MafDb.1Kgenomes.phase3.hs37d5

February 28, 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 for single derived from the single nucleotide variant set of the Phase 3 of the 1000 Genomes Project. The data is loaded automatically in the form of a MafDb2 object. The name of the exposed object matches the name of the package and part of the filename that contained the data imported into the package. The class definition and methods to access MafDb2 objects are found in the VariantFiltering software package.

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

Format

MafDb.1Kgenomes.phase3.hs37d5 MafDb2 object containing MAF values from the 1000 Genomes Project Phase 3 do

Author(s)

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Source

The 1000 Genomes Project Consortium. A global reference for human genetic variation. Nature, 526:68-74, 2015. (URL: http://www.1000genomes.org) [July, 2016, accessed]

See Also

MafDb2-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>
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