MafH5.gnomAD.v4.0.GRCh38

November 11, 2026

`MafH5.gnomAD.v4.0.GRCh38-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 version 4.0 of the Genome Aggregation Database (gnomAD). The data are exposed to the user in the form of a `GScores` object named after the package. 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 drive (e.g., through NFS access).

This package stores MAF values derived from the global AF values (denoted as the `AF` population) and the maximum MAF value among all the populations in gnomAD (denoted as the `AF_allpopmax`). The `AF_allpopmax` MAF values are different from those that could be derived from the gnomAD `AF_popmax` values in that they do not exclude any population when taking the maximum MAF value; see https://github.com/broadinstitute/gnomad-browser/issues/893#issuecomment-1092142309. If you have any question or request regarding the gnomAD data stored in this package, please open an issue at https://github.com/rcastelo/GenomicScores/issues.

Please consult the gnomAD FAQ page at https://gnomad.broadinstitute.org/help#frequently-asked-questions before you use these data for your own research.

Format

`MafH5.gnomAD.v4.0.GRCh38` `GScores` object containing MAF values from gnomAD genomes downloaded on February 202

Author(s)

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Source

Chen et al. A genomic mutational constraint map using variation in 76,156 human genomes. Nature, 625:92-100, 2024.

The Genome Aggregation Database (gnomAD), Cambridge, MA (URL: https://gnomad.broadinstitute.org) [February 2024, accessed]

See Also

GScores-class gscores GenomicScores

Examples

```
library(SNPlocs.Hsapiens.dbSNP149.GRCh38)
library(MafH5.gnomAD.v4.0.GRCh38)

ls("package:MafH5.gnomAD.v4.0.GRCh38")

mafh5 <- MafH5.gnomAD.v4.0.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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