MafDb.1Kgenomes.phase3.hs37d5

October 20, 2017

MafDb.1Kgenomes.phase3.hs37d5-package

Annotation package for minor allele frequency data from the 1000 Genomes Project Phase 3

Description

This annotation package stores minor allele frequency (MAF) data from the Phase 3 of the 1000 Genomes Project. The data are exposed to the user in the form of a MafDb object, named after the package and loaded into memory only as different chromosomes and populations are being queried. The class definition and methods to access MafDb objects are found in the VariantFiltering software package. To minimize disk space and memory requirements, MAF values larger or equal than 0.1 are stored using two significant digits, while MAF values smaller than 0.1 are stored using one significant digit.

Please consult the 1000 Genomes Project FAQ page at http://www.internationalgenome.org/faq before you use these data for your own research.

Format

MafDb .1Kgenomes .phase 3 . hs37d5 MafDb object containing MAF values from the 1000 Genomes Project Phase 3 down

Author(s)

R. Castelo

Source

The 1000 Genomes Project Consortium. A global reference for human genetic variation. Nature, 526:68-74, 2015.

The International Genome Sample Resource (IGSR), Hinxton, UK (URL: http://www.internationalgenome.org) [March, 2017, accessed]

See Also

MafDb-class mafByOverlaps mafById VariantFiltering

Examples

```
library(MafDb.1Kgenomes.phase3.hs37d5)

ls("package:MafDb.1Kgenomes.phase3.hs37d5")

mafdb <- MafDb.1Kgenomes.phase3.hs37d5
mafdb

populations(mafdb)

## lookup allele frequencies for rs1129038, a SNP associated to blue and brown eye colors
## as reported by Eiberg et al. Blue eye color in humans may be caused by a perfectly associated
## founder mutation in a regulatory element located within the HERC2 gene inhibiting OCA2 expression.
## Human Genetics, 123(2):177-87, 2008 [http://www.ncbi.nlm.nih.gov/pubmed/18172690]
rng <- GRanges("15", IRanges(28356859, 28356859))
mafByOverlaps(mafdb, rng)
mafByOverlaps(mafdb, "15:28356859-28356859")
mafByOverlaps(mafdb, "15:28356859")
mafByOverlaps(mafdb, "rs1129038")</pre>
```

Index

```
*Topic data
MafDb.1Kgenomes.phase3.hs37d5-package,

1
*Topic package
MafDb.1Kgenomes.phase3.hs37d5-package,

1
mafById, 2
mafByOverlaps, 2
MafDb, I
MafDb-class, 2
MafDb.1Kgenomes.phase3.hs37d5

(MafDb.1Kgenomes.phase3.hs37d5-package),

1
MafDb.1Kgenomes.phase3.hs37d5-package,

1
VariantFiltering, I, 2
```