MafDb.gnomADex.r2.1.hs37d5

April 22, 2024

`MafDb.gnomADex.r2.1.hs37d5-package`
Annotation package for minor allele frequency data from exomes of the Genome Aggregation Database

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Description

This annotation package stores minor allele frequency (MAF) data derived from the exome 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.gnomADex.r2.1.hs37d5` `GScores` object containing MAF values from gnomAD exomes downloaded on October 20

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`) [October, 2018, accessed]

See Also

GScores-class gscores GenomicScores

Examples

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

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

mafdb <- MafDb.gnomADex.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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