# Package: zalpha (via r-universe)

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Type Package

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create\_LDprofile

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## **Description**

An LD (linkage disequilibrium) profile is a look-up table containing the expected correlation between SNPs given the genetic distance between them. The use of an LD profile can increase the accuracy of results by taking into account the expected correlation between SNPs. This function aids the user in creating their own LD profile.

#### Usage

```
create_LDprofile(dist, x, bin_size, max_dist = NULL, beta_params = FALSE)
```

## Arguments

dist	A numeric vector, or a list of numeric vectors, containing the genetic distance for each SNP.
х	A matrix of SNP values, or a list of matrices. Columns represent chromosomes; rows are SNP locations. Hence, the number of rows should equal the length of the dist vector. SNPs should all be biallelic.
bin_size	The size of each bin, in the same units as dist.
max_dist	Optional. The maximum genetic distance to be considered. If this is not supplied, it will default to the maximum distance in the dist vector.
beta_params	Optional. Beta parameters are calculated if this is set to TRUE. Default is FALSE.

## **Details**

The input for dist and x can be lists. This allows multiple datasets to be used in the creation of the LD profile. For example, using all 22 autosomes from the human genome would involve 22 different distance vectors and SNP matrices. Both lists should be the same length and should correspond exactly to each other (i.e. the distances in each element of dist should go with the SNPs in the same element of x)

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In the output, bins represent lower bounds. The first bin contains pairs where the genetic distance is greater than or equal to 0 and less than bin\_size. The final bin contains pairs where the genetic distance is greater than or equal to max\_dist-bin\_size and less than max\_dist. If the max\_dist is not an increment of bin\_size, it will be adjusted to the next highest increment. The final bin will be the bin that max\_dist falls into. For example, if the max\_dist is given as 4.5 and the bin\_size is 1, the final bin will be 4. max\_dist should be big enough to cover the genetic distances between pairs of SNPs within the window size given when the  $Z_{\alpha}$  statistics are run. Any pairs with genetic distances bigger than max\_dist will be assigned the values in the maximum bin of the LD profile.

By default, Beta parameters are not calculated. To fit a Beta distribution to the expected correlations, needed for the Zalpha\_BetaCDF and Zbeta\_BetaCDF statistics, beta\_params should be set to TRUE and the package 'fitdistrplus' must be installed.

Ideally, an LD profile would be generated using data from a null population with no selection, For example by using a simulation if the other population parameters are known. However, often these are unknown or complex, so generating an LD profile using the same data as is being analysed is acceptable, as long as the bins are large enough.

#### Value

A data frame containing an LD profile that can be used by other statistics in this package.

#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

## See Also

Zalpha\_expected, Zalpha\_rsq\_over\_expected, Zalpha\_log\_rsq\_over\_expected, Zalpha\_Zscore, Zalpha\_BetaCDF, Zbeta\_expected, Zbeta\_rsq\_over\_expected, Zbeta\_log\_rsq\_over\_expected, Zbeta\_Zscore, Zbeta\_BetaCDF, Zalpha\_all.

#### **Examples**

```
## load the snps example dataset
data(snps)
## Create an LD profile using this data
create_LDprofile(snps$cM_distances,as.matrix(snps[,3:12]),0.001)
## To get the Beta distribution parameter estimates, the fitdistrplus package is required
if (requireNamespace("fitdistrplus", quietly = TRUE)==TRUE) {
    create_LDprofile(snps$cM_distances,as.matrix(snps[,3:12]),0.001,beta_params=TRUE)
}
```

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LDprofile

Dataset containing an example LD profile

## **Description**

A simulated LD profile, containing example LD statistics for genetic distances of 0 to 0.0049, in bins of size 0.0001.

#### Usage

```
data(LDprofile)
```

#### **Format**

A data frame with 50 rows and 5 variables:

bin the lower bound of each bin

 $\mathbf{rsq}$  the expected  $r^2$  value for a pair of SNPs, where the genetic distance between them falls in the given bin

sd the standard deviation of the expected  $r^2$  value

Beta\_a the first shape parameter for the Beta distribution fitted for this bin

Beta\_b the second shape parameter for the Beta distribution fitted for this bin

LR

Runs the LR function

## Description

Returns the |L||R| value for each SNP location supplied to the function, where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws. For more information about the |L||R| diversity statistic, please see Jacobs (2016).

## Usage

```
LR(pos, ws, X = NULL)
```

## Arguments

pos	A numeric vector of SNP locations
WS	The window size which the LR statistic will be calculated over. This should be on the same scale as the pos vector.
X	Optional. Specify a region of the chromosome to calculate LR for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate LR for every SNP in the pos vector.

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## Value

A list containing the SNP positions and the LR values for those SNPs

#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

## **Examples**

```
## load the snps example dataset
data(snps)
## run LR over all the SNPs with a window size of 3000 bp
LR(snps$bp_positions,3000)
## only return results for SNPs between locations 600 and 1500 bp
LR(snps$bp_positions,3000,X=c(600,1500))
```

L\_plus\_R

Runs the L\_plus\_R function

## Description

Returns the  $\binom{|L|}{2} + \binom{|R|}{2}$  value for each SNP location supplied to the function. |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws. For more information about the L\_plus\_R diversity statistic, please see Jacobs (2016).

## Usage

```
L_plus_R(pos, ws, X = NULL)
```

## **Arguments**

pos	A numeric vector of SNP locations
WS	The window size which the L_plus_R statistic will be calculated over. This should be on the same scale as the pos vector.
X	Optional. Specify a region of the chromosome to calculate L_plus_R for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate L_plus_R for every SNP in the pos vector.

#### Value

A list containing the SNP positions and the L\_plus\_R values for those SNPs

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#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

## **Examples**

```
## load the snps example dataset
data(snps)
## run L_plus_R over all the SNPs with a window size of 3000 bp
L_plus_R(snps$bp_positions,3000)
## only return results for SNPs between locations 600 and 1500 bp
L_plus_R(snps$bp_positions,3000,X=c(600,1500))
```

snps

Dataset containing details on simulated SNPs

## **Description**

A dataset containing the positions, genetic distances and alleles for 20 SNPs, across 10 simulated chromosomes.

