

# Package ‘bigsnpr’

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**Type** Package

**Title** Analysis of Massive SNP Arrays

**Version** 1.3.0

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**Description** Easy-to-use, efficient, flexible and scalable tools  
for analyzing massive SNP arrays <doi:10.1093/bioinformatics/bty185>.

**License** GPL-3

**LazyData** TRUE

**Language** en-US

**ByteCompile** TRUE

**SystemRequirements** Package 'bigsnpr' includes a few functions that wrap existing software such as 'PLINK' <www.cog-genomics.org/plink2>. Functions are provided to download these software. Note that these external software might not work for some operating systems (e.g. 'PLINK' might not work on Solaris).

**Depends** R (>= 3.3), bigstatsr (>= 1.2.2)

**Imports** bigassertr, bigparallelr, bigreadr, bigutilsr (>= 0.3), data.table, foreach, ggplot2, magrittr, Matrix, methods, Rcpp, stats

**LinkingTo** bigstatsr, Rcpp, RcppArmadillo (>= 0.9.600), rmio

**Suggests** bindata, covr, dbplyr (>= 1.4), dplyr, gaston, glue, Hmisc, pcadapt (>= 4.1), RSQLite, spelling, testthat, xgboost

**RoxygenNote** 7.0.2

**URL** <https://privefl.github.io/bigsnpr>

**BugReports** <https://github.com/privefl/bigsnpr/issues>

**NeedsCompilation** yes

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**Repository** CRAN

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bigsnpr-package	<i>bigsnpr: Analysis of Massive SNP Arrays</i>
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## Description

Easy-to-use, efficient, flexible and scalable tools for analyzing massive SNP arrays <doi:10.1093/bioinformatics/bty185>.

## Arguments

G	A <a href="#">FBM.code256</a> (typically <bigSNP>\$genotypes). <b>You shouldn't have missing values.</b> Also, remember to do quality control, e.g. some algorithms in this package won't work if you use SNPs with 0 MAF.
Gna	A <a href="#">FBM.code256</a> (typically <bigSNP>\$genotypes). You can have missing values in these data.
x	A <a href="#">bigSNP</a> .
infos.chr	Vector of integers specifying each SNP's chromosome. Typically <bigSNP>\$map\$chromosome.
infos.pos	Vector of integers specifying the physical position on a chromosome (in base pairs) of each SNP. Typically <bigSNP>\$map\$physical.pos.
nploidy	Number of trials, parameter of the binomial distribution. Default is 2, which corresponds to diploidy, such as for the human genome.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>

ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .
is.size.in.bp	Deprecated.
obj.bed	Object of type bed, which is the mapping of some bed file. Use <code>obj.bed &lt;-bed(bedfile)</code> to get this object.

### Author(s)

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### See Also

Useful links:

- <https://privefl.github.io/bigsnpr>
- Report bugs at <https://github.com/privefl/bigsnpr/issues>

---

bed-class

*Class bed*

---

### Description

A reference class for storing a pointer to a mapped version of a bed file.

### Usage

`bed(bedfile)`

### Arguments

`bedfile` Path to file with extension ".bed" to read. You need the corresponding ".bim" and ".fam" in the same directory.

### Details

A bed object has many field:

- \$address: address of the external pointer containing the underlying C++ object, to be used internally as a `XPtr<bed>` in C++ code
- \$extptr: use \$address instead
- \$bedfile: path to the bed file
- \$bimfile: path to the corresponding bim file
- \$famfile: path to the corresponding fam file
- \$prefix: path without extension

- \$nrow: number of samples in the bed file
- \$ncol: number of variants in the bed file
- \$map: data frame read from \$bimfile
- \$fam: data frame read from \$famfile
- \$.map: use \$map instead
- \$.fam: use \$fam instead
- \$light: get a lighter version of this object for parallel algorithms to not have to transfer e.g. \$.map.

## Examples

```
bedfile <- system.file("extdata", "example-missing.bed", package = "bigsnpr")
(obj.bed <- bed(bedfile))
```

---

bed-methods

*Methods for the bed class*

---

## Description

Methods for the bed class

Dimension methods for class bed. Methods nrow() and ncol() are automatically defined with dim().

## Usage

```
## S4 method for signature 'bed'
dim(x)

## S4 method for signature 'bed'
length(x)
```

## Arguments

x                    Object of type bed.

---

`bed_clumping`*LD clumping*

---

## Description

For a bigSNP:

- `snp_pruning()`: LD pruning. Similar to "--indep-pairwise (size+1) 1 thr.r2" in **PLINK**. **This function is deprecated (see [this article](#))**.
- `snp_clumping()` (and `bed_clumping()`): LD clumping. If you do not provide any statistic to rank SNPs, it would use minor allele frequencies (MAFs), making clumping similar to pruning.
- `snp_indLRLDR()`: Get SNP indices of long-range LD regions for the human genome.

## Usage

```
bed_clumping(  
  obj.bed,  
  ind.row = rows_along(obj.bed),  
  S = NULL,  
  thr.r2 = 0.2,  
  size = 100/thr.r2,  
  exclude = NULL,  
  ncores = 1  
)
```

```
snp_clumping(  
  G,  
  infos.chr,  
  ind.row = rows_along(G),  
  S = NULL,  
  thr.r2 = 0.2,  
  size = 100/thr.r2,  
  infos.pos = NULL,  
  is.size.in.bp = NULL,  
  exclude = NULL,  
  ncores = 1  
)
```

```
snp_pruning(  
  G,  
  infos.chr,  
  ind.row = rows_along(G),  
  size = 49,  
  is.size.in.bp = FALSE,  
  infos.pos = NULL,
```

```

    thr.r2 = 0.2,
    exclude = NULL,
    nploidy = 2,
    ncores = 1
)

snp_indLRDR(infos.chr, infos.pos, LD.regions = LD.wiki34)

```

## Arguments

obj.bed	Object of type bed, which is the mapping of some bed file. Use obj.bed <-bed(bedfile) to get this object.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
S	A vector of column statistics which express the importance of each SNP (the more important is the SNP, the greater should be the corresponding statistic). For example, if S follows the standard normal distribution, and "important" means significantly different from 0, you must use abs(S) instead. <b>If not specified, MAFs are computed and used.</b>
thr.r2	Threshold over the squared correlation between two SNPs. Default is 0.2.
size	For one SNP, window size around this SNP to compute correlations. Default is 100 / thr.r2 for clumping (0.2 -> 500; 0.1 -> 1000; 0.5 -> 200). If not providing infos.pos (NULL, the default), this is a window in number of SNPs, otherwise it is a window in kb (genetic distance). I recommend that you provide the positions if available.
exclude	Vector of SNP indices to exclude anyway. For example, can be used to exclude long-range LD regions (see Price2008). Another use can be for thresholding with respect to p-values associated with S.
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .
G	A <a href="#">FBM.code256</a> (typically <bigSNP>\$genotypes). <b>You shouldn't have missing values.</b> Also, remember to do quality control, e.g. some algorithms in this package won't work if you use SNPs with 0 MAF.
infos.chr	Vector of integers specifying each SNP's chromosome. Typically <bigSNP>\$map\$chromosome.
infos.pos	Vector of integers specifying the physical position on a chromosome (in base pairs) of each SNP. Typically <bigSNP>\$map\$physical.pos.
is.size.in.bp	Deprecated.
nploidy	Number of trials, parameter of the binomial distribution. Default is 2, which corresponds to diploidy, such as for the human genome.
LD.regions	A data.frame with columns "Chr", "Start" and "Stop". Default use the table of 34 long-range LD regions that you can find <a href="#">there</a> .

**Value**

- `snp_clumping()` (and `bed_clumping()`): SNP indices that are **kept**.
- `snp_indLRLDR()`: SNP indices to be used as (part of) the 'exclude' parameter of `snp_clumping()`.

**References**

Price AL, Weale ME, Patterson N, et al. Long-Range LD Can Confound Genome Scans in Admixed Populations. *Am J Hum Genet.* 2008;83(1):132-135. <http://dx.doi.org/10.1016/j.ajhg.2008.06.005>

**Examples**

```
test <- snp_attachExtdata()
G <- test$genotypes

# clumping (prioritizing higher MAF)
ind.keep <- snp_clumping(G, infos.chr = test$map$chromosome,
                        infos.pos = test$map$physical.pos,
                        thr.r2 = 0.1)

# keep most of them -> not much LD in this simulated dataset
length(ind.keep) / ncol(G)
```

---

bed\_counts

*Counts*


---

**Description**

Counts the number of 0s, 1s, 2s and NAs by variants in the bed file.

**Usage**

```
bed_counts(
  obj.bed,
  ind.row = rows_along(obj.bed),
  ind.col = cols_along(obj.bed),
  byrow = FALSE,
  ncores = 1
)
```

**Arguments**

`obj.bed` Object of type `bed`, which is the mapping of some bed file. Use `obj.bed <- bed.bedfile)` to get this object.

`ind.row` An optional vector of the row indices (individuals) that are used. If not specified, all rows are used.  
**Don't use negative indices.**



ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>
byrow	Whether to count by individual rather than by variant? Default is FALSE (count by variant).
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

**Value**

A matrix of with 4 rows and `length(ind.col)` columns.

**Examples**

```
bedfile <- system.file("extdata", "example-missing.bed", package = "bigsnpr")
obj.bed <- bed(bedfile)

bed_counts(obj.bed, ind.col = 1:5)

bed_counts(obj.bed, ind.row = 1:5, byrow = TRUE)
```

---

bed_cprodVec	<i>Cross-product with a vector</i>
--------------	------------------------------------

---

**Description**

Cross-product between a "bed" object and a vector.

Missing values are replaced by 0 (after centering), as if they had been imputed using parameter `center`.

**Usage**

```
bed_cprodVec(
  obj.bed,
  y.row,
  ind.row = rows_along(obj.bed),
  ind.col = cols_along(obj.bed),
  center = rep(0, length(ind.col)),
  scale = rep(1, length(ind.col)),
  ncores = 1
)
```

**Arguments**

obj.bed	A <a href="#">bed</a> object.
y.row	A vector of same size as ind.row.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>
center	Vector of same length of ind.col to subtract from columns of X.
scale	Vector of same length of ind.col to divide from columns of X.
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

**Value**

$$X^T \cdot y.$$

**Examples**

```
bedfile <- system.file("extdata", "example.bed", package = "bigsnpr")
obj.bed <- bed(bedfile)

y.row <- rep(1, nrow(obj.bed))
str(bed_cprodVec(obj.bed, y.row))
```

---

bed_MAF	<i>Allele frequencies</i>
---------	---------------------------

---

**Description**

Allele frequencies of a [bed](#) object.

