Package 'ldblock'

May 19, 2021

Title data structures for linkage disequilibrium measures in populations

Version 1.22.0

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Description Define data structures for linkage disequilibrium measures in populations.

Suggests RUnit, knitr, BiocStyle, gwascat

Imports Matrix, snpStats, VariantAnnotation, GenomeInfoDb, httr,
ensembldb, EnsDb.Hsapiens.v75,
Rsamtools, GenomicFiles (>= 1.13.6), BiocGenerics (>= 0.25.1)

Depends R (>= 3.5), methods

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License Artistic-2.0

LazyLoad yes

LazyData yes

BiocViews genetics, SNP, GWAS, LinkageDisequilibrium

VignetteBuilder knitr

RoxygenNote 7.1.1

Encoding UTF-8

git_url https://git.bioconductor.org/packages/ldblock

git_branch RELEASE_3_13

git_last_commit 86439b2

git_last_commit_date 2021-05-19

Date/Publication 2021-05-19

R topics documented:

`ldblock-package` *c("\Sexpr[results=rd,stage=build]tools:::Rd_package_title(\"#1\")", "ldblock")data structures for linkage disequilibrium measures in populations*

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")\tabular{ll}{ Package: \tab ldblock\cr Title: \tab data structures for linkage disequilibrium measures in populations\cr Version: \tab 1.22.0\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, gwascat\cr Imports: \tab Matrix, snpStats, VariantAnnotation, GenomeInfoDb, httr, ensembldb, EnsDb.Hsapiens.v75, Rsamtools, GenomicFiles (>= 1.13.6), BiocGenerics (>= 0.25.1)\cr Depends: \tab R (>= 3.5), methods\cr Maintainer: \tab VJ Carey <stvjc@channing.harvard.edu>\cr License: \tab Artistic-2.0\cr LazyLoad: \tab yes\cr LazyData: \tab yes\cr BiocViews: \tab genetics, SNP, GWAS, LinkageDisequilibrium\cr VignetteBuilder: \tab knitr\cr RoxygenNote: \tab 7.1.1\cr Encoding: \tab UTF-8\cr git_url: \tab https://git.bioconductor.org/packages/ldblock\cr git_branch: \tab RELEASE_3_13\cr git_last_commit: \tab 86439b2\cr git_last_commit_date: \tab 2021-05-19\cr Date/Publication: \tab 2021-05-19\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 ldmat,ldstruct-method accessor for matrix component ldstruct-class Class '"ldstruct"' s3_1kg Create a URL referencing 1000 genomes content in AWS S3. 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 *download hapmap resource with LD estimates*

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 *singletons from EUR*

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

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 ldstruct-class
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 — *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*

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 genemodel
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 readVcf
stats	passed to ld
depth	passed to ld

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 LDLINK_TOKEN

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 *Class "ldstruct"*

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 *Create a URL referencing 1000 genomes content in AWS S3.*

Description

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,
  tag = "20130502",
  wrap = function(x) TabixFile(x),
  tmpl = NULL,
  dropchr = TRUE
)
```

Arguments

chrnum	a character string denoting a chromosome, such as '22'
tag	a character string identifying the version, ignored if tmpl is non-null; valid tag values are the default or "20101123"
wrap	The URL is returned after evaluating wrap on it; default is useful when Tabix indexing is to be used
tmpl	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

Examples

```
s3_1kg("22")
## Not run:
 require(VariantAnnotation)
 scanVcfHeader(s3_1kg("22"))

## End(Not run)
```

---

sampinf_1kg *population and relationship information for 1000 genomes*

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` *couple together a group of VCFs*

Description

couple together a group of VCFs

Usage

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

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 TRUE ... use tabix-indexed vcf from EBI

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.

Examples

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

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