## Usage

snps

#### **Format**

A data frame with 20 rows and 12 variables:

bp\_positions location of the SNP on the chromosome e.g. in base pairs

cM\_distances genetic distance of the SNP from the start of the chromosome e.g. in centimorgans

chrom\_1 allele of the SNP on the first example chromosome

chrom\_2 allele of the SNP on the second example chromosome

**chrom\_3** allele of the SNP on the third example chromosome

chrom\_4 allele of the SNP on the fourth example chromosome

**chrom** 5 allele of the SNP on the fifth example chromosome

chrom\_6 allele of the SNP on the sixth example chromosome

chrom\_7 allele of the SNP on the seventh example chromosome

chrom\_8 allele of the SNP on the eighth example chromosome

chrom\_9 allele of the SNP on the ninth example chromosome

**chrom\_10** allele of the SNP on the tenth example chromosome

## **Examples**

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Zalpha	Runs the Zalpha function
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## Description

Returns a  $Z_{\alpha}$  value for each SNP location supplied to the function. For more information about the  $Z_{\alpha}$  statistic, please see Jacobs (2016). The  $Z_{\alpha}$  statistic is defined as:

$$Z_{\alpha} = \frac{{\binom{|L|}{2}}^{-1} \sum_{i,j \in L} r_{i,j}^2 + {\binom{|R|}{2}}^{-1} \sum_{i,j \in L} r_{i,j}^2}{2}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws, and  $r^2$  is equal to the squared correlation between a pair of SNPs

## Usage

Zalpha(pos, ws, x, minRandL = 4, minRL = 25, 
$$X = NULL$$
)

## **Arguments**

pos	A numeric vector of SNP locations
WS	The window size which the $Z_{\alpha}$ statistic will be calculated over. This should be on the same scale as the pos vector.
х	A matrix of SNP values. Columns represent chromosomes; rows are SNP locations. Hence, the number of rows should equal the length of the pos vector. SNPs should all be biallelic.
minRandL	Minimum number of SNPs in each set $R$ and $L$ for the statistic to be calculated. Default is 4.
minRL	Minimum value for the product of the set sizes for R and L. Default is 25.
X	Optional. Specify a region of the chromosome to calculate $Z_{\alpha}$ for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate $Z_{\alpha}$ for every SNP in the pos vector.

## Value

A list containing the SNP positions and the  $Z_{\alpha}$  values for those SNPs

#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

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## **Examples**

```
## load the snps example dataset
data(snps)
## run Zalpha over all the SNPs with a window size of 3000 bp
Zalpha(snps$bp_positions,3000,as.matrix(snps[,3:12]))
## only return results for SNPs between locations 600 and 1500 bp
Zalpha(snps$bp_positions,3000,as.matrix(snps[,3:12]),X=c(600,1500))
```

Zalpha\_all

Runs all the statistics in the zalpha package

#### **Description**

Returns every statistic for each SNP location, given the appropriate parameters. See Details for more information.

## Usage

```
Zalpha_all(
  pos,
  ws,
  x = NULL,
  dist = NULL,
  LDprofile_bins = NULL,
  LDprofile_rsq = NULL,
  LDprofile_sd = NULL,
  LDprofile_Beta_a = NULL,
  LDprofile_Beta_b = NULL,
  minRandL = 4,
  minRL = 25,
  X = NULL
)
```

## **Arguments**

pos	A numeric vector of SNP locations
WS	The window size which the statistics will be calculated over. This should be on the same scale as the pos vector.
X	Optional. A matrix of SNP values. Columns represent chromosomes; rows are SNP locations. Hence, the number of rows should equal the length of the pos vector. SNPs should all be biallelic.
dist	Optional. A numeric vector of genetic distances (e.g. $cM$ , $LDU$ ). This should be the same length as pos.
LDprofile_bins	Optional. A numeric vector containing the lower bound of the bins used in the LD profile. These should be of equal size.

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LDprofile\_rsq Optional. A numeric vector containing the expected  $r^2$  values for the corresponding bin in the LD profile. Must be between 0 and 1.

LDprofile\_sd Optional. A numeric vector containing the standard deviation of the  $r^2$  values for the corresponding bin in the LD profile.

LDprofile\_Beta\_a

Optional. A numeric vector containing the first estimated Beta parameter for the corresponding bin in the LD profile.

LDprofile\_Beta\_b

Optional. A numeric vector containing the second estimated Beta parameter for the corresponding bin in the LD profile.

minRandL Minimum number of SNPs in each set R and L for the statistics to be calculated.

L is the set of SNPs to the left of the target SNP and R to the right, within the

If not supplied, the function will calculate the statistics for every SNP in the pos

given window size ws. Default is 4.

minRL Minimum value for the product of the set sizes for R and L. Default is 25.

Optional. Specify a region of the chromosome to calculate the statistics for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector.

vector.

#### **Details**

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Not all statistics will be returned, depending on the parameters supplied to the function. If x is not supplied, only Zalpha\_expected, Zbeta\_expected, LR and L\_plus\_R will be calculated. For any of the statistics which use an expected  $r^2$  value, the parameters dist, LDprofile\_bins and LDprofile\_rsq must be supplied. This includes the statistics: Zalpha\_expected, Zalpha\_rsq\_over\_expected, Zalpha\_log\_rsq\_over\_expected, Zalpha\_Zscore, Zalpha\_BetaCDF, Zbeta\_expected, Zbeta\_rsq\_over\_expected, Zbeta\_log\_rsq\_over\_expected, Zbeta\_Zscore and Zbeta\_BetaCDF.

- For Zalpha\_Zscore and Zbeta\_Zscore to be calculated, the parameter LDprofile\_sd must also be supplied.
- For Zalpha\_BetaCDF and Zbeta\_BetaCDF to be calculated, the parameters LDprofile\_Beta\_a and LDprofile\_Beta\_b must also be supplied.

The LD profile describes the expected correlation between SNPs at a given genetic distance, generated using simulations or real data. Care should be taken to utilise an LD profile that is representative of the population in question. The LD profile should consist of evenly sized bins of distances (for example 0.0001 cM per bin), where the value given is the (inclusive) lower bound of the bin. Ideally, an LD profile would be generated using data from a null population with no selection, however one can be generated using this data. See the create\_LDprofile function for more information on how to create an LD profile. For more information about the statistics, please see Jacobs (2016).