**Usage**

```
bed_MAF(
  obj.bed,
  ind.row = rows_along(obj.bed),
  ind.col = cols_along(obj.bed),
  ncores = 1
)
```

**Arguments**

obj.bed	Object of type bed, which is the mapping of some bed file. Use <code>obj.bed &lt;- bed(bedfile)</code> to get this object.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

**Value**

A data.frame with

- \$ac: allele counts,
- \$mac: minor allele counts,
- \$af: allele frequencies,
- \$maf: minor allele frequencies,
- \$N: numbers of non-missing values.

**Examples**

```
bedfile <- system.file("extdata", "example-missing.bed", package = "bigsnpr")
obj.bed <- bed(bedfile)

bed_MAF(obj.bed, ind.col = 1:5)
```

---

bed_prodVec	<i>Product with a vector</i>
-------------	------------------------------

---

**Description**

Product between a "bed" object and a vector.

Missing values are replaced by 0 (after centering), as if they had been imputed using parameter center.

**Usage**

```
bed_prodVec(
  obj.bed,
  y.col,
  ind.row = rows_along(obj.bed),
  ind.col = cols_along(obj.bed),
  center = rep(0, length(ind.col)),
```

```

    scale = rep(1, length(ind.col)),
    ncores = 1
  )

```

### Arguments

obj.bed	A <a href="#">bed</a> object.
y.col	A vector of same size as ind.col.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>
center	Vector of same length of ind.col to subtract from columns of X.
scale	Vector of same length of ind.col to divide from columns of X.
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

### Value

$X \cdot y$ .

### Examples

```

bedfile <- system.file("extdata", "example.bed", package = "bigsnpr")
obj.bed <- bed(bedfile)

y.col <- rep(1, ncol(obj.bed))
str(bed_prodVec(obj.bed, y.col))

```

---

bed_projectPCA	<i>Projecting PCA</i>
----------------	-----------------------

---

### Description

Computing and projecting PCA of reference dataset to a target dataset.

### Usage

```

bed_projectPCA(
  obj.bed.ref,
  obj.bed.new,
  k = 10,
  ind.row.new = rows_along(obj.bed.new),
  ind.row.ref = rows_along(obj.bed.ref),

```

```

ind.col.ref = cols_along(obj.bed.ref),
strand_flip = TRUE,
join_by_pos = TRUE,
match.min.prop = 0.5,
build.new = "hg19",
build.ref = "hg19",
liftOver = NULL,
...,
verbose = TRUE,
ncores = 1
)

```

### Arguments

obj.bed.ref	Object of type bed, which is the mapping of the bed file of the reference data. Use <code>obj.bed &lt;- bed(bedfile)</code> to get this object.
obj.bed.new	Object of type bed, which is the mapping of the bed file of the target data. Use <code>obj.bed &lt;- bed(bedfile)</code> to get this object.
k	Number of principal components to compute and project.
ind.row.new	Rows to be used in the target data. Default uses them all.
ind.row.ref	Rows to be used in the reference data. Default uses them all.
ind.col.ref	Columns to be potentially used in the reference data. Default uses all the ones in common with target data.
strand_flip	Whether to try to flip strand? (default is TRUE) If so, ambiguous alleles A/T and C/G are removed.
join_by_pos	Whether to join by chromosome and position (default), or instead by rsid.
match.min.prop	Minimum proportion of variants in the smallest data to be matched, otherwise stops with an error. Default is 50%.
build.new	Genome build of the target data. Default is hg19.
build.ref	Genome build of the reference data. Default is hg19.
liftOver	Path to liftOver executable. Binaries can be downloaded at <a href="https://bit.ly/2KvHugi">https://bit.ly/2KvHugi</a> for Mac and at <a href="https://bit.ly/2TbSaEI">https://bit.ly/2TbSaEI</a> for Linux.
...	Arguments passed on to <code>bed_autoSVD</code>
fun.scaling	A function that returns a named list of mean and sd for every column, to scale each of their elements such as followed:

$$\frac{X_{i,j} - mean_j}{sd_j}$$

Default is `snp_scaleBinom()`.

`roll.size` Radius of rolling windows to smooth log-p-values. Default is 50.

`int.min.size` Minimum number of consecutive outlier SNPs in order to be reported as long-range LD region. Default is 20.

`thr.r2` Threshold over the squared correlation between two SNPs. Default is 0.2. Use NA if you want to skip the clumping step.

alpha.tukey	Default is 0.1. The type-I error rate in outlier detection (that is further corrected for multiple testing).
min.mac	Minimum minor allele count (MAC) for variants to be included. Default is 10.
max.iter	Maximum number of iterations of outlier detection. Default is 5.
size	For one SNP, window size around this SNP to compute correlations. Default is 100 / thr.r2 for clumping (0.2 -> 500; 0.1 -> 1000; 0.5 -> 200). If not providing infos.pos (NULL, the default), this is a window in number of SNPs, otherwise it is a window in kb (genetic distance). I recommend that you provide the positions if available.
verbose	Output some information on the iterations? Default is TRUE.
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

### Value

A list of 3 elements:

- \$obj.svd.ref: big\_SVD object computed from reference data.
- \$simple\_proj: simple projection of new data into space of reference PCA.
- \$OADP\_proj: Online Augmentation, Decomposition, and Procrustes (OADP) projection of new data into space of reference PCA.

---

bed\_projectSelfPCA      *Projecting PCA*

---

### Description

Projecting PCA using individuals from one dataset to other individuals from the same dataset.

### Usage

```
bed_projectSelfPCA(
  obj.svd,
  obj.bed,
  ind.row,
  ind.col = attr(obj.svd, "subset"),
  ncores = 1
)
```

### Arguments

obj.svd	List with v, d, center and scale. Typically the an object of type "big_SVD".
obj.bed	Object of type bed, which is the mapping of the bed file of the data containing both the individuals that were used to compute the PCA and the other individuals to be projected.
ind.row	Rows (individuals) to be projected.

ind.col	Columns that were used for computing PCA. If <code>bed_autoSVD</code> was used, then <code>attr(obj.svd, "subset")</code> is automatically used by default. Otherwise (e.g. if <code>bed_randomSVD</code> was used), you have to pass <code>ind.col</code> .
ncores	Number of cores used. Default doesn't use parallelism. You may use <code>nb_cores</code> .

**Value**

A list of 3 elements:

- `$obj.svd.ref`: `big_SVD` object computed from reference data.
- `$simple_proj`: simple projection of new data into space of reference PCA.
- `$OADP_proj`: Online Augmentation, Decomposition, and Procrustes (OADP) projection of new data into space of reference PCA.

---

bed_randomSVD	<i>Randomized partial SVD</i>
---------------	-------------------------------

---

**Description**

Partial SVD (or PCA) of a genotype matrix stored as a PLINK (.bed) file.#'

**Usage**

```
bed_randomSVD(
  obj.bed,
  fun.scaling = bed_scaleBinom,
  ind.row = rows_along(obj.bed),
  ind.col = cols_along(obj.bed),
  k = 10,
  tol = 1e-04,
  verbose = FALSE,
  ncores = 1
)
```

**Arguments**

obj.bed	Object of type <code>bed</code> , which is the mapping of some <code>bed</code> file. Use <code>obj.bed &lt;- bed.bedfile)</code> to get this object.
fun.scaling	A function that returns a named list of mean and <code>sd</code> for every column, to scale each of their elements such as followed:

$$\frac{X_{i,j} - mean_j}{sd_j}.$$

	Default doesn't use any scaling.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>

ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>
k	Number of singular vectors/values to compute. Default is 10. <b>This algorithm should be used to compute only a few singular vectors/values.</b>
tol	Precision parameter of <a href="#">svds</a> . Default is 1e-4.
verbose	Should some progress be printed? Default is FALSE.
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

**Value**

A named list (an S3 class "big\_SVD") of

- d, the singular values,
- u, the left singular vectors,
- v, the right singular vectors,
- niter, the number of the iteration of the algorithm,
- nops, number of Matrix-Vector multiplications used,
- center, the centering vector,
- scale, the scaling vector.

Note that to obtain the Principal Components, you must use [predict](#) on the result. See examples.

**Examples**

```
bedfile <- system.file("extdata", "example.bed", package = "bigsnpr")
obj.bed <- bed(bedfile)

str(bed_randomSVD(obj.bed))
```

---

bed\_scaleBinom      *Binomial(2, p) scaling*

---

**Description**

Binomial(2, p) scaling where p is estimated.

**Usage**

```
bed_scaleBinom(
  obj.bed,
  ind.row = rows_along(obj.bed),
  ind.col = cols_along(obj.bed),
  ncores = 1
)
```



**Arguments**

obj.bed	Object of type bed, which is the mapping of some bed file. Use obj.bed <-bed.bedfile) to get this object.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

**Details**

You will probably not use this function as is but as parameter fun.scaling of other functions (e.g. bed\_autoSVD and bed\_randomSVD).

**Value**

A data frame with \$center and \$scale.

**References**

This scaling is widely used for SNP arrays. Patterson N, Price AL, Reich D (2006). Population Structure and Eigenanalysis. PLoS Genet 2(12): e190. <http://dx.doi.org/10.1371/journal.pgen.0020190>.

**Examples**

```
bedfile <- system.file("extdata", "example-missing.bed", package = "bigsnpr")
obj.bed <- bed.bedfile)

str.bed_scaleBinom(obj.bed)

str.bed_randomSVD(obj.bed, bed_scaleBinom))
```

---

bed\_tcrossprodSelf      *Tcrossprod*

---

**Description**

Compute  $X.rowX.row^T$  for a Filebacked Big Matrix  $X$  after applying a particular scaling to it.

**Usage**

```
bed_tcrossprodSelf(
  obj.bed,
  fun.scaling = bed_scaleBinom,
  ind.row = rows_along(obj.bed),
  ind.col = cols_along(obj.bed),
  block.size = block_size(length(ind.row))
)
```

**Arguments**

`obj.bed` Object of type `bed`, which is the mapping of some `bed` file. Use `obj.bed <- bed(bedfile)` to get this object.

`fun.scaling` A function that returns a named list of mean and sd for every column, to scale each of their elements such as followed:

$$\frac{X_{i,j} - mean_j}{sd_j}.$$

Default doesn't use any scaling.

`ind.row` An optional vector of the row indices that are used. If not specified, all rows are used. **Don't use negative indices.**

`ind.col` An optional vector of the column indices that are used. If not specified, all columns are used. **Don't use negative indices.**

`block.size` Maximum number of columns read at once. Default uses `block_size`.

**Value**

A temporary `FBM`, with the following two attributes:

- a numeric vector center of column scaling,
- a numeric vector scale of column scaling.

