

MafDb.ExAC.r0.3.1.snvs.hs37d5

February 28, 2017

MafDb.ExAC.r0.3.1.snvs.hs37d5-package

Annotation package for minor allele frequency data from the Exome Aggregation Consortium

Description

This annotation package stores minor allele frequency (MAF) data for single derived from the single nucleotide variant set release 0.3.1 of the Exome Aggregation Consortium. The data is loaded automatically in the form of a MafDb2 object. The name of the exposed object matches the name of the package and part of the filename that contained the data imported into the package. The class definition and methods to access MafDb2 objects are found in the [VariantFiltering](#) software package.

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

Format

[MafDb.ExAC.r0.3.1.snvs.hs37d5](#) MafDb2 object containing MAF values from the Exome Aggregation Consortium d

Author(s)

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Source

Lek M et al. Analysis of protein-coding genetic variation in 60,706 humans. *Nature*, 536:285-291, 2016.

The Exome Aggregation Consortium (ExAC), Cambridge, MA (URL: <http://exac.broadinstitute.org>) [September, 2016, accessed]

See Also

[MafDb2-class mafByOverlaps mafById VariantFiltering](#)

Examples

```
library(MafDb.ExAC.r0.3.1.snvs.hs37d5)

ls("package:MafDb.ExAC.r0.3.1.snvs.hs37d5")

mafdb <- MafDb.ExAC.r0.3.1.snvs.hs37d5
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.
## Human Genetics, 123(2):177-87, 2008 [http://www.ncbi.nlm.nih.gov/pubmed/18172690]
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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