

Package ‘ldblock’

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Title data structures for linkage disequilibrium measures in populations

Version 1.32.0

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Description Define data structures for linkage disequilibrium measures in populations.

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

Imports BiocGenerics (>= 0.25.1), httr, Matrix

Depends R (>= 3.5), methods, rlang

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License Artistic-2.0

LazyData no

BiocViews genetics, SNP, GWAS, LinkageDisequilibrium

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ldblock-package	<i>c("\Sexpr[results=rd,stage=build]tools:::Rd_package_title(\`#\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")`

```
{ Package: \tab ldblock\cr Title: \tab data structures for linkage disequilibrium measures in populations\cr Version: \tab 1.32.0\cr Author: \tab VJ Carey <stvjc@channing.harvard.edu>\cr Description: \tab Define data structures for linkage disequilibrium measures in populations.\cr Suggests: \tab RUnit, knitr, BiocStyle, gwascats, rmarkdown, snpStats, VariantAnnotation, GenomeInfoDb, ensemblDb, EnsDb.Hsapiens.v75, Rsamtools, GenomicFiles (>= 1.13.6)\cr Imports: \tab BiocGenerics (>= 0.25.1), httr, Matrix\cr Depends: \tab R (>= 3.5), methods, rlang\cr Maintainer: \tab VJ Carey <stvjc@channing.harvard.edu>\cr License: \tab Artistic-2.0\cr LazyData: \tab no\cr BiocViews: \tab genetics, SNP, GWAS, LinkageDisequilibrium\cr VignetteBuilder: \tab knitr\cr RoxygenNote: \tab 7.2.0\cr Encoding: \tab UTF-8\cr git_url: \tab https://git.bioconductor.org/packages/ldblock\cr git_branch: \tab RELEASE_3_18\cr git_last_commit: \tab 398acc9\cr git_last_commit_date: \tab 2023-10-24\cr Date/Publication: \tab 2023-10-24\cr }
```

`c("\Sexpr[results=rd,stage=build]tools:::Rd_package_indices(\`#\1\`)", "ldblock")` Index of help topics: `\preformatted{ EUR_singletons singletons from EUR downloadPopByChr download hapmap resource with LD estimates expandSnpSet Given a set of SNP identifiers, use LD to expand the set to include linked loci hmlD 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 ldByGene Obtain LD statistics in region specified by a gene model. ldblock-package c("\Sexpr[results=rd,stage=build]tools:::Rd_package_title(\`#\1\`)", "ldblock")data structures for linkage disequilibrium measures in populations ldmat use LDmat API`

from NCI LDlink service ldmatrix,ldstruct-method accessor for matrix component ldstruct-class container for LD data s3_1kg 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. sampinf_1kg population and relationship information for 1000 genomes stack1kg couple together a group of VCFs }

Author(s)

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Examples

see vignette

downloadPopByChr	<i>download hapmap resource with LD estimates</i>
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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, 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

rs1	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 <code>ldstruct-class</code>
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 = hmlD(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 hapmap.ncbi.nlm.nih.gov/downloads/ld_data/2009-02_phaseIII_r2/ . It will be processed by read.delim .
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

Obtain LD statistics in region specified by a gene model.

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	<i>use LDmat API from NCI LDlink service</i>
-------	--

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 LDLINK_TOKEN |

Value

data.frame

Examples

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

ldmat,ldstruct-method	<i>accessor for matrix component</i>
-----------------------	--------------------------------------

Description

accessor for matrix component

Usage

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

Arguments

x instance of ldstruct

ldstruct-class	<i>container for LD data</i>
----------------	------------------------------

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'

tmp1 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 tmp1 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, to which superpopulation codes were added

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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