**Examples**

```
bedfile <- system.file("extdata", "example.bed", package = "bigsnpr")
obj.bed <- bed(bedfile)
```

```
K <- bed_tcrossprodSelf(obj.bed)
K[1:4, 1:6] / ncol(obj.bed)
```

---

bigSNP-class	<i>Class bigSNP</i>
--------------	---------------------

---

### Description

An S3 class for representing information on massive SNP arrays.

### Details

A named list with at least 4 slots:

**genotypes** A [FBM.code256](#) which is a special Filebacked Big Matrix encoded with type raw (one byte unsigned integer), representing genotype calls and possibly imputed allele dosages. Rows are individuals and columns are SNPs.

**fam** A data.frame containing some information on the SNPs (read from a ".fam" file).

**map** A data.frame giving some information on the individuals (read from a ".bim" file).

### See Also

[snp\\_readBed](#)

---

CODE_012	<i>CODE_012: code genotype calls (3) and missing values.</i>
----------	--

---

### Description

CODE\_012: code genotype calls (3) and missing values.

CODE\_DOSAGE: code genotype calls and missing values (4), and imputed calls (3) and imputed allele dosages rounded to two decimal places (201).

CODE\_IMPUTE\_PRED: code genotype calls and missing values (4), and imputed calls (3).

### Usage

CODE\_012

CODE\_DOSAGE

CODE\_IMPUTE\_PRED

### Format

An object of class `numeric` of length 256.

---

download_1000G	<i>Download 1000G</i>
----------------	-----------------------

---

**Description**

Download 1000 genomes project (phase 3) data in PLINK bed/bim/fam format, including 2490 (mostly unrelated) individuals and ~1.7M SNPs in common with either HapMap3 or the UK Biobank.

**Usage**

```
download_1000G(dir, overwrite = FALSE, delete_zip = TRUE)
```

**Arguments**

dir	The directory where to put the downloaded files.
overwrite	Whether to overwrite files when downloading and unzipping? Default is FALSE.
delete_zip	Whether to delete zip after decompressing the file in it? Default is TRUE.

**Value**

The path of the downloaded bed file.

---

download_beagle	<i>Download Beagle 4.1</i>
-----------------	----------------------------

---

**Description**

Download Beagle 4.1 from <https://faculty.washington.edu/browning/beagle/beagle.html>

**Usage**

```
download_beagle(dir = tempdir())
```

**Arguments**

dir	The directory where to put the Beagle Java Archive. Default is a temporary directory.
-----	---

**Value**

The path of the downloaded Beagle Java Archive.

---

download_plink	<i>Download PLINK</i>
----------------	-----------------------

---

**Description**

Download PLINK 1.9 from <http://www.cog-genomics.org/plink2>.

Download PLINK 2.0 from <http://www.cog-genomics.org/plink/2.0/>.

**Usage**

```
download_plink(dir = tempdir(), overwrite = FALSE, verbose = TRUE)
```

```
download_plink2(
  dir = tempdir(),
  AVX2 = TRUE,
  overwrite = FALSE,
  verbose = TRUE
)
```

**Arguments**

dir	The directory where to put the PLINK executable. Default is a temporary directory.
overwrite	Whether to overwrite file? Default is FALSE.
verbose	Whether to output details of downloading. Default is TRUE.
AVX2	Whether to download the AVX2 version? This is only available for 64 bits architectures. Default is TRUE.

**Value**

The path of the downloaded PLINK executable.

---

LD.wiki34	<i>Long-range LD regions</i>
-----------	------------------------------

---

**Description**

34 long-range Linkage Disequilibrium (LD) regions for the human genome based on some [wiki table](#).

**Usage**

```
LD.wiki34
```

**Format**

A data frame with 34 rows (regions) and 4 variables:

- Chr: region's chromosome
- Start: starting position of the region (in bp)
- Stop: stopping position of the region (in bp)
- ID: some ID of the region.

---

same_ref	<i>Determine reference divergence</i>
----------	---------------------------------------

---

**Description**

Determine reference divergence while accounting for strand flips. **This does not remove ambiguous alleles.**

**Usage**

```
same_ref(ref1, alt1, ref2, alt2)
```

**Arguments**

ref1	The reference alleles of the first dataset.
alt1	The alternative alleles of the first dataset.
ref2	The reference alleles of the second dataset.
alt2	The alternative alleles of the second dataset.

**Value**

A logical vector whether the references alleles are the same. Missing values can result from missing values in the inputs or from ambiguous matching (e.g. matching A/C and A/G).

**See Also**

[snp\\_match\(\)](#)

**Examples**

```
same_ref(ref1 = c("A", "C", "T", "G", NA),
         alt1 = c("C", "T", "C", "A", "A"),
         ref2 = c("A", "C", "A", "A", "C"),
         alt2 = c("C", "G", "G", "G", "A"))
```

SCT

*Stacked C+T (SCT)***Description**

Polygenic Risk Scores for a grid of clumping and thresholding parameters.

Stacking over many Polygenic Risk Scores, corresponding to a grid of many different parameters for clumping and thresholding.

**Usage**

```
snp_grid_clumping(
  G,
  infos.chr,
  infos.pos,
  lpS,
  ind.row = rows_along(G),
  grid.thr.r2 = c(0.01, 0.05, 0.1, 0.2, 0.5, 0.8, 0.95),
  grid.base.size = c(50, 100, 200, 500),
  infos.imp = rep(1, ncol(G)),
  grid.thr.imp = 1,
  groups = list(cols_along(G)),
  exclude = NULL,
  ncores = 1
)

snp_grid_PRS(
  G,
  all_keep,
  betas,
  lpS,
  n_thr_lpS = 50,
  grid.lpS.thr = 0.9999 * seq_log(max(0.1, min(lpS)), max(lpS), n_thr_lpS),
  ind.row = rows_along(G),
  backingfile = tempfile(),
  type = c("float", "double"),
  ncores = 1
)

snp_grid_stacking(
  multi_PRS,
  y.train,
  alphas = c(1, 0.01, 1e-04),
  ncores = 1,
  ...
)
```

**Arguments**

G	A <a href="#">FBM.code256</a> (typically <code>&lt;bigSNP&gt;\$genotypes</code> ). <b>You shouldn't have missing values.</b> Also, remember to do quality control, e.g. some algorithms in this package won't work if you use SNPs with 0 MAF.
infos.chr	Vector of integers specifying each SNP's chromosome. Typically <code>&lt;bigSNP&gt;\$map\$chromosome</code> .
infos.pos	Vector of integers specifying the physical position on a chromosome (in base pairs) of each SNP. Typically <code>&lt;bigSNP&gt;\$map\$physical.pos</code> .
lpS	Numeric vector of $-\log_{10}(\text{p-value})$ associated with betas.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
grid.thr.r2	Grid of thresholds over the squared correlation between two SNPs for clumping. Default is <code>c(0.01, 0.05, 0.1, 0.2, 0.5, 0.8, 0.95)</code> .
grid.base.size	Grid for base window sizes. Sizes are then computed as <code>base.size / thr.r2</code> (in kb). Default is <code>c(50, 100, 200, 500)</code> .
infos.imp	Vector of imputation scores. Default is all 1 if you do not provide it.
grid.thr.imp	Grid of thresholds over <code>infos.imp</code> (default is 1), but you should change it (e.g. <code>c(0.3, 0.6, 0.9, 0.95)</code> ) if providing <code>infos.imp</code> .
groups	List of vectors of indices to define your own categories. This could be used e.g. to derive C+T scores using two different GWAS summary statistics, or to include other information such as functional annotations. Default just makes one group with all variants.
exclude	Vector of SNP indices to exclude anyway.
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .
all_keep	Output of <code>snp_grid_clumping()</code> (indices passing clumping).
betas	Numeric vector of weights (effect sizes from GWAS) associated with each variant (column of G). If alleles are reversed, make sure to multiply corresponding effects by -1.
n_thr_lpS	Length for default <code>grid.lpS.thr</code> . Default is 50.
grid.lpS.thr	Sequence of thresholds to apply on <code>lpS</code> . Default is a grid (of length <code>n_thr_lpS</code> ) evenly spaced on a logarithmic scale, i.e. on a log-log scale for p-values.
backingfile	Prefix for backingfiles where to store scores of C+T. As we typically use a large grid, this can result in a large matrix so that we store it on disk. Default uses a temporary file.
type	Type of <code>backingfile</code> values. Either "float" (the default) or "double". Using "float" requires half disk space.
multi_PRS	Output of <code>snp_grid_PRS()</code> .
y.train	Vector of phenotypes. If there are two levels (binary 0/1), it uses <a href="#">big_splLogReg()</a> for stacking, otherwise <a href="#">big_splLinReg()</a> .



`alphas` Vector of values for grid-search. See `big_spLogReg()`. Default for this function is `c(1, 0.01, 0.0001)`.

`...` Other parameters to be passed to `big_spLogReg()`. For example, using `covar.train`, you can add covariates in the model with all C+T scores. You can also use `pf.covar` if you do not want to penalize these covariates.

### Value

`snp_grid_PRS()`: An FBM (matrix on disk) that stores the C+T scores for all parameters of the grid (and for each chromosome separately). It also stores as attributes the input parameters `all_keep`, `betas`, `lpS` and `grid.lpS.thr` that are also needed in `snp_grid_stacking()`.

---

<code>seq_log</code>	<i>Sequence, evenly spaced on a logarithmic scale</i>
----------------------	---

---

### Description

Sequence, evenly spaced on a logarithmic scale

### Usage

```
seq_log(from, to, length.out)
```

### Arguments

`from` the starting and (maximal) end values of the sequence. Of length 1 unless just `from` is supplied as an unnamed argument.

`to` the starting and (maximal) end values of the sequence. Of length 1 unless just `from` is supplied as an unnamed argument.

`length.out` desired length of the sequence. A non-negative number, which for `seq` and `seq.int` will be rounded up if fractional.

### Examples

```
seq_log(1, 1000, 4)
seq_log(1, 100, 5)
```

snp\_assocBGEN

*Compute quick association statistics from BGEN files***Description****THIS FUNCTION WILL BE MODIFIED IN THE FUTURE.****Usage**

```
snp_assocBGEN(
  bgenfiles,
  list_snp_id,
  y_row,
  ind_row,
  bgi_dir = dirname(bgenfiles),
  ncores = 1
)
```

**Arguments**

bgenfiles	Character vector of paths to files with extension ".bgen". The corresponding ".bgen.bgi" index files must exist.
list_snp_id	List (same length as the number of BGEN files) of character vector of SNP IDs to read. These should be in the form "<chr>_<pos>_<a1>_<a2>" (e.g. "1_88169_C_T" or "01_88169_C_T"). <b>This function assumes that these IDs are uniquely identifying variants.</b>
y_row	A vector corresponding to ind_row and representing the trait with which to compute correlations. Missing values in either ind_row or y_row are removed.
ind_row	A vector of the row indices (individuals) that are used. Missing values in either ind_row or y_row are removed. <b>Make sure to use indices corresponding to your training set only.</b>
bgi_dir	Directory of index files. Default is the same as bgenfiles.
ncores	Number of cores used. Default doesn't use parallelism. You may use <code>nb_cores()</code> .

