# Package 'PolyHaplotyper'

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Type Package

Polyploids
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Description Infer the genetic composition of individuals in terms of haplotype dosages for a haploblock, based on bi-allelic marker dosages, for any ploidy level.  Reference: Voorrips and Tumino: PolyHaplotyper: haplotyping in polyploids based on bi-allelic marker dosage data.  Submitted to BMC Bioinformatics (2021).
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# Description

Find all possible haplotypes for a haploblock from the haplotyping result

# Usage

```
allhap(hapresults, haploblock)
```

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## Arguments

hapresults list as returned by inferhaplotypes, or one element of such a list (i.e. the results

for one haploblock)

haploblock if hapresults is one element of the return value of inferHaplotypes, haploblock

should be missing of NULL; else haploblock is a single value indicating the

haploblock: either its name of its index in hapresults

#### **Details**

This function works with the results of inferHaplotypes; the setting of dropUnused does not affect this function

#### Value

an array with all possible haplotypes. The haplotypes are in columns, with the haplotype numbers as colnames; the markers are in rows.

## **Examples**

```
data(PolyHaplotyper_small)
# show the composition of all possible haplotypes with the markers
# in the first haploblock:
allhap(hapresults=phresults, haploblock=1)
```

allHaplotypes

get all haplotypes for the given markers

## **Description**

Given a set of bi-allelic (SNP) marker names, generate all possible haplotypes

#### Usage

```
allHaplotypes(mrknames)
```

## **Arguments**

mrknames

the names of the (bi-allelic) markers in the haploblock (contig)

## Value

a matrix with markers in columns and all possible (2 ^ nmrk) haplotypes in rows. 0: haplotype contains the non-dosage-counted marker allele (the reference allele); 1: haplotype contains the dosage-counted (alternative) marker allele. The colnames are the marker names.

```
# show the 8 possible haplotypes with 3 bi-allelic markers:
allHaplotypes(mrknames=c("mrkA", "mrkB", "mrkC"))
```

build\_ahccompletelist generate a list with all haplotype combinations

#### **Description**

generate a list which contains for each marker dosage combination at a given ploidy all matching haplotype combinations

#### Usage

build\_ahccompletelist(ploidy, maxmrk, savesec=1800, printsec=300, overwrite, shorten=FALSE)

#### **Arguments**

ploidy	ploidy; may be even or odd, but inferHaplotypes only works with even ploidy if FS families are present, so building an ahccompletelist for odd ploidy is probably not useful
maxmrk	the list will countain all haplotype combinations for haploblocks of 1maxmrk markers
savesec	default 1800: number of seconds between successive saves of the intermediate results
printsec	default 300: number of seconds between printout of the current set of haplotypes. NA or 0 suppresses printing.
overwrite	the new file ahccompletelist_ <ploidy>x.RData is written in the working directory. If a file with that name already exists and would be changed, this is aborted and the old file maintained if overwrite is FALSE</ploidy>
shorten	if file ahccompletelist_ <ploidy>x.RData already exists and covers haploblocks of lengths longer than maxmrk, this file is left unchanged if shorten is FALSE (default), but is shortened to maxmrk if shorten is TRUE</ploidy>

#### **Details**

An ahccompletelist reduces the processing time of inferHaplotypes enormously but takes a long time to build. This should therefore be done when PolyHaplotyper will be used multiple times. If an ahccompletelist file already exists in the working directory, this file is used as starting point; if maxmrk is larger than the length of the existing list only the additional items are calculated. If this function crashes (due to exceeding the memory limits of the computer or of R) the file ahccompletelist\_<ploidy>x.RData in the working directory contains an intact version of the ahccompletelist for the last finished marker count. Also, the temporary file "buildAHCcompletelist\_<datetime>"\_"<nmrk>.RData" contains a part of the list item for the number of markers being calculated at that moment.

Note that this function takes lots of time and memory already for ploidy 4 / maxmrk 7 and ploidy 6 / maxmrk 5, and is not practicable above ploidy 4 / maxmrk 8, ploidy 6 / maxmrk 6, ploidy 8 / maxmrk 5. Also, the files are large and take up considerable memory, and most of the size is used for the largest haploblock size. Therefore maxmrk should not be taken larger than necessary.

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## Value

NULL (invisible); the actual result is a file in the working directory named ahccompletelist\_<ploidy>x.RData

## **Examples**

```
# this example will create a file ahccompletelist_4x.RData
# in the working directory, for ploidy=4x and 1 - 4 markers:
build_ahccompletelist(ploidy=4, maxmrk=4, overwrite=FALSE)
```

calcMrkHaptable

produce a table of nr of markers vs nr of haplotypes

## **Description**

produce a table of nr of markers vs nr of haplotypes

#### Usage

```
calcMrkHaptable(ovwFS)
```

## **Arguments**

ovwFS

a list as produced by overviewFS. Only the first 2 columns of list item ovw are used

## Value

a frequency table with the numbers of haploblocks, with all combinations of marker counts and inferred haplotype counts per haploblock. The column with haplotype count NA (if any) shows the haploblocks for which no haplotype solution was found (the reason for that would usually be found in column 1 of ovwFS\$messages)

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calcStatistics calculate some statistics of the solutions of all haploblocks
------------------------------------------------------------------------------

#### **Description**

calculate some statistics of the solutions of all haploblocks

## Usage

```
calcStatistics(pedchk, ovwFS, indiv, haploblocks)
```

#### **Arguments**

pedchk a list as generated by pedigreeHapCheck

ovwFS a list as produced by overviewFS, or NA if no FS families available. If ovwFS

is specified, pedchk and ovwFS should have been calculated from the same un-

derlying data

indiv missing, NULL or a vector of names of individuals. If present, the component

pedstats is calculated only over the individuals in the vector, else over all indi-

viduals. Component FSstats is not affected by indiv

haploblocks missing, NULL or a vector of names or numbers of haploblocks, or a list of

haploblocks as in the inferHaplotypes input. If present, components pedstats and FSstats are calculated only over the specified haploblocks, else over all hap-

loblocks.

#### Value

a list with one or two items:

pedstats: a data.frame with one row per haploblock, showing numbers of individuals in columns

- mrk: individuals with complete marker data
- hap: individuals with an assigned haplotype combination
- match.NA: individuals where a match between the haplotypes of the individual and its parents could not be verified (either the individual itself or both parents have no haplotype combination assigned)
- noDR.TRUE: individuals whose haplotype combination matches that of its parents, assuming polysomic inheritance without Double Reduction
- noDR.FALSE: individuals whose haplotype combination doesn't match that of its parents, assuming polysomic inheritance without Double Reduction
- with DR. TRUE: individuals whose haplotype combination matches that of its parents, assuming polysomic inheritance, allowing Double Reduction
- with DR. FALSE: individuals whose haplotype combination doesn't match that of its parents, assuming polysomic inheritance, allowing Double Reduction

The total number of individuals is

match.NA + noDR.TRUE + noDR.FALSE == match.NA + withDR.TRUE + withDR.FALSE; the number of individuals with missing marker data or without assigned haplotype dosages can be calculated by subtracting mrk or hap from that total.