#### Value

A list containing the SNP positions and the statistics for those SNPs

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#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

#### See Also

Zalpha, Zalpha\_expected, Zalpha\_rsq\_over\_expected, Zalpha\_log\_rsq\_over\_expected, Zalpha\_Zscore, Zalpha\_BetaCDF, Zbeta, Zbeta\_expected, Zbeta\_rsq\_over\_expected, Zbeta\_log\_rsq\_over\_expected, Zbeta\_Zscore, Zbeta\_BetaCDF, LR, L\_plus\_R, create\_LDprofile.

## **Examples**

```
## load the snps and LDprofile example datasets
data(snps)
data(LDprofile)
## run Zalpha_all over all the SNPs with a window size of 3000 bp
## will return all 15 statistics
Zalpha_all(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq,LDprofile$sd,LDprofile$Beta_a,LDprofile$Beta_b)
## only return results for SNPs between locations 600 and 1500 bp
Zalpha_all(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq,LDprofile$sd,LDprofile$Beta_a,LDprofile$Beta_b,X=c(600,1500))
## will only return statistics not requiring an LD profile
Zalpha_all(snps$bp_positions,3000,as.matrix(snps[,3:12]))
```

Zalpha\_BetaCDF

Runs the Zalpha function using a cumulative beta distribution function on the r-squared values for the region

#### **Description**

Returns a  $Z_{\alpha}^{BetaCDF}$  value for each SNP location supplied to the function, based on the expected  $r^2$  values given an LD profile and genetic distances. For more information about the  $Z_{\alpha}^{BetaCDF}$  statistic, please see Jacobs (2016). The  $Z_{\alpha}^{BetaCDF}$  statistic is defined as:

$$Z_{\alpha}^{BetaCDF} = \frac{{{{{|L|}}}\choose{2}}^{-1} \sum_{i,j \in L} \frac{B(r_{i,j}^2;a,b)}{B(a,b)} + {{{|R|}}\choose{2}}^{-1} \sum_{i,j \in R} \frac{B(r_{i,j}^2;a,b)}{B(a,b)}}{2}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws,  $r^2$  is equal to the squared correlation between a pair of SNPs, and  $\frac{B(r_{i,j}^2;a,b)}{B(a,b)}$  is the cumulative distribution function for the Beta distribution given the estimated a and b parameters from the LD profile.

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#### Usage

```
Zalpha_BetaCDF(
  pos,
  ws,
  x,
  dist,
  LDprofile_bins,
  LDprofile_Beta_a,
  LDprofile_Beta_b,
  minRandL = 4,
  minRL = 25,
  X = NULL
)
```

#### **Arguments**

pos	A numeric vect	or of SNP locations

ws The window size which the  $Z_{\alpha}^{BetaCDF}$  statistic will be calculated over. This

should be on the same scale as the pos vector.

x A matrix of SNP values. Columns represent chromosomes; rows are SNP lo-

cations. Hence, the number of rows should equal the length of the pos vector.

SNPs should all be biallelic.

dist A numeric vector of genetic distances (e.g. cM, LDU). This should be the same

length as pos.

LDprofile\_bins A numeric vector containing the lower bound of the bins used in the LD profile.

These should be of equal size.

LDprofile\_Beta\_a

A numeric vector containing the first estimated Beta parameter for the corre-

sponding bin in the LD profile.

LDprofile\_Beta\_b

A numeric vector containing the second estimated Beta parameter for the corre-

sponding bin in the LD profile.

minRandL Minimum number of SNPs in each set R and L for the statistic to be calculated.

Default is 4.

minRL Minimum value for the product of the set sizes for R and L. Default is 25.

X Optional. Specify a region of the chromosome to calculate  $Z_{\alpha}^{BetaCDF}$  for in

the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate  $Z_{\alpha}^{BetaCDF}$  for every SNP in the pos

vector.

#### **Details**

The LD profile describes the expected correlation between SNPs at a given genetic distance, generated using simulations or real data. Care should be taken to utilise an LD profile that is representative of the population in question. The LD profile should consist of evenly sized bins of

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distances (for example 0.0001 cM per bin), where the value given is the (inclusive) lower bound of the bin. Ideally, an LD profile would be generated using data from a null population with no selection, however one can be generated using this data. See the create\_LDprofile function for more information on how to create an LD profile.

#### Value

A list containing the SNP positions and the  $Z_{\alpha}^{BetaCDF}$  values for those SNPs

#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

#### See Also

create\_LDprofile

## **Examples**

```
## load the snps and LDprofile example datasets
data(snps)
data(LDprofile)
## run Zalpha_BetaCDF over all the SNPs with a window size of 3000 bp
Zalpha_BetaCDF(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$Beta_a,LDprofile$Beta_b)
## only return results for SNPs between locations 600 and 1500 bp
Zalpha_BetaCDF(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$Beta_a,LDprofile$Beta_b,X=c(600,1500))
```

Zalpha\_expected

Runs the Zalpha function on the expected r-squared values for the region

## Description

Returns a  $Z_{\alpha}^{E[r^2]}$  value for each SNP location supplied to the function, based on the expected  $r^2$  values given an LD profile and genetic distances. For more information about the  $Z_{\alpha}^{E[r^2]}$  statistic, please see Jacobs (2016). The  $Z_{\alpha}^{E[r^2]}$  statistic is defined as:

$$Z_{\alpha}^{E[r^2]} = \frac{{{|L| \choose 2}}^{-1} \sum_{i,j \in L} E[r_{i,j}^2] + {{|R| \choose 2}}^{-1} \sum_{i,j \in R} E[r_{i,j}^2]}{2}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws, and  $E[r^2]$  is equal to the expected squared correlation between a pair of SNPs, given an LD profile.

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#### Usage

```
Zalpha_expected(
  pos,
  ws,
  dist,
  LDprofile_bins,
  LDprofile_rsq,
  minRandL = 4,
  minRL = 25,
  X = NULL
)
```

## **Arguments**

pos	A numeric vector of SNP locations
WS	The window size which the $Z_{\alpha}^{E[r^2]}$ statistic will be calculated over. This should be on the same scale as the pos vector.
dist	A numeric vector of genetic distances (e.g. cM, LDU). This should be the same length as pos.
LDprofile_bins	A numeric vector containing the lower bound of the bins used in the LD profile. These should be of equal size.
LDprofile_rsq	A numeric vector containing the expected $r^2$ values for the corresponding bin in the LD profile. Must be between $0$ and $1$ .
minRandL	$\label{eq:minimum number of SNPs in each set R and L for the statistic to be calculated.} \\ Default is 4.$
minRL	Minimum value for the product of the set sizes for R and L. Default is 25.
X	Optional. Specify a region of the chromosome to calculate $Z_{\alpha}^{E[r^2]}$ for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate $Z_{\alpha}^{E[r^2]}$ for every SNP in the pos vector.

