downloaded from<br/><a href="http://compbio.mit.edu/encode-motifs/motifs.txt">http://compbio.mit.edu/encode-motifs/motifs.txt</a>.</i> |
|--------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|

---

**Description**

This motif library can be loaded by 'data(encode\_library)'.

**Format**

A list object.

**Author(s)**

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

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|                  |                                                                                                                                                                                 |
|------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| encode_motifinfo | <i>The information for the motif library downloaded from<br/><a href="http://compbio.mit.edu/encode-motifs/motifs.txt">http://compbio.mit.edu/encode-motifs/motifs.txt</a>.</i> |
|------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|

---

**Description**

This is a character vector that be loaded by 'data(encode\_library)'. The names of this vector are the same as the names for [encode\\_motif](#). The entries of this vector are the corresponding motif information parsed from the raw file.

**Format**

A character vector.

**Author(s)**

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

---

|                  |                                           |
|------------------|-------------------------------------------|
| GetIUPACSequence | <i>Get the IUPAC sequence of a motif.</i> |
|------------------|-------------------------------------------|

---

### Description

Convert the position weight matrix of a motif to the IUPAC sequence.

### Usage

```
GetIUPACSequence(pwm, prob = 0.25)
```

### Arguments

|      |                                                                       |
|------|-----------------------------------------------------------------------|
| pwm  | The position weight matrix, with the columns representing A, C, G, T. |
| prob | The probability threshold. Default: 0.25.                             |

### Value

A character string.

### Author(s)

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

### Examples

```
data(example)
GetIUPACSequence(motif_library[[1]], prob = 0.2)
```

---

|              |                                                                                                                                                                                                                                               |
|--------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| jaspar_motif | <i>A motif library containing 593 motifs downloaded from <a href="http://jaspar.genereg.net/html/DOWNLOAD/JASPAR_CORE/pfm/nonredundant/pfm_all.txt">http://jaspar.genereg.net/html/DOWNLOAD/JASPAR_CORE/pfm/nonredundant/pfm_all.txt</a>.</i> |
|--------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|

---

### Description

This motif library can be loaded by 'data(jaspar\_library)'.

### Format

A list object.

### Author(s)

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

---

|                  |                                                                                                                                                                                                                                               |
|------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| jaspar_motifinfo | <i>The information for the motif library downloaded from <a href="http://jaspar.genereg.net/html/DOWNLOAD/JASPAR_CORE/pfm/nonredundant/pfm_all.txt">http://jaspar.genereg.net/html/DOWNLOAD/JASPAR_CORE/pfm/nonredundant/pfm_all.txt</a>.</i> |
|------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|

---

### Description

This is a character vector that be loaded by 'data(jaspar\_library)'. The names of this vector are the same as the names for [jaspar\\_motif](#). The entries of this vector are the corresponding motif information parsed from the raw file.

### Format

A character vector.

### Author(s)

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

---

|               |                                            |
|---------------|--------------------------------------------|
| LoadFastaData | <i>Load the SNP data from fasta files.</i> |
|---------------|--------------------------------------------|

---

### Description

Load SNP data.

### Usage

```
LoadFastaData(ref.filename = NULL, snp.filename = NULL,
              ref.urlname = NULL, snp.urlname = NULL, snpids = NULL,
              default.par = FALSE)
```

### Arguments

|              |                                                                            |
|--------------|----------------------------------------------------------------------------|
| ref.filename | a fastq file name for the reference allele sequences.                      |
| snp.filename | a fastq file name for the SNP allele sequences.                            |
| ref.urlname  | URL of a fastq file for the reference allele sequences.                    |
| snp.urlname  | URL of a fastq file for the SNP allele sequences.                          |
| snpids       | SNP IDs                                                                    |
| default.par  | A boolean for whether using the default Markov parameters. Default: FALSE. |

**Value**

A list object containing the following components:

|                 |                                                                                    |
|-----------------|------------------------------------------------------------------------------------|
| sequence_matrix | A list of integer vectors representing the deoxyribose sequence around each SNP.   |
| a1              | An integer vector for the deoxyribose at the SNP location on the reference genome. |
| a2              | An integer vector for the deoxyribose at the SNP location on the SNP genome.       |

The results are coded as: "A"-1, "C"-2, "G"-3, "T"-4.

