# MafDb.ESP6500SI.V2.SSA137.hs37d5

February 28, 2017

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`MafDb.ESP6500SI.V2.SSA137.hs37d5-package`
*Annotation package for minor allele frequency data from the NHLBI ESP project*

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## 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 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.

## Format

`MafDb.ESP6500SI.V2.SSA137.hs37d5` MafDb object containing MAF values from 6503 exomes downloaded in Septer

## Author(s)

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## Source

Tennessen JA, et al. Evolution and functional impact of rare coding variation from deep sequencing of human exomes. Science, 337:64-69, 2012.

Exome Variant Server, NHLBI GO Exome Sequencing Project (ESP), Seattle, WA (URL: `http://evs.gs.washington.edu/EVS`) [September, 2016, accessed]

## See Also

MafDb.1Kgenomes.phase1.hs37d5 MafDb.1Kgenomes.phase3.hs37d5 MafDb2-class mafByOverlaps mafById 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.
## 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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