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FSstats (only if ovwFS specified): a data.frame with for each FS family, "rest" and "all" one row, with columns

- pop: the FS family numbers, "rest" (all individuals not belonging to the FSs or their parents) and "all" (all individuals)
- bothparmrk: for how many haploblocks have both parents complete marker data?
- FSdone: for how many haploblocks has a solution based on polysomic inheritance been found for this FS family?
- mrk: the average number of (FS) individuals with complete marker data
- imp: the average number of (FS) individuals with imputed marker data
- hap: the average number of (FS) individuals with an assigned haplotype combination

The numbers for "all" include also the FS parents (each parent counted only once, even if some parents produced more than one FS family), which are not included in any of the other rows; therefore "all" is not equal to the sum of the rows above.

## **Examples**

checkmrkDosage

check a marker dosages matrix or data.frame

## **Description**

check a marker dosages matrix or data frame, select columns and rows, convert to matrix

## Usage

```
checkmrkDosage(mrkDosage, ploidy, indiv=NULL, markers=NULL,
generateMarkernames=TRUE)
```

If NULL, all individuals are selected

## Arguments

mrkDosage	has a marker dosage. If mrkDosage is a matrix, the colnames are the individual names and the rownames are the marker names. If mrkDosage is a data.frame and the name of the first column starts with "marker" (upper/lowercase not relevant) the contents of this column are used as marker names, else the rownames are used as marker names. The (other) column names are the individual names.	
ploidy	All marker dosages must be in 0:ploidy or NA. an integer value; all dosages are checked to be in 0:ploidy or NA	
, ,		
indiv	NULL (default) or a character vector with names of individuals to be selected.	

markers

NULL (default) or a character vector with names of markers to be selected. If NULL, all markers are selected.

generateMarkernames

if TRUE (default) and mrkDosage has no markernames specified, markernames are generated automatically. Markernames must be either present or be generated, and none may be missing.

#### **Details**

This function is called by inferHaplotypes and by mergeReplicates, so normally there is no need for the user to call this function directly.

#### Value

a matrix with the selected columns in the order of indiv and the selected rows in order of markers (or all columns or rows in the original order if indiv or markers are not specified), with names of individuals as column names, marker names as row names

## **Examples**

compareHapMrkDosages compare haplotyping results with observed markers dosages

#### **Description**

compare haplotyping results with observed markers dosages

## Usage

```
compareHapMrkDosages(mrkDosage, hapresults)
```

#### Arguments

mrkDosage

a data.frame or matrix with the marker dosages, in the format of inferHaplotypes()

hapresults

a list as returned by inferHaplotypes, with one item (itself a list) per haploblock with at least a matrix hapdos and a character vector markers. All markers in all haploblocks must also occur in mrkDosages, and all individuals in the hapdos matrices must also occur in mrkDosages

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#### Value

a 3-D array with dimensions haploblock, individual, and chkresult: mrkNA (ALL markers in the haploblock have missing data for the individual), hapNA (the haplotype dosages do not sum to ploidy and/or are missing), match (TRUE if the non-missing marker dosages match the haplotype dosages, FALSE is there is a conflict, NA if mrkNA and/or hapNA are TRUE) Each element is itself a list with elements:

#### **Examples**

```
data(PolyHaplotyper_small)
chmd <- compareHapMrkDosages(mrkDosage=phdos, hapresults=phresults)
# show results for first haploblock, first 8 individuals:
chmd[1, 1:8,]</pre>
```

compareHapresults

compare two haplotyping results

#### **Description**

compare two haplotyping results, e.g. PolyHaplotyper and SATlotyper

#### Usage

compareHapresults(haploblock, hapresultsA, hapresultsB)

#### **Arguments**

haploblock a list of character vectors. The names are the names of the haploblocks, the

character vectors have the names of the markers in each haploblock.

hapresultsA and

hapresultsB two list as returned by inferHaplotypes, with one item (itself a list) per hap-

loblock with at least a matrix hapdos and a character vector markers. All haploblocks in param haploblock must occur in hapresultsA and in hapresultsB. The individual names (colnames of the hapdos items for each haploblock) must be

identical and in the same order in hapresultsA and hapresultsB

## Value

a list with one element per haploblock in param haploblock. Each element is itself a list with elements: \$identical: TRUE or FALSE

\$message: a single string, "" if the comparison is possible, else the reason why not (if \$message is not "", \$identical is always FALSE). The next elements are only present if \$message is "":

\$compindiv: a matrix comparing the two hapdos, with one column per individual and 5 rows: Both\_NA, A\_NA, B\_NA, Equal, Uneq. The last 2 have NA values if A and/or B is NA \$haplofreq: a matrix with one row per haplotype occurring in A and/or B, and columns A and B, with the total frequency of each haplotype in hapdos A or hapdos B

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## **Description**

A data.frame with a pedigree; used in vignette, stored in PolyHaplotyper\_demo.RData

#### **Format**

a data.frame with 661 rows (samples) and 4 columns: genotype, mother, father, sample\_nr

demo_snpdos	dosages of SNP alleles

## **Description**

A data.frame with the allele dosages for 30 SNPs in 661 samples; used in vignette, stored in Poly-Haplotyper\_demo.RData

## **Format**

a data.frame with 30 rows (SNPs) and 663 columns. The first two columns contain the SNP name and contig; the remaining columns contain the allele dosages (integers in 0..6 or NA) of the SNPs in 661 samples

expandHapdos	Add dropped rows back to haplotype dosage matrix	
expanunapu05	Add dropped rows back to haptotype dosage matrix	

## **Description**

Haplotype dosage matrices generated with the dropUnused=TRUE lack some haplotypes that are required by pedigreeHapCheck; this function adds them back.

## Usage

```
expandHapdos(hapdos, nhap, hbname)
```

## Arguments

hapdos	a haplotype dosage matrix as returned by a.o. inferHaplotypes. The rownames are assumed to have all the same length: they are composed of the haploblock
	name to which the 0-padded haplotype number is appended, separated by "_"
nhap	the total number of haplotypes for the haploblock: $nhap = 2^n mrk$ with $nmrk$ the number of markers in the haploblock
hbname	the haploblock name (only used when hapdos has 0 rows)

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#### Value

a matrix similar to hapdos with rows for the dropped haplotypes re-inserted, with the correct rownames and containing only 0 and NA values

## **Examples**

getFSfreqs

get all FS haplotype combinations expected from two parental haplotype combinations, with their frequencies

#### **Description**

get all FS haplotype combinations expected from two parental haplotype combinations, with their frequencies. Different from getFScombs in that it returns the unique FS combinations as well as their frequencies, and that it also considers Double Reduction

#### Usage

```
getFSfreqs(parhac, DRrate)
```

#### Arguments

parhac matrix with one column for each parent and <ploidy> rows, giving the haplotype

combinations for each parent

DRrate the rate of double reduction per meiosis (NOT per allele!); e.g. with a DRrate

of 0.04, a tetraploid parent with genotype ABCD will produce a fraction of 0.04 of DR gametes AA, BB, CC and DD (each with a frequency of 0.01), and a fraction of 0.96 of the non-DR gametes AB, AC, AD, BC, BD, CD (each with a

frequency of 0.16)

#### Value

a list of 2 elements:

FShac: a matrix with one column per unique FS haplotype combination and the same row count as parhac, giving the FS haplotype combinations. There are no duplicated columns but several (not necessarily adjacent) columns may correspond to the same mrkdid

freq: a vector of length ncol(hapcomb), with for each FS haplotype combination in hapcomb its frequency (0.0 ... 1.0). The colnames are NULL: haplotype combinations are not named.