**Value**

A list of vectors of  $\log_{10}(\text{p-values})$  corresponding to the statistic  $n \times r^2$ , where  $r$  is the correlation of each variant with `y_row`.

---

snp_attach	<i>Attach a "bigSNP" from backing files</i>
------------	---

---

**Description**

Load a [bigSNP](#) from backing files into R.

**Usage**

```
snp_attach(rdsfile)
```

**Arguments**

rdsfile            The path of the ".rds" which stores the bigSNP object.

**Details**

This is often just a call to [readRDS](#). But it also checks if you have moved the two (".bk" and ".rds") backing files to another directory.

**Value**

The bigSNP object.

**Examples**

```
(bedfile <- system.file("extdata", "example.bed", package = "bigsnpr"))  
  
# Reading the bedfile and storing the data in temporary directory  
rds <- snp_readBed(bedfile, backingfile = tempfile())  
  
# Loading the data from backing files  
test <- snp_attach(rds)  
  
str(test)  
dim(G <- test$genotypes)  
G[1:8, 1:8]
```

---

snp_attachExtdata	<i>Attach a "bigSNP" for examples and tests</i>
-------------------	---

---

**Description**

Attach a "bigSNP" for examples and tests

**Usage**

```
snp_attachExtdata(bedfile = c("example.bed", "example-missing.bed"))
```

**Arguments**

bedfile            Name of one example bed file. Either

- "example.bed" (the default),
- "example-missing.bed".

**Value**

The example "bigSNP", filebacked in the "/tmp/" directory.

---

snp_autoSVD	<i>Truncated SVD while limiting LD</i>
-------------	--

---

**Description**

Fast truncated SVD with initial pruning and that iteratively removes long-range LD regions.

**Usage**

```
snp_autoSVD(
  G,
  infos.chr,
  infos.pos = NULL,
  ind.row = rows_along(G),
  ind.col = cols_along(G),
  fun.scaling = snp_scaleBinom(),
  thr.r2 = 0.2,
  size = 100/thr.r2,
  k = 10,
  roll.size = 50,
  int.min.size = 20,
  alpha.tukey = 0.05,
  min.mac = 10,
  max.iter = 5,
  is.size.in.bp = NULL,
  ncores = 1,
  verbose = TRUE
)
```

```
bed_autoSVD(
  obj.bed,
  ind.row = rows_along(obj.bed),
  ind.col = cols_along(obj.bed),
  fun.scaling = bed_scaleBinom,
  thr.r2 = 0.2,
  size = 100/thr.r2,
  k = 10,
```

```

roll.size = 50,
int.min.size = 20,
alpha.tukey = 0.05,
min.mac = 10,
max.iter = 5,
ncores = 1,
verbose = TRUE
)

```

## Arguments

G	A <a href="#">FBM.code256</a> (typically <code>&lt;bigSNP&gt;\$genotypes</code> ). <b>You shouldn't have missing values.</b> Also, remember to do quality control, e.g. some algorithms in this package won't work if you use SNPs with 0 MAF.
infos.chr	Vector of integers specifying each SNP's chromosome. Typically <code>&lt;bigSNP&gt;\$map\$chromosome</code> .
infos.pos	Vector of integers specifying the physical position on a chromosome (in base pairs) of each SNP. Typically <code>&lt;bigSNP&gt;\$map\$physical.pos</code> .
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>
fun.scaling	A function that returns a named list of mean and sd for every column, to scale each of their elements such as followed: $\frac{X_{i,j} - mean_j}{sd_j}$ Default is <code>snp_scaleBinom()</code> .
thr.r2	Threshold over the squared correlation between two SNPs. Default is 0.2. Use NA if you want to skip the clumping step.
size	For one SNP, window size around this SNP to compute correlations. Default is $100 / thr.r2$ for clumping (0.2 -> 500; 0.1 -> 1000; 0.5 -> 200). If not providing <code>infos.pos</code> (NULL, the default), this is a window in number of SNPs, otherwise it is a window in kb (genetic distance). I recommend that you provide the positions if available.
k	Number of singular vectors/values to compute. Default is 10. <b>This algorithm should be used to compute a few singular vectors/values.</b>
roll.size	Radius of rolling windows to smooth log-p-values. Default is 50.
int.min.size	Minimum number of consecutive outlier SNPs in order to be reported as long-range LD region. Default is 20.
alpha.tukey	Default is 0.1. The type-I error rate in outlier detection (that is further corrected for multiple testing).

<code>min.mac</code>	Minimum minor allele count (MAC) for variants to be included. Default is 10.
<code>max.iter</code>	Maximum number of iterations of outlier detection. Default is 5.
<code>is.size.in.bp</code>	Deprecated.
<code>ncores</code>	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .
<code>verbose</code>	Output some information on the iterations? Default is TRUE.
<code>obj.bed</code>	Object of type bed, which is the mapping of some bed file. Use <code>obj.bed &lt;- bed(bedfile)</code> to get this object.

### Details

If you don't have any information about SNPs, you can try using

- `infos.chr = rep(1, ncol(G))`,
- `size = ncol(G)` (if SNPs are not sorted),
- `roll.size = 0` (if SNPs are not sorted).

### Value

A named list (an S3 class "big\_SVD") of

- `d`, the singular values,
- `u`, the left singular vectors,
- `v`, the right singular vectors,
- `niter`, the number of the iteration of the algorithm,
- `nops`, number of Matrix-Vector multiplications used,
- `center`, the centering vector,
- `scale`, the scaling vector.

Note that to obtain the Principal Components, you must use [predict](#) on the result. See examples.

### Examples

```
ex <- snp_attachExtdata()

obj.svd <- snp_autoSVD(G = ex$genotypes,
                      infos.chr = ex$map$chromosome,
                      infos.pos = ex$map$physical.position)

str(obj.svd)
```

---

snp\_beagleImpute      *Imputation*

---

## Description

Imputation using **Beagle** version 4.

## Usage

```
snp_beagleImpute(  
  beagle.path,  
  plink.path,  
  bedfile.in,  
  bedfile.out = NULL,  
  memory.max = 3,  
  ncores = 1,  
  extra.options = "",  
  plink.options = "",  
  verbose = TRUE  
)
```

## Arguments

beagle.path	Path to the executable of Beagle v4+.
plink.path	Path to the executable of PLINK 1.9.
bedfile.in	Path to the input bedfile.
bedfile.out	Path to the output bedfile. Default is created by appending "_impute" to prefix.in (bedfile.in without extension).
memory.max	Max memory (in GB) to be used. It is internally rounded to be an integer. Default is 3.
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .
extra.options	Other options to be passed to Beagle as a string. More options can be found at Beagle's website.
plink.options	Other options to be passed to PLINK as a string. More options can be found at <a href="http://www.cog-genomics.org/plink2/filter">http://www.cog-genomics.org/plink2/filter</a> .
verbose	Whether to show PLINK log? Default is TRUE.

## Details

Downloads and more information can be found at the following websites

- [PLINK](#),
- [Beagle](#).

**Value**

The path of the new bedfile.

**References**

Browning, Brian L., and Sharon R. Browning. "Genotype imputation with millions of reference samples." *The American Journal of Human Genetics* 98.1 (2016): 116-126.

**See Also**

[download\\_plink](#) [download\\_beagle](#)

---

snp\_cor

*Correlation*


---

**Description**

Get significant correlations between nearby SNPs of the same chromosome (p-values are computed using a two-sided t-test).

**Usage**

```
snp_cor(
  Gna,
  ind.row = rows_along(Gna),
  ind.col = cols_along(Gna),
  size = 500,
  alpha = 0.05,
  fill.diag = TRUE,
  infos.pos = NULL,
  ncores = 1
)
```

**Arguments**

Gna	A <a href="#">FBM.code256</a> (typically <code>&lt;bigSNP&gt;\$genotypes</code> ). You can have missing values in these data.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>
size	For one SNP, window size around this SNP to compute correlations. Default is 500. If not providing <code>infos.pos</code> (NULL, the default), this is a window in number of SNPs, otherwise it is a window in kb (genetic distance).



alpha	Type-I error for testing correlations.
fill.diag	Whether to fill the diagonal with 1s (the default) or to keep it as 0s.
infos.pos	Vector of integers specifying the physical position on a chromosome (in base pairs) of each SNP. Typically <code>&lt;bigSNP&gt;\$map\$physical.pos</code> .
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

**Value**

The correlation matrix. This is a sparse symmetric matrix.

**Examples**

```
test <- snp_attachExtdata()

corr <- snp_cor(test$genotypes, ind.col = 1:1000)
corr[1:10, 1:10]

# Sparsity
length(corr@x) / length(corr)
```

---

snp_fastImpute	<i>Fast imputation</i>
----------------	------------------------

---

**Description**

Fast imputation algorithm based on local XGBoost models.

**Usage**

```
snp_fastImpute(
  Gna,
  infos.chr,
  alpha = 1e-04,
  size = 200,
  p.train = 0.8,
  n.cor = nrow(Gna),
  seed = NA,
  ncores = 1
)
```

**Arguments**

Gna	A <a href="#">FBM.code256</a> (typically <bigSNP>\$genotypes). You can have missing values in these data.
infos.chr	Vector of integers specifying each SNP's chromosome. Typically <bigSNP>\$map\$chromosome.
alpha	Type-I error for testing correlations. Default is 1e-4.
size	Number of neighbor SNPs to be possibly included in the model imputing this particular SNP. Default is 200.
p.train	Proportion of non missing genotypes that are used for training the imputation model while the rest is used to assess the accuracy of this imputation model. Default is 0.8.
n.cor	Number of rows that are used to estimate correlations. Default uses them all.
seed	An integer, for reproducibility. Default doesn't use seeds.
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

**Value**

An [FBM](#) with

- the proportion of missing values by SNP (first row),
- the estimated proportion of imputation errors by SNP (second row).

