

# Package ‘ldblock’

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**Title** data structures for linkage disequilibrium measures in populations

**Version** 1.39.1

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**Description** Define data structures for linkage disequilibrium measures in populations.

**Suggests** RUnit, knitr, BiocStyle, gwascats, rmarkdown, snpStats, VariantAnnotation, GenomeInfoDb, ensemblDb, EnsDb.Hsapiens.v75, Rsamtools, GenomicFiles (>= 1.13.6)

**Imports** BiocGenerics (>= 0.25.1), Seqinfo, httr, Matrix

**Depends** R (>= 3.5), methods, rlang

**Maintainer** VJ Carey <stvjc@channing.harvard.edu>

**License** Artistic-2.0

**LazyData** no

**BiocViews** genetics, SNP, GWAS, LinkageDisequilibrium

**VignetteBuilder** knitr

**RoxygenNote** 7.3.2

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|                 |                                                                                                                                                          |
|-----------------|----------------------------------------------------------------------------------------------------------------------------------------------------------|
| ldblock-package | <i>c("\Sexpr[results=rd,stage=build]tools::Rd_package_title(\"&gt;#1\"),"ldblock")data structures for linkage disequilibrium measures in populations</i> |
|-----------------|----------------------------------------------------------------------------------------------------------------------------------------------------------|

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**Description**

c("\Sexpr[results=rd,stage=build]tools::Rd\_package\_description(\">#1\"),"ldblock")Define data structures for linkage disequilibrium measures in populations.

**Details**

The DESCRIPTION file: c("\Sexpr[results=rd,stage=build]tools::Rd\_package\_DESCRIPTION(\">#1\"),"ldblock")This package was not yet installed at build time.\cr c("\Sexpr[results=rd,stage=build]tools::Rd\_package\_index(\">#1\"),"ldblock") Index: This package was not yet installed at build time.\cr

**Author(s)**

c("\Sexpr[results=rd,stage=build]tools::Rd\_package\_author(\">#1\"),"ldblock")VJ Carey <stvjc@channing.harvard.edu>  
Maintainer: c("\Sexpr[results=rd,stage=build]tools::Rd\_package\_maintainer(\">#1\"),"ldblock")VJ Carey <stvjc@channing.harvard.edu>

**Examples**

# see vignette

---

|                  |                                                   |
|------------------|---------------------------------------------------|
| downloadPopByChr | <i>download hapmap resource with LD estimates</i> |
|------------------|---------------------------------------------------|

---

**Description**

download hapmap resource with LD estimates

**Usage**

```
downloadPopByChr(  
  chrname = "chr1",  
  popname = "CEU",  
  
  urlTemplate = "http://hapmap.ncbi.nlm.nih.gov/downloads/ld_data/2009-02_phaseIII_r2/ld_%CHRN%",  
  targfolder = Sys.getenv("LDBLOCK_TXTGZ_DIR")  
)
```

**Arguments**

|             |                                                     |
|-------------|-----------------------------------------------------|
| chrname     | UCSC format tag for chromosome                      |
| popname     | hapmap three letter code for population, e.g. 'CEU' |
| urlTemplate | pattern for creating URL given chr and pop          |
| targfolder  | destination                                         |

**Details**

delivers HapMap LD data to 'targfolder'

**Value**

just run for side effect of download.file

**Examples**

```
## Not run:  
downloadPopByChr()  
  
## End(Not run)
```

---

|                |                            |
|----------------|----------------------------|
| EUR_singletons | <i>singletons from EUR</i> |
|----------------|----------------------------|

---

**Description**

singletons from EUR

**Usage**

```
EUR_singletons
```

**Format**

character vector

**Source**

[ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/working/20130606\\_sample\\_info/20130606\\_sample\\_info.xlsx](ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/working/20130606_sample_info/20130606_sample_info.xlsx), to which superpopulation codes were added

---

|              |                                                                                        |
|--------------|----------------------------------------------------------------------------------------|
| expandSnpSet | <i>Given a set of SNP identifiers, use LD to expand the set to include linked loci</i> |
|--------------|----------------------------------------------------------------------------------------|

---

### Description

Given a set of SNP identifiers, use LD to expand the set to include linked loci

### Usage

```
expandSnpSet(
  rsl,
  lb = 0.8,
  ldstruct,
  chrn = "chr17",
  popn = "CEU",
  txtgzfn = dir(system.file("hapmap", package = "ldblock"), full.names = TRUE)
)
```

