MafDb.ESP6500SI.V2.SSA137.hs37d5

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MafDb.ESP6500SI.V2.SSA137.hs37d5-package

Annotation package for minor allele frequency data from the NHLBI ESP project

Description

This annotation package stores minor allele frequency (MAF) data values from the release ESP6500SI-V2 of the NHLBI Exome Sequencing project (ESP). The data are exposed to the user in the form of a MafDb 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 MafDb objects are found in the VariantFiltering 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.

Format

MafDb.ESP6500SI.V2.SSA137.hs37d5 MafDb object containing MAF values from 6503 exomes downloaded in March 2017 from http://evs.gs.washington.edu/evs_bulk_data/ESP6500SI-V2-SSA137.GRCh38-liftover.snps_indels.vcf.tar.gz. See the inst/extdata/README file from the source code for more information on how to update these data.

Author(s)

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Source


Exome Variant Server, NHLBI GO Exome Sequencing Project (ESP), Seattle, WA (URL: http://evs.gs.washington.edu/EVS) [March, 2017, accessed]

See Also

MafDb-class mafByOverlaps mafByld VariantFiltering
Examples

library(MafDb.ESP6500SI.V2.SSA137.hs37d5)

ls("package:MafDb.ESP6500SI.V2.SSA137.hs37d5")

mafdb <- MafDb.ESP6500SI.V2.SSA137.hs37d5
mafdb

populations(mafdb)

## lookup allele frequencies for rs1129038, an SNP associated to blue and brown eye colors
## as reported in 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.
rng <- GRanges("15", IRanges(28356859, 28356859))
mafByOverlaps(mafdb, rng)
mafByOverlaps(mafdb, "15:28356859-28356859")
mafByOverlaps(mafdb, "15:28356859")
mafById(mafdb, "rs1129038")
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