#### **Details**

The LD profile describes the expected correlation between SNPs at a given genetic distance, generated using simulations or real data. Care should be taken to utilise an LD profile that is representative of the population in question. The LD profile should consist of evenly sized bins of distances (for example 0.0001 cM per bin), where the value given is the (inclusive) lower bound of the bin. Ideally, an LD profile would be generated using data from a null population with no selection, however one can be generated using this data. See the create\_LDprofile function for more information on how to create an LD profile.

## Value

A list containing the SNP positions and the  $Z^{E[r^2]}_{lpha}$  values for those SNPs

#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

#### See Also

```
create_LDprofile
```

## **Examples**

```
## load the snps and LDprofile example datasets
data(snps)
data(LDprofile)
## run Zalpha_expected over all the SNPs with a window size of 3000 bp
Zalpha_expected(snps$bp_positions,3000,snps$cM_distances,LDprofile$bin,LDprofile$rsq)
## only return results for SNPs between locations 600 and 1500 bp
Zalpha_expected(snps$bp_positions,3000,snps$cM_distances,LDprofile$bin,LDprofile$rsq,X=c(600,1500))
```

```
Zalpha_log_rsq_over_expected
```

Runs the Zalpha function on the log of the r-squared values over the expected r-squared values for the region

#### **Description**

Returns a  $Z_{\alpha}^{log_{10}(r^2/E[r^2])}$  value for each SNP location supplied to the function, based on the expected  $r^2$  values given an LD profile and genetic distances. For more information about the  $Z_{\alpha}^{log_{10}(r^2/E[r^2])}$  statistic, please see Jacobs (2016). The  $Z_{\alpha}^{log_{10}(r^2/E[r^2])}$  statistic is defined as:

$$Z_{\alpha}^{log_{10}(r^2/E[r^2])} = \frac{{\binom{|L|}{2}}^{-1} \sum_{i,j \in L} log_{10}(r_{i,j}^2/E[r_{i,j}^2]) + {\binom{|R|}{2}}^{-1} \sum_{i,j \in R} log_{10}(r_{i,j}^2/E[r_{i,j}^2])}{2}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws,  $r^2$  is equal to the squared correlation between a pair of SNPs, and  $E[r^2]$  is equal to the expected squared correlation between a pair of SNPs, given an LD profile.

## Usage

```
Zalpha_log_rsq_over_expected(
  pos,
  ws,
  x,
  dist,
  LDprofile_bins,
  LDprofile_rsq,
  minRandL = 4,
```

```
minRL = 25,
X = NULL
```

#### **Arguments**

pos	A numeric vector of SNP locations
WS	The window size which the $Z_{\alpha}^{log_{10}(r^2/E[r^2])}$ statistic will be calculated over. This should be on the same scale as the pos vector.
Х	A matrix of SNP values. Columns represent chromosomes; rows are SNP locations. Hence, the number of rows should equal the length of the pos vector. SNPs should all be biallelic.
dist	A numeric vector of genetic distances (e.g. $cM$ , $LDU$ ). This should be the same length as $pos$ .
LDprofile_bins	A numeric vector containing the lower bound of the bins used in the LD profile. These should be of equal size.
LDprofile_rsq	A numeric vector containing the expected $r^2$ values for the corresponding bin in the LD profile. Must be between 0 and 1.
minRandL	Minimum number of SNPs in each set R and L for the statistic to be calculated. Default is $4. $
minRL	Minimum value for the product of the set sizes for R and L. Default is 25.
X	Optional. Specify a region of the chromosome to calculate $Z_{\alpha}^{log_{10}(r^2/E[r^2])}$ for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate $Z_{\alpha}^{log_{10}(r^2/E[r^2])}$ for every SNP in the pos vector.

## **Details**

The LD profile describes the expected correlation between SNPs at a given genetic distance, generated using simulations or real data. Care should be taken to utilise an LD profile that is representative of the population in question. The LD profile should consist of evenly sized bins of distances (for example 0.0001 cM per bin), where the value given is the (inclusive) lower bound of the bin. Ideally, an LD profile would be generated using data from a null population with no selection, however one can be generated using this data. See the <code>create\_LDprofile</code> function for more information on how to create an LD profile.

## Value

A list containing the SNP positions and the  $Z_{\alpha}^{log_{10}(r^2/E[r^2])}$  values for those SNPs

#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

#### See Also

```
create_LDprofile
```

#### **Examples**

```
## load the snps and LDprofile example datasets
data(snps)
data(LDprofile)
## run Zalpha_log_rsq_over_expected over all the SNPs with a window size of 3000 bp
Zalpha_log_rsq_over_expected(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq)
## only return results for SNPs between locations 600 and 1500 bp
Zalpha_log_rsq_over_expected(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq,X=c(600,1500))
```

Zalpha\_rsq\_over\_expected

Runs the Zalpha function on the r-squared values over the expected r-squared values for the region

## Description

Returns a  $Z_{\alpha}^{r^2/E[r^2]}$  value for each SNP location supplied to the function, based on the expected  $r^2$  values given an LD profile and genetic distances. For more information about the  $Z_{\alpha}^{r^2/E[r^2]}$  statistic, please see Jacobs (2016). The  $Z_{\alpha}^{r^2/E[r^2]}$  statistic is defined as:

$$Z_{\alpha}^{r^2/E[r^2]} = \frac{{\binom{|L|}{2}}^{-1} \sum_{i,j \in L} r_{i,j}^2 / E[r_{i,j}^2] + {\binom{|R|}{2}}^{-1} \sum_{i,j \in R} r_{i,j}^2 / E[r_{i,j}^2]}{2}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws,  $r^2$  is equal to the squared correlation between a pair of SNPs, and  $E[r^2]$  is equal to the expected squared correlation between a pair of SNPs, given an LD profile.