**Author(s)**

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

**Examples**

```
LoadFastaData(
  ref.urlname="http://pages.stat.wisc.edu/~keles/atSNP-Data/sample_1.fasta",
  snp.urlname="http://pages.stat.wisc.edu/~keles/atSNP-Data/sample_2.fasta")
```

---

|                  |                                       |
|------------------|---------------------------------------|
| LoadMotifLibrary | <i>Load position weight matrices.</i> |
|------------------|---------------------------------------|

---

**Description**

Load the file for position weight matrices for motifs.

**Usage**

```
LoadMotifLibrary(filename = NULL, urlname = NULL, tag = "MOTIF",
  transpose = FALSE, field = 2, sep = c("\t", " "), skipcols = 0,
  skiprows = 2, pseudocount = 0)
```

**Arguments**

|             |                                                                                                                                                                  |
|-------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| filename    | a MEME format file name.                                                                                                                                         |
| urlname     | URL containing a MEME format file.                                                                                                                               |
| tag         | A string that marks the description line of the position weight matrix.                                                                                          |
| transpose   | If TRUE (default), then the position weight matrix should have 4 columns. Otherwise, it should have 4 rows.                                                      |
| field       | The index of the field in the description line, separated by space, that indicates the motif name.                                                               |
| sep         | A vector of chars for the string separators to parse each lines of the matrix. Default: c(" ", "\t").                                                            |
| skipcols    | Number of columns to be skipped in the position weight matrix.                                                                                                   |
| skiprows    | Number of description lines before each position weight matrix.                                                                                                  |
| pseudocount | An integer for the pseudocount added to each of the original matrices. Default: 0. Recommended to be 1 if the original matrices are position frequency matrices. |

## Details

This function reads the formatted file containing motif information and convert them into a list of position weight matrices. The list of arguments should provide enough flexibility of importing a varying number of formats. Some examples are the following: For MEME format, the suggested arguments are: tag = 'Motif', skiprows = 2, skipcols = 0, transpose = FALSE, field = 2, sep = ' '; For motif files from JOHNSON lab (i.e. [http://johnsonlab.ucsf.edu/mochi\\_files/JASPAR\\_motifs\\_H\\_sapiens.txt](http://johnsonlab.ucsf.edu/mochi_files/JASPAR_motifs_H_sapiens.txt)), the suggested arguments are: tag = '/NAME', skiprows = 1, skipcols = 0, transpose = FALSE, field = 2, sep = "\t"; For JASPAR pfm matrices (i.e. [http://jaspar.genereg.net/download/CORE/JASPAR\\_2018\\_CORE\\_vertbrates\\_non-redundant\\_pfms\\_jaspar.txt](http://jaspar.genereg.net/download/CORE/JASPAR_2018_CORE_vertbrates_non-redundant_pfms_jaspar.txt)), the suggested arguments are: tag = ">", skiprows = 1, skipcols = 0, transpose = TRUE, field = 1, sep = "\t"; For the TRANSFAC library provided by UCF bioinformatics groups (i.e. <http://gibbs.biomed.ucf.edu/PreDREM/download/nonredundantmotif.transfac>), the suggested arguments are: tag = "DE", skiprows = 1, skipcols = 1, transpose = FALSE, field = 2, sep = "\t".

## Value

A list object of position weight matrices.

## Author(s)

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## Examples

```
pwms <- LoadMotifLibrary(
  urlname="http://pages.stat.wisc.edu/~keles/atSNP-Data/pfm_vertbrates.txt",
  tag = ">", transpose = FALSE, field = 1, sep = c("\t", " ", ">"),
  skipcols = 1, skiprows = 1, pseudocount = 1)
```

---

|             |                                                                                         |
|-------------|-----------------------------------------------------------------------------------------|
| LoadSNPData | <i>Load the SNP information and code the genome sequences around the SNP locations.</i> |
|-------------|-----------------------------------------------------------------------------------------|

---

## Description

Load the SNP data.

