## Package 'HaploVar'

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Title Defining Local Haplotype Variants for Use in Trait Association

and Trait Prediction Analyses

Mitchell Bestry [ctb], Jacob Marsh [ctb], David Edwards [ctb]

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Type Package

Version 0.1.1 **Description** A local haplotyping tool for use in trait association and trait prediction analyses pipelines. 'HaploVar' enables users take single nucleotide polymorphisms (SNPs) (in VCF format) and a linkage disequilibrium (LD) matrix, calculate local haplotypes and format the output to be compatible with a wide range of trait association and trait prediction tools. The local haplotypes are calculated from the LD matrix using a clustering algorithm called density-based spatial clustering of applications with noise ('DBSCAN') (Ester et al., 1996) <ISBN: 1577350049>. **License** MIT + file LICENSE **Encoding UTF-8** RoxygenNote 7.3.2 Imports dplyr, tidyr, tibble, magrittr, dbscan **Depends** R (>= 4.00) LazyData true LazyDataCompression xz Suggests knitr, rmarkdown VignetteBuilder knitr NeedsCompilation no Author Tessa MacNish [aut, cre] (ORCID: <a href="https://orcid.org/0009-0008-8985-7348">https://orcid.org/0009-0008-8985-7348</a>), Hawlader Al-Mamun [ctb], Thomas Bergmann [ctb],

Maintainer Tessa MacNish <tessa.macnish@research.uwa.edu.au>

## **Contents**

collate_define_haplotypes	2
collate_haplotype_variants	2
define_haplotypes	3
define_haplotypes_globally	4
haplotype_variants	5
haplotype_variants_global	$\epsilon$
LD	7
vcf	7

Index 35

collate\_define\_haplotypes

Collate define\_haplotypes Lists

## Description

This function collates a list of output files from define\_haplotypes.

## Usage

```
collate_define_haplotypes(haplotype_list)
```

## Arguments

haplotype\_list A list of the lists created by the define\_haplotypes function.

#### Value

A collated list of all haplotype tables.

```
collate_haplotype_variants
```

Collate haplotype\_variants Tables

## Description

This function collates a list of output files from haplotype\_variants.

## Usage

```
collate_haplotype_variants(haplotype_variants_list, format = 1)
```

define\_haplotypes 3

#### **Arguments**

haplotype\_variants\_list

A list of the tables created by the define\_haplotypes function.

format The format you want the output table to be in. This should be the same number

you used when running define\_haplotypes.

#### Value

A collated table of haplotype variants.

define\_haplotypes

Define Haplotypes

#### **Description**

This function requires a VCF and an LD matrix. It will then define local haplotypes and return a list of tables. Each table within the list represents one haplotype. These haplotype tables display the SNP genotypes within the haplotype.

#### Usage

```
define_haplotypes(
  vcf,
  LD,
  epsilon = 0.6,
  MGmin = 30,
  hetmiss_as = "allele",
  keep_outliers = FALSE
)
```

#### **Arguments**

vcf A VCF file.

LD A LD matrix file.

epsilon Affects haplotype size. It is a parameter of the DBSCAN clustering tool. The

default is 0.6.

MGmin The minimum number of SNPs within a cluster for it to be defined as a haplo-

type. The default is 30.

hetmiss\_as Affects how missing data is handled for all instances where one allele in a geno-

type is missing. If hetmiss\_as = "allele" the genotype is assumed to be heterozy-

gous. If hetmiss\_as = "miss" the genotype is treated as NA.

keep\_outliers If FALSE, removes SNPs that are determined to be outliers.

#### Value

A list of haplotype tables.

```
\label{lem:define_haplotypes_globally} Define\ Haplotypes\ Globally
```

## Description

This function requires a list of VCF files and an LD matrices. The list of VCF files and LD matrices must be the same length. It will then define local haplotypes for each pair of files (VCF and LD matrix) and return a list of tables. Each table within the list represents one haplotype. These haplotype tables display the SNP genotypes within the haplotype.