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#### **Examples**

```
# specify combinations of haplotypes in two tetraploid parents:
hapcomb <- matrix(c(2,2,5,6, 1,2,5,5), ncol=2)
# FS frequencies without double reduction:
getFSfreqs(parhac=hapcomb, DRrate=0)
# FS frequencies with 5\% double reduction:
getFSfreqs(parhac=hapcomb, DRrate=0.05)</pre>
```

getGameteFreqs

get all gametes and their frequencies for a parental haplotype combi-

nation

## **Description**

get all gametes and their frequencies for a parental haplotype combination

## Usage

getGameteFreqs(parhac, DRrate)

#### **Arguments**

parhac vector of the parental haplotype combination of length <ploy>, giving the

<ploidy> haplotype numbers present per haplotype giving the dosage of that

haplotype, summing to ploidy

DRrate the rate of double reduction per meiosis (NOT per allele!); e.g. with a DRrate

of 0.04, a tetraploid parent with genotype ABCD will produce a fraction of 0.04 of DR gametes AA, BB, CC and DD (each with a frequency of 0.01), and a fraction of 0.96 of the non-DR gametes AB, AC, AD, BC, BD, CD (each with a

frequency of 0.16)

## **Details**

for hexaploids the DR gametes consist of a duplication of one of the 6 parental alleles, combined with one copy of one of the other 5 alleles.

Calculation is faster if DRrate is 0.0

## Value

a list of 2 elements:

hapcomb: a matrix with one column per unique gamete and ploidy/2 rows. Each element is the number (ID) of a haplotype). Within columns the haplotypes are sorted from low to high; the columns are ordered from left to right, first on row 1, then on row 2 etc

freq: a vector of length ncol(hapcomb), with for each gamete in hapcomb its frequency (0.0 ... 1.0)

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## **Examples**

```
# specify combination of haplotypes in a tetraploid parent:
hapcomb <- c(2,2,5,6) # 2 copies of haplotype 2, 1 each of 5 and 6
# gamete frequencies without double reduction:
getGameteFreqs(parhac=hapcomb, DRrate=0)
# gamete frequencies with 5\% double reduction:
getGameteFreqs(parhac=hapcomb, DRrate=0.05)</pre>
```

hapcomb2hapdos

convert haplotype combinations to haplotype dosages

## **Description**

converts matrices that contain haplotype combinations in columns (as the matrices in ahccompletelist or ahclist) to matrices with the haplotype dosages

## Usage

hapcomb2hapdos(hapcomb, nhap)

#### **Arguments**

hapcomb matrix with <ploidy> rows and any number of columns. Each column contains a

set of haplotypes (numbers in 1 ... nhap). NAs allowed but no values outside this range (not checked). E.g. a column with numbers 1-1-3-12 means two copies of

haplotype 1 and one of haplotypes 3 and 12 each.

nhap the total number of possible haplotypes: 2^nmrk where nmrk is the number of

bi-allelic markers in the haploblock

#### Value

matrix with nhap rows (one row for each possible haplotype) and as many columns as in hapmat, giving the dosages of each haplotype in each column of hapmat.

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han	dos2ha	ncomb
παρ	UUSZIIC	ahcoilin

convert haplotype dosages to haplotype combinations

#### **Description**

convert a vector or matrix of haplotype dosages to a vector or matrix of haplotype combinations

#### Usage

```
hapdos2hapcomb(hapdos, ploidy)
```

## **Arguments**

hapdos a vector with one item for some or all possible haplotypes, with the dosage of

the haplotypes, summing to ploidy; or a matrix where each column is such a vector, each representing an individual. Missing data are allowed (but any NA makes the entire vector or matrix column unknown); if a matrix, 0 rows indicate missing values for all individuals. When hapdos is a matrix, the haplotype numbers must be given as rownames; when a vector, as names. Not all possible haplotype numbers need to be present but the ones that are present must be in

ascending order, no duplicates or missing values allowed.

ploidy the ploidy (one number)

#### Value

a sorted vector of length ploidy with all haplotype numbers present, or a matrix where each column is such a vector, with the same colnames as hapdos

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hapdos2mrkdos	calculate the marker dosages resulting from haplotype dosage combinations

#### **Description**

calculate the marker dosages resulting from haplotype dosage combinations

#### Usage

```
hapdos2mrkdos(hapdos, allhap)
```

## **Arguments**

hapdos a matrix with one column per combination of haplotypes and one row for each

possible haplotype (corresponding to the rows of allhap) with dosage of the haplotypes in each combination. A vector is interpreted as a one-column matrix; all columns must sum to ploidy. The rownames of the matrix (or names of the

vector) must contain the haplotype numbers

allhap a matrix as returned by allHaplotypes

## **Details**

if hapdos contains NA values, all values in the corresponding column of the result will also be NA

## Value

a matrix with columns corresponding to the columns of hapdos and one row for each marker, with the dosages of each marker in each combination; colnames are the mrkdids (marker dosage IDs), rownames are the marker names taken from allhap

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haploblock\_df2list

convert a haploblock-defining data frame to a list

#### **Description**

convert a haploblock-defining data frame to a list as needed by inferHaplotypes

## Usage

```
haploblock_df2list(df, mrkcol, hbcol, sorted=TRUE)
```

## **Arguments**

a data.frame with at least the following two columns

mrkcol the name or number of the column with the markers

the name or number of the column with the haploblocks

sorted if TRUE (default) the haploblock list is sorted in alphabetical order of haploblock name (the markers within haploblocks are not sorted); if FALSE the

haploblocks will be in order of first occurrence in df

## Details

function inferHaplotypes needs a list where each item is a vector of all markers in one haploblock. This function produces such a list from a data.frame where the markers are in one column and the haploblocks in another column. The markers and haploblocks columns may be character, factor or numeric, and the columns may be indicated by name or number.