**See Also**

[snp\\_fastImputeSimple\(\)](#)

**Examples**

```
## Not run:

fake <- snp_attachExtdata("example-missing.bed")
G <- fake$genotypes
CHR <- fake$map$chromosome
infos <- snp_fastImpute(G, CHR)
infos[, 1:5]

# Still missing values
big_counts(G, ind.col = 1:10)
# You need to change the code of G
# To make this permanent, you need to save (modify) the file on disk
fake$genotypes$code256 <- CODE_IMPUTE_PRED
fake <- snp_save(fake)
big_counts(fake$genotypes, ind.col = 1:10)

# Plot for post-checking
## Here there is no SNP with more than 1% error (estimated)
pvals <- c(0.01, 0.005, 0.002, 0.001); colvals <- 2:5
df <- data.frame(pNA = infos[1, ], pError = infos[2, ])
```

```

# base R
plot(subset(df, pNA > 0.001), pch = 20)
idc <- lapply(seq_along(pvals), function(i) {
  curve(pvals[i] / x, from = 0, lwd = 2,
        col = colvals[i], add = TRUE)
})
legend("topright", legend = pvals, title = "p(NA & Error)",
      col = colvals, lty = 1, lwd = 2)

# ggplot2
library(ggplot2)
Reduce(function(p, i) {
  p + stat_function(fun = function(x) pvals[i] / x, color = colvals[i])
}, x = seq_along(pvals), init = ggplot(df, aes(pNA, pError))) +
  geom_point() +
  coord_cartesian(ylim = range(df$pError, na.rm = TRUE)) +
  theme_bigstatsr()

## End(Not run)

```

---

snp\_fastImputeSimple *Fast imputation*

---

## Description

Fast imputation via mode, mean, sampling according to allele frequencies, or 0.

## Usage

```

snp_fastImputeSimple(
  Gna,
  method = c("mode", "mean0", "mean2", "random"),
  ncores = 1
)

```

## Arguments

Gna	A <a href="#">FBM.code256</a> (typically <bigSNP>\$genotypes). You can have missing values in these data.
method	Either "random" (sampling according to allele frequencies), "mean0" (rounded mean), "mean2" (rounded mean to 2 decimal places), "mode" (most frequent call).
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

## Value

A new [FBM.code256](#) object (same file, but different code).

**See Also**

[snp\\_fastImpute\(\)](#)

**Examples**

```
big SNP <- snp_attachExtdata("example-missing.bed")
G <- big SNP$genotypes
G[, 2] # some missing values
G2 <- snp_fastImputeSimple(G)
G2[, 2] # no missing values anymore
G[, 2] # imputed, but still returning missing values
G$copy(code = CODE_IMPUTE_PRED)[, 2] # need to decode imputed values

G$copy(code = c(0, 1, 2, rep(0, 253)))[, 2] # "imputation" by 0
```

---

snp\_gc

*Genomic Control*

---

**Description**

Genomic Control

**Usage**

```
snp_gc(gwas)
```

**Arguments**

**gwas** A mhtest object with the p-values associated with each SNP. Typically, the output of [big\\_univLinReg](#), [big\\_univLogReg](#) or [snp\\_pcadapt](#).

**Value**

A ggplot2 object. You can plot it using the print method. You can modify it as you wish by adding layers. You might want to read [this chapter](#) to get more familiar with the package **ggplot2**.

**References**

Devlin, B., & Roeder, K. (1999). Genomic control for association studies. *Biometrics*, 55(4), 997-1004.

**Examples**

```

set.seed(9)

test <- snp_attachExtdata()
G <- test$genotypes
y <- rnorm(nrow(G))

gwas <- big_univLinReg(G, y)
snp_qq(gwas)
gwas_gc <- snp_gc(gwas) # change attr(gwas_gc, "transfo")

snp_qq(gwas_gc)
# The next plot should be prettier with a real dataset
snp_manhattan(gwas_gc,
               infos.chr = test$map$chromosome,
               infos.pos = test$map$physical.pos)

p <- snp_qq(gwas_gc) + ggplot2::aes(text = asPlotlyText(test$map))
## Not run: plotly::ggplotly(p, tooltip = "text")

```

---

snp\_getSampleInfos      *Get sample information*

---

**Description**

Get information of individuals by matching from an external file.

**Usage**

```

snp_getSampleInfos(
  x,
  df.or.files,
  col.family.ID = 1,
  col.sample.ID = 2,
  col.infos = -c(1, 2),
  pair.sep = "-_",
  ...
)

```

**Arguments**

x	A <a href="#">bigSNP</a> .
df.or.files	Either <ul style="list-style-type: none"> <li>• A data.frame,</li> <li>• A character vector of file names where to find at the information you want. You should have one column for family IDs and one for sample IDs.</li> </ul>

col.family.ID	Index of the column containing the family IDs to match with those of the study. Default is the second one.
col.sample.ID	Index of the column containing the sample IDs to match with those of the study. Default is the first one.
col.infos	Indices of the column containing the information you want. Default is all but the first and the second columns.
pair.sep	Separator used for concatenation family and sample IDs in order to match easier. Default is "-_".
...	Any additional parameter to pass to <code>bigreadr::fread2()</code> . Particularly, option <code>header = FALSE</code> is sometimes needed.

**Value**

The requested information as a `data.frame`.

**See Also**

[list.files](#)

**Examples**

```
test <- snp_attachExtdata()
# Just after reading
rle(test$fam$family.ID)
# Get populations clusters from external files
files <- system.file("extdata", paste0("cluster", 1:3), package = "bigsnpr")
bigreadr::fread2(files[1])
# need header option
bigreadr::fread2(files[1], header = FALSE)
infos <- snp_getSampleInfos(test, files, header = FALSE)
rle(infos[[1]])
```

---

snp\_MAF

*MAF*


---

**Description**

Minor Allele Frequency.

**Usage**

```
snp_MAF(
  G,
  ind.row = rows_along(G),
  ind.col = cols_along(G),
  nploidy = 2,
  ncores = 1
)
```

**Arguments**

G	A <a href="#">FBM.code256</a> (typically <bigSNP>\$genotypes). <b>You shouldn't have missing values.</b> Also, remember to do quality control, e.g. some algorithms in this package won't work if you use SNPs with 0 MAF.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>
nploidy	Number of trials, parameter of the binomial distribution. Default is 2, which corresponds to diploidy, such as for the human genome.
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

**Value**

A vector of MAFs, corresponding to ind.col.

**Examples**

```
obj.bigsnp <- snp_attachExtdata()
str(maf <- snp_MAF(obj.bigsnp$genotypes))
```

---

snp\_manhattan

*Manhattan plot*


---

**Description**

Creates a manhattan plot.

**Usage**

```
snp_manhattan(
  gwas,
  infos.chr,
  infos.pos,
  colors = c("black", "grey60"),
  dist.sep.chrs = 1e+07,
  ind.highlight = integer(0),
  col.highlight = "red",
  labels = NULL,
  npoints = NULL,
  coeff = 1
)
```

## Arguments

gwas	A mhtest object with the p-values associated with each SNP. Typically, the output of <code>big_univLinReg</code> , <code>big_univLogReg</code> or <code>snp_pcadapt</code> .
infos.chr	Vector of integers specifying each SNP's chromosome. Typically <code>&lt;bigSNP&gt;\$map\$chromosome</code> .
infos.pos	Vector of integers specifying the physical position on a chromosome (in base pairs) of each SNP. Typically <code>&lt;bigSNP&gt;\$map\$physical.pos</code> .
colors	Colors used for each chromosome (they are recycled). Default is an alternation of black and gray.
dist.sep.chrs	"Physical" distance that separates two chromosomes. Default is 10 Mbp.
ind.highlight	Indices of SNPs you want to highlight (of interest). Default doesn't highlight any SNPs.
col.highlight	Color used for highlighting SNPs. Default uses red.
labels	Labels of the x axis. Default uses the number of the chromosome there are in <code>infos.chr(sort(unique(infos.chr)))</code> . This may be useful to restrict the number of labels so that they are not overlapping.
npoints	Number of points to keep (ranked by p-value) in order to get a lighter object (and plot). Default doesn't cut anything. If used, the resulting object will have an attribute called <code>subset</code> giving the indices of the kept points.
coeff	Relative size of text. Default is 1.

## Details

If you don't have information of chromosome and position, you should simply use `plot` instead.

## Value

A `ggplot2` object. You can plot it using the `print` method. You can modify it as you wish by adding layers. You might want to read [this chapter](#) to get more familiar with the package `ggplot2`.

## Examples

```
set.seed(9)

test <- snp_attachExtdata()
G <- test$genotypes
y <- rnorm(nrow(G))

gwas <- big_univLinReg(G, y)
snp_qq(gwas)
gwas_gc <- snp_gc(gwas) # change attr(gwas_gc, "transfo")

snp_qq(gwas_gc)
# The next plot should be prettier with a real dataset
snp_manhattan(gwas_gc,
              infos.chr = test$map$chromosome,
```



```
infos.pos = test$map$physical.pos)

p <- snp_qq(gwas_gc) + ggplot2::aes(text = asPlotlyText(test$map))
## Not run: plotly::ggplotly(p, tooltip = "text")
```

---

snp\_match

*Match alleles*

---

## Description

Match alleles between summary statistics and SNP information. Match by ("chr", "a0", "a1") and ("pos" or "rsid"), accounting for possible strand flips and reverse reference alleles (opposite effects).

## Usage

```
snp_match(
  sumstats,
  info_snp,
  strand_flip = TRUE,
  join_by_pos = TRUE,
  match.min.prop = 0.5
)
```

## Arguments

sumstats	A data frame with columns "chr", "pos", "a0", "a1" and "beta".
info_snp	A data frame with columns "chr", "pos", "a0" and "a1".
strand_flip	Whether to try to flip strand? (default is TRUE) If so, ambiguous alleles A/T and C/G are removed.
join_by_pos	Whether to join by chromosome and position (default), or instead by rsid.
match.min.prop	Minimum proportion of variants in the smallest data to be matched, otherwise stops with an error. Default is 50%.

## Value

A single data frame with matched variants.

## See Also

[snp\\_modifyBuild](#)

## Examples

```
sumstats <- data.frame(
  chr = 1,
  pos = c(86303, 86331, 162463, 752566, 755890, 758144),
  a0 = c("T", "G", "C", "A", "T", "G"),
  a1 = c("G", "A", "T", "G", "A", "A"),
  beta = c(-1.868, 0.250, -0.671, 2.112, 0.239, 1.272),
  p = c(0.860, 0.346, 0.900, 0.456, 0.776, 0.383)
)

info_snp <- data.frame(
  id = c("rs2949417", "rs115209712", "rs143399298", "rs3094315", "rs3115858"),
  chr = 1,
  pos = c(86303, 86331, 162463, 752566, 755890),
  a0 = c("T", "A", "G", "A", "T"),
  a1 = c("G", "G", "A", "G", "A")
)

snp_match(sumstats, info_snp)
snp_match(sumstats, info_snp, strand_flip = FALSE)
```

---

snp\_MAX3

*MAX3 statistic*

---

## Description

Compute the MAX3 statistic, which tests for three genetic models (additive, recessive and dominant).

