MafDb.ExAC.r1.0.GRCh38
May 8, 2024

Description

This annotation package stores minor allele frequency (MAF) data derived from the variant set release 1.0 of the Exome Aggregation Consortium (ExAC). 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 ExAC FAQ page at http://exac.broadinstitute.org/faq before you use these data for your own research.

Format

MafDb.ExAC.r1.0.GRCh38 GScores object containing MAF values from ExAC downloaded on March 2018 from http://exac.broadinstitute.org/downloads

Author(s)

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Source


The Exome Aggregation Consortium (ExAC), Cambridge, MA (URL: http://exac.broadinstitute.org) [March, 2018, accessed]
See Also

GScores-class gscores GenomicScores

Examples

library(SNPlocs.Hsapiens.dbSNP149.GRCh38)
library(MafDb.ExAC.r1.0.GRCh38)

ls("package:MafDb.ExAC.r1.0.GRCh38")

mafdb <- MafDb.ExAC.r1.0.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"))
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