## Usage

```
Zalpha_rsq_over_expected(
  pos,
  ws,
  x,
  dist,
  LDprofile_bins,
  LDprofile_rsq,
  minRandL = 4,
  minRL = 25,
  X = NULL
)
```

#### **Arguments**

A numeric vector of SNP locations
The window size which the $Z_{\alpha}^{r^2/E[r^2]}$ statistic will be calculated over. This should be on the same scale as the pos vector.
A matrix of SNP values. Columns represent chromosomes; rows are SNP locations. Hence, the number of rows should equal the length of the pos vector. SNPs should all be biallelic.
A numeric vector of genetic distances (e.g. cM, LDU). This should be the same length as pos.
A numeric vector containing the lower bound of the bins used in the LD profile. These should be of equal size.
A numeric vector containing the expected $r^2$ values for the corresponding bin in the LD profile. Must be between 0 and 1.
Minimum number of SNPs in each set $R$ and $L$ for the statistic to be calculated. Default is 4.
Minimum value for the product of the set sizes for R and L. Default is 25.
Optional. Specify a region of the chromosome to calculate $Z_{\alpha}^{r^2/E[r^2]}$ for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate $Z_{\alpha}^{r^2/E[r^2]}$ for every SNP in the pos vector.

#### **Details**

The LD profile describes the expected correlation between SNPs at a given genetic distance, generated using simulations or real data. Care should be taken to utilise an LD profile that is representative of the population in question. The LD profile should consist of evenly sized bins of distances (for example 0.0001 cM per bin), where the value given is the (inclusive) lower bound of the bin. Ideally, an LD profile would be generated using data from a null population with no selection, however one can be generated using this data. See the create\_LDprofile function for more information on how to create an LD profile.

#### Value

A list containing the SNP positions and the  $Z_{lpha}^{r^2/E[r^2]}$  values for those SNPs

#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

## See Also

create\_LDprofile

Zalpha\_Zscore

#### **Examples**

```
## load the snps and LDprofile example datasets
data(snps)
data(LDprofile)
## run Zalpha_rsq_over_expected over all the SNPs with a window size of 3000 bp
Zalpha_rsq_over_expected(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq)
## only return results for SNPs between locations 600 and 1500 bp
Zalpha_rsq_over_expected(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq,X=c(600,1500))
```

Zalpha\_Zscore

Runs the Zalpha function using the Z score of the r-squared values for the region

## **Description**

Returns a  $Z_{\alpha}^{Zscore}$  value for each SNP location supplied to the function, based on the expected  $r^2$  values given an LD profile and genetic distances. For more information about the  $Z_{\alpha}^{Zscore}$  statistic, please see Jacobs (2016). The  $Z_{\alpha}^{Zscore}$  statistic is defined as:

$$Z_{\alpha}^{Zscore} = \frac{{{{{\left( {\frac{{|L|}}{2}} \right)}^{ - 1}}\sum\nolimits_{i,j \in L} {\frac{{r_{i,j}^2 - E[r_{i,j}^2]}}{{\sigma [r_{i,j}^2]}}} + {{{{\left( {\frac{{|R|}}{2}} \right)}^{ - 1}}}\sum\nolimits_{i,j \in R} {\frac{{r_{i,j}^2 - E[r_{i,j}^2]}}{{\sigma [r_{i,j}^2]}}} }}{2}}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws,  $r^2$  is equal to the squared correlation between a pair of SNPs,  $E[r^2]$  is equal to the expected squared correlation between a pair of SNPs, given an LD profile, and  $\sigma[r^2]$  is the standard deviation.

## Usage

```
Zalpha_Zscore(
  pos,
  ws,
  x,
  dist,
  LDprofile_bins,
  LDprofile_rsq,
  LDprofile_sd,
  minRandL = 4,
  minRL = 25,
  X = NULL
)
```

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## Arguments

pos	A numeric vector of SNP locations
ws	The window size which the $Z_{\alpha}^{Zscore}$ statistic will be calculated over. This should be on the same scale as the pos vector.
X	A matrix of SNP values. Columns represent chromosomes; rows are SNP locations. Hence, the number of rows should equal the length of the pos vector. SNPs should all be biallelic.
dist	A numeric vector of genetic distances (e.g. $cM$ , $LDU$ ). This should be the same length as $pos$ .
LDprofile_bins	A numeric vector containing the lower bound of the bins used in the LD profile. These should be of equal size.
LDprofile_rsq	A numeric vector containing the expected $r^2$ values for the corresponding bin in the LD profile. Must be between $0$ and $1$ .
LDprofile_sd	A numeric vector containing the standard deviation of the $\it r^2$ values for the corresponding bin in the LD profile.
minRandL	$\label{eq:minimum number of SNPs in each set $R$ and $L$ for the statistic to be calculated.}$ Default is 4.
minRL	Minimum value for the product of the set sizes for R and L. Default is 25.
X	Optional. Specify a region of the chromosome to calculate $Z_{\alpha}^{Zscore}$ for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate $Z_{\alpha}^{Zscore}$ for every SNP in the pos vector.

## **Details**

The LD profile describes the expected correlation between SNPs at a given genetic distance, generated using simulations or real data. Care should be taken to utilise an LD profile that is representative of the population in question. The LD profile should consist of evenly sized bins of distances (for example 0.0001 cM per bin), where the value given is the (inclusive) lower bound of the bin. Ideally, an LD profile would be generated using data from a null population with no selection, however one can be generated using this data. See the create\_LDprofile function for more information on how to create an LD profile.