## Usage

```
snp_MAX3(Gna, y01.train, ind.train = rows_along(Gna), val = c(0, 0.5, 1))
```

## Arguments

Gna	A <a href="#">FBM.code256</a> (typically <bigSNP>\$genotypes). You can have missing values in these data.
y01.train	Vector of responses, corresponding to ind.train. <b>Must be only 0s and 1s.</b>
ind.train	An optional vector of the row indices that are used, for the training part. If not specified, all rows are used. <b>Don't use negative indices.</b>
val	Computing $\max_{x \in \text{val}} Z_{CATT}^2(x)$ . <ul style="list-style-type: none"> <li>• Default is <math>c(0, 0.5, 1)</math> and corresponds to the <i>MAX3</i> statistic.</li> <li>• Only <math>c(0, 1)</math> corresponds to <i>MAX2</i>.</li> <li>• And only <math>0.5</math> corresponds to the Armitage trend test.</li> <li>• Finally, <math>\text{seq}(0, 1, \text{length.out} = L)</math> corresponds to <i>MAXL</i>.</li> </ul>

## Details

**P-values associated with returned scores are in fact the minimum of the p-values of each test separately. Thus, they are biased downward.**

## Value

An object of classes `mhtest` and `data.frame` returning one score by SNP. See `methods(class = "mhtest")`.

## References

Zheng, G., Yang, Y., Zhu, X., & Elston, R. (2012). Robust Procedures. Analysis Of Genetic Association Studies, 151-206. [http://dx.doi.org/10.1007/978-1-4614-2245-7\\_6](http://dx.doi.org/10.1007/978-1-4614-2245-7_6).

## Examples

```
set.seed(1)

# constructing a fake genotype big.matrix
N <- 50; M <- 1200
fake <- snp_fake(N, M)
G <- fake$genotypes
G[] <- sample(as.raw(0:3), size = length(G), replace = TRUE)
G[1:8, 1:10]

# Specify case/control phenotypes
fake$fam$affection <- rep(1:2, each = N / 2)

# Get MAX3 statistics
y01 <- fake$fam$affection - 1
str(test <- snp_MAX3(fake$genotypes, y01.train = y01))
# p-values are not well calibrated
snp_qq(test)
# genomic control is not of much help
snp_qq(snp_gc(test))

# Armitage trend test (well calibrated because only one test)
test2 <- snp_MAX3(fake$genotypes, y01.train = y01, val = 0.5)
snp_qq(test2)
```

---

snp\_modifyBuild

*Modify genome build*

---

## Description

Modify the physical position information of a data frame when converting genome build using executable *liftOver*.

**Usage**

```
snp_modifyBuild(info_snp, liftOver, from = "hg18", to = "hg19")
```

**Arguments**

info_snp	A data frame with columns "chr" and "pos".
liftOver	Path to liftOver executable. Binaries can be downloaded at <a href="https://bit.ly/2KvHugi">https://bit.ly/2KvHugi</a> for Mac and at <a href="https://bit.ly/2TbSaEI">https://bit.ly/2TbSaEI</a> for Linux.
from	Genome build to convert from. Default is hg18.
to	Genome build to convert to. Default is hg19.

**Value**

Input data frame info\_snp with column "pos" in the new build.

**References**

Hinrichs, Angela S., et al. "The UCSC genome browser database: update 2006." *Nucleic acids research* 34.suppl\_1 (2006): D590-D598.

---

snp\_pcadapt

*Outlier detection*

---

**Description**

Method to detect genetic markers involved in biological adaptation. This provides a statistical tool for outlier detection based on Principal Component Analysis. This corresponds to the statistic based on mahalanobis distance, as implemented in package **pcadapt**.

**Usage**

```
snp_pcadapt(  
  G,  
  U.row,  
  ind.row = rows_along(G),  
  ind.col = cols_along(G),  
  ncores = 1  
)  
  
bed_pcadapt(  
  obj.bed,  
  U.row,  
  ind.row = rows_along(obj.bed),  
  ind.col = cols_along(obj.bed),  
  ncores = 1  
)
```

**Arguments**

G	A <a href="#">FBM.code256</a> (typically <code>&lt;bigSNP&gt;\$genotypes</code> ). <b>You shouldn't have missing values.</b> Also, remember to do quality control, e.g. some algorithms in this package won't work if you use SNPs with 0 MAF.
U.row	Left singular vectors (not scores, $U^T U = I$ ) corresponding to <code>ind.row</code> .
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .
obj.bed	Object of type <code>bed</code> , which is the mapping of some <code>bed</code> file. Use <code>obj.bed &lt;- bed(bedfile)</code> to get this object.

**Value**

An object of classes `mhtest` and `data.frame` returning one score by SNP. See `methods(class = "mhtest")`.

**References**

Luu, K., Bazin, E., & Blum, M. G. (2017). `pcadapt`: an R package to perform genome scans for selection based on principal component analysis. *Molecular ecology resources*, 17(1), 67-77.

**See Also**

[snp\\_manhattan](#), [snp\\_qq](#) and [snp\\_gc](#).

**Examples**

```
test <- snp_attachExtdata()
G <- test$genotypes
obj.svd <- big_SVD(G, fun.scaling = snp_scaleBinom(), k = 10)
plot(obj.svd) # there seems to be 3 "significant" components
pcadapt <- snp_pcadapt(G, obj.svd$u[, 1:3])
snp_qq(pcadapt)
```

---

snp\_plinkIBDQC

*Identity-by-descent*


---

**Description**

Quality Control based on Identity-by-descent (IBD) computed by **PLINK 1.9** using its method-of-moments.

**Usage**

```
snp_plinkIBDQC(
  plink.path,
  bedfile.in,
  bedfile.out = NULL,
  pi.hat = 0.08,
  ncores = 1,
  pruning.args = c(100, 0.2),
  do.blind.QC = TRUE,
  extra.options = "",
  verbose = TRUE
)
```

**Arguments**

plink.path	Path to the executable of PLINK 1.9.
bedfile.in	Path to the input bedfile.
bedfile.out	Path to the output bedfile. Default is created by appending "_norel" to prefix.in (bedfile.in without extension).
pi.hat	PI_HAT value threshold for individuals (first by pairs) to be excluded. Default is 0.08.
ncores	Number of cores used. Default doesn't use parallelism. You may use <code>nb_cores</code> .
pruning.args	A vector of 2 pruning parameters, respectively the window size (in variant count) and the pairwise $r^2$ threshold (the step size is fixed to 1). Default is <code>c(100, 0.2)</code> .
do.blind.QC	Whether to do QC with pi.hat without visual inspection. Default is TRUE. If FALSE, return the data.frame of the corresponding ".genome" file without doing QC. One could use <code>ggplot2::qplot(Z0, Z1, data = mydf, col = RT)</code> for visual inspection.
extra.options	Other options to be passed to PLINK as a string (for the IBD part). More options can be found at <a href="http://www.cog-genomics.org/plink/1.9/ibd">http://www.cog-genomics.org/plink/1.9/ibd</a> .
verbose	Whether to show PLINK log? Default is TRUE.

**Value**

The path of the new bedfile. If no sample is filter, no new bed/bim/fam files are created and then the path of the input bedfile is returned.

**References**

Chang, Christopher C, Carson C Chow, Laurent CAM Tellier, Shashaank Vattikuti, Shaun M Purcell, and James J Lee. 2015. *Second-generation PLINK: rising to the challenge of larger and richer datasets*. GigaScience 4 (1): 7. <http://dx.doi.org/10.1186/s13742-015-0047-8>.

**See Also**

[download\\_plink](#) [snp\\_plinkQC](#) [snp\\_plinkKINGQC](#)

## Examples

```
## Not run:

bedfile <- system.file("extdata", "example.bed", package = "bigsnpr")
plink <- download_plink()

bedfile <- snp_plinkIBDQC(plink, bedfile,
                        bedfile.out = tempfile(fileext = ".bed"),
                        ncores = 2)

df_rel <- snp_plinkIBDQC(plink, bedfile, do.blind.QC = FALSE, ncores = 2)
str(df_rel)

library(ggplot2)
qplot(Z0, Z1, data = df_rel, col = RT)
qplot(y = PI_HAT, data = df_rel) +
  geom_hline(yintercept = 0.2, color = "blue", linetype = 2)
snp_plinkRmSamples(plink, bedfile,
                  bedfile.out = tempfile(fileext = ".bed"),
                  df.or.files = subset(df_rel, PI_HAT > 0.2))

## End(Not run)
```

---

snp_plinkKINGQC	<i>Relationship-based pruning</i>
-----------------	-----------------------------------

---

## Description

Quality Control based on KING-robust kinship estimator. More information can be found at [http://www.cog-genomics.org/plink/2.0/distance#king\\_cutoff](http://www.cog-genomics.org/plink/2.0/distance#king_cutoff).

## Usage

```
snp_plinkKINGQC(
  plink2.path,
  bedfile.in,
  bedfile.out = NULL,
  thr.king = 2^-3.5,
  make.bed = TRUE,
  ncores = 1,
  extra.options = "",
  verbose = TRUE
)
```

## Arguments

plink2.path      Path to the executable of PLINK 2.

bedfile.in	Path to the input bedfile.
bedfile.out	Path to the output bedfile. Default is created by appending "_norel" to prefix.in (bedfile.in without extension).
thr.king	Note that KING kinship coefficients are scaled such that duplicate samples have kinship 0.5, not 1. First-degree relations (parent-child, full siblings) correspond to ~0.25, second-degree relations correspond to ~0.125, etc. It is conventional to use a cutoff of ~0.354 ( $2^{-1.5}$ , the geometric mean of 0.5 and 0.25) to screen for monozygotic twins and duplicate samples, ~0.177 ( $2^{-2.5}$ ) to remove first-degree relations as well, and ~0.0884 ( $2^{-3.5}$ , <b>default</b> ) to remove second-degree relations as well, etc.
make.bed	Whether to create new bed/bim/fam files (default). Otherwise, returns a table with coefficients of related pairs.
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .
extra.options	Other options to be passed to PLINK2 as a string.
verbose	Whether to show PLINK log? Default is TRUE.

### Value

See parameter `make-bed`.

### References

Manichaikul, Ani, Josyf C. Mychaleckyj, Stephen S. Rich, Kathy Daly, Michele Sale, and Wei-Min Chen. "Robust relationship inference in genome-wide association studies." *Bioinformatics* 26, no. 22 (2010): 2867-2873.