## Usage

```
LoadSNPData(filename = NULL,
  genome.lib = "BSgenome.Hsapiens.UCSC.hg38",
  snp.lib = "SNPlocs.Hsapiens.dbSNP144.GRCh38", snpids = NULL,
  half.window.size = 30, default.par = FALSE, mutation = FALSE, ...)
```

## Arguments

|                  |                                                                                                                                                                                                                                                                                                                                                                                                                                                               |     |             |     |                                     |       |                        |    |                                 |    |                                       |
|------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----|-------------|-----|-------------------------------------|-------|------------------------|----|---------------------------------|----|---------------------------------------|
| filename         | A table containing the SNP information. Must contain at least five columns with exactly the following names: <table data-bbox="568 399 1055 567"> <tr><td>chr</td><td>chromosome.</td></tr> <tr><td>snp</td><td>The nucleotide position of the SNP.</td></tr> <tr><td>snpid</td><td>The names of the SNPs.</td></tr> <tr><td>a1</td><td>The deoxyribose for one allele.</td></tr> <tr><td>a2</td><td>The deoxyribose for the other allele.</td></tr> </table> | chr | chromosome. | snp | The nucleotide position of the SNP. | snpid | The names of the SNPs. | a1 | The deoxyribose for one allele. | a2 | The deoxyribose for the other allele. |
| chr              | chromosome.                                                                                                                                                                                                                                                                                                                                                                                                                                                   |     |             |     |                                     |       |                        |    |                                 |    |                                       |
| snp              | The nucleotide position of the SNP.                                                                                                                                                                                                                                                                                                                                                                                                                           |     |             |     |                                     |       |                        |    |                                 |    |                                       |
| snpid            | The names of the SNPs.                                                                                                                                                                                                                                                                                                                                                                                                                                        |     |             |     |                                     |       |                        |    |                                 |    |                                       |
| a1               | The deoxyribose for one allele.                                                                                                                                                                                                                                                                                                                                                                                                                               |     |             |     |                                     |       |                        |    |                                 |    |                                       |
| a2               | The deoxyribose for the other allele.                                                                                                                                                                                                                                                                                                                                                                                                                         |     |             |     |                                     |       |                        |    |                                 |    |                                       |
|                  | If this file exists already, it is used to extract the SNP information. Otherwise, SNP information extracted using argument 'snpids' is outputted to this file.                                                                                                                                                                                                                                                                                               |     |             |     |                                     |       |                        |    |                                 |    |                                       |
| genome.lib       | A string of the library name for the genome version. Default: "BSgenome.Hsapiens.UCSC.hg38".                                                                                                                                                                                                                                                                                                                                                                  |     |             |     |                                     |       |                        |    |                                 |    |                                       |
| snp.lib          | A string of the library name to obtain the SNP information based on rs ids. Default: "SNPlocs.Hsapiens.dbSNP144.GRCh38".                                                                                                                                                                                                                                                                                                                                      |     |             |     |                                     |       |                        |    |                                 |    |                                       |
| snpids           | A vector of rs ids for the SNPs. This argument is overridden if the file with name filename exists.                                                                                                                                                                                                                                                                                                                                                           |     |             |     |                                     |       |                        |    |                                 |    |                                       |
| half.window.size | An integer for the half window size around the SNP within which the motifs are matched. Default: 30.                                                                                                                                                                                                                                                                                                                                                          |     |             |     |                                     |       |                        |    |                                 |    |                                       |
| default.par      | A boolean for whether using the default Markov parameters. Default: FALSE.                                                                                                                                                                                                                                                                                                                                                                                    |     |             |     |                                     |       |                        |    |                                 |    |                                       |
| mutation         | A boolean for whether this is mutation data. See details for more information. Default: FALSE.                                                                                                                                                                                                                                                                                                                                                                |     |             |     |                                     |       |                        |    |                                 |    |                                       |
| ...              | Other parameters passed to <a href="#">read.table</a> .                                                                                                                                                                                                                                                                                                                                                                                                       |     |             |     |                                     |       |                        |    |                                 |    |                                       |

## Details

This function extracts the nucleotide sequence within a window around each SNP and code them using 1-A, 2-C, 3-G, 4-T.