## Usage

```
define_haplotypes_globally(
  vcf_list,
  LD_list,
  epsilon = NULL,
  MGmin = 30,
  hetmiss_as = "allele",
  keep_outliers = FALSE
)
```

## Arguments

vcf_list	A list of VCF files.
LD_list	A list LD matrix files.
epsilon	A list of epsilon values the same length as the list of VCF files. The epsilon affects haplotype size. It is a parameter of the DBSCAN clustering tool. The default is 0.6.
MGmin	The minimum number of SNPs within a cluster for it to be defined as a haplotype. The default is 30.
hetmiss_as	Affects how missing data is handled for all instances where one allele in a genotype is missing. If hetmiss_as = "allele" the genotype is assumed to be heterozygous. If hetmiss_as = "miss" the genotype is treated as NA.
keep_outliers	If FALSE, removes SNPs that are determined to be outliers.

#### Value

A collated list of haplotype tables for all VCF files provided.

haplotype\_variants 5

## Description

This function requires a VCF and an LD matrix. It will then define local haplotypes and identify the variants for each haplotype. The output can be formatted in six ways, to be compatible with a wide range of GWAS and genomic selection tools.

## Usage

```
haplotype_variants(
  vcf,
  LD,
  epsilon = 0.6,
  MGmin = 30,
  minFreq = 2,
  hetmiss_as = "allele",
  keep_outliers = FALSE,
  format = 1
)
```

## Arguments

vcf	A VCF file.
LD	A LD matrix file.
epsilon	Affects haplotype size. It is a parameter of the DBSCAN clustering tool. The default is 0.6.
MGmin	The minimum number of SNPs within a cluster for it to be defined as a haplotype. The default is 30.
minFreq	The minimum number of individuals a haplotype variant must be present in to be considered a valid haplotype variant. The default is 2.
hetmiss_as	Affects how missing data is handled for all instances where one allele in a geno- type is missing. If hetmiss_as = "allele" the genotype is assumed to be heterozy- gous. If hetmiss_as = "miss" the genotype is treated as NA.
keep_outliers	If FALSE removes SNPs, that are determined to be outliers.
format	The output format. There are six different output formats (1,2,3,4,5,6).

#### Value

A table of haplotype genotypes in your chosen format.

```
haplotype_variants_global
```

Identify Haplotype Variants Globally

## Description

This function requires a list of VCF files and an LD matrices. It will then define local haplotypes and identify the variants for each haplotype. The output can be formatted in six ways, to be compatible with a wide range of GWAS and genomic selection tools.

### Usage

```
haplotype_variants_global(
  vcf_list,
  LD_list,
  epsilon = NULL,
  MGmin = 30,
  minFreq = 2,
  hetmiss_as = "allele",
  keep_outliers = FALSE,
  format = 1
)
```

## Arguments

vcf_list	A list of VCF files.
LD_list	A list of LD matrix files.
epsilon	A list of epsilon values the same length as the list of VCF files. The epsilon affects haplotype size. It is a parameter of the DBSCAN clustering tool. The default is 0.6.
MGmin	The minimum number of SNPs within a cluster for it to be defined as a haplotype. The default is 30.
minFreq	The minimum number of individuals a haplotype variant must be present in to be considered a valid haplotype variant. The default is 2.
hetmiss_as	Affects how missing data is handled for all instances where one allele in a genotype is missing.If hetmiss_as = "allele" the genotype is assumed to be heterozygous. If hetmiss_as = "miss" the genotype is treated as NA.
keep_outliers	If FALSE removes SNPs, that are determined to be outliers.
format	The output format. There are six different output formats (1,2,3,4,5,6).

## Value

A table of haplotype genotypes in your chosen format.

LD

LD

Linkage Disequilibrium Matrix

7

### **Description**

Pairwise R^2 values for 490 Brassica napus single nucleotide polymorphisms (SNPs).

#### Usage

LD

#### **Format**

An object of class data. frame with 490 rows and 490 columns.

