

# Package ‘Summix’

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**Version** 2.0.0

**LazyData** false

**Title** Summix: A method to estimate and adjust for population structure in genetic summary data

**Description** This package contains the Summix method for estimating and adjusting for ancestry in genetic summary allele frequency data. The function `summix` estimates reference ancestry proportions using a mixture model. The `adjAF` function produces ancestry adjusted allele frequencies for an observed sample with ancestry proportions matching a target person or sample.

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**RoxygenNote** 7.1.1

**Suggests** rmarkdown, markdown, knitr

**biocViews** StatisticalMethod, WholeGenome, Genetics

**VignetteBuilder** knitr

**Encoding** UTF-8

**Depends** R (>= 4.1)

**Imports** nloptr, methods

**BugReports** <https://github.com/Bioconductor/Summix/issues>

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## R topics documented:

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| adjAF | <i>Ancestry adjusted allele frequencies</i> |
|-------|---------------------------------------------|

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**Description**

Adjusts allele frequencies for heterogeneous populations in genetic data given proportion of reference ancestry groups

**Usage**

```
adjAF(data, reference, observed, pi.target, pi.observed)
```

**Arguments**

|             |                                                                                                                                                                                                                              |
|-------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| data        | dataframe of unadjusted allele frequency for observed group, K-1 reference ancestry allele frequencies for N SNPs                                                                                                            |
| reference   | character vector of the column names for K-1 reference ancestry groups. The name of the last reference ancestry group is not included as that group is not used to estimate the adjusted allele frequencies.                 |
| observed    | character value for the column name of observed data group                                                                                                                                                                   |
| pi.target   | numeric vector of the mixture proportions for K reference ancestry groups in the target sample or subject. The order must match the order of the reference columns with the last entry matching the missing reference group. |
| pi.observed | numeric vector of the mixture proportions for K reference ancestry groups for the observed group. The order must match the order of the reference columns with the last entry matching the missing reference group.          |

**Value**

pi: table of input reference ancestry groups, pi.observed, and pi.target  
 observed.data: name of the data column for the observed group from which adjusted ancestry allele frequency is estimated  
 Nsnps: number of SNPs for which adjusted AF is estimated  
 adjusted.AF: data frame of original data with an appended column of adjusted allele frequencies

**Author(s)**

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**See Also**

[summix](#) for estimating the proportion of reference ancestry groups and <https://github.com/hendriau/Summix> for further documentation

**Examples**

```
data(ancestryData)
tmp.aa<-adjAF(data = ancestryData,
  reference = c("ref_AF_eur_1000G"),
  observed = "gnomad_AF_afr",
  pi.target = c(0, 1),
  pi.observed = c(.15, .85))
tmp.aa$adjusted.AF[1:5,]
```

ancestryData

*Example allele frequency data***Description**

reference data is 1000 Genomes and NAM. 1000 Genomes data was downloaded from <ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/> on May 31, 2018 The IAM Affymetrix 6.0 data were downloaded from <ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/working/> (data accessed October 2018) and had been previously harmonized with the 1000 Genomes data. Observed data is from gnomAD. gnomAD v2 data was downloaded from <https://gnomad.broadinstitute.org/downloads> on Oct. 11, 2018

**Usage**

```
data(ancestryData)
```

**Format**

Chromosome, SNP, base pair, reference and alternate alleles, reference allele frequencies, observed allele frequencies

**Examples**

```
data("ancestryData")
```

summix

*Summix: estimating mixture proportions of reference group***Description**

Summix: estimating mixture proportions of reference groups from large (N SNPs>10,000) genetic AF data

**Usage**

```
summix(data, reference, observed, pi.start = c())
```

## Arguments

|                        |                                                                                                                                                                                                                                |
|------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <code>data</code>      | a dataframe of the observed and reference allele frequencies for N genetic variants. See data formatting document at <a href="https://github.com/hendriau/Summix">https://github.com/hendriau/Summix</a> for more information. |
| <code>reference</code> | a character vector of the column names for the reference ancestries.                                                                                                                                                           |
| <code>observed</code>  | a character value that is the column name for the observed group.                                                                                                                                                              |
| <code>pi.start</code>  | length K numeric vector of the starting guess for the ancestry proportions. If not specified, this defaults to 1/K where K is the number of reference ancestry groups.                                                         |

## Value

data frame with the following columns  
 objective: least square value at solution  
 iterations: number of iterations for SLSQP algorithm  
 time: time in seconds of SLSQP algorithm  
 filtered: number of SNPs not used in estimation due to missing values  
 K columns of mixture proportions of reference ancestry groups input into the function

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## See Also

`adjAF` for adjusting allele frequencies and <https://github.com/hendriau/Summix> for further documentation. `slsqp` function in the `nloptr` package for further details on Sequential Quadratic Programming <https://www.rdocumentation.org/packages/nloptr/versions/1.2.2.2/topics/slsqp>

## Examples

```
# load the data
data("ancestryData")

# Estimate 5 reference ancestry proportion values for the gnomAD African/African American group
# using a starting guess of .2 for each ancestry proportion.
summix( data = ancestryData,
  reference=c("ref_AF_afr_1000G",
    "ref_AF_eur_1000G",
    "ref_AF_sas_1000G",
    "ref_AF_iam_1000G",
    "ref_AF_eas_1000G"),
  observed="gnomad_AF_afr",
  pi.start = c(.2, .2, .2, .2, .2) )
```

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