There are two ways of obtaining the nucleotide sequences. If filename is not NULL and the file exists, it should contain the positions and alleles for each SNP. Based on such information, the sequences around SNP positions are extracted using the Bioconductor annotation package specified by genome.lib. Users should make sure that this annotation package corresponds to the correct species and genome version of the actual data. Alternatively, users can also provide a vector of rs ids via the argument snpids. The SNP locations and allele information is then obtained via the Bioconductor annotation package specified by snp.lib, and passed on to the package specified by genome.lib to further obtain the nucleotide sequences.

If mutation=FALSE (default), this function assumes that the data is for SNP analysis, and the reference genome should be consistent with either the a1 or a2 nucleotide. When extracting the genome sequence around each SNP position, this function compares the nucleotide at the SNP location on the reference genome with both a1 and a2 to distinguish between the reference allele and the SNP allele. If the nucleotide extracted from the reference genome does not match either a1 or a2, the SNP is discarded. The discarded SNPs are in the 'rsid.rm' field in the output.

Alternatively, if mutation=TRUE, this function assumes that the data is for general single nucleotide mutation analysis. After extracting the genome sequence around each SNP position, it replaces

the nucleotide at the SNP location by the a1 nucleotide as the 'reference' allele sequence, and by the a2 nucleotide as the 'snp' allele sequence. It does NOT discard the sequence even if neither a1 or a2 matches the reference genome. When this data set is used in other functions, such as [ComputeMotifScore](#), [ComputePValues](#), all the results (i.e. affinity scores and their p-values) for the reference allele are indeed for the a1 allele, and results for the SNP allele are indeed for the a2 allele.

If the input is a list of rsid's, the SNP information extracted from `snp.lib` may contain more than two alleles for a single location. For such cases, [LoadSNPData](#) first extracts all pairs of alleles associated with those locations. If `'mutation=TRUE'`, all those pairs are considered as pairs of reference and SNP alleles, and their information is contained in `'sequence_matrix'`, `'a1'`, `'a2'` and `'snpid'`. If `'mutation=FALSE'`, [LoadSNPData](#) further filters these pairs based on whether one allele matches to the reference genome nucleotide extracted from `genome.lib`. Only those pairs with one allele matching the reference genome nucleotide is considered as pairs of reference and SNP alleles, with their information contained in `'sequence_matrix'`, `'a1'`, `'a2'` and `'snpid'`.

## Value

A list object containing the following components:

|                              |                                                                                                                                                                                               |
|------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <code>sequence_matrix</code> | A list of integer vectors representing the deoxyribose sequence around each SNP.                                                                                                              |
| <code>a1</code>              | An integer vector for the deoxyribose at the SNP location on the reference genome.                                                                                                            |
| <code>a2</code>              | An integer vector for the deoxyribose at the SNP location on the SNP genome.                                                                                                                  |
| <code>snpid</code>           | A string vector for the SNP rsids.                                                                                                                                                            |
| <code>rsid.missing</code>    | If the data source is a list of rsids, this field records rsids for SNPs that are discarded because they are not in the reference genome.                                                     |
| <code>rsid.duplicate</code>  | If the data source is a list of rsids, this field records rsids for SNPs that based on the SNPlocs package, this field records rsids for SNPs that are discarded because they are duplicated. |
| <code>rsid.na</code>         | This field records rsids for SNPs that are discarded because the nucleotide sequences contain none ACGT.                                                                                      |
| <code>rsid.rm</code>         | If the data source is a table and <code>mutation=FALSE</code> , this field records rsids for SNPs that are discarded because they are not in the reference genome.                            |

The results are coded as: "A"-1, "C"-2, "G"-3, "T"-4.

