

MafH5.gnomAD.r3.0.GRCh38

August 27, 2020

MafH5.gnomAD.r3.0.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 3.0 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. MAF data are stored using a HDF5 backend, which minimizes de main memory footprint but whose performance depends on the hard disk drive (HDD) reading performance. For this reason, this packages will perform faster if it is installed in a local HDD rather than in network HDD (e.g., through NFS access).

Please consult the gnomAD FAQ page at <http://gnomad.broadinstitute.org/faq> before you use these data for your own research.

Format

[MafH5.gnomAD.r3.0.GRCh38](#) [GScores](#) object containing MAF values from gnomAD genomes downloaded on October

Author(s)

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Source

Karczewski et al. The mutational constraint spectrum quantified from variation in 141,456 humans. bioRxiv, 531210, 2020.

The Genome Aggregation Database (gnomAD), Cambridge, MA (URL: <http://gnomad.broadinstitute.org>) [October 2019, accessed]

See Also

[GScores-class gscores](#) [GenomicScores](#)

Examples

```
library(SNPlocs.Hsapiens.dbSNP149.GRCh38)
library(MafH5.gnomAD.r3.0.GRCh38)

ls("package:MafH5.gnomAD.r3.0.GRCh38")

mafH5 <- MafH5.gnomAD.r3.0.GRCh38
mafH5
citation(mafH5)

populations(mafH5)

## lookup allele frequency for rs1129038, a SNP associated with blue and brown eye colors
## (Eiberg et al., Human Genetics, 2008). 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(mafH5, rng)
gscores(mafH5, GRanges("15:28111713"))

## lookup allele frequency for rs333, a deletion of 32 nucleotides (delta 32) within the CCR5 gene
## associated with resistance to an infection by HIV, the virus that causes AIDS. The homozygous
## state of this delta 32 allele has been reported to be highly protective against HIV-1
## infection (Huang et al., Nature Medicine, 1996) but with no effects on lifespan (Maier et al.,
## bioRxiv, 2019, Gudbjartsson et al., bioRxiv, 2019; Karczewski et al., bioRxiv 2019).

gscores(mafH5, GRanges("3:46373452-46373484"), type="nonsnrs")
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