### See Also

[download\\_plink2](#) [snp\\_plinkQC](#)

### Examples

```
## Not run:

bedfile <- system.file("extdata", "example.bed", package = "bigsnpr")
plink2 <- download_plink2(AVX2 = FALSE)

bedfile2 <- snp_plinkKINGQC(plink2, bedfile,
                           bedfile.out = tempfile(fileext = ".bed"),
                           ncores = 2)

df_re1 <- snp_plinkKINGQC(plink2, bedfile, make.bed = FALSE, ncores = 2)
str(df_re1)

## End(Not run)
```



snp\_plinkQC

*Quality Control***Description**

Quality Control (QC) and possible conversion to *bed/bim/fam* files using **PLINK 1.9**.

**Usage**

```
snp_plinkQC(
  plink.path,
  prefix.in,
  file.type = "--bfile",
  prefix.out = paste0(prefix.in, "_QC"),
  maf = 0.01,
  geno = 0.1,
  mind = 0.1,
  hwe = 1e-50,
  autosome.only = FALSE,
  extra.options = "",
  verbose = TRUE
)
```

**Arguments**

plink.path	Path to the executable of PLINK 1.9.
prefix.in	Prefix (path without extension) of the dataset to be QCed.
file.type	Type of the dataset to be QCed. Default is "--bfile" and corresponds to bed/bim/fam files. You can also use "--file" for ped/map files or "--vcf" for a VCF file. More information can be found at <a href="http://www.cog-genomics.org/plink/1.9/input">http://www.cog-genomics.org/plink/1.9/input</a> .
prefix.out	Prefix (path without extension) of the bed/bim/fam dataset to be created. Default is created by appending "_QC" to prefix.in.
maf	Minimum Minor Allele Frequency (MAF) for a SNP to be kept. Default is 0.01.
geno	Maximum proportion of missing values for a SNP to be kept. Default is 0.1.
mind	Maximum proportion of missing values for a sample to be kept. Default is 0.1.
hwe	Filters out all variants which have Hardy-Weinberg equilibrium exact test p-value below the provided threshold. Default is 1e-50.
autosome.only	Whether to exclude all unplaced and non-autosomal variants? Default is FALSE.
extra.options	Other options to be passed to PLINK as a string. More options can be found at <a href="http://www.cog-genomics.org/plink2/filter">http://www.cog-genomics.org/plink2/filter</a> . If using PLINK 2.0, you could e.g. use "--king-cutoff 0.0884" to remove some related samples at the same time of quality controls.
verbose	Whether to show PLINK log? Default is TRUE.

**Value**

The path of the newly created bedfile.

**References**

Chang, Christopher C, Carson C Chow, Laurent CAM Tellier, Shashaank Vattikuti, Shaun M Purcell, and James J Lee. 2015. *Second-generation PLINK: rising to the challenge of larger and richer datasets*. GigaScience 4 (1): 7. <http://dx.doi.org/10.1186/s13742-015-0047-8>.

**See Also**

[download\\_plink](#) [snp\\_plinkIBDQC](#)

**Examples**

```
## Not run:

bedfile <- system.file("extdata", "example.bed", package = "bigsnpr")
prefix <- sub_bed(bedfile)
plink <- download_plink()
test <- snp_plinkQC(plink.path = plink,
                   prefix.in = prefix,
                   prefix.out = tempfile(),
                   file.type = "--bfile", # the default (for ".bed")
                   maf = 0.05,
                   geno = 0.05,
                   mind = 0.05,
                   hwe = 1e-10,
                   autosome.only = TRUE)

test

## End(Not run)
```

---

snp\_plinkRmSamples      *Remove samples*

---

**Description**

Create new *bed/bim/fam* files by removing samples with PLINK.

**Usage**

```
snp_plinkRmSamples(
  plink.path,
  bedfile.in,
  bedfile.out,
  df.or.files,
```

```

col.family.ID = 1,
col.sample.ID = 2,
...,
verbose = TRUE
)

```

### Arguments

<code>plink.path</code>	Path to the executable of PLINK 1.9.
<code>bedfile.in</code>	Path to the input bedfile.
<code>bedfile.out</code>	Path to the output bedfile.
<code>df.or.files</code>	Either <ul style="list-style-type: none"> <li>• A data frame,</li> <li>• A character vector of file names where to find at the information you want. You should have one column for family IDs and one for sample IDs.</li> </ul>
<code>col.family.ID</code>	Index of the column containing the family IDs to match with those of the study. Default is the second one.
<code>col.sample.ID</code>	Index of the column containing the sample IDs to match with those of the study. Default is the first one.
<code>...</code>	Any additional parameter to pass to <code>bigreadr::fread2()</code> . Particularly, option <code>header = FALSE</code> is sometimes needed.
<code>verbose</code>	Whether to show PLINK log? Default is TRUE.

### Value

The path of the new bedfile.

### See Also

[download\\_plink](#)

---

snp\_PRS

*PRS*

---

### Description

Polygenic Risk Scores with possible clumping and thresholding.

**Usage**

```
snp_PRS(
  G,
  betas.keep,
  ind.test = rows_along(G),
  ind.keep = cols_along(G),
  same.keep = rep(TRUE, length(ind.keep)),
  lpS.keep = NULL,
  thr.list = 0
)
```

**Arguments**

G	A <a href="#">FBM.code256</a> (typically <code>&lt;bigSNP&gt;\$genotypes</code> ). <b>You shouldn't have missing values.</b> Also, remember to do quality control, e.g. some algorithms in this package won't work if you use SNPs with 0 MAF.
betas.keep	Numeric vector of weights associated with each SNP corresponding to <code>ind.keep</code> . You may want to see <a href="#">big_univLinReg</a> or <a href="#">big_univLogReg</a> .
ind.test	The individuals on whom to project the scores. Default uses all.
ind.keep	Column (SNP) indices to use (if using clumping, the output of <a href="#">snp_clumping</a> ). Default doesn't clump.
same.keep	A logical vector associated with <code>betas.keep</code> whether the reference allele is the same for G. Default is all TRUE (for example when you train the betas on the same dataset). Otherwise, use <a href="#">same_ref</a> .
lpS.keep	Numeric vector of $-\log_{10}(\text{p-value})$ associated with <code>betas.keep</code> . Default doesn't use thresholding.
thr.list	Threshold vector on <code>lpS.keep</code> at which SNPs are excluded if they are not significant enough. Default doesn't use thresholding.

**Value**

A matrix of scores, where rows correspond to `ind.test` and columns correspond to `thr.list`.

**Examples**

```
test <- snp_attachExtdata()
G <- big_copy(test$genotypes, ind.col = 1:1000)
CHR <- test$map$chromosome[1:1000]
POS <- test$map$physical.position[1:1000]
y01 <- test$fam$affection - 1

# PCA -> covariables
obj.svd <- snp_autoSVD(G, infos.chr = CHR, infos.pos = POS)

# train and test set
ind.train <- sort(sample(nrow(G), 400))
ind.test <- setdiff(rows_along(G), ind.train) # 117
```

```

# GWAS
gwas.train <- big_univLogReg(G, y01.train = y01[ind.train],
                           ind.train = ind.train,
                           covar.train = obj.svd$u[ind.train, ])

# clumping
ind.keep <- snp_clumping(G, infos.chr = CHR,
                       ind.row = ind.train,
                       S = abs(gwas.train$score))

# -log10(p-values) and thresholding
summary(lpS.keep <- -predict(gwas.train)[ind.keep])
thrs <- seq(0, 4, by = 0.5)
nb.pred <- sapply(thrs, function(thr) sum(lpS.keep > thr))

# PRS
prs <- snp_PRS(G, betas.keep = gwas.train$estim[ind.keep],
              ind.test = ind.test,
              ind.keep = ind.keep,
              lpS.keep = lpS.keep,
              thr.list = thrs)

# AUC as a function of the number of predictors
aucs <- apply(prs, 2, AUC, target = y01[ind.test])
library(ggplot2)
qplot(nb.pred, aucs) +
  geom_line() +
  scale_x_log10(breaks = nb.pred) +
  labs(x = "Number of predictors", y = "AUC") +
  theme_bigstatsr()

```

snp\_qq

*Q-Q plot***Description**

Creates a quantile-quantile plot from p-values from a GWAS study.

**Usage**

```
snp_qq(gwas, lambdaGC = TRUE, coeff = 1)
```

**Arguments**

gwas	A mhtest object with the p-values associated with each SNP. Typically, the output of <code>big_univLinReg</code> , <code>big_univLogReg</code> or <code>snp_pcadapt</code> .
lambdaGC	Add the Genomic Control coefficient as subtitle to the plot?
coeff	Relative size of text. Default is 1.

**Value**

A ggplot2 object. You can plot it using the print method. You can modify it as you wish by adding layers. You might want to read [this chapter](#) to get more familiar with the package **ggplot2**.

**Examples**

```
set.seed(9)

test <- snp_attachExtdata()
G <- test$genotypes
y <- rnorm(nrow(G))

gwas <- big_univLinReg(G, y)
snp_qq(gwas)
gwas_gc <- snp_gc(gwas) # change attr(gwas_gc, "transfo")

snp_qq(gwas_gc)
# The next plot should be prettier with a real dataset
snp_manhattan(gwas_gc,
               infos.chr = test$map$chromosome,
               infos.pos = test$map$physical.pos)

p <- snp_qq(gwas_gc) + ggplot2::aes(text = asPlotlyText(test$map))
## Not run: plotly::ggplotly(p, tooltip = "text")
```

---

snp\_readBed

*Read PLINK files into a "bigSNP"*


---

**Description**

Functions to read bed/bim/fam files into a [bigSNP](#).

**Usage**

```
snp_readBed(bedfile, backingfile = sub_bed(bedfile))

snp_readBed2(
  bedfile,
  backingfile = sub_bed(bedfile),
  ind.row = rows_along(obj.bed),
  ind.col = cols_along(obj.bed),
  ncores = 1
)
```

**Arguments**

bedfile            Path to file with extension ".bed" to read. You need the corresponding ".bim" and ".fam" in the same directory.

backingfile	The path (without extension) for the backing files for the cache of the <a href="#">bigSNP</a> object. Default takes the bedfile without the ".bed" extension.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .

### Details

For more information on these formats, please visit [PLINK webpage](#). For other formats, please use PLINK to convert them in bedfiles, which require minimal space to store and are faster to read. For example, to convert from a VCF file, use the `--vcf` option. See [snp\\_plinkQC](#).

### Value

The path to the RDS file that stores the bigSNP object. Note that this function creates one other file which stores the values of the Filebacked Big Matrix.