#### Value

A list containing the SNP positions and the  $Z^{Zscore}_{\alpha}$  values for those SNPs

## References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

## See Also

create\_LDprofile

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## **Examples**

```
## load the snps and LDprofile example datasets
data(snps)
data(LDprofile)
## run Zalpha_Zscore over all the SNPs with a window size of 3000 bp
Zalpha_Zscore(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq,LDprofile$sd)
## only return results for SNPs between locations 600 and 1500 bp
Zalpha_Zscore(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq,LDprofile$sd,X=c(600,1500))
```

Zbeta

Runs the Zbeta function

## **Description**

Returns a  $Z_{\beta}$  value for each SNP location supplied to the function. For more information about the  $Z_{\beta}$  statistic, please see Jacobs (2016). The  $Z_{\beta}$  statistic is defined as:

$$Z_{\beta} = \frac{\sum_{i \in L, j \in R} r_{i,j}^2}{|L||R|}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws, and  $r^2$  is equal to the squared correlation between a pair of SNPs

## Usage

```
Zbeta(pos, ws, x, minRandL = 4, minRL = 25, X = NULL)
```

#### **Arguments**

pos	A numeric vector of SNP locations
WS	The window size which the $Z_{\beta}$ statistic will be calculated over. This should be on the same scale as the pos vector.
х	A matrix of SNP values. Columns represent chromosomes; rows are SNP locations. Hence, the number of rows should equal the length of the pos vector. SNPs should all be biallelic.
minRandL	Minimum number of SNPs in each set $R$ and $L$ for the statistic to be calculated. Default is 4.
minRL	Minimum value for the product of the set sizes for R and L. Default is 25.
X	Optional. Specify a region of the chromosome to calculate $Z_{\beta}$ for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate $Z_{\beta}$ for every SNP in the pos vector.

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#### Value

A list containing the SNP positions and the  $Z_{\beta}$  values for those SNPs

#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

#### **Examples**

```
## load the snps example dataset
data(snps)
## run Zbeta over all the SNPs with a window size of 3000 bp
Zbeta(snps$bp_positions,3000,as.matrix(snps[,3:12]))
## only return results for SNPs between locations 600 and 1500 bp
Zbeta(snps$bp_positions,3000,as.matrix(snps[,3:12]),X=c(600,1500))
```

Zbeta\_BetaCDF

Runs the Zbeta function using a cumulative beta distribution function on the r-squared values for the region

## **Description**

Returns a  $Z_{\beta}^{BetaCDF}$  value for each SNP location supplied to the function, based on the expected  $r^2$  values given an LD profile and genetic distances. For more information about the  $Z_{\beta}^{BetaCDF}$  statistic, please see Jacobs (2016). The  $Z_{\beta}^{BetaCDF}$  statistic is defined as:

$$Z_{\beta}^{BetaCDF} = \frac{\sum_{i \in L, j \in R} \frac{B(r_{i,j}^2; a, b)}{B(a, b)}}{|L||R|}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws,  $r^2$  is equal to the squared correlation between a pair of SNPs, and  $\frac{B(r_{i,j}^2;a,b)}{B(a,b)}$  is the cumulative distribution function for the Beta distribution given the estimated a and b parameters from the LD profile.

#### Usage

```
Zbeta_BetaCDF(
  pos,
  ws,
  x,
  dist,
  LDprofile_bins,
  LDprofile_Beta_a,
  LDprofile_Beta_b,
  minRandL = 4,
```

Zbeta\_BetaCDF

```
minRL = 25,
  X = NULL
)
```

## Arguments

pos	A numeric vector of SNP locations	
WS	The window size which the $Z_{\beta}^{BetaCDF}$ statistic will be calculated over. This should be on the same scale as the pos vector.	
х	A matrix of SNP values. Columns represent chromosomes; rows are SNP locations. Hence, the number of rows should equal the length of the pos vector. SNPs should all be biallelic.	
dist	A numeric vector of genetic distances (e.g. cM, LDU). This should be the same length as pos.	
LDprofile_bins	A numeric vector containing the lower bound of the bins used in the LD profile. These should be of equal size.	
LDprofile_Beta_	_a	
	A numeric vector containing the first estimated Beta parameter for the corresponding bin in the LD profile.	
LDprofile_Beta_b		
	A numeric vector containing the second estimated Beta parameter for the corresponding bin in the LD profile.	
minRandL	Minimum number of SNPs in each set $R$ and $L$ for the statistic to be calculated. Default is 4.	
minRL	Minimum value for the product of the set sizes for R and L. Default is 25.	
X	Optional. Specify a region of the chromosome to calculate $Z_{\beta}^{BetaCDF}$ for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate $Z_{\beta}^{BetaCDF}$ for every SNP in the pos	

## **Details**

The LD profile describes the expected correlation between SNPs at a given genetic distance, generated using simulations or real data. Care should be taken to utilise an LD profile that is representative of the population in question. The LD profile should consist of evenly sized bins of distances (for example 0.0001 cM per bin), where the value given is the (inclusive) lower bound of the bin. Ideally, an LD profile would be generated using data from a null population with no selection, however one can be generated using this data. See the create\_LDprofile function for more information on how to create an LD profile.

#### Value

A list containing the SNP positions and the  $Z_{\beta}^{BetaCDF}$  values for those SNPs

vector.

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#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

#### See Also

```
create_LDprofile
```

#### **Examples**

```
## load the snps and LDprofile example datasets
data(snps)
data(LDprofile)
## run Zbeta_BetaCDF over all the SNPs with a window size of 3000 bp
Zbeta_BetaCDF(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$Beta_a,LDprofile$Beta_b)
## only return results for SNPs between locations 600 and 1500 bp
Zbeta_BetaCDF(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$Beta_a,LDprofile$Beta_b,X=c(600,1500))
```

Zbeta\_expected

Runs the Zbeta function on the expected r-squared values for the region

## Description

Returns a  $Z_{\beta}^{E[r^2]}$  value for each SNP location supplied to the function, based on the expected  $r^2$  values given an LD profile and genetic distances. For more information about the  $Z_{\beta}^{E[r^2]}$  statistic, please see Jacobs (2016). The  $Z_{\beta}^{E[r^2]}$  statistic is defined as:

$$Z_{\beta}^{E[r^2]} = \frac{\sum_{i \in L, j \in R} E[r_{i,j}^2]}{|L||R|}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws, and  $E[r^2]$  is equal to the expected squared correlation between a pair of SNPs, given an LD profile.