#### Value

the desired list

```
df1 <- data.frame(
  marker=paste0("mrk",1:9),
  block=LETTERS[c(1,2,3,2,3,1,1,2,2)],
  extracol=runif(9))
haploblock_df2list(df=df1, mrkcol="marker", hbcol=2)</pre>
```

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# Description

infer haplotypes for one or more haploblocks, for all individuals, using FS family(s) (with parents) if present, and infer haplotypes for non-FS material as well

## Usage

```
inferHaplotypes(mrkDosage, indiv=NULL, ploidy, haploblock,
parents=NULL, FS=NULL, minfrac=c(0.1, 0.01), errfrac=0.025, DRrate=0.025,
maxmrk=0, dropUnused=TRUE, maxparcombs=150000, minPseg=1e-8,
knownHap=integer(0), progress=TRUE, printtimes=FALSE, ahcdir)
```

# Arguments

mrkDosage	matrix or data.frame. Markers are in rows, individuals in columns, each cell has a marker dosage. Names of individuals are the column names, marker names are the row names or (if a data.frame) in a column named MarkerNames. All marker dosages must be in 0:ploidy or NA.
indiv	NULL (default) or a character vector with names of all individuals to be considered. If NULL, all columns of mrkDosage are selected.  All indivs that are not in parents or FS vectors (see below) are considered unrelated, i.e. we have no implementation for pedigrees (yet).
ploidy	all marker dosages should be in 0:ploidy or NA
haploblock	a list of character vectors. The names are the names of the haploblocks, the character vectors have the names of the markers in each haploblock. Haplotype names are constructed from the haploblock names, which are used as prefixes to which the (zero-padded) haplotype numbers are are appended with separator '.'.
parents	a matrix with one row for each FS family and two columns for the two parents, containing the names of the female and male parent of each family.
FS	a list of character vectors. Each character vector has the names of the individuals of one FS family. The items of the list should correspond to the rows of the parents matrix, in the same order.
minfrac	vector of two fractions, default 0.1 and 0.01. A haplotype is considered to be certainly present if it must occur in at least a fraction minfrac[1] of all individuals; in the final stage for the "other" individuals (those that do not belong to the FS or its parents) this fraction is lowered to minfrac[2]; see also inferHaps_noFS
errfrac	default 0.025. The assumed fraction marker genotypes with an error (over all markers in the haploblock). The errors are assumed to be uniformly distributed over all except the original marker dosage combinations (mrkdids)

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DRrate default 0.025. The rate of double reduction per meiosis (NOT per allele!); e.g. with a DRrate of 0.04, a tetraploid parent with genotype ABCD will produce a fraction of 0.04 of DR gametes AA, BB, CC and DD (each with a frequency of 0.01), and a fraction of 0.96 of the non-DR gametes AB, AC, AD, BC, BD, CD

(each with a frequency of 0.16)

maxmrk Haploblocks with more than maxmrk markers will be skipped. Default 0: no

haploblocks are skipped

dropUnused TRUE (default) if the returned matrix should only contain rows for haplotypes

that are present; if FALSE matrix contains rows for all possible haplotypes

maxparcombs Parent 1 and 2 both may have multiple possible haplotype combinations. For

each pair of haplotype combinations (one from P1 and one from P2) the expected FS segregation must be checked against the observed. This may take a long time if many such combinations need to be checked. This parameter sets a limit to the number of allowed combinations per haploblock; default 150000 takes about

45 min.

minPseg default 1e-8. The minimum P-value of a chisquared test for segregation in FS

families. The best solution for an FS family is selected based on a combination of P-value and number of required haplotypes, among all candidate solutions with a P-value of at least minPseg. If no such solution is found the FS and its

parents are treated as unrelated material

knownHap integer vector with haplotype numbers (haplotypes that must be present accord-

ing to prior inference or knowledge, numbers refer to rows of matrix produced

by allHaplotypes); default integer(0), i.e. no known haplotypes

progress if TRUE, and new haplotype combinations need to be calculated, and the num-

ber of markers and the ploidy are both >= 6, progress is indicated by printed

messages

printtimes if TRUE, the time needed to process each haploblock is printed

ahcdir a single directory, or not specified. inferHaplotypes uses lists that for each com-

bination of marker dosages give all possible combinations of haplotype dosages. These lists (ahclist and ahccompletelist) are loaded and saved at the directory specified by ahcdir. If no ahcdir is specified it is set to the current working di-

rectory.

If an ahclist or ahccompletelist for the correct ploidy is already in GlobalEnv

this is used and no new list is loaded, even if ahcdir is specified.

## **Details**

First we consider the case where one or more FS families and their parents are present in the set of samples. In that case, initially the possible haplotype configurations of the parents are determined. From that, all their possible gametes (assuming polysomic inheritance) are calculated and all possible FS haplotype configurations. Comparing this with the observed FS marker dosages the most likely parental and FS configurations are found.

It is possible that multiple parental combinations can explain the observed marker dosages in the FS. In that case, if one is clearly more likely and/or needs less haplotypes, that one is chosen. If there is no clear best solution still the parents and FS individuals that have the same haplotype configuration over all likely solutions are assigned that configuration.

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For FS where no good solution is found (because of an error in the marker dosages of a parent, or because the correct solution was not considered) the parents and individuals will be considered as unrelated material.

If several FS families share common parents they are treated as a group, and only solutions are considered that are acceptable for all families in the group.

Finally (or if no FS families are present, immediately) the other samples are haplotyped, which are considered as unrelated material. If FS families have been solved the haplotypes in their parents are considered "known", and known haplotypes can also be supplied (parameter knownHap). For these samples we consecutively add haplotypes that must be present in a minimum number of individuals, always trying to minimize the number of needed haplotypes.

InferHaplotypes uses tables that, for each combination of dosages of the markers in the haploblock, list all haplotype combinations (ahc) that result in these marker dosages. In principle inferHaplotypes uses a list (ahccompletelist) that, for a given ploidy, has all the haplotype combinations for haploblocks from 1 up to some maximum number of markers. This list can be computed with function build\_ahccompletelist. If this list is not available (or is some haploblocks contain more markers than the list), the ahc for the (extra) marker.

See the PolyHaplotyper vignette for an illustrated explanation.

#### Value

a list with for each haploblock one item that itself is a list with items:

message; if this is "" the haploblock is processed and further elements are present; else this message says why the haploblock was skipped (currently only if it contains too many markers)

hapdos: a matrix with the dosages of each haplotype (in rows) for each individual (in columns). For each individual the haplotype dosages sum to the ploidy. If dropUnused is TRUE Only the haplotypes that occur in the population are shown, else all haplotypes

mrkdids: a vector of the mrkdid (marker dosage ID) for each individual (each combination of marker dosages has its own ID; if any of the markers has an NA dosage the corresponding mrkdid is also NA).

