

ldblock package: linkage disequilibrium data structures

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Contents

1	Introduction	2
2	Import of HapMap LD data	2
3	A view of the block structure	2
4	Collecting SNPs exhibiting linkage to selected SNP	3

1 Introduction

There is a nice vignette in *snpStats* concerning linkage disequilibrium (LD) analysis as supported by software in that package. The purpose of this package is to simplify handling of existing population-level data on LD for the purpose of flexibly defining LD blocks.

2 Import of HapMap LD data

The `hmld` function imports gzipped tabular data from hapmap's repository hapmap.ncbi.nlm.nih.gov/downloads/ld_data/2009-02_phaseIII_r2/.

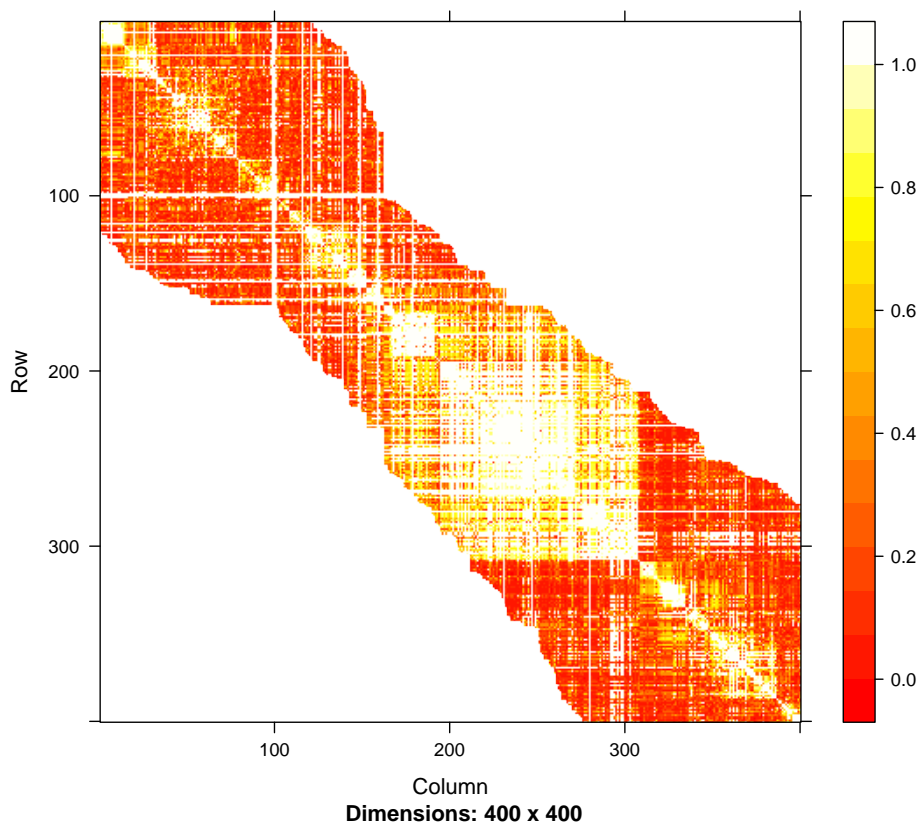
```
suppressPackageStartupMessages(library(ldblock))
path = dir(system.file("hapmap", package="ldblock"), full=TRUE)
ceu17 = hmld(path, poptag="CEU", chrom="chr17")
## importing /tmp/RtmpTPPBXD/Rinst3eed5eaa8677/ldblock/hapmap/ld_chr17-CEU.txt.gz
## done.
ceu17
## ldstruct for population CEU, chrom chr17
## dimensions: 36621 x 36621 ; statistic is Dprime
## object structure:
##      ldmatrix      chrom      genome      allpos      poptag      statInUse
## "dsCMatrix" "character" "character" "numeric" "character" "character"
##      allrs
## "character"
```

3 A view of the block structure

For some reason knitr/render will not display this image nicely.

```
library(Matrix)
##
## Attaching package: 'Matrix'
## The following object is masked from 'package:S4Vectors':
##
##      expand
image(ceu17@ldmatrix[1:400,1:400],
      col.reg=heat.colors(120), colorkey=TRUE, useRaster=TRUE)
```

ldblock package: linkage disequilibrium data structures



This ignores physical distance and MAF. The bright stripes are probably due to SNP with low MAF.

4 Collecting SNPs exhibiting linkage to selected SNP

We'll use `ceu17` and the `gwascat` package to enumerate SNP that are in LD with GWAS hits.

```
library(gwascat)
## gwascat loaded. Use data(ebicat38) for hg38 coordinates;
## data(ebicat37) for hg19 coordinates.
data(ebicat37)
seqlevelsStyle(ebicat37) = "NCBI"
## Warning in scan(file = file, what = what, sep = sep, quote = quote, dec =
## dec, : EOF within quoted string
e17 = ebicat37[ which(as.character(seqnames(ebicat37)) == "17") ]
```

Some dbSNP names for GWAS hits on chr17 are

```
rsh17 = unique(e17$SNPS)
head(rsh17)
## [1] "rs11078895" "rs11891" "rs7501939" "rs9905704" "rs4796793"
## [6] "rs78378222"
```

ldblock package: linkage disequilibrium data structures

We will use `expandSnpSet` to obtain names for SNP that were found in HapMap CEU to have which $D' > .9$ with any of these hits. These names are added to the input set.

```
length(rsh17)
## [1] 530
exset = expandSnpSet( rsh17, ldstruct= ceu17, lb=.9 )
## Warning in expandSnpSet(rsh17, ldstruct = ceu17, lb = 0.9): dropping 149 rsn
## not matched in ld matrix
length(exset)
## [1] 13209
all(rsh17 %in% exset)
## [1] TRUE
```

Not all GWAS SNP are in the HapMap LD resource. You can use your own LD data as long as the format agrees with that of the HapMap distribution.