**You shouldn't read from PLINK files more than once.** Instead, use [snp\\_attach](#) to load the "bigSNP" object in any R session from backing files.

### Examples

```
(bedfile <- system.file("extdata", "example.bed", package = "bigsnpr"))

# Reading the bedfile and storing the data in temporary directory
rds <- snp_readBed(bedfile, backingfile = tempfile())

# Loading the data from backing files
test <- snp_attach(rds)

str(test)
dim(G <- test$genotypes)
G[1:8, 1:8]
```

---

snp\_readBGEN

*Read BGEN files into a "bigSNP"*


---

### Description

Function to read the UK Biobank BGEN files into a [bigSNP](#).

**Usage**

```
snp_readBGEN(
  bgenfiles,
  backingfile,
  list_snp_id,
  ind_row = NULL,
  bgi_dir = dirname(bgenfiles),
  read_as = c("dosage", "random"),
  ncores = 1
)
```

**Arguments**

bgenfiles	Character vector of paths to files with extension ".bgen". The corresponding ".bgen.bgi" index files must exist.
backingfile	The path (without extension) for the backing files for the cache of the <a href="#">bigSNP</a> object.
list_snp_id	List (same length as the number of BGEN files) of character vector of SNP IDs to read. These should be in the form "<chr>_<pos>_<a1>_<a2>" (e.g. "1_88169_C_T" or "01_88169_C_T"). <b>This function assumes that these IDs are uniquely identifying variants.</b>
ind_row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
bgi_dir	Directory of index files. Default is the same as bgenfiles.
read_as	How to read BGEN probabilities? Currently implemented: <ul style="list-style-type: none"> <li>• as dosages (rounded to two decimal places), the default,</li> <li>• as hard calls, randomly sampled based on those probabilities (similar to PLINK option '--hard-call-threshold random').</li> </ul>
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores()</a> .

**Details**

For more information on this format, please visit [BGEN webpage](#).

This function is designed to read UK Biobank imputation files. This assumes that variants have been compressed with zlib, that there are only 2 possible alleles, and that each probability is stored on 8 bits.

**Value**

The path to the RDS file that stores the bigSNP object. Note that this function creates one other file which stores the values of the Filebacked Big Matrix.

**You shouldn't read from BGEN files more than once.** Instead, use [snp\\_attach](#) to load the "bigSNP" object in any R session from backing files.



**Examples**

```
# See e.g. https://github.com/privefl/UKBiobank/blob/master/10-get-dosages.R
```

---

snp_readBGI	<i>Read variant info from one BGI file</i>
-------------	--

---

**Description**

Read variant info from one BGI file

**Usage**

```
snp_readBGI(bgifile, snp_id)
```

**Arguments**

bgifile	Path to one file with extension ".bgi".
snp_id	Character vector of SNP IDs. These should be in the form "<chr>_<pos>_<a1>_<a2>" (e.g. "1_88169_C_T" or "01_88169_C_T"). <b>This function assumes that these IDs are uniquely identifying variants.</b>

**Value**

A data frame containing variant information.

---

snp_save	<i>Save modifications</i>
----------	---------------------------

---

**Description**

Save a bigSNP after having made some modifications to it. As bigSNP is an S3 class, you can add any slot you want to an object of this class, then use snp\_save to save these modifications in the corresponding ".rds" backing file.

**Usage**

```
snp_save(x, version = NULL)
```

**Arguments**

x	A <a href="#">bigSNP</a> .
version	the workspace format version to use. NULL specifies the current default version (3). The only other supported value is 2, the default from R 1.4.0 to R 3.5.0.

**Value**

The (saved) bigSNP.

**Examples**

```
set.seed(1)

# Reading example
test <- snp_attachExtdata()

# I can add whatever I want to an S3 class
test$map$`p-values` <- runif(nrow(test$map))
str(test$map)

# Reading again
test.savedIn <- sub_bk(test$genotypes$backingfile, ".rds")
test2 <- snp_attach(rdsfile = test.savedIn)
str(test2$map) # new slot wasn't saved

# Save it
test <- snp_save(test)

# Reading again
test3 <- snp_attach(rdsfile = test.savedIn)
str(test3$map) # it is saved now

# The complicated code of this function
snp_save
```

---

snp\_scaleBinom

*Binomial(n, p) scaling*

---

**Description**

Binomial(n, p) scaling where n is fixed and p is estimated.

**Usage**

```
snp_scaleBinom(nploidy = 2)
```

**Arguments**

nploidy            Number of trials, parameter of the binomial distribution. Default is 2, which corresponds to diploidy, such as for the human genome.

**Details**

You will probably not use this function as is but as the `fun.scaling` parameter of other functions of package `bigstatsr`.

**Value**

A new **function** that returns a data.frame of two vectors "center" and "scale" which are of the length of ind.col.

**References**

This scaling is widely used for SNP arrays. Patterson N, Price AL, Reich D (2006). Population Structure and Eigenanalysis. PLoS Genet 2(12): e190. <http://dx.doi.org/10.1371/journal.pgen.0020190>.

**Examples**

```
set.seed(1)

a <- matrix(0, 93, 170)
p <- 0.2
a[] <- rbinom(length(a), 2, p)
X <- add_code256(big_copy(a, type = "raw"), code = c(0, 1, 2, rep(NA, 253)))
X.svd <- big_SVD(X, fun.scaling = snp_scaleBinom())
str(X.svd)
plot(X.svd$center)
abline(h = 2 * p, col = "red")
plot(X.svd$scale)
abline(h = sqrt(2 * p * (1 - p)), col = "red")
```

snp\_split

*Split-parApply-Combine***Description**

A Split-Apply-Combine strategy to parallelize the evaluation of a function on each SNP, independently.

**Usage**

```
snp_split(infos.chr, FUN, combine, ncores = 1, ...)
```

**Arguments**

infos.chr	Vector of integers specifying each SNP's chromosome. Typically <bigSNP>\$map\$chromosome.
FUN	The function to be applied. It must take a <a href="#">FBM.code256</a> as first argument and ind.chr, an another argument to provide subsetting over SNPs. You can access the number of the chromosome by using attr(ind.chr, "chr").

combine	function that is used by <a href="#">foreach</a> to process the tasks results as they generated. This can be specified as either a function or a non-empty character string naming the function. Specifying 'c' is useful for concatenating the results into a vector, for example. The values 'cbind' and 'rbind' can combine vectors into a matrix. The values '+' and '*' can be used to process numeric data. By default, the results are returned in a list.
ncores	Number of cores used. Default doesn't use parallelism. You may use <a href="#">nb_cores</a> .
...	Extra arguments to be passed to FUN.

### Details

This function splits indices for each chromosome, then apply a given function to each part (chromosome) and finally combine the results.

### Value

The result of [foreach](#).

### Examples

```
# parallelize over chromosomes made easy
# examples of functions from this package
snp_pruning
snp_clumping
snp_fastImpute
```

---

snp\_subset

*Subset a bigSNP*

---

### Description

Subset (copy) of a bigSNP, also stored on disk.

### Usage

```
snp_subset(
  x,
  ind.row = rows_along(x$fam),
  ind.col = rows_along(x$map),
  backingfile = NULL
)

## S3 method for class 'bigSNP'
subset(
  x,
  ind.row = rows_along(x$fam),
```

```

    ind.col = rows_along(x$map),
    backingfile = NULL,
    ...
)

```

### Arguments

x	A <a href="#">bigSNP</a> .
ind.row	Indices of the rows (individuals) to keep. Negative indices <b>can</b> be used to exclude row indices. Default: keep them all.
ind.col	Indices of the columns (SNPs) to keep. Negative indices <b>can</b> be used to exclude column indices. Default: keep them all.
backingfile	Prefix of the two new files created (".bk" and ".rds"). By default, it is automatically determined by appending "_sub" and a number to the prefix of the input bigSNP backing files.
...	Not used.

### Value

The path to the RDS file that stores the bigSNP object.

### See Also

[bigSNP](#)

### Examples

```

str(test <- snp_attachExtdata())

# keep only first 50 samples and SNPs
rdsfile <- snp_subset(test, ind.row = 1:50, ind.col = 1:50)
str(snp_attach(rdsfile))

# remove only first 50 samples and SNPs
rdsfile2 <- snp_subset(test, ind.row = -(1:50), ind.col = -(1:50))
str(snp_attach(rdsfile2))

```

---

snp\_writeBed

*Write PLINK files from a "bigSNP"*

---

### Description

Function to write bed/bim/fam files from a [bigSNP](#). This will use the slot code **rounded** to write 0s, 1s, 2s or NAs.

**Usage**

```
snp_writeBed(x, bedfile, ind.row = rows_along(G), ind.col = cols_along(G))
```

**Arguments**

x	A <a href="#">bigSNP</a> .
bedfile	Path to file with extension ".bed" to create.
ind.row	An optional vector of the row indices (individuals) that are used. If not specified, all rows are used. <b>Don't use negative indices.</b>
ind.col	An optional vector of the column indices (SNPs) that are used. If not specified, all columns are used. <b>Don't use negative indices.</b>

**Value**

The input bedfile path.

**Examples**

```
N <- 17
M <- 911

fake <- snp_fake(N, M)
G <- fake$genotypes
G[] <- sample(as.raw(0:3), size = length(G), replace = TRUE)

# Write the object as a bed/bim/fam object
tmp <- tempfile(fileext = ".bed")
bed <- snp_writeBed(fake, tmp)

# Read this new file for the first time
rds <- snp_readBed(bed, backingfile = tempfile())
# Attach object in R session
fake2 <- snp_attach(rds)

# Same content
all.equal(fake$genotypes[], fake2$genotypes[])
all.equal(fake$fam, fake2$fam)
all.equal(fake$map, fake2$map)

# Two different backingfiles
fake$genotypes$backingfile
fake2$genotypes$backingfile
```

---

sub_bed	<i>Replace extension '.bed'</i>
---------	---------------------------------

---

**Description**

Replace extension '.bed'

**Usage**

```
sub_bed(path, replacement = "", stop_if_not_ext = TRUE)
```

**Arguments**

path	String with extension '.bed'.
replacement	Replacement of '.bed'. Default replaces by nothing. Can be useful to replace e.g. by '.bim' or '.fam'.
stop_if_not_ext	If replacement != "", whether to error if replacement is not an extension (starting with a '.').

**Value**

String with extension '.bed' replaced by replacement.

**Examples**

```
path <- "toto.bed"
sub_bed(path)
sub_bed(path, ".bim")
sub_bed(path, ".fam")
sub_bed(path, "_QC", stop_if_not_ext = FALSE)
```

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