# Package 'BIGr'

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Title (B)reeding (I)nsight (G)enomics Functions for Polypoid and Diploid Species
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<b>Description</b> This package contains the functions developed within Breeding Insight to analyze diploid and polyploid breeding and genetic data.
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calculate\_Het

Calculate Observed Heterozygosity from a Genotype Matrix

## **Description**

This function calculates the observed heterozygosity from a genotype matrix. It assumes that the samples are the columns, and the genomic markers are in rows. Missing data should be set as NA, which will then be ignored for the calculations. All samples must have the same ploidy.

### Usage

```
calculate_Het(geno, ploidy)
```

### **Arguments**

geno Genotype matrix or data.frame

ploidy The ploidy of the species being analyzed

#### Value

A dataframe of observed heterozygosity values for each sample

calculate\_MAF

Calculate Minor Allele Frequency from a Genotype Matrix

## Description

This function calculates the allele frequency and minor allele frequency from a genotype matrix. It assumes that the Samples are the columns, and the genomic markers are in rows. Missing data should be set as NA, which will then be ignored for the calculations. All samples must have the same ploidy.

## Usage

```
calculate_MAF(df, ploidy)
```

## Arguments

df Genotype matrix or data.frame

ploidy The ploidy of the species being analyzed

#### Value

A dataframe of AF and MAF values for each marker

```
capture_diversity.Gmat
```

Estimate Minimum Number of Individuals to Sample to Capture Population Genomic Diversity (Genotype Matrix)

### **Description**

This function can be used to estimate the number of individuals to sample from a population in order to capture a desired percentage of the genomic diversity. It assumes that the samples are the columns, and the genomic markers are in rows. Missing data should be set as NA, which will then be ignored for the calculations. All samples must have the same ploidy. This function was adapted from a previously developed Python method (Sandercock et al., 2023) (https://github.com/alex-sandercock/Capturing\_genomic\_diversity/)

## Usage

```
capture_diversity.Gmat(
   df,
   ploidy,
   r2_threshold = 0.9,
   iterations = 10,
   sample_list = NULL,
   parallel = FALSE,
   batch = 1,
   save.result = TRUE
)
```

## Arguments

Genotype matrix or data.frame with the count of alternate alleles (0=homozygous reference, 1 = heterozygous, 2 = homozygous alternate)
The ploidy of the species being analyzed
The ratio of diversity to capture (default = $0.9$ )
The number of iterations to perform to estimate the average result (default = $10$ )
The list of samples to subset from the dataset (optional)
Run the analysis in parallel (True/False) (default = FALSE)
Save the results to a .txt file? (default = TRUE)

#### Value

A data frame with minimum number of samples required to match or exceed the input ratio

## References

Sandercock, A. M., Westbrook, J. W., Zhang, Q., & Holliday, J. A. (2024). The road to restoration: Identifying and conserving the adaptive legacy of American chestnut. PNAS (in press).

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check\_ped

Evaluate Pedigree File for Accuracy

#### **Description**

Check a pedigree file for accuracy and output suspected errors

#### Usage

```
check_ped(ped.file)
```

#### **Arguments**

ped.file

path to pedigree text file. The pedigree file is a 3-column pedigree tab separated file with columns labeled as id sire dam in any order

#### **Details**

check\_ped takes a 3-column pedigree tab separated file with columns labeled as id sire dam in any order and checks for:

- Ids that appear more than once in the id column
- Ids that appear in both sire and dam columns
- Direct (e.g. parent is a offspring of his own daughter) and indirect (e.g. a great grandparent is son of its granchild) dependencies within the pedigree.
- Individuals included in the pedigree as sire or dam but not on the id column and reports them back with unknown parents (0).

When using check\_ped, do a first run to check for repeated ids and parents that appear as sire and dam. Once these errors are cleaned run the function again to check for dependencies as this will provide the most accurate report.

Note: This function does not change the input file but prints any errors found in the console.

#### Value

A list of dataframes of error types, and the output printed to the console

#### **Examples**

```
##Get list with a dataframe for each error type
#ped_errors <- check_ped(ped.file = "example_ped.txt")
##Access the "messy parents" dataframe result
#ped_errors$messy_parents

##Get list of sample IDs with messy parents error
#messy_parent_ids <- ped_errors$messy_parents$id
#print(messy_parent_ids)</pre>
```

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dosage2vcf Convert DArTag Dosage and Counts to VCF
--

## Description

This function will convert the DArT Dosage Report and Counts files to VCF format

## Usage

```
dosage2vcf(dart.report, dart.counts, ploidy, output.file)
```

## **Arguments**

dart.report	Path to the DArT dosage report .csv file. Typically contains "Dosage Report" in the file name.
dart.counts	Path to the DArT counts .csv file. Typically contains "Counts" in the file name.
ploidy	The ploidy of the species being analyzed
output.file	output file name and path

## **Details**

This function will convert the Dosage Report and Counts files from DArT into a VCF file. These two files are received directly from DArT for a given sequencing project. The output file will be saved to the location and with the name that is specified. The VCF format is v4.3

#### Value

A vcf file

# **Examples**

##The function will output the converted VCF using information from the DArT files

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dosage\_ratios

Calculate the Percentage of Each Dosage Value

#### **Description**

This function calculates the percentage of each dosage value within a genotype matrix. It assumes that the samples are the columns, and the genomic markers are in rows. Missing data should be set as NA, which will then be ignored for the calculations. All samples must have the same ploidy.

#### Usage

```
dosage_ratios(matrix_data, ploidy)
```

### **Arguments**

ploidy The ploidy of the species being analyzed

df Genotype matrix or data.frame

#### Value

A data.frame with percentages of dosage values in the genotype matrix

flip\_dosage

Switch Dosage Values from a Genotype Matrix

### **Description**

This function converts the dosage count values to the opposite value. This is primarily used when converting dosage values from reference based (0 = homozygous reference) to alternate count based (0 = homozygous alternate). It assumes that the Samples are the columns, and the genomic markers are in rows. Missing data should be set as NA, which will then be ignored for the calculations. All samples must have the same ploidy.

### Usage

```
flip_dosage(df, ploidy, is_reference = TRUE)
```

## **Arguments**

df Genotype matrix or data.frame

ploidy The ploidy of the species being analyzed

#### Value

A genotype matrix

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get\_countsMADC

Obtain Read Counts from MADC File

#### **Description**

This function takes the MADC file as input and retrieves the ref and alt counts for each sample, and converts them to ref, alt, and size(total count) matrices for dosage calling tools. At the moment, only the read counts for the Ref and Alt target loci are obtained while the additional loci are ignored.

## Usage

```
get_countsMADC(madc_file)
```

## **Arguments**

madc\_file

Path to MADC file

#### Value

A list of read count matrices for reference, alternate, and total read count values

updog2vcf

Export Updog Results as VCF

#### **Description**

This function will convert an Updog output to a VCF file

#### **Usage**

```
updog2vcf(multidog.object, ploidy, output.file)
```

### Arguments

 $\verb|multidog.object|$ 

updog output object with class "multidog" from dosage calling

ploidy The ploidy of the species being analyzed

output.file output file name and path

#### **Details**

When performing dosage calling for multiple SNPs using Updog, the output file contains information for all loci and all samples. This function will convert the updog output file to a VCF file, while retaining the information for the values that are commonly used to filter low quality and low confident dosage calls.

#### Value

A vcf file

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# References

Updog R package

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