# Package 'polyBreedR'

December 20, 2023
Title Genomics-assisted breeding for polyploids (and diploids)
Version 0.36
Author Jeffrey B. Endelman
Maintainer Jeffrey Endelman <endelman@wisc.edu></endelman@wisc.edu>
<b>Description</b> Genomics-assisted breeding for polyploids (and diploids)
<b>Depends</b> R (>= $4.0$ )
License GPL-3
LazyData true
RoxygenNote 7.2.3
Encoding UTF-8
<b>Imports</b> AGHmatrix, ggplot2, ggrepel, pedigree, grDevices, utils, tidyr, Matrix, methods, rlang, up dog, randomForest, vcfR, rrBLUP, data.table
Suggests knitr, rmarkdown, asreml
VignetteBuilder knitr

## R topics documented:

ray2vcf	2
_mat	2
eck_ploidy	3
eck_trio	4
rt2vcf	4
8	5
no_call	6
t_pedigree	7
Γ2DS	7
vsA	8
_mat	9
ppute	9
npute_L2H	10
npute_PO	11
adc	12
erge_impute	12
adXY	13
odate_alias	14
f2csv	14
rite_vcf	15

2 A\_mat

Index 16

array2vcf	SNP array to VCF

#### **Description**

Converts output from Genome Studio (Final Report or Wide) to VCF

#### Usage

```
array2vcf(array.file, map.file, model.file = NULL, ploidy, vcf.file)
```

#### **Arguments**

array.file	name of input file with SNP array allele intensities
map.file	vcf file with map positions for the markers
model.file	normal mixture model parameters for genotype calls
ploidy	sample ploidy, for use with model.file
vcf.file	output vcf file

#### **Details**

Auto-detects whether the input file is a Genome Studio Final Report, which is a "long" format with 9-row header, or in "wide" format, where all the data for each marker is one row. XY values are multiplied by 100

Genotype calls will attempt to be imported from the GS Final Report when model.file=NULL. For diploids, columns named "Allele 1 - AB" and "Allele 2 - AB" are expected. For tetraploids, a single column named "Alleles - AB" is expected.

It is assumed that the parameters in model.file lead to genotype calls for the dosage of allele B. For a VCF file, genotype calls need to be based on the dosage of ALT. By default, it is assumed that A is the REF allele. For variants where B is REF, include "REF=B" as INFO in the VCF map.file.

A_mat	Additive relationship matrix from pedigree

#### **Description**

Additive relationship matrix from pedigree

## Usage

```
A_mat(ped, ploidy, order.ped = TRUE)
```

#### **Arguments**

ped	Pedigree in three column format: id, mother, father

ploidy 2 or 4

order.ped TRUE/FALSE does the pedigree need to be ordered so that progeny follow par-

ents

check\_ploidy 3

#### **Details**

This is a wrapper that prepares the pedigree in the format required for R package AGHmatrix by Amadeu et al. (2016) (cite them if you use this function). A random bivalents model for tetraploid meiosis is assumed.

#### Value

Additive relationship matrix (dim: indiv x indiv)

#### References

Amadeu et al. (2016) Plant Genome 9, doi:10.3835/plantgenome2016.01.0009

check\_ploidy

Check ploidy

#### **Description**

Fraction of simplex or triplex markers

#### Usage

```
check_ploidy(geno, map)
```

#### **Arguments**

geno Genotype matrix (markers x indiv)

map Data frame with marker map (Marker, Chrom, Position)

#### **Details**

For every indiv in the genotype matrix, the fraction of markers per chromosome called as simplex or triplex is calculated, which should be low for diploids. A small amount of missing genotype data can be tolerated.