The mrkdids can be converted to the marker dosages with function mrkdid2mrkdos.

markers: a vector with the names of the markers in the haploblock

imputedGeno: a matrix in the same format as param mrkDosage, with one row for each marker in the haploblock and one column per imputed individual, with the dosages of the markers. These are the individuals that have incomplete data in mrkDosage but where the available marker dosages match only one of the expected marker genotypes in the FS family (only individuals in FS families are imputed). It is possible that an individual with imputed marker dosages is not haplotyped (as is the case for individuals with complete marker data) if the marker dosages match different possible haplotype combinations. The next elements are only present if one or more FS families were specified:

FSfit: a logical vector with one element per FS family; TRUE if a (or more than one) acceptable solution for the FS is found (although if multiple solution are found they might not be used if unclear which one is the best solution). (Even if no solution was found for an FS, still its individuals may have a haplotype combination assigned ignoring their pedigree)

FSmessages: a character vector with one item per FS family: any message relating to the fitting of a model for that FS, not necessarily an error

FSpval: a vector of the chi-squared P-value associated with the selected FS model for each FS family, or the maximum P value over all models in case none was selected

If for new combinations of marker dosages the possible haplotype combinations have to be calculated, an ahclist file is written to ahcdir

20 make.Happyinf.input

#### **Examples**

```
# this example takes about 1 minute to run:
data(PolyHaplotyper_small)
results <- inferHaplotypes(mrkDosage=phdos, ploidy=6,
haploblock=phblocks, parents=phpar, FS=phFS)
names(results)
names(results[[1]])</pre>
```

make.Happyinf.input

convert PolyHaplotyper input data to Happy-inf format

#### **Description**

convert PolyHaplotyper input data to Happy-inf format for a single haploblock

## Usage

```
make.Happyinf.input(mrkDosage, indiv=NULL, haploblock,
ploidy, fname)
```

## **Arguments**

mrkDosage	matrix or data.frame of allele dosages; same as input for inferHaplotypes. Markers are in rows, individuals in columns, each cell has a marker dosage. All marker dosages must be in 0:ploidy or NA
indiv	the names of the individuals to include in the Happy-inf input data. Default NULL includes all individuals
haploblock	a list of character vectors. The names are the names of the haploblocks, the character vectors have the names of the markers in each haploblock. Only the markers in haploblock will be included in the Happy-inf input data

ploidy single integer: the ploidy level

fname filename of a tab-separated output file: this will contain the data in Happy-inf

format (the saved data.frame is also the return value). If "" no file is written

## Value

a data.frame in the Happy-inf input format: a header row with "SNPID", "block" and names of the individuals and one row per marker, with only the individuals, markers and blocks as specified. #'SNPID has the marker names, "Block" the haploblock names. All markers of a haploblock are in contiguous rows. Missing dosages are represented by "NA"

make.SATlotyper.input convert PolyHaplotyper marker data to SATlotyper format

## **Description**

convert PolyHaplotyper marker data (input) to SATlotyper format for a single haploblock.

## Usage

```
make.SATlotyper.input(mrkDosage, indiv=NULL, markers,
ploidy, phenotype=0, fname)
```

## **Arguments**

mrkDosage	matrix or data.frame of allele dosages; same as input for inferHaplotypes. Markers are in rows, individuals in columns, each cell has a marker dosage. All marker dosages must be in 0:ploidy or NA
indiv	the names of the individuals to include in the ShesisPlus input data. Default NULL includes all individuals
markers	character vector with the names of the markers in the haploblock; all must occur in mrkDosage
ploidy	single integer: the ploidy level
phenotype	vector with the phenotypes of all individuals, in order of the columns of dosmat; default $\boldsymbol{0}$
fname	filename of a *.csv output file: this will contain the data in the SATlotyper format (the saved data.frame is also the return value). If "" no file is written

## Value

a data.frame in the SATlotyper input format: a header row with "Genotype" and the marker names, and one row per individual with the individual name plus for each marker the genotype as a sorted string of <ploidy> A's and B's, or <ploidy> N's

make. ShesisPlus.input convert PolyHaplotyper marker data to ShesisPlus format

## **Description**

convert PolyHaplotyper marker data (input) to ShesisPlus format for a single haploblock.

## Usage

```
make.ShesisPlus.input(mrkDosage, indiv=NULL, markers,
ploidy, phenotype=0, fname="")
```

## Arguments

mrkDosage	matrix or data.frame of allele dosages; same as input for inferHaplotypes. Markers are in rows, individuals in columns, each cell has a marker dosage. All marker dosages must be in 0:ploidy or NA
indiv	the names of the individuals to include in the ShesisPlus input data. Default NULL includes all individuals
markers	character vector with the names of the markers in the haploblock; all must occur in mrkDosage
ploidy	single integer: the ploidy level
phenotype	vector with the phenotypes of all individuals, in order of the columns of dosmat; default $\boldsymbol{0}$
fname	filename of the output file: this will contain the data in the ShesisPlus format (the saved data,frame is also the return value). If "" (default) no file is written

## **Details**

ShesisPlus needs the data formatted as: 1 column with names of individuals, 1 column with the phenotypes (with missing values represented as "NA"), and for each of the selected markers <ploidy> columns, with the (unphased) marker alleles. Here we use only biallelic markers; their alleles are indicated by 1 and 2 for the ref and alt allele, and 0 for missing data.

The contents of file fname can be pasted into the input data box of the ShesisPlus web interface at http://shesisplus.bio-x.cn/SHEsis.html

#### Value

a data.frame in the described ShesisPlus input format

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mergeReplicates	
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merge replicate samples in dosage matrix

#### Description

merge replicate samples in dosage matrix

## Usage

```
mergeReplicates(mrkDosage, replist, solveConflicts=TRUE)
```

## **Arguments**

mrkDosage a dosage matrix with markers in rows and individuals in columns. row names

are marker names, column names are individual names.

replist a list of character vectors, each of which has the sample names of a set of repli-

cates

solveConflicts if TRUE (default) and there are conflicting dosage assignments between repli-

cates for the same marker, the one with highest frequency is used, provided the total freq of other dosages =  $1 \text{ OR} \le 10\%$  of the frequency of the most frequent dosage. If solveConflicts is FALSE and there are conflicting dosages, the

consensus for that marker will be NA

## **Details**

This function merges all sets of replicates, each to one column. The column name of the one retained column is the first one for that set in replist.

For each set of replicates it calls getConsensusmrkDosage

#### Value

a version of mrkDosage in which only one column of each set of replicates is retained; this column (the first in its set as specified in replist) now has the consensus scores over all replicates. Also, if mrkDosage was a data.frame, it is converted into a matrix.

