MafDb.ExAC.r0.3.sites

MafDb.ExAC.r0.3.sites-package

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

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Description

This annotation package stores minimum allele frequency (MAF) data derived from the variant set release 0.3 of the Exome Aggregation Consortium. The data is stored in the form of a SQLite database and is loaded automatically in the form of a MafDb 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 MafDb 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.sites-package   MafDb object containing MAF values from the Exome Aggregation Consortium downl

Author(s)

R. Castelo

Source

The Exome Aggregation Consortium (ExAC), Cambridge, MA (URL: http://exac.broadinstitute.org) [March, 2015, accessed]

See Also

makeMafDbPackageExAC MafDb.ALL.wgs.phase1.release.v3.20101123 MafDb.ALL.wgs.phase3.release.v5a.20130502
MafDb.ESP6500SI.V2.SSA137 MafDb-class snpid2maf VariantFiltering

Examples

```
library(MafDb.ExAC.r0.3.sites)

ls("package:MafDb.ExAC.r0.3.sites")

mafdb <- MafDb.ExAC.r0.3.sites
mafdb

knownVariantsMAFcols(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]
snpid2maf(mafdb, "rs1129038")
```

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makeMafDbPackageExAC   *Make a MafDb annotation data package from the Exome Aggregation*
                              Consortium

Description

This function creates an annotation data package for a MafDb object. Its primary purpose is to ease
the task of fetching and packaging newer minimum allele frequency (MAF) data. Note that MAF
values are processed and stored in a way to reduce their space in disk. Please consult the manual
page for the MafDb-class to know the details of these processing steps.

Usage

```
makeMafDbPackageExAC(destDir=path.expand("~"), MafDbURL=MafDbExACdefaultURL, MafDbPkgName=MafDbExACd
                 genome="hg19", version=NULL, author=NULL, maintainer=NULL, license=NULL, yieldSize=10
```

Arguments

destDir	Destination directory for the newly created package.
MafDbURL	URL to the source VCF file(s). By default, it points to the URL holding the data currently stored in the package that defines this function.
MafDbPkgName	Name of the newly created package. This will define as well the name of the exported MafDb object.
genome	Version of the human genome, following UCSC nomenclature. Necessary for the internal call to the readVcf() function from the VariantAnnotation package.

version	Version to put on the new package. By default, the version corresponds to the version of the package that defines this function, bumping the second version number.
author	Author to put on the new package. By default, the author corresponds to the author of the package that defines this function.
maintainer	Maintainer to put on the new package. By default, the maintainer corresponds to the maintainer of the package that defines this function.
license	License to put on the new package. By default, the license corresponds to the license of the package that defines this function.
yieldSize	In the case source tabix VCF files, they are not read at once, but scanned in batches whose size is determined by this argument. By default is set to one million variants but it may be reduced to lower main memory requirements.

Value

Path to the folder containing the created data package.

Author(s)

R. Castelo

See Also

[MafDb-class](#) [snpid2maf](#) [MafDb.ExAC.r0.3.sites](#)

Examples

```
MafDbExACdefaultURL ## default URL from where makeMafDbPackageExAC() fetches the MAF data

MafDbExACdefaultPkgName ## default name for the package that makeMafDbPackageExAC() creates

## Not run:
## the previous two default values can be overridden when calling makeMafDbPackageExAC()
makeMafDbPackageExAC()

## End(Not run)
```

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