## Author(s)

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## Examples

```
## Not run: LoadSNPData(snpids = c("rs53576", "rs7412"),
genome.lib = "BSgenome.Hsapiens.UCSC.hg38", snp.lib =
"SNPlocs.Hsapiens.dbSNP144.GRCh38", half.window.size = 30, default.par = TRUE
, mutation = FALSE)
## End(Not run)
```

---

|                  |                                          |
|------------------|------------------------------------------|
| MatchSubsequence | <i>Compute the matching subsequence.</i> |
|------------------|------------------------------------------|

---

### Description

This function combines the SNP set, the motif library and the affinity score table and produce the matching subsequence found at each SNP location for each motif.

### Usage

```
MatchSubsequence(snp.tbl, motif.scores, motif.lib, snpids = NULL,
                 motifs = NULL, ncores = 1)
```

### Arguments

`snp.tbl` A data.frame with the following information:

|                          |                                                             |
|--------------------------|-------------------------------------------------------------|
| <code>snpid</code>       | SNP id.                                                     |
| <code>ref_seq</code>     | Reference allele nucleotide sequence.                       |
| <code>snp_seq</code>     | SNP allele nucleotide sequence.                             |
| <code>ref_seq_rev</code> | Reference allele nucleotide sequence on the reverse strand. |
| <code>snp_seq_rev</code> | SNP allele nucleotide sequence on the reverse strand.       |

`motif.scores` A data.frame with the following information:

|                                             |                                                                                                                    |
|---------------------------------------------|--------------------------------------------------------------------------------------------------------------------|
| <code>motif</code>                          | Name of the motif.                                                                                                 |
| <code>motif_len</code>                      | Length of the motif.                                                                                               |
| <code>ref_start, ref_end, ref_strand</code> | Location of the best matching subsequence on the reference allele.                                                 |
| <code>snp_start, snp_end, snp_strand</code> | Location of the best matching subsequence on the SNP allele.                                                       |
| <code>log_lik_ref</code>                    | Log-likelihood score for the reference allele.                                                                     |
| <code>log_lik_snp</code>                    | Log-likelihood score for the SNP allele.                                                                           |
| <code>log_lik_ratio</code>                  | The log-likelihood ratio.                                                                                          |
| <code>log_enhance_odds</code>               | Difference in log-likelihood ratio between SNP allele and reference allele based on the best matching subsequence. |
| <code>log_reduce_odds</code>                | Difference in log-likelihood ratio between reference allele and SNP allele based on the best matching subsequence. |

`motif.lib` A list of the position weight matrices for the motifs.

`snpids` A subset of snpids to compute the subsequences. Default: NULL, when all snps are computed.

`motifs` A subset of motifs to compute the subsequences. Default: NULL, when all motifs are computed.

`ncores` The number of cores used for parallel computing.

**Value**

A data.frame containing all columns in both `snp.tbl` and `motif.scores`. In addition, the following columns are added:

|                                |                                                                                                    |
|--------------------------------|----------------------------------------------------------------------------------------------------|
| <code>ref_match_seq</code>     | Best matching subsequence on the reference allele.                                                 |
| <code>snp_match_seq</code>     | Best matching subsequence on the SNP allele.                                                       |
| <code>ref_seq_snp_match</code> | Subsequence on the reference allele corresponding to the best matching location on the SNP allele. |
| <code>snp_seq_ref_match</code> | Subsequence on the SNP allele corresponding to the best matching location on the reference allele. |

**Author(s)**

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

**Examples**

```
data(example)
MatchSubsequence(motif_scores$snp.tbl, motif_scores$motif.scores,
motif_library, ncores=2)
```

---

|                            |                                |
|----------------------------|--------------------------------|
| <code>motif_library</code> | <i>A sample motif library.</i> |
|----------------------------|--------------------------------|

---

**Description**

A list of the position weight matrices corresponding to motifs, loaded by `'data(example)'`.

**Format**

A list object.

**Author(s)**

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

---

|             |                                                                                                                                                            |
|-------------|------------------------------------------------------------------------------------------------------------------------------------------------------------|
| motif_match | <i>Composit logo plotting input containing motif scores, the matching subsequences and the augmented matching subsequences on SNP and reference allele</i> |
|-------------|------------------------------------------------------------------------------------------------------------------------------------------------------------|

---

**Description**

This data.frame object loaded by 'data(example)' contains information about MYC\_disc1 match to rs53576.