## Value

List containing

mat Matrix (indiv x chrom) of results

plot ggplot2 barplot

4 dart2vcf

check\_trio

Check markers for parent-offspring trio

#### **Description**

Check markers for parent-offspring trio

## Usage

```
check_trio(parentage, geno, ploidy)
```

#### **Arguments**

parentage Data frame with three columns: id, mother, father

geno Matrix of allele dosages: markers x indiv

ploidy 2 or 4

#### **Details**

Computes the percentage of markers at which the two parents and offspring have incompatible allele dosages (for tetraploids, the random bivalents model is used). For dihaploid offspring of a single tetraploid parent, use ploidy = 4 and "haploid" for the father in parentage, as well as a diploid (0,1,2) genotype for the offspring. A small amount of missing genotype data can be tolerated.

#### Value

Data frame with the percentage of incompatible markers for each trio

dart2vcf

Convert DArTag to VCF

#### **Description**

Convert DArTag to VCF

## Usage

```
dart2vcf(counts.file, dosage.file, vcf.file, ploidy, first.data.row = 9)
```

#### **Arguments**

counts.file DArTag collapsed counts file

dosage.file DArTag dosage file

vcf.file name of VCF output file (uncompressed)

ploidy ploidy

first.data.row default is 9 for DArTag format

gbs 5

#### **Details**

Two input files expected. counts.file is the two-row collapsed counts file, whereas dosage.file has one row per target, with chrom and position in columns 4 and 5. DArT reports dosage of REF, whereas VCF standard is based on dosage of ALT. The dosage is exported as GT field in VCF.

Duplicate samples are renamed by appending the "Target ID".

gbs Genotype calls for GBS

## Description

Genotype calls for genotype-by-sequencing (GBS) data

## Usage

```
gbs(in.file, out.file, ploidy, prior = "norm", bias = TRUE, n.core = 1)
```

## **Arguments**

in.file	VCF input file
out.file	VCF output file
ploidy	ploidy
prior	model for prior (see Details)
bias	TRUE/FALSE, whether to estimate allelic bias
n.core	number of cores

#### **Details**

VCF input file must contain AD field. Posterior mode and mean genotypes are added as GT and DS fields. GQ is also added based on probability of posterior mode. Binomial calculation uses R/updog package (Gerard et al. 2018). Previous INFO is discarded; adds NS, DP.AVG, AF.GT, AB, OD, SE.

## Value

marker x indiv matrix of read depths

geno\_call

geno\_call Genotype calls

## **Description**

Genotype calls based on a normal mixture model

#### Usage

```
geno_call(
  data,
  filename,
  model.ploidy = 4L,
  sample.ploidy = 4L,
  min.posterior = 0,
  transform = TRUE
)
```

## Arguments

data matrix (markers x id) of input values for the normal mixture model

filename CSV filename with the model parameters

model.ploidy 2 or 4 (default)

sample.ploidy 2 or 4 (default)

min.posterior minimum posterior probability (default 0) for genotype call

TRUE (default) or FALSE whether to apply arcsin square root transformation

## Details

transform

The first column of the CSV input file should be the SNP ID, followed by columns for the normal distribution means, standard deviations, and mixture probabilities. Genotype calls are based on the maximum a posteriori (MAP) method. If the posterior probability of the MAP genotype is less than min.posterior, then NA is returned for that sample. By default, an arcsin square root transformation is applied to the input values to match the approach used by R package fitPoly. To use a tetraploid mixture model for diploid samples, set sample.ploidy = 2 and model.ploidy = 4.

#### Value

matrix of allele dosages (0,1,2,...ploidy) with dimensions markers x individuals

get\_pedigree 7

get_pedigree	Generate pedigree	
--------------	-------------------	--

#### **Description**

Generate pedigree for a set of individuals

#### Usage

```
get_pedigree(id, pedfile, delim = ",", na.string = "NA", trim = TRUE)
```

#### **Arguments**

id Vector of names of individuals

pedfile Name of pedigree file

delim Delimiter for the pedigree file (default is "," for CSV)

na.string String used for NA in the pedigree file (default is "NA")

trim TRUE/FALSE whether to trim pedigree (see Details)

#### **Details**

Finds ancestors of individuals in a three-column pedigree file (id,mother,father). The id column can be the identifier for an individual or cross. String matches must be exact or based on the naming convention crossID-progenyID. The returned pedigree is ordered using R package pedigree so that offspring follow parents. When trim is TRUE (default), the pedigree is trimmed to remove ancestors with only one offspring (which are not needed to compute the pedigree relationship matrix).

