MafDb.1Kgenomes.phase1.GRCh38

March 17, 2021

`MafDb.1Kgenomes.phase1.GRCh38-package`
Annotation package for minor allele frequency data from the 1000 Genomes Project Phase 1

Description

This annotation package stores minor allele frequency (MAF) data from the Phase 1 of the 1000 Genomes Project. The data are exposed to the user in the form of a `GScores` object, named after the package and loaded into main memory only as different chromosomes and populations are being queried. The class definition and methods to access `GScores` objects are found in the `GenomicScores` 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 on your own research.

Format

`MafDb.1Kgenomes.phase1.GRCh38` `GScores` object containing MAF values from the 1000 Genomes Project Phase 1 do

Author(s)

R. Castelo

Source

The 1000 Genomes Project Consortium. An integrated map of genetic variation from 1,092 human genomes. Nature, 491:56-65, 2012.

The International Genome Sample Resource (IGSR), Hinxton, UK (URL: `http://www.internationalgenome.org`) [March, 2018, accessed]

See Also

GScores-class gscores GenomicScores

Examples

```
library(SNPlocs.Hsapiens.dbSNP149.GRCh38)
library(MafDb.1Kgenomes.phase1.GRCh38)

ls("package:MafDb.1Kgenomes.phase1.GRCh38")

mafdb <- MafDb.1Kgenomes.phase1.GRCh38
mafdb
citation(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]

snpdb <- SNPlocs.Hsapiens.dbSNP149.GRCh38
rng <- snpsById(snpdb, ids="rs1129038")
rng
gscores(mafdb, rng)
gscores(mafdb, GRanges("15:28111713"))
```

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