**Format**

A data.frame object.

**Author(s)**

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

---

|              |                                                                         |
|--------------|-------------------------------------------------------------------------|
| motif_scores | <i>Scores for the sample snp data computed based on the motif data.</i> |
|--------------|-------------------------------------------------------------------------|

---

**Description**

This list object loaded by 'data(example)' contains two fields:

snp.tbl        A data.frame containing the sequence of nucleobases around each SNP.  
motif.scores   A data.frame containing the likelihood scores computed for each SNP and each motif.

**Format**

A data.frame object.

**Author(s)**

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

---

|                |                                                                                                                                                                               |
|----------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| plotMotifMatch | <i>Plot sequence logos of the position weight matrix of the motif and sequences of its corresponding best matching augmented subsequence on the reference and SNP allele.</i> |
|----------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|

---

## Description

Plot the best matching augmented subsequences on the reference and SNP alleles. Plot sequence logos of the position weight matrix of the motif to the corresponding positions of the best matching subsequences on the references and SNP alleles.

## Usage

```
plotMotifMatch(motif.match, motif.lib, cex.main = 2, ...)
```

## Arguments

|             |                                                          |
|-------------|----------------------------------------------------------|
| motif.match | a single row of dtMotifMatch output in data.frame format |
| motif.lib   | A list of position weight matrices                       |
| cex.main    | The size of the main title.                              |
| ...         | Other parameters passed to plotMotifLogo.                |

## Value

Sequence logo stacks: Reference subsequences, sequence logo of reference allele matching position weight matrix, SNP subsequences, sequence logo of SNP allele matching position weight matrix

## Author(s)

Sunyoung Shin<sunyoung.shin@utdallas.edu>

## Examples

```
data(example)
plotMotifMatch(motif_match, motif.lib = motif_library)
```

---

|       |                                                                                          |
|-------|------------------------------------------------------------------------------------------|
| prior | <i>Default stationary distribution for nucleotide sequences in the reference genome.</i> |
|-------|------------------------------------------------------------------------------------------|

---

### Description

This parameter is fitted using 61bp windowns around the SNPs in the NHGRI catalog. Loaded by 'data(default\_par)'.

### Format

A numeric vector.

### Author(s)

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

---

|         |                                        |
|---------|----------------------------------------|
| snpInfo | <i>A data set for SNP information.</i> |
|---------|----------------------------------------|

---

### Description

This list object loaded by 'data(example)' contains three fields :

|                 |                                                                                                         |
|-----------------|---------------------------------------------------------------------------------------------------------|
| sequence_matrix | A sequence matrix, coded by 1-A, 2-C, 3-G, 4-T, with each column corresponding to a subsequence of 61 b |
| transition      | The transition matrix used in Markov model.                                                             |
| prior           | The stationary distribution used in the Markov model.                                                   |

### Format

A list object.

### Author(s)

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

---

|         |                                          |
|---------|------------------------------------------|
| snp_tbl | <i>A data frame for SNP information.</i> |
|---------|------------------------------------------|

---

**Description**

This data frame is loaded by 'data(example)'. It is a table including the following columns:

|       |                                                 |
|-------|-------------------------------------------------|
| chr   | The chromosome.                                 |
| snp   | The SNP location coordinate.                    |
| snpid | The SNP label.                                  |
| a1,a2 | The nucleotide on the reference and SNP allele. |

**Format**

A data.frame object.

**Author(s)**

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

---

|            |                                                                                                |
|------------|------------------------------------------------------------------------------------------------|
| transition | <i>Default transition probability matrix for nucleotide sequences in the reference genome.</i> |
|------------|------------------------------------------------------------------------------------------------|

---

**Description**

This parameter is fitted using 61bp windows around the SNPs in the NHGRI catalog. Loaded by 'data(default\_par)'.

**Format**

A 4 by 4 numeric matrix.

**Author(s)**

Sunyoung Shin <sunyoung.shin@utdallas.edu>, Chandler Zuo <chandler.c.zuo@gmail.com>

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