24 mrkdid2mrkdos

```
mergeReplicates(mrkDosage=dosmat, replist=reps)
# introduce a conflicting dosage:
dosmat[3,2] <- 2
# merge:
mergeReplicates(mrkDosage=dosmat, replist=reps)</pre>
```

mrkdid2mrkdos

get the marker dosages from mrkdids (marker dosage IDs)

## **Description**

get the marker dosages from mrkdids (marker dosage IDs)

#### Usage

```
mrkdid2mrkdos(dosageIDs, nmrk, ploidy, mrknames=NULL)
```

## **Arguments**

dosageIDs vector of marker-dosage-combination IDs (mrkdid)
nmrk character vector of marker names in the haploblock

ploidy the ploidy level, a single positive integer

mrknames a vector of nmrk marker names (default NULL): if not NULL these are used as

rownames of the returned matrix

#### Value

a matrix with in columns the marker dosages corresponding to the marker dosageIDs, with these mrkdids as colnames, and one row per marker, with marker names as rownames if mrknames are specified

mrkdos2mrkdid 25

mrkdos2mrkdid get marker dosage IDs from marker dosages	mrkdos2mrkdid	get marker dosage IDs from marker dosages
---------------------------------------------------------	---------------	-------------------------------------------

## **Description**

get marker dosage IDs (mrkdid) from marker dosages

## Usage

```
mrkdos2mrkdid(mrkDosage, indiv=NULL, ploidy, check=TRUE)
```

#### **Arguments**

mrkDosage	matrix or data.frame. Markers are in rows, individuals in columns, each cell has a marker dosage. Names of individuals are the column names, marker names are the row names or (if a data.frame) in a column named MarkerNames. All marker dosages must be in 0:ploidy or NA. If a data.frame, additional columns may be present.
indiv	NULL (default) or a character vector with names of individuals to be selected. If NULL, all columns are selected; if mrkDosage is a data.frame, that is probably not what is intended.
ploidy	all marker dosages are checked to be in 0:ploidy or NA
check	if TRUE (default) checkmrkDosage is called. If FALSE it is assumed that mrk-Dosage is a matrix (not a data.frame) and it is not checked.

#### **Details**

with ploidy==1 and (of course) all dosages 0 or 1 this function returns the haplotype numbers for the haplotype specified by each column

#### Value

a vector of marker dosage IDs, one for each column of mrkDosage: each a number in 1:((ploidy+1)^nrow(mrkDosage)), NA for each column in dosages where any of the dosages are NA

26 overviewByFS

#### **Description**

generate an overview of the results by haploblock and by FS family

## Usage

```
overviewByFS(haploblock, parents, FS, hapresults)
```

#### **Arguments**

haploblock a list of character vectors. The names are the names of the haploblocks, the

character vectors have the names of the markers in each haploblock.

parents a matrix of two colums and one row per FS, containing the names of the two

parents of each FS

FS a list of character vectors with the names of the samples for each FS

hapresults a list as returned by inferHaplotypes, with one item per haploblock, containing

at least all those in the list specified by haploblock

#### Value

- a list with two items:
- ovw: an integer matrix with one row per haploblock and the following columns:
- \* nmrk: the number of markers in the haploblock
- \* nhap: the number of different haplotypes assigned over all individuals (NA if no solution was found for this haplotype; the reason for that is listed in the first column of item messages of the return value)
- \* for each FS family a set of 6 columns:
- + parmrk (0, 1 or 2: the number of parents with complete marker data)
- + fit (0=FALSE or 1=TRUE), indicating if a solution for the FS was found based on polysomic inheritance)
- + mrk: the number of FS progeny with complete marker data
- + imp: the number of FS progeny where complete marker data were imputed
- + hap: the number of FS progeny with assigned haplotype combinations. hap will be less than the mrk value if the same FS marker genotype can be produced with different combinations of haplotypes that are all compatible with the parental haplotype combinations) or if some FS marker genotypes cannot be produced by the inferred parental haplotype combinations
- + P: the chi-squared P-value of the best fitting solution, even if this is discarded because of lack of fit).
- \* For "rest" (all individuals that are not part of the FS's or their parents) and "all" (all individuals) there are also columns mrk and hap, and for "all" there is also a column imp, similar to those for the FS families. The numbers for "all" are the sums of those for the FS families, the FS parents (some FS may share a parent but shared parents are counted only one) and the "rest".
- messages: a character matrix with one row per haploblock and the following columns:

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\* haploblock: the reason why there is no solution for the haploblock ("" if there is a solution) \* one column for each FS family with a possible message or "". A message can indicate a failure to find a solution for the FS family but may also describe less significant problems, such as some progeny with unexpected marker dosages etc.

# Examples

padded

pad an integer (prefix with zeroes to a fixed length)

## **Description**

pad an integer (prefix with zeroes to a fixed length)

## Usage

```
padded(x, maxx=0)
```

# Arguments

x vector of non-negative integers

maxx a single integer to whose nchar all x elements will be padded; if 0 (default) the

largest value in x will be used

#### Value

a character vector representing the values of x left-padded with 0's to the length of integer maxx or of max(x)

```
padded(c(21, 1, 121, NA, 0))
padded(c(21, 1, 121, NA, 0), maxx=1000)
```

28 pedigreeHapCheck

pedigreeHapCheck	check the consistency of haploblock genotypes over a pedigree

## **Description**

For all haploblocks, check whether the inheritance of inferred haplotypes over the pedigree is consistent without or with allowing for double reduction (DR), and assuming polysomic inheritance.

## Usage

pedigreeHapCheck(ped, mrkDosage, haploblock, hapresults)

#### **Arguments**

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ped	data.frame or matrix listing the pedigree. Column 1 has names of all individuals (no duplicates, no NA), column 2 and 3 have the parents (duplicates and NA allowed, also individuals with one known parent or with two identical parents allowed). No sorting of the pedigree is needed. All parents should also be listed as individuals; if that is not the case the missing parents will be added as founders and a warning will be issued.
mrkDosage	a matrix with one row for each marker and one column for each individual, with the observed dosages. Individuals with one or more NA dosages are considered to have no marker data. Individual names are the colnames, no duplicates allowed; the set of individuals needs not be the same as those in ped; individual names common to mrkDosage and ped must be exactly identical (upper/lower case and whitespace included)
haploblock	a list of character vectors. The names are the names of the haploblocks, the character vectors have the names of the markers in each haploblock
hapresults	a list as returned by inferHaplotypes, with one item per haploblock, containing at least all those in the list specified by haploblock

#### Value

- a list with two items:
- ped\_arr: a 3D logical array with dimensions individuals, diagnostics and haploblocks. For each individual and each haploblock there are 4 diagnostics:
- \* mrk: does the individual have complete marker dosage data?
- \* imp: were the marker dosages for this individual imputed?
- \* hap: is there a haplotype combination assigned to the individual?
- \* noDR: does the haplotype genotype of the individual match that of its parents, assuming polysomic inheritance but no Double Reduction? NA if the individual or both its parents do not have a haplotype genotype assigned
- \* withDR: as noDR, but allowing Double Reduction
- parents\_arr: a 3D integer array with dimensions parents (all individuals that occur as parents in the pedigree), diagnostics and haploblocks. For each parent and each haploblock there are 7 diagnostics:

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- \* par\_mrkdata: 0=FALSE, 1=TRUE, does this parent have complete marker data?
- \* par\_hapdata: 0=FALSE, 1=TRUE, does this parent have a haplotype genotype assigned?
- \* totprogeny: how many first-generation progeny (children) does this parent have (combined over all its matings, both as mother and as father)
- \* prog\_mrkdata: how many progeny have complete marker dosage data?
- \* prog\_hapdata: how many progeny have a haplotype combination assigned?
- \* nonDRmatch: how many progeny have a haplotype combination that is compatible with their parent's haplotype combinations, assuming polysomic inheritance but no Double Reduction
- \* DRmatch: as nonDRmatch, but also allowing DR

Both ped\_arr and parents\_arr contain all haploblocks in haploblock, also those skipped because of too many markers and those without any haplotyped individuals. These can be excluded by excluding them from the haploblock list.