### Arguments

|          |                                                                                                                                              |
|----------|----------------------------------------------------------------------------------------------------------------------------------------------|
| rsl      | input list – SNPs not found in the LD structure are simply returned along with those found, and the expansion list, all combined in a vector |
| lb       | lower bound on statistic used to retrieve loci in LD                                                                                         |
| ldstruct | instance of <a href="#">ldstruct-class</a>                                                                                                   |
| chrn     | chromosome identifier                                                                                                                        |
| popn     | population identifier (one of 'CEU', 'MEX', ...)                                                                                             |
| txtgzfn  | path to gzipped hapmap file with LD information                                                                                              |

### Details

direct use of elementwise arithmetic comparison

### Value

character vector

### Note

As of 2015, it appears that locus names are more informative than addresses for determining SNP identity across resources.

### Examples

```
og = Sys.getenv("LDBLOCK_TXTGZ_DIR")
on.exit( Sys.setenv("LDBLOCK_TXTGZ_DIR" = og ) )
Sys.setenv("LDBLOCK_TXTGZ_DIR"=system.file("hapmap", package="ldblock"))
ld17 = hmlld(chr="chr17", pop="CEU")
ee = expandSnpSet( ld17@allrs[1:10], ldstruct = ld17 )
```

---

|      |                                                                                                                                                                                            |
|------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| hmlD | <i>import hapmap LD data and create a structure for its management; generates a sparse matrix representation of pairwise LD statistics and binds metadata on variant name and position</i> |
|------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|

---

## Description

import hapmap LD data and create a structure for its management; generates a sparse matrix representation of pairwise LD statistics and binds metadata on variant name and position

## Usage

```
hmlD(hmgztxt, poptag, chrom, genome = "hg19", stat = "Dprime")
```

## Arguments

|         |                                                                                                                                                                                                                                                       |
|---------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| hmgztxt | name of gzipped text file as distributed at <a href="http://hapmap.ncbi.nlm.nih.gov/downloads/ld_data/2009-02_phaseIII_r2/">hapmap.ncbi.nlm.nih.gov/downloads/ld_data/2009-02_phaseIII_r2/</a> . It will be processed by <a href="#">read.delim</a> . |
| poptag  | heuristic tag identifying population                                                                                                                                                                                                                  |
| chrom   | heuristic tag for chromosome name                                                                                                                                                                                                                     |
| genome  | genome tag                                                                                                                                                                                                                                            |
| stat    | statistic to use, "Dprime", "R2", and "LOD" are options                                                                                                                                                                                               |

## Value

instance of ldstruct class

## Examples

```
getClass("ldstruct")
# see vignette
```

---

|          |                                                                  |
|----------|------------------------------------------------------------------|
| ldByGene | <i>Obtain LD statistics in region specified by a gene model.</i> |
|----------|------------------------------------------------------------------|

---

## Description

Obtain LD statistics in region specified by a gene model.

## Usage

```
ldByGene(
  sym = "MMP24",
  vcf = system.file("vcf/c20exch.vcf.gz", package = "ldblock"),
  flank = 1000,
  vcfSLS = "NCBI",
  genomeSLS = "hg19",
  stats = "D.prime",
  depth = 10
)
```

**Arguments**

|           |                                                                   |
|-----------|-------------------------------------------------------------------|
| sym       | A standard gene symbol for use with <code>genemodel</code>        |
| vcf       | Path to a tabix-indexed VCF file                                  |
| flank     | number of basepairs to flank gene model for search                |
| vcfSLS    | seqlevelsStyle (SLS) token for VCF; will be imposed on gene model |
| genomeSLS | character tag for genome, to be used with <code>readVcf</code>    |
| stats     | passed to <code>ld</code>                                         |
| depth     | passed to <code>ld</code>                                         |

**Value**

sparse matrix representation of selected LD statistic, as returned by `ld`

**Note**

Uses an internal function `genemod4ldblock`, that relies on `EnsDb.Hsapiens.v75` to get gene model.

