MafDb.gnomAD.r2.1.GRCh38

April 12, 2023

`MafDb.gnomAD.r2.1.GRCh38-package`
Annotation package for minor allele frequency data from the Genome Aggregation Database

---

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.GRCh38` `GScores` object containing MAF values from gnomAD genomes downloaded on April 2019 fi

Author(s)

R. Castelo

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) [April 2019, accessed]

See Also

GScores-class gscores GenomicScores

Examples

```
library(SNPlocs.Hsapiens.dbSNP149.GRCh38)
library(MafDb.gnomAD.r2.1.GRCh38)

ls("package:MafDb.gnomAD.r2.1.GRCh38")

mafdb <- MafDb.gnomAD.r2.1.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"))
```

Index