#### Value

Data frame with columns id, mother, father

GT2DS	Convert GT to ALT allele dosage (DS)	

#### Description

Convert GT to ALT allele dosage (DS)

#### Usage

```
GT2DS(GT, diploidize = FALSE, n.core = 1)
```

## Arguments

GT	GT string
diploidize	TRUE/FALSE
n.core	number of cores

8 GvsA

#### **Details**

If diploidize is TRUE, data are recoded as a diploid 0,1,2.

#### Value

integer data with same dimensions as GT

GvsA Plot G vs. A

## Description

Plot marker-based vs. pedigree-based additive relationship coefficients

## Usage

```
GvsA(
  parentage,
  G,
  A,
  filename = NULL,
  thresh.G = Inf,
  thresh.A = 0.5,
  Gmax = NULL,
  Amax = NULL
)
```

## **Arguments**

parentage	Data frame of individuals to plot, with 3 columns: id,mother,father
G	Genomic relationship matrix
A	Pedigree relationship matrix
filename	Name of PDF file to save the results (optional for one individual)
thresh.G	Threshold above which names are displayed (default Inf)
thresh.A	Threshold above which names are displayed (default 0.5)
Gmax	Upper limit for y-axis for plotting. If NULL, maximum value in G is used.
Amax	Upper limit for x-axis for plotting. If NULL, maximum value in A is used.

## **Details**

Useful for finding and correcting pedigree errors. If the G or A coefficient for an individual exceeds the threshold, its name is displayed in the figure. If parentage contains one individual, by default a ggplot2 variable will be returned, but the result can also be written to file. If multiple individuals are present, a filename is required.

G\_mat 9

G\_mat

Additive genomic relationships

## Description

Relationship matrix for additive effects with bi-allelic markers

## Usage

```
G_mat(geno, ploidy)
```

#### **Arguments**

geno Matrix of allele dosages (markers x indiv) ploidy Any even integer (2,4,6,...)

#### **Details**

Additive effects are based on the traditional orthogonal decomposition of genetic variance in panmictic populations (Fisher 1918; Kempthorne 1957; Endelman et al. 2018). Missing genotype data is replaced with the population mean.

#### Value

G matrix

#### References

```
Fisher (1918) Trans. Roy. Soc. Edin. 52:399-433.
Kempthorne (1957) An Introduction to Genetic Statistics.
Endelman et al. (2018) Genetics 209:77-87.
```

impute

Impute missing data for bi-allelic markers

#### **Description**

Impute missing data for bi-allelic markers

## Usage

```
impute(
   in.file,
   out.file,
   ploidy,
   method,
   geno,
   min.DP = 1,
   max.missing,
   params = NULL,
   n.core = 1
```

10 impute\_L2H

#### **Arguments**

in.file	VCF input file
out.file	VCF output file
ploidy	ploidy
method	One of the following: "pop", "EM", "RF"
geno	One of the following: "GT","DS"
min.DP	genotypes below this depth are set to missing (default=1)
max.missing	remove markers above this threshold, as proportion of population
params	list of method-specific parameters
n.core	multicore processing

#### **Details**

Assumes input file is sorted by position. Markers with no genetic variance are removed. method="pop" imputes with the population mean for geno="DS" and population mode for geno="GT". method="EM" uses parameter "tol" (default is 0.02, see rrBLUP A.mat documentation). Imputed values are truncated if needed to fall in the interval [0,ploidy].

method="RF" uses parameters "ntree" (default 100) for number of trees and "nflank" (default 100) for the number of flanking markers (on each side) to use as predictors. Because RF first uses EM to generate a complete dataset, parameter "tol" is also recognized.

