MafDb.gnomAD.r2.1.hs37d5

April 23, 2025

MafDb.gnomAD.r2.1.hs37d5-package

Annotation package for minor allele frequency data from the Genome Aggregation Database

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Description

This annotation package stores minor allele frequency (MAF) data derived from the whole genome variant set release 2.1 of the Genome Aggregation Database (gnomAD). 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 gnomAD FAQ page at http://gnomad.broadinstitute.org/faq before you use these data for your own research.

Format

MafDb.gnomAD.r2.1.hs37d5   GScores object containing MAF values from gnomAD downloaded on March 2018 from http

Author(s)

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Source

Karczewski et al. Variation across 141,456 human exomes and genomes reveals the spectrum of loss-of-function intolerance across human protein-coding genes. bioRxiv, 531210, 2019.

The Genome Aggregation Database (gnomAD), Cambridge, MA (URL: http://gnomad.broadinstitute.org) [March, 2018, accessed]

See Also

[GScores-class gscores GenomicScores](#)

Examples

```
library(SNPlocs.Hsapiens.dbSNP144.GRCh37)
library(MafDb.gnomAD.r2.1.hs37d5)

ls("package:MafDb.gnomAD.r2.1.hs37d5")

mafdb <- MafDb.gnomAD.r2.1.hs37d5
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.dbSNP144.GRCh37
rng <- snpsById(snpdb, ids="rs1129038")
rng
gscores(mafdb, rng)
gscores(mafdb, GRanges("15:28356859"))
```

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