Package 'SNPtools'

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Title S4 Tools for Reading and Organizing Genetic Data

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Description Provides S4-based structures to encapsulate the import, organization, and processing of genetic data from PLINK and FImpute files, with customizable arguments. Includes tools for combining SNP panels, summarizing genotype data, and facilitating downstream quality control and analysis workflows.

Depends R (>= 4.1.0),
    snpStats,
    tidyverse

Imports methods,
    data.table,
    fQC

Suggests knitr,
    rmarkdown

VignetteBuilder knitr

Encoding UTF-8

License GPL-3

RoxygenNote 7.3.2

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check.sample.call.rate

Check Sample Call Rate

Description

Identifies samples with call rate below a given threshold.

Usage

```
check.sample.call.rate(sample.summary, min.call.rate)
```

Arguments

sample.summary A data frame with a "Call.rate" column for each sample.

min.call.rate   Minimum acceptable call rate (between 0 and 1).

Value

A character vector with the names of samples to remove.

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combinarSNPData   *Combine multiple SNPDataLong objects*

Description

This function merges a list of SNPDataLong objects, typically representing different SNP panels or datasets, into a single unified SNPDataLong object. It ensures that all genotype matrices have the same set of SNPs (filling missing SNPs with NA), and merges the marker map information while removing duplicate SNP entries.

Usage

```
combinarSNPData(lista)
```

Arguments

lista   A list of SNPDataLong objects to be combined.

Details

The function performs the following steps internally:

1. Computes the union of all SNPs across input objects.
2. Fills missing SNP columns in each genotype matrix with NA-coded columns.
3. Combines genotype matrices by rows (individuals).
4. Merges marker maps, removing duplicates (retaining the first occurrence).
5. Creates and returns a new `SNPDataLong` object with the combined data.

Value

A single `SNPDataLong` object containing the combined genotype matrix, merged map, and a concatenated path string.

Examples

```
## Not run:
# Example usage:
combined_data <- combinarSNPData(list(snp_data1, snp_data2, snp_data3))

## End(Not run)
```

FImputeRunner *Build FImputeRunner object*

Description

A convenience function to construct a 'FImputeRunner' object from basic inputs.

Usage

```
FImputeRunner(object, path, exec_path = "FImpute3", name = "data")
```

Arguments

path	A character string indicating the directory to save FImpute files.
exec_path	Path to the FImpute executable (default = "FImpute3").
name	Name for the dataset (used internally, default = "gen_data").
geno	A SnpMatrix object.
map	A data.frame with SNP metadata (columns: Name, Chromosome, Position).

Value

An object of class 'FImputeRunner'.

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getGeno   *Flexible and efficient genotype file reading with autodetection using fread*

Description

This generic and method allow flexible import of SNP genotype data from Illumina FinalReport files, supporting fast initial column detection using `data.table::fread`, followed by full genotype matrix construction via `snpStats::read.snps.long`.

Usage

```
getGeno(...)
```

Arguments

path	Path to the directory containing `FinalReport.txt`
fields	A list specifying column indices for sample, SNP, allele1, allele2, and confidence
codes	A character vector with allele codes (e.g., `c("A", "B")`)
threshold	Confidence threshold for genotype calling
sep	Field separator used in the files
skip	Number of lines to skip at the start of the file
verbose	Logical; if `TRUE`, displays progress messages
every	Frequency of progress update (number of SNPs)

Value

An `SNPDataLong` object containing the genotype matrix and map, or `NULL` if an error occurs

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importAllGenos   *Import and combine multiple genotype configurations*

Description

This generic and method import genotype data from multiple configurations defined in an `SNPImportList` object, then combine them into a single unified `SNPDataLong` object.

Usage

```
importAllGenos(object)
```

Arguments

object   An object of class `SNPImportList` containing import configurations

Value

A single combined `SNPDataLong` object

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import_geno_list   *Import multiple genotype datasets from a list of configurations*

Description

This function iterates over a list of configuration lists (each specifying parameters such as path, fields, separators, etc.), imports each genotype dataset using `getGeno()`, and then combines them into a single `SNPDataLong` object.