impute\_L2H

Impute from low to high density markers by Random Forest

## **Description**

Impute from low to high density markers by Random Forest

## Usage

```
impute_L2H(
  high.file,
  low.file,
  out.file,
  params = list(),
  exclude = NULL,
  n.core = 1
)
```

## **Arguments**

high.file	name of high density file
low.file	name of low density file
out.file	name of CSV output file for imputed data
params	list of parameters (see Details)
exclude	optional, vector of high density samples to exclude
n.core	multicore processing

impute\_PO 11

#### **Details**

Argument params is a list with three named elements: format, n.tree, n.mark. format can have values "GT" (integer dosage) or "DS" (real numbers between 0 and ploidy). Classification trees are used for GT and regression trees for DS. n. tree is the number of trees (default = 100). n.mark is the number of markers to use as predictors (default = 100), chosen based on minimum distance to the target.

The exclude argument is useful for cross-validation.

Both VCF and CSV are allowable input file formats—they are recognized based on the file extension. For CSV, the first three columns should be marker, chrom, pos. The output file is CSV.

Any missing data are imputed separately for each input file at the outset, using the population mean (DS) or mode (GT) for each marker.

#### Value

matrix of OOB error with dimensions markers x trees

impute_PO	Impute from low to high density markers with PolyOrigin

#### **Description**

Impute from low to high density markers by linkage analysis with PolyOrigin

#### Usage

```
impute_PO(high.file, low.file, low.format = "GT", ped.file, out.prefix)
```

## **Arguments**

high.file	name of high density file with phased parents	
low.file	name of low density VCF file with progeny	
low.format	either "GT" (default) or "AD"	
ped.file	pedigree file for progeny (must follow PO format)	
out.prefix	prefix for output files	

#### **Details**

You must have separately installed PolyOrigin and Julia for this function to work.

#' The high density file contains phased parental genotypes in PolyOrigin format. The first 3 columns are the genetic map in cM: marker, chrom, position. To output imputed data with physical rather than genetic map positions, including a fourth column named "bp". Subsequent columns are the phased parental genotypes.

VCF is assumed for the low-density file. The pedigree file must follow PolyOrigin format.

Two output files are generated, corresponding to the posterior maximum and mean genotypes.

12 merge\_impute

m	1	М	_

Multi-allelic genotype calls from DArTag

#### **Description**

Genotype calls from the DArTag MADC (Missing Allele Discovery Count) file

#### Usage

```
madc(madc.file, marker = "CDF1", min.AD = 5)
```

#### **Arguments**

madc.file MADC filename

marker Name of marker ("CDF1" is only option so far)

min. AD minimum AD for discovery

#### **Details**

Due to multi-allelism, for some trait markers a correct interpretation is not possible using the collapsed counts file; the MADC file is needed. Currently, the only marker implemented is CDF1 for potato DArTag, which assumes samples are diploid and there are no CDF1.1 homozygotes. The min.AD parameter establishes the threshold at which an allele is considered present, which is needed to account for sequencing and other errors.

#### Value

data frame of counts and diploid GT

merge_	imp	ute
c. 5c_	- T III P	ucc

Merge two genotype matrices and impute missing data

#### **Description**

Merge two genotype matrices and impute missing data by BLUP

## Usage

```
merge_impute(geno1, geno2, ploidy)
```

## Arguments

geno1	Genotype matrix (coded 0ploidy) with dimensions markers x indiv
geno2	Genotype matrix (coded 0ploidy) with dimensions markers x indiv
ploidy	Either 2 or 4

readXY 13

#### **Details**

Designed to impute from low to high density markers. The BLUP method is equivalent to Eq. 4 of Poland et al. (2012), but this function is not iterative. Additional shrinkage toward the mean is applied if needed to keep the imputed values within the range [0,ploidy]. Missing data in the input matrices are imputed with the population mean for each marker. If an individual appears in both input matrices, it is renamed with suffixes ".1" and ".2" and treated as two different individuals. Monomorphic markers are removed.