#### **Source**

Wu, D., Liang, Z., Yan, T., Xu, Y., Xuan, L., Tang, J., Zhou, G., Lohwasser, U., Hua, S., Wang, H., Chen, X., Wang, Q., Zhu, L., Maodzeka, A., Hussain, N., Li, Z., Li, X., Shamsi, I. H., Jilani, G., ... Jiang, L. (2019). Whole-Genome Resequencing of a Worldwide Collection of Rapeseed Accessions Reveals the Genetic Basis of Ecotype Divergence. Molecular Plant, 12(1), 30–43. https://doi.org/10.1016/j.molp.2018.11.007

vcf

Brassica napus genotype data in VCF format

#### **Description**

A subset of Brassica napus genotype data for chromosome C01. The genotype data reports single nucleotide polymorphism (SNP) data. The variables are as follows:

## Usage

vcf

#### **Format**

A data frame with 490 rows and 1000 variables:

#CHROM The chromsome where the SNP is located

**POS** The reference position of the SNP (bp)

**ID** The name/ID of the SNP

**REF** Reference base

ALT Alternate base

QUAL Phred-scaled quality score of the alternate base

FILTER PASS if the SNP has passed all filters

**INFO** Additional information

**FORMAT** The data type of the genotype

**R4155\_R4155** Genotypes for sample R4155\_R4155

**R4156 R4156** Genotypes for sample R4156 R4156

**R4157\_R4157** Genotypes for sample R4157\_R4157

**R4158\_R4158** Genotypes for sample R4158\_R4158

**R4159\_R4159** Genotypes for sample R4159\_R4159

**R4160\_R4160** Genotypes for sample R4160\_R4160

**R4161\_R4161** Genotypes for sample R4161\_R4161

**R4162\_R4162** Genotypes for sample R4162\_R4162

**R4163\_R4163** Genotypes for sample R4163\_R4163

**R4164\_R4164** Genotypes for sample R4164\_R4164

**R4165\_R4165** Genotypes for sample R4165\_R4165

**R4166 R4166** Genotypes for sample R4166 R4166

**R4167\_R4167** Genotypes for sample R4167\_R4167

**R4168\_R4168** Genotypes for sample R4168\_R4168

**R4169\_R4169** Genotypes for sample R4169\_R4169

**R4170\_R4170** Genotypes for sample R4170\_R4170

**R4171\_R4171** Genotypes for sample R4171\_R4171

**R4172\_R4172** Genotypes for sample R4172\_R4172

**R4173\_R4173** Genotypes for sample R4173\_R4173

**R4174\_R4174** Genotypes for sample R4174\_R4174

**R4176\_R4176** Genotypes for sample R4176\_R4176

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**R4178 R4178** Genotypes for sample R4178 R4178

**R4179\_R4179** Genotypes for sample R4179\_R4179

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**R4181\_R4181** Genotypes for sample R4181\_R4181

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**R4183\_R4183** Genotypes for sample R4183\_R4183

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**R4186\_R4186** Genotypes for sample R4186\_R4186