## **Examples**

pedigreeSim2PH

convert the PedigreeSim true haplotypes to marker dosages and haplotype dosages

#### Description

convert the PedigreeSim true haplotypes to marker dosages and haplotype dosages

#### Usage

```
pedigreeSim2PH(ps_geno, haploblock, indiv=NULL, dropUnused=TRUE)
```

## Arguments

ps\_geno

the filename of a \*\_genotypes.dat file as produced by PedigreeSim, or a data.frame read from such a file (with a column marker, followed by <ploidy> columns per individual with the marker alleles for each homolog; the names of these columns must be <indivname>\_<homolog number>). ps\_geno should not contain missing data (this will be true unless the PedigreeSim output is midified)

haploblock

a list of character vectors. The names are the names of the haploblocks, the character vectors have the names of the markers in each haploblock. Haplotype names are constructed from the haploblock names, which are used as prefixes to which the (zero-padded) haplotype numbers are are appended with separator

All markers in haploblock must be present in ps\_geno. If haploblock is NULL only the marker dosages are generated, not the haplotype dosages.

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indiv the names of the individuals to be extracted; default NULL: all individuals in

ps\_geno. If not NULL, all indiv must be present in ps\_geno

dropUnused TRUE (default) if the returned matrix should only contain rows for haplotypes

that are present; if FALSE matrix contains rows for all possible haplotypes

#### **Details**

if all alleles are in 0/1, these are kept. If only 2 allele symbols occur in ps\_geno all are converted (alphabetically) to 0/1 in the same way (e.g. if the alleles are A and B, A -> 0 and B -> 1, even in markers with only B's). If different allele symbols are used between markers, per marker the (alphabetically or numerically) lowest -> 0 and the highest -> 1. So if more than two different allele symbols occur in ps\_geno, and one marker has only A and another only B alleles, both are converted to 0's.

in mrkDosage, the dosage of the alleles (converted to) 1 is reported

#### Value

a list with 2 items:

\$mrkDosage: a matrix of marker dosages in the input format of inferHaplotypes, with all markers that occur in haploblock, sorted according to haploblock, and for the selected indiv in the specified order

\$haplist: (only if haploblock is not NULL) a list with one element for each haploblock, similar to the inferHaplotypes output). Each element is itself a list, with two components:

\$hapdos is a matrix with the haplotype dosages for that haploblock for each individual

\$markers: a vector with the names of the markers in the haploblock in the output of inferHaplotypes

## Description

A list with for each haploblock the names of the markers it contains; used in manuals, stored in PolyHaplotyper\_small.RData

#### **Format**

a list with 2 character vectors

phdos 31

phdos dosages of SNP alleles

## **Description**

A matrix with the allele dosages for 8 SNPs in 661 samples; used in manuals, stored in PolyHaplotyper\_small.RData

#### **Format**

a data.frame with 30 rows (SNPs) and 663 columns. The first two columns contain the SNP name and contig; the remaining columns contain the allele dosages (integers in 0..6 or NA) of the SNPs in 661 samples

phFS

members of FS families

## **Description**

A list with for each FS family the names of the individuals it contains; used in manuals, stored in PolyHaplotyper\_small.RData

#### **Format**

a list with 4 vectors, each with the (here numeric) names of the FS members

phpar parents of FS families

# Description

A matrix with the names of the parents of each FS family; used in manuals, stored in PolyHaplo-typer\_small.RData

## **Format**

a matrix with 4 rows, 1 per FS family, and 2 columns, 1 for each parent

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## Description

A data.frame with a pedigree; used in manuals, stored in PolyHaplotyper\_small.RData

#### **Format**

a data.frame with 661 rows (samples) and 4 columns: genotype, mother, father, sample\_nr

|--|

## **Description**

A list with the results of function inferHaplotypes; used in manuals, stored in PolyHaplotyper\_small.RData

#### **Format**

a list with 2 elements, one per haploblock; each element itself a list with multiple components

```
read.Happyinf.output read the haplotyping results from the Happy-inf output
```

## **Description**

read the haplotyping results from the Happy-inf output

## Usage

```
read.Happyinf.output(file_prefix, dropUnused=TRUE)
```

# Arguments

file_prefix	ne output files generat		

eter)

dropUnused TRUE (default) if the returned matrix should only contain rows for haplotypes

that are present; if FALSE matrix contains rows for all possible haplotypes

#### **Details**

This function reads the <file\_prefix>.stat.dat and <file\_prefix>.stats.dat files. The first contains one row per SNP marker, grouped per haploblock and <ploidy> columns per individual with the SNP haplotypes, where SNP alleles are represented as 0 or 1, or NA for unknown. For one SNP, the <ploidy> alleles in an individual are all known or all unknown It is possible that of the different SNPs in a haploblock some are known and some are not; in the conversion to PolyHaplotyper format all these partially known haplotypes are made unknown. The latter file has a statistics "mismatch" and "ratio" for each individual / haploblock combination; for their meaning see the Happyinf readme file.

#### Value

a list with 2 items:

\$hapdos is a matrix with individuals in columns and haplotypes in rows, giving the dosages of the haplotypes in each individual (summing to ploidy). This is the same format as the hapdos components of the inferHaplotypes results of PolyHaplotyper except that

\$stats is a matrix with individuals in rows (matching dosmat) and two rows named mismatch and ratio.

read.SATlotyper.output

read the haplotyping results from the SATlotyper output

## Description

read the haplotyping results from the SATlotyper output

## Usage

```
read.SATlotyper.output(fname, output="", allelecodes=c("A", "B"),
sep, haploblockname="")
```

#### **Arguments**

fname	filename of an xml file produced by SATlotyper
output	character vector, console output of SATlotyper; this contains a table of calculated haplotypes with more info than the xml file
allelecodes	the codes used for the ref and alt SNP alleles in make. SATlotyper.input, default "A" and "B" $$
sep	the separator used in make.SATlotyper.input (SATlotyper cleverly identifies this separator and uses it for its output), default tab
haploblockname	if not "" (default) the haplotype IDs are given as <haploblockname>_<hapnr>, else just as <hapnr>, left_padded with zeroes</hapnr></hapnr></haploblockname>

#### **Details**

The xml file is parsed using the package XML. The resulting list has 3 items named source, bootstrapping and haplotyping. In this function source is ignored and the results are obtained from haplotypings[[1]] (i.e. even if multiple haplotypings were done only the first is extracted), with the haplotype score from the bootstrapping item added to the haplotype.info.