**Examples**

```
if (interactive()) { # there is a warning owing to non-SNV present
  ld1 = ldByGene(depth=150)
  image(ld1[1:200,1:200], col.reg=heat.colors(120), colorkey=TRUE,
        main="SNPs in MMP24 (chr20)")
}
```

---

ldmat

---

*use LDmat API from NCI LDlink service*


---

**Description**

use LDmat API from NCI LDlink service

**Usage**

```
ldmat(rsvec, pop = "CEU", type = "d", token = Sys.getenv("LDLINK_TOKEN"))
```

**Arguments**

|       |                                                                                                    |
|-------|----------------------------------------------------------------------------------------------------|
| rsvec | character vector of SNP ids                                                                        |
| pop   | three letter code for HapMap population, defaults to CEU                                           |
| type  | 'r2' or 'd', defaults to 'd' implying d-prime                                                      |
| token | the API token provided by NCI, defaults to value of environment variable <code>LDLINK_TOKEN</code> |

**Value**

data.frame

**Examples**

```
if (interactive()) ldmat(c("rs77749396", "rs9303279", "rs9303280", "rs9303281"))
```

---

ldmat, ldstruct-method    *accessor for matrix component*

---

### Description

accessor for matrix component

### Usage

```
## S4 method for signature 'ldstruct'
ldmat(x)
```

### Arguments

x                      instance of ldstruct

---

ldstruct-class            *container for LD data*

---

### Description

Manage information about LD statistics as reported by HapMap.

### Objects from the Class

Objects can be created by calls of the form `new("ldstruct", ...)`.

### Examples

```
showClass("ldstruct")
```

---

|        |                                                                                                                                                                                                                           |
|--------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| s3_1kg | <i>Create a URL referencing 1000 genomes content in AWS S3. stack1kg produces a VcfStack instance with references to VCF for 1000 genomes autosomal chrs. S3-resident VCF files with version "v5a.20130502" are used.</i> |
|--------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|

---

### Description

Create a URL referencing 1000 genomes content in AWS S3. stack1kg produces a VcfStack instance with references to VCF for 1000 genomes autosomal chrs. S3-resident VCF files with version "v5a.20130502" are used.

### Usage

```
s3_1kg(chrnum, tmp1, dropchr = TRUE)
```

**Arguments**

|         |                                                                                   |
|---------|-----------------------------------------------------------------------------------|
| chrnum  | a character string denoting a chromosome, such as '22'                            |
| tpl     | alternate template for full URL, useful if versions prior to 2010 are of interest |
| dropchr | if TRUE chrnum will have 'chr' removed if present                                 |

**Value**

by default, a TabixFile instance

**Note**

The "wrap" parameter has been removed. A TabixFile structure will be returned. The tag parameter has been removed. Supply a tpl argument if you are not using 20130502 version.

**Examples**

```
requireNamespace("Rsamtools")
s3_1kg("22") # try scanVcfHeader from VariantAnnotation
```

---

|             |                                                                 |
|-------------|-----------------------------------------------------------------|
| sampinf_1kg | <i>population and relationship information for 1000 genomes</i> |
|-------------|-----------------------------------------------------------------|

---

**Description**

population and relationship information for 1000 genomes

**Usage**

```
sampinf_1kg
```

**Format**

```
data.frame
```

**Source**

[ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/working/20130606\\_sample\\_info/20130606\\_sample\\_info.xlsx](ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/working/20130606_sample_info/20130606_sample_info.xlsx), to which superpopulation codes were added



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|          |                                        |
|----------|----------------------------------------|
| stack1kg | <i>couple together a group of VCFs</i> |
|----------|----------------------------------------|

---

**Description**

couple together a group of VCFs

**Usage**

```
stack1kg(chrs = as.character(1:22), index = FALSE, useEBI = FALSE)
```

**Arguments**

|        |                                                                                                                                                                               |
|--------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| chrs   | a vector of chromosome names for extraction from 1000 genomes VCF collection                                                                                                  |
| index  | logical telling whether VcfStack should attempt to create the local index; for 1000 genomes, the tbi are in the cloud and will be used by readVcf so FALSE is appropriate     |
| useEBI | logical(1) defaults to FALSE ... if TRUE, use tabix-indexed vcf from EBI, but in July 2022 the EBI FTP site does not respond. If FALSE, the AWS Open Data access path is used |

**Value**

VcfStack instance

**Note**

The seqinfo component of returned stack will have NA for genome. Please set it manually; for useEBI=TRUE this would be GRCh38; very likely so for useEBI=FALSE, but this should be checked.

**Examples**

```
if (interactive()) {  
  st1 = stack1kg()  
  st1  
}
```

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