**R4187\_R4187** Genotypes for sample R4187\_R4187

**R4188 R4188** Genotypes for sample R4188 R4188

R4189\_R4189 Genotypes for sample R4189\_R4189

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R4190_R4190 Genotypes for sample R4190_R4190
R4191_R4191 Genotypes for sample R4191_R4191
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R4224_R4224 Genotypes for sample R4224_R4224
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R4226_R4226 Genotypes for sample R4226_R4226
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R4872_R4872 Genotypes for sample R4872_R4872
R4873 R4873 Genotypes for sample R4873 R4873
R4874_R4874 Genotypes for sample R4874_R4874
R4875_R4875 Genotypes for sample R4875_R4875
R4876_R4876 Genotypes for sample R4876_R4876
R4877_R4877 Genotypes for sample R4877_R4877
R4878 R4878 Genotypes for sample R4878 R4878
R4879 R4879 Genotypes for sample R4879 R4879
R4880 R4880 Genotypes for sample R4880 R4880
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R4882_R4882 Genotypes for sample R4882_R4882
R4883_R4883 Genotypes for sample R4883_R4883
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R4885_R4885 Genotypes for sample R4885_R4885
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R4887_R4887 Genotypes for sample R4887_R4887
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R4889 R4889 Genotypes for sample R4889 R4889
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R4893_R4893 Genotypes for sample R4893_R4893
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R4913_R4913 Genotypes for sample R4913_R4913
R4914_R4914 Genotypes for sample R4914_R4914
R4915 R4915 Genotypes for sample R4915 R4915
R4916 R4916 Genotypes for sample R4916 R4916
R4917 R4917 Genotypes for sample R4917 R4917
R4918 R4918 Genotypes for sample R4918 R4918
R4919_R4919 Genotypes for sample R4919_R4919
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R4938_R4938 Genotypes for sample R4938_R4938
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R5097 R5097 Genotypes for sample R5097 R5097
R5098 R5098 Genotypes for sample R5098 R5098
R5099_R5099 Genotypes for sample R5099_R5099
R5100 R5100 Genotypes for sample R5100 R5100
R5101_R5101 Genotypes for sample R5101_R5101
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R5117 R5117 Genotypes for sample R5117 R5117
R5118 R5118 Genotypes for sample R5118 R5118
R5119 R5119 Genotypes for sample R5119 R5119
R5120_R5120 Genotypes for sample R5120_R5120
R5121_R5121 Genotypes for sample R5121_R5121
R5122_R5122 Genotypes for sample R5122_R5122
R5123_R5123 Genotypes for sample R5123_R5123
R5124_R5124 Genotypes for sample R5124_R5124
R5125_R5125 Genotypes for sample R5125_R5125
R5126_R5126 Genotypes for sample R5126_R5126
R5127_R5127 Genotypes for sample R5127_R5127
R5128 R5128 Genotypes for sample R5128 R5128
R5129_R5129 Genotypes for sample R5129_R5129
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**R5130\_R5130** Genotypes for sample R5130\_R5130 **R5131\_R5131** Genotypes for sample R5131\_R5131 **R5132\_R5132** Genotypes for sample R5132\_R5132 **R5133 R5133** Genotypes for sample R5133 R5133 **R5134\_R5134** Genotypes for sample R5134\_R5134 **R5135\_R5135** Genotypes for sample R5135\_R5135 **R5136\_R5136** Genotypes for sample R5136\_R5136 **R5137\_R5137** Genotypes for sample R5137\_R5137 **R5138\_R5138** Genotypes for sample R5138\_R5138 R5139\_R5139 Genotypes for sample R5139\_R5139 **R5140\_R5140** Genotypes for sample R5140\_R5140 **R5141\_R5141** Genotypes for sample R5141\_R5141 **R5142\_R5142** Genotypes for sample R5142\_R5142 **R5143\_R5143** Genotypes for sample R5143\_R5143 **R5144\_R5144** Genotypes for sample R5144\_R5144 **R5145\_R5145** Genotypes for sample R5145\_R5145 **R5146\_R5146** Genotypes for sample R5146\_R5146 **R5147\_R5147** Genotypes for sample R5147\_R5147 R5148\_R5148 Genotypes for sample R5148\_R5148 **R5149\_R5149** Genotypes for sample R5149\_R5149 **R5150 R5150** Genotypes for sample R5150 R5150 R5151\_R5151 Genotypes for sample R5151\_R5151 **R5152\_R5152** Genotypes for sample R5152\_R5152 **R5153\_R5153** Genotypes for sample R5153\_R5153 **R5154 R5154** Genotypes for sample R5154 R5154 **R5155 R5155** Genotypes for sample R5155 R5155 **R5156\_R5156** Genotypes for sample R5156\_R5156 **R5157\_R5157** Genotypes for sample R5157\_R5157 **R5158 R5158** Genotypes for sample R5158 R5158 **R5159 R5159** Genotypes for sample R5159 R5159 **R5160\_R5160** Genotypes for sample R5160\_R5160 **R5161\_R5161** Genotypes for sample R5161\_R5161

#### Source

Wu, D., Liang, Z., Yan, T., Xu, Y., Xuan, L., Tang, J., Zhou, G., Lohwasser, U., Hua, S., Wang, H., Chen, X., Wang, Q., Zhu, L., Maodzeka, A., Hussain, N., Li, Z., Li, X., Shamsi, I. H., Jilani, G., ... Jiang, L. (2019). Whole-Genome Resequencing of a Worldwide Collection of Rapeseed Accessions Reveals the Genetic Basis of Ecotype Divergence. Molecular Plant, 12(1), 30–43. https://doi.org/10.1016/j.molp.2018.11.007

# **Index**

```
* datasets
    LD, 7
    vcf, 7

collate_define_haplotypes, 2
collate_haplotype_variants, 2

define_haplotypes, 3
    define_haplotypes_globally, 4

haplotype_variants, 5
haplotype_variants_global, 6

LD, 7

vcf, 7
```