## Usage

```
Zbeta_expected(
  pos,
  ws,
  dist,
  LDprofile_bins,
  LDprofile_rsq,
  minRandL = 4,
```

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```
minRL = 25,
 X = NULL
)
```

#### **Arguments**

A numeric vector of SNP locations pos

The window size which the  $Z_{\beta}^{E[r^2]}$  statistic will be calculated over. This should be on the same scale as the pos vector. WS

dist A numeric vector of genetic distances (e.g. cM, LDU). This should be the same

length as pos.

LDprofile\_bins A numeric vector containing the lower bound of the bins used in the LD profile.

These should be of equal size.

A numeric vector containing the expected  $r^2$  values for the corresponding bin in LDprofile\_rsq

the LD profile. Must be between 0 and 1.

minRandL Minimum number of SNPs in each set R and L for the statistic to be calculated.

Default is 4.

Minimum value for the product of the set sizes for R and L. Default is 25. minRL

Χ

Optional. Specify a region of the chromosome to calculate  $Z_{\beta}^{E[r^2]}$  for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate  $Z^{E[r^2]}_{\beta}$  for every SNP in the pos vector.

#### **Details**

The LD profile describes the expected correlation between SNPs at a given genetic distance, generated using simulations or real data. Care should be taken to utilise an LD profile that is representative of the population in question. The LD profile should consist of evenly sized bins of distances (for example 0.0001 cM per bin), where the value given is the (inclusive) lower bound of the bin. Ideally, an LD profile would be generated using data from a null population with no selection, however one can be generated using this data. See the create\_LDprofile function for more information on how to create an LD profile.

#### Value

A list containing the SNP positions and the  $Z_{\beta}^{E[r^2]}$  values for those SNPs

#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps. Genetics, 2016. 203(4): p. 1807

#### See Also

create\_LDprofile

#### **Examples**

```
## load the snps and LDprofile example datasets
data(snps)
data(LDprofile)
## run Zbeta_expected over all the SNPs with a window size of 3000 bp
Zbeta_expected(snps$bp_positions,3000,snps$cM_distances,LDprofile$bin,LDprofile$rsq)
## only return results for SNPs between locations 600 and 1500 bp
Zbeta_expected(snps$bp_positions,3000,snps$cM_distances,LDprofile$bin,LDprofile$rsq,X=c(600,1500))
```

Zbeta\_log\_rsq\_over\_expected

Runs the Zbeta function on the log of the r-squared values over the expected r-squared values for the region

## Description

Returns a  $Z_{\beta}^{log_{10}(r^2/E[r^2])}$  value for each SNP location supplied to the function, based on the expected  $r^2$  values given an LD profile and genetic distances. For more information about the  $Z_{\beta}^{log_{10}(r^2/E[r^2])}$  statistic, please see Jacobs (2016). The  $Z_{\beta}^{log_{10}(r^2/E[r^2])}$  statistic is defined as:

$$Z_{\beta}^{\log_{10}(r^2/E[r^2])} = \frac{\sum_{i \in L, j \in R} \log_{10}(r_{i,j}^2/E[r_{i,j}^2])}{|L||R|}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws,  $r^2$  is equal to the squared correlation between a pair of SNPs, and  $E[r^2]$  is equal to the expected squared correlation between a pair of SNPs, given an LD profile.

#### Usage

```
Zbeta_log_rsq_over_expected(
  pos,
  ws,
  x,
  dist,
  LDprofile_bins,
  LDprofile_rsq,
  minRandL = 4,
  minRL = 25,
  X = NULL
)
```

#### **Arguments**

ws

pos A numeric vector of SNP locations

The window size which the  $Z_{\beta}^{log_{10}(r^2/E[r^2])}$  statistic will be calculated over. This should be on the same scale as the pos vector.

Х	A matrix of SNP values. Columns represent chromosomes; rows are SNP locations. Hence, the number of rows should equal the length of the pos vector. SNPs should all be biallelic.
dist	A numeric vector of genetic distances (e.g. cM, LDU). This should be the same length as pos.
LDprofile_bins	A numeric vector containing the lower bound of the bins used in the LD profile. These should be of equal size.
LDprofile_rsq	A numeric vector containing the expected $r^2$ values for the corresponding bin in the LD profile. Must be between $0$ and $1$ .
minRandL	Minimum number of SNPs in each set R and L for the statistic to be calculated.

Default is 4.

Minimum value for the product of the set sizes for R and L. Default is 25.

minRL Minimum value for the product of the set sizes for R and L. Default is 25.

Optional. Specify a region of the chromosome to calculate  $Z_{\beta}^{log_{10}(r^2/E[r^2])}$  for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate  $Z_{\beta}^{log_{10}(r^2/E[r^2])}$  for every SNP in the

pos vector.

#### **Details**

Χ

The LD profile describes the expected correlation between SNPs at a given genetic distance, generated using simulations or real data. Care should be taken to utilise an LD profile that is representative of the population in question. The LD profile should consist of evenly sized bins of distances (for example 0.0001 cM per bin), where the value given is the (inclusive) lower bound of the bin. Ideally, an LD profile would be generated using data from a null population with no selection, however one can be generated using this data. See the create\_LDprofile function for more information on how to create an LD profile.

#### Value

A list containing the SNP positions and the  $Z_{\beta}^{log_{10}(r^2/E[r^2])}$  values for those SNPs

## References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

#### See Also

```
create_LDprofile
```

## **Examples**

```
## load the snps and LDprofile example datasets
data(snps)
data(LDprofile)
## run Zbeta_log_rsq_over_expected over all the SNPs with a window size of 3000 bp
Zbeta_log_rsq_over_expected(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
```

```
LDprofile$bin,LDprofile$rsq)
## only return results for SNPs between locations 600 and 1500 bp
Zbeta_log_rsq_over_expected(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq,X=c(600,1500))
```

Zbeta\_rsq\_over\_expected

Runs the Zbeta function on the r-squared values over the expected r-squared values for the region

## **Description**

Returns a  $Z_{\beta}^{r^2/E[r^2]}$  value for each SNP location supplied to the function, based on the expected  $r^2$  values given an LD profile and genetic distances. For more information about the  $Z_{\beta}^{r^2/E[r^2]}$  statistic, please see Jacobs (2016). The  $Z_{\beta}^{r^2/E[r^2]}$  statistic is defined as:

$$Z_{\beta}^{r^2/E[r^2]} = \frac{\sum_{i \in L, j \in R} r_{i,j}^2/E[r_{i,j}^2]}{|L||R|}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws,  $r^2$  is equal to the squared correlation between a pair of SNPs, and  $E[r^2]$  is equal to the expected squared correlation between a pair of SNPs, given an LD profile.

#### Usage