#### Value

a list with 3 items:

\$hapdos is a matrix with individuals in columns and haplotypes in rows, giving the dosages of the haplotypes in each individual (summing to ploidy). This is the same format as the hapdos components of the inferHaplotypes results of PolyHaplotyper

\$haplotype.info is a data.frame that is a combination of information from two or three tables: columns Haplotype and necessity are from element haplotypings[[1]]\$haplotypes in the xml file, column hapnr gives the PolyHaplotyper equivalents of the Haplotypes,

column score is from element bootstrapping in the xml file,

columns id, number, frequency, homozygous, necessary, distance and neighbours are from (the console) output if present.

The haplotype.info data.frame is ordered by hapnr.

\$HaplotypingScore is the single number in \$haplotyping[[1]]\$score.

read.ShesisPlus.output

Read the haplotyping results from the ShesisPlus output

#### **Description**

Read the haplotyping results from the ShesisPlus output

## Usage

```
read.ShesisPlus.output(SSPout, order.by=c("", "hapnr", "count")[2])
```

#### **Arguments**

SSPout filename of a text file with ShesisPlus output (as copied from the web page pro-

duced by running ShesisPlus web interface at http://shesisplus.bio-x.cn/SHEsis.html);

or a character vector with the same output

order.by how to order the rows of the hapstat data.frame. "hapnr" means ordering by

haplotype number, "count" means ordering by decreasing Total.count, anything

else results in no reordering. By default order by hapnr.

## Details

If present, the markernames and the haplotype statistics are read from the file. ShesisPlus does not provide haplotype combinations for individuals

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#### Value

a list with 2 items: \$markernames has the marker names in the markers in the haploblock (if present in the file); hapstat contains the haplotype statistics as read from the file with an additional (first) column hapnr: the haplotype numbers as defined in PolyHaplotyper. The other columns are Haplotype (as sequences of marker alleles), Total.count (of the haplotype over the whole population) and, according to an email from Zhiqiang Li of 13-04-2020) BETA: Regression coefficient, SE: Standard error, R2: Regression r-squared, T: t-distribution statistics, P: p-value

## **Examples**

```
# we give a typical SSP output as character vector; instead we could also
# give the name of a text file
SSPout <- c(
" Please cite:",
     Shen, J. et al. SHEsisPlus, a toolset for genetic studies ...",
     Shi, Y. et al. SHEsis, a powerful software platform ...",
     Li, Z. et al. A partition-ligation-combination-subdivision ...",
""
"if you find this tool useful in your research. Thanks!",
"Haplotype Analysis:",
"Haplotypes with frequency <0.03 are ignored.",
"Loci chosen for haplotype analysis: m1, m2, m3, m4",
"Haplotype Total count Beta SE R2 T p",
"1122 2232 0.002 0.01 1.01e-04 0.252 0.8",
"1222 230 -0.019 0.017 0.002 -1.15 0.25",
"1221 152 -0.01 0.024 2.94e-04 -0.43 0.667",
"2222 288 0.008 0.02 2.93e-04 0.429 0.667",
"1121 142 -0.009 0.022 2.81e-04 -0.42 0.674"
read.ShesisPlus.output(SSPout)
```

run.SATlotyper

A simple interface to run SATlotyper

#### **Description**

A simple interface to run SATlotyper

# Usage

```
run.SATlotyper(path_to_SATlotyper, infile, outfile,
SAT_solver="sat4j.conf")
```

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#### **Arguments**

path\_to\_SATlotyper

path to the folder where SATlotyper.jar and the \*.conf files of the SAT-solvers

are located

infile name (and path) to the SATlotyper input file

outfile name (and path) for the SATlotyper output file (an xml file)

SAT\_solver name of the \*.conf file for the SAT-solver to use. Default "sat4j.conf" because

this works under both Windows and Linux.

#### **Details**

This function issues a system command to invoke SATlotyper. Java and SATlotyper must be installed. This is just a simple interface for convenience; for more control run SATlotyper directly from the command window.

#### Value

The return is a list with elements:

\$cmd: the command passed to the system

\$result: screen output from SATlotyper; this contains some extra info not present in the output xml

file)

The main result is the outfile

show0neFS	show marker and haplotype dosages for one FS family

## Description

show marker and haplotype dosages for one FS family and its parents

#### Usage

```
showOneFS(FSnr, hbresults, mrkDosage, FS, parents)
```

## **Arguments**

FSnr the number of the FS family (indexes the FS list and parents)

hbresults a list with the haplotyping results for one haploblock: one element of a list as

returned by inferHaplotypes

mrkDosage a matrix of marker dosages, may contain rows for more markers than only those

in the current haploblock (the relevant markers are specified in the hbresults list)

FS a list of which each item is a (character) vector with the names of the individuals

in that FS family

parents a (character) matrix with 2 columns and one row for each FS family in FS, with

the names of the two parents of each family

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#### Value

a list with 3 elements:

\$mrkdat: a matrix with info on the marker dosages distribution in the FS. The first two columns are for the parents: the parent name is between (brackets) in the column name, their mrkdid (marker dosage ID) in row 1 and their marker dosages below that. The remaining columns are for the different mrkdids observed in the FS: the mrkdid itself in the column name, its frequency in row 1 and its marker dosages below that. The final column gives the frequency of individuals with one or more missing marker dosages.

\$hapdat: a matrix with similar layout as mrkdat, but now with the haplotype dosages rather than the marker dosages. Some mrkdids (columns) may not have a haplotype dosage combination assigned (if multiple possible haplotype combinations result in the same marker dosages)

usedhap: a matrix with the dosage (0 or 1) of each marker in each of the used haplotypes; haplotype nrs in columns, markers in rows

## **Examples**

totHapcombCount

calculate the total nr of possible haplotype combinations

## Description

calculate the total nr of possible haplotype combinations

#### Usage

```
totHapcombCount(ploidy, nmrk)
```

# Arguments

ploidy a vector of 1 or more ploidy levels

nmrk a vector of 1 or more numbers of markers per haploblock

#### **Details**

nmrk is used to calculate the total number of possible haplotypes = 2^nmrk. The shorter vector of ploidy and nmrk is recycled.

## Value

a vector with the number of possible haplotype combinations for each pair of ploidy and nmrk values

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#### **Examples**

```
totHapcombCount(ploidy=4, nmrk=c(1:8))
```

usedhap

Find all used (inferred) haplotypes

## **Description**

Find all haplotypes for a haploblock that were inferred to be present in the population (i.e. all haplotypes used for haplotyping any of the individuals)

## Usage

```
usedhap(hapresults, haploblock)
```

## **Arguments**

hapresults list as returned by inferhaplotypes, or one element of such a list (i.e. the results

for one haploblock)

haploblock if hapresults is one element of the return value of inferHaplotypes, haploblock

should be missing of NULL; else haploblock is a single value indicating the

haploblock: either its name of its index in hapresults

## **Details**

This function works with the results of inferHaplotypes; the setting of dropUnused does not affect this function

#### Value

an array with the haplotypes that are used in the population. The haplotypes are in columns, with the haplotype numbers as colnames; the markers are in rows.

```
data(PolyHaplotyper_small)
# show the composition of haplotypes inferred to be present
# in the first haploblock:
usedhap(hapresults=phresults, haploblock=1)
```

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