Usage

```
import_geno_list(config_list)
```

Arguments

config_list   A list of configuration lists. Each element must include at least `path` and `fields`. Optional elements include `codes`, `threshold`, `sep`, `skip`, and `verbose`.

Value

A unified `SNPDataLong` object containing combined genotype data from all configurations.

Examples

```
## Not run:
configs <- list(
 list(path = "panel1", fields = list(sample = 2, snp = 1, allele1 = 7, allele2 = 8, confidence = 9)),
 list(path = "panel2", fields = list(sample = 2, snp = 1, allele1 = 7, allele2 = 8, confidence = 9), threshold = 0.10
)
combined_data <- import_geno_list(configs)

## End(Not run)
```

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qcSamples   *Quality control on samples*

Description

Applies quality control (QC) procedures to samples in a 'SNPDataLong' object, based on heterozygosity and call rate thresholds.

Usage

```
qcSamples(x, ...)

## S4 method for signature 'SNPDataLong'
qcSamples(
  x,
  heterozygosity = NULL,
  smp_cr = NULL,
  action = c("report", "filter", "both")
)
```

Arguments

x	An object of class 'SNPDataLong'.
heterozygosity	A numeric threshold or range for heterozygosity. Samples outside this threshold are removed.
smp_cr	Minimum acceptable sample call rate (between 0 and 1). Samples below this value are removed.
action	Character string indicating the action to perform. One of: - '"report"': only returns a list of samples to remove and those kept; - '"filter"': returns a filtered object without reporting; - '"both"': performs filtering and returns the filtered object.

Value

Depending on the 'action' argument: - '"report"': returns a list with removed and kept samples; - '"filter"': returns a new 'SNPDataLong' object with filtered genotypes; - '"both"': returns a list with: - 'filtered': the filtered 'SNPDataLong' object; - 'report': a list of removed and kept samples.

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qcSNPs   *Quality Control for SNPDataLong with optional criteria*

Description

Allows applying genotypic quality filters with user-defined criteria, including call rate, MAF, HWE, monomorphism, chromosome filtering, and removal of SNPs at the same genomic position.

Usage

```
qcSNPs(x, ...)
```

Arguments

x	An object of class SNPDataLong.
missing_ind	Maximum allowed proportion of missing data per individual (optional). *[Currently not implemented in this function]*
missing_snp	Maximum allowed proportion of missing data per SNP (optional). *[Currently not implemented in this function]*
min_snp_cr	Minimum acceptable call rate for SNPs.
min_maf	Minimum minor allele frequency allowed for SNPs (optional).
hwe	p-value threshold for Hardy-Weinberg equilibrium test (optional).
snp_position	Logical. If TRUE, removes SNPs mapped to the same position, keeping the one with the highest MAF.
snp_mono	Logical. If TRUE, identifies and removes monomorphic SNPs.
remove_chr	Character vector of chromosomes to exclude (optional).
action	One of "report" (returns a list of removed SNPs), "filter" (returns a filtered SNPDataLong object), or "both" (returns both).

Value

Depending on the action argument: - "report": list with SNPs removed by each criterion and SNPs kept. - "filter": filtered SNPDataLong object. - "both": list with the filtered object and detailed report.

Examples

```
## Not run:
set.seed(123)
mat <- matrix(sample(c(0, 1, 2, NA), 100, replace = TRUE, prob = c(0.4, 0.4, 0.15, 0.05)),
              nrow = 10, ncol = 10)
colnames(mat) <- paste0("snp", 1:10)
rownames(mat) <- paste0("ind", 1:10)
map <- data.frame(Name = colnames(mat), Chrom = 1, Position = 1:10)
x <- new("SNPDataLong", geno = mat, map = map)
```

```
qcSNPs(x, min_snp_cr = 0.8, min_maf = 0.05, snp_mono = TRUE, snp_position = TRUE, action = "report")

## End(Not run)
```

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qc_header   *Formatted header message*

Description

Prints a formatted message with a border for section titles in the console.

Usage

```
qc_header(title)
```

Arguments

title   Character string to be printed inside the header box.

Value

No return value. Used for side effects (message).

