MafDb.gnomAD.r2.1.GRCh38
March 13, 2024

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 from http://gnomad.broadinstitute.org/downloads. The original data were derived from the human genome release GRCh37 and the data in this package were derived by lifting the associated genomic positions over to GRCh38. See the inst/extdata/README file from the source code for more information on how these data have been stored into this annotation package.

Author(s)

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Source


The Genome Aggregation Database (gnomAD), Cambridge, MA (URL: http://gnomad.broadinstitute.org) [April 2019, accessed]
See Also

GScores-class gscores GenomicScores

Examples

```r
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.

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

* data
  MafDb.gnomAD.r2.1.GRCh38-package, 1

* package
  MafDb.gnomAD.r2.1.GRCh38-package, 1

GenomicScores, /, 2
GScores, /
gscores, 2
GScores-class, 2

MafDb.gnomAD.r2.1.GRCh38, /
MafDb.gnomAD.r2.1.GRCh38
  (MafDb.gnomAD.r2.1.GRCh38-package), 1
MafDb.gnomAD.r2.1.GRCh38-package, 1