#### Value

Imputed genotype matrix (markers x indiv)

#### References

Poland et al. (2012) Plant Genome 5:103-113.

readXY

Read SNP array intensity data

#### **Description**

Read SNP array intensity data

## Usage

```
readXY(filename, skip = 9, output = "ratio")
```

## **Arguments**

filename filename

skip number of lines to skip before the header line with the column names

output One of three options: "ratio", "theta", "AD"

#### **Details**

The first two columns of the tab-delimited input file should be the SNP and Sample ID. Columns labeled "X" and "Y" contain the signal intensities for the two alleles. Use output to specify whether to return the ratio = Y/(X+Y) or theta = atan(Y/X)\*2/pi. Option "AD" exports the XY data in the allele depth format for a VCF file ("X,Y"), with the X and Y values multiplied by 100 and rounded to the nearest integer.

#### Value

matrix with dimensions markers x individuals

14 vcf2csv

update\_alias

Update names based on alias

## **Description**

Update names based on data frame with alias and preferred name

#### Usage

```
update_alias(x, alias, remove.space = TRUE, filename = NULL)
```

#### **Arguments**

x Vector of names to update

alias Data frame with two columns: first is the preferred name and second is the alias

remove.space TRUE/FALSE

filename update names in CSV file

#### **Details**

Parameter remove. space indicates whether blank spaces should be removed before string matching.

#### Value

Vector with updated names

vcf2csv

Convert VCF to CSV

## Description

Convert VCF to CSV

## Usage

```
vcf2csv(vcf.file, csv.file, format)
```

## Arguments

vcf.file Input file csv.file Output file

format Name of FORMAT key to export, either "GT" or "DS"

#### Value

none

write\_vcf

write_vcf
-----------

#### **Description**

Create VCFv4.3 file

#### Usage

```
write_vcf(filename, fixed, geno, other.meta = NULL)
```

#### **Arguments**

filename VCF file name

fixed character matrix with 8 columns: CHROM, POS, ID, REF, ALT, QUAL, FIL-

TER, INFO

geno named list of genotype matrices, see Details

other .meta optional, other metadata (without ##) besides INFO and FORMAT keys

#### Details

Several standard INFO key are recognized: ##INFO=<ID=REF,Number=A,Type=Character,Description=\"Array allele (A/B) in reference genome\"> ##INFO=<ID=NS,Number=1,Type=Integer,Description="Number of samples with data"> ##INFO=<ID=DP.AVG,Number=1,Type=Float,Description="Average Sample Depth"> ##INFO=<ID=DP,Number=1,Type=Integer,Description="Total Depth"> ##INFO=<ID=AB,Number=1,Type=Integer,Description="Total Depth"> ##INFO=<ID=AB,Number=1,Type=Integer,Description="Sequencing Error (PHRED)"> ##INFO=<ID=AB,Number=1,Type=Integer,Description="Allele Frequency"> ##INFO=<ID=AF,GT,Number=1,Type=Integer,Description="Allele Count in genotypes"> ##INFO=<ID=AC,Number=A,Type=Integer,Description="Total number of alleles"> ##INFO=<ID=AN,Number=1,Type=Integer,Description="Total n

Any additional metadata should be included without the ## prefix.

## **Index**

```
A_{mat, 2}
array2vcf, 2
{\sf check\_ploidy}, {\color{red} 3}
check_trio, 4
dart2vcf, 4
G_mat, 9
gbs, 5
geno_call, 6
get_pedigree, 7
GT2DS, 7
GvsA, 8
impute, 9
impute\_L2H,\, \textcolor{red}{10}
impute\_PO,\, \color{red} 11
\mathsf{madc},\, \textcolor{red}{12}
merge_impute, 12
readXY, 13
update\_alias, \\ 14
vcf2csv, 14
write_vcf, 15
```