Examples

```
qc_header("Quality Control on Samples")
```

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read.fimpute   *Read imputed genotypes from FImpute output*

Description

Reads imputed genotypes from FImpute's output folder and returns a SnpMatrix object. It expects the files 'genotypes_imp.txt' and 'snp_info.txt' to be present.

Usage

```
read.fimpute(file)
```

Arguments

file   Character. Path to the FImpute output directory (usually "output_fimpute").

Value

An object of class `SnpMatrix` containing the imputed genotypes.

Examples

```
## Not run:
snp_mat <- read.fimpute("output_fimpute")

## End(Not run)
```

---

runFImpute   *Run FImpute from a FImputeRunner object*

Description

This function runs the external FImpute software using a 'FImputeRunner' object, ensuring that all required input files are present and the results are imported.

Usage

```
runFImpute(object, verbose = TRUE)

## S4 method for signature 'FImputeRunner'
runFImpute(object, verbose = TRUE)
```

Arguments

object	An object of class 'FImputeRunner'.
verbose	Logical. If TRUE (default), FImpute output will be printed to the console.

Value

An updated 'FImputeRunner' object with the 'results' slot populated (SnpMatrix).

Examples

```
## Not run:
# Example: Running FImpute from a FImputeRunner object

path_fimpute <- "fimpute_run_example"
param_file <- file.path(path_fimpute, "fimpute.par")
fimpute_exec <- "FImpute3"  # assuming it is in PATH

export_obj <- new("FImputeExport",
                  geno = geno_obj@geno,
                  map = geno_obj@map,
                  path = path_fimpute)
```

```
runner <- new("FImputeRunner",
              export = export_obj,
              par_file = param_file,
              exec_path = fimpute_exec)

runner <- runFImpute(runner, verbose = TRUE)
head(runner@results)

## End(Not run)
```

---

saveFImpute *Save genotype and map files in FImpute format*

Description

S4 method to export genotype ('.gen'), map ('.map'), and parameter ('data.par') files compatible with the [FImpute](https://www.aps.uoguelph.ca/~msargol/fimpute/) software.

Usage

```
saveFImpute(object, ...)

## S4 method for signature 'FImputeExport'
saveFImpute(object)

## S4 method for signature 'SNPDataLong'
saveFImpute(object, path = NULL)
```

Arguments

object	An object of class 'FImputeExport' or 'SNPDataLong'.
...	Further arguments passed to methods.
path	Character. Output directory where files will be written (only for 'SNPDataLong' method; default = '"fimpute_run"').

Value

No return value. Files are saved to disk.

Examples

```
## Not run:
if (requireNamespace("snpStats", quietly = TRUE)) {
  mat <- matrix(sample(c(0L, 1L, 2L), 50, replace = TRUE), nrow = 5)
  colnames(mat) <- paste0("snp", 1:10)
  rownames(mat) <- paste0("ind", 1:5)

  sm <- new("SnpMatrix", data = as.raw(mat))
```

```
  map <- data.frame(Name = colnames(mat), Chromosome = 1, Position = 1:10)
  x <- new("SNPDataLong", geno = sm, map = map)

  saveFImpute(x, path = tempdir())
}

## End(Not run)
```

---

saveFImputeRaw   *Export genotypes and map using basic arguments*

Description

Convenience function to export FImpute files directly from a 'SnpMatrix' and map 'data.frame'.

Usage

```
saveFImputeRaw(geno, map, path)
```

Arguments

geno   A 'SnpMatrix' object (from the 'snpStats' package).

map   A 'data.frame' with columns 'Name', 'Chromosome', and 'Position'.

path   Path where the files will be saved.

Value

No return value. Files are saved to disk.

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summary,SNPDataLong-method
                    Summary for SNPDataLong objects

Description

Provides a detailed summary of an SNPDataLong object, including sample and SNP counts, proportion of missing data, and SNP distribution by chromosome if mapping information is available.

Usage

```
## S4 method for signature 'SNPDataLong'
summary(object, ...)
```

Arguments

object   An object of class SNPDataLong.

Value

Prints a summary to the console. Returns `NULL` (invisible).

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