```
Zbeta_rsq_over_expected(
  pos,
  ws,
  x,
  dist,
  LDprofile_bins,
  LDprofile_rsq,
  minRandL = 4,
  minRL = 25,
  X = NULL
)
```

## **Arguments**

pos	A numeric vector of SNP locations
WS	The window size which the $Z_{\beta}^{r^2/E[r^2]}$ statistic will be calculated over. This should be on the same scale as the pos vector.
X	A matrix of SNP values. Columns represent chromosomes; rows are SNP locations. Hence, the number of rows should equal the length of the pos vector. SNPs should all be biallelic

dist A numeric vector of genetic distances (e.g. cM, LDU). This should be the same

length as pos.

LDprofile\_bins A numeric vector containing the lower bound of the bins used in the LD profile.

These should be of equal size.

A numeric vector containing the expected  $r^2$  values for the corresponding bin in LDprofile\_rsq

the LD profile. Must be between 0 and 1.

minRandL Minimum number of SNPs in each set R and L for the statistic to be calculated.

Default is 4.

minRL Minimum value for the product of the set sizes for R and L. Default is 25.

Χ

Optional. Specify a region of the chromosome to calculate  $Z_{\beta}^{r^2/E[r^2]}$  for in the format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate  $Z_{\beta}^{r^2/E[r^2]}$  for every SNP in the pos

#### **Details**

The LD profile describes the expected correlation between SNPs at a given genetic distance, generated using simulations or real data. Care should be taken to utilise an LD profile that is representative of the population in question. The LD profile should consist of evenly sized bins of distances (for example 0.0001 cM per bin), where the value given is the (inclusive) lower bound of the bin. Ideally, an LD profile would be generated using data from a null population with no selection, however one can be generated using this data. See the create\_LDprofile function for more information on how to create an LD profile.

## Value

A list containing the SNP positions and the  $Z_{\beta}^{r^2/E[r^2]}$  values for those SNPs

#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps. Genetics, 2016. 203(4): p. 1807

#### See Also

```
create_LDprofile
```

## **Examples**

```
## load the snps and LDprofile example datasets
data(snps)
data(LDprofile)
## run Zbeta_rsq_over_expected over all the SNPs with a window size of 3000 bp
Zbeta_rsq_over_expected(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq)
## only return results for SNPs between locations 600 and 1500 bp
Zbeta_rsq_over_expected(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
```

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LDprofile\$bin,LDprofile\$rsq,X=c(600,1500))

Zbeta\_Zscore

Runs the Zbeta function using the Z score of the r-squared values for the region

## Description

Returns a  $Z_{\beta}^{Zscore}$  value for each SNP location supplied to the function, based on the expected  $r^2$  values given an LD profile and genetic distances. For more information about the  $Z_{\beta}^{Zscore}$  statistic, please see Jacobs (2016). The  $Z_{\beta}^{Zscore}$  statistic is defined as:

$$Z_{\beta}^{Zscore} = \frac{\sum_{i \in L, j \in R} \frac{r_{i,j}^2 - E[r_{i,j}^2]}{\sigma[r_{i,j}^2]}}{|L||R|}$$

where |L| and |R| are the number of SNPs to the left and right of the current locus within the given window ws,  $r^2$  is equal to the squared correlation between a pair of SNPs,  $E[r^2]$  is equal to the expected squared correlation between a pair of SNPs, given an LD profile, and  $\sigma[r^2]$  is the standard deviation.

## Usage

```
Zbeta_Zscore(
  pos,
  ws,
  x,
  dist,
  LDprofile_bins,
  LDprofile_rsq,
  LDprofile_sd,
  minRandL = 4,
  minRL = 25,
  X = NULL
)
```

#### **Arguments**

pos	A numeric vector of SNP locations
WS	The window size which the $Z_{\beta}^{Zscore}$ statistic will be calculated over. This should be on the same scale as the pos vector.
X	A matrix of SNP values. Columns represent chromosomes; rows are SNP locations. Hence, the number of rows should equal the length of the pos vector. SNPs should all be biallelic.
dist	A numeric vector of genetic distances (e.g. cM, LDU). This should be the same length as pos.

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LDprofile\_bins A numeric vector containing the lower bound of the bins used in the LD profile.

These should be of equal size.

LDprofile\_rsq A numeric vector containing the expected  $r^2$  values for the corresponding bin in

the LD profile. Must be between 0 and 1.

LDprofile\_sd A numeric vector containing the standard deviation of the  $r^2$  values for the cor-

responding bin in the LD profile.

minRandL Minimum number of SNPs in each set R and L for the statistic to be calculated.

Default is 4.

minRL Minimum value for the product of the set sizes for R and L. Default is 25.

X Optional. Specify a region of the chromosome to calculate  $Z_{\beta}^{Zscore}$  for in the

format c(startposition, endposition). The start position and the end position should be within the extremes of the positions given in the pos vector. If not supplied, the function will calculate  $Z_{\mathcal{B}}^{Zscore}$  for every SNP in the pos vector.

#### **Details**

The LD profile describes the expected correlation between SNPs at a given genetic distance, generated using simulations or real data. Care should be taken to utilise an LD profile that is representative of the population in question. The LD profile should consist of evenly sized bins of distances (for example 0.0001 cM per bin), where the value given is the (inclusive) lower bound of the bin. Ideally, an LD profile would be generated using data from a null population with no selection, however one can be generated using this data. See the create\_LDprofile function for more information on how to create an LD profile.

#### Value

A list containing the SNP positions and the  $Z_{\beta}^{Zscore}$  values for those SNPs

#### References

Jacobs, G.S., T.J. Sluckin, and T. Kivisild, *Refining the Use of Linkage Disequilibrium as a Robust Signature of Selective Sweeps*. Genetics, 2016. **203**(4): p. 1807

#### See Also

```
create_LDprofile
```

## **Examples**

```
## load the snps and LDprofile example datasets
data(snps)
data(LDprofile)
## run Zbeta_Zscore over all the SNPs with a window size of 3000 bp
Zbeta_Zscore(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq,LDprofile$sd)
## only return results for SNPs between locations 600 and 1500 bp
Zbeta_Zscore(snps$bp_positions,3000,as.matrix(snps[,3:12]),snps$cM_distances,
LDprofile$bin,LDprofile$rsq,LDprofile$sd,X=c(600,1500))
```

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